

RECOMMENDED NEW JERSEY NBS PANEL		
Category	Abbreviation	Name of the Disorders
13 Organic Acidemias (OA)	IVA	Isovaleric Acidemia
	GAI	Glutaric Acidemia Type I
	HMG	3-hydroxy 3-methyl glutaric aciduria
	MCD	Multiple Carboxylase Deficiency
	MUT	Methylmalonic Acidemia (mutase deficiency)
	Cbl A,B	Methylmalonic Acidemia (Cbl A, B)
	3 MCC	3-Methylcrotonyl-CoA carboxylase deficiency
	PROP	Propionic acidemia
	BKT	Beta Ketothiolase deficiency
	3MGA*	3-Methylglutaconic Aciduria
	2MBG*	2-Methylbutryl-CoA dehydrogenase deficiency
	IBG*	Isobutryl-CoA dehydrogenase Deficiency
	Cbl C,D*	Methylmalonic Acidemia (Cbl C, D)
10 Fatty Acid Oxidation Disorders (FAOD)	MCAD	Medium-chain acyl-CoA dehydrogenase deficiency
	VLCAD	Very-long-chain acyl-CoA dehydrogenase deficiency
	LCAD	Long-chain acyl-CoA dehydrogenase deficiency
	SCAD	Short-chain acyl-CoA dehydrogenase deficiency
	LCHAD	Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
	TFP	Trifunctional protein deficiency
	CUD	Carnitine uptake deficiency
	GA-II*	Glutaric Acidemia Type II
	CPT-II*	Carnitine Palmitoyltransferase II deficiency
	CACT*	Carnitine/Acylcarnitine translocase deficiency
13 Amino Acid Disorders (AA)	PKU	Phenylketonuria
	H-PHE*	Benign Hyperphenylalaninemia
	BIOPT-BS*	Defects of bipterin cofactor biosynthesis
	BIOPT-REG*	Defects of bipterin cofactor regeneration
	MSUD	Maple syrup urine disease
	HCY	Homocystinuria
	TYR I	Tyrosinemia type I
	TYR II*	Tyrosinemia type II
	TYR III*	Tyrosinemia type III
	CIT	Citrullinemia
	ASA	Argininosuccinic acidemia
	CIT-II*	Citrullinemia type II
MET*	Hypermethioninemia	

4 Hemoglobinopathies	HbSS	Sickle cell anemia (Hb SS disease)
	Hb S/βTh	Hb S/β-thalassemia
	Hb S/C	Hb S/C disease
	VARIANT Hb	Variant Hb
5 Other	CH	Congenital Hypothyroidism
	BIOT	Biotinidase deficiency
	CAH	Congenital Adrenal Hyperplasia
	GALT	Galactosemia
	CF	Cystic Fibrosis
TOTAL: 45		
Footnote: * Secondary targets which are recommended to be mandated.		

NON-MANDATED DISORDERS WHICH MAY VERY RARELY BE DETECTED	
Abbreviations	Name of the Disorder
MS/MS Detectable disorders	
ARG	Argininemia
MAL	Malonic Aciduria
CPT I-A	Carnitine Palmitoyl transferase I-A deficiency
M/SCHAD	Medium/Short Chain 3-OH acyl-CoA DH deficiency
MCKAT	Medium Chain Ketoacyl-CoA thiolase deficiency
2M3HBA	2-Methyl 3-hydroxy butyric aciduria
DE RED	Dienoyl-CoA reductase deficiency
Non MS/MS Disorders	
GALE	Galactose Epimerase deficiency
GALK	Galactokinase deficiency