carieM™ METABOLIC TESTING PROGRAM

SUJANAN A LILAC INSIGHTS INITIATIVE Good Birth. Good Life.

To: **Max superspeciality Hospital**
2, Press Enclave Road
Saket New Delhi
Delhi
New Delhi - 110017
Contact: 9818005382
Report Of: B/O SWETA BAIROLIYA
Pt. Contact: 9312274244

Sample ID	2460001354
Patient ID	160243948
Collected on	20/05/2024
Received on	21/05/2024 11:32
Registered on	21/05/2024 12:50
Reported on	23/05/2024 12:16
Referred by	**Dr. PIYUSH CHANDEL**

carIeM™ Newborn Screening - Hepta

Patient Name: B/O SWETA BAIROLIYA Patient DOB: 17/05/2024

Weight: 2.80 Kg Gender: FEMALE City: New Delhi Hospital ID: ML04696820

Sample Type:DBS **Method:**Time-resolved Fluroimmunoassay

- carieM™ is curated to ensure early detection of IEMs (Inborn Errors of Metabolism) so that they can be identified and managed appropriately at an early stage and the adverse outcomes associated with IEMs can be prevented.
- Through carIeM™ Metabolic Testing program we want to ensure that all your metabolic testing requirements from screening to management receive multidisciplinary advice from our expert team of Metabolic Geneticists, Metabolic Dietitian, and Genetic Counsellors collaboratively at one place and aid you in your decision to achieve the best possible outcomes for your loved ones.

Clinical History

- Newborn screening test was offered to screen for Inborn Errors of Metabolism in the baby.

Screening for Common 7 Parameters

Metabolites	Results	Normal Ranges	Observed Levels	Interpretations
Congenital Hypothyroidism (TSH)	1.97 µU/mL	N: 0.1 to 10 µU/mL	Normal level of TSH.	**Screen Negative**
Congenital Adrenal Hyperplasia (17-OHP)	7.39 nmol/L	N: 0.1 to 30 nmol/L	Normal level of 17-OHP.	**Screen Negative**
Galactosemia (Total Galactose)	5.00 mg/dL	N: 0.1 to 15 mg/dL	Normal level of total Galactose.	**Screen Negative**
Cystic Fibrosis (IRT)	13.70 ng/mL	N: 0.1 to 70 ng/mL	Normal level of IRT.	**Screen Negative**
Phenylketonuria (PKU)	1.30 mg/dL	N: 0.1 to 2 mg/dL	Normal level of Phenylalanine.	**Screen Negative**

Enzymes	Results	Normal Ranges	Observed Levels	Interpretations
G6PD Deficiency (G6PD enzyme)	5.65 U/g Hb	N: 2.5 U/g Hb & Above	Normal activity of G6PD enzyme.	**Screen Negative**
Biotinidase Deficiency (Biotinidase enzyme)	158.40 Units	N: 60 Units & Above	Normal activity of Biotinidase enzyme.	**Screen Negative**

TSH: Thyroid-stimulating Hormone; IRT: ImmunoreactiveTrypsinogen; 17- OHP: 17-hydroxyprogesterone; G6PD: Glucose-6-phosphate dehydrogenase

Patient name : B/O SWETA BAIROLIYA **Sample ID** : 2460001354

Impression

The given blood sample shows all the metabolites/enzymes studied to be within normal limits.

Suggestion

Clinical correlation is recommended.

Rakhee P. Vishwakarma
MSc. Mphil., Scientific Officer

Verified by
Mr. Pradip Kadam
In-charge Biochemistry

Dr. Chaitanya Datar
MBBS, MD (Medical Genetics),
Consultant, Clinical & Metabolic Geneticist

***Note:** All the above results would pertain to the level of the metabolites at the time of sample collection. It must be noted that the clinical condition, dietary intake, medical supplementation etc. at the time of sample collection does have an impact on metabolite levels. Therefore consideration of these factors is essential while interpreting these results.

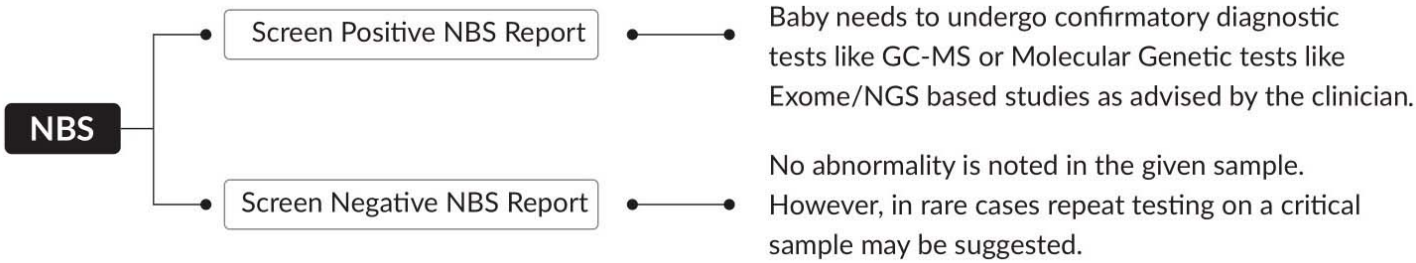
Understanding and Interpreting carIeM™ Hepta Newborn Screening Test Report

Screening for Common 7 Parameters

- A "negative" or "in-range" result means that the baby's blood test did not show any signs of the conditions included on the newborn screening panel.
- A "positive" or "out-of-range" result means that the baby's screening exam did show signs that the baby may be at higher risk of having one or more of the conditions included in the newborn screening panel. This does not mean that the baby definitely has a medical condition. However, follow-up testing must be performed immediately to determine if a condition is actually present after consultation with a qualified medical practitioner.

Positive Newborn screening results give out the abnormal levels of metabolites and enzymes in the body indicative of a particular type of Inborn Error of Metabolism.

The final report interpretation involves correlation between the abnormal metabolites/enzymes and the medical history, family history, clinical presentation, etc.



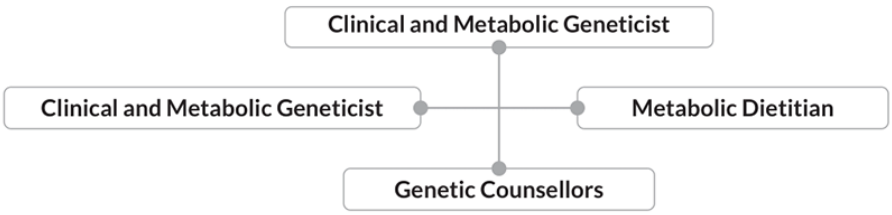
Patient name : B/O SWETA BAIROLIYA **Sample ID** : 2460001354

The carIeM Advantage

carIeM™ is an unique metabolic testing program which offers you the added value of 40+ years of leading metabolic disorder experts of the country.

carIeM™ program is the only metabolic testing program in India to offer extensive care consisting of:

Team at Sujanan



Laboratory:

State-of-the-art diagnostic facility with capability to perform screening to advance metabolic testing such as TMS, GC-MS, Disorder specific advanced metabolic panels to Molecular testing like Exomes or other NGS based testing options, as advised by the clinician.

Clinical & Metabolic Geneticists:

- Metabolic disorders can be complex and it requires understanding of complex biochemical pathways and various diagnostic options available for diagnosis of IEMs.
- Post diagnosis management of IEMs also require expertise to advise guideline based management to the patient.

Metabolic Dietitian:

Many IEMs require strict nutritional intervention. Metabolic dietitian at carIeM™ ensures guidelines and protocol based nutritional guidance for such disorders.

Genetic Counsellors across India:

- Given the inherited nature of most IEMs and the necessary long-term management for these disorders, the genetic counsellor's role in clinical setting is integral in providing ongoing support and education for patients and their families.
- This includes coping with the disease burden, helping patients and families adapt to a condition in the family and ensuring adequate understanding of the genetic risks and the available prenatal diagnostic and reproductive choices.

General Disclaimers

- Genetic/metabolic testing may have technical limitations. These limitations pertaining to different assays have been mentioned in the respective reports.
- It is assumed that the specimen belongs to the person undergoing the test.
- The above results must be interpreted in conjunction with the clinical profile of the patient by the referring Clinician
- Genetic counselling pertaining to the report must be considered. It is the patient/ relative's responsibility to seek further guidance.
- Isolated laboratory investigations may not confirm the diagnosis of a disease. They help in arriving at a diagnosis in conjunction with the clinical presentation and other investigations.
- Some of the special tests may be outsourced to some of our referral laboratories and the reports may be transcribed on our letterhead.
- Partial reproduction of this report is not considered valid.
- This report is not valid for medico-legal purpose.
- Each sample received at Lilac Insights' processing centre is handled with the utmost sensitivity and care. All samples received on Sundays and National holidays are stored as per specific guidelines for the respective specimens and processed on the next day.

Neither the lab nor its employees/representatives are liable / responsible for any loss or damage that may be incurred to any person/s as a result of the incorrect use of the report or inaction thereof.

END OF REPORT