Special Issue

Functional Studies and Deep Phenotyping in Monogenic Forms of Diabetes, Severe Insulin Resistance and Hypoglycemia

Message from the Guest Editor

Monogenic forms of diabetes (MD), severe insulin resistance (SIR), and hyperinsulinemic hypoglycemia (HH) represent a sizeable number of cases in neonates, children, and young adults with these conditions. With the advent of next generation sequencing, many genetic variants with uncertain significance are discovered in genetic labs every day. One of the tools used to ascertain the pathogenic role of a genetic variant in a given disease are functional studies. Due to the intrinsic nature of proteins involved in MD, SIR, and HH that span enzymes, transciption factors, hormones, receptors, ion channels, and transfer-RNAs, functional studies may consist of biochemical assays, cell biology experiments, in vitro assays of transcription factor activity, animal models like mouse knockouts, and studies in human induced pluripotent stem cells (iPSCs) or human embryonic stem cells (hECSs) manipulted by CRISPR. This Special Issue focuses on any type of functional study commonly applied to support (or exclude) the pathogenic role of a genetic defect in the aforementioned conditions.

Guest Editor

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The International Journal of Molecular Sciences (*IJMS*, ISSN 1422-0067) is an open access journal, which was established in 2000. The journal aims to provide a forum for scholarly research on a range of topics, including biochemistry, molecular and cell biology, molecular biophysics, molecular medicine, and all aspects of molecular research in chemistry. *IJMS* publishes both original research and review articles, and regularly publishes special issues to highlight advances at the cutting edge of research. We invite you to read recent articles published in *IJMS* and consider publishing your next paper with us.

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