



Genetic Steroid Disorders: Chapter 13F. Genetic Factors in Cushing Disease Pathogenesis

Alan A. Parsa, Shlomo Melmed

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Cushing disease (CD), the clinical manifestation of excess endogenous glucocorticoids derived from the adrenal glands, is caused by increased ACTH secretion from pituitary corticotroph adenomas. Cell signaling, tumor suppressor genes, and cell cycle regulators are implicated in the pathogenesis of ACTH-secreting tumors, as well as some rare familial syndromes. Tumors typically present clinically as microadenomas caused by effects of excess glucocorticoids and rarely as macroadenomas owing to pituitary compressive symptoms. Discovery of novel treatment options is challenging, especially since patient numbers are limited. Accordingly, animal models recapitulating human CD are ideal to study pituitary corticotroph tumor pathogenesis.

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