Newborn Screening Program - 2024 Annual Report
Jan 1-Dec 31 2024
New York State Department of Health
Wadsworth Center
120 New Scotland Ave
Albany, NY

Repeat Requests	22,498
Borderlines	16,213
Referrals	1,866

Specimens Received

Initial Valid	186,085
Initial Invalid	20,409
Total Newborns	206,728
Repeat Specimens	40,001
Total Specimens	246,895

Screened Disorders	Analytes	Borderline	Referrals	Number Confirmed with Disease (Preliminary)	
Endocrine Disorders					
Congenital Adrenal Hyperplasia (CAH)	17-Hydroxyprogesterone (17-OHP)	1197	118	Congenital Adrenal Hyperplasia - 21-Hydroxylase Deficiency	6
				Congenital Adrenal Hyperplasia, Other Enzyme Deficiency	0
Congenital Hypothyroidism (CH)	Thyroid Stimulating Hormone (TSH)	5878	633	Primary Congenital Hypothyroidism	116
				Secondary Congenital Hypothyroidism	0
				Other	216
Hemoglobin Disorders					
Hemoglobin Disorders	Hemoglobin SS	N/A	111	Hemoglobin S + S (Sickle Cell) Disease	96
	Hemoglobin SC	N/A	68	Hemoglobin S + C Disease	64
	Hemoglobin CC	N/A	28	Hemoglobin C + C Disease	21
	Other Hemoglobins	N/A	29	Other Hemoglobinopathies	28
	Hemoglobin AS Trait	4247	N/A		N/A
	Hemoglobin AC Trait	1224	N/A		N/A
	Hemoglobin AD Trait	121	N/A		N/A
	Hemoglobin A VAR Trait	128	N/A		N/A
Infectious Disease					
Human Immunodeficiency Virus (HIV)	HIV Antibodies	N/A	274	Confirmed by diagnosis developed by the AIDS Institute.	
Amino Acid Disorders					
Maple Syrup Urine Disease (MSUD)	Leucine	55	2	Maple Syrup Urine Disease (MSUD)	0
				Hydroxyprolinemia	0
Homocystinuria (HCY)	Methionine	411	5	Homocystinuria (HCY)	0
				Hypermethioninemia (MET)	1
Phenylketonuria (PKU)	Phenylalanine	155	18	Phenylketonuria (PKU)	15
				Hyperphenylalaninemia (HPA)	2
Tyrosinemia Type I (TYR I)	Succinylacetone	0	0	Tyrosinemia Type I (TYR I)	0
Tyrosinemia Type II, III (TYR II, III)	Tyrosine	187	11	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 (SUM AC)	353	15	Carnitine Uptake Defect (CUD)	1
Carnitine Palmitoyltransferase I (CPT I) Deficiency	C0/(C16 + C18)	86	4	Carnitine Palmitoyltransferase I (CPT I) Deficiency	1
Carnitine Palmitoyltransferase II (CPT II) Deficiency/Carnitine-Acylcarnitine Translocase Deficiency	Hexadecanoylcarnitine (C16), Octadecenoylcarnitine (C18:1)	3	12	Carnitine Palmitoyltransferase II (CPT II) Deficiency	2
2,4-Dienoyl-CoA Reductase Deficiency (DECRD)	Decadienoylcarnitine (C10:2)	45	0	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	3	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency	1
				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)	174	12	Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	9
				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 Deficiency (VLCAD)	Tetradecanoylcarnitine (C14), Tetradecenoylcarnitine (C14:1)	5	8	Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	2

Screened Disorders	Analytes	Borderline	Referrals	Number Confirmed with Disease (Preliminary)	
Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)	Butyrylcarnitine (C4)	70	19	Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	6
Medium/Short-Chain Hydroxyl CoA Dehydrogenase (M/SCHAD) Deficiency	Hydroxybutyrylcarnitine (C4OH), Hydroxyhexanoylcarnitine (C6OH)	44	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)	18	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)	105	8	Glutaryl-CoA Dehydrogenase Deficiency/Glutaric Aciduria Type I (GA I)	1
Isovaleric Acidemia (IVA)/2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBCD)	Isovalerylcarnitine (C5)	390	3	Isovaleryl CoA Dehydrogenase Deficiency/Isovaleric Acidemia (IVA)	2
				2-Methylbutyrylglycinuria (2MBG)/2-Methylbutyryl-CoA Dehydrogenase (2MBCD) Deficiency/Short/Branched Chain Acyl-CoA Dehydrogenase (SBCAD) Deficiency	0
3-Methylcrotonyl-CoA Carboxylase (3MCC) Deficiency/2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase (MHBD) Deficiency/3-Methylglutaconic Aciduria (3MGA)	Hydroxyisovalerylcarnitine (C5OH)	46	57	3-Methylcrotonyl-CoA Carboxylase (3MCC) Deficiency	8
				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)	4	1	Malonyl-CoA Decarboxylase Deficiency/Malonic Aciduria (MA)	1
Propionyl-CoA Carboxylase Deficiency (PA)/Methylmalonyl-CoA Mutase Deficiency (MMA)	Propionylcarnitine (C3), Methylmalonylcarnitine (C4DC)	272	28	Propionyl-CoA Carboxylase Deficiency (PA)	2
				Methylmalonyl-CoA Mutase Deficiency (MMA)	0
				Cobalamin A/B Deficiency	0
				Cobalamin C/D/F Deficiency	2
				Multiple Carboxylase Deficiency (MCD)	0
Isobutyryl-CoA Dehydrogenase Deficiency (IBCD)	Butyrylcarnitine (C4)	70	19	Isobutyryl-CoA Dehydrogenase (IBCD) Deficiency	2
Urea Cycle Disorders					
Argininosuccinic Aciduria (ASA)/Citrullinemia (CIT)	Citrulline	45	6	Argininosuccinic Aciduria (ASA)	2
				Citrullinemia Type I (CTLN1)	0
Argininemia (ARG)	Arginine	17	0	Argininemia (ARG)	0
Lysosomal Storage Disorders					
Krabbe Disease	Galactocerebrosidase	0	9	Infantile-Onset Krabbe Disease	2
				Krabbe Disease Possible Late-Onset	0
Mucopolysaccharidosis Type I (MPS I)	Alpha-L-iduronidase	0	9	Mucopolysaccharidosis Type I (MPS I)	1
Pompe Disease	Alpha-glucosidase	0	10	Infantile-Onset Pompe Disease	0
				Pompe Disease Possible Late-Onset	5
Other Genetic Conditions					
X-linked Adrenoleukodystrophy (X-ALD)	C26:0 Lysophosphatidylcholine (C26:0 LPC)	17	12	Male with X-linked Adrenoleukodystrophy (X-ALD)	6
				Female Carrier of X-ALD	5
				Zellweger Syndrome	1
				Other Peroxisomal Biogenesis Disorder	0
Biotinidase Deficiency (BTD)	Biotinidase	14	2	Biotinidase Deficiency (BTD)	2
Cystic Fibrosis (CF)	Immunoreactive Trypsinogen	N/A	111	Cystic Fibrosis (CF)	22
Spinal Muscular Atrophy (SMA)	*SMN1* Gene, Exon 7 Deletion	N/A	13	Spinal Muscular Atrophy (SMA)	13
Guanidinoacetate Methyltransferase Deficiency (GAMT)	Guanidinoacetate	38	1	Guanidinoacetate Methyltransferase Deficiency (GAMT)	0
Galactosemia (GALT)	Galactose Transferase	8	3	Galactosemia (GALT)	2
Severe Combined Immunodeficiency (SCID)	T-cell Receptor Excision Circles (TRECS)	782	202	Classic SCID	5
				Leaky SCID	0
				Variant SCID	0
Total		16,213	1,866		671

Data is based on infants born in 2024 whose specimens were received before 3/1/25.

Borderline result counts are provided per specimen. An infant with multiple specimens with borderline results for a given condition may be counted more than once.

Screen-positive referrals, confirmed cases and hemoglobin trait counts are provided per infant.