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3-Beta-Hydroxysteroid Dehydrogenase Deficiency

National Cancer Institute

Source

National Cancer Institute. *3-Beta-Hydroxysteroid Dehydrogenase Deficiency*. NCI Thesaurus. Code C131088.

Congenital adrenal hyperplasia due to presumed mutation(s) in the HSD3B2 gene, which results in decreased activity of the enzyme 3-beta-hydroxysteroid dehydrogenase. The clinical manifestations of the deficiency are dependent on the degree of reduction in enzymatic activity: 46,XY infants may have incomplete development of the genitalia, while 46,XX infants may have virilization.