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Selected Abstracts of the 19th International Workshop on Neonatology and Pediatrics

FROM WOMB TO AGING, FROM MEDICAL HISTORY TO ARTIFICIAL INTELLIGENCE

CAGLIARI (ITALY) • OCTOBER 18TH-21ST, 2023

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ABS 1

TOTAL HEAD GROWTH IN PATIENTS WITH PKU DIAGNOSED BY NEWBORN SCREENING TEST

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BACKGROUND

Phenylketonuria (PKU) is an inborn error of phenylalanine (PHE) metabolism caused by phenylalanine hydroxylase (PAH) enzyme deficiency. It is a relatively common disorder that is inherited as autosomal recessive disease. PKU may be diagnosed by newborn screening test. The diagnosis is established when plasma PHE concentration is persistently above 2 mg/dL (120 μmol/L). PKU can cause microcephaly, intellectual disability, attention deficit hyperactive disorder, epilepsy and eczema due to accumulated toxic effect of PHE in the blood. The study looks for whether PHE control can improve head growth in early diagnosed and treated children.

METHODS

The head circumference records of 28 early-detected patients with PKU at diagnosis and at 3 month intervals until the age of 3 years were studied. These measurements were compared between the following 2 groups: those with PHE median level below 360 μmol/L and never above 600 μmol/L, and those with levels higher than 600 μmol/L.

RESULTS

In patients with PKU diagnosed by newborn screen test, higher occipitofrontal circumference Z-score readings and hence further head growth were noticed among children with relatively lower PHE levels at diagnosis (27.28% and 38.89%, respectively) than those with higher levels (10% and 20%, respectively) with a significant difference. The same finding was also observed at 3 years of age, but was not statistically significant: the difference exceeded the acceptable level of significance (p-value = 0.057).

CONCLUSION

In patients with PKU diagnosed by newborn screen test, the higher PHE level at diagnosis, the smaller the head size compared with those with lower PHE. The head growth at 3 year of age of those with stricter PHE control does not vary considerably from patients with less strict control.

ABS 2

ARTIFICIAL INTELLIGENCE AND POSTPARTUM DEPRESSION: THE "TALKING ABOUT" ALGORITHM

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Postpartum depression (PD) is the most widespread perinatal psychiatric disorder, also representing the non-obstetric most frequent birth-related complication. From an epidemiological point of view, it has an average prevalence of 17-18% worldwide. The consequences of this pathology affect the health of both the mother and the child, who could have to face altering of its affective, behavioral and social development, but also the relationship with the partner (including paternal PD). Therefore, an early diagnosis appears of fundamental importance to promptly introduce adequate therapy.

Talking About, by the company GPI (Trento, Italy), is a project focused on voice analysis as a medium to access human emotions. It consists of a series of speech emotional recognition (SER) algorithms. The aim of the study is to evaluate the application of the artificial intelligence (AI) algorithm Talking About on the mothers' emotions analysis. Talking About investigates the unconscious aspects of voice that usually cannot be controlled or voluntarily modified, aiming at identifying the subjects' emotions. Thus, all biases, characterizing all classic screening questionnaires, should be neutralized, achieving a sharper overview of the mothers' emotional state.

The mother's emotions results are displayed in 5 main categories: 2 positive, 3 negative.

In this study, a total 154 mothers – who gave birth at the "Policlinico Universitario D. Casula" and/or carried out a pediatric examination at the "Ambulatorio SOS MAMI" (which is a PostNatal Care Service) – were enrolled. They underwent both the Edinburgh Postnatal Depression Scale (EPDS) and Talking About voice tests.

Despite this study's sample limitations, preliminary data related to PD symptoms identification are promising and encouraging, leading the way to further investigations related to the application of AI as a medium of PD screening support. Further studied in the next future are needed to explore this topic, even considering PD of the father.

REFERENCES

- Braun S, Tashev I. On training targets for noise-robust voice activity detection. arXiv. 2021;2021:2102.07445. [Preprint].
- Fanos V, Dessì A, Deledda L, Lai A, Ranzi P, Avellino I, Marinaro F, Petza S, Pintus R, Oliverio G, Vitale SG, Angioni S, Colangelo A. Postpartum depression screening through artificial intelligence: preliminary data through the Talking About algorithm. J Pediatr Neonat Individual Med. [In press].
- Fanos V, Fanos T, Corridori M. Come riconoscere e affrontare la depressione post partum. Le risposte del pediatra e dello psichiatra alle domande dei genitori. Quartu Sant'Elena: Hygeia Press, 2023.
- Fanos V, Fanos T. La depressione post partum. Cause, sintomi e diagnosi. Quartu Sant'Elena: Hygeia Press, 2013.
- Luo Y, Mesgarani N. Conv-TasNet: Surpassing Ideal Time-Frequency Magnitude Masking for Speech Separation. IEEE/ACM Trans Audio Speech Lang Process. 2019;27(8):1256-66.
- Zheng C, Peng X, Zhang Y, Srinivasan S, Lu Y. Interactive Speech and Noise Modeling for Speech Enhancement. arXiv. 2021;2021:2012.09408. [Preprint].

ABS 3

PEDIATRIC ONCOLOGICAL SPONDYLOLISTHESIS: THE CONTRIBUTION OF PHYSICAL AND REHABILITATION MEDICINE AND ORTHODONTICS

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Spondylolisthesis (SP) can arise from a congenital defect in the lumbar region or from acute trauma-induced injury. In active children and adolescents, it can result from stress fractures due to overuse. The initial symptom is typically pain. Although rare, SP can also occur as a result of cancer, potentially causing physical impairment and postural changes in the spine, including flatback and hyperkyphosis. These alterations can extend to the cervical region (forward neck) and head (forward head). Timely evaluation of pediatric oncological SP is essential, involving clinical and radiological assessments. A multidisciplinary team, including spinal specialists, pediatricians, oncologists, and psychologists, manages these cases, with orthodontists contributing to head posture evaluation [1]. This review provides a comprehensive overview, focusing on clinical assessment and rehabilitative possibilities for pediatric oncological SP. The clinical approach encompasses detailed medical history, thorough physical examination, and comprehensive radiological assessment. As for the lumbar spine, the evaluation of joint mobility, muscle strength, muscle palpation, peripheral neurological examination, balance, and gait is crucial. A comprehensive examination of the entire spine and head is advised to identify additional postural deviations, such as forward neck and head, their mobility (including temporomandibular joint), and neck/head muscle contractures. The Scoliosis Research Society-22 Patient Questionnaire (SRS-22), a self-administered outcome measure, is recommended to assess health-related quality of life in these cases [2]. When SP due to cancer is diagnosed in young individuals, surgery, and radiotherapy are often necessary to prevent significant morbidity and potential lower limb paralysis. Following oncological treatments, a combination of orthoses, exercise-based therapy, and psychological support is recommended. The engagement of various healthcare professionals, including spinal specialists, pediatricians, oncologists, psychologists, and orthodontists, is essential. Orthodontists assess postural changes related to teeth and jaw irregularities and temporomandibular disorders [3]. In conclusion, proper clinical and radiological evaluations are crucial for accurate assessments. Evidence-based multidisciplinary rehabilitation, incorporating braces, exercise therapy, and cognitive-behavioral therapy, plays a vital role in treating these children. A

multidisciplinary team involving spinal specialists, pediatricians, oncologists, psychologists, and orthodontists is highly recommended [4].

REFERENCES

[1] Frosch M, Mauritz MD, Bielack S, Blödt S, Dirksen U, Dobe M, Geiger F, Häfner R, Höfel L, Hübner-Möhler B, von Kalle T, Lawrenz B, Leutner A, Mecher F, Mladenov K, Norda H, Stahlschmidt L, Steinborn M, Stücker R, Trauzeddel R, Trollmann R, Wager J, Zernikow B. Etiology, Risk Factors, and Diagnosis of Back Pain in Children and Adolescents: Evidence- and Consensus-Based Interdisciplinary Recommendations. Children (Basel). 2022;9(2):192.

[2] Asher MA, Min Lai S, Burton DC. Further development and validation of the Scoliosis Research Society (SRS) outcomes instrument. Spine (Phila Pa 1976). 2000;25(18):2381-6.

[3] Bian C, Chen N, Li XL, Zhou XG, Lin H, Jiang LB, Liu WM, Chen Q, Dong J. Surgery Combined with Radiotherapy to Treat Spinal Tumors: A Review of Published Reports. Orthop Surg. 2016;8(2): 97-104.

[4] Scribante A, Pellegrini M, Ghizzoni M, Nardi MG, Rocca B, Monticone M. Pediatric oncological spondylolisthesis: the contribution of the Physical Medicine and Rehabilitation and Orthodontics. A narrative review. J Pediatr Neonat Individual Med. 2023;12(2):e120218.

ABS 4

A NEW PEDIATRIC INITIATIVE FOR CLINICAL GOVERNANCE: THE ITALIAN ASSOCIATION OF CHIEFS OF PEDIATRIC DIVISIONS

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The role of chief of a Pediatric Division has undergone profound changes over the years, which have led to a substantial modification of the professional duties of healthcare managers. Today, in addition to common medical duties towards patients, the modern chief has to pursue the objectives indicated by the healthcare company strategic management, to discuss the assigned budget, to take care of the performances of the assigned personnel and to settle staff disputes, to manage disciplinary procedures, to manage health technology assessment, to know all the different work contracts, to interact with risk managers, to manage public complaints and lawsuits. Moreover, he needs to be skilled with media companies, he has to be able to give good news and bad news, and to handle frequent audits and incident reports. In order to get all these abilities, in Italy he attends specific courses to become a healthcare manager.

A "scientific society" or a "syndicate" cannot handle properly all the previous duties of a modern chief, that's why on the 4th of October 2022, in Rome, a group of 10 hospital chiefs of Pediaric Divisions started a new association (*Coordinamento Nazionale dei Primari di Pediatria* – CONAPP) which could gather and support healthcare managers of private or public, teaching or not, Pediatric Divisions. In the previous years, other medical specialties (e.g., oncology, vascular surgery, etc.), started national associations of chiefs, but in pediatrics no one was active.

Up to now, 131 Italian chiefs of Pediatric or Neonatal Divisions joined the association, a national conference was held in June 2023 in Rome, a first national board was nominated, and a statute was validated and authenticated by a notary public. A database of all the Pediatric Divisions in Italy was started and some documents with healthcare organization proposals were addressed to the Ministry of Health, and to the Director of the National Agency for Regional Healthcare Systems (*Agenzia Nazionale per i Servizi Sanitari Regionali* – AGENAS).

Among its main objectives, this association takes care of the professional training of its members about healthcare management, clinical governance and occupational safety. By the interaction with the Italian Society of Pediatrics (*Società Italiana di Pediatria* – SIP) and other national pediatric associations and scientific societies, the association also contributes to identifying and solving the problems of health policies relating to children and their families. In particular, it intends to strengthen the Pediatric Divisions, the reorganization of the pediatric area and the relationships with the territorial health authorities.

In conclusion, the new association was founded to give adequate support to the new modern chiefs of

Pediatric and Neonatal Divisions in carrying out their work, considering that the future horizon will no longer be that of treating children in the best possible way only, but also of interpreting this new "managerial" aspect of the chief's activity, which has become mandatory.

REFERENCES

- Gafoor A, Thaker S, Gupta H, Botchu R. Life as a clinical director: an insight. Clin Radiol. 2020;75(8):640.e13-640.e16.
- Perez J. Leadership in Healthcare: Transitioning From Clinical Professional to Healthcare Leader. J Healthc Manag. 2021;66(4):280-302.
- van Rossum L, Aij KH, Simons FE, van der Eng N, Ten Have WD. Lean healthcare from a change management perspective. J Health Organ Manag. 2016;30(3):475-93.

ABS 5

A RARE VARIANT OF BECKER MUSCULAR DYSTROPHY

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CASE REPORT

A child, third born, son of a mother affected by minor psychiatric problems and a father of Hispanic origin, was born via spontaneous delivery and was left in the custody of the mother with supervision by the social services. At 2 months of age, a 1/6 systolic murmur was highlighted (at the cardiological exam, the diagnosis of mitral insufficiency was made). He was fed with adapted milk; he had a stunting growth from 6 months of age, which improved after 2 years. At 15 months, he had COVID-19, from which he recovered without relics. At 16 months, a neuropsychiatric evaluation for mild psychomotor retardation was demanded. At 23 months, a second check-up in the Child Neuropsychiatry Department with generic diagnosis of mild psychomotor retardation was done. At 27 months, the mother reported that the child had a further regression in language, and that he trips over himself when running, falls from his chair and aggressive attitudes had been reported from school. A day after this interview, he fell out of a chair and suffered a minor head injury. Blood test control was performed, and a creatine phosphokinase (CPK) value of 1,200 IU/L was found. So the hospitalization in the Neuropsychiatry Department for further diagnostic investigations was programmed. While waiting for the hospitalization, a convulsive crisis appeared in apyrexia during an intercurrent episode of acute streptococcal tonsillitis (diagnosis with swab) under antibiotic therapy. When the child was admitted to hospital, the CPK value was confirmed at 1,300 IU/L. The value persisted in subsequent checks, and, even if the motor delay was slight, a possible muscular dystrophy due to dystrophin protein deficiency was suspected. The molecular investigation Multiplex Ligation-dependent Probe Amplification (MLPA) [1], which allows the recognition of deletions or duplications (in our case) of the dystrophin gene, was performed. The gene encodes the formation of the protein essential for the functioning of the muscular districts of our body. In this case, the presence of the duplication of exon 4 was identified. The diagnosis was Becker muscular dystrophy (BMD), which is caused by a malfunction of the dystrophin gene due to deletions, mutations or in-frame duplications in the *DMD* gene (Xp21.2). The frequency is 1:18,500 of male births. This duplication of exon 4 is poorly described in the scientific literature and is very rare [2]. This suggests an intermediate clinical phenotype, as it allows the formation of a smaller protein than it should be, but functional. This variant of BMD is expected to be the mildest form of the dystrophin defect. The symptom onset, which is usually late, was early. In its form, the course will be slowly progressive, with loss of walking which may occur after the age of 30 (approximately 1/3 of patients). The most significant problem in terms of prognosis will be the heart problem (cardiomyopathy). Transmission is of the recessive type linked to the X chromosome. In about 30% of cases, the mutations are not transmitted from the mother (*de novo*). In our case, a muscle biopsy quantified the presence of the dystrophin protein in the muscle tissue, and this will be useful for treatments with prednisone, ACE inhibitors and/or beta blockers, which will be symptomatic for now; we will evaluate in the future whether it will be possible to use Givinostat [3] until exon skipping.

REFERENCES

[1] Huamán-Dianderas FD, Guevara-Fujita ML, Málaga DR, Estrada-Cuzcano A, Fujita R. [Detection of mutations causing Duchenne and Becker muscular dystrophies: multiplex polymerase chain reaction vs. multiplex ligation dependent probe amplification]. [Article in Spanish]. Rev Peru Med Exp Salud Publica. 2019;36(3):475-80.

[2] Lipatova NA, Krakhmaleva IN, Shishkin SS, Shakhovskaia NI, Podnikova NI, Lunga IN, Tarksh MA, Gerasimova NL. [Polymorphism of exon 4 in the CANP-3 gene in patients with primary myopathies]. [Article in Russian]. Genetika. 1999;35(12):1713-7.

[3] Comi GP, Niks EH, Vandenborne K, Cinnante CM, Kan HE, Willcocks RJ, Velardo D, Magri F, Ripolone M, van Benthem JJ, van de Velde NM, Nava S, Ambrosoli L, Cazzaniga S, Bettica PU. Givinostat for Becker

muscular dystrophy: A randomized, placebo-controlled, double-blind study. Front Neurol. 2023;14:1095121.

ABS 6

HUMAN HERPESVIRUS-6 INFECTION AS A CAUSE OF RHABDOMYOLYSIS IN TODDLER

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INTRODUCTION

Human Herpes virus-6 (HHV6) is the sixth member of the Herpes virus family, and it is responsible for a ubiquitous infection in the first 2 years of life. Beyond the classic clinical presentation, the potential complications include neurological diseases, hepatitis, thrombocytopenia, myocarditis, and rhabdomyolysis [1]. Rhabdomyolysis is defined by an increase in creatine phosphokinase (CK) values more than 1,000 IU/L, secondary to skeletal muscle damage. It is characterized by a classic triad: myalgia, weakness, and dark urine. As for etiology, trauma, exercise, drugs, infections, and hereditary diseases are the most common causes. In children, it is frequently linked to viral myositis with a specific muscle tropism. An aggressive and timely hydration can preserve kidney function and prevent complications [2].

CASE REPORT

A 3-years-old Sardinian girl was admitted to our hospital for a hyperpyrexia for 5 days, not responsive to therapy, with lower limb pain and vomiting. The patient's medical history and her family history were unremarkable. When admitted, blood tests showed neutropenia, mild thrombocytopenia and high values of CK (1,439 IU/L; normal values: 0-145 IU/L) with negative C-reactive protein. The urine test showed dark yellow color, clear appearance, urobilinogen 1, and albumin 10 mg/L. Serologic tests for Adenovirus, Cytomegalovirus, Parvovirus B19, HHV7 and Epstein-Barr virus were all negative. Abdominal ultrasound was performed, with increased spleen finding. A microscopic examination of blood was performed, with evidence of some atypical activated lymphocytes, as for a mononucleosis-like condition. Moreover, for a differential diagnosis, a blood test for HHV6 was performed, with a positive response to this virus (63,281 copies/ml; detection limit: 250 copies/ml). Based on the high values of CK, the patient was diagnosed with rhabdomyolysis. Intravenous hydration with balance saline solution was started until the third day of hospitalization. During the clinical course, there was a progressive improvement in clinical symptoms, and a normalization of CK values and CBC. The patient did not develop renal failure, and her neurodevelopmental status was normal after hospital discharge.

CONCLUSION

Currently, rhabdomyolysis in children with primary HHV6 infection has been reported in only 2 cases [3]. The aim of our work was to highlight a correlation between HHV6 and rhabdomyolysis in another pediatric case, emphasizing a different clinical presentation of HHV6 and the possible prevention of complications with appropriate therapies.

REFERENCES

[1] Tesini BL, Epstein LG, Caserta MT. Clinical impact of primary infection with roseoloviruses. Curr Opin Virol. 2014;9:91-6.

[2] Aricò MO, Belleri P, Cattazzo E, Del Vecchio L, Guariento M, Pagano G, La Fauci G. Approccio al bambino con iperCKemia e rabdomiolisi in pronto soccorso pediatrico. Quaderni ACP. 2022;29(5):200-5.

[3] Murakami R, Adachi S, Koga H. Rhabdomyolysis Associated With Primary Human Herpesvirus-6 Infection. Pediatr Infect Dis J. 2019;38(12): e341.

ABS 7

BREASTFEEDING FOLLOW-UP: FROM BIRTH TO 6 MONTHS OF CORRECT AGE IN A COHORT OF VLBW NEWBORNS

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INTRODUCTION

Breastfeeding provides multiple health and development benefits even for preterm infants. Breast milk is universally considered the best nutritional source for preterm infant, as it has been shown to be able to provide adequate amounts of macronutrients to ensure appropriate growth and reduce incidence and severity of complication related to prematurity. Nevertheless, there are few studies in the literature investigating the duration of breastfeeding and the type of nutrition after discharge from the Neonatal Intensive Care Unit (NICU).

AIMS
We carried out a retrospective cohort study at the Neonatal Unit of University of Turin with the aim of evaluating whether the breastfeeding promotion and support techniques practiced in the NICU were actually able to encourage breast milk nutrition and breastfeeding, not only during the hospitalization, but up to 6 months of corrected age (CA) of the infants.
METHODS
We conducted a retrospective observational study. We included in the study all infants born below 31 completed weeks of gestational age and/or with birth weight lower than 1,500 g during the 4-year period 2019-2022. By consulting medical records, we investigated maternal, pregnancy and delivery, neonatal and breastfeeding clinical variables.
RESULTS
Among 309 infants included in the study, 193 were very preterm and 58 extremely preterm, 170 were very low birth weight and 81 extremely low birth weight. We observed a significant reduction in human milk feeding and breastfeeding in the infant cohort during the first 6 months of CA: 47.7% took human milk at discharge and only 7.6% continued until 6 months of CA. We also noted stability in the frequency of exclusively breastfed infants over the follow-up period, suggesting that once successful breastfeeding is initiated, it is continued until the first 6 months of CA. Multivariate analysis showed how medically assisted reproduction, intrauterine growth restriction, type of labor and type of delivery and, among neonatal variables, being small for gestational age, late sepsis, surfactant administration, bronchopulmonary dysplasia and patent ductus arteriosus are correlated with the incidence of breastfeeding, making it more difficult.
CONCLUSION
It is possible to state that the breastfeeding promotion and support techniques practiced in the NICU guarantee a high intake of breast milk at discharge; however, a reduction in human milk nutrition can be observed from the first few weeks. This demonstrates, on the one hand, the importance of providing adequate intra-hospital assistance to initiate breastfeeding as soon as possible and, on the other hand, the need to provide support especially after hospital discharge to promote breastfeeding until weaning age.

ABS 8

SIXTH CRANIAL NERVE PALSY: NOT ALL IS AS IT SEEMS!

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INTRODUCTION
Petrous apicitis (PA) is an infectious process of the petrous apex and represents a rare complication of acute otitis media (AOM). The extradural inflammation caused by PA can involve the V and VI cranial nerve, causing Gradenigo syndrome with otorrhea, trigeminal oculo-orbital pain and abducens palsy, associated with fever and general discomfort [1, 2].
We describe an unusual case of PA in a child who arrived to the Emergency Room (ER) with suggestive features of meningoencephalitis associated with VI cranial nerve paralysis without signs or history of AOM.
CASE REPORT
A 4-year-old male presented to ER with fever and rhinitis lasting 4 days, associated with headache and vomiting during the last 24 hours. The physical examination showed normal vital signs, preferred lateral decubitus, GCS of 14/15, convergent strabismus in the left eye (acute onset), photophobia, weakly positive meningeal signs. Blood tests: WBC 23,100/mmc, N 83%, CRP 10 mg/dl, PCT 0.8 ng/ml, other laboratory tests normal. To rule out meningoencephalitis, we performed a brain CT scan which was normal, a lumbar puncture with clear CSF, rapid drop, and increased leukocytes (WBC 766/mm^3, N 98%, normal glucose and proteins) and EEG with slowed background activity on both hemispheres. Therefore, i.v. ceftriaxone and acyclovir were started. Microbiological investigations on CSF and blood were negative (blood culture, CSF, and viral PCR). Nasopharyngeal swab resulted positive for Adenovirus and Rhinovirus. During the admission there was a clinical improvement with apyrexia and negativization of the inflammation indices. However, due to the persistence of left esotropia, a control lumbar puncture was performed with clear CSF, and slow drop; its chemical-physical analysis, autoimmunity panel and oligoclonal bands were negative. An orthoptic examination confirmed paralysis of the left VI cranial nerve and brain MRI was suggestive of left PA with phlebitis of the cavernous sinus and ipsilateral chronic mastoiditis. Therefore, he discontinued acyclovir and performed an ENT

evaluation, which highlighted intact left tympanic membrane, with no signs of mastoid inflammation, and it was recommended to continue conservative therapy (ceftriaxone and methylprednisolone/prednisone x 14 days). Follow-up showed EEG normal and brain angio MRI improving. Due to persistence of VI cranial nerve paralysis, in the absence of other neurological deficits, ophthalmological follow-up was undertaken.

CONCLUSIONS

In our case, VI cranial nerve paralysis appears to be induced by two infectious processes whose correlation and etiology remained, however, unknown: acute meningoencephalitis (with clinical, EEG and CSF supportive findings) and PA associated with cavernous sinus phlebitis and chronic mastoiditis. Even without objective and anamnestic signs of AOM, in the presence of VI cranial nerve palsy, it is suggested to perform brain MRI for early identification of PA, which requires targeted medical treatment and sometimes a surgical approach.

REFERENCES

[1] Choi KY, Park SK. Petrositis with bilateral abducens nerve palsies complicated by acute otitis media. Clin Exp Otorhinolaryngol. 2014;7(1):59-62.

[2] Price T, Fayad G. Abducens nerve palsy as the sole presenting symptom of petrous apicitis. J Laryngol Otol. 2002;116(9):726-9.

ABS 9

THE EMERGENCE OF CONSCIOUSNESS-STATE DEPENDENT COMPLEXITY: SEPs AND PERTURBATION COMPLEXITY INDEX IN NEWBORNS AND YOUNG INFANTS

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INTRODUCTION

In adults, sensory and nonsensory stimuli are processed differently during sleep and wakefulness, with greater complexity occurring during wakefulness.

AIMS

This study aims to investigate whether this way of processing stimuli is innate or experience-dependent; therefore, we assessed whether this difference between physiological behavioral states is already present at birth. Secondly, we examined whether this difference, if present, overlaps with the one observed in adult subjects and, if not, when it occurs.

METHODS

A between and within subjects analysis (2X2 Design) was performed in two samples: the first one composed of 46 newborns between 12 and 76 hours of age, the second one comprising 22 infants between 2 and 4 months of age; in both cases, subjects were selected based on the fact that they were physiological. At these two different time points, starting from the electroencephalic activity, two measures were extracted and analyzed: Somatosensory Evoked Potentials (SEPs) and the Perturbational Complexity Index (PCIst) following tactile stimulation of the median nerve, both in the waking and sleeping states.

RESULTS

In the newborns sample, the comparison between sleep and wakefulness showed a significant difference, represented by higher PCIst values in sleep than in wakefulness, for each parameter combination. In the infants sample, the comparison between sleep and wakefulness showed a significant difference, represented by greater PCIst values in wakefulness than in sleep, for each parameter combination. The difference observed between the two subjects samples is determined by the PCIst value during sleep, which is high in newborns and then decreases in infants. Correlational analysis by age revealed a significant positive correlation between the PCIst and age in the infant sample, for each parameter combination.

CONCLUSIONS

Even in newborns, there is a difference in the complexity of stimuli processing between the two physiological behavioral states under investigation. However, this complexity is greater during sleep compared to wakefulness, indicating that the established pattern of stimuli processing observed in adults is not inherent from birth. The mature pattern begins to be observed from the third month of age, as the PCIst is higher during wakefulness and exhibits an increase with advancing age. This suggests that experience plays a crucial role in the

developmental progression of this process during this stage of life. The elevated PCIst value observed during sleep in the sample of newborns has been correlated with the distinct role that sleep plays during the different stages of life. In newborns, specifically, it seems to promote the maturation of functional connections in the brain.

ABS 10

URINARY GC-MS METABOLOMICS OF SARDINIAN CYSTIC FIBROSIS PATIENTS REVEALS UNIQUE MUTATION-CLASS DEPENDENT SIGNATURES

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Cystic fibrosis (CF) is an autosomal recessive disease caused by mutations in the CF transmembrane conductance regulator (*CFTR*) gene [1]. Its product, the CFTR protein, mainly acts as an ion channel, and its impairment results in an increased density and viscosity of secretion [2]. CF displays high phenotypic variability because of the intervention of genetic and environmental factors [3]. In this context, metabolomics represents a useful tool to investigate pathology's complexity, providing further insights into the pathophysiology and contributing to clinical biomarkers identification. GC-MS is one of the most widely used techniques for metabolomics studies due to its high resolution, sensitivity, reproducibility, and relatively low cost. Urine samples from 35 patients affected by CF and with different genotypes (F508del/F508del, T338I/T338I, and F508del/T338I) were collected. An aliquot of urine was evaporated to dryness, derivatized, and diluted with hexane. Finally, samples were analyzed with GC-MS technique. Identification of metabolites was performed using the standard NIST 08 library. Chromatogram analysis was performed using MassHunter software, and the resulting data were normalized by total area before undergoing statistical analysis. The supervised multivariate statistical analysis, based on the GC-MS data, allowed the differentiation of the samples based on the metabolomic profile. In particular, a good separation between F508del/F508del vs T338I/T338I genotypes, and F508del/F508del vs F508del/T338I genotypes was observed. Moreover, the comparison of the two groups T338I/T338I vs F508del/T338I did not highlight significant differences. The variable of importance responsible for the separation were sugars (glucose, maltose), organic acid (citric acid, 3-[3-hydroxyphenyl]-3-hydroxypropionic acid and 3-hydroxyisovaleric acid), and polyols, such as inositol isoforms. The classification obtained corresponded to the different phenotypes under investigation. So, the different metabolomic profiles are consistent with pathological conditions and clinical symptoms. In conclusion, urine metabolomics has proven to be a useful tool to discriminate among the different subclasses of CF, mirroring the complexity of the pathological condition. Overall, this study creates a basis for future metabolic indicators of CF patients.

REFERENCES

[1] Bierlaagh MC, Muilwijk D, Beekman JM, van der Ent CK. A new era for people with cystic fibrosis. Eur J Pediatr. 2021;180(9):2731-9.

[2] Figueira MF, Ribeiro CMP, Button B. Mucus-targeting therapies of defective mucus clearance for cystic fibrosis: A short review. Curr Opin Pharmacol. 2022;65:102248.

[3] Butnariu LI, Țarcă E, Cojocaru E, Rusu C, Moisă ȘM, Leon Constantin MM, Gorduza EV, Trandafir LM. Genetic Modifying Factors of Cystic Fibrosis Phenotype: A Challenge for Modern Medicine. J Clin Med. 2021;10(24):5821.

ABS 11

EXPLORING THE RELATIONSHIP AMONG MRI SCORING SYSTEMS AND NEURODEVELOPMENTAL OUTCOME AT 2 YEARS IN INFANTS WITH HIE AFTER THERAPEUTIC HYPOTHERMIA

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INTRODUCTION

Hypoxic-ischemic encephalopathy (HIE) is a major worldwide cause of neonatal mortality and morbidity. HIE occurs in 1 to 8 per 1,000 live births in developed countries [1]. The introduction of therapeutic hypothermia (TH) as treatment for HIE has decreased mortality and increased disability-free survival rate in neonates with moderate to severe encephalopathy [2]. Nevertheless, around 40% of survivors still demonstrate an increased risk for cognitive deficits and behavioural problems both across toddler age and childhood [3].

AIMS

This study aimed to explore the relationship between Barkovich (BSS) and Okereafor (OSS) magnetic resonance scorings and neurodevelopmental outcome at 24 months of age in infants with neonatal HIE treated with TH; it also aimed to investigate prognostic accuracy of MRI scoring combined with the Hammersmith Infant Neurological Examination (HINE) at 12 months to detect neurodevelopmental impairments.

METHODS

Observational, prospective, single-centre study design. Thirty-nine infants born at ≥ 35 weeks of gestational age with moderate-to-severe HIE who underwent TH from 2017 to 2021 were recruited. Infants were scanned after rewarming and within the first postnatal week of life and underwent HINE at 12 months of age and Griffiths scales at 24 months.

RESULTS

Both OSS and BSS were associated with gross motor skills. The OSS was also associated with social, cognitive and linguistic outcomes. The area under the ROC curve for the OSS was 0.9062 (Youden index cut-point = 5, sensitivity = 69%, specificity = 100%). The BSS showed that AUC was 0.819 (Youden index cut-point = 1, sensitivity = 57%, specificity = 100%). The OSS combined with HINE at 12 months increased its sensitivity (100%).

CONCLUSIONS

The OSS could be a suitable tool to investigate emerging social, cognitive and linguistic deficits in HIE infants through the first 2 years of life and prevent high-order cognitive deficits. Moreover, the OSS combined with HINE at 12 months of age provides optimal identification of neurodevelopmental impairments at 24 months. Further research could compare the prognostic accuracy of the existing MRI scoring systems using large samples of newborns and multi-centric study design. While the frequency of major motor sequelae has decreased in infants with HIE treated with TH, other neurodevelopmental impairments emerged. This study highlights that the OSS is sensitive in cognitive, social and linguistic skills. The OSS, added to the neurological follow-up program, could improve prognostic accuracy of neurodevelopmental outcome at 24 months. The results of this study are useful for clinicians involved in the follow-up of HIE infants to adequately counsel parents about their child's development and implement early focused interventions.

REFERENCES

[1] Douglas-Escobar M, Weiss MD. Hypoxic-ischemic encephalopathy: a review for the clinician. JAMA Pediatr. 2015;169(4):397-403.

[2] Azzopardi D, Strohm B, Marlow N, Brocklehurst P, Deierl A, Eddama O, Goodwin J, Halliday HL, Juszczak E, Kapellou O, Levene M, Linsell L, Omar O, Thoresen M, Tusor N, Whitelaw A, Edwards AD; TOBY Study Group. Effects of hypothermia for perinatal asphyxia on childhood outcomes. N Engl J Med. 2014;371(2):140-9.

[3] Schreglmann M, Ground A, Vollmer B, Johnson MJ. Systematic review: long-term cognitive and behavioural outcomes of neonatal hypoxic-ischaemic encephalopathy in children without cerebral palsy. Acta Paediatr. 2020;109(1):20-30.

ABS 12

A FEMALE INFANT WITH GESTATIONAL AGE 22^{+4} WEEKS. A CASE REPORT

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INTRODUCTION

Extremely low birth weight (ELBW) neonates comprise less than 1% of all births. However, due to the various medical interventions such as respiratory, cardiovascular, and nutrition support, and a long duration of hospital stay, these neonates are a major proportion of the Neonatal Intensive Care Unit (NICU) census. Survival till discharge is not the only endpoint. Considering the high incidence of neurosensory disabilities such as motor or mental developmental delay, deafness and other long-term disabilities, the Units that report the outcome of ELBW neonates should also report the long-term neurological outcome. A case report of a female infant with gestational age 22^{+4} weeks in our NICU is presented below.

CASE REPORT

A female infant with gestational age 22^{+4} weeks was delivered by C-section due to premature rupture

of the amniotic membranes. A cervical cerclage was performed at 22^{+2} weeks due to cervical insufficiency. A uterine hemorrhage was reported on the same day. An antenatal dexamethasone treatment was received at 22^{+2} weeks.

Respiratory system

Due to complete lack of respiratory movements, a cardiovascular resuscitation with endotracheal intubation was necessary immediately after birth. Surfactant (Curosurf® 200 mg/kg) was administrated within the first hour due to respiratory distress syndrome. Mechanical ventilation (pressure support ventilation and HFO as well) was performed until the 53th day. A successful extubation was achieved after a low dose (1 mg/kg/day) hydrocortisone therapy over 13 days. The following respiratory support (nasal CPAP and high-flow nasal cannula) was necessary until 40^{+4} weeks. For prevention of bronchopulmonary dysplasia, we used an inhalative steroid therapy with fluticasone.

Cardiovascular system

An arterial hypotension in the first days was managed with domapine, dobutamine and noradrenalin. Due to signs of pulmonary hypertension, NO was used over 7 days. A treatment with paracetamol was necessary due to lack of spontaneous closure of ductus arteriosus. Severe RBC transfusions were performed due to anemia of prematurity.

Intestinal system

Enteral nutrition was started on the 7th day. No complications were observed, so that parenteral nutrition was stopped after 46 days. In addition, the infant received a therapy with ursodeoxycholic acid because of a direct hyperbilirubinemia.

Infections

Because of the preterm premature rupture of membranes, we initially administered an antibiotic prophylaxis with ampicillin, gentamicin and cefotaxime. Due to a nosocomial infection with *Staphylococcus haemolyticus*, a therapy with piperacillin/tazobactam and vancomycin was applied. A systemic infection with *Candida parapsilosis* at the 13th day of life was successfully treated with fluconazole and micafungin over 4 weeks.

Other findings

A laser photocoagulation was performed due to retinopathy of prematurity.

Discharge

The infant was discharged home at the corrected age of 22 days. A neurological examination as well as the brain ultrasound and MRI findings were normal at the point of discharge.

CONCLUSION

Rates of survival for infants born at the border of viability are still low and vary considerably among NICUs. Almost 1 in 4 extremely preterm infants dies during the birth hospitalization. Among those who survive, respiratory and other morbidities as well as long-term neurodevelopmental impairment are a large concern for patients, clinicians, and families. Active follow-up information is required to determine childhood outcomes.

REFERENCES

- Chawla D. Survival of Extremely-Low-Birth-Weight Neonates in India. Indian J Pediatr. 2023;90(3):217-8.
- Mehler K, Oberthuer A, Keller T, Becker I, Valter M, Roth B, Kribs A. Survival Among Infants Born at 22 or 23 Weeks' Gestation Following Active Prenatal and Postnatal Care. JAMA Pediatr. 2016;170(7):671-7.
- Patel RM. Short- and Long-Term Outcomes for Extremely Preterm Infants. Am J Perinatol. 2016;33(3):318-28.

ABS 13

DIAGNOSTIC-THERAPEUTIC PATHWAYS OF NTDs

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INTRODUCTION

Neural tube defects (NTDs) are the second most common congenital malformations in humans that affect the development of the central nervous system. Classically, NTDs are divided into two main subgroups (open defects, such as craniorachischisis, exencephaly-anencephaly and myelomeningoceles, and closed defects, including encephalocele, meningocele and spina bifida occulta [also known as occult spinal dysraphism – OSD]). The two most common NTDs are spina bifida and anencephaly. Spina bifida is a birth defect in which the spinal cord does not develop properly due to incomplete closure of the neural tube [1]. There is not a common consensus on NTDs pathogenesis; many risk factors (genetic and environmental) seem to be involved [2].

CLINICAL MANIFESTATION AND MANAGEMENT

The clinical spectrum of NTDs is broad, ranging from skin anomalies to motor, urinary, or bowel

dysfunctions. Notably, however, symptoms related to OSD are often not clinically evident at birth [3]. If newborns or infants have obvious clinical signs or atypical dimples, it is recommended to have a careful periodic clinical evaluation and possibly MRI to identify the pathology and allow for adequate management with a preventive intervention to avoid irreversible neurological deterioration [3]. Some of NTDs are evident from the gestational period. Myelomeningocele diagnosis can be prenatal and is based on the use of ultrasound from the end of the first trimester of pregnancy onwards. A detailed systematic examination of the sagittal, axial and coronal planes is necessary, along the entire length of the spine, from the cervical to the sacral tract. Instead, covered lesions such as OSD are found in the uterus rarely [4].

The management of patients with spina bifida is divided into several levels: neurological, urological, musculoskeletal, endocrinological, dermatological and rehabilitation [1]. As the life expectancy of these patients has increased, guidelines are required to protect normal kidney function, develop strategies for urinary continence, and promote independence during adulthood [5]. People with spina bifida are at risk of progressive kidney damage secondary to a hostile neurogenic bladder. Improper management can result in deterioration of the upper urinary tract, hydronephrosis, recurrent pyelonephritis and kidney scarring. Some patients may progress to the end stage of kidney disease, requiring dialysis or kidney transplant [5].

OBJECTIVES AND TREATMENT

The objectives to be achieved in the pediatric area are the maintenance of appropriate intravescical pressure, the preservation of the renal function and urinary continence. Clean intermittent bladder catheterization and antimuscarinic therapy can be used to achieve these goals for most children, but surgery is sometimes required. Patient monitoring requires periodic kidney and bladder ultrasound, urodynamic testing and serum creatinine with cystatin C to supervise hydronephrosis and avoid bladder wall thickening [1].

CONCLUSION

The children's psyche and quality of life are negatively impacted by NTDs and their treatment; if undiagnosed, they can lead to severe and irreversible complications. Improving quality of life and avoiding irreversible complications require early diagnosis and treatment. Further research is required in all aspects of urologic management to achieve these objectives.

REFERENCES

[1] Phillips LA, Burton JM, Evans SH. Spina Bifida Management. Curr Probl Pediatr Adolesc Health Care. 2017;47(7):173-7.

[2] Avagliano L, Massa V, George TM, Qureshy S, Bulfamante GP, Finnell RH. Overview on neural tube defects: From development to physical characteristics. Birth Defects Res. 2019;111(19):1455-67.

[3] Choi SJ, Yoon HM, Hwang JS, Suh CH, Jung AY, Cho YA, Lee JS. Incidence of Occult Spinal Dysraphism Among Infants With Cutaneous Stigmata and Proportion Managed With Neurosurgery: A Systematic Review and Meta-analysis. JAMA Netw Open. 2020;3(7):e207221.

[4] Copp AJ, Adzick NS, Chitty LS, Fletcher JM, Holmbeck GN, Shaw GM. Spina bifida. Nat Rev Dis Primers. 2015;1:15007.

[5] Joseph DB, Baum MA, Tanaka ST, Frimberger DC, Misseri R, Khavari R, Baillie S, Yerkes EB, Wood H. Urologic guidelines for the care and management of people with spina bifida. J Pediatr Rehabil Med. 2020;13(4):479-89.

ABS 14

IDIOPATHIC SCOLIOSIS: HOW ARTIFICIAL INTELLIGENCE CAN ASSIST CURRENT AND FUTURE CLINICAL PRACTICE

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Adolescent idiopathic scoliosis is a structural deformity of the spine with an uncertain etiology (it is hypothesized to have a multifactorial origin). It is a progressive condition that tends to worsen during adolescence but stabilizes when the vertebral epiphyseal cartilage fuses. It typically occurs between the ages of 10 and 18, with a prevalence of 2-3%. The deformity is manifested in three planes: frontal, axial and sagittal. Prognosis is influenced by the patient's age, sex, puberty, curve level, and degree of bone maturation. Treatment options include observation and scoliosis-specific physiotherapy exercises, use of braces and surgical correction of the deformity [1]. Artificial intelligence (AI) is progressively becoming more prevalent in the fields of radiology and healthcare, as radiology AI matures and seamlessly integrates into the daily radiology workflow. Over the past few years, the radiology community has witnessed a rapid surge in the potential of AI to revolutionize various aspects of radiology, ranging from the interpretation of medical images to informed clinical and operational decision-making [2]. This remarkable progress has

been made possible by AI's capacity to effectively model complex multivariate data through precise and robust machine learning techniques. Notably, within the realm of radiology, the most promising advancements have manifested in streamlining radiologic imaging processes and enhancing imaging devices, thereby optimizing image acquisition and reconstruction [3]. In the analysis of radiographs, the AI system exhibited sensitivity and a negative predictive value (NPV) equal to or greater than spine surgeons. In the analysis of back photos of patients in suspicion of scoliosis, the AI system showed higher sensitivity and NPV than surgeons. These results advocate for the immediate integration of AI into clinical practice to assist surgeons and, in the future, reduce radiation exposure for patients [2].

REFERENCES

[1] Negrini S, Donzelli S, Aulisa AG, Czaprowski D, Schreiber S, de Mauroy JC, Diers H, Grivas TB, Knott P, Kotwicki T, Lebel A, Marti C, Maruyama T, O'Brien J, Price N, Parent E, Rigo M, Romano M, Stikeleather L, Wynne J, Zaina F. 2016 SOSORT guidelines: orthopaedic and rehabilitation treatment of idiopathic scoliosis during growth. Scoliosis Spinal Disord. 2018;13:3.

[2] Zhang T, Zhu C, Zhao Y, Zhao M, Wang Z, Song R, Meng N, Sial A, Diwan A, Liu J, Cheung JPY. Deep Learning Model to Classify and Monitor Idiopathic Scoliosis in Adolescents Using a Single Smartphone Photograph. JAMA Netw Open. 2023;6(8):e2330617.

[3] Pianykh OS, Langs G, Dewey M, Enzmann DR, Herold CJ, Schoenberg SO, Brink JA. Continuous Learning AI in Radiology: Implementation Principles and Early Applications. Radiology. 2020;297(1):6-14.

ABS 15

CLINICAL-PATHOLOGICAL CORRELATIONS BETWEEN ABNORMAL PLACENTAL VASCULAR DEVELOPMENT AND UNFAVORABLE NEONATAL OUTCOME: A MULTIFACTORIAL STUDY

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INTRODUCTION

The placenta is the organ that nourishes, protects and supports the fetus during gestation. Its vascular development is crucial, as various alterations can lead to both fetal and maternal dysfunction. Histopathological diagnosis of major pathological vascular abnormalities (chorangiosis [CR], chorangioma [CM], chorangiomatosis [CMT]) during the placental development and the related hypervascularization can have a significant impact on the neonatal outcome. The early identification of at-risk pregnancies may reduce the possible related complications [1-3].

Based on our series of placentas analyzed between 2020 and 2023, we investigated possible clinical-pathological correlations and related neonatal outcomes. In particular, we correlated the placental pathologies with the maternal (hypertension gestational diabetes, pre-eclampsia, placental abruption, flow alterations, fever during pregnancy, mode of delivery) and the newborn (weight, Apgar score, admission to the Neonatal Intensive Care Unit [NICU], IUGR) health conditions.

METHODS

Our study included 41 over 28-week-old placentas carrying the following histopathological findings: CR, CM and CMT. In the multifactorial analysis we considered: gestational age, maternal age, comorbidities, type of lesions (subdividing CR and CMT into focal and diffuse), clinical diagnosis, possible hospitalization in the NICU of the newborn, and Apgar score at birth.

RESULTS

Our data showed a prevalence of focal (63%) followed by diffuse (13%) CMT, CR, and CM (8%), which were more prevalent in women between 35 and 45 years old. All our cases were characterized by placental hypervascularization. 54% of the newborn were delivered at or near term (> 37 weeks). 39/41 of fetuses were delivered alive. After delivery, 30% of the newborns were admitted to the NICU. In newborns admitted to the NICU, focal CMT were clinically diagnosed before birth in 72% of cases; CR-related cases (focal, diffuse, multifocal) were clinically diagnosed in 26% of cases; CM were clinically diagnosed in 2% of cases.

CONCLUSIONS

From the histopathological point of view, while all displaying placental hypervascularization, CMT, CM, and CR are identified by more detailed pathological findings: CMT by coagulopathy with thrombosis of the funicular veins, CM by necrosis, microcalcifications, blood extravasations, intervillous and perivillous fibrin, and CR by blood extravasations, areas of hypotrophy, late maturation of the villi, Wharton's jelly edema, and thrombosis. Our multifactorial analysis correlated the pathological findings of placental hypervascularization with an unfavorable neonatal outcome. Thus, the histopathological investigation is paramount for the appropriate clinical-diagnostic framing of the neonatal pathology, also for the planning of possible future pregnancies.

REFERENCES

[1] Stanek J. Chorangiosis of Chorionic Villi: What Does It Really Mean? Arch Pathol Lab Med. 2016;140(6):588-93.

[2] Amer HZ, Heller DS. Chorangioma and related vascular lesions of the placenta – a review. Fetal Pediatr Pathol. 2010;29(4):199-206.

[3] Carlucci S, Stabile G, Catagini S, Borghi C, Scutiero G, Morano D, Greco P. Fetal disseminated intravascular coagulopathy, hydrops and massive umbilical vein thrombosis consequence of a rare placental condition: multifocal chorangiomatosis. J Matern Fetal Neonatal Med. 2022;35(20):4009-13.

ABS 16

GASTROINTESTINAL DISORDERS IN ATRX SYNDROME: HOW TO DEAL WITH THEM?

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INTRODUCTION

Alpha-thalassemia mental retardation syndrome (ATRX) is an X-linked genetic disease caused by a mutation in the *ATRX* gene. The ATRX protein acts on chromatin remodeling by regulating gene expression. The syndrome is manifested by delayed psychomotor development, typical facial dysmorphism, genital abnormalities, alpha thalassemia and convulsions. In addition, gastrointestinal disorders, sometimes of such an entity to require surgery, can complicate the clinical picture [1]. Cases of gastric pseudo-volvulus, intermittent volvulus of the small or large intestine and ultrashort Hirschsprung's disease are reported in the literature. Other patients present feeding difficulties, regurgitation, vomiting, constipation, pain, and marked abdominal distension [2]. In non-surgical cases, there are other therapeutic solutions that we point out.

CASE REPORT

M. is a 10-year-old male affected by ATRX syndrome. He was born at term to non-consanguineous parents. The child, who has come to our attention, manifested important gastrointestinal disorders, such as gastroesophageal reflux, dysphagia, and chronic constipation with extreme family discomfort. Since January 2023, the symptoms had become so disabling to require 3 hospitalizations. Upper intestinal transit X-ray and abdominal X-ray showed a tendency to gastroesophageal stagnation, slowed gastric emptying time, and marked distension of intestinal loops. In addition, barium enema examination showed a substenotic area at the rectum-sigmoid level, compatible with Hirschsprung's disease. In April 2023, despite macrogol and regular evacuative enemas, due to the persistence and intensity of the abdominal pains, he was admitted to our clinic, and after cardiological evaluation, he started therapy with pyridostigmine, initially administered at a dosage of 30 mg/day, subsequently increased to 60 mg/day, continued on discharge. Since the beginning of the therapy there has been an improvement in the bowel habit and an abdominal pain reduction, with a marked reduction of irritability and consequent well-being.

CONCLUSIONS

There are various therapeutic strategies for patients with rare diseases and gastrointestinal disorders, such as chronic constipation (laxatives, evacuative enemas), but pyridostigmine, by increasing the levels of acetylcholine in the neuromuscular junction, helps intestinal peristalsis. In the literature there are few studies on the role of pyridostigmine in gastrointestinal disorders in children. These studies show a reduction of abdominal distension and an increase of intestinal peristalsis [3]. Through this case, we want to focus attention on the efficacy of pyridostigmine also in patients with ATRX syndrome, in which gastrointestinal complications often significantly affect the quality of life of young patients and their families.

REFERENCES

[1] Martucciello G, Lombardi L, Savasta S, Gibbons RJ. Gastrointestinal phenotype of ATR-X syndrome. Am J Med Genet A. 2006;140(11):1172-6.

[2] Horesh N, Pery R, Amiel I, Shwaartz C, Speter C, Guranda L, Gutman M, Hoffman A. Volvulus and bowel obstruction in ATR-X syndrome – clinical report and review of literature. Am J Med Genet A. 2015;167A(11): 2777-9.

[3] Manini ML, Camilleri M, Grothe R, Di Lorenzo C. Application of Pyridostigmine in Pediatric Gastrointestinal Motility Disorders: A Case Series. Paediatr Drugs. 2018;20(2):173-80.

ABS 17

ACCESSES TO PEDIATRIC EMERGENCY UNIT CAUSED BY DOMESTIC ACCIDENT: A RETROSPECTIVE STUDY

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The aim of this study is to analyze accesses to the Paediatric Emergency Unit of "AOOR Villa Sofia V. Cervello" Hospital (Palermo, Italy) caused by domestic accidents: from 1/1/2017 to 30/6/2023, there were 127,670 accesses to the emergency area, 33,384 (26.1%) for traumatic conditions due to accidents, and the majority of these accidents happened inside the home, 12,859 accesses. Before the COVID-19 pandemic, the total number of accesses to the Emergency Room was around 30,000 per year; accesses due to domestic accidents were around 9%. During the pandemic, general accesses decreased by 68.5% (9,137 accesses in 2020; 8,070 accesses in 2021), but domestic accidents increased in percentage up to 14%. This is explained by effects of the lock down: fewer social contacts, less contagion of seasonal pathologies, more presence of children at home. In 2022 and the first 6 months of 2023, there was a growth in general accesses, with fewer green codes and a trend towards a further increase in yellow codes. In the circumstance of a domestic accident, green codes are always more than 77%, yellow codes are 17-22%, red codes are 1-2%. Approximately 70% of the consultations, requested by paediatricians who take care of children in these conditions, are of an orthopedic nature. The majority of parents who take their children to hospital in a green or yellow code condition caused by domestic accident arrive with their own car; under code red conditions, the request for an ambulance is prevalent only in road accidents, school accidents and sports accidents, but in domestic traumas condition the majority of parents don't call the ambulance but go to the hospital directly with their own car. Analyzing data of red code accesses due to domestic accidents, in 29% of these accesses prolonged hospitalization was necessary and 45% were head injuries. In 20% of these red code accesses, it is noted the refusal of short intensive observation phase inside the Emergency Room by the parents; this is significantly higher in this specific population, compared to the general: it has had a clear increase during 2023 and mainly refers to minor head traumas. These data highlight that, after the pandemic, Paediatric Emergency Room use has changed: green codes decreased, while yellow and red codes are returning to pre-pandemic levels; people influx is lower in the morning and increases towards the evening, just like pre pandemic. Parents tend to avoid staying in the Paediatric Emergency Room during the observation phase if they have been reassured that their child condition is not serious; this can be a risk if the child is not completely stable and the parents overestimate their coping skills. Another risk factor is using their own cars in yellow or red code conditions, as it is thought that waiting for the ambulance could be a risk, but it is often more risky to move the child independently and take him to hospital by car. A psychoeducational process is important to accompany the parents to think about their parental competencies and prevent future risks.

ABS 18

HYALINE MEMBRANE DISEASE. HALLMARK OF NEONATAL RESPIRATORY DISTRESS SYNDROME

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INTRODUCTION

Hyaline membrane disease (HMD) is an acute lung condition that affects premature infants. The cause is insufficient levels of alveolar surfactant, which primarily occurs in infants who are born before 32 weeks of gestation and weigh less than 1,200 grams. The more premature the baby is, the greater the risk and severity of HMD. Type II pneumocytes produce alveolar surfactant, which is a complex lipoprotein composed of 6 phospholipids and 4 apoproteins. Ultrastructurally, it exists in three distinct forms within the lung: intracellular and extracellular lamellar bodies, extracellular tubular myelin, and a surface monolayer. Lamellar bodies are expelled from type II pneumocytes, where they are produced, and then transformed into tubular myelin.

HMD is the historical term for neonatal respiratory distress syndrome (RDS). The name is after the eosinophilic membrane that lines the distal airspaces, typically the terminal bronchioles or alveolar ducts, as observed in autopsies of neonates with RDS. Macroscopically, the lungs of these infants appear similar to the liver, with a reddish appearance. The hyaline membrane is composed of fibrin, cellular debris from lung epithelium, red blood cells, and leukocytes. Histological examination reveals pulmonary tissue with few dilated alveoli and diffuse areas of atelectasis.

The formation of hyaline membranes is driven by the collapse of alveoli that are not adequately coated with surfactant, reducing the pulmonary surface area. Gas exchange occurs only through the walls of

alveolar ducts and terminal bronchioles, which are unsuitable for this purpose. That leads to hypoxia and hypercapnia. Subsequently, vascular disruption occurs, resulting in the leakage of plasma into the alveolar spaces and the deposition of fibrin and necrotic cells along the surfaces of alveolar ducts and respiratory bronchioles, leading to the formation of hyaline membranes [1-3].

CASE REPORT

We present a case of a premature male newborn born at 25 weeks of gestation, who was affected by neonatal RDS. A chest radiography showed diffuse bilateral lung parenchymal hypodensity. The infant passed away in the Neonatal Intensive Care Unit, with a PO_2 value of 21.4 mmHg and a pCO_2 value of 123 mmHg. During the post-mortem examination, the infant weighed 803 grams and appeared normally shaped upon external and internal organ examination. His biometric measurements corresponded to 25 weeks of gestational age. Microscopic examination revealed lungs in the late canalicular phase, with a wide-spreading presence of dense eosinophilic material deposits along the alveolar walls (hyaline membranes). High-grade prematurity with hyaline membrane pneumopathy was the cause of death.

CONCLUSION

In premature infants, the lungs may have an immature surfactant production, leading to the formation of hyaline membranes. HMD is an acute lung condition that should be quickly diagnosed to reduce the often fatal complications.

REFERENCES

[1] Locci G, Fanos V, Gerosa C, Faa G. Hyaline membrane disease (HMD): the role of the perinatal pathologist. J Pediatr Neonat Individual Med. 2014;3(2):e030255.

[2] deMello DE, Chi EY, Doo E, Lagunoff D. Absence of tubular myelin in lungs of infants dying with hyaline membrane disease. Am J Pathol. 1987;127(1):131-9.

[3] Yadav S, Lee B, Kamity R. Neonatal Respiratory Distress Syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing, 2023.

ABS 19

IMPACT OF TWINNING ON SHORT-TERM CLINICAL OUTCOMES OF PRETERM INFANTS: A PROTOCOL STUDY

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INTRODUCTION

In the last decades, the frequency of twin pregnancies has increased steadily in all developed countries. Currently, the data present in the literature highlight how twinning is a risk factor for preterm birth, with an increase in mortality and cumulative morbidity. On the other hand, there are no studies that evaluate how much this increased risk is a direct effect of twinning, rather than the low gestational age (GA) at birth.

AIMS

The aim of this protocol study is to evaluate, in a population of preterm infants (< 37 weeks of GA), the association between twinning and two short-term outcomes with high clinical relevance: anaemia and cranial ultrasound alterations associated with unfavorable long-term outcome.

MATERIAL AND METHODS

An observational longitudinal study with retrospective data collection was planned. To have a sufficient number of cases and, at the same time, to assume that the treatment conditions are similar enough, it was decided to collect the data of all preterm newborns of the last 2 years (2021-2022), born in the Neonatology Unit, University of Turin, Italy. Neonates with major malformations, genetic/metabolic disorders, neonatal hemolytic disease were excluded. All maternal/neonatal variables that could be associated with outcomes or are useful for descriptive purposes, were collected. Odds ratio and 95% confidence interval were calculated using a conditional logistic regression model and a multivariate model by the creation of a directed acyclic graph (www.dagitty.net).

CONCLUSION

Our first goal is to evaluate the trend of the hemochromocytometric parameters associated with anemia in our population, putting them in relation to the reference values in the literature. Subsequently, it will be assessed if twinning affects these values in terms of z-score, time of onset of any pathological change and possible duration of therapy. In premature infants, the problem of when, how, and to whom to perform the cranial ultrasound develops. In order to answer these questions for the premature twins, we decided to consider all the ultrasound abnormalities that can affect neurodevelopment. In conclusion, our study could be the first step to highlight the possible link between twinning and these two clinical outcomes.

ABS 20

PHENOTYPICAL EXPRESSION OF *AHDC1 DE NOVO* TRUNCANT MUTATIONS: A RARE DISEASE

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A 2-month-old infant arrives at our clinic, the second child of a normal pregnancy with a negative neonatal anamnesis. The mother, with the experience of her 3 years older brother and the cousin of the same age, reports to the pediatrician that the child has an absent attitude and is not very sensitive to environmental stimuli. The growth in height and weight is normal and the child does not show particular signs of neurological deficit for his age. The pediatrician sends him to a check-up at the Child Neuropsychiatry Unit of the Regional Reference Hospital, where nothing obvious is detected. In subsequent pediatric clinical observations during the first years of life, non-specific dysmorphisms appeared, as well as anomalies of psychomotor development and learning difficulties in walking, in language and in relationships, and a serious and progressive neuropsychic retard. Therefore, visits to the Child Neuropsychiatry Unit of the Regional Reference Hospital continued. Metabolic investigations, transfontanellar ultrasound, cardiological visit and echocardiogram, electroretinography and visual evoked potentials were progressively performed, all in the norm. The video electro encephalogram gave as a result "notes of suffering prevailing in the posterior bihemispheric site, in the context of dysregulation of cerebral electrical activity in wakefulness and sleep". Magnetic resonance imaging (MRI) of the brain showed reduced white matter thickness of the semioval centers and thinning of the corpus callosum; myelination improved (MRI diagnosis) in subsequent years. The ophthalmological exam diagnosed hypermetropic astigmatism and papillary pallor. The sweat test was negative. A subsequent cardiological visit revealed moderate ascending aortic ectasia with a tricuspid aortic valve. After 1 year of age, growth slowed down with short stature evident, arginine stimulation tests were negative. After 2 years of follow-ups, towards the age of 3, the FT4 value was one-third lower than normal, and therapy with levothyroxine was started. Genetic tests were performed: array CGH, sialotransferrins, search for deletions and analysis of the region 15-q11-q13 methylation pattern (Prader Willi syndrome), methylation of exons 3 and 7 of the *TPRS1* gene (tricho-rhinophalangeal syndrome), all negative. With the polysonography performed due to the continuous recurrent diseases of the upper respiratory tract, a slight hearing loss on the right and obstructive sleep apnea syndrome (OSAS) were diagnosed, with subsequent surgery to remove the tonsils and adenoids. The diagnosis of Say Barber Biesecker syndrome (congenital hypothyroidism, facial dysmorphism, microcephaly, blepharophimosis, bulbous nose, thin lips, low-set ears and micrognathia, postaxial polydactyly and severe mental retardation) is proposed as a variant of Ohdo-Sonoda syndrome (congenital syndrome with multiple malformations), both unconfirmed. Genetic testing for short stature revealed the c.4285C>T p.(Arg429Cys) variant in the *PCNT* gene. The variant has never been described in the literature (replacement of arginine with cysteine). In the following 5 years, a slight psychomotor and neurological improvement was observed (also in cerebral myelination, subsequent MRI), and the child was placed in the community, continuing all the medical and physical support therapies. At the age of 6, the patient was included in the research project of a Foundation aimed at the diagnosis and treatment of rare diseases. The truncating mutation of the *de novo AHDC1* gene is found. This explains the clinical picture of the child, who falls under Xia Gibbs syndrome 3 [1]. Worldwide, there are 250 known cases of this disease, first described in 2014. The clinical course cannot be predicted. The mutations lead to a picture of syndromic intellectual disability characterized by phenotypic variability [2]. There are some common manifestations: hypotonia, delay in the acquisition of motor milestones, intellectual disability, language delay (but present with different severity) [3]. The clinical case highlights the need, in the case of syndromes with severe intellectual disability, to use broad-spectrum diagnostic techniques, such as whole exome sequencing. There is no therapy that improves the prognosis, except a social network of assistance that can improve the quality of life of the child and of the family.

REFERENCES

[1] Xia F, Bainbridge MN, Tan TY, Wangler MF, Scheuerle AE, Zackai EH, Harr MH, Sutton VR, Nalam RL, Zhu W, Nash M, Ryan MM, Yaplito-Lee J, Hunter JV, Deardorff MA, Penney SJ, Beaudet AL, Plon SE, Boerwinkle EA, Lupski JR, Eng CM, Muzny DM, Yang Y, Gibbs RA. De novo truncating mutations in AHDC1 in individuals with syndromic expressive language delay, hypotonia, and sleep apnea. Am J Hum Genet. 2014;94(5): 784-9.

[2] Jiang Y, Wangler MF, McGuire AL, Lupski JR, Posey JE, Khayat MM, Murdock DR, Sanchez-Pulido L, Ponting CP, Xia F, Hunter JV, Meng Q, Murugan M, Gibbs RA. The phenotypic spectrum of Xia-Gibbs syndrome. Am J Med Genet A. 2018;176(6):1315-26.

[3] Goyal C, Naqvi WM, Sahu A, Aujla AS. Xia-Gibbs Syndrome: A Review of Literature. Cureus. 2020;12(12):e12352.

ABS 21

IS THE IRRITABLE BOWEL SYNDROME A MANIFESTATION OF DISTRESS IN PARENTS OF PRETERM INFANTS? PRELIMINARY FINDINGS FROM A LONGITUDINAL FOLLOW-UP STUDY

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INTRODUCTION

Irritable bowel syndrome (IBS) represents one of the most common functional gastrointestinal disorders, ranging from 7% to 21% within the general population. It is a symptom-based condition defined by the presence of abdominal pain or discomfort, with altered bowel habits and lack of any other disease that can cause these symptoms [1]. IBS showed a high overlap with psychological symptoms. In particular, subjects suffering of IBS usually report higher rate of anxiety and depressive symptoms as compared to general population. In addition, a growing number of studies demonstrated that IBS patients frequently report past stressful life events, suggesting that life stressors and psychological issues may play a major pathophysiological role in IBS development [2]. The birth of an infant requiring an admission to the Neonatal Intensive Care Unit (NICU) is frequently associated with parental anxiety, stress and depression, both during NICU stay and after discharge [3]. This stressful experience can indeed strongly interfere with the normal transition into parenthood and lead to psychopathological outcomes in both mothers and fathers.

AIMS

This study aims to examine IBS symptoms prevalence and psychosocial variables associated with them during NICU hospitalization and 1 year after discharge.

MATERIALS AND METHODS

Parents of preterm infants admitted to the NICU were prospectively enrolled. IBS, parental stress, psychophysical well-being, psychopathological symptoms (i.e., anxiety and depression) were longitudinally assessed, during NICU hospitalization and 1 year after discharge, using standardized questionnaires.

RESULTS

Forty-four parents (mothers = 24, fathers = 20) participated to the study. Both mothers and fathers experienced high rates of IBS clinically significant symptoms during NICU hospitalization (mothers = 75%, fathers = 50%) and 1-year after discharge (mothers = 75%, fathers = 40%). Mothers reported higher scores as compared to fathers both during hospitalization ($p = 0.015$) and after discharge ($p = 0.013$). At the follow-up visit, paternal IBS symptoms were associated with being first-time father ($R^2 = 0.299$, $\beta = -0.547$, $t = -2.770$, $p < 0.05$), stress related to infant's look and behavior ($R^2 = 0.472$, $\beta = 0.687$, $p < 0.01$) and anxiety ($R^2 = 0.610$, $\beta = 0.781$, $t = 5.309$, $p < 0.01$). Maternal general health perception was associated with IBS score after discharge ($R^2 = 0.251$, $\beta = -0.501$, $t = -2.718$, $p < 0.05$).

CONCLUSIONS

Those findings highlight that parents of preterm infants experience high rates of IBS symptoms both during NICU hospitalization and 1 year after discharge. In particular, mothers showed higher scores than fathers at the 2 time points. Our study newly demonstrated a gender-related different influence of individual, situational and psychological health dimensions on IBS symptoms onset among mothers and fathers of preterm infants. The wide range of parental stress manifestations should be considered to improve Family-Centered Care interventions.

REFERENCES

[1] Schmulson MJ, Drossman DA. What Is New in Rome IV. J Neurogastroenterol Motil. 2017;23(2):151-63.

[2] Grover M, Kolla BP, Pamarthy R, Mansukhani MP, Breen-Lyles M, He JP, Merikangas KR. Psychological, physical, and sleep comorbidities and functional impairment in irritable bowel syndrome: Results from a national survey of U.S. adults. PLoS One. 2021;16(1): e0245323.

[3] Salomè S, Mansi G, Lambiase CV, Barone M, Piro V, Pesce M, Sarnelli G, Raimondi F, Capasso L. Impact of psychological distress and psychophysical wellbeing on posttraumatic symptoms in parents of preterm infants after NICU discharge. Ital J Pediatr. 2022;48(1):13.

ABS 22

EXPERIMENTAL PILOT PROJECT: "LET'S HELP EACH OTHER: INTEGRATED PSYCHOLOGIST-PEDIATRICIAN HEALTH INTERVENTIONS IN THE PEDIATRIC FIRST AID POST OF AUGUSTA". A SERVICE FOR FAMILIES

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The project is implemented in the pediatric first aid post (FAP) (in Italian: *punto di primo intervento*) of Augusta (Siracusa, Italy) through the co-presence work of the pediatrician and the pediatric psychologist operating according to the integrated work model [1]. It has as its general objective the valorization and humanization of primary care from a "Well-Being" perspective, understood as health promotion through integrated psychologist-pediatrician interventions, for children and caregivers who turn to pediatric FAP, as prescribed by the directives of the *Supplementary Regional Agreement on Pediatrics* (2011) in order to intervene with early diagnoses, provide health education, reduce clinical risk, enhance the management of chronicity through collaboration between local and hospital structures and between professional figures. Specific objectives are: reduction of the emotional and adaptive stress of the child and family members resulting from emergency situations; early detection of neglect situations; returning the role of support and help to parents; improving compliance between family members and healthcare professionals; improvement of chronicity management; increase and improvement of integration between the professional figures of the psychologist and the pediatrician. The methodology used is that of intervention-research and is based on: direct observation [2]; user data collection form; short psychological interventions (max 4 meetings); check-list for detecting intensity, frequency, duration of behavioral or phenomenal traits; human satisfaction evaluation questionnaires for users, psychologists and pediatricians [3]. The total sample is composed of 2,772 subjects belonging to the pediatric FAP, of which 1,591 subjects aged between 1 and 10 years visited by psychologists and pediatricians. Of these, it was possible to note in particular: frequency of access; reason for the consultation (67.5% flu symptoms; 11.3% allergies, gastro-intestinal disorders, ear pain; 21% psychological counseling). The project overturns the usual setting in which, driven by a need, the family turns to the professional to get an opinion from the expert psychologist who offers his availability for an observation that does not arise from an explicit and conscious need of the user and implies a final restitution. Advantages: taking charge of the subject and his family centered on listening and recognizing them as active subjects of care; early diagnosis of discomfort or developmental problems for patients assisted by the pediatrician and summoned by him; reduction of hospitalizations in the case of chronic diseases or rare diseases to improve therapeutic compliance, through parental support and containment of emotions; integration as a meeting of professionalism, exchange of knowledge and enrichment of languages. Critical issues: poor institutional recognition; training not always adequate for the integrated work model by some pediatricians.

REFERENCES

[1] Perricone G, Polizzi C, Morales MR. Un servizio di counseling per la trasformazione dei costrutti delle mamme dei bambini ospedalizzati. In: Polizzi C (Ed.). Pensarsi psicologo pediatrico. Modelli, percorsi e strategie di una formazione. Milan: Franco Angeli, 2011.

[2] Perricone Briulotta G. Agire l'osservazione. Modelli e percorsi. Milan: McGraw-Hill, 2003.

[3] Pedon A, Gnisci A. Metodologia della ricerca psicologica. Bologna: Il Mulino, 2005.

ABS 23

A SEVERE HYPERLACTACIDEMIA IN A DOLL FACE INFANT: DIAGNOSIS OF GLYCOGENOSIS 1B

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CASE REPORT

A 5-month-old female infant was referred to hospital for inconsolable crying and respiratory distress. The infant appeared pale, irritable, dehydrated,

hypotonic, with subcostal and intercostal retractions. Blood gas analysis revealed metabolic acidosis with severe lactic acidemia (pH 7.219, pCO_2 61.5 mmHg, HCO_3- 6.9 mmol/L, BE -22.3 mmol/L, lactate 26.19 mmol/L) and blood glucose of 37 mg/dL. IV fluid replacement and oral dextrose therapy were started. Her laboratory tests showed normal CBC, absent signs of infection, triglycerides 724 mg/dL, AST 272 IU/L, ALT 146 IU/L. Abdominal USD detected hepatomegaly. EEG, brain MRI, urinary organic acids, blood acylcarnitine and amino acids were normal. A strong suspicion of defective glycogenolysis and gluconeogenesis was raised, leading to request genetic test for glycogenosis, subtype 1 (GSD1). Meanwhile, a diet with lactose-free milk and maltodextrins was started. The patient was fed every 3 hours daytime and at night with enteral feeding through an NG tube.

MATERIALS AND METHODS

Peripheral blood samples of proband and her parents were sent for Trio whole exome sequencing (Trio-WES). DNA was fragmented, and the exons and their exon-intron boundaries were enriched using Roche KAPA capture technology, amplified, and sequenced on Illumina NextSeq™ 550. Sequencing was performed with 2 x 150 base paired-end sequencing reads. Secondary and tertiary analysis of genetic data was carried out through the GenomeUP (JuliaOmix™) platform. The pathogenicity of the variants identified was evaluated according to the American College of Medical Genetics and Genomics (ACMG) guidelines criteria.

RESULTS AND DISCUSSION

Trio-WES revealed a compound heterozygote on *SLC37A4* gene consistent with the phenotype: a nonsense variant on exon 6, c.652C>T(Q218X), was transmitted by the father and a splicing variant on intron 10, c.1124-2-1124-1del, was transmitted by the mother. Both variants are described in various databases as pathogenetic or probably pathogenetic, therefore confirming the diagnosis of glycogenosis, subtype 1b (GSD1b).

In GSD1b, the defective neo-gluconeogenesis is the main cause of severe lactic acidemia during fasting and hypoglycemic crisis [1]. The typical neutropenia was not present at diagnosis, but presented a month after hospital discharge. Lactic acidosis regressed in about 2 weeks. The patient will continue her own specific diet and will start vitamin integration, G-CSF injections and ACE inhibitors. Empagliflozin will be considered, for the shown positive effect on neutrophil dysfunction-related symptoms [2].

CONCLUSIONS

GSD1b should be suspected in infants showing hypoglycemia and metabolic acidosis with severe hyperlactacidemia. Dietetic therapy, restricted of galactose and fructose, must be started as soon as possible to prevent hypoglycemic events between feeds and to reduce the risk of life-threatening complications, cerebral damage and abnormal cognitive development, as well as preventing long-term storage of glycogen in the affected organs [3].

REFERENCES

[1] Parikh NS, Ahlawat R. Glycogen Storage Disease Type I. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing, 2023.

[2] Guerra F, Gasperini S, Bonanomi S, Crescitelli V, Da Dalt L, Norata GD, Balzarini M, Biondi A, Baragetti A, Saettini F. Empagliflozin Modifies Neutrophils' Subsets in Patients with Glycogen Storage Disease Type Ib By Increasing Mature Neutrophils and Decreasing Immature Neutrophils. Blood. 2022;140(1):2614-6.

[3] Kaczor M, Wesół-Kucharska D, Greczan M, Kierus K, Kałużny Ł, Duś-Żuchowska M, Ehmke Vel Emczyńska-Seliga E, Ciara E, Książyk J, Rokicki D. Clinical characteristics and long-term outcomes of patients with glycogen storage disease type 1b: a retrospective multi-center experience in Poland. Pediatr Endocrinol Diabetes Metab. 2022;28(3): 207-12.

ABS 24

LESS INVASIVE SURFACTANT ADMINISTRATION (LISA). RESULTS IN APPLYING THIS METHOD IN OUR NICU (BETWEEN APRIL 2019 AND DECEMBER 2022)

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BACKGROUND

Less invasive surfactant administration (LISA) is widely and increasingly used in Europe for the treatment of respiratory distress syndrome in preterm infants. There is a growing body of evidence that LISA-treated infants are at a decreased risk for bronchopulmonary dysplasia (BPD) compared to intubation and mechanical ventilation and other non-invasive strategies of respiratory support. LISA reduces duration of ventilation and risk of BPD, both well-defined risk factors of adverse development. Data on long-term outcome after LISA compared to intubation are scarce. The results of LISA in the Neonatal Intensive Care Unit (NICU) of Rea Maternity

Hospital (Athens, Greece) over the last 3 years are presented below.

METHODS

We compared the standard procedure of surfactant administration, that was applied via endotracheal intubation while the infant was ventilated, with a gentler approach called LISA. With LISA technique, surfactant was applied less invasively via a thin catheter while the infant was breathing spontaneously on CPAP.

We have administrated surfactant to 750 infants over the period April 2019-December 2022.

The infants were further classified in 3 categories according to their gestational age (GA):

- Category 1: mature infants, > 37 GA;
- Category 2: late preterm infants, 34^{+0}-36^{+6} GA;
- Category 3: preterm infants, < 34 GA.

We used Curosurf® at a dose of 200 mg/kg. No premedication was used.

RESULTS

350 (47%) of the infants received surfactant with LISA, while 400 (53%) via endotracheal intubation.

36 (5%) of the infants that received surfactant with LISA needed secondary intubation due to lack of respiratory improvement.

181 children of Category 1 (mature infants > 37 GA) received surfactant, 173 (96%) via intubation, while 8 (4%) via LISA.

294 children of Category 2 (late preterm infants) received surfactant, 173 (59%) via intubation, while 121 (41%) via LISA.

275 children of Category 3 (preterm infants) received surfactant, 54 (20%) via intubation, while 221 (80%) via LISA.

COMMENTS

Complications: pneumothorax in 4 infants after LISA; all of them were mature infants.

The majority of infants in Category 1 have received surfactant via intubation because nCPAP is not preferred as a method of RDS therapy in mature infants at our NICU, and that is why LISA was performed less in this group.

Among the 350 infants that received surfactant with LISA, intubation could be avoided in 314 of them, while 36 needed a secondary intubation.

CONCLUSION

Without doubt, LISA patients showed an immediate improvement of the oxygenation, a shorter duration of any other form of ventilatory support as a lower percentage of intubation and mechanical ventilation in the first 72 hours. Data on long-term outcome after LISA compared to intubation are rare, so that further follow-up is mandatory.

REFERENCES

- Kribs A, Wieg C, Roth B. Noninvasive Surfactant-Applikation (LISA) – Erkenntnisstand 2016. Neonatologie Scan. 2016;05(03):197-217.
- Mosca F, Colnaghi M, Fumagalli M. BPD: old and new problems. J Matern Fetal Neonatal Med. 2011;24:80-2.
- Teig N, Weitkämper A, Rothermel J, Bigge N, Lilienthal E, Rossler L, Hamelmann E. Observational Study on Less Invasive Surfactant Administration (LISA) in Preterm Infants < 29 Weeks – Short and Long-term Outcomes. Z Geburtshilfe Neonatol. 2015;219(6):266-73.

ABS 25

MONITORING OF URINARY TRACT COMPLICATIONS IN PATIENTS WITH SPINA BIFIDA

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Myelomeningocele, commonly known as spina bifida (SB), is the most common birth defect affecting the central nervous system, in which the spinal cord does not develop properly due to incomplete closure of the neural tube at 28 days of gestation. The overlying bones and skin are incompletely formed and the corresponding area is exposed to amniotic fluid in uterus. The diagnosis is usually obvious at birth because of the grossly visible lesion. It results in permanent disability. Patients can experience complications secondary to the neurologic abnormalities like urinary tract complications, learning disabilities, tethered cord and orthopedic problems.

Patients with neurogenic bladder may develop chronic renal disease (CRD) [1]. As the life expectancy of these patients has increased, a greater need for urological care guidelines has emerged to protect normal kidney function, develop strategies for urinary continence, and promote independence during adulthood [2].

If SB is not diagnosed before delivery, it is necessary to perform in the newborn an ultrasound of the kidneys and bladder and evaluate the function of the bladder with voiding cystourethrogram, cystometrogram and electromyogram (urodynamic studies).

Ultrasounds exams (US) detect the presence of hydronephrosis, hydroureter, abnormal bladder wall thickness, while urodynamic studies detect

excessive post-void residual volume. These findings suggest a risk for CRD.

Throughout life, patients with SB should be evaluated with US and serum markers for monitoring changes in urinary tract and guiding treatment decisions.

Serum creatinine is one of the biomarkers that are used to calculate the estimated glomerular filtration rate, but it is affected by multiple variables (age, sex, race). Consequently, another protein, cystatin C, is also assayed to provide more accurate data [2].

Blood exams should be performed in infancy and every few years thereafter. If there are urinary tract infections (UTIs) or significant hydronephrosis, US and blood exams should be done more often. Urinalysis should be performed only if there are symptoms of a UTI.

Renal scans (using DMSA or MAG3) should be performed in patients with abnormalities of the upper tract on the US, or those with unfavorable urodynamics, in order to assess renal scars in case of recurrent UTIs [3].

In conclusion, with good urologic management, preservation of kidney function is an attainable goal. It requires comprehensive and multidisciplinary management to improve quality of life and reduce the risk of renal complications.

REFERENCES

[1] Burke R, Liptak GS; Council on Children with Disabilities. Providing a primary care medical home for children and youth with spina bifida. Pediatrics. 2011;128(6):e1645-57.

[2] Joseph DB, Baum MA, Tanaka ST, Frimberger DC, Misseri R, Khavari R, Baillie S, Yerkes EB, Wood H. Urologic guidelines for the care and management of people with spina bifida. J Pediatr Rehabil Med. 2020;13(4):479-89.

[3] Shiroyanagi Y, Suzuki M, Matsuno D, Yamazaki Y. The significance of 99mtechnetium dimercapto-succinic acid renal scan in children with spina bifida during long-term followup. J Urol. 2009;181(5):2262-6; discussion 2266.

ABS 26

PEDIATRIC FUNCTIONAL NEUROLOGICAL DISORDER: THE COMMUNICATION OF DIAGNOSIS IS THE START OF THE TREATMENT

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BACKGROUND

Functional neurological disorder (FND) is a mental disorder characterized by neurologic symptoms (either motor or sensory) that suggest the presence of an underlying neurological condition, without organic disease. Common symptoms include weakness and/or paralysis, non-epileptic seizures, movement disorders, speech or visual impairment, swallowing difficulty, sensory disturbances or cognitive symptoms. Many clinicians use different names to describe the disorder's symptoms: "functional", "psychogenic" or conversion disorder, hysteria, dissociative disorders and health anxiety/hypochondriasis. FNDs are common in the pediatric population. DSM-5 diagnostic criteria are the following:

A. one or more symptoms of altered voluntary motor or sensory function;
B. clinical findings provide evidence of incompatibility between the symptom and recognized neurological or medical conditions;
C. the symptom or deficit is not better explained by another medical or mental disorder;
D. the symptom or deficit causes clinically significant distress or impairment in social, occupational, or other important areas of functioning or warrants medical evaluation.

Although the exact incidence is unknown, a 2021 *JAMA Neurology* study looking at the prevalence and costs of FNDs from 2008 to 2017 found that approximately 1% of Pediatric Emergency Department visits with neurologic symptoms were related to FNDs (3,800 out of 328,609 patients, US health care databases).

METHODS

In our health system at Rovigo Hospital (Rovigo, Italy), FND is typically diagnosed by a neurologist and neuropsychiatrist. A careful physical examination and medical history can be diagnostic, but in some cases, we can do further tests to confirm diagnosis (e.g. EEG, MRI) or look for other health conditions. Further psychological tests (SAFA, MMPI-A) complete the assessment.

The first most important therapeutic act is how to explain the diagnosis of FND. We proceed as the scientific evidence suggests:

- give a name to the condition: FND;
- emphasize that physical examination signs are the basis for diagnosis;
- underline that FND is real, common, brain-based and treatable in many patients;
- underline that the patient is not "crazy" nor "making up their symptoms";
- provide an explanatory model, such a framing that FND is a "software rather than a hardware problem";

- provide educational materials;
- focus on "what" the diagnosis is; speculations regarding "why" should generally be deferred to guided exploration in the course longitudinal treatment;
- leave time for the patient and their family members to ask questions.

CONCLUSION

In our clinical cases, we observed that the support of the patient and the family during hospitalization, combined with a careful and respectful explanation, have determined the rapid resolution of the symptoms. Psychological support continues to identify triggers and facilitate the mentalization process, essential in the developmental age.

REFERENCES

- Aybek S, Perez DL. Diagnosis and management of functional neurological disorder. BMJ. 2022;376:o64.
- Garcia A. Pediatric functional neurologic disorder. Pediatr Clin N Am. 2023;70(3):589-601.
- Stephen C, Fung V, Lungu CI, Espay AJ. Assessment of Emergency Department and inpatient use and costs in adult and pediatric functional neurological disorders. JAMA Neurol. 2021;78(1):88-101.

ABS 27

THE IMPORTANCE OF FINE-NEEDLE ASPIRATION CYTOLOGY IN THE PREOPERATIVE DIAGNOSIS OF PEDIATRIC MALIGNANT THYROID NODULES: A CASE SERIES

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INTRODUCTION

Thyroid nodules, quite common in the adult population, are rare in the pediatric population. The incidence of such nodules in the pediatric population ranges from 1% to 2%, with a predominance of benign nodules over malignant ones. However, the prevalence of malignant thyroid nodules in the pediatric population (especially in the case of papillary thyroid carcinoma) has increased in the last decades, reaching 22-26% compared to the 5% prevalence in the adult population. Usually, a pediatric thyroid nodule appears asymptomatic: it may manifest as a neck mass with or without lymphadenopathy, often in euthyroidism, with symptoms related solely to mass effect. Therefore, a timely histopathological diagnostic approach is crucial, which can be achieved primarily through fine-needle aspiration cytology (FNAC), a test with 94% sensitivity, 99% accuracy, and 100% specificity. According to The Bethesda System for Reporting Thyroid Cytopathology (TBSRTC), FNAC can identify the benign or malignant nature of a nodule through specific cytopathological diagnostic criteria, ensuring early diagnosis even in the pediatric population and surgical therapy to eradicate the neoplasm [1-3].

METHODS

Our study analyzes a series of 3 cases from 2021 to 2023 of children aged 11 to 13 years who, on ultrasound examination, presented with 3 nodules (all with hypoechoic components and all in a euthyroid state) ranging in size from 3.7 mm to 43.6 mm.

RESULTS

Following FNAC, our cytological diagnosis was Tir3B (Class IV according to TBSRTC) in 2/3 cases and Tir4 (Class V according to TBSRTC) in 1/3 cases. After performing surgical interventions on the children (1/3 thyroidectomy, 2/3 hemithyroidectomy), we confirmed the presence of malignant neoplasms in the aforementioned nodules. In 2/3 cases, corresponding to the cytological diagnosis of Tir3B (Class IV according to TBSRTC), our histological diagnosis identified, respectively, a differentiated follicular origin carcinoma with features of microinvasive encapsulated papillary carcinoma (pT2[s]Nx) and a microinvasive encapsulated follicular carcinoma (pT1b[s]Nx). In 1/3 cases, corresponding to the cytological diagnosis of Tir4 (Class V according to TBSRTC), our histological diagnosis identified a classic-type papillary carcinoma (pT1aNx).

CONCLUSIONS

Despite the rarity of pediatric thyroid tumors, the recent increase in their frequency and the often asymptomatic nature necessitate early diagnosis to ensure timely therapy. Our study highlights how FNAC is a first-line tool in the preoperative diagnosis of pediatric malignant thyroid nodules and how this method can support

the diagnosis and management of these pathological entities.

REFERENCES

[1] Wang H, Mehrad M, Ely KA, Liang J, Solórzano CC, Neblett WW 3rd, Coogan AC, Weiss VL. Incidence and malignancy rates of indeterminate pediatric thyroid nodules. Cancer Cytopathol. 2019;127(4):231-9.

[2] Lebbink CA, Links TP, Czarniecka A, Dias RP, Elisei R, Izatt L, Krude H, Lorenz K, Luster M, Newbold K, Piccardo A, Sobrinho-Simões M, Takano T, Paul van Trotsenburg AS, Verburg FA, van Santen HM. 2022 European Thyroid Association Guidelines for the management of pediatric thyroid nodules and differentiated thyroid carcinoma. Eur Thyroid J. 2022;11(6):e220146.

[3] Paulson VA, Rudzinski ER, Hawkins DS. Thyroid Cancer in the Pediatric Population. Genes (Basel). 2019;10(9):723.

ABS 28

UNDERSTANDING ELEMENTARY PLACENTAL LESIONS: DISTINGUISHING BETWEEN PATHOLOGICAL AND PHYSIOLOGICAL CHANGES IN THE CLINICAL PRACTICE

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It is becoming increasingly evident that the histomorphological study of the placenta can provide relevant and valuable information regarding the intrauterine life, especially in cases with adverse outcomes. The accurate analysis of placental changes may be very useful for the newborn as well as for the mother, both for herself (signs of placental infection) and for future pregnancies (pathological signs of placental rejection).

Not all placentas come under our observation, but only those identified through precise protocols that may change among different centers. Among the most important conditions in which placental histological examination is mandatory, there are pathological pregnancies (e.g., diabetes, preeclampsia), intrauterine growth restriction, placenta accreta, small or excessively large placentas, prolonged membrane rupture, umbilical cord knots or constrictions [1].

This selection is made in order to focus attention on pathological fetal and maternal conditions or those considered as such. With the consensus of Amsterdam in 2016, the following four main placental pathologies were defined: maternal vascular malperfusion, fetal vascular malperfusion, acute chorioamnionitis, villitis and intervillitis of unknown etiology, each characterized by specific patterns [2].

The pathologist's task is to proceed with careful and comprehensive sampling of all placental structures and to correlate the various histopathological changes observed with the clinical data. The final diagnosis should fall into one of the four categories described above. However, in daily practice, elementary lesions sometimes do not allow precise conclusions, leading to generic descriptions of morphological findings which, from a practical point of view, are devoid of any clinical significance. As a consequence, pathologists often face the dilemma of the clinical significance of the observed placental changes. This occurs more frequently in full-term placentas, but the doubts are not excluded, albeit more rarely, in preterm pregnancies.

The significance of increased perivillous fibrin, for example, of small infarcted areas, the finding of hypervascularization of terminal villi, the increase in syncytial knots as compared to the gestational age, all these changes, when taken individually, may not allow a correct diagnostic framework. Therefore, despite efforts to categorize elementary lesions into precise patterns that can provide useful information to clinicians, we still find ourselves questioning the significance of small alterations seemingly devoid of any clinical significance.

All these data taken together, the following questions arise spontaneously: do we need to return to the old hypothesis that the placenta is an organ that is born, grows, performs its functions, and then undergoes physiological aging [3]? Can we think that compensatory processes come into play when something goes awry during pregnancy, compromising its function?

In conclusion, we believe that the complex histopathological analysis of the placenta cannot disregard the dialogue between pathologists, gynecologists and neonatologists. An in-depth study of all clinical information, gathered during pregnancy and delivery, appears mandatory. Some elementary lesions (such as increased fibrin and hypervascularization of terminal villi, among the most common), which should be considered pathological in some contexts, should be classified as a normal physiological change in other conditions, according with the clinical data. Our proposal is to include in the group of major placental pathologies (maternal vascular malperfusion, fetal vascular malperfusion, acute chorioamnionitis, and villitis of unknown etiology) a new category, following the exclusion of all possible clinicopathological

correlations, which could be simplified as: "histomorphological alterations apparently devoid of any clinical significance, associated with physiological placental involution".

REFERENCES

[1] Fulcheri E, Grillo F, Musizzano Y. Il trattamento della placenta per l'esame istopatologico finalizzato allo studio ed alla diagnostica del danno neurologico feto-neonatale. Riv It Ost Gin. 2006;9:575-81.

[2] Khong TY, Mooney EE, Ariel I, Balmus NC, Boyd TK, Brundler MA, Derricott H, Evans MJ, Faye-Petersen OM, Gillan JE, Heazell AE, Heller DS, Jacques SM, Keating S, Kelehan P, Maes A, McKay EM, Morgan TK, Nikkels PG, Parks WT, Redline RW, Scheimberg I, Schoots MH, Sebire NJ, Timmer A, Turowski G, van der Voorn JP, van Lijnschoten I, Gordijn SJ. Sampling and Definitions of Placental Lesions: Amsterdam Placental Workshop Group Consensus Statement. Arch Pathol Lab Med. 2016;140(7):698-713.

[3] Macpherson TA, Szulman AE. The placenta and products of conception. In: Silverberg SG (Ed.). Principles and Practice of Surgical Pathology. New York: Churchill Livingstone, 1990.

ABS 29

USE OF SELUMETINIB IN PEDIATRIC PATIENTS WITH NF1 AND PLEXIFORM NEUROFIBROMAS

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INTRODUCTION

Neurofibromatosis type 1 (NF1) is a genetic disorder with an incidence of 1:3,000 live births [1]. The patients affected are more likely to develop malignant and benign tumors, including plexiform neurofibromas [2]. Selumetinib, acting on the Ras-Raf-MEK-ERK pathway, was recently approved by the European Medicines Agency (EMA) and the Italian Medicines Agency (*Agenzia Italiana del Farmaco* – AIFA) for its use in patients with NF1 and with symptomatic and inoperable plexiform neurofibromas starting from the third year of age [3].

AIMS OF THE STUDY

The aims of the study are: to help provide further data on the safety of this drug and to highlight the adverse effects; to study the response to treatment in terms of tumor volume, pain reduction and improvement of functional problems as emotional and psychological state of patients and their families; furthermore, to verify whether there is a correlation between the gene variant and response to treatment.

MATERIALS AND METHODS

We selected, among our 116 patients affected by NF1, 17 patients, who had one or more plexiform neurofibromas. Six of them were eligible to start treatment with selumetinib. At time 0, the patients underwent echocardiography, ophthalmological examinations and magnetic resonance imaging (MRI) of the site affected by the tumor. The administered dose of selumetinib was 25 mg/m^2, every 12 hours. The monitoring included: blood tests in the first 6 months, ophthalmologic evaluation every 3 months and in any case when symptoms appeared, cardiological examination every 3 months and MRI every 6 months for volumetric evaluation of the neurofibroma.

RESULTS

Selumetinib side effects were well tolerated, but psychological distress was reported in 2 patients. Acneiform eruption appeared in 50% of patients, while gastrointestinal effects were reported by 66.6% of them. Volumetric reduction of the plexiform neurofibroma was observed in 2 patients, 3 patients had no changes, while in 1 patient clinical improvement was reported.

CONCLUSION

Among the 6 cases, 2 of them had tumor shrinkage, and both had a frameshift mutation of *NF1* gene, suggesting a possible genotype-phenotype correlation. Selumetinib is a compassionate-use drug and treatment duration will be established by a multidisciplinary team based on the drug's adverse effects and MRI results. Further studies are necessary to demonstrate actual effectiveness on clinical improvement and volumetric reduction of plexiform neurofibromas and genotype-phenotype correlation for a possible positive response to selumetinib.

REFERENCES

[1] Tamura R. Current Understanding of Neurofibromatosis Type 1, 2, and Schwannomatosis. Int J Mol Sci. 2021;22(11):5850.

[2] Friedman JM. Neurofibromatosis 1. In: Adam MP, Mirzaa GM, Pagon RA (Eds.). GeneReviews®. Seattle (WA): University of Washington, Seattle, 1993-2023. Available at: https://www.ncbi.nlm.nih.gov/books/NBK1109/, date of publication: 2 October 1998, last access: September 2023.

[3] Mukhopadhyay S, Maitra A, Choudhury S. Selumetinib: the first ever approved drug for neurofibromatosis-1 related inoperable plexiform neurofibroma. Curr Med Res Opin. 2021;37(5):789-94.

ABS 30

A CASE STUDY OF THE EUROPEAN LIFE MILCH PROJECT: CORRELATION BETWEEN

MATERNAL NUTRITION, BREASTFEEDING AND GROWTH

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BACKGROUND

Breast milk is an individual-specific fluid, an essential source of nutritional factors and non-nutritive bioactive factors that promote the survival and development of the newborn. Characteristic of breast milk is its dynamism, which is expressed through qualitative changes resulting from factors of different nature, for example, the maternal genetic profile, the maternal diet, the physiology of the mammary gland, the state of maternal health, the type of birth, but also environmental factors and sampling-related factors [1].

OBJECTIVES

This observational study is based on the Life MILCH Project (Ref No. LIFE 18 ENV/000460), co-financed by the European Commission in 2019. The objectives of this study were: to study any correlations between the nutrition of mothers during pregnancy, the breastfeeding methods and body weight of their babies; to evaluate whether the food consumed by mothers, contained in plastic packaging or derivatives, could have an influence on the maternal body mass index (BMI) at the end of pregnancy and the weight of the child at birth; and finally, to compare the results obtained with the scientific data present in the literature.

METHODS

The study was carried out according to the Life MILCH Project protocol; 65 normal pregnant women participated, enrolled between the 36th and 41st week of gestation, whose anthropometric data were collected. The anthropometric data of their children were collected, as well, after birth up to the 12th month of life. To obtain data relating to maternal eating style, the food questionnaires filled out by each mother before giving birth were used, using the Mediterranean diet food pyramid as a reference model. The frequency of maternal exposure to plastics was obtained from the same questionnaires.

RESULTS

To study the correlation between maternal nutrition and the child's body weight, we considered the maternal diet at the end of pregnancy and studied the variation in the weight of their children (average weight) over time, considering the type of breastfeeding. The statistical analysis highlighted statistically significant differences between the two groups of children (exclusive breastfeeding and mixed breastfeeding). Regarding maternal exposure to plastic, our results are in line with those reported in the literature, despite the low significance, demonstrating a correlation between greater exposure to plastic, obesity (maternal BMI) and weight gain of the child.

CONCLUSIONS

Maternal nutrition can greatly influence the growth outcomes of the child, in particular its body weight. This depends on the qualitative and quantitative composition of the diet, but also on exposure to substances such as endocrine disruptors, which have the ability, through molecular and epigenetic mechanisms, to modify both the composition of breast milk and the endocrine structure of the child, in this case the lipid metabolism. From the results obtained, a horizon of possible future studies emerges.

REFERENCE

[1] Ballard O, Morrow AL. Human milk composition: nutrients and bioactive factors. Pediatr Clin North Am. 2013;60(1):49-74.

ABS 31

PEDIATRIC ONCOLOGICAL PAIN: THE INFLUENCE ON THE POSTURE OF THE HEAD AND THE SPINE

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Pediatric oncological pain (OP) is common in children with cancer, particularly those affected by osteosarcoma, a type of neoplasm that often targets the head and spine. This pain can lead to postural changes in these areas and negatively impacts the quality of life for affected children. Osteosarcoma frequently causes bone and muscle pain, setting off a potentially harmful cycle that may involve mood disorders and maladaptive thoughts. It is crucial to comprehensively assess children with OP, incorporating both clinical and psychological evaluations, to guide multidisciplinary rehabilitative strategies. A variety of healthcare professionals play essential roles in managing children with oncological pain. Experts directly addressing postural changes in the head and spine, such as orthodontists and physiatrists, collaborate with specialists in pediatrics, oncology, and psychology to provide comprehensive care for these vulnerable patients. Their combined efforts aim to mitigate the physical and mental impact of OP [1]. This review focuses on the comprehensive assessment of pediatric OP, particularly in terms of postural evaluation of the head and spine – a critical step in planning effective rehabilitative interventions. The assessment primarily involves evaluating motor functions, by encompassing various aspects such as joint mobility, muscle strength, muscle palpation, peripheral neurological examination, and other relevant clinical tests. While radiological examinations like ultrasonography, radiography, computed tomography, and magnetic resonance imaging may not be suitable for postural assessment, more advanced and non-invasive evaluations are currently recommended. It is important to acknowledge that mood disorders are prevalent among young individuals with OP stemming from head and spinal tumors. Literature highlights elevated levels of anxiety and depression in these cases compared to individuals without cancer-related pain [2]. Orthodontists and physiatrists commonly address the postural aspects of OP resulting from head and spine cancers. Orthodontists assess postural changes due to teeth and jaw irregularities and temporomandibular joint disorders, while physiatrists focus on functional alterations in the neck and spinal column [3]. In conclusion, thorough assessments based on robust clinical evaluations are essential. A collaborative multidisciplinary approach, involving orthodontists, physiatrists, psychologists, oncologists, and pediatricians, is recommended. Evidence-based multidisciplinary rehabilitation strategies, including education, exercise, and cognitive-behavioral therapy, play a significant role in the care of children with tumor-related OP.

REFERENCES

[1] Anghelescu DL, Tesney JM. Neuropathic Pain in Pediatric Oncology: A Clinical Decision Algorithm. Paediatr Drugs. 2019;21(2):59-70.

[2] Moreira R, Teles A, Fialho R, Baluz R, Santos TC, Goulart-Filho R, Rocha L, Silva FJ, Gupta N, Bastos VH, Teixeira S. Mobile Applications for Assessing Human Posture: A Systematic Literature Review. Electronics. 2020,9(8):1196.

[3] L'Hotta AJ, Beam IA, Thomas KM. Development of a comprehensive pediatric oncology rehabilitation program. Pediatr Blood Cancer. 2020,67(2): e28083.

ABS 32

USE OF GRIFFITHS III SCALES IN A COHORT OF PRETERM AND VLBW INFANTS: EVALUATION OF NEURODEVELOPMENTAL OUTCOMES

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INTRODUCTION

Preterm infants are at risk of atypical development, not only from a motor point of view, but also from a cognitive one. Although it is difficult to have sensitive and specific tests for this population in the first 2 years of life, early diagnosis is essential, because it allows habilitative interventions to start as soon as possible.

OBJECTIVES

Our study aimed to examine neurodevelopmental outcomes at 24 months of corrected age (CA) in a cohort of preterm infants, through the administration of the Griffiths III scale. Furthermore, the most compromised areas of neurodevelopment were identified through the analysis of the standardized obtained scores.

METHODS

A single-center retrospective study was conducted, focused on a population of preterm infants born at < 32 weeks of gestational age (GA) or < 1,500 g birth weight, who reached 24 months of CA in the recruitment period (01/09/2021-02/05/2023).

Within the cohort, we studied the possible relationship between neurological outcomes and various perinatal and neonatal variables, in order to evaluate which of these had the greatest influence on adverse outcomes.

RESULTS

Overall, the developmental quotients (DQ) obtained on the various subscales of the Griffiths III test were average compared to the general population, although those born between 23 and 25 weeks of GA obtained a language DQ lower than the average. The subscales that investigate cognitive development, in fact, obtained lower scores than those relating to the motor sphere. In particular, the areas of neurodevelopment that are most compromised concern learning, language and the personal-social-emotional sphere.

Adverse neurological outcomes at 24 months of CA were supported by severe neonatal distress, surgery for retinopathy of prematurity, bronchopulmonary dysplasia, early sepsis and chorioamnionitis, confirming data in the literature. Nonetheless, intraventricular hemorrhage (IVH) grades I and II were also found to be an important risk factor for major neurological outcomes. However, this topic is still the subject of current scientific debate.

CONCLUSIONS

The present study confirms that preterm birth is associated with a greater risk of adverse neurological outcome at 24 months of CA and this probability increases with decreasing GA. Among the neonatal disorders that most compromise neurodevelopment, IVH grades I and II stand out. However, in the past, it was not considered a particularly relevant factor in determining adverse neurological outcomes. This finding suggests that clinicians should not underestimate IVH, even if not large, during follow-up, in order to early diagnose any atypical development and offer timely support to the newborn.

Finally, it would be appropriate to investigate the results of this study with further research, conducted on a larger population of preterm infants, to confirm the results obtained.

ABS 33

TRAUMA IN PARENTS AND CHILDREN IN CASE OF HOSPITALIZATION OF THE NEWBORN IN THE NEONATAL INTENSIVE CARE UNIT

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This literature review aimed to verify the presence of studies regarding the psychological and emotional trauma of children and their parents in the event of hospitalization in Neonatal Intensive Care Units (NICU).

The bibliographic search on the PubMed search engine produced 5 results.

There has been a tendency to consider the child and parents separately rather than the family unit as affected by this adverse event, which is in fact considered an adverse childhood experience. In fact, only 1 article out of 5 deals with providing support to both parents and the child with concrete interventions aimed at building and safeguarding the relationship, because a healthy relationship is also considered a protective factor against the development of psychological problems in the future, both for parents and for the child. Furthermore, the child develops his relationship with the parents also through physical contact (kangaroo therapy and similar), which improves his cognitive abilities. Parents' involvement in the child's care helps create relationships and improve attachment [1].

In the past, it was believed that 60% of women whose newborns were admitted to NICU for any reason developed post-traumatic stress disorder (PTSD), a percentage comparable to that of PTSD after major catastrophes. Recently, however, the cause of hospitalization has been analyzed and it emerged that the cause of hospitalization could be decisive in the development of PTSD itself. It would seem that sudden events, such as asphyxia or an unexpected premature birth, trigger PTSD [2].

Fathers have only recently been taken into consideration in this scenario. Fathers and mothers experience their child's hospitalization differently, with greater stress on the part of the latter, but this does not mean that fathers are not equally at risk of developing psychological disorders [3]. Fathers should also be involved in this type of study and we should think about targeted interventions for them.

In general, the path should be that of involvement and support, both during hospitalization and in the first years of the child's life. In fact, it has been seen that follow-up visits, scheduled even just in the first year after discharge, lead to a reduction in stress on the parents' part.

REFERENCES

[1] Lakatos PP, Matic T, Carson M, Williams ME. Child-Parent Psychotherapy with Infants Hospitalized in the Neonatal Intensive Care Unit. J Clin Psychol Med Settings. 2019;26(4):584-96.

[2] Orovou E, Eskitzis P, Mrvoljak-Theodoropoulou I, Tzitiridou-Hatzopoulou M, Dagla M, Arampatzi C, Iliadou M, Antoniou E. The Relation between Neonatal Intensive Care Units and Postpartum Post-Traumatic

Stress Disorder after Cesarean Section. Healthcare (Basel). 2023;11(13): 1877.

[3] Matricardi S, Agostino R, Fedeli C, Montirosso R. Mothers are not fathers: differences between parents in the reduction of stress levels after a parental intervention in a NICU. Acta Paediatr. 2013;102(1):8-14.

ABS 34

AN ITCH… THAT GOES TO THE BONE

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INTRODUCTION

Osteomyelitis (OM) is an infectious process of bone tissue. In childhood, the most common form is acute hematogenous OM, which affects the long bones of the lower limbs and vertebrae. The etiology is predominantly bacterial and the most involved germ is *Staphylococcus aureus*, whose entrance door is often represented by skin lesions. Magnetic resonance imaging (MRI) represents the gold standard in the diagnosis of OM, being able to document bone edema, the nonspecific first sign of infection, already after 24-48 hours; however, its use is often limited to emergencies [1]. The onset of symptoms, especially in the vertebral location, is often inaccurately identified.

CASE REPORT

An 8-year-old male came to the Emergency Room for left abdominal and lumbar pain during the last 3 days and fever during the last 24 hours. No urinary symptoms, no previous traumas, with uneventful previous history. At physical examination: stable vital signs, fever, suffering clinical state, difficult walking, right side preferential position; abdominal palpation painful in the left side with ipsilateral lumbar irradiation without peritonism and negative Giordano's sign. Crusted lesions, scraping residue from insect bites, on the lower limbs. Laboratory blood test: WBC 10,000/mm^3 (N 88%), CRP 13 mg/dL, PCT 4.42 ng/mL, other lab tests normal. Abdomen X-ray and ultrasound were negative except for coprostasis. Dorsolumbar spine X-ray was negative. After blood culture, positive for multisensitive *Staphylococcus aureus*, we started i.v. ceftriaxone, replaced with piperacillin/tazobactam on the 3rd day due to hypertransaminasemia and bile sand at ultrasound control. Abdominal symptoms and lumbar pain resolved. Due to persistence of fever and difficulty in flexing the trunk due to stiffness, on the 4th day, to rule out bone localization, we performed technetium scintigraphy, which highlighted pathological hypercaptation on D9, and neurosurgical evaluation, which excluded neurosensory deficits and suggested immobilization. Given the microbiological data and the persistence of fever, we introduced vancomycin with fever reduction and resolution of the pain after 24 hours. On the 7th day, MRI confirmed vertebral OM D9-D10 (pathological inflammatory changes of spinous, transverse, pedicle, interapophyseal joints, posterior paravertebral soft tissues and epidural space). The patient was discharged on the 10th day of admission with an orthopedic bust and oral amoxicillin/clavulanic acid. Control MRI to be done after 6 weeks of therapy.

CONCLUSIONS

In our case, scintigraphy promptly clarified the clinical uncertainties relating to bone localization, allowing us to undertake targeted antibiotic treatment and immobilization pending MRI, which remains, however, the gold standard in OM to evaluate the extent of the inflammatory process and allow for differential diagnosis.

REFERENCE

[1] Lee YJ, Sadigh S, Mankad K, Kapse N, Rajeswaran G. The imaging of osteomyelitis. Quant Imaging Med Surg. 2016;6(2):184-98.

ABS 35

THE IMPORTANCE OF PLACENTAL ANALYSIS IN INTRAUTERINE GROWTH RETARDATION

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INTRODUCTION

The placenta is the interface for the supply of nutrients and oxygen between the maternal and fetal circulation. Placental weight and baby weight at birth are strongly correlated. In fact, the possibility of the fetus to grow *in utero* depends on placental function.

Intrauterine growth retardation (IUGR) is defined as the failure to reach the fetus's growth potential, which is difficult to define, evaluate and measure. Therefore, fetal, and neonatal size measurement is used as a surrogate marker of potential growth;

however, this parameter doesn't take into account the placental features.

AIMS

Our study aims to evaluate which morphological alterations are present at the placental level, both macroscopically and microscopically, and in fetuses diagnosed with IUGR [1, 2].

MATERIALS AND METHODS

The present work has been set up retrospectively, selecting 31 women from our digital archive, in the period between 2018 and 2022, whose fetuses had been diagnosed with IUGR. The placentas were sampled and processed as routine and subjected to histological analysis by 3 different pathologists to evaluate their morphology, the degree of maturation and the presence of focal lesions associated with the IUGR.

RESULTS

Our data showed that the average age, for all patients, was 34-35 years. The mean gestational age, for all patients, was approximately 27-28 weeks. As for the weight of the chorionic disc, the average was 279 g. In 94% of cases, there was inter- and perivillose fibrin, a characterizing element of these placentas. 58% of placentas presented with microcalcifications, infarction areas (52%) and increased syncytial nodes (32%), indicating a hypoxic condition. Wharton's jelly oedema was present in about 84% of cases.

DISCUSSION

According to the data in the literature, as regards both weight and placental volume, reduced in IUGR infants, and for the presence of multiple elementary lesions related to situations of maternal vascular malperfusion, the lesions observed are not specific to IUGR, as detectable even in situations with regular intrauterine growth of the fetus. Among the various histopathological findings that could contribute significantly to IUGR, compared to normal placentas, the following have been observed: peri- and intervillous fibrinoid necrosis, syncytial node increase, villi hypoplasia, microcalcifications, placental infarctions greater than 5%.

CONCLUSIONS

Although histological changes in the placenta are more common in IUGR infants, it is not possible to determine whether these abnormalities certainly contribute to IUGR. Therefore, we believe that IUGR should be inserted in a multifactorial context, in which placental factors, but also maternal and epigenetic factors, can intervene. The study of the placenta in all IUGR newborns may, therefore, be important to identify the most specific elementary lesions in order to direct the study of pregnancy to a prevention of risk factors.

REFERENCES

[1] Zur RL, Kingdom JC, Parks WT, Hobson SR. The Placental Basis of Fetal Growth Restriction. Obstet Gynecol Clin North Am. 2020;47(1):81-98.

[2] Sun C, Groom KM, Oyston C, Chamley LW, Clark AR, James JL. The placenta in fetal growth restriction: What is going wrong? Placenta. 2020;96:10-8.

ABS 36

A RARE CASE OF URTICARIA PIGMENTOSA

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Cutaneous mastocytosis is characterized by infiltration of mast cells in the papillary and upper reticular dermis. The maculopapular cutaneous mastocytosis variant, also known as urticaria pigmentosa, usually affects children and shows about 1 cm red-brown or yellow papules, tipically located in the trunk and extremities. The incidence of urticaria pigmentosa has been estimated to be 1:8,000 new dermatology patients. A systemic involvement of urticaria pigmentosa, described as mast cells infiltration of the bone marrow, has been reported in 10-70% of patients, though most cases show an indolent course.

Here, we present the case of a 6-year-old patient, clinically presenting with 3-10 mm brownish macules on the forehead and back. Lesions appear to be itchy after physical stimuli like temperature change. Histopathologically, we found an abundant cell infiltration in the papillary and upper reticular dermis. The positivity for C-kit/CD117 staining, and the presence of intracellular basophilic granules at the Giemsa staining, supported the diagnosis of urticaria pigmentosa.

Skin biopsy is needed to exclude other disordes, such as chronic dermatitis and nodular prurigo, in order to define cutaneous mastocytosis. The possibility of progression toward a malignant, systemic mastocytosis suggests the utility of bone marrow biopsy, as well as periodic dermatologic examination. In our database, this is the only case reported in the last 6 years [1-3].

REFERENCES

[1] Allison MA, Schmidt CP. Urticaria pigmentosa. Int J Dermatol. 1997;36(5):321-5.

[2] Di Raimondo C, Del Duca E, Silvaggio D, Di Prete M, Lombardo P, Mazzeo M, Spallone G, Campione E, Botti E, Bianchi L. Cutaneous mastocytosis: A dermatological perspective. Australas J Dermatol. 2021;62(1):e1-7.

[3] Topar G, Staudacher C, Geisen F, Gabl C, Fend F, Herold M, Greil R, Fritsch P, Sepp N. Urticaria pigmentosa: a clinical, hematopathologic, and serologic study of 30 adults. Am J Clin Pathol. 1998;109(3): 279-85.

ABS 37

EUROPEAN LIFE MILCH PROJECT: MATERNAL OBESITY AND NEONATAL GROWTH, BREASTFEEDING AND NEURODEVELOPMENT

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INTRODUCTION

The European Project Life MILCH is a longitudinal and multicenter study still ongoing, co-funded in 2019 by the European Commission, in which the Universities of Cagliari, Parma, Reggio-Emilia and Florence (Italy) participate, targeting mother-child dyads in the critical time window between gestation and the 1st year of life. From a prevention perspective, the aim is to quantify exposure to endocrine-disrupting chemicals (EDCs), ubiquitous chemical pollutants with possible adverse health effects, through careful human bio-monitoring of mothers and their infants, followed at birth and in follow-up at the 1st, 3rd, 6th, and 12th months of life in a pathway of neonatological examinations and neurodevelopmental testing [1].

OBJECTIVES

In the present study, we first investigated whether exposure in pregnancy, through diet, to plastics and EDCs present in them could influence maternal body mass index (BMI) and fetal and neonatal growth outcomes. Secondly, we compared the neurodevelopment of breastfed and non-breastfed infants at 6 months of age, through the Bayley-III edition (BSID-III) Scales, under the assumption that breast milk may favorably affect optimal brain development [2].

METHODS

For the first objective, 100 mothers (and their respective babies), recruited from the AOU of Monserrato (Cagliari, Italy), were selected and completed an online questionnaire inherent to the consumption of 15 foods packed in plastic during pregnancy, obtaining a food exposure score to plastic (range 1-15). By regression analysis, we examined the possible association of this score with maternal BMI at the beginning of pregnancy and with the parameters of the infants at birth and at follow-up visits (weight and length).

For the second objective, we compared by t-test the averages of the composite scores obtained at the 5 areas (cognitive, motor, language, social-emotional and adaptive behavior) of the BSID-III from a first group of 35 breastfed infants up to 6 months and from a second group of 8 non-breastfed infants up to 6 months who were fed exclusively with formula milk.

RESULTS

For the first objective, despite the presence of many outliers, there is a slight trend of increasing maternal BMI and decreasing infant weight and length at birth and at 1 month of age, in relation to the pregnancy plastic exposure score; in particular, this trend is statistically significant for infant length at 1 month (Spearman's rho = -0.35, p = 0.025). Furthermore, from multivariate analysis, the impact of plastic on neonatal parameters at each T appears to be an order of magnitude less than that of gestational age.

Regarding the second objective, statistically significant differences at t-test (p-value < 0.05) emerged for the mean composite scores obtained at BSID-III by long-term breastfed infants on the cognitive (103.57 ± 10.33) and motor (109.02 ± 17.22) scales, which were significantly higher, compared with those obtained on the same scales, cognitive (95 ± 7.07) and motor (92.87 ± 11.15), by formula-fed infants.

CONCLUSIONS

Although the power of the study is limited, the results suggest a possible impact, albeit modest, of plastic exposure on maternal BMI and fetal and neonatal growth outcomes and, albeit on a limited sample of the population, corroborate the favorable role of breastfeeding on child neurodevelopment [3].

REFERENCES

[1] LIFE18 ENV/IT/000460 – Life MILCH Mother and Infant dyads: Lowering the impact of endocrine disrupting Chemicals in milk for a Healthy Life. Call: LIFE 2018 – Call for proposals for LIFE ACTION GRANTS – Programme: LIFE Environment and Resource Efficiency.

[2] Uauy R, Peirano P. Breast is best: human milk is the optimal food for brain development. Am J Clin Nutr. 1999;70(4):433-4.
[3] Horta BL, Loret de Mola C, Victora CG. Breastfeeding and intelligence: a systematic review and meta-analysis. Acta Paediatr. 2015;104(467):14-9.

ABS 38

RARE DISEASES AND COMPLICATIONS: DEEP VEIN THROMBOSIS AND POMC DEFICIENCY. A CASE REPORT

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INTRODUCTION

POMC deficiency is a rare autosomal recessive disorder caused by mutations in the *POMC* gene (chromosome 2p23.3) [1, 2]. Deficiency of melanocortins, such as ACTH and MSH, underlies the clinical phenotype, which is characterized by: hypopigmentation, red hair, hyperphagia, early obesity, hypoglycemia, hyperbilirubinemia, neonatal cholestasis, and hypocortisolism [2]. Venous thromboembolism (VTE), related to venous stasis, endothelial damage, and hypercoagulability, is a rare event in the pediatric age. With an incidence of 1 case/100,000/year and a 2.2% mortality rate, VTE represents an increasing problem in hospitalized children [3].

CASE REPORT

F. is a 9-year-old female with POMC deficiency, presenting with hypergrowth, right somatic hemihypertrophy, global developmental delay, severe hypotonia, and inability to walk. Due to sudden onset of pain and swelling in the right lower limb, she was admitted to our department. Right lower limb echocolor-Doppler confirmed the diagnosis of deep vein thrombosis (DVT), showing total occlusion of the right femoral vein. Later, angio-CT revealed an extensive thrombus on the common femoral axis, deep femoral vein and superficial femoral vein up to the popliteal vein, with oedema of the soft tissues, and a clot in the distal tract of the external iliac vein. On the basis dosage of factor X value, which was below normal with a value of 58%, hydrocortisone therapy, low molecular weight heparin and elastocompression of both limbs were started, with progressive resolution of the pathological picture.

CONCLUSIONS

DVT is a rare occurrence in the pediatric age. Common causes include central venous catheters, chronic disease, thrombophilia, and different classes of drugs. According to the American Society of Hematology (ASH) guidelines, duration of the anticoagulant prophylaxis is unclear. Our case highlights the complexity of therapy in patients with rare diseases and the key role of follow-up in the context of anticoagulant prophylaxis. We also highlight the importance of a multidisciplinary approach to the rehabilitation of patients with rare syndromes and severe obesity.

REFERENCES

[1] Viakhireva I, Kalinchenko N, Vasilyev E, Chistousova GV, Filatova A, Marakhonov A, Rubtsov PM, Skoblov M, Tiulpakov A. A Founder Mutation in the POMC 5'-UTR Causes Proopiomelanocortin Deficiency Through Splicing-Mediated Decrease of mRNA. J Clin Endocrinol Metab. 2022;107(9):e3654-60.
[2] Gregoric N, Groselj U, Bratina N, Debeljak M, Zerjav Tansek M, Suput Omladic J, Kovac J, Battelino T, Kotnik P, Avbelj Stefanija M. Two Cases With an Early Presented Proopiomelanocortin Deficiency – A Long-Term Follow-Up and Systematic Literature Review. Front Endocrinol (Lausanne). 2021;12:689387.
[3] Witmer C, Raffini L. Treatment of venous thromboembolism in pediatric patients. Blood. 2020;135(5):335-43.

ABS 39

HEMORRHAGIC DIARRHEA: WHEN PURPURA TAKES OVER

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INTRODUCTION

Hemorrhagic diarrhea in children is often secondary to infectious enteritis, rarely to chronic inflammatory bowel disease (IBD), hemolytic uremic syndrome, and Shönlein-Henoch purpura (SHP). The most common bacteria are *Salmonella* and *Campylobacter*. The role of *Clostridium difficile* (CD) is controversial and often saprophytic. CD testing is recommended for children older than 2 years of age in case of prolonged diarrhea of unknown etiology or with risk factors (IBD, immunodeficiency, and recent antibiotic therapy).

CASE REPORT

An 8-year-old male presented to the Emergency Room for gastric vomiting that had lasted for 4 days and abdominal pain, accompanied by a single episode of melena followed by hemorrhagic diarrhea in the previous 24 hours. His medical history was consistent with regular growth and alternating bowel pattern.

Vital signs were within normal limits, abdomen soft to touch, with widespread tenderness, no signs of peritonism, and normal bowel movements.

Lab tests showed neutrophilic leukocytosis, increased platelet count, stools positive for CD/toxin (FILMARRAY™); signs of adenomesenteritis on abdominal ultrasound.

In suspicion of CD enterocolitis, endovenous hydration and antibiotic therapy were started. On the 4th day of hospitalization, due to bilious vomiting with persistent abdominal pain, a direct abdomen X-ray and abdomen contrast CT scan were performed: negative for intestinal occlusion or perforation. Oral vancomycin was then continued for a total of 10 days (replaced with metronidazole due to poor tolerance), with progressive resolution of the gastrointestinal symptoms. On the 8th day of hospitalization, the child presented arthritis of the right wrist associated with rare petechiae with rapid self-resolution on the feet. Laboratory tests were normal for ASCA, ANCA, IgA, C3, C4, FR, ANA, anti-transglutaminase, but fibrinogen and D-dimer were elevated.

The child remained in good general clinical condition for 10 days, then the abdominal pain returned, with loss of appetite associated with a 10% weight loss. Therefore, in the suspicion of IBD, a colonoscopy was performed, which showed a picture of non-specific colitis. Four weeks after the onset of symptoms, there was evidence of typical purpura in the lower limbs associated with left ankle arthritis compatible with SHP. During the follow-up, there was no evidence of renal involvement (hematuria or proteinuria).

CONCLUSIONS

In SHP, gastrointestinal manifestations may precede purpura in 10% of cases, making the diagnosis tricky [1]. In our case, the stool positivity for CD/toxin was a confounder, although we could not exclude its pathogenetic role in vasculitis inflammation. The high use of multiplex panels in microbiological diagnostics, with a consequent increase in the diagnosis of CD, requires us to discriminate against its pathogenic role to avoid useless treatments.

REFERENCE

[1] Leung AKC, Barankin B, Leong KF. Henoch-Schönlein Purpura in Children: An Updated Review. Curr Pediatr Rev. 2020;16(4):265-76.

ABS 40

HIGHLIGHTS FOR MANAGEMENT OF BLADDER BOWEL DYSFUNCTION IN CHILDREN

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INTRODUCTION

Bladder bowel dysfunction (BBD) is a generic term used to refer to a variety of lower urinary tract symptoms (LUTS) associated with constipation and/or encopresis. It is a common condition, as it occurs in 15-20% of kids during their school years, and makes up to almost 40% of consults in pediatric urology. Nonetheless, it often remains underestimated [1-3].

PATHOGENESIS

There's not a common consensus on BBD's pathogenesis, as both familiar and environmental factors seem to be important [1]. Generally, children with a familiar history of BBD are more likely to develop the same dysfunctions in the urinary tract as well as functional constipation. Nonetheless, scientific literature shows how living in a hostile family environment, bad habits in terms of diet and lifestyle, can also lead to BBD [2]. Finally, recent studies from Westwell-Roper et al., found an association of high OCD-related impairment and psychiatric comorbidities with BBD [4].

CLINICAL MANIFESTATIONS

LUTS include dysuria, urgency, urinary frequency, hesitancy, daytime incontinence, enuresis, dribbling, straining, voiding postponement, and urinary retention; overactive bladder, underactive bladder, and dysfunctional voiding can be present, as well [2].

BBD also counts as a risk factor for urinary tract infections (UTIs) in the pediatric population, affecting their prognosis as well, and often presents itself along with vesicoureteral reflux (VUR): this association has a higher risk of recurrent UTIs than those who only have VUR, and can eventually lead

to kidney failure and renal scarring [2]. Moreover, BBD also has an impact on the prognosis of UTIs, therefore making early diagnosis and treatment extremely important in the management of these patients.

Functional constipation is also a very common manifestations in these patients, with a reduced frequency of bowel movements, leading to painful defecation and, sometimes, encopresis. The latter is usually the result of stool impactions obstructing the bowel, forcing liquid feces to go around and be expelled with intestinal gases. Functional constipation may be caused by fault of fibers and vegetables in the child's diet, or by behavioral problems [1].

TREATMENT

First-line approach to BBD is usually urotherapy, a non-pharmacological and non-surgical line of treatment, which leads to resolution in about 50% of the patients [2]. It consists of educating the child and the caregivers, giving adequate instructions on hydration (6 glasses of water/die) and diet (balanced, rich in fibers, fruits and vegetables), defining specific times for voiding (every 2-3 hours more or less) and helping the child with pelvic floor muscles awareness. Although functional constipation is often treated with diets rich in fibers, probiotics, behavioral education and adequate hydration, administering macrogol has shown to be by far the most effective.

Pharmacological treatment must be considered in case of failure of urotherapy, and relies on anticholinergics in order to act upon the child's discomfort. Other options include: solifenacin, β3-adrenoceptor agonists, alpha blockers. Sometimes, supplements (e.g., cranberry) may be considered, especially in case of VUR. Finally, surgical treatment is reserved to cases of refractive detrusor overactivity and intractable constipation [2].

CONCLUSIONS

BBD negatively impacts the children's quality of life as well as their confidence. When left untreated, it can lead to severe complications regarding the patients' psyche and their kidneys and bladders' function, even resulting in incontinence and/or chronic illnesses. Therefore, early diagnosis and treatment are crucial in order to prevent further damage.

REFERENCES

[1] Masnata G, Cracolici E, Borrello S, Casano S, Madeddu E, Manca V. La disfunzione vescico-intestinale nel bambino. Riv Ital Pediatr Ospedal. 2020;7(2).

[2] Santos JD, Lopes RI, Koyle MA. Bladder and bowel dysfunction in children: An update on the diagnosis and treatment of a common, but underdiagnosed pediatric problem. Can Urol Assoc J. 2017;11(1-2 Suppl 1):S64-72.

[3] Ribeiro RS, Abreu GE, Dourado ER, Veiga ML, Lobo VA, Barroso U Jr. Bladder and bowel dysfunction in mothers and children: a population-based cross-sectional study. Arq Gastroenterol. 2020;57(2): 126-30.

[4] Westwell-Roper C, Best JR, Naqqash Z, Afshar K, MacNeily AE, Stewart SE. Bowel and Bladder Dysfunction Is Associated with Psychiatric Comorbidities and Functional Impairment in Pediatric Obsessive-Compulsive Disorder. J Child Adolesc Psychopharmacol. 2022;32(6): 358-65.

ABS 41

POLYCHONDRITIS COMPLICATED BY NEPHRITIS IN CHILDHOOD: DESCRIPTION OF A CLINICAL CASE WITH CUTANEOUS VASCULITIS AND PANCA POSITIVITY WITH ATYPICAL PATTERN

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INTRODUCTION

Relapsing polychondritis (RP) is a rare autoimmune disease characterized by recurrent, progressive inflammation and degeneration of the cartilaginous structures. Sites typically involved include the ear, nose, larynx, and tracheobronchial tree. Pediatric cases are rare and reported in the literature only as sporadic case reports, sometimes associated with autoimmune diseases [1]. We present the case of a 7-year-old boy with polychondritis associated with cutaneous vasculitis complicated by nephritis.

CASE REPORT

M. came to our attention on suspicion of Schönlein-Henoch purpura. He presented cutaneous vasculitis with ecchymosis and rare non-palpable confluent purpuric elements mainly affecting legs. He also had diffuse arthralgias, epistaxis, severe costochondral pain and scrotal edema.

Laboratory tests showed increased inflammation indices (ESR, CRP); mild urinary abnormalities were detected, but with normal renal function and isolated positivity for pANCA in immunofluorescence (atypical pattern; negativity

of antiMPO and PR3). Because of the appearance of intense abdominal pain and the positivity of occult blood in feces, therapy with prednisone 1 mg/kg/day was started.
The following day, M. presented a respiratory crisis with laryngeal constriction and chondritis of the left ear (which appeared edematous, erythematous), and of the thickness of the auricular cartilage. In suspicion of polychondritis, steroid therapy was increased, with the introduction of boluses of methylprednisolone.
The instrumental investigations (CAT and PET), performed to evaluate the inflammation of the cartilaginous structures, specially the respiratory tract, were negative (but the ongoing high-dose steroid therapy may have extinguished the inflammation).
The search for anti-collagen type 2 antibodies typical of RP (conducted in Sweden) was positive, but within the range for the Swedish adult population (not evaluable in pediatric age).
After a few weeks of treatment with cortisone and immunosuppressive therapy with methotrexate, he had, however, persistence of microhematuria (+++) and proteinuria.
Given the positivity of pANCA antibodies and the association of RP with pANCA-positive vasculitis, a renal biopsy was considered, but not yet performed because of the normalization of urine test and negativization of pANCA during follow-up.

CONCLUSIONS

RP is a little-known disease with a poor prognosis, which is associated in up to a third of cases with autoimmune diseases, in particular pANCA-related vasculitis. These can become manifest in various stages of the disease and involve vessels of any organ and caliber. In this case, the appearance of chondritis of the ear allowed prompt recognition of the pathology.

REFERENCE

[1] Alqanatish JT, Alshanwani JR. Relapsing polychondritis in children: A review. Mod Rheumatol. 2020;30(5):788-98.

ABS 42

TWINNING AS A RISK FACTOR FOR NEONATAL ACUTE INTESTINAL DISEASES

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OBJECTIVES

Acute intestinal diseases (AID), including necrotizing enterocolitis and spontaneous intestinal perforation, are a group of conditions that typically present in preterm infants, and are associated with an elevated mortality and morbidity rate. The risk factors for these diseases remain largely unknown. We hypothesized that twinning may represent a risk factor for AID.

AIMS

The aim of the study is to identify the correlation between twinning and the development of AID.

METHODS

A single-center retrospective case-control study was conducted. We recruited all infants with a diagnosis of AID, confirmed by anatomopathology when available, born in the Neonatal Intensive Care Unit at Sant'Anna Hospital, in Turin (Italy), between 2010-2020. Considering the rarity of the outcome, 4 matched controls for each subject were randomly chosen from the overall population of newborns. Odds ratio (OR) and 95% confidence interval (CI) were calculated using a conditional logistic regression model and a multivariate model by the creation of a directed acyclic graph (www.dagitty.net).

RESULTS

A total of 69 cases and 276 controls were recruited.
The comparison between the two groups shows an association between the development of AID and perinatal asphyxia, late-onset sepsis and feeding intolerance.
Regarding maternal data, medically assisted pregnancy appears to be a risk factor, while the administration of antenatal steroids seems to be protective.
Logistic regression assessed the total effect of twinning on the outcome, using the medically assisted pregnancy as a covariate, with an OR of 1.55 (95% CI: 0.86-1.80).
A multivariate analysis was then conducted using gestational age, intrauterine growth restriction, preterm premature rupture of membranes and maternal comorbidities as covariates, with an OR of 1.61 (95% CI: 0.91-2.87).

CONCLUSIONS

Data regarding the association between twin pregnancies and short-term neonatal outcomes are limited. Our study suggests that twinning may be a risk factor for the development of AID. Due to the small number of cases observed, further studies on larger populations are needed.

ABS 43

INHIBITED CATATONIA IN AN INSTITUTIONALIZED 17-YEAR-OLD MALE: A IATROGENIC CASE REPORT

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INTRODUCTION

Catatonia is a psychomotor syndrome characterized by disorders of motility, affectivity and behavior. It is due to organic and non-organic causes, such as psychiatric, neurological and metabolic disorders, drug intake or suspension. It is involved the prefrontal cortex and/or neurotransmitters (GABA, glutamate, serotonin and dopamine). It recognizes 3 subtypes: inhibited (mutacism, rigidity, incontinence, food refusal), malignant (fever, alterations of the autonomic system, muscular rigidity), and excited (aimless hyperactivity, stereotypes, aggression). Benzodiazepines (effective in 80%) are the first-line treatment, and electroconvulsive therapy is used in selected cases [1, 2].

CASE REPORT

A 17-year-old male was entrusted to social services for parental deficiencies from childhood and welcomed into communities for medium-mild cognitive retardation and conduct disorders. After 2 months from the inclusion in the last community, he presented loss of motor skills, food refusal and weight loss. Therefore, he was admitted to the Emergency Room (ER). The motor picture was interpreted as congenital and, in suspicion of gastroesophageal reflux dysphagia, he was treated with parenteral hydration and pantoprazole. Four days later, due to the onset of fever and dyspnea, he was taken to ER again. Blood tests showed neutrophilic leukocytosis, C-reactive protein 13.5 mg/dl, CPK normal; chest X-ray and pneumological evaluation confirmed diagnosis of bronchitis. Blood, urine and saliva culture were negative. He was pale, with increased heart and respiratory rate, $SatO_2$ 94%, scattered crackling rales, altered consciousness (mutacic, unreactive, wide-eyed), distal hypertonicity, labial tremors, absence of sphincter control, multiple bedsores. Piperacillin-tazobactam, i.v. hydration, pantoprazole and antithrombotic prophylaxis were started. Personal history collection was difficult; however, we learned that the previous motor and neurological status was normal up to a few days after the decompensation, and that he was receiving clotiapine, valproate, benzodiazepine and other neuroleptics as needed. Brain CT, brain MRI and echocardiogram were normal and, in suspicion of inhibited catatonia (a 24 score on the Bush-Francis Catatonia Rating Scale), he suspended neuroleptic therapy and performed challenge tests with i.v. lorazepam. We observed motor and relational response within 30 min. Thus, the drug was introduced into chronic oral therapy with resolution of the clinical picture.

CONCLUSIONS

In our case, the response to lorazepam confirmed the suspicion of inhibited catatonia. The anamnestic gaps do not allow us to define with certainty the triggering cause (neuroleptics or aggravation of the psychic picture from adaptation difficulties), which is, however, iatrogenic and invites us to pay more attention to the objective examination and the anamnesis.

REFERENCES

[1] Callari A, Bartolommei N, Lattanzi L, Mauri M. Clinical management of catatonic syndromes. Part I: diagnosis, clinical setting and treatment principles. J Psychopatol. 2014;20:144-52.

[2] Bartolommei N, Callari A, Lattanzi L, Mauri M. Clinical management of catatonic syndromes. Part II: therapeutic algorithms. J Psychopatol. 2014;20:244-51.

ABS 44

A RARE CASE OF NEUROLOGICAL COMPLICATIONS IN WAARDENBURG-SHAH SYNDROME

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INTRODUCTION

Waardenburg syndrome (WS) is a genetic disorder characterized by hypochromic spot, piebaldism, sensorineural hearing loss, iris heterochromia, and blue eyes. WS has a heterogeneous genetics: types I and III are associated with mutations of the *PAX3* gene and present with dystopia cantorum and limb

anomalies, respectively; type IV, or Waardenburg-Shah, manifests with Hirschsprung disease and is associated with mutations in the endothelin-3 genes; mutations of the *MITF* gene are found in 15% of WS type II cases. WS1 and WS2 are the most common variants, WS3 and WS4 are rarer. The incidence is around 1:40,000 [1, 2]. We present a rare case of WS4 with particular neurological complications.

CASE REPORT

A. is a 14-year-old female, born at term from a physiological pregnancy, with evident signs of piebaldism, heterochromia of the iris, horizontal nystagmus, hypotonia of the legs, deambulation delay (acquired at the age of 2) and ataxia. For the detection of severe sensorimotor polyneuropathy, exome analysis (WES) was performed, which revealed a *de novo* heterozygous mutation in the *SOX10* gene, associated with Waardenburg-Shah syndrome [3].

During the follow-up, the girl performed EMG/ENG, with evidence of widespread chronic sensorimotor neurogenic suffering of a predominantly myelinopathic type, more marked in the legs. She did EEG, which resulted normal. The girl then started a rehabilitation program with the aim of regaining muscle strength; today, the little girl walks with support.

CONCLUSIONS

With this case report, we want to underline the importance of the multidisciplinary approach to rare diseases, which are often complex; it is necessary to follow the patient over the years to intercept early some clinical manifestations.

REFERENCES

[1] Read AP, Newton VE. Waardenburg syndrome. J Med Genet. 1997;34(8):656-65.

[2] Hageman MJ, Delleman JW. Heterogeneity in Waardenburg syndrome. Am J Hum Genet. 1977;29(5):468-85.

[3] Pingault V, Zerad L, Bertani-Torres W, Bondurand N. SOX10: 20 years of phenotypic plurality and current understanding of its developmental function. J Med Genet. 2022;59(2):105-14.

ABS 45

EVOLUTION OF KNOWLEDGE ABOUT CONGENITAL HIP DYSPLASIA (CHD) IN THE HISTORY OF PEDIATRICS

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The clinical picture of congenital hip dysplasia (CHD) was well known to Hippocrates, who in *De articulis* described claudication and pain, unhealthy posture, muscular hypotrophy, and underdevelopment of the affected limb. For about twenty centuries, this knowledge remained almost unchanged. In particular, since ancient times, the anatomy of the area had been known about the bone apparatus, but muscles, tendons, and the real structure of the joints and that of ligaments were described in 1543 only, in *De humani corporis fabrica* by Andrea Vesalio (1514-1564), who also recognized the morphological and functional differences; on the other hand, Realdo Colombo (1516-1559) described femur ossification centers (*De re anatomica*, 1559). Ambrose Parè (1517-1597) was the first to attempt a differential diagnosis between CHD and claudications secondary to osteitis or acute osteomyelitis. Two centuries later, Nicolas Andry (who coined the term "orthopedics" to indicate the study and treatment of skeletal deformities in children) fixed the clinical picture of rickets, especially in early childhood. The spreading in medicine of Morgagni anatomical-clinical method allowed then G.B. Paletta (1747-1830) to rationally describe CHD on an anatomical-pathological basis in 1788 and, more completely, in 1820. The modern period in the history of CHD can be traced back to the work of Guillaume Dupuytren (1777-1835) in 1826 on claudication caused by the congenital displacement of the femoral head. In this work, the French Master completes Paletta's observations, reiterates the need, already known since antiquity, to make an early diagnosis, indicates as a characteristic symptom the ease of longitudinal movement of the limb suspected of dislocation, the so-called "Dupuytren's sign", which proved to be more indicative than the various measurements of the diameters of the trunk proposed by Roser and Nelaton (1854) and by Peter (1856) or the Bryant triangle or the Schoemacher line, which are shown only at the time of loading. The discovery of X-rays by W.C. Röntgen (1895) provided an effective diagnostic tool in CHD, too. The first systematic study on the subject is owed to Boston (1902), and then we arrive to the masterful works of Vincenzo Putti (1880-1940), which made it possible to clarify the signs of subluxation (Putti's triad) and led the Bologna Master to propose a radiological screening for all newborns (1929). On a clinical level, the most pathognomonic and reliable maneuver for the diagnosis of CHD was identified in Ferrara by Marino Ortolani (1904-1983) in 1936, "click

sign" (as Ortolani himself defined it), which was perfected in 1962 by T.G. Barlow, thus allowing hip dislocation to be demonstrated as well. The Ortolani-Barlow sign still retains all its validity even after the studies by R. Graf (1983-1986) on the use of ultrasound in the pathology of the coxo-femoral joint of the infant, a method that currently represents the gold standard in the diagnosis of CHD.

ABS 46

HOW TO DEAL WITH NEONATAL POST-OPERATIVE PAIN: STRONGER TOGETHER

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CASE REPORT

V. is a preterm newborn (30 weeks + 5 days of gestational age) and weights 1,280 g (appropriate for gestational age). At 3 weeks of age, after the onset of necrotizing enterocolitis complicated by intestinal perforation, she requires surgical bowel resection and the placement of a double-barreled ileocolostomy.

After surgical treatment, the following analgesic therapy is administered:

- fentanyl 0.5 mcg/kg/h by continuous IV infusion for the first 48 hours of the post-operative period;
- paracetamol IV 7.5 mg/kg every 6 hours starting the day after surgery and keeping it for 15 days;
- fentanyl 1.1 mcg/kg IV bolus every 6 hours (alternating every 3 hours with paracetamol boluses) starting 48 hours after surgery and continuing for 1 week.

No adverse events or need to use naloxone for opioid overdoses are recorded throughout the duration of therapy. V. was constantly monitored with EDIN scale, and the therapy effectively controlled the post-operative pain and allowed a good adaptation to the invasive ventilation with ET tube that V. needed to overcome the critical phase of the post-surgical period.

DISCUSSION

Analgesic coverage in the newborn during the post-operative period is strongly recommended, and should be carried out continuously for at least 48-72 hours. Despite this, nowadays there is not enough evidence to give precise indications on therapeutic options. Opioids are generally considered the first choice for major surgery, especially in the ventilated child. The studies published to date do not allow to determine which dosage, which route and which mode of administration are preferable. Fentanyl administered via boluses seems to be safer than its continuous infusion, although some studies have shown an increase in episodes of severe apnea. Paracetamol can be used as an opioid saver. Personalization of treatment through constant monitoring of pain and the effectiveness of therapy is essential.

CONCLUSIONS

Further studies are needed to better target our treatments and to understand long-term risks related to the use of pain medications in the neonatal period. However, we can say that alternating paracetamol and fentanyl boluses can be a useful strategy to reduce the dosage of opioids and, therefore, possible side effects.

REFERENCES

• Ceelie I, de Wildt SN, van Dijk M, van den Berg MM, van den Bosch GE, Duivenvoorden HJ, de Leeuw TG, Mathôt R, Knibbe CA, Tibboel D. Effect of intravenous paracetamol on postoperative morphine requirements in neonates and infants undergoing major noncardiac surgery: a randomized controlled trial. JAMA. 2013;309(2):149-54.

• Lago P, Garetti E, Pirelli A, Merazzi D, Savant Levet P, Bellieni CV, Pieragostini L, Ancora G. Linee Guida per la prevenzione ed il trattamento del dolore nel neonato. Milan: Biomedia, 2016.

• Vaughn PR, Townsend SF, Thilo EH, McKenzie S, Moreland S, Denver KK. Comparison of continuous infusion of fentanyl to bolus dosing in neonates after surgery. J Pediatr Surg. 1996;31(12):1616-23.

ABS 47

NEURODEVELOPMENTAL DISORDERS: ARE THERE LINKS OR SYMPTOMATOLOGICAL SIMILARITIES BETWEEN AUTISM SPECTRUM DISORDERS, ASPERGER SYNDROME, SAVANT SYNDROME?

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INTRODUCTION

Autism spectrum disorders, Asperger syndrome and savant syndrome are neurodevelopmental disorders

that can be easily confused if there is no deep knowledge and experience in their evaluation. Therefore, it is important to analyze them individually in order to assess the peculiar characteristics of each and any similarities with autism.

AUTISM SPECTRUM DISORDERS

These are heterogeneous neurodevelopmental disorders characterized by persistent deficits in communication and social interaction, with particular behavioral patterns. Affected individuals often have neurological, psychiatric and medical comorbidities. In Italy, currently 1 in 77 children, between the ages of 7 and 9, has an autism spectrum disorder. More male individuals are affected, 4.4 times more than female individuals. Very heterogeneous and variable is the clinical symptomatology, and we can basically summarize it in persistent deficits in communication and social interaction, repetitive behavior patterns, interests or activities, fixed, restricted, lacking flexibility. They may, in addition, present hyper- or hypo-reactivity as a response to sensory stimuli, or unusual interests in environmental aspects. In the very young child, shifty gaze, little interest in attempts to engage, lack of or limited seeking in relationships and play with other children, delayed or failed development of language and alternative ways of making oneself understood, such as through gestures or facial expressions. Those who manage to develop some communicative language tend to repeat certain expressions and have difficulty maintaining a conversation with another person. Above all, they show little ability to adapt to another.

ASPERGER'S SYNDROME

This syndrome is basically due to a neurodevelopmental disorder that limits the ability to socialize and communicate; however, unlike autism, it does not involve intellectual or language disabilities. Asperger's individuals usually exhibit repetitive behaviors and, more rarely, poor motor coordination. It usually manifests itself during the first 2-3 years of life; at school, they have difficulty socializing or conversing. Currently, Asperger's syndrome is considered among the autism spectrum disorders. Important to know is that an Asperger's individual, despite having reduced ability to socialize and communicate, may also have no additional intellectual and language disabilities. However, the most peculiar symptoms of the syndrome concern language, communication, socialization, daily interests and motor skills. They may speak in a monotonous and pedantic tone of voice, and often cannot distinguish between idioms and ironic phrases. Motor-wise, they resemble autistics, presenting with poorly coordinated movements. They generally have a normal IQ, more rarely they can appear "brilliant", showing exceptional gifts and qualities in the fields of music, computer science and mathematics.

SAVANT SYNDROME

This is a rarely encountered situation, characterized by mental/cognitive disability, sometimes severe, but exceptionally some subjects show exceptional abilities, above average, sometimes so talented as to make them reach remarkable levels. Savant subjects are called "islands of genius", and three different types have been described:

- splinter savant or splinter skills – subjects with peculiar abilities;
- talented savant – real talents in specific areas: musical, artistic, etc;
- prodigious savant – with exceptional talents and abilities that make them extraordinary prodigies in art, music, computation, mechanics and spatial abilities, mathematics.

This syndrome can occur at birth due to congenital neurodevelopmental abnormalities, but also be acquired as a consequence of brain injury or other diseases (fronto-temporal dementia). In about 50 percent of cases, savant syndrome is associated with autism. Hardly the opposite occurs, because only 1/10 with autism appears to have savant-like symptomatology. Very few autistic individuals can develop and manifest "savant-like abilities". In fact, in the majority of cases, it is high-functioning autism or Asperger's syndrome.

CONCLUSIONS

In the light of the above, it is clear that these disorders often present similar symptomatology, and therefore it may be difficult to differentiate and frame them clinically and therapeutically. Certainly, the experience of a pool of specialists may be helpful for a correct diagnosis.

ABS 48

DEEP VEIN THROMBOSIS IN A CHILD WITH POMC DEFICIENCY: CASE REPORT FROM THE POINT OF VIEW OF A PEDIATRIC EMERGENCY DEPARTMENT

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CASE REPORT

A 9-year-old female comes to our Pediatric Emergency Medicine Department for pain and swelling of the right lower limb, which has begun the same morning. The family's origins are not from Italy, so the child and her parent speak Italian with difficulty. Her father refers to a trauma to the limb in the previous days and a fracture of the right foot 5 years prior. The girl is affected by proopiomelanocortin (POMC) deficiency. POMC is a polypeptide that, through the activation of several hormones and receptors, regulates cortisol secretion. Congenital POMC deficiency is caused by a mutation in the *POMC* gene located in chromosome 2. This disorder is characterized by obesity, adrenal insufficiency and decreased skin pigmentation [1].

In our case, the patient has delayed psychomotor development, adrenal insufficiency and obesity. For her disease, she takes a therapy with hydrocortisone. On physical examination, fair general conditions were observed. Edema of the entire right lower limb is observed, with pain on mobilization and attitude of the thigh in abduction. The knee is swollen and warm. At the level of the right big toe, there is a small crusted lesion. The rest of clinical evaluation results normal.

The blood tests show: white blood cells 15,220/μL (of which NE 71.9%), CRP 4.97 mg/dL, ATIII 55%, fibrinogen 454 mg/dL, D-dimer 6,651 ng/ml. PT and APTT are normal. An X-ray of the right lower limb and a cardiological examination are required, but both exams result normal. The venous color Doppler ultrasound highlights a deep vein thrombosis (DVT) at the level of the right femur. The vascular surgeon recommends therapy with low molecular weight heparin, elastic compression and a venous color Doppler control after 15 days. The patient is admitted to the Department of Pediatrics and Rare Diseases of "Microcitemico Hospital" in Cagliari, Italy.

CONCLUSIONS

DVT refers to a blood clot that forms in the deep veins of the extremities, the thoraco-abdominal area, or the cerebral sinuses. Venous thrombosis is rare in pediatric population and most of the time it occurs in children who have certain risk factors, such as the presence of central venous catheters, immobility, infections, cancer, congenital heart disease. Steroid therapy, as in the case of our patient, is also considered a risk factor for venous thrombosis [2, 3]. The particularity of our case is in the difficulties faced for a correct diagnostic classification. The child's mental retardation and the presence of a linguistic barrier complicated the collection of anamnestic data and clinical evaluation. Furthermore, the presence of an ulceration on the right foot and, in the anamnesis, a trauma in the previous days could direct us towards numerous possible diagnoses. At the same time, however, the patient's obesity, her difficulty walking, and the long-term steroid therapy were decisive in raising the diagnostic suspicion of DVT.

REFERECES

[1] Çetinkaya S, Güran T, Kurnaz E, Keskin M, Sağsak E, Savaş Erdeve S, Suntharalingham JP, Buonocore F, Achermann JC, Aycan Z. A Patient with Proopiomelanocortin Deficiency: An Increasingly Important Diagnosis to Make. J Clin Res Pediatr Endocrinol. 2018;10(1):68-73.

[2] Jaffray J, Young G. Deep vein thrombosis in pediatric patients. Pediatr Blood Cancer. 2018;65(3):e26881.

[3] Stephens ET, Nguyen ATH, Jaffray J, Branchford B, Amankwah EK, Goldenberg NA, Faustino EVS, Zakai NA, Stillings A, Krava E, Young G, Fargo JH. Risk of venous thromboembolism in pediatric hospitalized patients undergoing noncardiac surgery: A report from the Children's Hospital-Acquired Thrombosis consortium. Res Pract Thromb Haemost. 2022;6(7):e12810.

ABS 49

IF YOU EAT WHEAT, IT IS BETTER TO SIT! A CASE OF WHEAT-DEPENDENT EXERCISE-INDUCED ANAPHYLAXIS

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BACKGROUND

Food-dependent exercise-induced anaphylaxis (FDEIA), or, more precisely, "summation" anaphylaxis, necessarily involves the association between the ingestion of a food (specific or non-specific) and physical exercise. There are also reported cases of FDEIA triggered by the presence of cofactors such as drugs, alcohol, infectious diseases, or emotional stress. The underlying pathogenesis of FDEIA is not yet well understood: it is hypothesized to involve an IgE-mediated mechanism characterized by a reduced tolerance, induced by physical exertion, towards an otherwise tolerated food. In this context, wheat-dependent exercise-induced anaphylaxis (WDEIA) represents the most common type of FDEIA [1, 2].

CASE REPORT

S. is an 11-year-old boy followed at the Pediatric Clinic of the Fondazione IRCCS Policlinico San Matteo in Pavia, Italy, for grape-induced anaphylaxis, equipped with an adrenaline auto-injector. He presented to the clinic with a new episode of anaphylaxis (urticaria, angioedema and wheezing) that occurred while he was in good health, approximately 90 minutes after consuming a sandwich with beef hamburger and Emmental cheese, foods he had always tolerated. S. had not taken any drugs and denied any insect stings. During a more thorough medical history collection, S. mentioned he played soccer shortly after his meal. In suspicion of FDEIA, specific IgE testing for wheat and its recombinant components was performed, revealing a positive result for Tri a 14 (0.46 kUa/l), while Tri a 19 was negative. These results suggested a rare case of WDEIA. Considering the highly indicative clinical history, it was decided to forgo the oral food challenge and, instead, recommend avoiding physical activity within 2 hours before and 4 hours after meals containing wheat. However, a few months later, S. returned to our clinic with a new anaphylactic reaction that occurred 2 hours after eating pasta and immediately engaging in physical activities with friends, thus confirming the diagnosis of WDEIA.

CONCLUSIONS

In WDEIA, both physical exercise and wheat consumption are individually tolerated, while their combination can trigger anaphylactic reactions. WDEIA is often underdiagnosed in Emergency Departments; however, understanding it is crucial to refer the patient to the Allergy Service for diagnostic confirmation and follow-up. Sensitization to Tri a 19 is the most frequently observed, while cases associated with Tri a 14 (commonly causing the so-called "baker's asthma") are rarer. Currently, there are no markers to determine the threshold of physical activity capable of precipitating anaphylaxis in these patients; therefore, it is essential to strictly adhere to the recommendations provided by healthcare professionals to prevent further episodes [3].

REFERENCES

[1] Shin M. Food allergies and food-induced anaphylaxis: role of cofactors. Clin Exp Pediatr. 2021;64(8):393-9.

[2] Indirli GC, Anania C, Cuomo B, D'Auria E, Decimo F, Mastrorilli V, Santoro A, Sartorio MUA, Veronelli E, Calvani M. Anafilassi da esercizio cibo-dipendente o meglio da sommazione. Riv Immun Allergol Ped. 2020;34(02):I-XVI.

[3] Capra ME, De Paulis NG, Montagni M, Peveri S, Savi E, Biasucci G. An unusual case of wheat dependent exercise induced anaphylaxis (WDEIA) triggered by Tri a 14 in a pediatric patient: a case report. Eur Ann Allergy Clin Immunol. 2018;50(4):187-9.

ABS 50

A BLOOD STORY

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CASE REPORT

A 5-year-old girl arrives at our Pediatric Emergency Department at 8:30 am, for 2 episodes of blood vomiting with bright red blood and traces of piceus blood, which occurred about an hour before in apparent full well-being. The child is suffering from pervasive developmental disorder; she had previously been hospitalized for accidental ingestion of medication, but parents, in this Emergency Room access, deny accidental or voluntary medication in the past 72 hours.

The girls's general condition immediately does not appear bright: the child is alert but has pale skin with cold ends and refill time of 3 seconds. Cardio-respiratory and abdominal objectivity are normal. Vital signs: body temperature 36°C, SaO_2 100% in ambient air, HR 106/min, blood pressure 93/66 mmHg. There are no lesions of the oral cavity or bleeding in other visible sites. Immediately and promptly, a peripheral venous access is placed, infusion of 0.9% saline is started and urgent blood chemistry tests are taken, from which a state of anemia emerges (Hb 10.3 g/dl, red blood cells 3,880,000/mm^3, MCV 76.4 fl) and mild metabolic acidosis (pH 7,314, pCO_2 37.3 mmHg, HCO_3 18.3 mmol/l, BE -7.7 mmol/l); the remaining laboratory tests and abdomen ultrasound are normal. At 10:55 am, an evacuation of piceo-colored feces is reported. In the suspicion of a hemorrhage of the upper digestive tract, therapy with esomeprazole is started and the pediatric surgeon is contacted, who recommends hospitalization for observation. Considering the progressive anemization of the patient (Hb 9.2 g/dl), during hospitalization, esophagogastroduodenoscopy is performed urgently, with visualization of caffean gastric stagnation, mixed with clots and bright red blood. In the antral area there is an ulcerative lesion with a bottom covered with fibrin and visible vessel. First, it was attempted an injective hemostasis with

saline solution, without success; then we proceeded with mechanical technique with the positioning of 3 hemoclips, finally obtaining a stable hemostasis. Only the following day the mother reported that she had found, in her home, a blister of ibuprofen, with several missing tablet, thus making the accidental ingestione of NSAID's palusible. Hematemesis is in most cases attributable to bleeding of the upper digestive tract, an event that in pediatric age is uncommon and is mainly attributable to esophageal varices and non-varicose causes. The etiology varies according to age [1]. Although most bleeding in children is not hemodynamically significant, the first approach involves patient stabilization. Endoscopy is indicated within the first 24 hours in case of stable patients [2] and represents, in addition to a diagnostic method, also the first line of treatment of high digestive bleeding, through several possible methods: injection of vasoactive substances (adrenaline), thermocoagulation and mechanical hemostasis (ligation, clips), among which the latter is particularly effective and lasting [3].

REFERENCES

[1] Romano C, Oliva S, Martellossi S, Miele E, Arrigo S, Graziani MG, Cardile S, Gaiani F, de'Angelis GL, Torroni F. Pediatric gastrointestinal bleeding: Perspectives from the Italian Society of Pediatric Gastroenterology. World J Gastroenterol. 2017;23(8):1328-37.

[2] Owensby S, Taylor K, Wilkins T. Diagnosis and management of upper gastrointestinal bleeding in children. J Am Board Fam Med. 2015;28(1):134-45.

[3] Sur LM, Armat I, Sur G, Tisa IB, Bordea MA, Lupan I, Samasca G, Lazar C. Practical Aspects of Upper Gastrointestinal Bleeding in Children. J Clin Med. 2023;12(8):2921.

ABS 51

A GASTROENTERITIS THAT RETURNS

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INTRODUCTION

The food protein-induced enterocolitis syndrome (FPIES) is a food allergy, not IgE mediated, to mainly gastrointestinal expression typical of the infant. Unlike most food allergies, it does not manifest itself immediately after taking the meal, and can also begin after a few hours. Any food can cause FPIES, but the most frequent are milk, soy, rice, meat, eggs and fish [1].

CASE REPORT

We report the case of a 6-month-old infant who came to our Emergency Room for repeated vomiting (about 6 episodes in the last hour). His older brother had gastrointestinal symptoms. On clinical evaluation, the child appeared pale, hyporeactive and dehydrated. The tests showed mild neutrophil leukocytosis, with negative phlogosis indices; Multistix® urine test strips showed presence of ketones; blood gas analysis: pH 7.31, HCO_3 16 mmol/L, BE -10.3, Lat 3.6, Na 136, K 7.7, glycemia 122. Intravenous rehydration with sodium chloride 0.9% was administered. After about 2 hours, he presented 2 evacuations of blood-streaked liquid stools. After 4 hours, he resumed feeding, with no vomiting nor diarrhea, so he was discharged with a diagnosis of gastroenteritis. Ten days later, symptoms reappeared, for which he was admitted. From history, it emerged the introduction of infant formula about 15 days before. Hospitalization and tests: blood, urine and feces cultures (including Adenovirus and Rotavirus research), abdominal ultrasound and skin prick test, all negative; fecal occult blood (FOB), positive. The clinical picture (vomiting and bloody diarrhea), medical history (onset of symptoms from the introduction of infant formula) and laboratory results (FOB positive) already pointed to the diagnostic suspicion of FPIES during the first day of hospitalization. It was decided to start a protein-free milk diet, favoring breastfeeding. In the following days, the infant did not have any more episodes of vomiting/diarrhea and fed regularly. He was discharged on the seventh day in good condition, with indications for follow-up.

CONCLUSION

The presentation of FPIES is often non-specific, and this can lead to diagnostic delay. The clinical picture may mimic a common gastroenteritis, acute abdomen, sepsis or inflammatory bowel disease. On the other hand, the history, the negativity of the prick tests, but especially the timing of presentation of the symptoms, associated with the positivity of FOB, are fundamental to address the diagnostic suspicion. Rehydration is the cornerstone of treatment in the acute phase, while the subtraction of triggering foods is the essential tool for the treatment of FPIES.

REFERENCE

[1] Calvani M, Anania C, Bianchi A, D'Auria E, Cardinale F, Votto M, Martelli A, Tosca M, Chiappini E, Brambilla I, Miraglia Del Giudice M, Caffarelli C. Update on Food protein-induced enterocolitis syndrome (FPIES). Acta Biomed. 2021;92(Suppl 7):e2021518.

ABS 52

WHEN A SEDATION CAN CHANGE LIFE. AN INCIDENTAL NEUROBLASTOMA

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CASE REPORT

We describe the case of an 11-month-old girl brought to the Emergency Department (ED) due to suspected ingestion of a foreign body (rubber balloon). Following the urgently performed esophagogastroduodenoscopy, which yielded negative results for foreign bodies, the patient was admitted to the ward.

Upon arrival, she still exhibited mild drowsiness post-sedation, with normal vital signs. Deep palpation of the abdomen revealed a mass on the left side, with a hard consistency and limited mobility in the deep planes, which was no longer appreciable in re-evaluations on the day after.

Therefore, an abdominal ultrasound was performed, confirming the suspicion of a new growth (measuring 6.5 x 6 x 4.5 cm), with a solid echogenic structure. Laboratory tests showed only neutrophilic leukocytosis, with a negative C-reactive protein result and a slight increase in creatine phosphokinase. Urinary catecholamine levels, including vanilmandelic acid and homovanillic acid, were elevated above normal, as were the levels of neuron-specific enolase and alpha-fetoprotein. Further diagnostic investigation with abdominal MRI revealed "an oval-shaped retroperitoneal formation in the mesogastric region, superficially surrounding the vertebral bodies, completely encircling the aorta with displacement of the inferior vena cava". A biopsy confirmed the suspected diagnosis of neuroblastoma, leading to the transfer of the child to the Pediatric Hematology-Oncology Unit. Neuroblastoma is a malignant neuroendocrine tumor, typical of the pediatric age group, originating from neural crest cells. It is the most common tumor of early childhood (comprising approximately 7-10% of tumors in children aged 0 to 5 years) and the most frequent extracranial solid tumor in the pediatric population. The average age at diagnosis is 18 months. Most cases of neuroblastoma are sporadic, but sometimes there is an association with chromosomal abnormalities in germ cells, hereditary predisposition, and congenital anomalies.

Clinical presentation varies widely depending on the patient's age and stage of the disease. Symptoms vary according to the affected site and the progressive involvement of adjacent organs. Some children with localized tumors have a favorable prognosis, while others with more advanced disease may face a more challenging outlook [1]. The diagnosis of neuroblastoma in its early stage, especially in asymptomatic children, may be associated with a good prognosis. Incidentally diagnosed cases of neuroblastoma (incidentalomas) in the pediatric age group are relatively rare and tend to have a more favorable prognosis compared to cases associated with clinical manifestations [2].

In our case, the general sedation the child underwent, allowed for a deeper and more comfortable abdominal palpation, enabling an early diagnosis of the tumor before clinical symptoms became evident.

REFERENCES

[1] Swift CC, Eklund MJ, Kraveka JM, Alazraki AL. Updates in Diagnosis, Management, and Treatment of Neuroblastoma. Radiographics. 2018;38(2):566-80.

[2] Armata J, Dłuniewska A, Garus K, Balwierz W, Mieyński W, Hnatko-Kołacz M. Incidental neuroblastoma. Pediatr Hematol Oncol. 2000;17(8):673-8.

ABS 53

AN UNUSUAL SWELLING

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CASE REPORT

An 8-year-old child presented to the Pediatric Emergency and Urgency Care Unit to perform tests for a lesion in right thigh and homolateral inguinal region, which had a hard consistency and a nodular appearance that was about 10 cm long in the previous 2 weeks and was covered by intact skin.

The ultrasound performed a few days before showed a gross nodal lesion extended from the proximal region of the right lower limb to the right iliac fossa. The child was in good general condition upon her arrival, with no signs of neurological involvement. Cardio-thoraco abdominal objectivity was normal, and the joints were unharmed. There were no signs of trauma. Medical history: no noteworthy

pathologies. Considering the clinical picture and the investigations already conducted, it was decided to repeat a complete abdominal ultrasound, which was likely to result in a probable arteriovenous (AV) malformation of the femoral artery/vein, and hematochemical examinations, obtained in the norm except for an increase in D-dimer (964 ng/ml; normal values between 0 and 500). The child was hospitalized in the Pediatric Surgery Unit of ARNAS Brotzu (Cagliari, Italy), where lower right and abdomen CT was performed. The examination revealed a potential occlusion of the external iliac vein and the possibility of lymphoproliferative nature of the neoformation; to exclude a sarcoma hypothesis, she was transferred to the Oncohematology Department, where she performed a full body MRI and a CT biopsy to complete the diagnostic assessment. The child was discharged after receiving prophylaxis with enoxaparin sodium 1,500 mg/day for suspicion of occlusion of the external iliac vein found on the CT.

The case was then entrusted to the colleagues of the Pediatric Surgery Unit of a third level center where, based on the clinical history and the investigations carried out, it was suspected that a venous lymphatic malformation caused bleeding and thrombosis after an acute episode. Because of the difficulty of the approach due to the location of the lesion, a conservative approach will be taken, and surgical approach with scleroembolization will only be performed if there are complications.

AV malformations are a type of vascular malformation that occurs due to developmental defects in the arterial and venous vasculature, without endothelial cell hyperplasia. AV malformations are infrequent, underdiagnosed and cause significant morbidity over the lifetime [1]. While AV malformations can affect any organ, the head and neck are the most frequently affected, with intracranial AV malformations being more frequent than extracranial AV malformations [2-6]. Next in frequency are AV malformations of the extremities, which have an equal distribution between the upper and lower extremities [7]. In addition, there have been reports of AV malformations in the trunk and viscera of the chest and abdomen [8, 9].

Noninvasive imaging (i.e., Doppler ultrasonography, computed tomography, or magnetic resonance imaging) in association with clinical findings are critical in establishing the diagnosis, evaluating the extent of the malformation, and planning appropriate treatment; proper treatment is considered, today, nidus ablation. Conservative treatment is recommended, but if a patient has clinical complications (e.g., ulceration, pain, hemorrhage, cardiac failure, or unacceptable cosmetic consequences), a multidisciplinary approach and a probable nidus sclerotherapy must be performed [10].

REFERENCES

[1] Tasnádi G. Epidemiology and etiology of congenital vascular malformations. Semin Vasc Surg. 1993;6(4):200-3.

[2] Lee JW, Chung HY. Vascular anomalies of the head and neck: current overview. Arch Craniofac Surg. 2018;19(4):243-7.

[3] Kobayashi K, Nakao K, Kishishita S, Tamaruya N, Monobe H, Saito K, Kihara A. Vascular malformations of the head and neck. Auris Nasus Larynx. 2013;40(1):89-92.

[4] Uller W, Alomari AI, Richter GT. Arteriovenous malformations. Semin Pediatr Surg. 2014;23(4):203-7.

[5] Ros de San Pedro J, Cuartero Pérez B, Ferri ÑÍguez B, Villanueva San Vicente V. Arteriovenous Malformations of the Temporalis Muscle: A Comprehensive Review. Oper Neurosurg (Hagerstown). 2018;14(4):325-40.

[6] Mulliken JB, Fishman SJ, Burrows PE. Vascular anomalies. Curr Probl Surg. 2000;37(8):517-84.

[7] Rosen RJ, Nassiri N, Drury JE, Interventional management of high-flow vascular malformations. Tech Vasc Interv Radiol. 2013;16(1):22.

[8] Miyasako Y, Kuwai T, Ishaq S. Case of a small intestinal arteriovenous malformation diagnosed by double-balloon enteroscopy preoperatively. Dig Endosc. 2019;31(3):e68.

[9] Gawecki F, Strangeways T, Amin A, Perks J, McKernan H, Thurainatnam S, Rizvi A, Jackson JE, Santhirapala V, Myers J, Brown J, Howard LSGE, Tighe HC, Shovlin CL. Exercise capacity reflects airflow limitation rather than hypoxaemia in patients with pulmonary arteriovenous malformations. QJM. 2019;112(5):335-42.

[10] Hyodoh H, Hori M, Akiba H, Tamakawa M, Hyodoh K, Hareyama M. Peripheral vascular malformations: imaging, treatment approaches, and therapeutic issues. Radiographics. 2005;25(Suppl 1):S159-71.

ABS 54

ACUTE URTICARIA IN PEDIATRIC EMERGENCY ROOM: WHAT CAUSES?

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INTRODUCTION

Urticaria is a frequent reason for access to pediatric first aid. The causes of acute urticaria in children are many, including: allergies to food or drugs, infections, physical factors, insect or idiopathic bites. The purpose of our study was to assess the etiology of acute urticaria in children who show up in the Emergency Room (ER).

MATERIALS AND METHODS
We conducted a retrospective study on ER cards and medical records of children hospitalized in ER with acute urticaria during a period of 8 months.
RESULTS
125 children were included, average age 7.15 years (0-13 years). In 52% of cases, urticaria was classified as idiopathic. The most frequent etiological factors were infections (30%), food (15%), drugs (10%) and insect bites (6%). After discharge, among the 30 patients sent for an allergy visit with suspicion of urticaria from allergy to drugs or food, 70% were allergic (17% of 125 children).
CONCLUSIONS
These data suggest that allergy is not the main trigger of acute urticaria in children who show up at the ER. Therefore, a detailed history, together with the objective examination, allows to screen patients with high probability of diagnosis for allergy, deserving of an allergy assessment.

ABS 55

WHEN THE FIRST DIAGNOSIS IS NOT ENOUGH: A STRANGE PEDIATRIC BRAIN TUMOR ONSET

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INTRODUCTION
Acute abdominal pain is a common complaint in children. The care of these patients is challenging for clinicians.
CASE REPORT
We describe a case of a 16-year-old boy who presented to our Emergency Department with a 3-day fever abdominal pain and vomiting, without diarrhea or headache. Physical examination on admission was unremarkable, except for fever and slight tenderness on palpation of the abdomen. Blood examinations didn't show an increase in systemic inflammation index. An abdominal ultrasound revealed the presence of enlarged lymph nodes and mesenteric inflammation, no wall thickening of the terminal ileum. After a surgical consultation, the diagnosis was directed towards a mesenteric lymphadenitis. Thus, the patient was hospitalized for deterioration of clinical conditions. In the following hours, the boy presented a lipothymia and alteration of the emotional state, reduced desire to communicate, maintaining an oriented language. With the suspicious of encephalitis, electroencephalography was performed, which showed the presence of irritative elements. The CT scan, performed urgently, brought to light a tetraventricular hypertensive hydrocephalus and a voluminous solid expansive lesion (diameter 4 x 5 cm), mass effect on the midbrain and midline shift. Brain MRI completed the study of the lesion. A ventricular shunt was placed in emergency with a simultaneous biopsy of the tumor, which revealed a high-grade glioma (grade IV according to WHO). Once stable clinical conditions were reached, the patient was transferred to the Pediatric Neurosurgery Center of the Gemelli Hospital in Rome.
DISCUSSION
Glioblastoma is a highly aggressive and malignant brain tumor, extremely rare in the pediatric population, commonly seen in older children [1]. Acute abdominal pain, in reverse, is a common complaint in children that can have a wide range of causes, from benign to more serious medical issues. The age of the child, physical examination, laboratory tests and imaging can help focus the differential diagnosis [2]. In our case, symptoms and ultrasound results pointed towards the diagnosis of mesenteric lymphadenitis. Mesenteric lymphadenitis in children is a not rare cause of acute abdominal pain, often caused by viral infections, and is a self-limiting condition. Differential diagnosis is crucial to rule out other conditions, such as appendicitis [3]. However, it is possible for a child to have an underlying gastrointestinal condition, unrelated to other emerging diagnoses. Abdominal pain and a brain tumor are typically unrelated symptoms, as they involve different parts of the body and have different underlying causes.
CONCLUSIONS
Fortunately, in our case, the boy was hospitalized due to deterioration of his general conditions and a careful clinical observation allowed us to highlight subtle signs of neurological alteration. After only 16 hours of hospitalization, a new unexpected diagnosis of hypertension hydrocephalus due to a brain tumor was made, which allowed a timely therapeutic approach.
REFERENCES
[1] Thomas DL. 2021 updates to the World Health Organization classification of adult-type and pediatric-type diffuse gliomas: a clinical practice review. Chin Clin Oncol. 2023;12(1):7.

[2] Raymond M, Marsicovetere P, DeShaney K. Diagnosing and managing acute abdominal pain in children. JAAPA. 2022;35(1):16-20.

[3] Benetti C, Conficconi E, Hamitaga F, Wyttenbach M, Lava SAG, Milani GP, Bianchetti MG, Simonetti GD, Helbling R. Course of acute nonspecific mesenteric lymphadenitis: single-center experience. Eur J Pediatr. 2018;177(2):243-6.

ABS 56

EATING DISORDERS IN CHILDREN AND ADOLESCENTS: INTEGRATED CLINICAL MANAGEMENT EXPERIENCE IN THE PROVINCE OF ROVIGO, A SINGLE CENTER STUDY

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INTRODUCTION

Eating disorders (EDs), including anorexia nervosa, bulimia, and binge-eating disorder, are a prevalent public health issue. Since 2018, national guidelines on ED have been issued by the Italian Ministry of Health, the so-called "Lilac path", for the admission, triage and evaluation of patients suffering from ED. EDs have a particular impact on both the physical and psychological development of children. During COVID-19 pandemic we had an increase of complex cases, and a lower age of onset of these disorders (prevalence of 6% at age < 12 years). We show the results of our multidisciplinary activity for ED patients.

MATERIALS, METHODS AND RESULTS

In order to treat children with EDs and to apply the national guidelines, we developed a specific program, so that a multifactorial and multidisciplinary assessment for these patients from outpatient clinic to hospital admission has been possible. Since treatment must address important nutritional, physical and mental comorbidities, a multidisciplinary team has been needed. The team, which brought together professionals of different services, included the following: neuropsychiatrist, psychologist, pediatrician, nutritionist and dietitian. Medical complications of ED may facilitate the first contact with health professionals and treatment initiation. We have 2 hospital beds for the management of malnutrition symptoms. During the admission, dietitians and nutritionists are involved, as well, to plan the assisted meals and to find the best approach for managing the anxiety that meal causes.

Patients are discharged when clinical status and meal management improve, and they may be handled at an outpatient clinic.

The outpatient path includes: 3 meetings for assessment; 3 meetings for psycho-nutrition with the family, and about 10 meetings for Enhanced Cognitive Behavioral Therapy with the patients, neuropsychiatric check-ups, and appointments with nutritionist and dietitian.

Somatic factors are part of care, but weight is not the only goal of improvement. The Children's Global Assessment Scale (C-GAS) is measured at the beginning of treatment and after 6 months and helps us to monitor adaptive changes. An increase of 20 points in C-GAS has been detected.

In the first semester of 2023, we treated 27 children: 25 females and 2 males, with age from 9 to 17 years. Twelve cases were new onsets. Admission to hospital in 8 cases, 3 after discharge had to be treated at specialized centers. In 13 cases, neuroleptic and/or antidepressant therapy was started.

CONCLUSIONS

EDs are an exemplary summary of the interdependence between body and mind, and it is therefore essential to measure the results in terms of both weight recovery and social functioning. Tracking the BMI during the recovery and the C-GAS has proved, in our experience, that early and multidisciplinary treatments with an hospital-territory continuity can improve clinical parameters.

REFERENCES

• Auger N, Steiger H, Luu TM, Chadi N, Low N, Bilodeau-Bertrand M, Healy-Profitos J, Ayoub A, Brousseau E, Israel M. Shifting age of child eating disorder hospitalizations during the COVID-19 pandemic. J Child Psychol Psychiatry. 2023;64(8):1176-84.

• Le Grange D, Eckhardt S, Dalle Grave R, Crosby RD, Peterson CB, Keery H, Lesser J, Martell C. Enhanced cognitive-behavior therapy and family-based treatment for adolescents with an eating disorder. Psychol Med. 2020;52(13):1-11.

• Ruocco G, Giacomini D, Romano G; Italian Ministry of Health. [Admission, triage, evaluation and treatment of patients with eating disorders – "The lilac path in emergency"]. [Article in Italian]. Available at: https://www.salute.gov.it/imgs/C_17_pubblicazioni_2961_allegato.pdf, date of publication: 2020, last access: September 2023.

ABS 57

ATYPICAL PLEOMORPHIC ADENOMA: A RARE CASE IN A CHILD

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INTRODUCTION

Pleomorphic adenoma (PA) accounts for 60-70% of all salivary glands tumors, in most cases occurring in parotid ones. Typically, it often shows up in people between 30 and 60 years, with a female increased predilection, occurring rarely also children [1]. Clinically, parotid PAs present as an irregular, nodular and slow-growing preauricular mass, whose symptoms mainly depend on size and location. Indeed, if not treated, they could result in a giant size, even compromising nearby tissues, such us facial nerve. Histologically, PA appears epithelial, myoepithelial and mesenchymal proliferation in a chondromyxoid matrix, surrounded by a pseudo-capsule with occasional metaplasia features [2, 3]. Surgical excision represents the mainstay of treatment [3].

CASE REPORT

We report the case of an 11-year-old patient with a 2.5 cm indolent and mobile parotid mass, whose radiological features were a central cystic-colliquated area carrying rim and intralesional in-homogeneous enhancement, and a possible capsule interruption. The whitish mass was circumscribed by a fibrous pseudocapsule with variable thickness and it was characterized by a proliferation of epithelial/myoepithelial cells in continuity with myxoid material, mainly found on the periphery of the nodule; centrally, connective and hyaline features were seen. Frequent keratinizing squamous metaplasia, calcifications, occasional chondroid metaplasia, and occasional aspects of adipose meta-plasia were also observed. In our case, epithelial/myoepithelial cells exhibited focal nuclear atypia with slightly pleomorphic nuclei, sometimes with a prominent nucleolus.

CONCLUSIONS

PAs are benign salivary gland tumors with the potential to grow to a larger size if left untreated. Therefore, early detection is crucial, not only to prevent malignant transformation, which can occur approximately in 6% of PAs, but also to avoid involvement of nearby anatomical structures. Complete excision of the tumor is the gold-standard treatment. A long-term follow-up is necessary to reduce the high recurrence rate, even after removal.

REFERENCES

[1] AlKindi M, Ramalingam S, Hakeem LA, AlSheddi MA. Giant Parotid Pleomorphic Adenoma with Atypical Histological Presentation and Long-Term Recurrence-Free Follow-Up after Surgery: A Case Report and Review of the Literature. Case Rep Dent. 2020;2020:8828775.

[2] Ethunandan M, Witton R, Hoffman G, Spedding A, Brennan PA. Atypical features in pleomorphic adenoma – a clinicopathologic study and implications for management. Int J Oral Maxillofac Surg. 2006;35(7):608-12.

[3] Valstar MH, Mast H, Ten Hove I, Moonen LR, Balm AJ, Smeele LE, Koljenović S, Dinjens WN, van Velthuysen MF. Malignant transformation of salivary gland pleomorphic adenoma: proof of principle. J Pathol Clin Res. 2021;7(5):432-7.

ABS 58

EXPERIENCES OF TELEMEDICINE: A RELYING SOLUTION TO THE PANDEMIC CHALLENGES IN PROVIDING HEALTHCARE SERVICES AND COMMUNICATION

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INTRODUCTION

The COVID-19 pandemic changed the models of healthcare delivery; in particular, children with medical complexity (CMC), and their family caregivers (FC), have been affected by these changes and the pandemic itself, with psychological distress. During the pandemic, to reduce virus exposure, telemedicine has proven to be an accepted tool by FC and healthcare providers (HP) for the management of CMC patients.

AIMS OF THE STUDY

We aim to analyse the impact of telemedicine on CMC patients and FC and to compare different experiences on telemedicine in different backgrounds.

RESULTS

We previously studied a telematic approach with the training of the FC for intermittent catheterization, trans-anal irrigation and applications for cystostomy buttons, and the sharing via mail of voiding charts to evaluate adherence to the procedures in patients affected by neurogenic bladder and neural tube defects. As result, telemedicine has helped both physical health, with equal number

of hospitalization and complications as before the pandemic, and psychological wellness, with FC perception of telemedicine to be a reliable source of assistance [1]. Telemedicine has proved to be a viable way to manage 21 CMC ventilated patients, by reducing hospital admission to 1 every 6 months, with only 1 problem out of 12 not fixed by remote assistance [2]. Another group studied the influence of the pandemic on FC experiences by collecting 12 FC interviews that highlight 3 common thematics: negative and positive aspects of the delivery of health services, the impact of the pandemic on physical and psychological wellness, and the common challenges during the pandemic. Despite the fact that they experienced inadequate access to healthcare services, financial problems and the necessity of balancing multiple roles, they praised the role of telemedicine with the reduction of physical burdens, financial expenses and risk of contamination [3].

CONCLUSION

The pandemic has strained FC, CMC and HP, with multiple challenges in providing healthcare services and communication, in groups already susceptible to mental health problems. However, telemedicine has proved to mitigate these issues in multiple clinical and social settings, and to meet the psychological needs of FC.

REFERENCES

[1] Masnata G, Dessì S, Sanna G, Corona L. Telemedicine in pediatric and adult patients with neurogenic bladder during the COVID-19 pandemic: to make a virtue out of necessity. In: Selected Abstracts of the 17th International Workshop on Neonatology and Pediatrics, Virtual Edition; Cagliari (Italy); October 28-29, 2021. J Pediatr Neonat Individual Med. 2021;10(2):e100229.

[2] Onofri A, Pavone M, De Santis S, Verrillo E, Caggiano S, Ullmann N, Cutrera R. Telemedicine in children with medical complexity on home ventilation during the COVID-19 pandemic. Pediatr Pulmonol. 2021;56(6):1395-400.

[3] Pitch N, Davidson L, Mekhuri S, Patel R, Patel S, Ambreen M, Amin R. Exploring the experience of family caregivers of children with medical complexity during COVID-19: a qualitative study. BMC Pediatr. 2023;23(1):160.

ABS 59

PSYCHOLOGICAL DIFFICULTIES EXPERIENCED BY SUBJECTS SUFFERING FROM TYPE 1 DIABETES MELLITUS

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Subjects suffering from type 1 diabetes mellitus (T1DM) can experience serious psychological and psychopathological issues directly deriving from their conditions. In these terms, the current state of the art highlights some main pathological domains, as in the case of alexithymia, affective psychopathology and maladaptive defences. In this case, the need to implement knowledge and interventions in the field of chronic conditions is clear. In our case, the commitment is centred in the emergence of affective dynamics typically emerging in children, adolescents and adults suffering from T1DM. Preliminary data obtained through the use of standardized and validated instruments on a group of 105 subjects (aged between 11- and 17-year-old; mean age: 13.88, standard deviation: 2.16; female prevalence: 60%) highlighted how consciousness about emotions and feelings of subjects represents an issue known as alexithymia. This condition appears to reach "borderline" levels in the examined subjects, leading to affective disturbances as anxiety, depression and somatization. Along with these results, uncertainty about clinical issues and alexithymia appeared to be correlated. The directions assumed by phenomena suggest the need for greater attention in diagnostic and therapeutical terms.

ABS 60

THE CHRONICITY IN CHILDHOOD AND ADOLESCENCE: NEW PERSPECTIVES OF TAKING CHARGE

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The present contribution focuses on the importance of integrated care of the child and the adolescent in chronic conditions. Chronicity represents a specific condition in which a disease does not reach recovery, but it reaches a condition with less serious aspects, which are extended over time. It is, therefore, a state of persistent disease that presents sharp phases and phases of quiescence. This is a condition of developmental risk that constitutes a real threat

to the health of the developmental trajectory of the child/adolescent, with possible outcomes with regard to all the developmental dimensions of the child/adolescent, also involving the family.

This contribution aims to be a promoter of integrated work between pediatricians and psychologists to safeguard the health and the well-being of the minor and all the aspects that define the pediatric condition, with the aim of improving the taking charge action in the accompaniment and in integrated support for children/adolescents/families who are hospitalized in the Pediatrics Unit and have undergone invasive procedures, through the definition of general criteria of responsibility and the operational path.

The integrated intervention moves in line with studies in the field's literature [1, 2] and it arises and develops from a precise observation and a careful analysis of the demand of the Pediatric Hospital contexts in which the intervention takes place.

The objective of the intervention is to improve the adherence and compliance to procedures and treatments of children/adolescents with chronic illness conditions, implementing an integrated approach between healthcare professionals and the psychologist. An intervention oriented in this way allows a better understanding of the diagnosis and treatment management by the users; it promotes less rigid mental constructs and stereotypes, with the advantage of greater flexibility; it redefines self-understanding and self-representation, mentalizing the experience of illness as part of oneself; and still, an integrated intervention allows to strengthen and build bonds. The experience of illness becomes, in this sense, "thinkable", to the extent that healthy and sick parts are integrated, building new resources and coping strategies in all family members [3]. Finally, the indirect user of the proposed integrated intervention in Pediatrics will be the healthcare system, in terms of possible positive effects on cure and care and in the approach to patients and their families.

REFERENCES

[1] Kazak AE, Noll RB. The integration of psychology in pediatric oncology research and practice: collaboration to improve care and outcomes for children and families. Am Psychol. 2015;70(2):146-58.

[2] Costello LH, Suh C, Burnett B, Kelsay K, Bunik M, Talmi A. Addressing Adolescent Depression in Primary Care: Building Capacity Through Psychologist and Pediatrician Partnership. J Clin Psychol Med Settings. 2021;28(1): 53-66.

[3] Fairfax A, Brehaut J, Colman I, Sikora L, Kazakova A, Chakraborty P, Potter BK; Canadian Inherited Metabolic Diseases Research Network. A systematic review of the association between coping strategies and quality of life among caregivers of children with chronic illness and/or disability. BMC Pediatr. 2019;19(1):215.

ABS 61

A *BARTONELLA HENSELAE* INFECTION, COMPLICATED BY HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, MIMICS SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS ONSET IN A 4-YEAR-OLD CHILD

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INTRODUCTION

Systemic Bartonellosis is a rare cause of fever of unknown origin and sometimes the diagnosis could be difficult. In case of rash and arthritis, it may be misdiagnosed as systemic juvenile idiopathic arthritis (JIA) at onset [1]. We describe a case of *Bartonella henselae* infection complicated by hemophagocytic lymphohistiocytosis (HLH).

CASE REPORT

D. is a 4-year-old child, admitted to our hospital for suspected JIA. He presented persistent and continuous-remitting fever for 14 days, evanescent skin rash, arthralgias and arthritis of the knees. Laboratory tests showed significant increase in inflammatory indices, while other exams (autoimmunity and infectious) were normal. Given the contact with domestic cats, a zoonosis panel was performed and a positive result for anti-*Bartonella* IgM (1:20) and IgG (1:80) was showed. Bone marrow aspiration, done to exclude lymphoproliferative disease or presence of histio-hemophagocytes figures, was negative, but on bone marrow blood, PCR for *Bartonella*-DNA was positive. This data allowed us to confirm the diagnosis of systemic Bartonellosis. However, the start of antibiotic therapy with azithromycin and rifampicin resulted in a worsening of the fever pattern and hyperferritinemia. In the suspicion of a condition of bacterial lysis due to antibiotic therapy, or in the hypothesis of an evolution in an initial HLH, steroid bolus therapy (methylprednisolone intravenous and subsequently prednisone *per os*) was introduced in association with antibiotic therapy, with prompt

defervescence of fever. After 10 days of treatment and initial clinical improvement, the steroid tapering was started. However, in a few days, the fever resumed. It was first attributed to Adenovirus overinfection, and then to poorly controlled Bartonellosis. Despite the addition of therapy with trimethoprim/cotrimoxazole and then doxycycline, the general condition gradually worsened to a full-blown picture of severe HLH, characterized by continuous fever, pancytopenia, collapse of the erythrocyte sedimentation rate, increase in ferritin and triglycerides, impaired liver function with hypertransaminasemia, hypoalbuminemia, coagulopathy, with the need for transfusions of red blood cells and infusions of albumin and plasma. Boluses of methylprednisolone were then reintroduced, but without benefit. Therefore, it was started an off-label therapy with anti-IL1 (anakinra) at 8 mg/kg/day, which however did not lead to the expected clinical improvement. Considering the incipient multiple organ failure, D. was transferred to a highly specialized Pediatric Intensive Care Unit, where he continued with ongoing therapy at higher doses and intensive care, until clinical improvement.

CONCLUSIONS

JIA is a diagnosis of exclusion, that requires a differential diagnostic approach, which allows to exclude infectious and/or proliferative pathologies, which, due to common symptoms, like fever, arthritis/arthralgias, have always to be considered.

REFERENCE

[1] Hayem F, Chacar S, Hayem G. Bartonella henselae infection mimicking systemic onset juvenile chronic arthritis in a 2 1/2-year-old girl. J Rheumatol. 1996;23(7):1263-5.

ABS 62

PARENTING STYLE IN ADOLESCENT RESTRICTIVE ANOREXIA NERVOSA

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INTRODUCTION

Anorexia nervosa (AN) is a complex disorder with multifactorial pathogenesis, associated with high psychiatric comorbidity, mainly represented by internalizing disorders.

AIMS

The study aims to evaluate the presence of a typical parental profile in a sample of adolescent subjects with AN aged between 11 and 17 years.

METHODS

18 subjects with AN were recruited if age was at least 11 years and IQ was between 85 and 115 points.

The Parents Preference Test (PPT) was used to assess and evaluate the parenting styles. The final score determines the parenting style based on 4 dimensions:

- Attentional Focus (8 items): it describes the way in which the parent pays attention to the child and her needs during the interaction, if he is mainly concentrated on his own activities or those of his child;
- Experiential Mode (8 items): it investigates whether the parent focuses more on the emotional (emotional orientation) or rational (rational orientation) aspects of the relationship in the experience with the child;
- Behavior Regulation (8 items): it measures how much the management of the child's behavior is based on the characteristics of the situation and context or whether it is influenced by rules established *a priori* within the family;
- Energy: this dimension determines an active or passive profile, measuring how much the parent takes the initiative and is receptive in interacting with the child.

RESULTS

In recruited subjects, mean age was 13.61 years (SD 1.70), with clear prevalence of females (83%). The average body mass index (BMI) value was 14.68, and more than half of the subjects had a BMI ≤ 5th. 83% presented restrictive AN subtype, while 17% presented binge eating/purging behaviors. Moreover, 27.7% underwent pharmacological treatment with antipsychotic and/or antidepressant drugs (SSRIs). The comparison analysis between the categorical variables reported statistically significant differences in the Behavior Regulation dimension, with a prevalence of a rule-based parenting style greater in cases than in controls.

CONCLUSION

The present study confirms the presence in AN adolescents of an affective dimension characterized by deficient interpersonal skills, which cannot be interpreted only in the light of an anxious or depressive state. Furthermore, an overall active, emotional and flexible maternal parenting style was found, although more assertive and authoritarian compared to the control population.

ABS 63

PILOT STUDY ON PERSONALITY ASPECTS IN RESTRICTIVE ANOREXIA NERVOSA

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INTRODUCTION

Eating disorders (EDs) are constantly growing as pathological behaviors with a typical onset during adolescence and relevant negative impact on health and psychosocial functioning and on the whole parental and familiar system.

AIMS

Aim of this pilot case-control study is identifying a putative personality profile in patients diagnosed with restrictive anorexia nervosa (AN) and in their mothers.

METHODS

For this study, 20 subjects with restrictive AN and 28 mothers of AN subjects have been recruited. The average disease duration was at least 24 months; the evaluation was conducted with the MMPI-2 test and MMPI-A test for daughters. For the interpretation of the results, only the Basic and the Content scales were considered. The Basic scale is composed of 10 items: hypochondriasis (Hs), depression (D), hysteria (Hy), psychopathic deviation (Pd), masculinity/femininity (Mf), paranoia (Pa), psychasthenia (Pt), schizophrenia (Sc), hypomania (Ma), social introversion (Yes). The Content scale consists of 15 items: anxiety (ANX), fears (FRS), obsessiveness (OBS), depression (DEP), health problems (HEA), bizarre mind (BIZ), anger (ANG), cynicism (CYN), antisocial practices (ASP), type A (TPA), low self-esteem (LSE), social distress (SOD), family problems (FAM), work interference (WRK) and treatment resistance (TRT). To compare the two groups, the t-test or the Wilcoxon-Mann-Whitney test was used and Cohen's d was calculated to verify the effect size; p-values < 0.05 were considered significant.

RESULTS

The comparison between the group of mothers of patients with AN and those of the control group shows no significant differences for age, socioeconomic and educational level.

The statistical analysis highlights higher scores for the AN group in Hs (t-value[54] = 3.22; p = 0.002; d = 0.86), DEP (t-value[54] = 2.50; p = 0.016; d = 0.67), Hy (t-value[54] = 3.40; p = 0.001; d = 0.90), Pa (t-value[54] = 3.87; $p < 0.001$; d = 1.03), Si (t-value[54] = 3.76; $p < 0.001$; d = 1.00), ANX (t-value[54] = 2.64; p = 0.011; d = 0.70), HEA (t-value [54] = 3.16; p = 0.003; d = 0.84), with significant effect size. A different statistical test was carried out for the daughters as the normality for the distribution was violated. The statistical analysis revealed significant scores in Hs (p = 0.005), D (p = 0.001), Pd (p = 0.032), Pt (p = 0.003), Sc (p = 0.007), ANX (p = 0.008), OBS (p = 0.025), DEP (p = 0.005), HEA (p = 0.007), BIZ (p = 0.042), ANG (p = 0.022), LSE (p = 0.001), FAM (p = 0.008), TRT (p = 0.004).

CONCLUSIONS

The study highlighted the personality aspects of mothers and daughters with restrictive AN, finding different personality aspects, but with a strong correlation in certain subscales. Our results suggest the mandatory assessment of the personality of the mothers (and in general taking care of them) and not just of AN girls, because of the pathological family unit that can maintain the severity of the disorder.

ABS 64

SOCIAL PERCEPTION AND THEORY OF MIND AMONG ADOLESCENTS WITH ANOREXIA NERVOSA

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INTRODUCTION

Eating and nutrition disorders (EDs) in developmental age are increasingly widespread disorders. Anorexia nervosa (AN) is characterized by caloric restriction with body weight $< 5^{th}$ percentile, intense fear of gaining weight or becoming fat, and alteration of body weighting or shaping.

METHODS

47 females with restrictive AN, aged 8-16 years, have been recruited and evaluated with Neuropsychological Battery for Children, Second Edition (NEPSY-II). Exclusion criteria were: ED different from AN; medical condition not related to ED; neurological and/or psychiatric diseases.

Control (C) group consists of 44 adolescents matched for age, mother tongue and level of education. The aforementioned assessment was carried out after obtaining written informed consent from the legal guardian/parent and assent from AN subjects.

RESULTS

No significant differences were found between AN and C groups for age ($p = 0.552$), IQ ($p = 0.201$), but only for BMI ($p < 0.001$). Among AN subjects, with respect to the C group, NEPSY-II assessment reports lower scores in the following tests: Theory of Mind (ToM) (t-value[89] = -3.36; $p = 0.001$), emotion recognition (ER) (t-value[89] = -2.039; $p = 0.022$).

DISCUSSION AND CONCLUSION

Social perception is part of the cognitive process, and the delay in social relationships, social aversion, disinterest in social interactions and poor social skills are distinctive signs that characterize very specific clinical conditions. Specifically, in the AN group, it has been reported a low score in ToM domain for the part A that evaluates the ability to understand mental constructs (e.g., beliefs, intentions, deceptions, emotions, fantasy and pretense), as well as the ability to understand that others have their own thoughts, ideas, feelings, which may be different from ours. On the other hand, a low score was found also in the ER test, highlighting a poor ability to identify and decode facial emotional expressions. These difficulties are reflected in mutual relationships with the possibility of anxiety arising in social situations, suggesting the relevance in the development and maintenance of AN symptoms [1], linked to specific neural pathways dysfunctioning, such as the right temporoparietal junction [2]. The present prospective observational study may be considered original in the field of EDs, providing the need to take charge also of the neuropsychological evaluation of these subjects as part of clinical assessment.

REFERENCES

[1] Nalbant K, Kalaycı BM, Akdemir D, Akgül S, Kanbur N. Emotion regulation, emotion recognition, and empathy in adolescents with anorexia nervosa. Eat Weight Disord. 2019;24(5):825-34.

[2] McAdams CJ, Krawczyk DC. Impaired neural processing of social attribution in anorexia nervosa. Psychiatry Res. 2011;194(1):54-63.

ABS 65

DEVELOPMENTAL GOALS AND EDUCATIONAL STRATEGIES IN ITALIAN AND SUB-SAHARAN AFRICAN MIGRANT MOTHERS' NARRATIVES

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INTRODUCTION

In the host society, migrant parents face educational approaches that diverge from what they are used to expecting. Child education in Western countries is focused on the belief that the child is a single and self-determined agent, whose wills and wishes are of supreme importance. Most migrants to European societies have a low level of formal education and come from a culture where multigenerational families are the rule [1]. Their parental ethno-theories focus on action autonomy to develop a community-oriented behavior rather than psychological independence. Their relational model is hierarchical, and educational strategies are characterized by highly structured teaching schemes based on recurrence, amendment, and punishment [2].

AIMS

The present study wants to contribute to understanding the different educational goals and strategies between Italian and Sub-Saharan African migrant mothers to improve the well-being of their children and better support migrant mothers in their integration process.

METHODS

Trained psychologists interviewed 188 mothers with children between 2 and 6 years old, unlike in educational degree (40 low degrees, 48 medium degrees, 53 high degrees, 47 very high degrees) and ethnic background (106 Italian and 82 Sub-Saharan African [Senegal and Nigeria]; all living in Northern Italy). In the interviews, mothers were asked to talk about their beliefs, experiences, and expectations concerning everyday situations, such as eating, sleeping, playing, and learning. The interview data from mothers were analyzed by qualitative content analysis, resulting in two coding frames, one for each ethnic background. The coding categories were derived inductively from the interview responses. These coding frames were tested, revised, and checked for internal consistency.

RESULTS AND CONCLUSIONS

Results indicate that more educated Italian mothers are focused on autonomy-oriented developmental goals and a constructivist approach to education. On the contrary, migrant mothers and less-

educated Italian mothers are concentrated on community-oriented developmental goals and a didactic approach to education. They are also more concentrated on their babies' physical health. The divergences were most salient between Italian-graduated mothers and migrant mothers. Notwithstanding the significant correlation between parenting beliefs and socioeconomic status [3], other dimensions must also be considered to guarantee child well-being in migrant families: the parents' education and acculturation process. In our study, we focused on parents' education. In future studies, it will be essential to assess different generations of migrant families and analyze generational changes in developmental aims. Our results can contribute to broadening educational perspectives and practices so that the best interest of all children can be secured.

REFERENCES

[1] ISMU (Initiatives and Studies on Multiethnicity) Foundation. The Twenty-eight Italian Report on migrations. Milan: ISMU, 2022.

[2] Valtolina GG. [Physical punishment and the role of cultural context: A review]. [Article in Italian]. Maltrattamento Abuso Infanzia. 2013;15(1): 11-35.

[3] Emmen RA, Malda M, Mesman J, van Ijzendoorn MH, Prevoo MJ, Yeniad N. Socioeconomic status and parenting in ethnic minority families: testing a minority family stress model. J Fam Psychol. 2013;27(6):896-904.

ABS 66

EXTRA-UTERINE GROWTH RESTRICTION IN PRETERM INFANTS AND NEURODEVELOPMENTAL OUTCOMES: A SCOPING REVIEW

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INTRODUCTION

Extra-uterine growth restriction (EUGR) is a condition affecting premature infants, characterized by poor postnatal growth compared to their intrauterine growth potential. There is not currently unanimous agreement on the EUGR definition. There are two current definitions in the literature: "cross-sectional" and "longitudinal". According to the first definition, an infant can be considered growth-restricted if the weight at a specific time point is below a standardized cut-off. The latter definition indeed assesses differences in growth, measuring how the weight of the infant varies between two predefined time points. The incidence of EUGR varies according to the definition chosen [1]. Neurodevelopmental outcomes in premature babies can vary widely. Thus, early intervention and close monitoring are crucial for optimizing their neurodevelopmental outcome.

PURPOSE

To evaluate neurodevelopmental outcomes in EUGR children.

METHODS

A systematic search using PubMed was conducted, encompassing studies published up to September 2023. The search strategy included keywords related to EUGR and neurological outcomes, resulting in a total of 296 studies. Then, these studies were analyzed for data on neurological outcomes in EUGR patients.

RESULTS

In several studies, EUGR was associated with an increased risk of adverse neurodevelopmental outcomes. A number of studies reported a delay in the acquisition of language and motor milestones. Furthermore, in some studies, EUGR children tended to develop cognitive deficits and behavioral alterations [2]. There is not a clear association between EUGR and motor deficits. It was also highlighted the importance of nutrition to improve EUGR, leading to less neurological consequences.

DISCUSSION

The results of this review highlight possible adverse neurodevelopmental outcomes in EUGR infants. Those findings emphasize the importance of early identification of EUGR to reduce the impact of possible neurological manifestations and other adverse events. Indeed, close monitoring of growth, nutritional support and developmental assessment could have beneficial effects. However, the included studies used different parameters for the assessment of neurological outcomes as well as for the definition of EUGR in neonates. Moreover, in most studies the sample size was relatively small, and they had limited follow-up time points [3]. Therefore, it will be crucial to define specific diagnostic criteria and to conduct studies with large sample sizes and long-lasting follow-ups.

REFERENCES

[1] Fenton TR, Kim JH. A systematic review and meta-analysis to revise the Fenton growth chart for preterm infants. BMC Pediatr. 2013;13:59.

[2] Fenton TR, Cormack B, Goldberg D, Nasser R, Alshaikh B, Eliasziw M, Hay WW, Hoyos A, Anderson D, Bloomfield F, Griffin I, Embleton N, Rochow N, Taylor S, Senterre T, Schanler RJ, Elmrayed S, Groh-Wargo S, Adamkin D, Shah PS. "Extrauterine growth restriction" and "postnatal growth failure" are misnomers for preterm infants. J Perinatol. 2020;40(5): 704-14.

[3] De Rose DU, Cota F, Gallini F, Bottoni A, Fabrizio GC, Ricci D, Romeo DM, Mercuri E, Vento G, Maggio L. Extra-uterine growth restriction in preterm infants: Neurodevelopmental outcomes according to different definitions. Eur J Paediatr Neurol. 2021;33:135-45.

ABS 67

IS CEREBELLAR DAMAGE IN PREMATURE INFANTS INVOLVED IN LONG-TERM COGNITIVE, LEARNING, AND BEHAVIORAL DISABILITY?

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INTRODUCTION

The cerebellum is a structure of the central nervous system which normally undergoes its most important developmental stage in the last trimester of pregnancy and is highly susceptible to damage in premature infants. Thanks to the progress in diagnostic techniques, such as mastoid window in transfontanellar ultrasound and magnetic resonance imaging (MRI), many cerebellar abnormalities have been identified, including hemorrhages, ischemia, and hypotrophy. Cerebellar abnormalities are often accompanied by cerebral lesions, making it difficult to establish a direct association between cerebellar damage and outcomes [1]. The cerebellum is involved in motor control; moreover, it plays a role in cognition, learning and behavior.

PURPOSE

The purpose was assessing the impact of cerebellar lesions on neurodevelopment outcome and distinguish the effects of cerebellar damage from cerebral lesions in order to formulate a more accurate prognosis.

METHODS

A systematic search of PubMed was conducted, encompassing studies published in the last 5 years. The search strategy included keywords related to cerebellum and neurological outcomes.

RESULTS

Most studies focus primarily on assessing the impact of cerebellar hemorrhages, attempting to establish a correlation between the extent of the hemorrhages and their outcomes. They introduced a classification system: massive, limited and focal. Massive hemorrhages evolve in atrophy and less favorable outcomes, including cerebral palsy and impairment of language and cognition. Focal hemorrhages do not seem to cause atrophy and no neurodevelopmental abnormalities have been found at 2 years of age except hypotonia, hypertonia and abnormalities of osteotendinous reflexes [2]. Limited hemorrhages are still under investigation. Prognosis tends to improve from bilateral lesions to unilateral lesions with or without vermian involvement, which is associated with underdeveloped socialization skills and autism [3]. Reduced vermian volume is connected to cognitive impairment and cerebellar atrophy (reduced transverse cerebellar diameter), and reduced cerebellar volume of the lateral hemispheres has been associated with less executive functioning, visuospatial function, and language abnormalities.

DISCUSSION

It is essential to conduct a proper follow-up to address any neurological and behavioral abnormalities in infants with cerebellar damage. Most studies have been conducted with a small sample size and a short follow-up period, so one of the current challenges is to extend the follow-up until adolescence, to obtain reliable results regarding the impact of cerebellar lesions on neurodevelopment. That is important to ensure appropriate and tailored therapies.

REFERENCES

[1] Boswinkel V, Steggerda SJ, Fumagalli M, Parodi A, Ramenghi LA, Groenendaal F, Dudink J, Benders MN, Knol R, de Vries LS, van Wezel-Meijler G. The CHOPIn Study: a Multicenter Study on Cerebellar Hemorrhage and Outcome in Preterm Infants. Cerebellum. 2019;18(6): 989-98.

[2] Limperopoulos C, Bassan H, Gauvreau K, Robertson RL Jr, Sullivan NR, Benson CB, Avery L, Stewart J, Soul JS, Ringer SA, Volpe JJ, duPlessis AJ. Does cerebellar injury in premature infants contribute to the high prevalence of long-term cognitive, learning, and behavioral disability in survivors? Pediatrics. 2007;120(3):584-93.

[3] Brossard-Racine M, Limperopoulos C. Cerebellar injury in premature neonates: Imaging findings and relationship with outcome. Semin Perinatol. 2021;45(7):151470.

ABS 68

IMPACT OF MATERNAL DIET DURING AND AFTER PREGNANCY ON GROWTH AND DISTRIBUTION OF ADIPOSITY IN THE FIRST 12 MONTHS OF LIFE: PRELIMINARY DATA FROM THE EUROPEAN LIFE-MILCH PROJECT

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INTRODUCTION

The ongoing European LIFE-MILCH project (www.lifemilch.eu) focuses on detecting endocrine-disrupting chemicals in mothers, in breast and formula milk and in the urine of mothers and infants up to 12 months of age, studying relationships with neurodevelopment, growth, distribution of adiposity, pubertal stages, to establish a risk assessment model to prepare safety guidelines.

OBJECTIVE

We have evaluated the effects of maternal diet during and after pregnancy and of duration of breastfeeding on longitudinal growth, head circumference, body mass index (BMI), and skinfold thickness (SFT).

METHODS

The analyses were carried out on the 254/654 mother-infant dyads enrolled at one site. The mothers were enrolled at 36-40 weeks of gestation, all in good health. Pregnancies were uncomplicated. All women filled out questionnaires related to lifestyle and nutritional habits (daily and weekly frequency of intake of cereals, meat, eggs, fruit, vegetables, fatty food and dairy products) at recruitment (T0), 1 (T1), 3 (T2), and 6 months (T3) after delivery. The duration and type of feeding were registered for all babies. In infants, anthropometric measurements were taken at birth, T1, T2, T3 and 12 months (T4); bicipital, tricipital, subscapular and supra-iliac SFT were evaluated at T3 and T4. The series included 125 males and 129 females.

RESULTS

After delivery, mothers ate significantly fewer vegetables and fruit ($p < 0.001$) and cereals ($p < 0.001$), and were taking more fat food ($p < 0.001$), whereas meat intake was unchanged and intake of dairy products was variable. The intake of cereals started to decrease at T2 only for mothers who stopped exclusive breastfeeding ($p = 0.029$). The effect of mothers' diet before and after delivery was analyzed separately; the average of frequency of intake of each food category was considered. Whereas at T3 we did not observe any effect on BMI standard deviation scores (SDS), these were increased at T4 in the infants of mothers reporting a lower intake of cereals during pregnancy ($p = 0.0257$), and a higher intake of dairy products after delivery ($p = 0.002$). BMI SDS showed a trend to decrease in those who received formula milk after 3 months of age. Growth velocity showed a trend to decrease in breastfed children. In females, bicipital SFT was greater at T4 if mothers reported a greater meat ($p = 0.022$) and dairy intake ($p = 0.006$) after pregnancy. Tricipital SFT was smaller both at T3 and T4 if the intake of cereals was reduced during pregnancy ($p = 0.025$ and $p = 0.002$, respectively). Tricipital SFT was smaller at T3 when the cereal intake was reduced after pregnancy ($p = 0.022$). At T4, supra-iliac SFT was smaller when the cereal intake was greater during pregnancy ($p = 0.003$). In males, increased supra-iliac SFT was observed at T3 when the mothers reported a greater intake of cereals during pregnancy ($p = 0.016$).

CONCLUSION

Maternal nutrition during pregnancy and lactation has effects mostly on weight and distribution of fat in infants. This highlights the need to improve maternal nutrition.

ABS 69

ATYPICAL TERATOID RHABDOID TUMOR IN PEDIATRIC AGE

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INTRODUCTION

It is a malignant and rapidly aggressive tumor that generally affects the central nervous system (CNS)

and often, simultaneously, attacks other organs and tissues. Anglo-Saxon authors designate it by the acronym AT/RT (atypical teratoid/rhabdoid tumor). It affects very young children and often, as in the case we observed, can be present already in the first weeks of life, in both the brain and the liver.

In the majority of cases (60%), it begins in the CNS, with particular prevalence and localization in the posterior cranial fossa (cerebellum); in the remaining cases (40%), in other brain regions. Characteristic of this neoplasm is its very rapid diffusibility not only in the CNS but also in other organs. Metastases can attack the liver, kidneys, lungs, soft tissues and other organs and, despite precise and early treatment being practiced, they become evident in a few weeks. The terminology AT/RT specifically indicates forms with localization in the CNS. The very rare primary forms in the liver or other organs, are called teratoid or rhabdoid tumor of the child.

Symptoms are most easily seen in children older than 2 years of age and are the following: drowsiness, vomiting, macrocrania, diplopia, facial paralysis or other facial changes due to the involvement of several cranial nerves. More rarely, balance disturbances, seizures, hemiplegia occur. In the infant and young child of a few months, the diagnosis is very difficult because, although the neoplasm is already evident at the level of the CNS or liver, there is no obvious symptomatology. The physician examining such a patient a few weeks old, may consider himself lucky if he appreciates an increase in volume and consistency of the liver for a localization in that organ, or a macrocrania with mild diastasis of the sutures. The diagnosis is however very arduous, in the first weeks of life.

Most patients with a teratoid/rhabdoid tumor have inactivated *SMARCB1* gene. This gene has "onco-suppressive" activity and, therefore, has an active "regulation" in both cell proliferation and differentiation. When the *SMARCB1* gene is inactivated, the cells have no control, they replicate actively and expand, forming a tumor. More rarely it is another mutated gene, *SMARCA4*, which, similarly to *SMARCB1*, governs the mechanisms of cell proliferation and differentiation.

CASE REPORT

A 41-day-old baby girl came to normal checkup in apparent full well-being. First born, born at term by cesarean section, after normal pregnancy. Normal evolution of perinatal phenomena, formula feeding, rosy complexion, absence of jaundice, regular diuresis and alvus, normal cardiorespiratory activity. Absence of tremors, hypertone, nystagmus, etc. Normal blood pressure. Weight: 4.5 kg. Abdomen was mildly globular due to meteorism. On careful palpation, the right lobe of the liver of increased volume and consistency was appreciated, and percussion of the affected area confirmed this evidence. Ultrasound of the abdomen was immediately requested, which showed dimensional increase of the right lobe and the presence of two focal changes. Between segments V and VI, a cystic structure with little solid component and no vascularization of dimensions 5.5 x 5 x 4.7 cm. In segment VII, adjacent to the right suprahepatic vein that is imprinted but not infiltrated, a solid structure, with hypoechogenic appearance of size 2 x 2.2 cm. The child was transferred to a specialized department for such diseases and after further investigations (CT, MRI, ultrasound, biopsy, etc.) the diagnosis of rhabdoid/teratoid tumor in the brain, liver, lung was confirmed. The child is currently intubated in the Neonatal Intensive Care Unit and practicing all therapies prescribed by protocols for this difficult case.

REFERENCES

- Cerrato S, Vendemmia S, Pezone I, Vendemmia M. Rhabdomyosarcoma pelvis. In: Vendemmia S, Vendemmia M (Eds.). Atlante di Pediatria e Neonatologia per immagini. Naples: Cuzzolin Editore, 2022.
- Plesca DA. Embryonal Rabdomiosarcoma. In: Vendemmia S, Vendemmia M (Eds.). Atlante di Pediatria e Neonatologia per immagini. Naples: Cuzzolin Editore, 2022.

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BEHIND AN APPARENT SEPTIC SHOCK: A CASE REPORT

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CASE REPORT

In a full-term birth with uneventful delivery and regular fetal ultrasounds, the maternal rectovaginal swab was positive for Group B *Streptococcus* with complete antibiotic prophylaxis. Apgar scores were 6 and 8 at 1 and 5 minutes, respectively. The newborn exhibited appropriate growth for gestational age, and umbilical cord blood gas analysis was within normal limits. However, the infant developed severe respiratory distress and metabolic acidosis within the first 3 hours of life, necessitating the initiation of neonatal intensive care support.

In our Neonatal Intensive Care Unit, due to suspicion of sepsis, blood cultures were obtained, and empiric antibiotic therapy was initiated. To rule out a possible metabolic condition, serum ammonium levels were measured and found to be within the normal range. There was a progressive worsening of respiratory insufficiency, requiring invasive ventilation. Cardiological evaluation excluded structural heart abnormalities, but revealed severe heart failure (depressed left ventricular function with an ejection fraction of 30%, hypokinesis, and right ventricular dilation with suprasystemic pulmonary pressures), along with a significant elevation in ntproBNP and troponin levels. Treatment with milrinone and dopamine was initiated, with dopamine being discontinued due to stable blood pressure and active diuresis. Thrombocytopenia, anemia, and coagulation abnormalities were noted, leading to the administration of blood products. Sepsis remained suspected despite negative inflammatory markers. Metabolic acidosis only partially improved with bicarbonate administration. Serial echocardiographic evaluations showed gradual improvement in left ventricular function and reduced pulmonary pressures but persistent right ventricular dilation, as well as distension of the inferior vena cava and hepatic veins. Considering these findings, at approximately 20 hours of life, suspicion arose regarding an abdominal compartment syndrome, even in the absence of overt abdominal distension, that could explain the cardiac and acidosis picture. A comprehensive abdominal ultrasound revealed a large right paravertebral mass approximately 5 cm in diameter. A subsequent CT scan confirmed this finding, demonstrating significant compression and displacement of the inferior vena cava by the mass. To complete the diagnostic workup, urinary vanillylmandelic acid and homovanillic acid, neuron-specific enolase, serum βhCG, and α-fetoprotein were measured, all of which were markedly elevated. In suspicion of neuroblastoma, the neonate was transferred to a specialized referral center.

DISCUSSION

Initially, the clinical presentation, including metabolic acidosis, depressed cardiac function, and hematologic abnormalities, led to suspicion of sepsis. However, persistent negative inflammatory markers, stable blood pressures without the need for vasoactive agents, despite milrinone therapy, prompted consideration of an abdominal compartment syndrome. Neuroblastoma is the most common malignant solid tumor in pediatric patients, and cases diagnosed within the first 28 days of life represent 5% of all neuroblastomas. The clinical presentation of neuroblastoma in infants under 3 months is characterized by respiratory compromise, abdominal compartment syndrome, coagulopathy, and elevated blood pressure. Based on our experience, we believe that, in neonates presenting with a clinical picture suggestive of septic shock without the need for inotropic support, the possibility of neuroblastoma should be considered.

REFERENCES

- Davidoff AM. Neonatal neuroblastoma. Clin Perinatol. 2021;48(1):101-15.
- Fisher JPH, Tweddle DA. Neonatal neuroblastoma. Semin Fetal Neonatal Med. 2012;17(4):207-15.

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LEARNING FROM MISTAKES. WHICH CAME FIRST, THE EGG OR THE CHICKEN? NAVIGATING PARADOXES TO ARRIVE AT A DIAGNOSIS: A CASE REPORT

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CASE REPORT

S. was born at 41^{+1} weeks in another medical center via an urgent cesarean section due to inadequate engagement, maternal fever during labor, and meconium-stained amniotic fluid (M3). Apgar scores were 8 at 1 minute and 9 at 5 minutes, with no evidence of umbilical cord acidosis, and birth weight was below the 3rd percentile. On the second day of life, subentrant apneas emerged, necessitating intubation and transfer to our Neonatal Intensive Care Unit. An initial electroencephalogram confirmed the suspicion of status epilepticus, which was managed with phenobarbital, phenytoin, and midazolam, and ultimately discontinued on the 17th day of life. Subsequent brain ultrasound revealed a left-sided grade III intraventricular hemorrhage (IVH) according to Papile criteria, which was subsequently corroborated by magnetic resonance imaging (MRI) of the basal brain along with arteriography. On the 4th day of hospitalization, extubation was successfully performed following adequate seizure control. In the subsequent days, progressive post-hemorrhagic ventricular dilatation

(PHVD) occurred, leading to symptomatic hydrocephalus, which was managed through lumbar punctures. An urgent computed tomography scan identified superior sagittal sinus (SSS) thrombosis, which was further confirmed via venous angio-MRI. Simultaneously, a transfontanellar Doppler ultrasound was conducted, demonstrating the absence of blood flow within the SSS. Subsequent to the placement of an Ommaya reservoir, near-daily cerebrospinal fluid removals were performed (with prior ultrasound guidance), until ventriculoperitoneal shunt placement on the 67th day of life. Anticoagulant therapy, initiated upon the diagnosis of thrombosis, commenced with intravenous sodium heparin (for 23 days) and was followed by low-molecular-weight heparin, eventually discontinued after 57 days. Upon confirmation of blood flow within the SSS via Doppler ultrasound, a follow-up MRI on the 60th day of life revealed the restoration of normal flow signals within the venous sinuses, resolution of the thrombus, and a significant reduction in intraventricular blood.

DISCUSSION

In the existing literature, numerous cases of IVH in term neonates arising from cerebral venous sinus thrombosis (CVST) have been documented [1, 2]. In this particular case, the initial negative findings from the MRI performed upon admission suggest that CVST may have developed subsequent to PHVD, potentially exacerbated by decreased blood flow due to rapid ventricular dilatation and the concurrent inflammatory response associated with IVH. Alternatively, it is conceivable that the initial MRI study may have been prematurely conducted to detect SSS occlusion or may have benefited from supplementary neurodiagnostic investigations. The routine execution of transfontanelle Doppler ultrasound of the venous sinuses in patients with hydrocephalus is not currently a standard practice within our medical unit. In light of this case, we contemplate whether the expedited diagnosis of CVST could have been achieved through Doppler ultrasound assessments during routine PHVD evaluations. Moreover, in instances of isolated IVH in full-term neonates with predisposing risk factors for CSVT, consideration might be given to incorporating imaging sequences that enable a more precise evaluation of cerebral blood circulation.

REFERENCES

[1] Tan AP, Svrckova P, Cowan F, Chong WK, Mankad K. Intracranial hemorrhage in neonates: A review of etiologies, patterns and predicted clinical outcomes. Eur J Paediatr Neurol. 2018;22(4): 690-717.

[2] Kersbergen KJ, Groenendaal F, Benders MJ, de Vries LS. Neonatal cerebral sinovenous thrombosis: neuroimaging and long-term follow-up. J Child Neurol. 2011;26(9):1111-20.

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CONGENITAL ABSENCE OF PERICARDIUM: THE LARGEST ANALYSIS IN THE FIELD ON 247 WORLDWIDE CASES

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INTRODUCTION

Congenital absence of pericardium (CAP), also known as pericardial agenesis, is an extremely rare cardiac malformation occurring in less than 1/10,000 [1]. The first report dates back to 1559 [2]. Regarding embryology, incomplete development of the pleuro-pericardial membranes, fusing at the midline and separating the pericardial and pleural cavities, causes complete or partial CAP. Partial CAP is more common than complete CAP [3]. Left-sided CAP is way more common than right-sided and bilateral CAP. The defect is sometimes accompanied by other congenital cardiac abnormalities. Its diagnosis is at times suspected by means of echocardiography, but it needs to be refined with CT or cardiac MRI.

METHODS

Due to the rarity of the disease, just case reports and limited case series have been published so far. This study represents the largest analysis in the field on 247 worldwide cases. Age at diagnosis, gender prevalence, clinical presentation, electrocardiographic features, imaging (ultrasounds, CT, and cardiac MRI), associated cardiac abnormalities and outcome are here analysed.

RESULTS

The majority of reviewed CAP cases occurred in males (63.2%). Mean age at diagnosis was 31.8 ± 19.3 years; range 32 weeks of gestation-81 years). 23.5% of the patients were asymptomatic. The most common clinical presentations were chest pain (35.2%) and dyspnea (29.2%). The most commonly seen ECG changes were right axis deviation (28.7%) and right bundle branch block (23.9%). CAP was suspected or diagnosed by echocardiography in 20.1% of cases. Diagnosis was made by computed tomography and/or cardiac magnetic resonance imaging in 61.9% of cases. CAP was left sided in 71.2%, complete in 23.1%, and right sided in 5.7%. An associated congenital heart disease was found in 22.7%, especially in the form of atrial septal defect (6.5%) and patency of ductus arteriosus (2.8%). Pericardial repair was required in 12.9% of the incomplete forms of the disease. Never the complete form required surgical correction. The outcome was favorable in the majority of patients, with just a few deaths (7.3%).

CONCLUSION

Two hundred and forty-seven cases have been analysed in this systematic review. CAP can be detected at any age ranging from premature newborns to elderly patients. However, it is often detected incidentally during postmortem. Some patients – most often those with complete CAP – are asymptomatic, but especially those with partial CAP may suffer from vague atypical chest pain, which is thought to be caused by cardiac mobility or torsion or traction of cardiac structures. In extremely rare cases, the chest pain is angina-like or other symptoms can occur, such as arrhythmias or syncope. Some CAP patients also report shortness of breath on lying to one side – a phenomenon called trepopnea that is uniquely linked to partial defects. A few cases of serious complications potentially leading to death, including cardiac herniation of the left atrial appendage or left ventricle with subsequent muscle strangulation, occlusion of vena cava return, and compression of the coronary arteries, have been described. CAP is a rare entity varying from partial to complete absence. It is very important to differentiate partial from complete CAP because of the different symptoms and prognosis. CAP can be isolated or be a part of a syndrome (most of all the pentalogy of Cantrell). The most commonly seen associated congenital heart defects are atrial septal defect and patency of ductus arteriosus. The indication for surgical repair depends on symptoms, size, and location. Partial CAP is more at risk of herniation, thereby its closure reduces the risk. Conversely, complete CAP usually has a benign course and no intervention is required. Due to the rarity of CAP, the establishment of an International Registry would be desirable.

REFERENCES

[1] Faridah Y, Julsrud PR. Congenital absence of pericardium revisited. Int J Cardiovasc Imaging. 2002;18(1):67-73.

[2] Columbus R. De re anatomica libri XV. 1559.

[3] Glover LB, Barcia A, Reeves TJ. Congenital absence of the pericardium. A review of the literature with demonstration of a previously unreported fluoroscopic finding. J Roentgenol Radium Ther Nucl Med. 1969;106(3):542-9.

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NAILFOLD MICROVASCULAR DYSFUNCTION PRECEDES RETINOPATHY IN CHILDREN AND ADOLESCENTS WITH POORLY CONTROLLED TYPE 1 DIABETES

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INTRODUCTION

Nailfold capillaroscopy (NFC) is a non-invasive and easy-to-use imaging technique to study microvascular circulation under the nail. Nailfolds are the only body site allowing for an assessment of the capillary microvasculature at a horizontal plane. NFC study has been applied in a number of physiological and pathological conditions [1]. Type 1 diabetes (T1D) is the most common kind of diabetes in youth, and its incidence is on an upward trajectory. Approximately 18% of T1D new diagnoses is in children with age equal to or less than 9 [2]. According to another study, the number of T1D pediatric patients increases of 3.4%/year. As such, even T1D-related macro- and microvascular complications are increasing, including diabetic retinopathy (DR) [2]. The latter is one of the most common causes of blindness in developed countries [3]. The risk of DR can be decreased with a tight glycemic control, with hemoglobin A1c (HbA1c) being the best indicator of long-term glycemic

control with the ability to reflect the cumulative glycemic history of the preceding 2 to 3 months. The American Diabetes Association and the International Society of Pediatric and Adolescent Diabetes suggest a blood level of HbA1c < 7.0% in all T1D children. The current guidelines recommend a DR screening starting from the age of 11 and after 2-5 years following T1D diagnosis. The aims of the study are looking for early nailfold microcirculation abnormalities and abnormal retinal findings in children affected by T1D in comparison with healthy counterparts and searching for possible correlations between these abnormalities and the patients' HbA1c. Capillaries morphology, density, and how blood flows through the capillary system were evaluated. A comparison with a group of healthy peers (n = 55) was done as well.

METHODS

Twenty-six patients with T1D were enrolled (17 males and 9 females; mean age 12 ± 3 years; age range 6-18 years). None of the enrolled children was a smoker or was suffering from any other autoimmune conditions. All of them underwent retinal vessel analysis as well as NFC in a single 3-hour session.

RESULTS

The mean HbA1c was 8.1% ± 1.1%. Mean duration of T1D was 3.4 ± 0.9 years. None of the participants had early signs of retinopathy (p = ns compared to controls), but all of them had one or more abnormalities on NFC. Regarding T1D patients, in 18 out of 26 (69.2%), nailfold capillaries were tortuous, while tortuosity was seen just in 3/55 controls (5.4%, p < 0.00001). In 14/26 T1D subjects (53.8%), capillaries were dilated with or without apical aneurysms, while this abnormality was not seen in any of the controls (p < 0.00001). In 12 T1D subjects (46.1%), the number of capillaries was markedly reduced (< 7 capillaries/mm^2). Again, in 12/26 patients (46.1%), perivascular edema was seen (p < 0.00001 with control group). In all the sample size (100%), capillary blood flow was slowed down, while this abnormality was not seen in any of the controls (p < 0.00001). In slightly more than a half of the enrolled patients (53.8%), a blood "sludge" phenomenon (i.e. increased blood viscosity and tendency to red cell aggregation leading to a temporary blood flow interruption) was detected. Again, this was not noted in any of the healthy controls (p < 0.00001). No gender differences were seen. No correlation was detected with HbA1c (p = ns).

DISCUSSION

This is the first study investigating peripheral microcirculation in children and adolescents with T1D and no DR by using NFC. Although glycemic control was not ideal (mean HbA1c 8.1% with a suggested target < 7%), nobody of the patients in the study had DR. NFC is able to detect early microvascular changes (tortuous, ectatic/aneurysmatic capillaries, microhemorrhages, periungual swelling, sludge phenomenon) which precede the onset of DR. The latter is the leading cause of blindness in diabetic adults. In this respect, early identification of those T1D subjects at risk of developing DR before their sight is threatened is crucial. All the detected abnormalities are non-specific. No correlation was seen with disease duration. Interestingly, in 84.6% of the subjects in the study had HbA1c > 7.5%, which is the cut-off identified in the Diabetes Control and Complications Trial (DCCT) for the risk of developing T1D-related complications. The DCCT showed that intensive therapy against T1D in subjects aged 13-39 years had reduced the number of the disease-related complications. It is noteworthy to mention that 63% of the T1D were amid puberty. Vascular endothelial growth factor (VEGF) serum concentrations are increased in prepubertal and pubertal children with diabetes. Severity of microvascular complications is associated with a marked increase of VEGF concentrations in the serum of these patients. Overall, NFC proved to be capable of identifying some preclinical signs of T1D. The technique may predict microvascular dysfunction development in T1D children and adolescents.

REFERENCES

[1] Martino F, Agolini D, Aprigliano D, Guido F, Placanica G, Giardini O. [Nailfold capillaroscopy in normal children between 0 and 16 years of age]. [Article in Italian]. Minerva Pediatr. 1997;49(5):197-201.

[2] Lawrence JM, Divers J, Isom S, Saydah S, Imperatore G, Pihoker C, Marcovina SM, Mayer-Davis EJ, Hamman RF, Dolan L, Dabelea D, Pettitt DJ, Liese AD; SEARCH for Diabetes in Youth Study Group. Trends in Prevalence of Type 1 and Type 2 Diabetes in Children and Adolescents in the US, 2001-2017. JAMA. 2021;326(8):717-27.

[3] Sabanayagam C, Banu R, Chee ML, Lee R, Wang YX, Tan G, Jonas JB, Lamoureux EL, Cheng CY, Klein BEK, Mitchell P, Klein R, Cheung CMG, Wong TY. Incidence and progression of diabetic retinopathy: a systematic review. Lancet Diabetes Endocrinol. 2019;7(2):140-9.

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CAN INFLAMMATION DURING THE PRENATAL AND NEONATAL STAGES AFFECT HEALTH IN ADULTHOOD?

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The Developmental Origins of Health and Disease (DOHaD) hypothesis has illuminated the profound influence of early-life experiences on lifelong health. This review synthesizes key findings and connections in the context of the DOHaD hypothesis, highlighting the impact of prenatal and neonatal inflammation on adult well-being. Evidence points to the presence of microplastics and nanoplastics (NPs) in the human placenta, raising concerns about their effects on fetal development and immune responses [1]. These synthetic particles may incite immune responses, potentially interfering with fetal defenses against pathogens. Moreover, NPs have been shown to damage mitochondria and increase oxidative stress, accentuating the far-reaching impacts of environmental factors on early development. Even at low concentrations, such as those found in the environment, NPs have been demonstrated to exert these effects. The gut microbiota emerges as a central player, influencing gastrointestinal health, neuropsychiatric conditions, and cognitive functions through the microbiota-gut-brain axis [2]. Within this complex web of interactions, the gut microbiota's composition has been associated with cognitive function and behavior modulation by influencing neurotransmitter metabolism. In the context of psychiatric disorders, there is notable evidence of heightened gut inflammation in individuals with conditions such as schizophrenia and first-episode psychosis, which potentially influences systemic cytokine levels and, in turn, impacts brain function. Maternal factors, including cholesterol levels and microRNAs (miRNAs) in breast milk, play pivotal roles in shaping an individual's health trajectory. The evolution of breast milk is intrinsically tied to its capacity to provide not only essential nutrients but also a sophisticated array of substances, including miRNAs. These miRNAs, discovered in extracellular vesicles in human milk, play indispensable roles in regulating various biological processes in infants. They wield influence over lipid and glucose metabolism, gut development, neurogenesis, immunity, and even tissue-specific gene expression throughout life. Furthermore, there is a burgeoning interest in the potential impact of these miRNAs on viral infections in infants. Moreover, neurodevelopmental disorders are linked to microglial dysfunction, while mitochondrial dysfunction and oxidative stress underlie aging and disease [3]. Microglia develop from erythro-myeloid progenitors in the early embryonic yolk sac and migrate into the brain, where they remain throughout an individual's life. Microglia's association with various neuropsychiatric disorders, including schizophrenia, Rett syndrome, bipolar disorder, and major depressive disorder, underscores their critical role in synaptic modulation, learning, and memory processes. Microglia function and morphology may be altered in these conditions, potentially leading to imbalances in synaptic plasticity mechanisms. Proper synaptic pruning, vital for maintaining the balance between excitatory and inhibitory synapses in the brain, is necessary for healthy neurodevelopment. Despite some limitations, this review underscores the importance of early-life experiences and environmental factors in determining lifelong health, offering insights into potential preventive strategies and interventions.

REFERENCES

[1] Ragusa A, Svelato A, Santacroce C, Catalano P, Notarstefano V, Carnevali O, Papa F, Rongioletti MCA, Baiocco F, Draghi S, D'Amore E, Rinaldo D, Matta M, Giorgini E. Plasticenta: First evidence of microplastics in human placenta. Environ Int. 2021;146:106274.

[2] Khandaker GM, Cousins L, Deakin J, Lennox BR, Yolken R, Jones PB. Inflammation and immunity in schizophrenia: implications for pathophysiology and treatment. Lancet Psychiatry. 2015;2(3):258-70.

[3] Guzman-Villanueva D, Weissig V. Mitochondria-Targeted Agents: Mitochondriotropics, Mitochondriotoxics, and Mitocans. Handb Exp Pharmacol. 2017;240:423-38.

ABS 75

PHENOTYPIC VARIABILITY IN *PPP2R5D*-RELATED NEURODEVELOPMENTAL DISORDER

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The *PPP2R5D* gene encodes for the B56 regulatory subunit of protein phosphatase-2A (PP2A), an intracellular serine/threonine phosphatase. PP2A-B56 has been shown to inhibit AKT activity, through a process of dephosphorylation, leading to down-regulation of cell growth and proliferation linked to the PI3K/AKT pathway. Missense variants in *PPP2RD5* are described to be associated with macrocephaly, hypotonia, and intellectual disability.

We present the case of a child, currently 15 months of age, who showed from birth macrocrania, with stable values above the 99th percentile, sagittal and coronary suture diastasis with a wide anterior fontanel, asymmetry of the frontal bosses, and plagiocephaly. At 3 months, he had surgery for removal of angioma in the left pectoral site. At abdomen ultrasound, two angiomas at liver level were found. At re-evaluation at 6 months of age, persistence of macrocephaly was noted, with remaining auxological parameters between the 25th and 50th percentiles. Marked axial hypotonia, with absent head control and delayed acquisitions, was present. Neurological evaluation and MRI of the brain were performed, with no abnormalities found. SNP-array, which excluded the presence of CNV, and whole exome sequencing (WES) in trio were performed. The analysis detected the presence in heterozygosity of the *de novo* variant c.592G>A (p.Glu198Lys) at exon 5 of the *PPP2R5D* gene. The fact that the *PPP2RD5* gene is involved in neoangiogenesis could explain the presence of currently unreported vascular malformations in the rare cases described. Given the rarity of the neurodevelopmental disorder related to *PPP2R5D* and the recent identification of the condition, in patients with variants in the gene, a clinic-internal evaluation considering the possible presence of such malformations might therefore be appropriate in order to better follow-up along with overall monitoring, given also the reduced knowledge of the long-term outcome.

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IMPACT OF MATERNAL DIET ON PUBERTAL STAGES AND ANO-GENITAL DISTANCE IN THE FIRST 12 MONTHS OF LIFE: PRELIMINARY DATA FROM THE EUROPEAN LIFE-MILCH PROJECT

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INTRODUCTION

The European LIFE-MILCH study evaluates endocrine disrupting chemicals (EDC) in mothers and newborns up to 12 months of age, in particular in breast and formula milk and in the urine of both. The project will explore EDC relationships with growth, distribution of adiposity, pubertal stages and neurodevelopment, and ano-genital distance (AGD) to define a risk assessment model to produce safety guidelines.

OBJECTIVE

To observe the effects of maternal diet during and after pregnancy on pubertal stages, genital malformations, and AGD from birth up to 12 months of age.

METHODS

254 healthy mother-infant dyads were enrolled at 36-40 weeks of gestational age. The cohort included 125 males and 129 females. Pregnancies were physiological. Questionnaires related to nutritional habits (daily and weekly frequency of intake of cereals, meat, eggs, fruit, vegetables, fatty food and dairy products) at recruitment (T0), 1 (T1), 3 (T2), and 6 months (T3) were filled out by all women after delivery. The duration and type of feeding were recorded for all newborns. All auxological measurements were taken and in addition pubertal stages and AGD at TO, T1, T2, T3 and T4.

RESULTS

The intake of vegetables, fruit ($p < 0.001$) and cereals ($p < 0.001$) was reduced after delivery, whereas fat food consumption was increased ($p < 0.001$). The meat intake was constant and dairy products intake was variable. Cereals intake started to decrease at T2 only for mothers who stopped exclusive breastfeeding ($p = 0.029$). Thelarche disappeared earlier in the females whose mothers referred a higher cereal intake during pregnancy ($p = 0.006$), as well as in those who reported a higher intake of eggs ($p = 0.048$). In males, a slight correlation ($p = 0.062$) between the persistence of breast bud and a higher cereal intake was found after delivery. In females, maternal diet after delivery showed no relationship with thelarche. A lower probability of hydrocele ($n = 35/125$, $p = 0.011$) was associated with a higher dairy products intake during pregnancy. Generally, the mother's nutritional preferences were not correlated with AGD. However, boys at 3 and 6 months showed a weak negative association ($p = 0.038$) with meat consumption during pregnancy, although a strong negative association was observed with mother's fatty food intake at 3 months after delivery ($p = 0.039$).

CONCLUSION

The mother's nutrition during pregnancy and lactation influences the clinical signs of minipuberty, and possibly has an effect on the development of hydrocele, and on AGD. This underlines the need to control maternal diet.

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CLINICAL AND ONCOLOGIC OUTCOMES AFTER PAROTIDECTOMY FOR PEDIATRIC PAROTID TUMORS

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INTRODUCTION

Diagnosis and treatment of pediatric parotid tumors can be challenging [1] because of the heterogeneous nature of these tumors as well as their rarity. Currently, evidence-based guidelines for treatment are lacking [2]; as a consequence, the management of pediatric parotid tumors is mainly based on the experience gained with adults. The authors report their experience on the surgical management of pediatric parotid tumors.

MATERIALS AND METHODS

We conducted a retrospective analysis of 495 consecutive parotidectomies performed in our Department from December 2010 to December 2021. All patients younger than 18 years who underwent parotidectomy for benign or malignant parotid neoplasm were included. Patients' demographic data, clinical presentation, tumor size, surgical management, histopathologic diagnosis, postoperative complications, and oncologic outcomes were evaluated.

RESULTS

Eight patients younger than 18 years (4 males and 4 females, mean age 15 years, range of 11-18 years) underwent parotidectomy for parotid neoplasms. All patients presented a slow-growth pre-auricular mass, without any other symptoms in 7 cases (87.5%), while it was associated with pain in 1 case (12.5%). Preoperative facial palsy was never observed. All the tumors were located in the superficial lobe without clinical or radiological evidence of neck lymph node involvement; therefore, all patients underwent parotidectomy I-II according to Quer [3] (superficial parotidectomy) without neck dissection. We managed to spare the facial nerve in all patients, and no facial paralysis or paresis was seen immediately after surgery or during follow-up. Mean hospitalization time was 5 days. Only minor complications were recorded: 4 patients presented earlobe numbness, 3 patients developed Frey's syndrome and 1 patient presented a postoperative seroma. At definitive histology, 4 patients (50%) had benign epithelial tumors (pleomorphic adenoma in all cases), 2 patients (25%) had malignant epithelial tumors (low-grade mucoepidermoid carcinoma in 1 case and acinic cell carcinoma in the other case) and 2 patients (25%) had hematolymphoid tumors (Hodgkin lymphoma in both cases). Definitive histology confirmed the complete removal of the neoplasm with negative margins in all cases. Mean tumor size was 23.5 mm (range of 12-30 mm). No patient showed adverse pathologic features, such as perineural or lymphovascular invasion. Patients diagnosed with benign epithelial tumor were considered disease-free and no other care was indicated. Patients with malignant epithelial tumors were discussed in a multidisciplinary oncological board: adjuvant therapy was not deemed necessary, and patients were directed to close follow-up. Mean follow-up time for benign and malignant epithelial tumors was 5.4 years (range of 2-11 years). None of our patients developed recurrences during the follow-up and they were free of disease at the last follow-up. The 5-year recurrence-free survival of the patients with benign and malignant tumors was 100%. Patients with a diagnosis of lymphoma were referred for long-term oncologic follow-up to the Department of Onco-Haematology, and 5 and 6 years after surgery, they did not show any complications from the surgical procedure.

CONCLUSION

In our experience, pediatric parotid tumors show higher tendency to be malignant than in adults, but, when adequately treated, they are associated with an overall more favorable prognosis.

REFERENCES

[1] Liu B, Liu JY, Zhang WF, Jia J. Pediatric parotid tumors: clinical review of 24 cases in a Chinese population. Int J Pediatr Otorhinolaryngol. 2012;76(7):1007-11.

[2] Morse E, Fujiwara RJT, Husain Z, Judson B, Mehra S. Pediatric Salivary Cancer: Epidemiology, Treatment Trends, and Association of Treatment Modality with Survival. Otolaryngol Head Neck Surg. 2018;159(3): 553-63.

[3] Quer M, Guntinas-Lichius O, Marchal F, Vander Poorten V, Chevalier D, León X, Eisele D, Dulguerov P. Classification of parotidectomies: a proposal of the European Salivary Gland Society. Eur Arch Otorhinolaryngol. 2016;273(10):3307-12.

ABS 78

SUPRAVENTRICULAR TACHYCARDIA IN THE NEWBORN: A RETROSPECTIVE STUDY CONDUCTED IN CAGLIARI, ITALY

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INTRODUCTION

Supraventricular tachycardia (SVT) is considered among the most common causes of arrhythmia in infants and children. It is an increase in heart rate greater than 220 bpm in children under 1 year of age and greater than 180 bpm above 1 year of age. It can be detected during pregnancy, "fetal SVT", with a ventricular rate greater than 180 bpm and/or on fetal echocardiogram by analyzing the relationship between mechanical contractions of the atrial and ventricular chambers.

STUDY DESCRIPTION

A retrospective study was conducted at the AOU of Cagliari (Cagliari, Italy) at Dr. Neroni's Level 2 Pediatric Cardiology Outpatient Clinic, aimed at providing an analysis on the treatment and management of neonatal SVT found in a population sample of 20 patients, from 2014 to 2021. We evaluated the type and dosage of antiarrhythmic drugs used for each subclass of SVT, predictivity of success, duration of prophylactic therapy, and follow-up management. Criteria for inclusion: having presented with an episode of tachycardia within the first year of life, which had to have been followed by admission to the AOU of Cagliari. Data taken into consideration: gestational age and prematurity, birth weight, age at first onset of SVT, positive history of fetal tachycardia, presence of comorbidities such as congenital heart disease, severity of clinical picture, possible ventricular dysfunction and signs of decompensation, maximum heart rate, type of abortive treatment, type of antiarrhythmic drugs in maintenance therapy, duration of hospital stay, follow-up and outcome. Arrhythmias were classified as atrial flutter, atrial ectopic, atrial multifocal, atrioventricular reentrant tachycardia (with subclass including Wolff-Parkinson-White syndrome), permanent junctional reentrant tachycardia, and unspecified SVT, in case the infant had only a diagnosis of SVT; in addition, the study includes an infant with a diagnosis of ventricular tachycardia.

Therapy at onset includes amiodarone as an antiarrhythmic, while prophylactic therapy includes amiodarone, propranolol, flecainide, digoxin, ivabradine, and nadolol. Possible multidrug therapies and rates in antiarrhythmic drug combinations were analyzed.

CONCLUSIONS

SVT has a good prognosis with a low incidence of dramatic clinical pictures, the most frequent type being atrioventricular reentry tachycardia, in line with the literature. The most widely used drug therapy is flecainide, which has shown 88% efficacy in preventing short- and medium-term recurrence. Overall, prophylaxis was ineffective in 27% of infants, some of whom had additional comorbid conditions.

REFERENCES

• Capponi G, Belli G, Giovannini M, Remaschi G, Brambilla A, Vannuccini F, Favilli S, Porcedda G, De Simone L. Supraventricular tachycardias in the first year of life: what is the best pharmacological treatment? 24 years of experience in a single centre. BMC Cardiovasc Disord. 2021;21(1):137.

• Costantini M, Rosati E, Costantini L, De Jaco G, Amico A, Maffia M, Bronzetti G. Quadri aritmici in età pediatrica. Dai meccanismi al Pronto soccorso. Area Pediatrica. 2019;20(3):128-35.

• Srinivasan C, Balaji S. Neonatal supraventricular tachycardia. Indian Pacing Electrophysiol J. 2019;19(6):222-31.

ABS 79

A RARE CASE OF FULMINANT NEONATAL EARLY-ONSET SEPSIS DUE TO *S. PYOGENES*: AN EMERGING CHALLENGE?

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CASE DESCRIPTION

We present a fatal case of a neonate outborn at 35^{+6} weeks of gestational age by vaginal delivery for spontaneous preterm labor. Apgar scores at 1 and 5 min were 8 and 9, respectively. The pregnancy was

uneventful, except for isolated fetal mild bilateral pyelectasis. There was no history of prolonged rupture of membranes and the vagino-rectal swab for Group B *Streptococcus* (*GBS*) was negative. During the first hour of life, lethargy, respiratory distress and mixed acidosis were noted. Therefore, he was transferred to our Neonatal Intensive Care Unit. He required mechanical ventilation, surfactant administration and vasoactive drugs. The peripheral white blood cell count was 3,530/mmc, normal CRP (3.5 mg/L), elevated presepsin (2,216 pg/ml), procalcitonin (100 ng/ml) and INR (2.4). Early-onset neonatal sepsis was suspected and intravenous broad spectrum antibiotic therapy was promptly started. *Streptococcus pyogenes (S. pyogenes)* was later isolated from blood cultures. Fulminant septic shock with disseminated intravascular coagulation was confirmed, which progressed to worsening general condition; the newborn died of septic shock and multiple organ dysfunction in less than 30 hours of life. A detailed medical history showed tonsillitis in the older sibling, 2 weeks before delivery's day. Throat cultures obtained from both parents and the older sibling were positive and mother's vaginal culture was positive for *S. pyogenes*. According to our regional health protocol, the mother, considered to be a high risk category for invasive disease, received antibiotic therapy, and for all other family members monitoring has been proposed.

DISCUSSION

S. pyogenes (Group A *Streptococcus* – *GAS*) rarely causes early-onset neonatal sepsis. Differently from vaginal swab for *GBS*, no screening programs for *GAS* are normally performed nowadays, considering a significantly lower rate of *GAS* vaginal colonization, maternal and neonatal infections. The European Centre for Disease Prevention and Control (ECDC), nevertheless, confirmed an increase of invasive *GAS* infections (iGAS) in the first years of life in several European countries during 2022, in particular since September 2022 after COVID-19 pandemic [1].

Many different hypotheses have been formulated in this regard. Neither the involvement of a specific strain nor mechanisms of antibiotic resistance appear to be relevant [1].

COVID-19 pandemic could explain the increased risk of iGAS infections through a reduction of immune stimulation due to non-pharmaceutical interventions and lockdowns (immune debt) or an immune dysregulation (immune theft) after COVID-19 infection [2]. It has also been suggested that coinfections with respiratory viruses could play an important role [1]. In fact, it was already known that respiratory viruses, particularly influenza, can modify bacterial adherence and effector cell-mediated clearance, increasing the incidence of iGAS cases [3]. UK guidelines for the management of contacts of iGAS infections in community settings consider neonates and women within the first 28 days of delivery in close contact of iGAS cases at home, as high-risk patients. Antibiotic prophylaxis for high-risk close contacts had to start as soon as possible (within 24 hours, and preferably the same day) but not to commence beyond 10 days of iGAS diagnosis in the index case [4]. It is, therefore, significant to highlight the emerging problem of iGAS disease in the first years of life and increase the level of surveillance, considering the high rate of mortality and morbidity associated with this condition.

REFERENCES

[1] ECDC. Increase in Invasive Group A streptococcal infections among children in Europe, including fatalities. Press release. Available at: https://www.ecdc.europa.eu/en/news-events/increase-invasive-group-streptococcal-infections-among-children-europe-including, date of publication: December 2022, last access: October 2023.

[2] Jain N, Lansiaux E, Reinis A. Group A streptococcal (GAS) infections amongst children in Europe: Taming the rising tide. New Microbes New Infect. 2022;51:101071.

[3] Herrera AL, Huber VC, Chaussee MS. The Association between Invasive Group A Streptococcal Diseases and Viral Respiratory Tract Infections. Front Microbiol. 2016;7:342.

[4] UK Health Security Agency. UK guidelines for the management of contacts of invasive group A streptococcus (iGAS) infection in community settings. Available at: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/1140766/Management-of-contacts-of-invasive-group-a-streptococcus.pdf, date of publication: March 2023, last access: October 2023.

ABS 80

WILLIAMS SYNDROME, AORTIC RECOARCTATION AND MULTIVESSEL INVOLVEMENT: THE IMPORTANCE OF TIMELY DIAGNOSIS AND TREATMENT

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INTRODUCTION

Coarctation of the aorta is a narrowing of the descending aorta, which is typically found at the

insertion of the ductus arteriosus. It represents 6% to 8% of all congenital heart defects. In 90% of cases, it is sporadic, but it can be part of specific syndromes, such as Williams syndrome. Williams syndrome is characterized by a microdeletion of the long arm of chromosome 7, also containing the *ELN* gene, which codes for elastin: this involves, with a variable phenotype, cardio-vascular anomalies, such as the stenosis of medium and large caliber arteries, which appear thickened and stiff. In addition, there may be facial dysmorphisms, a peculiar neuropsychological, cognitive and linguistic profile and connective tissue anomalies.

CASE REPORT

We will describe the case of a newborn a few hours old, transferred to our Neonatal Intensive Care Unit from a peripheral hospital due to skin hypoperfusion, systolic murmur, weak femoral pulses. Given the clinical and echocardiographic diagnosis of tight aortic coarctation, IV therapy with PGE1 0.1 mcg/kg/min was administered and the baby underwent cardiac decoarctation surgery, performed on the fifth day of life. At the post-discharge check-up, she showed an initial narrowing of the caliber of the aortic arch, which worsened in the following days; a narrowing of the peripheral pulmonary branches was also found. The pressures measured in the right upper limb and lower limbs demonstrate an initial aortic re-coarctation: BP AS 104/80; BP AAII 95/61 mmHg. Furthermore, persistent eyelid edema, which was initially associated with neonatal heart failure, and a small chin are noted. Therefore, a clinical suspicion of Williams syndrome raised and genetic tests were performed.

CONCLUSIONS

Patients with Williams syndrome and aortic coarctation must be subject to close clinical monitoring, because for such patients several cases of rapidly progressive aortopathies have been described in the literature.

In these children, recoarctation seems to be linked to an active hypertrophy of the muscle cells, related to the absence of elastin at the level of the vessel wall, rather than to a failure of the surgery to maintain an adequate caliber of the vessel lumen. In addition to the increased risk of recoarctation, it is important to consider the anesthetic risk and the potential multivessel involvement as well, which can include, in addition to the aorta and pulmonary arteries, also renal, mesenteric and coronary arteries, all of them leading to increased mortality. Summarizing, the treatment of patients with aortic coarctation must mandatory take into account the potentially associated genetic syndromes because they have impact on the prognosis and therapeutic approach.

REFERENCES

- Kim YY, Andrade L, Cook SC. Aortic Coarctation. Cardiol Clin. 2020;38(3):337-51.
- Mannarino S, Keizman E, Pasotti M, Codazzi AC, De Sando E, Giamberti A. A rare case of discrete aortic coarctation in Williams-Beuren syndrome. Diagnostic and therapeutic considerations. Pediatr Med Chir. 2015;37(2):pmc.2015.120.
- Pober BR. Williams-Beuren syndrome. N Engl J Med. 2010;362(3):239-52.

ABS 81

PEER RELATIONSHIPS AND ADOLESCENTS WITH CHRONIC DISEASE

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Adolescence is a critical time for the development of peer relationships and the presence of a chronic disease complicates this process. There are four key aspects of youths' peer relationships: close friendships; one's position in the larger peer group; the presence of peer victimization (PV); and romantic relationships. There are bidirectional influences between peer relationships and chronic disease. Specifically, in each of the four areas, youths' relationships can interfere with disease management, and having a chronic disease can interfere with relationship development.

First, close friendships provide youth with support, companionship, and intimacy. They buffer the impact of stress and enhance self-esteem. Importantly, youths' close friends are a key source of support for chronic disease; they also provide companionship for lifestyle aspects of disease management (e.g., eating, exercising) and help youth feel accepted. However, youth with certain conditions, like neurological disorders or obesity, find it harder to establish close friendships than healthy youth and thus may be limited in their friendships.

Second, adolescents need to "fit in" with peers and find their position within the larger peer group. The peer group contributes to a sense of identity and belonging and provides opportunities for developing friendships and romantic relationships. However, peers can affect health behaviors. For example, youth who align with the "jocks" (athletes) or "brains" (studious youth) are likely to engage in healthy eating and exercise, which can positively affect a chronic disease. In contrast, youth who

affiliate with a "problem" crowd are more likely to drink, smoke, or engage in risky sexual behavior, with potential adverse health consequences. Also, youths' chronic disease might limit their ability to affiliate with other peers. Youth with visible defects or cognitive impairment, or who cannot participate in typical peer activities due to their illness, may find themselves neglected or rejected by peers.

Third, adolescents may experience stress from PV. The most common forms of PV are relational victimization (i.e., being left out or excluded by friends) and reputational victimization (i.e., being the target of rumors or efforts to damage one's reputation). PV can occur online and through social media. Not only does PV contribute to youths' psychological distress (e.g., feeling anxious and depressed) but also to sleep problems and somatic complaints, all of which could interfere with disease management. In turn, youth with certain chronic conditions are more likely to experience PV than others; this includes youth with visible illnesses or disabilities, such as craniofacial conditions, obesity, epilepsy, chronic skin disease, or visual impairments.

Finally, adolescents' romantic relationships are important. By 16 years of age, most youth have had a romantic relationship, which sets the stage for intimate adult relationships and provides considerable social support. However, romantic relationships can be a source of stress and conflict, especially when a break-up occurs. For youth with chronic conditions, romantic partners are an important source of support and can facilitate youths' disease management. Like friends, they can support either healthy or unhealthy behaviors. However, romantic conflict and break-ups are associated with poorer self-care and greater psychological distress in youth with chronic conditions. Although less well-studied than other types of peer relationships, youth with visible illnesses or disabilities may likely have a harder time developing romantic relationships than other youth.

In general, chronic conditions that are visible or associated with cognitive impairment may trigger PV or rejection and affect youths' peer acceptance and romantic appeal. Conditions with complex management tasks that affect one's lifestyle (e.g., eating, exercise) can limit youths' participation in normal peer activities. Similarly, conditions with intensive or extended treatments can limit youths' friendships and normal social activities.

To address the above concerns, providers are encouraged to monitor youths' peer relationships to better understand how they may affect disease management. First, ask about youths' friendships and romantic relationships (e.g., Who are their friends and romantic partners? How do they help or interfere with disease management?) and consider inviting youths' friends and romantic partners to assist with disease management and support. Second, ask about youths' peer acceptance and the crowds they affiliate with; help youth manage their chronic disease while also "fitting in" with peers. Third, ask youth if they experience PV or bullying, and if so, help them get help. Finally, ask about romantic relationships and breakups. In general, providers should help youth develop connections that promote peer support and positive relationships.