Newborn Screening Program - 2025 Annual Report

January 1-December 31, 2025

New York State Department of Health

Wadsworth Center

120 New Scotland Ave

Albany, NY

Repeat Requests	35,527
Borderline Results	18,065
Referrals	2,018

Specimens Received	
Initial Specimens	204,235
Repeat Specimens	43,825
Total Specimens	248,060
Total Newborns	204,235

Screened Disorders	Primary Analytes	Borderline	Referrals	Number Confirmed with Disease (Preliminary)	
Endocrine Disorders					
Congenital Adrenal Hyperplasia (CAH)	17-Hydroxyprogesterone (17-OHP)	1317	139	Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency	11
				Congenital Adrenal Hyperplasia, Other Enzyme Deficiency	0
Congenital Hypothyroidism (CH)	Thyroid Stimulating Hormone (TSH)	7038	789	Primary Congenital Hypothyroidism	130
				Secondary Congenital Hypothyroidism	0
				Other	215
Hemoglobin Disorders					
Hemoglobin Disorders	Hemoglobin SS	N/A	110	Hemoglobin S + S Disease	93
	Hemoglobin SC	N/A	46	Hemoglobin S + C Disease	43
	Hemoglobin CC	N/A	16	Hemoglobin C + C Disease	13
	Other Hemoglobinopathies	N/A	45	Other Hemoglobinopathies	40
	Hemoglobin S Trait	4020	N/A		N/A
	Hemoglobin C Trait	1172	N/A		N/A
	Hemoglobin D Trait	108	N/A		N/A
	Hemoglobin Variant Trait	104	N/A		N/A
Infectious Disease					
Human Immunodeficiency Virus (HIV)	HIV Antibodies	N/A	296	Confirmed perinatal HIV transmission data prepared by the AIDS Institute (HIV Annual Surveillance Reports)*	
Amino Acid Disorders					
Maple Syrup Urine Disease (MSUD)	Leucine	104	1	Maple Syrup Urine Disease (MSUD)	1
				Hydroxyprolinemia	0
Homocystinuria (HCY)	Methionine	387	8	Homocystinuria (HCY)	0
				Hypermethioninemia (MET)	1
Phenylketonuria (PKU)	Phenylalanine	187	22	Phenylketonuria (PKU)	18
				Hyperphenylalaninemia (HPA)	3
Tyrosinemia Type I (TYR I)	Succinylacetone	0	2	Tyrosinemia Type I (TYR I)	0
Tyrosinemia Type II, III (TYR II, III)	Tyrosine	231	12	Tyrosinemia Type II (TYR II)	0
				Tyrosinemia Type III (TYR III)	0
Fatty Acid Oxidation Disorders					
Carnitine Uptake Defect (CUD)	Free Carnitine (C0), Total Acylcarnitines	271	8	Carnitine Uptake Defect (CUD)	0
Carnitine Palmitoyltransferase I (CPT I) Deficiency	C0/(C16 + C18)	91	1	Carnitine Palmitoyltransferase I (CPT I) Deficiency	0
Carnitine Palmitoyltransferase II (CPT II) Deficiency/Carnitine-Acylcarnitine Translocase Deficiency	Hexadecanoylcarnitine (C16), Octadecenoylcarnitine (C18:1)	1	8	Carnitine Palmitoyltransferase II (CPT II) Deficiency	1
2,4-Dienoyl-CoA Reductase Deficiency (DECRD)	Decadienoylcarnitine (C10:2)	160	1	2,4-Dienoyl-CoA Reductase Deficiency (DECRD)	0
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency/Trifunctional Protein Deficiency (TFP)	Hydroxyhexadecanoylcarnitine (C16OH), Hydroxyoctadecenoylcarnitine (C18:1OH)	4	5	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency	0
				Trifunctional Protein (TFP) Deficiency	0
Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency/ Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency/Medium-Chain 3-Keto Acyl-CoA Thiolase (MCKAT) Deficiency	Hexanoylcarnitine (C6), Octanoylcarnitine (C8)	366	10	Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	4
				Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency/Glutaric Acidemia Type II (GA II)	0
				Medium-Chain 3-Keto Acyl-CoA Thiolase (MCKAT) Deficiency	0
Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	Tetradecanoylcarnitine (C14), Tetradecenoylcarnitine (C14:1)	2	13	Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	2

Screened Disorders	Primary Analytes	Borderline	Referrals	Number Confirmed with Disease (Preliminary)	
Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	Butyrylcarnitine (C4)	196	23	Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	0
Medium/Short-Chain Hydroxyl CoA Dehydrogenase (M/SCHAD) Deficiency	Hydroxybutyrylcarnitine (C4OH), Hydroxyhexanoylcarnitine (C6OH)	111	2	Medium/Short-Chain Hydroxyl CoA Dehydrogenase (M/SCHAD) Deficiency	0
Organic Acid Disorders					
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency/2-Methyl-3-Hydroxybutyryl-CoA-Dehydrogenase (MHBD) Deficiency	Tiglylcarnitine (C5:1)	25	0	Mitochondrial Acetoacetyl-CoA Thiolase Deficiency/Beta-Ketothiolase (BKT) Deficiency	0
				2-Methyl-3-Hydroxybutyryl-CoA-Dehydrogenase (MHBD) Deficiency	0
Glutaric Aciduria Type I (GA I)	Glutarylcarnitine (C5DC)	154	6	Glutaryl-CoA Dehydrogenase Deficiency/Glutaric Aciduria Type I (GA I)	2
Isovaleric Acidemia (IVA)/2-Methylbutyryl-CoA Dehydrogenase (2MBCD) Deficiency	Isovalerylcarnitine (C5)	586	11	Isovaleryl CoA Dehydrogenase Deficiency/Isovaleric Acidemia (IVA)	1
				2-Methylbutyrylglycinuria (2MBG)/2-Methylbutyryl-CoA Dehydrogenase (2MBCD) Deficiency/Short/Branched Chain Acyl-CoA Dehydrogenase (SBCAD) Deficiency	3
3-Methylcrotonyl-CoA Carboxylase (3MCC) Deficiency/2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase (MHBD) Deficiency/3-Methylglutaconic Aciduria (3MGA)	Hydroxyisovalerylcarnitine (C5OH)	26	68	3-Methylcrotonyl-CoA Carboxylase (3MCC) Deficiency	7
				3-Hydroxy-3-Methylglutaryl-CoA Lyase (HMG) Deficiency	0
				2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase (MHBD) Deficiency/2-Methyl-3-Hydroxybutric Acidemia (2M3HBA)	0
				3-Methylglutaconic Aciduria (3MGA)	0
Malonyl-CoA Decarboxylase Deficiency (MA)	Malonylcarnitine (C3DC)	2	1	Malonyl-CoA Decarboxylase Deficiency/Malonic Aciduria (MA)	0
Propionyl-CoA Carboxylase Deficiency (PA)/Methylmalonyl-CoA Mutase Deficiency (MMA)	Propionylcarnitine (C3), Methylmalonylcarnitine (C4DC)	444	22	Propionyl-CoA Carboxylase Deficiency (PA)	1
				Methylmalonyl-CoA Mutase Deficiency (MMA)	0
				Cobalamin A/B Deficiency	0
				Cobalamin C/D/F Deficiency	1
				Multiple Carboxylase Deficiency (MCD)	0
Isobutyryl-CoA Dehydrogenase (IBCD) Deficiency	Butyrylcarnitine (C4)	196	23	Isobutyryl-CoA Dehydrogenase (IBCD) Deficiency	1
Urea Cycle Disorders					
Argininosuccinic Aciduria (ASA)/Citrullinemia (CIT)	Citrulline	66	7	Argininosuccinic Aciduria (ASA)	1
				Citrullinemia Type I (CIT I)	2
Argininemia (ARG)	Arginine	32	1	Argininemia (ARG)	1
Lysosomal Disorders					
Krabbe Disease	Galactocerebrosidase	N/A	6	Krabbe Disease, Infantile-Onset	0
				Krabbe Disease, Possible Late-Onset	0
Mucopolysaccharidosis Type I (MPS I)	Alpha-L-iduronidase	N/A	8	Mucopolysaccharidosis Type I (MPS I)	0
Pompe Disease	Alpha-glucosidase	N/A	13	Pompe Disease, Infantile-Onset	1
				Pompe Disease, Possible Late-Onset	10
Other Genetic Conditions					
X-linked Adrenoleukodystrophy (X-ALD)	C26:0 Lysophosphatidylcholine (C26:0 LPC)	19	23	X-linked Adrenoleukodystrophy (X-ALD)	4
				Zellweger Syndrome	3
				Other Peroxisomal Biogenesis Disorder	0
Biotinidase Deficiency (BTD)	Biotinidase	9	4	Biotinidase Deficiency (BTD)	4
Cystic Fibrosis (CF)	Immunoreactive Trypsinogen	N/A	105	Cystic Fibrosis (CF)	21
Spinal Muscular Atrophy (SMA)	*SMN1* Gene, Exon 7 Deletion	N/A	13	Spinal Muscular Atrophy (SMA)	13
Guanidinoacetate Methyltransferase (GAMT) Deficiency	Guanidinoacetate	77	1	Guanidinoacetate Methyltransferase (GAMT) Deficiency	1
Galactosemia (GALT)	Galactose Transferase	1	3	Galactosemia (GALT)	3
Severe Combined Immunodeficiency (SCID)	T-cell Receptor Excision Circles (TRECS)	558	146	Classic SCID	6
				Leaky SCID	1
				Variant SCID	0
Total		18,065	2,018		662

Data is based on infants born in 2025 whose specimens were received before 6/1/2026. Repeats are requested for specimens of suboptimal or unsuitable quality or quantity and for specimens with borderline results.

Borderline result counts are provided per analyte; specimens with multiple analytes in the borderline range are counted more than once. Screen-positive referrals, confirmed cases, and hemoglobin trait counts are provided per infant.

*https://www.health.ny.gov/diseases/aids/general/statistics/