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, 2020 As of 8/28/2021 **Newborn Screening Disorders**		# of Babies with Confirmed Classic Disease	# of Babies with Variant Disease or Carrier Status
2001	Biotinidase Deficiency	BIOT	39	204
2001	Congenital Adrenal Hyperplasia	CAH	101	58
2001	Cystic Fibrosis	CF	256	545
1978	Congenital Hypothyroidism	CH	2124	168
1982, 2009, 2009	Galactosemia, Galactoepimerase Deficiency, Galactokinase Deficiency	GALT, GALE, GALK	112	1262
2001	Maple Syrup Urine Disease	MSUD	12	1
1964	Phenylketonuria, Hyperphenylalanemia (benign), Biopterin Cofactor defect of Biosynthesis or Regeneration	PKU, H-PHE, Biopt-Bio, Biopt-Reg	317	191
1990	Sickle Cell Anemia and Other Hemoglobinopathies	S/S, S/C, S/B-Thal, Var Hgb	1298	1288
2014	Severe Combined Immunodeficiency	SCID	8	38
	Amino Acid Disorders			
2009	Homocystinuria	HCY	3	1
2009	Hypermethioninemia	MET	1	5
2009	Tyrosinemia Types I, II, III	TYR I, II, III	2	94
	Fatty Acid Disorders			
2009	Carnitine/Acylcarnitine Translocase Deficiency	CACT	0	0
2009	Carnitine Palmitoyltransferase Deficiency, Type IA	CPT-1A	3	1
2009	Carnitine Palmitoyltransferase Deficiency, Type II	CPT-II	2	0
2009	Carnitine Uptake Defect	CUD	12	14
2009	Dienoyl-CoA Reductase Deficiency	DERED	0	0
2009	Glutaric Acidemia, Type II	GA-II	2	1
2002	Long/Very Long Chain Acyl-CoA Dehydrogenase Deficiency	LCAD/ VLCAD	30	17
2009	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD	2	0
2009	Medium/Short Chain 3-OH Acyl-CoA Dehydrogenase Deficiency	M/SCHAD	1	0
2002	Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD	89	17
2009	Medium Chain Ketoacyl-CoA Thiolase Deficiency	MCKAT	1	0
2002	Short Chain Acyl-CoA Dehydrogenase Deficiency	SCAD	96	29
2009	Trifunctional Protein Deficiency	TFP	3	1
	Organic Acid Disorders			
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	42	22
2009	3-Methylglutaconyl CoA Hydrastase Deficiency	3MGA	5	0
2009	Mitochondrial Acetoacyl CoA Thiolase Deficiency	BKT	1	0
2003	Glutaric Acidemia, Type I	GA-1	11	1
2003	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMG	9	0
2009	Isobutyryl-CoA Dehydrogenase Deficiency	IBD	8	2
2003	Isovaleryl-CoADehydrogenase Deficiency	IVA	13	2
2009	Malonyl-CoA Decarboxylase Deficiency	MAL	0	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	6	17
2003	Propionyl-CoA Carboxylase Deficiency	PROP	9	1
	Urea Cycle Disorders			
2009	Argininemia	ARG	4	0
2002	Argininosuccinate Lyase Deficiency	ASA	8	0
2002	Citrullinemia Types I, II	CIT I, II	6	3
	Lysosomal Storage Disorders			
2019	Fabry	Fabry	7	0
2019	Gaucher	Gaucher	6	0
2019	Krabbe	Krabbe	1	21
2019	Mucopolysaccharidosis Type 1	MPS1	2	10
2019	Niemann Pick	Nie-pick	0	1
2019	Pompe	Pompe	1	2
	TOTALS		**4637**	**3983**