Newborn Screening Program - 2022 Annual Report
Jan 1-Dec 31 2022
New York State Department of Health
Wadsworth Center
Biggs Laboratory
Albany, NY

Repeat Requests	21191
Borderlines	9201
Referrals	1775

Specimens Received

Initial Valid	188,872
Initial Invalid	19,642
Total Newborns	208,514
Repeat Specimens	38,328
Total Specimens	246,842

Screened Disorders	Analytes	Borderline	Referrals	# Confirmed with Disease (Preliminary)	
Endocrine Disorders					
Congenital Adrenal Hyperplasia	17-hydroxyprogesterone	976	98	Congenital adrenal hyperplasia - 21-Hydroxylase deficiency	12
				Congenital adrenal hyperplasia, other enzyme deficiency	0
Congenital Hypothyroidism	Thyroxine, TSH	4830	612	Primary Congenital Hypothyroidism	104
				Secondary Congenital Hypothyroidism	1
				Other	208
Hemoglobin Disorders					
Hemoglobin Disorders	Hemoglobin SS	N/A	106	Hemoglobin S + S (sickle cell) disease	91
	Hemoglobin SC	N/A	57	Hemoglobin S + C disease	56
	Hemoglobin CC	N/A	17	Hemoglobin C + C disease	13
	Other Hemoglobins	N/A	26	Other Hemoglobinopathies	22
Infectious Disease					
HIV	HIV Antibodies	N/A	258	Confirmed by diagnosis developed by the AIDS Institute	
Amino Acid Disorders					
Maple Syrup Urine Disease	Leucine	114	3	Maple Syrup Urine disease	1
				Hydroxyprolinemia	0
Homocystinuria	Methionine	518	11	Homocystinuria	0
				Hypermethioninemia	0
Phenylketonuria	Phenylalanine	183	21	Phenylketonuria (PKU)	16
				Hyperphenylalaninemia	3
Tyrosinemia Type I	Succinylacetone	0	0	Tyrosinemia Type 1	0
Tyrosinemia Type II, III	Tyrosine	256	11	Tyrosinemia Type 2	0
				Tyrosinemia Type 3	0
Fatty Acid Oxidation Disorders					
Carnitine uptake defect	Free Carnitine (C0), Total Acylcarnitines (SUM AC)	115	22	Carnitine uptake defect (CUD)	5
Carnitine palmitoyltransferase 1 deficiency	C0/(C16 + C18)	54	2	Carnitine palmitoyltransferase 1 (CPT1) deficiency	0
Carnitine palmitoyltransferase 2 deficiency/Carnitine/Acylcarnitine translocase deficiency	Hexadecanoylcarnitine (C16), Octadecenoylcarnitine (C18:1)	0	11	Carnitine palmitoyltransferase 2 (CPT2) deficiency	2
2,4-Dienoyl-CoA reductase deficiency	Decadienoylcarnitine (C10:2)	42	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)	2	5	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)	229	15	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	7
				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	9	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	3

Screened Disorders	Analytes	Borderline	Referrals	# Confirmed with Disease	
Short-chain acyl-CoA dehydrogenase deficiency	Butyrylcarnitine (C4)	172	23	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	8
				Isobutyryl-CoA dehydrogenase (IBCD) deficiency	1
Medium/short-chain hydroxyl CoA dehydrogenase deficiency	Hydroxybutyrylcarnitine (C4OH), Hydroxyhexanoylcarnitine (C6OH)	77	1	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)	21	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)	72	6	Glutaryl-CoA dehydrogenase deficiency - glutaric aciduria (GA-I)	1
Isovaleryl CoA dehydrogenase deficiency/2-methylbutyryl-CoA dehydrogenase deficiency	Isovalerylcarnitine (C5)	376	14	Isovaleryl CoA dehydrogenase deficiency - isovaleric acidemia (IVA)	0
				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)	70	63	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	Malonylcarnitine (C3DC)	4	0	Malonyl-CoA decarboxylase deficiency - Malonic Aciduria (MA)	0
Propionyl-CoA carboxylase deficiency/Methylmalonyl-CoA mutase deficiency	Propionylcarnitine (C3), Methylmalonylcarnitine (C4DC)	307	36	Propionyl-CoA carboxylase deficiency (PA)	1
				Methylmalonyl-CoA mutase deficiency (MMA)	1
				Cobalamin A/B deficiency	0
				Cobalamin C/D/F deficiency	2
				Multiple Carboxylase deficiency	0
Urea Cycle Disorders					
Argininosuccinic aciduria/Citrullinemia	Citrulline	64	10	Argininosuccinic aciduria	0
				Citrullinemia	1
Argininemia	Arginine	26	1	Argininemia	1
Lysosomal Storage Disorders					
Krabbe Disease	Galactocerebrosidase	0	19	Krabbe disease possible late onset *	1
Mucopolysaccharidosis Type I	alpha-L-iduronidase	0	4	MPS 1	0
Pompe Disease	Alpha-glucosidase	0	18	Infantile-onset Pompe Disease	0
				Possible late-onset Pompe disease	10
Other Genetic Conditions					
Adrenoleukodystrophy	C26:0 Lysophosphatidylcholine (C26:0 LPC)	15	11	Male with X-linked Adrenoleukodystrophy (X-ALD)	2
				Female carrier of X-ALD	4
				Zellweger Syndrome	2
				Other Peroxisomal Biogenesis Disorder	2
Biotinidase Deficiency	Biotinidase	6	7	Biotinidase Deficiency	6
Cystic Fibrosis (CF)	Immunoreactive Trypsinogen	N/A	125	Cystic Fibrosis	21
Spinal Muscular Atrophy (SMA)	SMN1 gene, exon 7 deletion	N/A	5	Spinal Muscular Atrophy	5
Guanidinoacetate methyltransferase deficiency (GAM	Guanidinoacetate	30	4	Guanidinoacetate methyltransferase deficiency	0
Galactosemia	Galactose Transferase	17	7	Galactosemia	2
Severe Combined Immunodeficiency (SCID)	T-cell receptor excision circles (TRECS)	625	137	Classic SCID	3
				Leaky SCID	0
				Variant SCID	0
Total		9201	1775		627

Data based on infants born in 2022 whose specimens were received before 4/19/23.

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