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Genomic Insights: Exploring the Applications of Sequencing Technologies in Genetics

Guest Editor:

Dr. Elena Bonora

Department of Medical and Surgical Sciences, DIMEC, St. Orsola-Malpighi Hospital, University of Bologna, 40138 Bologna, Italy

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Message from the Guest Editor

Dear Colleagues,

Next-Generation Sequencing technologies can improve our knowledge of the molecular genetics of human diseases, but may also lead to uncertainty during the interpretation of the results. Providing a definitive genetic diagnosis is critical in prenatal testing or management of severely affected paediatric patients and can be especially challenging in affected individuals in whom a broad unbiased sequencing approach such as whole-exome sequencing or whole-genome sequencing is potentially detrimental. Molecular studies are warranted to clarify the genetic basis of human diseases and emphasize the importance of functional studies in the diagnostic variant reclassification process.

Dr. Elena Bonora Guest Editor









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Editor-in-Chief

Prof. Dr. Maurizio Battino

Department of Odontostomatologic and Specialized Clinical Sciences, Sez-Biochimica, Faculty of Medicine, Università Politecnica delle Marche, Via Ranieri 65, 60100 Ancona, Italy

Message from the Editor-in-Chief

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International Journal of Molecular Sciences Editorial Office MDPI, St. Alban-Anlage 66 4052 Basel, Switzerland Tel: +41 61 683 77 34 www.mdpi.com mdpi.com/journal/ijms ijms@mdpi.com X@IJMS_MDPI