NAVIGENE
Genetics : Screening • Diagnosis • Research

Disorders detected on Dried Blood Spot

A. List for Disorder Screened by Tandem Mass Spectrometry (MS-MS)

Sr.No.	Group Name	Sr.No.	Disorder Name
1	Amino Acid Metabolism	1	Arginase deficiency/Argininemia
		2	Arginosuccinate synthase (ASS) deficiency
		3	Arginosuccinic aciduria
		4	Benign hyperphenylalaninemia
		5	Citrullinemia type-2 (Citrin deficiency)
		6	Classical citrullinemia
		7	Defects of biopterin cofactor biosynthesis
		8	Defects of biopterin cofactor regeneration
		9	Homocystinuria
		10	Hypermethioninemia
		11	Hyperornithinemia, Hyperammoninemia , Hyperhomocitrullinemia (HHH) syndrome
		12	Maple syrup urine disease
		13	Neonatal Tyrosinemia
		14	Phenylketonuria (PKU)
		15	Tyrosinemia I
		16	Tyrosinemia III
		17	Tyrosinemia type II
2	Fatty acid oxidation Disorders	18	Carnitine transporter deficiency (Carnitine uptake deficiency)
		19	Carnitine/acylcarnitine Translocase deficiency
		20	Carnitine palmityl transferase deficiency type I
		21	Carnitine palmityl transferase deficiency type II
		22	Short chain acyl co A dehydrogenase deficiency
		23	Short chain hydroxy acyl co A dehydrogenase deficiency
		24	Medium chain acyl CoA dehydrogenase deficiency
		25	Medium chain ketoacyl coA thiolase deficiency
		26	Very long chain acyl-CoA dehydrogenase deficiency
		27	Long chain L-3-hydroxy acyl-CoA dehydrogenase
		28	Trifunctional protein deficiency
		29	Multiple acyl-CoA dehydrogenase deficiency/Glutaric acidemia type II
		30	Medium chain L-3 hydroxy acyl coA dehydrogenase deficiency

List for Disorder Screened by Tandem Mass Spectrometry (MS-MS)

Sr.No.	Group Name	Sr.No.	Disorder Name
3	Organic Acid Metabolism	31	Glutaric acidemia type I
		32	Isovaleric acidemia
		33	2-Methylbutyryl-CoA-dehydrogenase deficiency
		34	Propionic acidemia
		35	Methylmalonyl-CoA mutase deficiency
		36	Methylmalonic acidemia
		37	Methylmalonic acidemia (mutase)
		38	3-Methylcrotonyl-CoA carboxylase deficiency
		39	Malonic aciduria
		40	3-Hydroxy 3-methyl glutaric acidemia
		41	β-Ketothiolase deficiency
		42	Multiple CoA carboxylase deficiency
		43	Isobutaryl CoA dehydrogenase deficiency
		44	Malonyl-CoA decarboxylase deficiency
		45	Holocarboxylase deficiency
		46	3- Methyl glutaconyl Co A hydratase deficiency

B. Disorder detected by other method/Technologies:

Sr.No.	Disorder Name
1	Congenital Hypothyroidism (CH)
2	Congenital Adrenal Hyperplasia (CAH)
3	Glucose 6 Phosphate Dehydrogenase Deficiency (G6PD)
4	Galactosemia (GALT)
5	Biotinidase deficiency (BIOT)
6	Phenylketonuria (PKU)
7	Cystic fibrosis (Immuno Reactive Trypsinogen/IRT)