Genetic Steroid Disorders: Chapter 6A. Apparent Mineralocorticoid Excess

Mabel Yau, Saroj Nimkarn

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Genetic Steroid Disorders: Chapter 6A. Apparent Mineralocorticoid Excess Mabel Yau, Saroj Nimkarn Apparent mineralocorticoid excess (AME) is a rare inherited form of hypertension caused by 11β-hydroxysteroid dehydrogenase type 2 (11β-HSD) deficiency. The disorder was first described biochemically and hormonally in 1977 by New et al. in a Native American girl with severe hypertension. AME defined an important “pre-receptor” pathway in steroid hormone action and their specificities to the receptor. The exploration of the pathogenesis of AME opened a new area in receptor biology as a result of the demonstration that the specificity of the mineralocorticoid receptor function depends on a metabolic enzyme (11β-HSD2) rather than the receptor itself. The clinical manifestations of AME mimic those of excessive mineralocorticoid activity, but plasma levels of aldosterone and other known mineralocorticoids are not elevated. Affected patients may present with low birthweight, failure to thrive, severe hypertension, hypercalciuria and renal failure. The hypertension is severe, with onset in early childhood.

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