Special Issue

Novel Genetic causes of Pituitary Hormone Deficiency

Message from the Guest Editor

Research over the last 20 years has elucidated the genetic etiologies of Combined Pituitary Hormone Deficiency (CPHD). The pituitary plays a central role in growth regulation, coordinating the multitude of central and peripheral signals to maintain the body's internal balance. Naturally occurring mutations in humans and in mice have demonstrated roles for several factors in the etiology of CPHD. Depending upon the expression patterns of these molecules, the phenotype may consist of isolated hypopituitarism, or more complex disorders such as septo-optic dysplasia (SOD) and holoprosencephaly. More recently, mutations in genes involved in Kallmann syndrome such as PROKR2 were also reported in CPHD, suggesting a potential role for the PROK2 pathway in pituitary development. Although numerous monogenic causes of CPHD have been identified, most patients remain with an unexplained etiology as shown by the relatively low mutation detection rate. The introduction of novel diagnostic approaches and NGS(next-generation sequencing) technology is now leading to the disclosure of novel genetic causes in disorders characterized by pituitary hormone defects.

Guest Editor

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