Congenital Adrenal Hyperplasia (CAH) Fact Sheet for Health Care Providers
Interpreting an Abnormal CAH Newborn Screening Result – November 2019

What is the screening methodology utilized by the Public Health Laboratory, Oklahoma State Department of Health (OSDH)?
A two-tier screen is utilized:

1. 17-hydroxyprogesterone (17-OHP) level is tested on each filter paper (FP).
2. If the 17-OHP level is elevated, a steroid profile by Tandem Mass Spectrometry (MS/MS) is performed. This test is performed on the same FP sample and will be done by Mayo Laboratories.

Note: MS/MS steroid profile includes testing for 17-OHP, androstenedione, and cortisol with determination of the following ratio: 17-OHP + androstenedione ÷ cortisol. Not all specimens receive MS/MS steroid profile.

What is the NORMAL range for CAH screening and how will it be reported?

Weight in Grams	17-OHP ng/ml	Steroid Profile	Report
≥ 2500	< 28	not performed	Not consistent with CAH
≥ 2500	< 55	normal	Not consistent with CAH
< 2500	< 75	not performed	Not consistent with CAH
< 2500	≥ 75	normal	Not consistent with CAH

How are ABNORMAL screen results reported?

Weight in Grams	17-OHP ng/ml	Steroid Profile	Report
≥ 2500	≥ 55	normal	Inconclusive
≥ 2500	≥ 28-54.9	abnormal	Inconclusive
≥ 2500	≥ 55	abnormal	Consistent with CAH
< 2500	≥ 75	abnormal	Inconclusive

Pre-release (results reported prior to completion of second-tier):

Weight in Grams	17-OHP ng/ml	Steroid Profile
≥ 2500	≥ 55	Pending
<2500	≥ 200	Pending

What are the diagnostic workup needs?
The follow-up program will provide detailed guidance on needed actions.

What is CAH?
CAH is a group of autosomal recessive genetic conditions. The OSDH program screens for the most common form of CAH which is due to 21-hydroxylase deficiency. This disorder is characterized by a deficiency in the hormones cortisol and aldosterone and an overproduction of androgen. Serious loss of body salt and water can result in death. In girls, the genitalia may appear like that of a male, and can result in incorrect sex assignment. Symptoms of adrenal insufficiency include: emesis, excessive weight loss relative to birth weight, diaphoresis, hyperventilation, pallor, dry mucosa, and lethargy.

What is my role in screening?
You have been listed as the infant's planned Health Care Provider on the filter paper requisition and are required by the Newborn Screening Program (NSP) *Regulations* to coordinate follow-up activities.

For more information or assistance, call (405) 271-6617 or 1-800-766-2223.