






Congenital adrenal hyperplasia - 17A Hydroxyprogesterone (17 OHP)

	Test Code	B0002
	Test Summary	This biochemical test analyzes the level of 17 alpha hydroxyprogesterone (17 OHP)
	Turn-Around-Time (TAT)*	3 days
	Acceptable Sample Types	Dried Blood Spots
	Acceptable Billing Types	Self (patient) Payment Institutional Billing

Indications for Testing

- Individuals suspected of having congenital adrenal hyperplasia.
- Individuals with a family history of 21-hydroxylase deficiency.

Test Description

Elevated 17 alpha hydroxyprogesterone levels are associated with congenital adrenal hyperplasia.

Condition Description

21-hydroxylase deficiency is a disease that causes excessive androgen production in the adrenal glands. There are three distinct types of 21-hydroxylase deficiency: salt wasting, simple virilizing, and non-classic. The salt-wasting form of the disease is the most severe, with an age of onset typically in the fetal or neonatal periods. It has symptoms of excessive loss of sodium in the urine, poor feeding, weight loss, dehydration, vomiting, ambiguous genitalia in females, small testes in males, an early growth spurt, shorter adult height, decreased fertility, hirsutism in females, irregular menstruation, and male pattern baldness. The simple virilizing form has an age of onset in the fetal or neonatal period, with symptoms of ambiguous genitalia in females, small testes in males, an early growth spurt, shorter adult height, decreased fertility, hirsutism in females, irregular menstruation, and male pattern baldness. The non-classic form has an age of onset in adolescence or adulthood with symptoms of hirsutism in females, male pattern baldness, irregular menstruation, decreased fertility, early beard growth, and small testes in males. The incidence of salt-wasting and simple virilizing forms for 21-hydroxylase deficiency is estimated to be ~ 1 in 15,000. The incidence of the non-classic form of 21-hydroxylase deficiency is estimated to be ~ 1 in 1000.

Test Methods and Limitations

Isoelectric Focusing techniques are utilized to physically separate proteins in a gel matrix. The sample of mixed proteins, including the analytes, will be isolated from other proteins because this electrophoresis method relies on an electric current and solutions of varied pH to drive the proteins through the gel according to their isoelectric points. Once the physical proteins are separated, their presence and location in the gel can be viewed for analysis and interpretation.

Detailed Sample Requirements

Dried Blood Spots