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# 11-Beta-Hydroxylase Deficiency

National Cancer Institute

## Source

National Cancer Institute. *11-Beta-Hydroxylase Deficiency*. NCI Thesaurus. Code C131085.

Decreased or absent activity of the enzyme 11-beta-hydroxylase caused by loss-of-function mutations in the CYP11B1 gene, resulting in congenital adrenal hyperplasia. Clinical manifestations of this condition include virilization in 46XX infants and hypertension.