

# Congenital adrenal hyperplasia (adrenogenital syndrome)

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Virilisation and adrenal insufficiency in children are pathognomonic of congenital adrenal hyperplasia (CAH). This is an autosomal recessive disorder caused by a variety of enzymatic defects in the synthetic pathway of cortisol and other steroids from cholesterol. The most frequent defect (95%) is the 21-hydroxylase deficiency, which has an incidence of 1 in 5000 live births. Excessive 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. Hypertension and short stature, caused by the premature epiphyseal plate closure, are common signs. Affected patients are treated by replacement of hydrocortisone and fludrocortisone. Large hyperplastic adrenals may need to be removed if symptomatic.

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