

The initial management and stabilisation of the newborn infant with a positive screen for congenital adrenal hyperplasia (CAH): An ANZSPED guideline.

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Key points

THIS IS A GUIDE FOR BABIES RECALLED FROM THE COMMUNITY

Newborn screening for CAH is aimed at detecting classical or severe CAH due to 21-hydroxylase deficiency; mild cases may not be detected.

The screening test, elevated 17 hydroxyprogesterone (170HP) level by immunoassay supported by an abnormal steroid profile on liquid chromatography-tandem mass spectrometry (LC-MS/MS) on the same sample, is highly specific for CAH.

Screen positive babies are at high risk of deterioration if treatment is delayed while further confirmatory biochemical or genetic tests are awaited.

Urgent consultation with a paediatric endocrinologist is recommended.

It is prudent to monitor glucose and electrolytes and start treatment as soon as possible.

Unwell babies should receive a stress dose of hydrocortisone (25 mg IV/IM) and resuscitation with 0.9% saline. If BG <3 mmol/l give 10% dextrose bolus (2 mls/kg). Hydrocortisone is continued as 100 mg/m²/d or 1 mg/kg IV/IM 4 hourly. Additional fludrocortisone is not needed alongside stress glucocorticoids as large doses have mineralocorticoid effect.

Well babies are treated with oral hydrocortisone and fludrocortisone, ie hydrocortisone 12-15 mg/m²/d (i.e. 1-1.5 mg po tds) and fludrocortisone 100-200 mcg po once daily. Salt replacement is started once feeding is established and prior to discharge (typically 4 mmol/kg/d in 4 divided doses).

Previously well babies on hydrocortisone who deteriorate should receive stress steroid dosing.

All CAH screen positive newborns should be admitted to hospital.

Actions Screening lab informs paediatric team about positive CAH screen Paediatric team contact parents to arrange for urgent review in ED (same day) Urgent blood tests; gas, electrolytes, BGL, 170HP + repeat bloodspot screening card Stress hydrocortisone 25 Admit and commence mg IV/IM and resuscitate oral hydrocortisone and with 0.9% saline (+/-Unwell? fludrocortisone (before glucose bolus depending definitive diagnosis) Yes No on BGL)

^{*}unwell eg hypotension, hypoglycaemia, hyponatraemia, hyperkalaemia