LABORATORY REPORT

Redcliffe labs

Name	CRM NO	Lab ID	Father Name

Package	Gender	DOB/Age	Referred By
TMS 52	Female		
Hospital name	**City**	**Pre Term / Full Term**	**Birth Weight (Kg)**
	NOIDA	Full Term	
Specimen Source	**Specimen Notes**	**Collection Date & Time**	**Received Date & Time**
Heel Prick	Acceptable		
Date of Report & time	**Blood Transfusion**	**Transfusion Date**	**Special Feeds / IVF / TPN/ Supplements**
	No	No	No

INBORN ERRORS OF METABOLISM (IEM) SUMMARY REPORT

Sr. No.	Test Methodology	Result	Test Type
1	TANDEM MASS SPECTROMETRY SCREENING REPORT	NEGATIVE	Screening

DISCLAIMER

All investigations have their limitations which are imposed by the limits of sensitivity and specificity of individual assay procedures as well as the quality of specimen received by the laboratory.

It is presumed that the received specimen belongs to the patient named or identified in the test request form.

Partial reproduction of this report is not permitted.

It is important to monitor baby's health and take a proactive approach should there be any unhealthy condition(s) within or beyond scope of this test.

A negative screening assay result does not rule out the possibility of an underlying metabolic / genetic disease.

The metabolites and health conditions mapping is not mutually unique and there can be other forms in which an abnormality may be apparent. Similarly, the health condition may not exclusively be an outcome of the disorders in consideration

If the newborn is premature, retest is suggested when adjusted gestational age is 40 weeks.

HB-Variant hemoglobins may require further testing by the laboratory to diagnose.

HB-Presence of Hb Bart's with any newborn screen result indicates the presence of alpha thalassemia of unknown severity.

HB-If an increased amount of adult hemoglobin is noted, the infant is presumed to have been transfused (Including Intrauterine transfusion).

HB-Carriers of beta thalassaemia and HbLepore cannot be excluded at this age.

Investigations performed is not intended to replace the medical advice and or treatment obtained from a qualified healthcare practitioner as it doesnot cover all the metabolic disorders

HB (Covered under Hemoglobinopathies package)

Dr. Ankur Jindal (Ph.D)
Consultant Cytogenomics

Name	CRM NO	Lab ID	Father Name

TANDEM MASS SPECTROMETRY SCREENING REPORT

REPORT- SUMMARY

Observations:
All the analytes are within the normal range.

Interpretation:
The tested sample is screened NEGATIVE for the list of disorders in Annexure 1.

Recommendation:
Please correlate the results with other clinical and diagnostic findings.

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Initial Report [y] Duplicate Report [y] Revision Report [ ] Version No [1]

Analyte Name	Analyte Result(µmol/L)	Analyte Reference	MoM*	MoM Value	MoM Reference
			0.01 0.1 1 10 100		
Alanine	280.000	71.00-1361.24		1.14	NMT 5.56
Arginine	13.200	0.78-60.00		5.43	NMT 24.69
Citrulline	13.800	3.86-62.11		1.21	NMT 5.45
Glutamine	936.000	48.24-1309.25		1.83	NMT 2.56
Glycine	358.000	117.35-1270.00		0.93	NMT 3.29
leucine\isoleucine\hydroxyproline	134.000	23.93-383.93		1.06	NMT 3.02
Methionine	14.200	4.63-48.00		0.87	NMT 2.93
Ornithine	90.100	22.75 - 330.00		1.03	NMT 3.77
Phenylalanine	56.300	17.10 - 193.00		1.05	NMT 3.61
Proline	156.000	30.00 - 448.00		1.14	NMT 3.27
Tyrosine	72.700	15.34-455.88		0.97	NMT 6.10
Valine	117.000	31.28 - 450.00		1.03	NMT 3.95
Free Carnitine	25.900	7.00-121.04		1.03	NMT 4.82
Acetylcarnitine	15.600	2.49-65.00		0.95	NMT 3.81
Propionylcarnitine	2.780	0.12-7.02		1.71	NMT 4.31
Butyrylcarnitine	0.230	0.06-1.30		1.05	NMT 6.27
Isovalerylcarnitine	0.090	0.00-0.60		0.75	NMT 5.00
Hexanoylcarnitine	0.020	0.00-0.30		0.5	NMT 7.50
Octanoylcarnitine	0.040	0.00-0.37		0.67	NMT 6.17
Octenoylcarnitine	0.030	0.00-0.32		1	NMT 10.67
Decenoylcarnitine	0.060	0.01-0.24		1.5	NMT 6.00
Decadienoylcarnitine	0.010	0.00-0.07		1	NMT 7.00
Tetradecenoylcarnitine	0.120	0.01-0.51		1.71	NMT 7.30
Hexadecanoylcarnitine	4.090	0.00-9.04		1.14	NMT 2.53
3-Hydroxy-hexadecanoylcarnitine	0.030	0.00-0.09		1.5	NMT 4.50
Octadecanoylcarnitine	0.990	0.00-2.41		1.06	NMT 2.59
Octadecenoylcarnitine	1.710	0.00-3.25		1.37	NMT 2.60
Succinylacetone	0.200	0.00-1.00		0.8	NMT 4.00
Adenosine	0.450	0.00-3.52		0.76	NMT 5.97
2-deoxyadenosine	0.020	0.00-0.10		1	NMT 5.00
C26:0-lysophosphatidylcholine	0.650	0.0 - 1.06		2.17	NMT 3.53
Argininosuccinic acid	0.150	0.00-2.02		0.65	NMT 8.70
Glutamic acid	300.000	47.00-915.00		1.2	NMT 3.65
Propionylcarnitine/Acetylcarnitine	0.180	0.00-0.55		1.64	NMT 5.00
Methylmalonyl	0.170	0.00-1.50		1	NMT 8.82
Glutarylcarnitine	0.050	0.00-0.35		0.71	NMT 5.00
Malonylcarnitine	0.280	0.00-1.00		2.8	NMT 10.00

* This Graph represents the value corresponding to Multiple of Median(MoM). Each box represents the interval between 1%ile and 99%ile. The MoM Shown as dot.

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ANNEXURE - 1 LIST OF DISORDERS

Amino Acid Disorders
Citrullinemia Type I
Citrullinemia Type II
Hyperornithinemia with Gyral Atrophy
5-Oxoprolinuria
Transient Neonatal Tyrosinemia
Argininosuccinic Acidemia
Argininemia
Defects of biopterin cofactor biosynthesis
Disorders of biopterin cofactor regeneration
Benign Hyperphenylalaninemia
Homocystinuria
Hypermethioninemia
Non-ketotic hyperglycinemia
Phenylketonuria
Tyrosinemia type I
Tyrosinemia type II
Tyrosinemia type III
Maple syrup (urine) disease

Fatty Acid Oxidation Disorders
Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency
Very long-chain acyl-CoA dehydrogenase deficiency
Short-Chain Acyl-CoA Dehydrogenase Deficiency
Trifunctional protein deficiency
Glutaric acidemia type II
Medium-Chain Acyl-CoA Dehydrogenase Deficiency
Medium-chain ketoacyl-CoA thiolase deficiency
Carnitine Acyl-Carnitine Translocase deficiency
Carnitine Palmitoyl-Transferase type II deficiency
2,4 Dienoyl-CoA reductase deficiency
Carnitine Palmitoyl Transferese Type I Deficiency
Carnitine Uptake Defect

Organic Acid Disorders
Isobutyryl-CoA-Dehydrogenase Deficiency
Glutaric Acidemia type I
Iso-Valeric Acidemia
Beta-KetoThiolase deficiency
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency

Organic Acid Disorders
Malonic acidemia
Methylmalonic acidemia (Cobalmin Disorders)
Methylmalonic acidemia (Mutase)
3-Methylglutaconic aciduria
2-Methyl-Butyryl-Glycinuria
2-Methyl-3-HydroxyButyric Aciduria
3-Methylcrotonyl-CoA carboxylase deficiency
Multiple Carboxylase Deficiency (also known as Holocarboxylase synthase deficiency)
3-Hydroxy-3-Methyl-Glutaric aciduria
Propionic acidemia

Other Conditions
Adenosine deaminase (ADA)–severe combined
Carbamoyl phosphate synthetase I
Hyperornithinemia-Hyperammonemia-Homocitrullinuria
X -linked adrenoleukodystrophy
Treatment with antibiotics/Pyvalic Acid
Treatment with Benzoate or Valproic Acid
Hyperalimentation (TPN)
Liver Complications
Medium Chain Triglyceride (MCT) Oil Administration
N-acetylglutamate synthase deficiency
Ornithine transcarbamylase deficiency

REFERENCES

1. Ombrone D, Giocaliere E, Forni G, Malvagia S, la Marca G. Expanded newborn screening by mass spectrometry: New tests, future perspectives. Mass Spectrom Rev. 2016 Feb;35(1):71?84.

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

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