RML MEHROTRA PATHOLOGY
Committed To Diagnostic Excellence

Newborn Screening Profile

- Amino Acids and Acylcarnitines in Dried Blood Spots (DBS) (28 Test)
- NEWBORN SCREENING TEST – Comprehensive Panel (07 Test)
- NEWBORN SCREENING TEST – Basic Panel (04 Test)

RML Pathology brings to you world class diagnostics for early detection of disorders and abnormalities in newborn babies

NEWBORN SCREENING PROFILE

Neonatal Screening is an important preventive measure to detect severe irreversible damages caused by congenital metabolism disorders and endocrinopathies at a very early stage. One elementary integral part of the screening programme is the determination of the amino acids and acylcarnitines profile. The defects detected in these profiles are aminoacidopathies, fatty acid oxidation defects, carnitine cycle defects and organoacidemias.

Sample Type: Dried Blood Spots (DBS)

NEWBORN SCREENING TEST – Amino Acids and Acylcarnities from Dried Blood (28 Test)

Of these target diseases those listed in below are screened by **TANDEM MASS SPECTROMETRY**

Amino Acids and Succinylacetone

Alanine	Arginine	Aspartic Acid	Citrulline	Glutamic Acid	Glycine
Leucine	Methionine	Ornithine	Phenylalanine	Proline	Tyrosine
		Valine	Succinylacetone		

Acylcarnities and free Carnitine

Free Carnitine	C2- Carnitine	C3- Carnitine	C4- Carnitine	C5- Carnitine	C5DC- Carnitine
C6- Carnitine	C8- Carnitine	C8- Carnitine	C10- Carnitine	C12- Carnitine	C14- Carnitine
		C16- Carnitine	C18- Carnitine		

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NEWBORN SCREENING TEST – Comprehensive Panel (07 Test)

Of these target diseases those listed in below are screened by **ELISA**

Glucose-6- phosphate dehydrogenase deficiency (G6-PD)	**Congential Andrenal Hyperplasia (CAH)**	**Congenital Hypothyroidism (TSH)**	**Total Galactose (TGAL)**
Phenylketonuria (PKU) Deficiency	**Cystic Fibrosis (IRT Screening Assay)**	**Biotinidase Deficiency (BTD Screening Assay)**	

NEWBORN SCREENING TEST – Basic Panel (04 Test)

Of these target diseases those listed in below are screened by **ELISA**

Glucose-6- phosphate dehydrogenase deficiency (G6-PD)	**Congential Andrenal Hyperplasia (CAH)**	**Congential Andrenal Hypothyrodism (TSH)**	**Total Galactose (TGAL)**
Glucose-6-Phosphate Dehydrogenenase (G6PD)	**17- Hydroxyprogesterone (17-OHP)**	**Thyroid Stimulating Hormone (TSH)**	

Table 1. Metabolites and suspicious metabolic disease

Metabolites	2nd Testing recommended	Suspicious metabolic disease
Phe		PKU, citrin deficiency (citrulline ↑)
[Leu+Ile] & Val	AA (plasma), OA (urine)	MSUD, ketosis
Met	AA (plasma), ammonia (blood)	Homocystinuria, MAT deficiency, citrin deficiency (citrulline (high))
Cit	AA (plasma), ammonia (blood)	Citrullinemia I, argininosuccinic aciduria, citrin deficiency
Cit (low)	ammonia (blood)	OTC deficiency, CPS1 deficiency
Arg	AA (plasma)	Hyperargininemia, citrin deficiency(citrulline (high))
Tyr	OA(urine)	Hypertyrosinemia (if succinylacetone (+ type 1), liver dysfunction
C0 (low)	Removal rate of carnitine	systemic carnitine deficiency, secondary carnitine deficiency
C0	OA (urine)	CPT1 deficiency, tissue degradation (hypoxia)
C2	OA (urine)	Ketosis
C2 (low)		(general fatty acid oxidation problem)
C3[38)]	OA (urine)	Methylmalonic aciduria, propionic aciduria
C4	OA (urine)	IBD deficiency, SCAD deficiency, glutaric aciduria type 2
C4-OH	OA (urine)	SCHAD (HAD) deficiency, ketosis
C5	OA (urine)	Isovaleric aciduria, under administraion of pivaloyl containing antibiotics , 2MBD deficiency
C5-DC	OA (urine)	Glutaric aciduria type 1, glutaric aciduria type 2
C5-OH	OA (urine)	Complex carboxylase deficiency, methylcrotonylglycineuria, HMG uria, beta-ketothiolase deficiency, MCG-CoA hydrolase enzyme deficiency, biotin deficiency
C8	OA (urine)	MCAD deficiency (C8>C10), glutaric aciduria type 2 (C8<C10)
C10–C12	OA (urine)	Glutaric aciduria type 2
C14:1	OA (urine)	VLCAD deficiency, ketosis
C16	OA (urine)	CPT2 deficiency, CACT deficiency, VLCAD deficiency, glutaric aciduria type 2
C16-OH	OA (urine)	TFP/LCHAD deficiency
C18:1(C18)	OA (urine)	CPT2 deficiency, CACT deficiency, VLCAD deficiency, glutaric aciduria type 2
C18:1-OH	OA (urine)	TFP/LCHAD deficiency
C16, C18 (low)	OA (urine)	CPT1 deficiency

PKU, phenylketonuria; AA, amino acid analysis; OA, organic acid analysis; MSUD, maple syrup urine disease; MAT, methionine adenosyl transferase ; OTC, ornithine transcarbamylase; CPS1, carbamoyl phosphate synthetase I; IBD, isobutyryl CoA dehydrogenase; SCAD, short chain acyl-CoA dehydrogenase; SCHAD, short chain 3-hydroxyacyl-CoA dehydrogenase; 2MBD, 2-methylbutyryl-CoA dehydrogenase; HMG, 3-hydroxy-3-methyl-glutaryl-CoA reductase; MCG, methylcrotonylglycine; MCAD, medium-chain acyl-coenzyme A dehydrogenase; VLCAD, very long-chain acyl CoA dehydrogenase; CPT, carnitine palmitoyltransferase; CACT, carnitine acylcarnitine translocase; TFP, trifunctional protein; LCHAD, long-chain 3-hydroxyacyl-CoA dehydrogenase.

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