Genetic Steroid Disorders: Chapter 3A. Congenital Adrenal Hyperplasia Owing to 21-Hydroxylase Deficiency

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Congenital adrenal hyperplasia (CAH) refers to a group of autosomal recessive genetic disorders that arise from defective steroidogenesis. The 21-hydroxylase deficiency (21OHD) is the most common form of CAH, accounting for more than 90% of cases. It is the most common disorder of sexual development (DSD) in females. The gene is encoded by CYP21A2, which is located on the short arm of chromosome 6 (6p21.3). The activity of the enzyme 21-hydroxylase, encoded by the CYP21A2 gene, is deficient, leading to an accumulation of 17-hydroxyprogesterone (17-OHP) and subsequent elevation of androgens. The three forms of 21OHD are the salt-wasting form, simple-virilizing form, and non-classical form. The first two forms are classical forms of the disease where the hallmark finding is ambiguity of the genitalia in affected female newborns. Patients with the non-classical form have normal genitalia, yet may present with signs of early sexual development and other symptoms of hyperandrogenemia such as short stature, hirsutism, acne, and impaired fertility. Hormonal testing is important in making the diagnosis of 21-hydroxylase deficiency, yet genetic testing is crucial to secure the diagnosis. More than 100 mutations have been identified caused by gene conversions, large scale gene deletions, and de novo mutations, and novel mutations are continuously being identified. Genotype–phenotype non-concordance is observed in a significant number of patients.

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