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Editorial

Special Issue "Genetics in Pediatric Endocrinology"

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The inception of pediatric endocrinology in the United States began little less than a century ago, but it has grown as a subspecialty field since the 1950s. Initial innovative research endeavors in 1980s leading to the development of radioimmunoassays (RIAs) for measuring hormones and polymerase chain reaction (PCR) technology provided the initial intersection of this discipline with genetics [1]. From the initial suspicion of the role of DNA and inheritance in endocrine disorders, to an era where the structures and functions of hormones and receptors, along with the evolving role of genetics in their synthesis has improved our understanding of how these affect human growth and development. Research in this area has helped advance diagnostic and treatment options benefitting thousands of pediatric endocrinologists and their patients worldwide. Pediatric Endocrinology Board certification requirements include not only working with a number of endocrine and diabetes patients but also understanding the laboratory assessment of patients, including formal training in performing immunoassays and completing a course in molecular genetics. In this Special Issue of *Endocrines*, we will explore the impact of the genetics revolution on endocrine function. The development of newborn endocrine screening antedated the explosion of applications of molecular genetic analysis and their enormous contributions to diagnosis and targeted treatments of endocrine disorders [2]. The brief review provides updates on the prevalence of congenital hypothyroidism and congenital adrenal hyperplasia. And, how lifesaving and life-enhancing role of screening helped with the discovery and expansion of the knowledge on genetic etiology of these two groups of disorders, and, also of other genetic disorders associated with them.

Improved genetic analysis of syndromes with endocrine manifestations and awareness of the actual prevalence of Turner syndrome (TS), Prader–Willi syndrome (PWS) and others have allowed for associated endocrine disorders to be better understood and helped in avoiding errors of diagnosis in cases mimicking TS or PWS, in which the manifestations differ.

Discovery of the genes involved in MODY (also known as monogenic diabetes) has been of great help in targeting the treatments needed. Multiple endocrine neoplasia management has been enhanced by a better understanding of which elements are involved and the timeline of development in various forms that can be discerned by the affected gene. Knowing that growth hormone insensitivity may be due to a mutation in the GH receptor or the IGF receptor has assisted in counseling and treatment.

The genetic and hormonal contributions to understanding the regulation of calcium and phosphorus have evolved together and facilitated targeted therapy. Congenital hyperinsulinism management, medical or surgical, can be better targeted based on the underlying mutation.

A glimpse of the role of epigenetic differences in altering the gene expression and phenotype presentations noted in TS and central precocious puberty (CPP), as well as the lesser-known role of whole-genome sequencing, may shed light on the genetic etiology of CPP.

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The recently available complete sequence of the human genome and advances in molecular technology, which have enabled whole-genome sequencing, will continue to enhance our understanding of the intersection between genetics and pediatric endocrinology, and may require its own Special Issue in future.

This collection of 11 reviews and a case report, though not an exhaustive review of all the potential topics in the intersection of genetics and pediatric endocrinology, helps address why knowledge of genetics is critical to understanding how endocrine systems develop and function.

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