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Complex Genetic Syndromes in Pediatric Age

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Message from the Guest Editors

Dear Colleagues,

The interest in rare genetic syndromes has greatly increased with the reduction of morbidity and mortality due to infectious diseases. Today, genetically transmitted conditions are a major cause of pediatric morbidity and mortality. A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population. Although they are singularly rare, these conditions affect a large part of the population. The European Organization for Rare Diseases (EURORDIS) estimates that as many as 5,000 to 7,000 distinct rare diseases exist, and as much as 6% to 8% of the population of the European Union is affected by one. Diagnosis means to avoid an excess of medical care, expensive diagnostic processes, and frustration for patients and their families. The advent of arrayCGH before and NGS technologies then has created a great shift in our approach to both the discovery of new disease genes and the time for diagnosis of genetic disease. In this Special Issue we look for examples of the diagnostic complexity of genetic syndromes in pediatric age.