



Genetic Steroid Disorders: Chapter 6C. Glucocorticoid-Remediable Aldosteronism

Florencia Halperin, Robert G. Dluhy

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Glucocorticoid-remediable aldosteronism (GRA) is a heritable form of primary hyperaldosteronism. As a result of a chimeric gene duplication, aldosterone synthase is expressed in the cortisol-producing zona fasciculata of the adrenal cortex and is regulated by adrenocorticotropin (ACTH). Clinically, GRA is characterized by early onset of hypertension, which may be severe and refractory to standard therapies. In the absence of treatment with diuretics, hypokalemia is uncommon. GRA is associated with a high prevalence of intracranial aneurysms and hemorrhagic stroke. Diagnostic testing for the presence of the chimeric gene is available. The mainstay of treatment is glucocorticoid suppression of ACTH, and alternatives include mineralocorticoid receptor antagonism.

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