

# Positive Result:

## Blood Spot Screen Result Notification



## Elevated 17-hydroxyprogesterone (17-OHP)

### Next Steps

Today, you should take the following recommended actions:

- **Consult** with a pediatric endocrinologist. Contact information for endocrinologists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Evaluate** infant (ambiguous genitalia or non-palpable testes, lethargy, vomiting, poor feeding); arrange emergency treatment if symptomatic.
- **Arrange** laboratory testing as recommended by the endocrinologist.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

### Review with Family

Discuss this result with the family as MDH has **not** notified them. Share the follow-up plan with them. Educate family about signs, symptoms, and need for urgent treatment to prevent adrenal crisis.

### False Positives

Screening result can be impacted by prematurity, specimen collection before 24 hours, and perinatal stress.

### Differential Diagnosis

An elevated 17-OHP is primarily associated with:

- Congenital adrenal hyperplasia (CAH) — Incidence of 1 in 18,000

### Clinical Summary

CAH is an endocrine disorder involving impaired synthesis of cortisol by the adrenal cortex. There are three types of CAH: non-classical, simple virilizing, and salt-wasting. Non-classical CAH is the least severe and salt-wasting CAH is the most severe.

Infants with non-classical CAH typically experience early puberty. Symptoms are treatable with low-dose glucocorticoids and may not be life-long.

The term “classical” CAH encompasses both the salt-wasting and simple virilizing types. Most cases of classical CAH are the salt-wasting type. Newborns with salt-wasting CAH can experience poor feeding, weight loss, dehydration, and vomiting. Affected newborns can lose large amounts of sodium in their urine, which can be life-threatening in early infancy. In contrast, infants with simple virilizing CAH do not experience salt loss. In both types, female newborns often have ambiguous genitalia while affected males have normal genitalia. Children with classical CAH require life-long glucocorticoid treatment. Ambiguous genitalia in females can be surgically addressed. Fludrocortisone treats sodium loss for children with salt-wasting CAH.