



Newborn Screening ACT Sheet [Elevated 17-hydroxyprogesterone (17-OHP)] Congenital Adrenal Hyperplasia (CAH)

Differential Diagnosis:

- Congenital Adrenal Hyperplasia (CAH),
- 21-OH deficiency; stress or
- prematurity are possible secondary causes of increased 17-OHP.

Condition Description:

- Lack of adequate adrenal cortisol and aldosterone, and increased androgen production.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
- Consult with pediatric endocrinologist, having the following information available (sex, age at NBS sampling, birth weight) and refer, if needed.
- Examine the newborn (ambiguous genitalia or non palpable testes, lethargy, vomiting, poor feeding).
- Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- Emergency treatment as indicated (e.g. IV fluids, IM/IV hydrocortisone).
- Educate family about signs, symptoms and need for urgent treatment of adrenal crisis.
- Report findings to newborn screening program.

(See flowchart for information concerning the specific actions)

Diagnostic Evaluation:

Diagnostic tests include serum 17-OHP (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced). Additional tests may be recommended by the specialist.

Clinical Expectations:

Ambiguous genitalia in females who may appear to be male with nonpalpable testes. At risk for life threatening adrenal crises, shock, and death in males and females. Finding could also be a false positive associated with stress or prematurity.

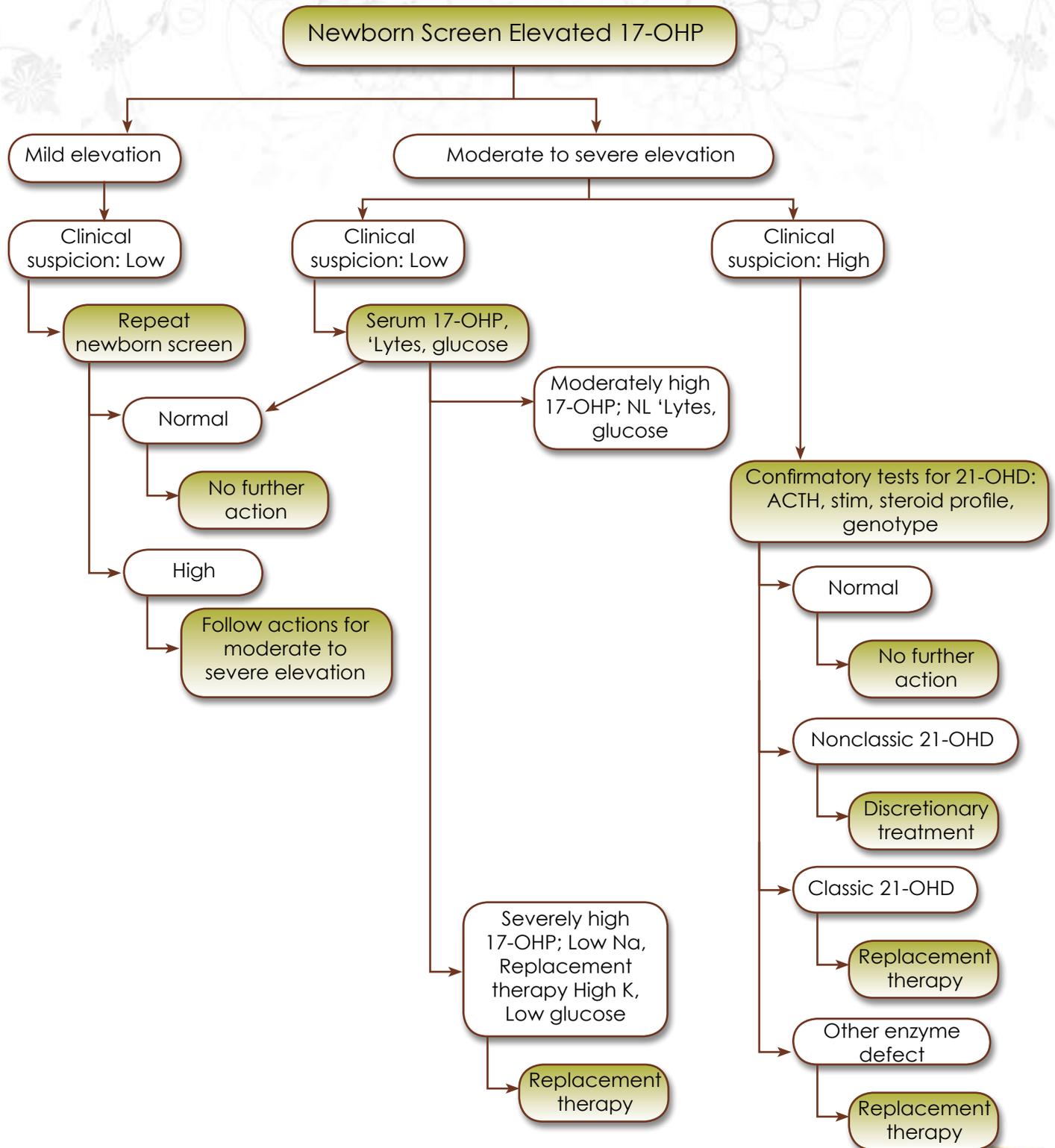
Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen





Screening Today for a Safer Tomorrow

Congenital Adrenal Hyperplasia (Elevated 17-OHP)



NEWBORN



SCREENING

Screening Today for a Safer Tomorrow

Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key

17-OHP = 17-hydroxyprogesterone

'Lytes = Serum electrolytes

ACTH stim = Adrenocorticotrophic hormone stimulation test

21-OHD = 21-hydroxylase deficiency

Steroid profile = Complete adrenal cortical hormone profile, e.g., by MS/MS

Discretionary treatment = Consult pediatric endocrinology to determine if hydrocortisone therapy is necessary

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