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

2009 Data Newborn Screening Disorders		# of Babies with Confirmed Classic Disease	# of Babies with Variant Disease or Carrier Status	# of Babies with cleared results
Endocrine Disorders				
Congenital Adrenal Hyperplasia	CAH	3	4	1067
Congenital Hypothyroidism	CH	70	9	1727
Metabolic Disorders				
Biotinidase Deficiency	BIOT	1	3	37
Galactosemia	GALT	1	20	37
Galactoepimerase Deficiency, Galactokinase Deficiency	GALE, GALK	1		
Other Disorders				
Cystic Fibrosis	CF	10	42	190
Sickle Cell Anemia and Other Hemoglobinopathies	S/S, S/C, S/B-Thal, Var Hb	34	46	7
Hemoglobin Traits			(2919)	
Amino Acid and Urea Cycle Disorders				
Argininemia	ARG	0	0	5
Argininosuccinate Lyase Deficiency	ASA	0	0	
Citrullinemia Types I, II	CIT I, II	1	0	
Homocystinuria	HCY	1	0	131
Hypermethioninemia	MET	0	0	
Tyrosinemia Types I, II, III	TYR I, II, III	1	7	
Maple Syrup Urine Disease	MSUD	1	0	0
Phenylketonuria, Hyperphenylalanemia (benign), Biopterin Cofactor defect of Biosynthesis or Regeneration	PKU, H-PHE, Biopt-Bio, Biopt-Reg	3	7	7
Fatty Acid Disorders				
Carnitine Palmitoyltransferase Deficiency, Type IA	CPT-1A	0	0	62
Carnitine Palmitoyltransferase Deficiency, Type II	CPT-II	1	0	
Carnitine Uptake Defect	CUD	0	1	
Carnitine/Acylcarnitine Translocase Deficiency	CACT	0	0	
Dienoyl-CoA Reductase Deficiency	DERED	0	0	
Glutaric Aciduria, Type II	GA-II	0	0	
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD	1	0	
Long/Very Long Chain Acyl-CoA Dehydrogenase Deficiency	LCAD/VLCAD	1	0	
Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD	4	0	
Medium Chain Ketoacyl-CoA Thiolase Deficiency	MCKAT	0	0	
Medium/Short Chain 3-OH Acyl-CoA Dehydrogenase Deficiency	M/SCHAD	0	0	
Short Chain Acyl-CoA Dehydrogenase Deficiency	SCAD	9	3	
Trifunctional Protein Deficiency	TFP	0	0	
Organic Acid Disorders				
2-Methyl-3-Hydroxybutyric Acidemia	2M3HBA	0	0	**101**
2-Methylbutyryl-CoA Dehydrogenase Deficiency	2MBG	0	0	
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMG	1	0	
3-Methylcrotonyl-CoA Carboxylase Deficiency	3MCC	6	2	
3-Methylglutaconyl CoA Hydrastase Deficiency	3MGA	0	0	
Glutaric Acidemia, Type I	GA-1	1	0	
Isobutyryl-CoA Dehydrogenase Deficiency	IBD	1	0	
Isovaleryl-CoADehydrogenase Deficiency	IVA	1	1	
Malonyl-CoA Decarboxylase Deficiency	MAL	0	0	
Methylmalonic Acidemia [Mutase Deficiency or Defects in Cobalamin A/B, or C/D]	MUT, CBL A/B, CBL C/D	0	1	
Mitochondiral Acetoacyl CoA Thiolase Deficiency	BKT	0	0	
Multiple Carboxylase Deficiency	MCD	0	0	
Propionyl-CoA Carboxylase Deficiency	PROP	0	0	
TOTALS		**152**	**147**	**3371**