Genetic Steroid Disorders: Chapter 13A. Animal Models of Adrenal Genetic Disorders

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Adrenal disorders that are caused by specific genetic alterations comprise a heterogeneous group of diseases with mostly low incidence that can affect patients from birth to adulthood. These conditions include failure of proper adrenal development resulting in adrenal agenesis or, conversely, adrenal tumorigenesis. Furthermore, deficiencies of adrenal steroidogenesis result in a lack or a shift of adrenal steroid production that can cause a specific clinical phenotype. For functional studies of gene products, mouse models remain to be intensively utilized as an experimental system owing to the similarity to humans with respect to genome organization, development, and physiology. For the majority of adrenal genetic disorders, mouse models exist that in most instances resemble the clinical phenotype observed in affected patients. Here we provide an overview of these models that allows for both mechanistic and therapeutically relevant investigations that will eventually translate into improved patient care.

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