Diseases affecting the endocrine system can result in increased mortality and morbidity

**RARE DISEASES: PART 5 OF 6**

**Treating congenital adrenal hyperplasia**

In this article...

- The aetiology of congenital adrenal hyperplasia
- Types of CAH and their effects on patients
- Screening and treatment strategies

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Genetic alterations associated with rare endocrine diseases disrupt the body’s normal chemical communication system. Faulty genes can affect any part of the hormone pathway by altering the way the body recognises a hormone, or how a hormone acts on a target organ. One example is congenital adrenal hyperplasia.

CAH results from an inherited alteration in a gene that blocks an essential enzyme (usually 21-hydroxylase) in the adrenal hormone pathway. Synthesis of cortisol (the “stress” hormone) and aldosterone, which regulates the blood pressure through sodium, potassium and fluid balance, is impaired or absent. The adrenal glands enlarge as they work harder to correct the imbalance, resulting in the overproduction of androgens (male hormones); the more severe the enzyme block, the greater the hormone production. In 95% of people with CAH, it is due to an alteration in the 21-hydroxylase gene, which is recessively inherited (Perrin et al, 2000).

One in 55 people is a carrier for CAH (Baumgartner-Parzer et al, 2005).

**Classical CAH**

Classical CAH can be divided into two subtypes and affects both sexes:

- **Salt-wasting CAH** accounts for 75% of those affected and involves a severe enzyme block, with absence of cortisol and aldosterone. Neonates may develop poor feeding and vomiting leading to life-threatening dehydration, hypotension and collapse. Urgent treatment with hydrocortisone and fludrocortisone is required and must be continued for life. Females may be born with ambiguous external genitalia having been exposed to high male hormones levels in utero. In extreme cases, girls can be mistaken for boys.

- **Simple virilising/non-salt-wasting CAH** affects the remaining 25% of people. This is a less severe enzyme block, causing absence of cortisol. Due to an excess of male hormones, children with CAH experience early puberty and accelerated early growth, but often end up shorter than their peers. Cortisol replacement in childhood aims to balance normal growth and pubertal development.

Treatment of classical CAH involves lifelong hydrocortisone replacement with or without fludrocortisone, and management of adrenal crisis. Increased oral steroid dosage, plus emergency intravenous or intramuscular hydrocortisone and treatment of hypoglycaemia are essential during illness, surgery and acute stress.

Patients should carry a steroid card or medic-alert bracelet and emergency hydrocortisone kit. Girls may need corrective genital surgery and psychosexual counselling.

**Non-classical CAH**

This involves a milder enzyme block with only a slight hormone imbalance. Teenage girls may present with delayed or irregular periods, facial hair and greasy skin. Both sexes may experience fertility problems in adulthood, shorter final height, obesity and hypertension. Treatment aims to maintain optimum health and fertility. Patients may also need hydrocortisone replacement for potentially stressful situations.

**Screening and treatment**

Where there is a risk that a foetus could inherit a faulty copy of the gene responsible for CAH from each parent, the following treatment is offered:

- **Dexamethasone** (a steroid that crosses the placenta) can be prescribed at 5–6 weeks’ gestation, protecting a female foetus from exposure to excess male hormones in utero;
- A foetal DNA test to determine the baby’s sex can be performed from seven weeks’ gestation – dexamethasone is stopped if the foetus is male;
- Chorionic villus sampling for genetic testing can be done from 11 weeks’ gestation – dexamethasone is stopped if the foetus is an unaffected female.

At-risk babies are checked by a paediatrician at birth, a cord blood sample is taken for genetic testing (if not already checked) and a short synacthen test is recommended to confirm adequate adrenal hormone production.

All affected children and adults require lifelong steroid treatment and endocrine specialist follow-up, but treated patients should lead normal healthy lives. NT

**References**


**NURSING PRACTICE REVIEW**

**Rare diseases**

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