Special Issue

Clinical and Molecular Diagnosis of Congenital Adrenal Hyperplasia (CAH)

Message from the Guest Editor

Congenital adrenal hyperplasia (CAH) represents one of the most challenging endocrine disorders caused by a group of autosomal recessive diseases related to pathogenic variants in genes encoding enzymes involved in cortisol biosynthesis: 21-hydroxylase (21OH), 11β-hydroxylase (11βOH), 17α-hydroxylase (17OH, also known as 17,20-lyase), 3β-hydroxysteroid dehydrogenase type 2 (3βHSD2), steroidogenic acute regulatory protein (StAR), P450 cholesterol side-chain cleavage enzyme (SCC), and P450 oxidoreductase (POR). Clinical presentation includes a range of clinical and biochemical phenotypes related to severe (classic) or mild (non-classic) forms. Challenges in the treatment of CAH include the avoidance of glucocorticoid overtreatment and the control of sex hormone imbalances. Molecular genetic testing for CAH is offered worldwide, and is of importance for differential diagnosis, carrier detection, and adequate genetic counseling, particularly for family planning. New approaches based on next-generation sequencing (NGS) allow the simultaneous screening of all candidate genes, increasing the efficiency of molecular diagnosis.

Guest Editor

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