Newborn Screening Program - 2023 Annual Report
Jan 1-Dec 31 2023
New York State Department of Health
Wadsworth Center
David Axelrod Institute
120 New Scotland Avenue, Albany, NY 12208

Repeat Requests	21438
Borderlines	14269
Referrals	1711

Specimens Received

Initial Valid	184,838
Initial Invalid	19,667
Total Newborns	204,505
Repeat Specimens	37,872
Total Specimens	242,779

Screened Disorders	Analytes	Borderline	Referrals	# Confirmed with Disease	
Endocrine Disorders					
Congenital Adrenal Hyperplasia	17-hydroxyprogesterone	1170	118	Congenital adrenal hyperplasia - 21-Hydroxylase deficiency	8
				Congenital adrenal hyperplasia, other enzyme deficiency	0
Congenital Hypothyroidism	Thyroxine, TSH	4405	608	Primary Congenital Hypothyroidism	114
				Secondary Congenital Hypothyroidism	0
				Other	198
Hemoglobin Disorders					
Hemoglobin Disorders	Hemoglobin SS	N/A	92	Hemoglobin S + S (sickle cell) disease	80
	Hemoglobin SC	N/A	59	Hemoglobin S + C disease	58
	Hemoglobin CC	N/A	14	Hemoglobin C + C disease	12
	Other Hemoglobins	N/A	36	Other Hemoglobinopathies	
	Hemoglobin AS Trait	4168	N/A		
	Hemoglobin AC Trait	1221	N/A		
	Hemoglobin AD Trait	87	N/A		
	Hemoglobin A VAR Trait	100	N/A		27
Infectious Disease					
HIV	HIV Antibodies	N/A	297	Confirmed by diagnosis developed by the AIDS Institute	
Amino Acid Disorders					
Maple Syrup Urine Disease	Leucine	91	2	Maple Syrup Urine disease	0
				Hydroxyprolinemia	0
Homocystinuria	Methionine	417	5	Homocystinuria	1
				Hypermethioninemia	1
Phenylketonuria	Phenylalanine	186	25	Phenylketonuria (PKU)	11
				Hyperphenylalaninemia	10
Tyrosinemia Type I	Succinylacetone	0	1	Tyrosinemia Type 1	0
Tyrosinemia Type II, III	Tyrosine	187	8	Tyrosinemia Type 2	0
				Tyrosinemia Type 3	0
Fatty Acid Oxidation Disorders					
Carnitine uptake defect	Free Carnitine (C0), Total Acylcarnitines (SUM AC)	308	9	Carnitine uptake defect (CUD)	1
Carnitine palmitoyltransferase 1 deficiency	C0/(C16 + C18)	88	4	Carnitine palmitoyltransferase 1 (CPT1) deficiency	0
Carnitine palmitoyltransferase 2 deficiency/Carnitine/Acylcarnitine translocase deficiency	Hexadecanoylcarnitine (C16), Octadecenoylcarnitine (C18:1)	1	5	Carnitine palmitoyltransferase 2 (CPT2) deficiency	0
2,4-Dienoyl-CoA reductase deficiency	Decadienoylcarnitine (C10:2)	50	0	2,4-Dienoyl-CoA (2,4Di) reductase deficiency	0
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency/Trifunctional protein deficiency	Hydroxyhexadecanoylcarnitine (C16OH), Hydroxyoctadecenoylcarnitine (C18:1OH)	1	2	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	0
				Trifunctional protein (TFP) deficiency	0
Multiple acyl-CoA dehydrogenase deficiency/ Medium-chain acyl-CoA dehydrogenase deficiency/Medium-chain 3-keto acyl-CoA thiolase deficiency	Hexanoylcarnitine (C6), Octanoylcarnitine (C8)	215	20	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	6
				Multiple acyl-CoA dehydrogenase (MAD) deficiency - glutaric acidemia type II (GA-II)	1
				Medium-chain 3-keto acyl-CoA thiolase (MCKAT) deficiency	0
Very long-chain acyl-CoA dehydrogenase deficiency	Tetradecanoylcarnitine (C14), Tetradecenoylcarnitine (C14:1)	0	13	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	5

Screened Disorders	Analytes	Borderline	Referrals	# Confirmed with Disease	
Short-chain acyl-CoA dehydrogenase deficiency	Butyrylcarnitine (C4)	104	19	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	8
				Isobutyryl-CoA dehydrogenase (IBCD) deficiency	0
Medium/short-chain hydroxyl CoA dehydrogenase deficiency	Hydroxybutyrylcarnitine (C4OH), Hydroxyhexanoylcarnitine (C6OH)	69	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 deficiency	Tiglylcarnitine (C5:1)	15	0	Mitochondrial acetoacetyl-CoA thiolase deficiency - beta-ketothiolase (BKT) deficiency	0
				2-Methyl-3-hydroxybutyryl-CoA-dehydrogenase (MHBD) deficiency	0
Glutaryl-CoA dehydrogenase deficiency	Glutarylcarnitine (C5DC)	93	10	Glutaryl-CoA dehydrogenase deficiency - glutaric aciduria (GA-I)	4
Isovaleryl CoA dehydrogenase deficiency/2-methylbutyryl-CoA dehydrogenase deficiency	Isovalerylcarnitine (C5)	385	8	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 deficiency/2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency/3-Methylglutaconic aciduria	Hydroxyisovalerylcarnitine (C5OH)	49	54	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	Malonylcarnitine (C3DC)	4	1	Malonyl-CoA decarboxylase deficiency - Malonic Aciduria (MA)	1
Propionyl-CoA carboxylase deficiency/Methylmalonyl-CoA mutase deficiency	Propionylcarnitine (C3), Methylmalonylcarnitine (C4DC)	256	30	Propionyl-CoA carboxylase deficiency (PA)	3
				Methylmalonyl-CoA mutase deficiency (MMA)	1
				Cobalamin A/B deficiency	0
				Cobalamin C/D/F deficiency	0
				Multiple Carboxylase deficiency	0
Urea Cycle Disorders					
Argininosuccinic aciduria/Citrullinemia	Citrulline	52	13	Argininosuccinic aciduria	0
				Citrullinemia	4
Argininemia	Arginine	30	1	Argininemia	1
Lysosomal Storage Disorders					
Krabbe Disease	Galactocerebrosidase	0	13	Krabbe disease possible late onset *	3
Mucopolysaccharidosis Type I	Alpha-L-iduronidase	0	13	MPS 1	2
Pompe Disease	Alpha-glucosidase	0	12	Infantile-onset Pompe Disease	0
				Possible late-onset Pompe disease	8
Other Genetic Conditions					
Adrenoleukodystrophy	C26:0 Lysophosphatidylcholine (C26:0 LPC)	8	11	Male with X-linked Adrenoleukodystrophy (X-ALD)	4
				Female carrier of X-ALD	4
				Zellweger Syndrome	0
				Other Peroxisomal Biogenesis Disorder	0
Biotinidase Deficiency	Biotinidase	9	4	Biotinidase Deficiency	4
Cystic Fibrosis (CF)	Immunoreactive Trypsinogen	N/A	104	Cystic Fibrosis	25
Spinal Muscular Atrophy (SMA)	SMN1 gene, exon 7 deletion	N/A	5	Spinal Muscular Atrophy	5
Guanidinoacetate methyltransferase deficiency (GAMT)	Guanidinoacetate	43	0	Guanidinoacetate methyltransferase deficiency	0
Galactosemia	Galactose Transferase	11	2	Galactosemia	2
Severe Combined Immunodeficiency (SCID)	T-cell receptor excision circles (TRECS)	446	91	Classic SCID	2
				Leaky SCID	1
				Variant SCID	2
Total		14269	1711		626

Data based on infants born in 2023 whose specimens were received before 4/4/24.

* Infants classified as confirmed for Krabbe disease include those at high risk for disease based on confirmatory enzyme activity testing