Special Issue

Application of Genetic Diagnosis in Pediatrics

Message from the Guest Editor

Traditional diagnostic approaches are now being replaced by more granular frameworks capable of dissecting anomalies at single-cell resolution. This shift has led to the more effective diagnosis of rare or complex pediatric conditions that previously involved years of inconclusive evaluations. Today, robust high-throughput computational methods are enabling genomic approaches that significantly shorten diagnostic timelines, offering earlier clarity and enabling personalized interventions. This Special Issue will bring together cutting-edge studies that demonstrate the translational value of genetic diagnosis in pediatric care, from identifying actionable mutations to unraveling dysregulated transcriptomic and epigenomic landscapes, thereby informing prognosis, guiding precision therapies, and supporting informed family planning. We particularly welcome contributions exploring the integration of genomic, transcriptomic, and epigenomic data into routine pediatric care, the development of novel diagnostic algorithms, machine learning applications, population-scale pediatric genomics, and ethical or policy considerations in early genetic testing.

Guest Editor

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Message from the Editor-in-Chief

Journal of Personalized Medicine (*JPM*; ISSN 2075-4426) is an international, open access journal aimed at bringing all aspects of personalized medicine to one platform. *JPM* publishes cutting edge, innovative preclinical and translational scientific research and technologies related to personalized medicine (e.g., precision medicine, pharmacogenomics/proteomics, systems biology, 'omics association analysis). *JPM* is covered in Scopus, the Science Citation Index Expanded (SCIE), PubMed, PMC, Embase, and other databases.

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