**Figure 2. Testing Options for Follow-up of a Positive Newborn Screen for Congenital Adrenal Hyperplasia (CAH)**

**Elevated 17-hydroxyprogesterone (17-OHP) newborn screen**

**Option A**
- Diagnose 21-hydroxylase deficiency: 17-Hydroxyprogesterone Response to ACTH [17682(X)]
  - Normal response
  - 21-OHD unlikely; consider alternative diagnosis

**Option B**
- Differential diagnosis of CAH: Steroid Panel, Congenital Adrenal Hyperplasia (CAH) [90398]
  - No abnormality identified
  - CAH unlikely; consider alternative diagnosis
- CAH Panel 11, Neonatal Random Urine [10046(X)]
  - Abnormality identified
  - CAH likely; confirm with ACTH stimulation test

**Option C**
- Rule out stress as cause of increased 17-OHP: Steroid Panel, 21-Hydroxylase Deficiency vs Stress [90397]
  - ↑ 17-OHP
  - ↓ cortisol
  - ↑ androstenedione
  - N androstenedione

17-OHP post-ACTH response equivocal
- 17-OHP post-ACTH: 1000 to 10,000 ng/dL
  - Nonclassic 21-hydroxylase deficiency diagnosed
- 17-OHP post-ACTH >10,000 ng/dL
  - Classic 21-hydroxylase deficiency diagnosed

ACTH stimulation testing referred to in option B can be performed with test code 90398 or with individual test codes (see Table 2); refer to Table 4 for aid in interpreting results. “Stress” referred to in option C may be caused by prolonged birth, infection, or blood collection. Test codes are in brackets. ACTH indicates adrenocorticotropic hormone; 21-OHD, 21-hydroxylase deficiency; and N, normal.

This figure was developed by Quest Diagnostics based on references 1 and 2. It is provided for informational purposes only and is not intended as medical advice. A physician’s test selection and interpretation, diagnosis, and patient management decisions should be based on his/her education, clinical expertise, and assessment of the patient.

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