



Congenital Adrenal Hyperplasia (Monographs on Endocrinology)

M.I. New, L.S. Levine

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Congenital adrenal hyperplasia (CAH) consists of a group of disorders of adrenal steroidogenesis. Each disorder results from an inherited deficiency of one of the several enzymes necessary for normal steroid synthesis. The different enzyme deficiencies produce characteristic patterns of hormonal abnormalities; the clinical symptoms of the different forms of CAH depend on the particular hormones that are deficient or that are produced in excess. The earliest documented description of CAH was by DeCrecchio in 1865 (DeCrecchio 1865). This Neapolitan anatomist described a cadaver having a penis with first degree hypospadias but no externally palpable gonads. Dis section revealed a vagina, uterus, fallopian tubes, ovaries, and markedly enlarged adrenals. It is interesting that the subject suffered a confusion of sex assignment, being declared a female at birth and a male 4 years later. He conducted himself as a male sexually and socially. Since the original description of this case, investigators have unravelled the pathophysiology of the inborn errors of steroidogenesis. 1 Steroidogenesis and Enzymatic Conversions of Adrenal Steroid Hormones A. Steroidogenesis The adrenal synthesizes three main classes of hormones: mineralocorticoids (17-deoxy pathway), glucocorticoids (17-hydroxy pathway), and sex steroids.

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