CYP11B1 wt Allele

National Cancer Institute

Source

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Human CYP11B1 wild-type allele is located within 8q21 and is approximately 7 kb in length. This allele, which encodes cytochrome P450 11B1, mitochondrial protein, plays a role in the conversion of progesterone to cortisol in the adrenal cortex. Functional mutations in the CYP11B1 gene can result in 11-beta-hydroxylase deficiency which, in turn, causes congenital adrenal hyperplasia. CYP11B1/CYP11B2 gene fusion causes glucocorticoid-remediable aldosteronism.