Congenital adrenal hyperplasia (adrenogenital synd

Congenital adrenal hyperplasia (adrenogenital syndrome)

Virilisation and adrenal insu ffi ciency in children are pathog - nomonic of congenital adrenal hyperplasia (CAH). This is an autosomal recessive disorder caused by a variety of enzy - matic defects in the synthetic pathway of cortisol and other steroids from cholesterol. The most frequent defect (95%) is the 21-h ydroxylase deficiency , which has an incidence of 1 in 5000 live births. Excessiv e ACTH secretion secondary to the loss of cortisol leads to an increase in androgenic cortisol precursors and to CAH. CAH may present in girls at birth with - ambiguous genitalia or as late-onset disease at puberty . Hyper - tension and short stature, caused by the premature epiphyseal plate closure, are common signs. A ff ected patients are treated by replacement of hydr ocortisone and fludrocortisone. Large hyperplastic adrenals may need to be removed if symptomatic.

Revision #1
Created 2025-12-31 15:21:15 UTC by Omar Ayman
Updated 2025-12-31 15:21:15 UTC by Omar Ayman