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11-Beta-Hydroxysteroid Dehydrogenase Type 1 Deficiency

National Cancer Institute

Source

National Cancer Institute. *11-Beta-Hydroxysteroid Dehydrogenase Type 1 Deficiency*.
NCI Thesaurus. Code C131084.

Decreased activity of the enzyme 11-beta-hydroxysteroid dehydrogenase type 1 due to inactivating mutation(s) in the HSD11B1 gene. The condition is characterized by hyperandrogenism as a result of increased adrenocorticotrophic hormone stimulation of the adrenal gland due to failure of cortisol-mediated down-regulation, and is clinically indistinguishable from H6PD deficiency.