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Hexose-6-phosphate Dehydrogenase Deficiency

National Cancer Institute

Source

National Cancer Institute. *Hexose-6-phosphate Dehydrogenase Deficiency*. NCI Thesaurus. Code C131849.

Decreased activity of hexose-6-phosphatase due to autosomal recessive mutation(s) in the H6PD gene. This enzyme is necessary to generate NADPH, a cofactor in the 11-beta-hydroxysteroid dehydrogenase pathway required for conversion of cortisone to cortisol. 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 11-beta HSD type 1 deficiency.