11 Beta-Hydroxylase Deficiency

11 beta-hydroxylase deficiency is the second most common variant of congenital adrenal hyperplasia characterized by a defect in the enzyme 11 beta-hydroxylase in the steroid biosynthesis pathway. This enzyme is necessary for the conversion of 11 deoxycorticosterone (11 DOC) to corticosterone in the mineralocorticoid synthesis pathway and 11 deoxycortisol to cortisol in the glucocorticoid synthesis pathway. Therefore, deficiency of this enzyme leads to decreased cortisol and aldosterone synthesis. Low levels of cortisol causes increased ACTH stimulation of the steroid biosynthesis pathway prior to 11 beta hydroxylase and leads to increase androgen synthesis as accumulated cortisol precursors are shunted into the androgen synthesis pathway. This also results in the buildup of 11 deoxycorticosterone. These individuals have clinical features of androgen excess including masculization and premature sexual maturation in boys and virilization in females. Although there is loss of aldosterone, 11 deoxyocorticosterone has mineralocorticoid activity and excess accumulation can lead to hypertension.



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Pathophysiology

Decreased Cortisol
Down-arrow Court-of-Sol
11 beta-hydroxylase is necessary for the conversion of 11 deoxycorticosterone (11 DOC) to corticosterone in the mineralocorticoid synthesis pathway and 11 deoxycortisol to cortisol in the glucocorticoid synthesis pathway. Therefore, deficiency of this enzyme leads to decreased cortisol and aldosterone synthesis.

Decreased Aldosterone
Down-arrow on Aldo-stereo
11 beta-hydroxylase is necessary for the conversion of 11 deoxycorticosterone (11 DOC) to corticosterone in the mineralocorticoid synthesis pathway and 11 deoxycortisol to cortisol in the glucocorticoid synthesis pathway. Therefore, deficiency of this enzyme leads to decreased cortisol and aldosterone synthesis.

Increased Sex Hormones
Up-arrow Harmonica with Sex-symbols
Low levels of cortisol cause increased ACTH stimulation of the steroid biosynthesis pathway prior to 11 beta-hydroxylase and lead to increased androgen synthesis as accumulated cortisol precursors are shunted into the androgen synthesis pathway.

Increased 11 Deoxycorticosterone (11 DOC)
Up-arrow Doctor with (11) Label
Although there is a loss of aldosterone, 11 deoxycorticosterone has mineralocorticoid activity, and excess accumulation can lead to hypertension.

Signs and Symptoms

Hypertension
Hiker-BP
Although there is a loss of aldosterone, 11 deoxycorticosterone has mineralocorticoid activity, and excess accumulation can lead to hypertension.

Masculinization
Mask-with-large-beard
These individuals have clinical features of androgen excess, including masculinization and premature sexual maturation in boys and virilization in females.

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