

**New Jersey Department of Health
Division of Family Health Services
Newborn Screening and Genetic Services Program**

Year Screening Began	Program to Date Data from 1964 to December, 2015 As of 7/28/2016		# of Babies with Confirmed Classic Disease	# of Babies with Variant Disease or Carrier Status
<u>Newborn Screening Disorders</u>				
2001	Biotinidase Deficiency	BIOT	28	184
2001	Congenital Adrenal Hyperplasia	CAH	85	48
2001	Cystic Fibrosis	CF	217	443
1978	Congenital Hypothyroidism	CH	1743	141
1982, 2009, 2009	Galactosemia, Galactosepimerase Deficiency, Galactokinase Deficiency	GALT, GALE, GALK	91	980
2001	Maple Syrup Urine Disease	MSUD	11	1
1964	Phenylketonuria, Hyperphenylalanemia (benign), Biopterin Cofactor defect of Biosynthesis or Regeneration	PKU, H-PHE, Biopt-Bio, Biopt-Reg	357	173
1990	Sickle Cell Anemia and Other Hemoglobinopathies	S/S, S/C, S/B-Thal, Var Hgb	1022	1019
2014	Severe Combined Immunodeficiency	SCID	3	20
<i>Amino Acid Disorders</i>				
2009	Homocystinuria	HCY	5	0
2009	Hypermethioninemia	MET	2	3
2009	Tyrosinemia Types I, II, III	TYR I, II, III	2	65
<i>Fatty Acid Disorders</i>				
2009	Carnitine/Acylcarnitine Translocase Deficiency	CACT	0	0
2009	Carnitine Palmitoyltransferase Deficiency, Type IA	CPT-1A	2	0
2009	Carnitine Palmitoyltransferase Deficiency, Type II	CPT-II	4	0
2009	Carnitine Uptake Defect	CUD	15	8
2009	Dienoyl-CoA Reductase Deficiency	DERED	0	0
2009	Glutaric Acidemia, Type II	GA-II	4	1
2002	Long/Very Long Chain Acyl-CoA Dehydrogenase Deficiency	LCAD/VLCAD	23	8
2009	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD	3	0
2009	Medium/Short Chain 3-OH Acyl-CoA Dehydrogenase Deficiency	M/SCHAD	1	0
2002	Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD	69	9
2009	Medium Chain Ketoacyl-CoA Thiolase Deficiency	MCKAT	1	0
2002	Short Chain Acyl-CoA Dehydrogenase Deficiency	SCAD	80	25
2009	Trifunctional Protein Deficiency	TFP	2	0
<i>Organic Acid Disorders</i>				
2009	2-Methyl-3-Hydroxybutyric Acidemia	2M3HBA	0	0
2009	2-Methylbutyryl-CoA Dehydrogenase Deficiency	2MBG	1	0
2003	3-Methylcrotonyl-CoA Carboxylase Deficiency	3MCC	33	12
2009	3-Methylglutaconyl CoA Hydrastase Deficiency	3MGA	0	2
2009	Mitochondrial Acetoacyl CoA Thiolase Deficiency	BKT	1	0
2003	Glutaric Acidemia, Type I	GA-1	6	0
2003	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMG	8	0
2009	Isobutyryl-CoA Dehydrogenase Deficiency	IBD	8	2
2003	Isovaleryl-CoA Dehydrogenase Deficiency	IVA	8	2
2009	Malonyl-CoA Decarboxylase Deficiency	MAL	1	0
2009	Multiple Carboxylase Deficiency	MCD	0	0
2003	Methylmalonic Acidemia [Mutase Deficiency or Defects in Cobalamin A/B, or Cobalamin C/D]	MUT, CBL A/B, CBL C/D	9	18
2003	Propionyl-CoA Carboxylase Deficiency	PROP	7	1
<i>Urea Cycle Disorders</i>				
2009	Argininemia	ARG	2	0
2002	Argininosuccinate Lyase Deficiency	ASA	4	0
2002	Citrullinemia Types I, II	CIT I, II	5	2
TOTALS			3901	3148