# 17-Alpha-Hydroxylase/17,20 Lyase Deficiency

National Cancer Institute

## Source

*National Cancer Institute. 17-Alpha-Hydroxylase/17,20 Lyase Deficiency. NCI Thesaurus. Code C131086.*

Decreased or absent activity of the enzyme 17-alpha-hydroxylase/17,20 lyase due to loss-of-function mutation(s) in the CYP17A1 gene. The clinical manifestations of the deficiency are dependent on whether one or both activities of the enzyme are affected, and may include hypertension due to reduced 17-hydroxylase activity and incomplete genital masculinization in 46,XY infants due to reduced 17,20 lyase activity.