17-HYDROXYPROGESTERONE (17-OHP), NEWBORN SCREEN; CAH SCREEN (Fluoro Immuno Assay)	ng/mL	(<55.00)

Interpretation

RESULT IN ng/mL	REMARKS
<55	Normal
>55	High

Note: 1.Results should be clinically correlated as individual / biological variations can affect the test results

2. Test conducted on heel prick blood

3. An early morning sample is preferred specially to detect Non–classic variants of the disease

4. Genetic counseling available with prior appointment at Department of Genetics, National Reference Lab, New Delhi

Comments

Congenital Adrenal Hyperplasia (CAH) , an autosomal recessive disease, occurs in 2 forms namely Classic form, and Non- classic form. Majority of CAH cases (90%) are due to 21-hydroxylase deficiency leading to elevations of 17-hydroxyprogesterone levels. Less commonly it is due to 11-hydroxylase deficiency.

The ***classic*** variety presents in the newborns or early childhood with adrenal insufficiency and virilization with or without salt wasting.

The ***non-classic form*** presents in late childhood or young adulthood as hirsutism, amenorrhea and infertility in females and precocious puberty in males.

Impression

Advised