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Editorial

Genetics and acronyms

Giovanni Corsello

Pediatrics and Neonatal Medicine, University of Palermo, Italy

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"Nomen omen."
Old Latin phrase

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Corresponding author

Giovanni Corsello, Pediatrics and Neonatal Medicine, University of Palermo, Italy; email: giocors@alice.it.

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In a global society as the present, the nomenclature and terminology of diseases must be universally accepted among the specialists. This sentence is particularly true in some fields of medicine, as genetics, in which the progress of knowledge has been particularly rapid in last years. The high number of diagnostic tests in genetics has also changed in many cases the approach to patients affected in terms of clinical management and prevention.

Many genetic disorders were termed using the names of the doctor (or the doctors) who discovered and described them. This happened in many other fields of medicine and in the XX century became a common mode to declare most of diseases. In this way many diseases became eponymic. The doctors publishing the paper with the description of a new disorder had their names as eponyms, because the scientific community attributed the names to that disease. Sometimes this right was not so clearly accepted and debates about the time of publication arose among different groups of researchers. When the debate prolonged, a multiple choice with more names one after the other was frequently adopted to avoid public conflicts.

The name of doctors and specialist were also frequently used to term sign and symptoms of diseases, including genetic syndromes. This revealed particularly useful when its description was difficult to perform. The eponym achieves the result of a better comprehension of clinical data in patients affected by rare as well as common diseases. In some cases, an eponymic disease showed some eponymic signs in its clinical spectrum, useful to a better recognition of diseases and patients. It is easier to understand and communicate a clinical combination of features when we say Babinski or Brushfield than the prolonged description of the events.

More rarely, a new disease received the name of the first patients described. The Pierre Robin sequence was one of the rare cases with the semantic origin of its eponym related to patients. Its name was given to this disease from the name of the first young man with the typical malformations of mandible and oral region. When the syndrome of Smith-Lemli-Opitz was delineated in 1963 as an autosomal recessive disorder with multiple congenital anomalies, mental retardation and severe growth delay, John Opitz proposed to term it as RSH syndrome, from the initial of the names of the first three patients described in the original paper. In a recent paper the disease has been defined as RSH/Smith-Lemli-Opitz to put together the acronym and the eponym. The times were not mature to understand this ethical point of view and only later medicine evolved towards bioethics and communication. Winckler in his book "The Sach's disease" defines the use of eponyms to mark the diseases as the symbol of a primacy of doctors with respect of patients and asks to forget this method and to change this use in the future.

In effect, when the number of genetic diseases began to be particularly high and rapidly increasing, due to the results of diagnostic cytogenetic and molecular testing, it was always more difficult to find eponyms. In some cases the authors clearly proposed the acronyms, originated from the initial of main signs and symptoms. as a useful and efficacious system to term new diseases. One of the advantages of the acronyms was the flexibility, the possibility to modify it in relation to a new sign discovered in subsequent papers. This happened for example to the VATER association, in which the term originates from the initial of Vertebral, Anorectal, Tracheo-Esophageal and Renal abnormalities, modified in the end of XX century in VACTERL association, adding the Cardiac and Limbs abnormalities so frequent in this multifactorial and heterogeneous disorders of blastogenesis. Other associations of congenital malformations were classified using acronyms. CHARGE association, due to the contemporary presence in a patient of CHoanal Atresia, growth Retardation, Genital and Ear abnormalities. This name remained also when genetic molecular tests showed the genetic basis of this disease with microdeletion of the long arm of chromosome 8, now noted as CHARGE syndrome. In some instances the acronym reproduces a word with other kind of semantic suggestions. It is the example of the LEOPARD syndrome, in which LEntigo, Ocular abnormalities, Pulmonary stenosis, Abnormalities of the cardiac Rhythm and Deafness. We know this syndrome as a clinical and genetic variant of the Noonan syndrome. Also the CFC syndrome, with Cardiac, Facial and Cutaneous abnormalities is now in the spectrum of the Noonan phenotype.

Moreover, some acronyms in their list of initials show also numbers. It is the case of the CATCH22 syndrome, in which Cleft of palate, Aplasia of Thymus, Cardiac defects and Hypocalcaemia are listed with chromosome 22 to mark the related locus of chromosome 22 in which may be find the microdeletion 22q11.21. This syndrome is an example of great both clinical and

semantic heterogeneity. In fact, it may be called as CATCH22, as well as velo-cardio-facial syndrome, Shprintzen syndrome and Di George sequence. The conotruncal hearth defects are the clinical marker of disease also on prenatal diagnosis.

Some acronyms show among the initials also the words related to the physiopathology of disease. It is the case of the APECED syndrome, in which the letter A means Autoimmunity, the letters P and E mean PolyEndocrinopathies, the letter C means Candidiasis of skin and mucosae, in addition to Ectodermal Dystrophia. Also the CAPS syndrome share an acronym related to the pathogenesis of this group of autoinflammatory diseases: it means Cryopyrin-Associate Periodic Syndromes and includes Muckle-Wells syndrome and Familial Cold Auto-inflammatory Syndrome, whose acronym is FCAS.

Acronyms rapidly diffused as a good method to term genetic diseases and syndromes. It was simple, rapidly accepted among the scientific community and easier to modify, if necessary. Recently, some authors observed the risk of an abuse of acronyms. They tend to become too many and sometimes too complex. As in most of the human things, it is necessary avoid the abuse of acronyms. There is a place for historical eponyms, which has been routinely accepted as well as there is a place for acronyms and other system to identify genetic syndrome and diseases. It is necessary that we all always remember that the name of a disease is not a medal or symbol for doctors or scientific societies. On the other hand it is a system to improve all over the world the diagnostic opportunity and clinical management for patients with genetic diseases, carrying out disabilities and handicaps, and for their families.

In more recent years acronyms were proposed to mark multicentric studies, to simplify the terminology and comprehension. Many examples we have of these more transient acronyms, lasting the time of the study although present for a longer time in the medical literature. Some name were chosen with the scope to be attractive: DIAMOND for example means DHA Intake And Measurement Of Neural Development!

In conclusion, among the advantages of the use of acronyms in medical genetics must be listed the simplification of terminology, the easier comprehension, the universal use and the flexibility in terms of possible changes due to an expansion of the phenotype. Numerous potential risks and disadvantages may be related to the use of acronyms. They are their difficult comprehension in medical circles different from those of clinical genetics, possible errors due to overlapping with other acronyms, the looseness of the historical link of diseases. As in most of the scientific areas, it is necessary to avoid oppositions and accept both eponyms and acronyms in synergic or alternative use, considering their helpful contribution to term and define many genetic diseases and various phenotypes with multiple malformations.

Declaration of interest

The Author declares that there is no conflict of interest.