



Name	Master VIHAAN	Collected	14/9/2019 3:04:00PM
Lab No.	149532270	Received	14/9/2019 3:22:09PM
A/c Status	P	Gender:	Male
	Ref By :	Report Status	Final

Test Name	Results	Units	Bio. Ref. Interval
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#### NEWBORN SCREENING

17-HYDROXYPROGESTERONE (17-OHP), NEWBORN SCREEN; CAH SCREEN @ (Delfia)	3.55	ng/mL serum	<22.00
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#### Interpretation

17-OHP (ng/mL Serum)	REMARKS
<22	Presumptive Negative
22-66	Equivocal zone
>66	Presumptive Positive

#### Note

1. The concentration of 17 OHP in newborn varies with age, weight, prematurity and twinning
2. Test conducted on heel prick blood
3. Premature, sick or stressed infants have higher 17 OHP values leading to false positive screen results
4. Antenatal corticosteroid treatment may reduce 17 OHP levels resulting in false negative screen
5. Positive screen test should be followed by confirmatory test on LCMS/MS

#### 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. 17-OHP levels are normally high at birth and decrease rapidly during the first few postnatal days. In contrast, 17-OHP levels increase with time in infants affected with CAH

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. These cases may not be detected by newborn screening

#### Impression

#### Advised





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Test Name	Results	Units	Bio. Ref. Interval
BIOTINIDASE, NEWBORN SCREEN @ (DELFIA)	232.30	Units	>58.50

#### Note

1. High albumin levels interfere with the assay and may give false negative results
2. Results should be clinically correlated as individual / biological variations can affect the test results
3. Test conducted on heel prick blood

#### Comments

Biotinidase deficiency is an autosomal recessive disorder caused by mutations in the biotinidase gene (BTD). Patients with profound deficiency usually present around 3 months of age but may present as early as first week of life and as late as 10 years of age. Untreated children show alopecia, periorificial skin rash, irreversible sensorineural deafness, conjunctivitis, development delays and hypotonia. Partial deficiency may be asymptomatic or produce milder symptoms. Certain organic acidurias may present similarly as biotinidase deficiency. Patients are successfully treated with oral doses of biotin, a water soluble form of vitamin B.





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Test Name	Results	Units	Bio. Ref. Interval
CYSTIC FIBROSIS, NEWBORN, SCREEN @ (DELFIA)	42.90	ng/mL	<65.00

#### Note

1. IRT levels decline with age
2. IRT levels above cut-off should be confirmed by second line test (Sweat chloride or mutation analysis)
3. Test conducted on heel prick blood

#### Comments

Cystic fibrosis is one of the most common autosomal recessive diseases due to mutation in Cystic fibrosis transmembrane conductance regulator ( CFTR) gene seen mainly in people of Northern European ancestry. It is a multisystem disorder affecting pulmonary, gastrointestinal and reproductive organs. The phenotypic expression of the disease is heterogenous ranging from meconium ileus to severe respiratory disease in infants. Immunoreactive trypsinogen ( IRT) is used to screen newborns for increased risk of Cystic fibrosis. Screening for Cystic Fibrosis has helped in increasing survival age largely due to organ transplantation, improved nutrition, new drug therapies and shall continue to do so with a potential of successful gene therapy.

#### Increased IRT Levels

Cystic fibrosis - Delta F 508 heterozygotes ( Commonest mutation ), Hypoxic insult to pancreas, Renal insufficiency, Congenital heart disease, Spina bifida, Gastrochisis, Viral infections, Trisomy 13,18 & Galactosemia.

**False Positive results:** Transient neonatal hypertrypsinemia, Low Apgar score, African-American infants, Healthy carriers.

**False negative results:** Meconium ileus (Sweat chloride test is preferred test for screening).





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Test Name	Results	Units	Bio. Ref. Interval
GALACTOSEMIA, NEWBORN SCREEN @ (Delfia)	3.90	mg/dL	<10.00

### Interpretation

#### Note

1. Results should be clinically correlated as individual / biological variations can affect the test results
2. Test conducted on heel prick blood
3. False Negative results can be observed If sample is taken from infant who has not ingested breast milk/within 4 months of blood transfusion/ from an Infant on lactose free formula feed or on IV fluids

#### Comments

Classic galactosemia is an autosomal recessive disorder of galactose metabolism caused by mutations in GALT gene. The complete or near complete deficiency of GALT enzyme can be life threatening. Galactosemia is treated by a galactose free diet, but despite adequate treatment from an early age, children with galactosemia remain at an increased risk for development delays, speech problems and abnormalities of motor function.

TYPE OF DISEASE	DEFICIENCY	INHERITANCE	CLINICAL SYMPTOMS
Classic Galactosemia (GALT deficiency)	Deficiency of Galactose-1-phosphate uridyl transferase	Autosomal recessive	Hyperglycemia, vomiting, diarrhea, irritability, feeding difficulty, failure to thrive
GALK deficiency	Deficiency of Galactokinase	Rare	Cataract, Pseudotumor cerebri
GALE deficiency	Deficiency of Uridine diphosphate galactose-4-epimerase	-	Benign form: Asymptomatic Severe form: Hypotonia, Sensorineural deafness

### Impression

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Test Name	Results	Units	Bio. Ref. Interval
PHENYLALANINE, NEWBORN SCREEN @ (DELFIA)	0.90	mg/dL	<2.10

#### Interpretation

PHENYLALANINE VALUE	COMMENTS
<2.10	Normal
2.10-3.00	Equivocal
>3.00	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. To avoid missing a Phenylketonuria (PKU) case, infants should not be screened until at least 48 hours after birth

#### Comments

Phenylalanine is an essential amino acid which is converted to Tyrosine by Phenylalanine hyroxylase. Deficiency of this enzyme results in Phenylketonuria ( PKU) which is inherited as an autosomal recessive trait with Classic PKU being the commonest. Phenylalanine levels are normal in the cord blood of neonates and only rise after milk feedings have been initiated, hence screens obtained prior to 48 hours may give false negative results. The urine of affected individuals has a peculiar musty odour. Brain myelination is abnormal. PKU should be considered in an infant who loses developmental milestones in the first 6 to 12 months of life. Untreated PKU leads to severe brain damage with intellectual impairment, behavior abnormalities, seizures & spasticity.

#### Impression

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Test Name	Results	Units	Bio. Ref. Interval
TSH, NEWBORN SCREEN @ (Delfia)	2.56	miU/L	

#### Interpretation

#### As per Newborn Screening Guidelines for Congenital Hypothyroidism in India (2018)

TSH IN mIU/L (SERUM UNITS) ON FIRST SCREEN			REMARKS
24 to 48 hours after birth	>48 hours to 2 weeks after birth	>2 weeks after birth	
<34	<20	<10	Normal
34-40	20-40	—	Borderline increase; Recommended for retest at age 2 weeks
>40	>40	>10	Highly suggestive of Congenital Hypothyroidism; Send venous sample for TSH and FT4 after 72 hours of age for confirmation
>80	>80	>80	Send venous sample for TSH and FT4 for confirmation and start treatment immediately

#### Note

1. Results should be clinically correlated as individual / biological variations can affect the test results
2. Test conducted on heel prick blood
3. Ideal sampling time is between postnatal day 3 to day 5 or Cord blood
4. False positive result may be observed if sample is collected within 72 hours of birth
5. Presence of thyroid hormone from the mother in newborn circulation at birth can mask Congenital hypothyroidism
6. Genetic counseling available with prior appointment at Department of Genetics, National Reference Lab, New Delhi

#### Comment

Congenital hypothyroidism (CH) is the commonest cause of preventable mental retardation. The crucial point to remember when screening for CH is the neonatal surge of TSH and T4. The TSH surge starts 30 min after birth (T4 some hours later), is most marked for the next 24 hours, but may persist for 48 to 72 hours. Thus, cord blood is largely spared of the neonatal surge. If the screen sample is taken during the surge, a false





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positive result will follow. High risk neonates such as multiple birth, preterm, low birthweight and sick neonates are at increased risk of inappropriate TSH levels (both false positive and false negative). It is recommended to do second screening test at 2-4 weeks of age for high risk babies.			





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Test Name	Results	Units	Bio. Ref. Interval
G-6-PD, NEWBORN SCREEN @ (Delfia)	7.10	U/g of Hb	>2.20

### Interpretation

#### Note

1. Results should be clinically correlated as individual / biological variations can affect the test results
2. Test conducted on heel prick blood
3. Recommended confirmation of deficient state is by quantitative estimation of G-6-PD in whole blood
4. False negative results may be observed in heterozygous G6PD deficient females

#### Comments

Glucose -6-Phosphate dehydrogenase (G-6-PD) deficiency is the most common enzymopathy affecting 400 million people worldwide. The disease is X-linked and more than 300 different types of G-6-PD variants have been described. Majority of G-6-PD deficient individuals are usually asymptomatic and develop hemolysis only when oxidative stress occurs as with bacterial / viral infections and after ingestion of certain drugs or fava beans.

#### Classification of G-6-PD deficiency

Class I	Severe deficiency associated with Chronic hemolytic anemia
Class II	Severe deficiency (<10% residual activity)usuallywithout Hemolytic anemia
Class III	Moderate to mild deficiency (10-60% residual activity)
Class IV	very mild or no deficiency
Class V	Increased activity

#### BLOOD GROUP, ABO & RH TYPING @ (Erythrocyte Magnetized Technology)

ABO Group A

Rh Factor Positive





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**Note:** 1. Both forward and reverse grouping performed

2. Test conducted on EDTA whole blood



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<b>BILIRUBIN TOTAL, DIRECT AND INDIRECT, SERUM</b> (DPD, Calculated)			
Bilirubin Total	15.61	mg/dL	0.30 - 1.20
Bilirubin Direct	0.73	mg/dL	Not Established
Bilirubin Indirect	14.88	mg/dL	Not Established

#### Comment

Unconjugated hyperbilirubinemia poses a risk for development of Kernicterus, especially in low birth weight infants. Most neonates develop mild unconjugated hyperbilirubinemia between days 2 and 5 after birth. Peak levels are typically 5-10 mg/dL and decline to normal level within 2 weeks. It is recommended to interpret bilirubin levels according to infant's age in hours. Physiological jaundice generally is not harmful but bilirubin concentration above 10 mg/dL coupled with prematurity, low serum albumin, acidosis, and substances that compete with binding site albumin (eg. ceftriaxone, sulfisoxazole, aspirin), may increase the risk for Kernicterus.



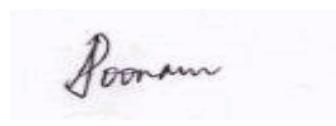
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Dr Poonam Yadav  
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Dr Anil Arora  
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 NRL - Dr Lal PathLabs Ltd



Dr Parul Chopra  
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-----End of report-----





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IMPORTANT INSTRUCTIONS

\*Test results released pertain to the specimen submitted.\*All test results are dependent on the quality of the sample received by the Laboratory.\*Laboratory investigations are only a tool to facilitate in arriving at a diagnosis and should be clinically correlated by the Referring Physician.\*Sample repeats are accepted on request of Referring Physician within 7 days post reporting.\*Report delivery may be delayed due to unforeseen circumstances. Inconvenience is regretted.\*Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit request within 72 hours post reporting.\*Test results may show interlaboratory variations.\*The Courts/Forum at Delhi shall have exclusive jurisdiction in all disputes/claims concerning the test(s) & or results of test(s).\*Test results are not valid for medico legal purposes. \*Contact customer care Tel No. +91-11-39885050 for all queries related to test results.

(#) Sample drawn from outside source.



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