Journal of
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Paediatric Genomics: Diagnosing Rare Diseases in Children

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Message from the Guest Editors

Paediatric rare diseases can impose a substantial medical, financial, and psychosocial burden on patients and families, as a result of the various implications of these conditions. The past decade has been full of medical diagnostic and therapeutic promises because of the completion of human genome project (HGP), a great scientific achievement. Paediatric genomics is rapidly evolving in the 21st century with the advent of advanced genetic testing technology, including Next Generation Sequencing (NGS). A wide variety of sequencing platforms are available such as whole exome sequencing (WES) and whole genome sequencing (WGS) to diagnose and treat rare diseases. For this reason, this special issue will explore the use of such technologies in diagnosing rare genetic conditions in children. Additionally, newer disease specific treatments are being developed, changing the outcome for children with previously untreatable genetic diseases. Scientist and clinicians are encouraged to submit their scientific contributions in Paediatrics genomics, which will further the field of rare disease diagnostics in children.

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Message from the Editor-in-Chief

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