

Genetics and acronyms

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The last ten years, the next ten years in Neonatology

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*“Nomen omen.”
Old Latin phrase*

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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 heart 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.