Pediatric Endocrine Society Kenya

Guideline for Management of
CONGENITAL ADRENAL HYPERPLASIA

Clinical Features

Salt-Wasting CAH

Symptoms
Vomiting
Poor feeding
Excessive thirst
Poor weight gain
Lethargy
Irritability

Signs
Dehydration
Hypotension
Ambiguous genitalia
Enlarged phallus
Hyperpigmentation of genitalia and nipples

Simple Virilizing CAH

Virilization
Precocious puberty
Early development of adult body odour
Increased growth velocity

Non-Classic CAH

Oligomenorrhea
Hirsutism
Subfertility
Delayed puberty in girl
Hypertension

Biochemical Features

Hyponatremia
Hyperkalemia
Hypoglycemia
Hypocortisolism
Metabolic acidosis

Patients with 11β-hydroxylase or 17α-hydroxylase deficiencies may have hypernatremia, hypokalemia and metabolic alkalosis.

Neonatal Screening

Elevated 17 hydroxyprogesterone (17-OHP)
- After 72 hours of life
- 8.00 am – 9.00 am

SUSPECTED CONGENITAL ADRENAL HYPERPLASIA

Manage Adrenal Crisis if present (see below)

Refer to Paediatric Endocrinologist

Other investigations

Pelvic U/S to assess for presence of müllerian structures
Karyotype to determine chromosomal sex
Testosterone, androstenedione, plasma renin activity, aldosterone
Genotyping

Basal 17-hydroxyprogesterone (17-OHP) 8.00 am
Dehydroepiandrosterone Sulfate (DHEAS)
Bone Age (BA) X-ray of non-dominant hand for children over 2 years of age

Basal 17-OHP >200 ng/dL (6 nmol/L)
BA ≥2SD for age
DHEAS normal or elevated

ACTH Stimulation Test (250 mcg/m2)
Obtain 17-OHP and 11-deoxycortisol

Elevated 11-deoxycortisol
Diagnosis: 11β-hydroxylase deficiency
Treat as CAH but without mineralocorticoid or salt replacement
Manage hypertension if present

Stimulated 17-OHP <1000 ng/dL (31 nmol/L)
Diagnosis: Premature Adrenarche

Stimulated 17-OHP 1000-10000 ng/dL (31-300 nmol/L)
Diagnosis: Non-Classic CAH 21α-hydroxylase deficiency

Stimulated 17-OHP >10000 ng/dL (300 nmol/L)
Diagnosis: Classic CAH 21α-hydroxylase deficiency

Basal 17-OHP <200 ng/dL (6 nmol/L)
BA ≥2SD for age
DHEAS elevated

ACTH Stimulation Test (250 mcg/m2)
Obtain 17-hydroxypregnenolone (Δ5-17P) and Cortisol

17-hydroxypregnenolone (Δ5-17P) >6678ng/dL (201nmol/L) and Δ5-17P:Cortisol >363
Diagnosis: Non-Classic CAH 3β-hydroxysteroid dehydrogenase deficiency

Basal 17-OHP <200 ng/dL (6 nmol/L)
BA ≤2SD for age
DHEAS low

Obtain 11-deoxycorticosterone and corticosterone levels.
If elevated:
Diagnosis: 17α-hydroxylase deficiency
Treat as CAH but without mineralocoriticoid or salt replacement
Manage hypertension if present

TREAT AS CONGENITAL ADRENAL HYPERPLASIA

MAINTENANCE TREATMENT

Treatment	Dose	Dose Splitting
Glucocorticoid (Hydrocortisone tablets) Hydrocortisone is administered in tablet form, to be crushed for young children and mixed with milk or liquid as a fresh preparation, and not as suspension, since the distribution of the drug in liquid media is uneven and unstable. Prednisolone and Dexamethasone should not be used in children due to adverse effect on growth.	Neonates and Adolescents: 10-20 mg/m^2/day Children: 10-15 mg/m^2/day	3 times a day (half of the total daily dose at 6am, quarter at 2pm and quarter at 6pm)
Mineralocorticoid (Fludrocortisone tablets)	0.05-0.2 mg/day	Once a day
Sodium Chloride (table salt) during exclusive breastfeeding period	2-4 mEq/kg/day (1-3 g/day)	Several times a day

Stress Treatment

Patients with severe CAH do not produce adequate cortisol in stressful situations such as illness and surgery and are at increased risk of adrenal crisis.

The hydrocortisone dose should be doubled or tripled in these situations for 3-4 days or until the clinical situation allows reduction to the usual dose.

All children should be given a disease identification card , which mentions their condition. The card should always accompany the child at school, home, picnic, hospital etc.

Adrenal Crisis

This is a medical emergency!
Clinical features: Lethargy, poor feeding, nausea, vomiting, abdominal pain, shock
Biochemical features: Hyponatremia, hyperkalemia, metabolic acidosis, hypoglycemia
Management:

1. Life Support: Airway (patency, position), Breathing (oxygen if in shock), Circulation (insert a large bore cannula).
2. Take blood for full blood count, renal chemistry, blood glucose, and blood gas analysis.
3. Determine weight and height.
4. If signs of shock are present, restore intravenous hydration with normal saline at 20 ml/kg over 15 minutes. Maximum is 60 ml/kg (3 boluses). Further fluid replacement to be guided by the hydration status.
5. If hypoglycemia is present, administer 5 ml/kg of 10% dextrose (2ml/kg for neonates).
6. Administer intravenous hydrocortisone at 50-100 mg/m^2 bolus followed by 50-100 mg/m^2/d in four divided doses (6 hourly). Usual dose in newborn babies is 25 mg bolus followed by 5-6 mg every 6 hours.
7. Continue intravenous route till patient is able to take orally.
8. Correct any electrolyte imbalances (hyponatremia, hyperkalemia, metabolic acidosis).
9. For patients with impaired sensorium, insert urethral catheter and nasogastric tubes. Monitor vital signs, intake, output and sensorium.
10. Mineralocorticoid replacement may be resumed when patient is stable and shifted to oral hydrocortisone maintenance doses.

MONITORING OF CHILDREN WITH CONGENITAL ADRENAL HYPERPLASIA

Age	Frequency	Investigations
First 3 months	Monthly	Serum electrolytes
3-12 months	3 monthly	Serum electrolytes Serum 17-OHP If feasible: total testosterone, androstenedione and ACTH Optional: plasma renin acitvity and aldosterone:renin ratio
12-30 months	4 monthly	Serum electrolytes Serum 17-OHP If feasible: total testosterone, androstenedione and ACTH Optional: plasma renin acitvity and aldosterone:renin ratio
	Annually	Vitamin D, Calcium, Bone densitometry
After 24 months	Annually	Bone age assessment
After 5 years	Annually	Ultrasound to screen for testicular and ovarian adrenal rest tumors
All visits	All visits	Assessment of weight, length, blood pressure, signs of virilization, skin pigmentation, Cushingoid features Serum electrolytes for children on fludrocortisone

References:
1. Rodríguez A, Ezquieta B, Labarta JI, Clemente M, Espino R, Rodriguez A, Escribano A. Recommendations for the diagnosis and treatment of classic forms of 21-hydroxylase-deficient congenital adrenal hyperplasia. An Pediatr (Barc). 2017 Aug;87(2):116.e1-116.e10.
2.Dabas A, Vats P, Sharma R, Singh P, Seth A, Jain V, Batra P, Gupta N, Kumar R, Kabra M, Kapoor S, Yadav S. Management of Infants with Congenital Adrenal Hyperplasia. Indian Pediatr. 2020 Feb 15;57(2):159-164.

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