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Genetic Testing for Rare Diseases

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Message from the Guest Editor

Rare diseases are defined by their low prevalence but, as a whole, they affect 8–10% of the population in Western countries. Six to seven thousand rare diseases have been reported in the literature, and the number is regularly increasing. There are many differences among them in terms of etiopathology, age of onset, affected organs and tissues, etc., but about 80% of them have a genetic cause.

Therefore, the genetic testing of rare diseases is a key point for confirming the clinical diagnosis, offering the correct genetic counseling to the patients and their relatives and, more recently, the possibility of receiving a

This Special Issue will be focused on clinical genetics, genetic counselling, rare metabolic diseases, sensorineural disorders, neuromuscular diseases, and bioinformatics, among other topics.

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Message from the Editor-in-Chief

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