

# Selected Abstracts of the 6<sup>th</sup> International Congress of UENPS • Session “Other organs (eye, kidney, blood)”

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**ABS 1****NEPHROLITHIASIS IN AN EXTREMELY LOW BIRTH WEIGHT INFANT. A CASE REPORT**

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**INTRODUCTION**

Presence of kidney stones in the neonatal period is an especially rare disease, which usually manifests itself as a complication of nephrocalcinosis in very preterm infants.

**CASE REPORT**

We describe a clinical case of an extremely preterm girl (23 weeks of gestation, birth weight 590 grams) diagnosed with kidney stones. The girl's adaptation was complicated due to respiratory distress syndrome as well as chronic pulmonary disease. The initial congenital and the later acquired infections were treated with antibiotics. Diuretics, hormones and parenteral nutrition were also administered. She underwent patent ductus arteriosus ligation and laser photocoagulation for the treatment of retinopathy of prematurity. The infant was diagnosed with renal nephrocalcinosis in her right kidney by ultrasound at 35 days of age. Ultrasound monitoring of kidney stones was carried out in two weeks when a urinary tract infection complicated the condition. Stones were also seen radiographically. As this condition worsened, acute renal failure progressed. The condition had improved after intensive treatment. Upon discharging the patient from the hospital, ultrasound scans of the right kidney were showing large numbers of small calcium stones coalescing in some places. The girl went home in satisfactory condition at 126 days of her life (corrected age of 41 weeks).

**CONCLUSIONS**

In this case, kidney stones complicated the already difficult state of the extremely preterm infant, increasing the risk of morbidity and survival during the first months of her life.

**ABS 2****A CASE OF SEVERE HEMOLYTIC DISEASE DUE TO Rh-ISOIMMUNIZATION**

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**INTRODUCTION**

Hemolytic disease of the newborn is a consequence of maternal alloimmunization against fetal red blood cells antigens. The Rhesus (Rh) blood group is particularly important because of its strong immunogenicity, with the ability to cause severe hemolysis and consequent anemia and hyperbilirubinemia in the first hours after birth.

**METHODS**

Herein we report the case of a male newborn, following a nearly term uneventful gestation (36 weeks). It was a second gestation and the maternal blood group, typed in the first gestation, was A Rh positive. An elective caesarean was decided after a low variability pattern on the cardiotocogram.

**RESULTS**

The newborn needed resuscitation following apnea, bradycardia and extreme skin pallor, being immediately admitted in the neonatal intensive care unit. Blood studies showed a severe anemia (hemoglobin 3 g/dl) and a positive direct antiglobulin test. Red blood cells transfusion and immunoglobulin were administered to overcome the hemolysis. The maternal blood type was repeated and was found to be A Rh negative, which confirmed a wrong blood typing on the first gestation. During the hospitalization period, 3 more blood transfusions were needed, as well as albumin replacement and an antibiotic course (due to a neonatal sepsis with negative blood cultures). The total bilirubin increased to a maximum of 26.9 mg/dl (with direct bilirubin of 19 mg/dl). A cranial ultrasonography done in the first week suggested a left frontoparietal stroke, which was conservatively managed.

**CONCLUSIONS**

Despite the severity of hemolytic disease presented in our case, the outcome has been, at this point, good. Due to the possibility of late adverse neurodevelopment manifestations, this case requires a rigorous follow-up. It is also important to remind that Rh D alloimmunization remains a current issue. Antenatal prophylaxis with specific anti D-immunoglobulin is crucial during pregnancy in such cases.

**ABS 3****TENDENCY OF RETINOPATHY OF PREMATURITY. ARE OUR PRETERM BABIES SAFE FROM IT?**

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*Pediatric Department, Neonatal Unit, Reina Sofia University Hospital, Córdoba, Spain***BACKGROUND**

Retinopathy of prematurity (ROP) is a proliferative disease that affects the premature infants. High  $\text{FiO}_2$ , blood transfusions and sepsis have been related to the etiology. It has been described a stop in the normal vascular growth, fibro vascular tissue and retinal detachment.

**OBJECTIVE AND METHODS**

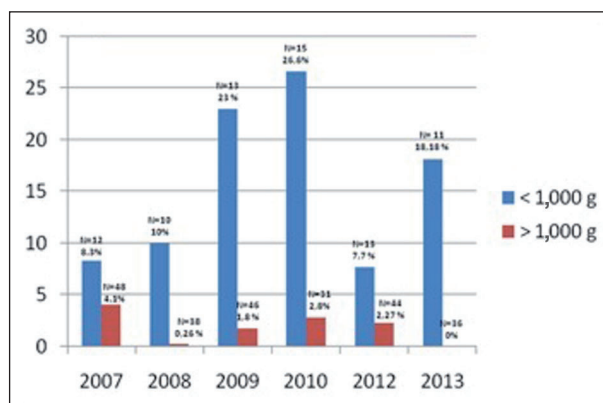
To analyze the incidence and severity of ROP in a third level hospital. We present a retrospective descriptive study of the most severe ROP, divided according sections weight from 2007 to 2013. All the premature infants who survived  $\geq 28$  days of life were included. Inclusion criteria: preterm  $\leq 32$  weeks and/or  $< 1,500$  g and hemodynamic instability or faculty decision. Data were compared to the results published by a working group of the Spanish Society of Neonatology (SEN1500).

**RESULTS**

**Fig. 1** shows the incidence of severe retinopathy according to birth weight from 2007 to 2013.

**CONCLUSIONS**

Even though the number of cases is not big enough, the incidence of global ROP shows a lower incidence but severe retinopathy remains stable. It is more frequent among infants with lower birth weight. In Spain we do not have data to analyze the repercussion of the new prevention strategies such as the better prenatal assistance, the minimal use of  $\text{FiO}_2$  and invasive ventilation, blood transfusions protocols. Data reported by SEN1500 suggest a lower incidence of severe ROP in the last years. Increased rates of survival of newborns with lower gestational ages might influence in the incidence of ROP. It is difficult to separate iatrogenic and intrinsic causes of ROP, especially in extremely low birth weight infants undergoing invasive treatments and procedures: however, these procedures are necessary for the survival of the premature infants. Finally, although further studies are needed, data about the successful treatment with intravitreal monoclonal antibodies (bevacizumab) or associated



**Figure 1 (ABS 3).** Incidence of severe retinopathy of prematurity (ROP) according to birth weight at Reina Sofia University Hospital in Córdoba, Spain.

to photocoagulation therapy are available. These treatments might reduce the more severe stages of ROP.

**ABS 4****TRANSIENT ABNORMAL MYELOPOIESIS AND HYDROPS FETALIS WITH DOWN SYNDROME IN A PRETERM INFANT AND ABNORMAL PLACENTAL FINDINGS**O. Turan<sup>1</sup>, D. Anuk Ince<sup>1</sup>, L. Olcay<sup>1</sup>, S. Agva<sup>2</sup>, O. Ote<sup>3</sup>, Z. Yılmaz Çelik<sup>3</sup>, A. Ecevit<sup>1</sup><sup>1</sup>Department of Pediatrics, Division of Neonatology, University of Hospital of Baskent, Ankara, Turkey<sup>2</sup>Department of Pathology, University of Hospital of Baskent, Ankara, Turkey<sup>3</sup>Department of Medical Genetics, University of Hospital of Baskent, Ankara, Turkey**INTRODUCTION**

Transient myeloproliferative disorder (TMD) of Down syndrome (DS), also known as transient abnormal myelopoiesis, is a characterized by proliferation of myeloblasts in peripheral blood and bone marrow. Its only occurs in the fetal or neonatal period of life and spontaneous resolution regardless of therapy in most cases. Herein, we present a case of preterm infant with DS and TMD successfully treated with exchange transfusion and chemotherapy. We also emphasize the importance of pathological examination of the placenta in newborn infants with TMD of Down syndrome.

**CASE REPORT**

A 27-year-old gravid 1 mother delivered by cesarean section a male baby at 32 of weeks ges-

tation, with a birth weight of 2,665 g. Antenatal ultrasound revealed hydrops fetalis. On admission our patient had ascites, hepatosplenomegaly, systolic murmur, abdominal distension, hypotonia, generalized skin edema and dyspnea. He showed typical signs consistent with trisomy 21 confirmed by karyotype analysis (47, XX, +21). However, cytogenetic analysis of *GATA1* mutation could not be performed in our case. Peripheral blood test results revealed a white blood cell (WBC) count of 139,000/mm<sup>3</sup> with 38% of blasts. At immunophenotype on flow cytometry, blasts were positive for CD34, CD41a, CD117 and CD61. Echocardiography revealed a patent ductus arteriosus and pericardial effusion. Placental pathology showed that weight of placenta: 1,101 g (placentomegaly), hydrops of chorionic villus, and atypical hematopoietic cells (positive for CD61 and CD34). The treatment of the newborn was initiated with exchange transfusion due to anuria and hyperleukocytosis. Moreover, he was put on treatment with cytosine-arabioside (Ara-C) and prednisolone. Peripheral blasts disappeared after the initiation of chemotherapy. The patient is still in remission for 10 months.

#### CONCLUSIONS

The patients with TMD may develop abnormal myelopoiesis within the first four years of life. The most significant factors for mortality in this disease are prematurity, hyperleukocytosis, ascites, coagulopathy, hepatic and renal dysfunction. Our patient had the vast majority of risk factors. The placenta of our patient's mother was enlarged and edema was marked. Chorionic villi contained proliferating blast cells. These proliferating blast cells were not present in the maternal space of the placenta. Our patient was probably born alive because of this reason. Even our patient was a very complicated case; he was successfully treated and discharged.

#### ABS 5

#### BLOOD TRANSFUSIONS: INDICATIONS AND COMPLICATIONS IN THE NEONATAL INTENSIVE CARE UNIT OF CAIRO UNIVERSITY PEDIATRIC HOSPITAL

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#### INTRODUCTION

Blood and blood products transfusions in the Neonatal Intensive Care Unit (NICU) are lifesaving. Transfusional policies in NICUs are not well established. Better practices rely not only on choosing the transfusion guidelines, but also on how to avoid associated risk factors in neonates. The aim of this work is to study prospectively blood transfusions in the NICU of Cairo University Pediatric Hospital, over a period of six months in order to identify the most prevalent indications and complications.

#### METHODS

Our study included 131 neonates whose guardians consented to enrollment. All patients included in the study were subjected to: comprehensive history, clinical examination of neonates on admission, before and after transfusion, laboratory investigations and imaging.

#### RESULTS

Forty-three patients were preterm infants and 88 full terms. Males were more common than females (73 versus 58). According to body weight, 23 had weights below 1.5 kg, 32 weighed between 1.5-2.5 kg and the 76 weighed more than 2.5 kg. Cesarean section was more predominant than vaginal delivery (82 versus 49). Important indications for blood transfusion included severe anemia, failure of weight gain, improvement while ventilated and heart failure. Complications included electrolyte imbalance, sepsis and thrombocytopenia University of Hospital of Baskent, Ankara, Turkey. Being

**Table 1 (ABS 5).** Frequency of blood transfusion and different parameters.

	Single blood transfusion	Multiple blood transfusion	p-value
Ventilated	21	20	<b>0.003</b>
Not ventilated	73	17	
Sepsis	17	15	<b>0.016</b>
No sepsis	74	25	
< 37 weeks gestation	28	15	0.147
> 37 weeks gestation	64	24	
Male	53	20	0.904
Female	39	19	
< 1.5 kg	15	8	0.202
1.5-2.5 kg	23	9	
> 2.5 kg	54	22	
Vaginal delivery	34	15	0.109
Cesarean section	58	24	

Significant p-values are highlighted in bold.



mechanically ventilated or septic were statistically relevant risk factors for frequency of transfusions while sex, age, weight and delivery mode were not (**Tab. 1**). Of the cases 88.7% were discharged alive and 11.8% died with the p-value of 0.0002.

#### CONCLUSIONS

Blood transfusion is common and necessary in NICU. Better-individualized protocols addressing also risks of transfusions are essential to minimize complications and improve outcome.

#### ABS 6

### EARLY TREATMENT OF HYPERBILIRUBIN-AEMIA WITH INTRAVENOUS IMMUNOGLOBULINS IN ABO OR Rh ISOSENSITIZATION OF THE TERM NEWBORNS

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#### INTRODUCTION

Haemolytic disease of the newborn secondary to ABO or Rh isosensitization represents a subgroup of newborns with hyperbilirubinaemia. Standard therapy includes phototherapy and exchange transfusion with a possibility of application of intravenous immunoglobulins (IVIG). The mechanism of action of IVIG is thought to reduce the breakdown of red blood cells by blocking Fc receptors on macrophages and enhancing clearance of maternal antibodies. Several studies have looked at the efficacy of IVIG, the potential to avoid exchange transfusion, side effects and optimal regime and systematic reviews have concluded it might be a useful intervention but the current data is limited and further research is needed. Nevertheless it is recommended by both, NICE and AAP.

#### METHODS

Case series of 4 term infants, 38-39 weeks of gestation and 3,180-3,630 g birth weight with positive Coombs test and low albumin between 27-28 g/l. Total bilirubin level was obtained within 6 hours (average 136  $\mu\text{mol/l}$ , **Tab. 1**) and all infants were started on phototherapy and hydration. In addition, all 4 infants received 0.5-1 mg/kg IVIG at average 8 hours as their bilirubin levels were within 34-51  $\mu\text{mol/l}$  of the exchange transfusion threshold (150  $\mu\text{mol/l}$  at 6 hours). They received the second dose 12 hours after the first.

**Table 1 (ABS 6).** Levels of bilirubin at 6, 12 and 24 hours after delivery in 4 term newborns with isosensitization. Exchange transfusion threshold is also presented [1].

Patient	Isosensitization type	Bilirubin ( $\mu\text{mol/l}$ )		
		6 hours	12 hours	24 hours
1	O-A	124.8	152.1	184.6
2	O-B	145.3	188.1	215.4
3	Rh	118.0	137.0	177.8
4	O-A	143.0	154.0	210.3
Exchange transfusion threshold		> 150	> 200	> 300

#### RESULTS

Bilirubin levels were monitored at 12 hours with average 156  $\mu\text{mol/l}$  and 24 hours with average 189  $\mu\text{mol/l}$  (**Tab. 1**). All levels were below the exchange transfusion threshold and further monitoring showed decline in bilirubin levels.

#### CONCLUSION

All 4 infants with confirmed haemolytic disease of the newborn and levels of bilirubin below the exchange transfusion but within IVIG threshold were treated with two doses of IVIG in addition to phototherapy and hydration. None of the infants required exchange transfusion, the time of phototherapy was shortened, time of hospitalization was lower and no side effects were noted.

#### REFERENCES

[1] American Academy of Pediatrics Subcommittee on Hyperbilirubinemia. Management of hyperbilirubinemia in the newborn infant 35 or more weeks of gestation. *Pediatrics*. 2004;114(1):297-316.

#### ABS 7

### EVALUATION OF INCIDENCE AND RISK FACTORS FOR RETINOPATHY OF PREMATURITY (ROP) IN GREEK PREMATURE INFANTS

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#### INTRODUCTION

Retinopathy of prematurity (ROP) is an eye disorder that affects the developing immature blood vessels

of the retina in premature neonates. It can result in mild and spontaneously resolved disease or in a progressive disease with retinal detachment and blindness. Low gestational age, low birth weight, sex, oxygen therapy and its duration, anemia and blood transfusion are known risk factors of this disease.

#### AIMS

To investigate the prevalence and risk factors for ROP among preterm infants in our neonatal intensive care unit.

#### METHODS

All neonates with gestational age < 32 weeks and birth weight  $\leq$  1,500 g that were hospitalized in our neonatal intensive care unit from January 2014 to March 2016 (n = 40) were enrolled in this study and were examined for ROP. Possible risk factors were recorded and analyzed.

#### RESULTS

Among 40 premature infants, which met the criteria for retinal examination, ROP was diagnosed in two (5%). One of them had ROP stage I and the other ROP stage II. Both neonates with ROP were males; they had history of blood transfusion and respiratory distress syndrome (one had bronchopulmonary dysplasia). ROP was treatable in both cases. None of the infants had severe ROP. Mean gestational age (GA) and birth weight (BW) was lower in the ROP-affected infants compared to non-affected infants,  $27.5 \pm 0.7$  weeks and  $965 \pm 77.8$  g versus  $28.9 \pm 1.7$  weeks and  $1,234 \pm 226.9$  g, respectively ( $p = 0.256$  and  $p = 0.064$ ). The mean oxygen therapy was  $89.1 \pm 79.2$  and  $14.0 \pm 19.9$  days ( $p < 0.05$ ), in ROP group and healthy controls, respectively.

#### CONCLUSIONS

The incidence of ROP is relatively low in premature children in our neonatal intensive care unit. This result could be attributed to our policy that involves strict control of  $\text{FiO}_2$  and  $\text{SaO}_2$ , as well as the duration of oxygen therapy. Very low birth weight and prolonged duration of oxygen therapy were significant risk factors for ROP.

#### ABS 8

#### EVALUATION OF CORRELATION BETWEEN PRENATAL AND POSTNATAL ULTRASONOGRAPHIC FINDINGS OF URINARY TRACT DILATION

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#### INTRODUCTION

Nephrourological anomalies are a major cause of renal dysfunction in childhood. Our objective was to determine the prevalence of urinary tract dilation (UTD) prenatally and assess the degree of correlation between pre- and postnatal ultrasonographic findings.

#### METHODS

Descriptive observational study of fetuses with renal pathologies in a tertiary hospital from January 2015-April 2016. Data concerning the ultrasonographic findings, gestational age (GA), presence of other malformations and prenatal additional studies were collected. All newborns were evaluated with abdominal ultrasound (US) examination in the first 6 weeks of life.

#### RESULTS

We evaluated 113 pregnant women with prenatal diagnosis of fetal malformation. The diagnosis was established at a mean age of 24.1 weeks of GA and received counseling in perinatal consultation in 48% of cases. Most infants were born at term ( $39.2 \pm 2$  weeks) with adequate weight ( $3,180 \pm 45$  g). Nephrourological abnormalities were detected in 45% of women; the most frequent was UTD (56.8%). The UTD was predominantly unilateral (69.3%) and mild (anterior-posterior renal pelvic diameter (APRPD) 16 mm in 28.2% and 23.8%, respectively). The 66.7% of UTD were confirmed. Most mild UTD disappeared (55%) or remained lower (30%) in early postnatal stage. Nevertheless, only 13.6% of moderate-severe UTD disappeared, remaining the majority (54%) with renal pelvis > 10 mm. Cardiovascular malformations were the most frequently associated with nephrourological abnormalities. No chromosomal abnormalities were detected.

#### CONCLUSIONS

The largest proportion of mild UTD was associated with spontaneous resolution and they were considered as transient dilatation. It was detected a progression to a greater degree in 15% of cases. On the contrary, moderate-severe dilations are mostly

confirmed, and they may correspond to diseases of the urinary tract in children. Prenatal US is a useful tool for anticipate the evolution of UTD.

## ABS 9

### NEONATAL ALLOIMMUNE THROMBOCYTOPE- NIA DUE TO MATERNAL ANTI-HLA B49 AND CREG BW4 ANTIBODIES: A CASE REPORT

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## INTRODUCTION

Although neonatal alloimmune thrombocytopenia (NAIT) due to human platelet antigens (HPA) is well described, the pathogenetic mechanism and complete identification of maternal anti-human lymphocyte antigen (HLA) antibodies causing NAIT is yet to be established.

## CASE REPORT

A 40<sup>+5</sup> week-old boy born to G2 P0 A0 mother developed early onset purpura (2 hours of life) due to thrombocytopenia without any evidence of other causes. Platelet alloantibody in mother's serum testing was performed using the Pak ELISA (Immucor GTI Diagnostics, WI, USA). The presence of HLA antibodies was examined in the mother's serum by LABScreen Mixed Kit and LABScreen Single Antigen bead test (One Lambda, CA, USA) in which a 1,000 Median Fluorescence Intensity (MFI) was considered positive. The alleles were characterized by the PCR-SSO method using the LabType SSO Kit (One Lambda, CA, USA) and a LABScan 100 (Luminex Corporation, Texas, USA). The detection of platelet alloantibodies in mother's serum showed no anti-HPA specific antibodies but very high MFI valued anti-HLA antibodies. In particular the tests showed that anti-HLA B49 and Cross-Reactive Group (CREG) Bw4 antibodies had the corresponding paternal antigen in the newborn's serum.

## CONCLUSIONS

This report documents a patient with NAIT induced by maternal IgG anti-HLA B49 and CREG Bw4 alloantibodies without any evidence of HPA alloantibodies. This is the first case described involving anti-HLA B49 antibodies in NAIT. In our report

we assume that the extremely high IgG MFI value has had a role in the pathogenetic mechanism of the NAIT. Therefore, the possibility that a high MFI value contributes to the onset of anti-HLA antibodies NAIT should be reconsidered. Further studies are necessary to identify the role of anti-HLA antibodies in NAIT.

## ABS 10

### DEEP VEIN THROMBOSIS IN A LATE PRE- TERM: A CASE REPORT

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## INTRODUCTION

Renal vein thrombosis is the most frequent thromboembolic event unrelated to vascular catheters thrombosis, with highest incidence in neonatal period. Neonatal incidence of thromboembolic accidents is 5 to 100,000 live birth; 40% of this are located in the renal veins. Clinical features include renal mass, hematuria, hypertension, thrombocytopenia, oligoanuria, and decreased renal function. Abnormal Doppler ultrasound scanning results confirm diagnosis. Risk factors include shock, dehydration, maternal diabetes mellitus, traumatic births, perinatal asphyxia, polycythemia, cyanotic heart disease, sepsis. There are few data in literature regards inherited prothrombotic abnormalities. Some of them have been described in case reports, but the prevalence of these disorders has not been studied in a cohort of patients.

## CASE REPORT

We report a premature female newborn, 36 weeks gestational age, from thrombophilic mother, with antithrombotic treatment during pregnancy, who developed, in the first 48 hours after birth, jaundice and thrombocytopenia, then hematuria and a renal mass in the left flank. Doppler ultrasound scanning highlighted left renal vein thrombosis. The treatment consisted in unfractionated heparin (APTT was checked every 4 hours), followed by low molecular weight heparin. In the next days the thrombotic process was extended to the inferior vena cava, left femoral vein and external left iliac vein. Finally right renal vein was affected.

Thrombophilic mutation analysis confirmed factor V (Leiden) mutation.

#### CONCLUSIONS

Although infants with this presentation are typically treated with anticoagulation, there is a lack of evidence-based guidelines. A case-control study is required to investigate whether maternal prothrombotic disorders may be a risk factor for neonatal thrombotic processes.

#### ABS 11

### RED BLOOD CELLS TRANSFUSIONS IN VERY LOW BIRTH WEIGHT NEONATES

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#### INTRODUCTION

Very low birth weight preterm infants are associated with greater risk of neonatal morbidity and mortality and are more susceptible to red blood cells transfusions. The aim of this study is to analyze the prevalence and risk factors associated with red blood cells transfusions.

#### METHODS

Retrospective study developed at a Level III Neonatal Intensive Care Unit including preterm infants with very low birth weight admitted between November 2011 and November 2015. Those who died were excluded. Demographic, perinatal and clinical data during hospitalization were recorded. Univariate and multivariate analysis were performed to compare neonates with and without need for red blood cells transfusion and to find predictor factors.

#### RESULTS

A total of 79 neonates were included, with median birth weight of 1,190 grams and a mean gestational age of  $29 \pm 2$  weeks. Forty-nine (62.02%) of them had received transfusion support with red blood cells. Higher need for red blood cells transfusions were significantly associated with lower birth weight (OR = 0.99; 95% CI: 0.990 to 0.999) and greater total milliliters of phlebotomy losses (OR = 1.17; 95% CI: 1.07 to 1.28) during their stay. Birth weight (B = -0.01; 95% CI: -0.008 to -0.003), hemoglobin at admission (B = -0.33; 95% CI: -0.53 to -0.13) and sepsis (B = 1.85; 95% CI: 0.72 to 2.98) were predictive factors for the number of red blood cells transfusions. Regarding the treatment with

erythropoietin, there were no differences between the two groups for the number of transfusions and other outcomes.

#### CONCLUSIONS

A considerable number of very low birth weight neonates were exposed to transfusions with red blood cells. Phlebotomy losses are one of the major factors for the need of transfusion in preterm infants, and therefore sampling should be minimized to the most.

#### ABS 12

### MAINZER-SALDINO SYNDROME: A RARE CAUSE OF CONGENITAL RENAL FAILURE

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#### INTRODUCTION

Mainzer-Saldino syndrome is an autosomal recessive inheritance disorder characterized by chronic kidney disease, eye problems and skeletal abnormalities.

#### CASE REPORT

Family history: consanguinity. A brother died due to malformation syndrome, with renal dysplasia, chronic renal failure, craniosynostosis and cholestasis. A 4-day-old female was admitted in our neonatal unit because of renal failure related to renal dysplasia. The patient presented stage V chronic renal failure since birth, and tubulointerstitial nephritis. Peritoneal dialysis started at 9 days of life. Renal ultrasound: small hyperecogenic kidneys, without cortical-medullary differentiation and small cortical cysts. Hepatomegaly and cholestasis since birth. Liver biopsy showed canalicular cholestasis, ductal proliferation and fibrosis. Continuous horizontal nistagmus and delayed ataxic sitting position. Retinal study was normal at follow up. Bone study showed metaphysis proximal phalanges first cup-shaped (**Fig. 1**), narrow thorax and 11 pairs of ribs. On echocardiography an interauricular communication were detected. Genetic study showed normal karyotype, *JAG1* gene was studied to discard Alagille syndrome; and CGHarray on fibroblasts culture, without change on genic dose. Many heterozygosity loss regions were observed,





**Figure 1 (ABS 12).** Typical radiological findings in the Mainzer-Saldino syndrome.

related to consanguinity and highly suggestive of recessive genetic defect.

#### CONCLUSIONS

Our patient showed the clinical and radiological findings compatible with Mainzer-Saldino syndrome. Without renal biopsy or genetic study, confirmation of diagnosis is not possible, although clinical presentation is highly suggestive. The main symptom is the nephronophthisis and the fast evolution to chronic renal disease, associated with nearly all the extra renal manifestations (hepatic cholestasis and fibrosis, cup-shaped phalanges and cerebellar ataxia). Consanguinity and the presence of a previous brother with non-filiated malformative syndrome support the diagnosis.

#### ABS 13

#### NEONATAL ALLOIMMUNE THROMBOCYTOPENIA: TWO CASE STUDIES

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#### INTRODUCTION

Incidence of thrombocytopenia in healthy term infants is around 1%, while in premature infants it is the most common hematological abnormality. Thrombocytopenia is defined as number of the thrombocytes below 150,000/ $\mu$ L, but many healthy newborns can have lower value of thrombocytes (100,000/ $\mu$ L), without clinical signs of disease. It can be caused by illness of the mother, conditions of placenta, illness of the newborn and dysfunction of thrombocytes. Neonatal alloimmune thrombocytopenia begins with alloimmunization of a mother during pregnancy with fetal thrombocytes antigens inherited from the father. The incidence is 1:11,000 newborns, 20-60% with first pregnancy and it is most common reason for cerebral hemorrhage linked with thrombocytopenia with on term born infants. Around 10% of newborns affected with this disease have neurological consequences. In Caucasian population HPA-1a antibodies are cause of thrombocytopenia in 80-90% of cases, anti HPA-5b in 5-15% of cases, while other antibodies are rarely found.

#### CASE REPORTS

We report two newborn infants who have been diagnosed isolated thrombocytopenia with thrombocytes value below 20,000/ $\mu$ L, with clinically visible petechiae and hematomas of soft tissue, without gastrointestinal or intracranial hemorrhage. An emergency transfusion of concentrate of irradiated thrombocytes was performed. After the transfusion there was no clinically visible new hematoma and laboratory results of thrombocytes did not require new transfusions. Serological laboratory testing on neonatal alloimmune thrombocytopenia (NATP) with both newborns was positive. With first infant it was proven the existence of HPA-1a antibodies and with second infant HPA-1b antibodies.

#### CONCLUSIONS

NATP is a rare occurrence: seriousness of the clinical condition and potential intracranial hemorrhage connected with thrombocytopenia impose an early diagnosis and treatment of this disease.

#### ABS 14

#### NEONATAL HEMOLYTIC ANEMIA REVEALED BY EFFUSION SYNDROME: REPORT OF TWO CASES

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## INTRODUCTION

Hemolytic disease of the newborn is a serious disease, affecting 3/100,000 to 80/100,000 patients per year. We report two cases of hemolytic anemia revealed by effusion syndrome.

## CASE REPORT

First case presentation: a premature newborn of 34 weeks was admitted for fetal hydrops with anemia (hemoglobin at 7.8 g/dl, MCV = 108, MCH = 36) followed by intense jaundice. The management consisted of a blood transfusion; phototherapy, a thoracentesis and ascites puncture with a good clinical outcome. No etiology was found.

Second case presentation: a newborn was admitted on second day of life for right pleural effusion. The analysis of the pleural fluid after aspiration confirmed a chylothorax. The day after his admission he developed jaundice with anemia (hemoglobin at 7.9 g/dl, MCV = 110, MCH = 36). The blood tests showed rhesus incompatibility with a positive Coombs test. He was treated with blood transfusion and phototherapy with a good clinical outcome.

## CONCLUSIONS

Neonatal hemolytic anemia is a particular entity due to risk of rapid onset of kernicterus and the mode of expression of this hemolysis. The onset of an effusion syndrome is an exceptional situation whose etiological research remains challenging.

## ABS 15

### **ABOUT A RARE CASE OF NEONATAL VOLKMANN SYNDROME REVEALING NEONATAL THROMBOPHILIA**

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## INTRODUCTION

Neonatal Volkmann syndrome is a rare and difficult diagnosis, suspected from the association of skin

lesions and motor weakness or paralysis of the wrist and hand.

## CASE REPORT

We report an unusual case of a newborn presenting a Volkmann Syndrome. He was admitted in neonatal intensive care unit at day 7 of life. He presented a rash in the first hour of life at the posterolateral side of the left forearm. The evolution was marked by necrosis of the hand and the forearm. In the past medical history, no necking by the umbilical cord or use of toxic drugs or intravenous fluids in the affected limb was reported. Examination found extended necrosis from the forearm to the back of the hand with total necrosis of the left fourth finger with one aspect of the claw fingers and the absence of flexion and active extension of the wrist and fingers. The blood tests revealed neonatal thrombophilia with protein C and S deficiency, the platelets level was normal and the swab skin was sterile. No amniotic band was found and the angio-CT scan did not reveal any thrombosis. We performed a necrosectomy and mechanical debridement and directed healing as well as the splint establishment posture with a thin skin transplant later after one month of life.

## CONCLUSIONS

This case reports an unusual association between neonatal Volkmann syndrome and neonatal thrombophilia, never reported before in other previous studies, and focuses on the challenging diagnosis of the neonatal Volkmann syndrome and its unknown cause and enhances the rapid evolution to necrosis in case of delayed care.

## ABS 16

### **FUNCTIONAL STUDY OF THE KIDNEY TUBULES IN IUGR NEWBORNS, USING NGAL, THE NOVEL BIOMARKER OF KIDNEY INJURY**

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## BACKGROUND

As very well known, the key role in the etiology of Intrauterine Growth Restriction (IUGR) is played by chronic hypoxia associated with placental insufficiency. IUGR it often leads to various morphological and functional changes in fetal organs and tissues. One of the most sensible organs to the chronic hypoxia is the kidney, and its injury often remains unidentified due to lack of symptoms.

**AIMS**

The aim of this investigation was the study of the functional kidney condition of IUGR newborns. For this purpose we used the novel biomarker of early kidney injury NGAL (Neutrophil Gelatinase-Associated Lipocalin).

**METHODS**

Clinical and laboratory analyses were carried out on 49 newborns with GA = 29-36 weeks. They were divided into 2 groups: main group – preterm newborns with IUGR (n = 33), and control group – term infants without IUGR (n = 16). Urinary and serum samples were collected on days 1-3 and 7-10 of their life. Serum and urinary NGAL were quantified by ELISA. Results were compared by Mann-Whitney test.

**RESULTS**

Levels of uNGAL and sNGAL were significantly higher in main group of infants compared to the control group,  $171.06 \pm 58.58$  ng/mL and  $205.31 \pm 43.61$  ng/mL accordingly on 1-3 days, and despite the moderate decrease at the end of early neonatal period, levels of uNGAL and sNGAL remained high  $170.03 \pm 67.46$  ng/mL and  $158.37 \pm 78.4$  ng/ml accordingly on the days 7-10 ( $p \leq 0.05$ ).

**CONCLUSIONS**

Chronic antenatal hypoxia and immaturity of renal tissues and cells are characteristic to IUGR newborns who are predisposed to the development of tubular dysfunction, and a biomarker such as NGAL could be used as a predictive and informative tool for defining a tubular injury.

**ABS 17****POSTNATAL OUTCOME OF ANTENATALLY DIAGNOSED RENAL PELVIS DILATATION: A RETROSPECTIVE STUDY**

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**AIMS**

The aim of this study was to determine the occurrence and postnatal outcomes of neonates with antenatally diagnosed renal pelvis dilatation (RPD) delivered at tertiary institution of South-East Asia.

**METHODS**

The study included infants with antenatal diagnosis of RPD > 5 mm born during period from January 2011 to

December 2014. Postnatally, using standardized departmental protocol, RPD was classified as mild (5-9.9 mm), moderate (10-14.9 mm), and severe ( $\geq 15$  mm). Isotope (MAG3) scan and micturating cystourethrogram (MCU) were performed, where appropriate. Outcomes evaluated were resolution of RPD, presence of structural urological anomalies and need for surgical intervention.

**RESULTS**

The study included 134 newborn infants. Bilateral RPD was present in 45 (33.6%) infants. The RPD was isolated finding in 117 (87.3%) infants, whereas it was associated with calyceal dilatation in 13 (9.7%) and ureteric dilatation in 4 (3.0%) cases. Mild renal pelvis dilatation was present in 85 (63.4%), moderate in 31 (23.1%) and severe in 18 (13.4%) cases. Complete resolution of RPD (< 5 mm) occurred in 55 (64.7%) in mild, 8 (25.8%) in moderate, and none in severe groups ( $p < 0.005$ ). Partial or complete pelviureteric junction obstruction was found in 12 cases, of which, 9 (75%) were from the moderate and severe groups. Structural uropathies observed in 14 (10.4%) cases include duplex kidney (3), horseshoe kidney (2), megaureter (1), multicystic dysplastic kidney (1), posterior urethral valve (2), vesico-ureteric reflux (3) and obstructed ureterocele (2). 9 (6.7%) infants needed surgical intervention for their structural urological anomalies.

**CONCLUSIONS**

This study showed that grade of antenatal RPD correlates well with severity of confirmed postnatal abnormalities, with higher rate of resolution of mild RPD. Also, additional abnormalities like dilated calyces and ureter demonstrated a greater association with postnatal urological pathologies, and need for further postnatal evaluation and surgical treatment.

**ABS 18****POLYCYTHEMIA. RISK FACTORS, MORBIDITY AND TREATMENT BEFORE AND AFTER BIRTH GUIDE CHANGE**

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**INTRODUCTION**

Neonatal polycythemia (NP) is frequently found in newborns (2-5%). Clinical findings and need

to treatment are, on the other hand, less common. One of the main risk factor is timing of umbilical cord clamping. In this study our aim was to analyze epidemiological, clinical and therapeutic factors in admitted newborns because of NP in our neonatal unit between January 2011 to December 2013.

#### METHODS

We studied the differences between two groups before and after changing our birth guidelines in January 2012 (group 1: cord clamping when cord beating ends, from January 2011 to December 2011; group 2: cord clamping until 2 minutes at most, from January 2012 to December 2013). Data reporting: digital medical history of pregnancy and birth. Perinatal risk factors, neonatal morbidity and treatment received. Statistical analysis: STATA 12.0

#### RESULTS

We reported 47 newborns of group 1 and 54 in group 2. Medium weight at birth was 3,023 g in group 1 and 2,915 g in group 2. Maternal mean

age was 32.2 and 33.17 years, respectively. No differences were found in perinatal risk factors, maternal pathology, birth and comorbidity (age and length of hospitalization and associated symptoms). Incidence of NP was reduced after new birth guide in 36% (RR 0.64, CI 95% 0.43-0.94;  $p = 0.023$ ). Compared with 2011, incidence in 2012 decreased in a 31% (RR 0.69, CI 95% 0.44-1.08;  $p = 0.112$ ) and in 2013 it decreased in a 43% (RR 0.58, CI 95% 0.35-0.94;  $p = 0.029$ ). Main results are presented in **Tab. 1**. Partial exchange transfusion was necessary in 14.9% of cases of group 1 versus 3.7% in group 2, with RR 0.22 (CI 95% 0.05-0.99), so risk decreased to 78%.

#### CONCLUSIONS

Changes in our birth guidelines, consisted of cord clamping at most 2 minutes (instead of at the end of cord beating), reduced symptomatic NP risk and need to partial exchange transfusion in our group of patients.

**Table 1 (ABS 18).** Comparison of number of neonatal polycythemia before and after the new birth guidelines in 2012.

	Polycythemia	No polycythemia	Total of births
Cord clamp when cord beating ends (2011)	47	2,442	2,487
Cord clamp at most 2 minutes (2012-2013)	54	4,427	4,481