



Genetic Steroid Disorders: Chapter 4A. Steroid 5α -Reductase 2 Deficiency

David W. Russell, Jean D. Wilson

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Loss-of-function mutations in the steroid 5 α -reductase 2 gene (SRD5A2) cause a disorder of male sexual differentiation in which the prostate does not form and external genitalia develop along female lines. Failure to synthesize dihydrotestosterone in fetal tissues that give rise to the male urogenital tract underlies the phenotype that characterizes this disorder. Studies of the SRD5A2 gene and its encoded enzyme at the molecular, biochemical, and endocrinological levels established the crucial role of dihydrotestosterone in formation of the male phenotype and in many other androgen actions and led to the development of drugs for the treatment of prostatic disease.

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