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Special Issue Reprint

Genetic Testing for Rare Diseases

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Rare diseases, or orphan diseases, are those that individually affect a small number of patients, but taken together affect over 300 million people worldwide. They are characterized by their etiological, diagnostic and evolutionary complexity, important morbi-mortality, with high levels of disability that entail and hinder the development of a normal vital subject, not only in those who suffer from them, but also their families; therefore, a comprehensive social health approach is necessary to address this problem.
About 80% of rare diseases have a genetic origin, mainly monogenic; thus, genetic testing is mandatory for the confirmation of clinical diagnostics and to ensure correct genetic counseling.
Next-generation sequencing (NGS) has enabled a revolution in genetic diseases, specially in rare diseases. However, their complexity makes diagnoses difficult even with the advent of NGS.
In this Special Issue, we present several examples of the complexity of genetic diagnosis for most of these diseases and the consequences that genetic testing implies for genetic counseling. There are examples of the genetic heterogeneity of hearing loss, some metabolic and lisosomal disorders, ataxia, Prader–Willi syndrome, and three comprehensive reviews on syndromic retinal dystrophies, the complexity of the molecular diagnosis of neuromuscular disorders, and the value of genetic counseling before and after a genetic test.

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