Newborn Screening Program - 2020 Annual Report
Jan 1-Dec 31 2020
New York State Department of Health
Wadsworth Center
Biggs Laboratory
Albany, NY

Specimens Received	
Initial Valid	201,566
Initial Invalid	7,141
Total Newborns	208,707
Repeat Specimens	38,229
Total Specimens	246,936

Screened Disorders	Analytes	Referrals	# Confirmed with Disease	
Endocrine Disorders				
Congenital Adrenal Hyperplasia	17-hydroxyprogesterone	99	Congenital adrenal hyperplasia - 21-Hydroxylase deficiency	7
			Congenital adrenal hyperplasia, other enzyme deficiency	1
Congenital Hypothyroidism	Thyroxine, TSH	496	Primary Congenital Hypothyroidism	87
			Secondary Congenital Hypothyroidism	0
			Other	219
Hemoglobin Disorders				
Hemoglobin Disorders	Hemoglobin SS	112	Hemoglobin S + S (sickle cell) disease	71
	Hemoglobin SC	62	Hemoglobin S + C disease	44
	Hemoglobin CC	20	Hemoglobin C + C disease	13
	Other Hemoglobins	46	Other Hemoglobinopathies	15
Infectious Disease				
HIV	HIV Antibodies	334	Confirmed by diagnosis developed by the AIDS Institute	
Amino Acid Disorders				
Maple Syrup Urine Disease	Leucine	9	Maple Syrup Urine disease	1
			Hydroxyprolinemia	0
Homocystinuria	Methionine	4	Homocystinuria	0
			Hypermethioninemia	0
Phenylketonuria	Phenylalanine	10	Phenylketonuria (PKU)	5
			Hyperphenylalaninemia	2
Tyrosinemia Type I	Succinylacetone	2	Tyrosinemia Type 1	2
Tyrosinemia Type II, III	Tyrosine	2	Tyrosinemia Type 2	0
			Tyrosinemia Type 3	0
Fatty Acid Oxidation Disorders				
Carnitine uptake defect	Free Carnitine (C0), Total Acylcarnitines (SUM AC)	27	Carnitine uptake defect (CUD)	3
Carnitine palmitoyltransferase 1 deficiency	C0/(C16 + C18)	2	Carnitine palmitoyltransferase 1 (CPT1) deficiency	0
Carnitine palmitoyltransferase 2 deficiency/Carnitine/Acylcarnitine translocase deficiency	Hexadecanoylcarnitine (C16), Octadecenoylcarnitine (C18:1)	18	Carnitine palmitoyltransferase 2 (CPT2) deficiency	0
2,4-Dienoyl-CoA reductase deficiency	Decadienoylcarnitine (C10:2)	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)	3	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	0
			Trifunctional protein (TFP) deficiency	1
Multiple acyl-CoA dehydrogenase deficiency/ Medium-chain acyl-CoA dehydrogenase deficiency/Medium-chain 3-keto acyl-CoA thiolase deficiency	Hexanoylcarnitine (C6), Octanoylcarnitine (C8)	10	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	3
			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	Tetradecanoylcarnitine (C14), Tetradecenoylcarnitine (C14:1)	13	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	3

Screened Disorders	Analytes	Referrals	# Confirmed with Disease	
Short-chain acyl-CoA dehydrogenase deficiency	Butyrylcarnitine (C4)	37	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	17
			Isobutyryl-CoA dehydrogenase (IBCD) deficiency	3
Medium/short-chain hydroxyl CoA dehydrogenase deficiency	Hydroxybutyrylcarnitine (C4OH), Hydroxyhexanoylcarnitine (C6OH)	0	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)	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)	4	Glutaryl-CoA dehydrogenase deficiency - glutaric aciduria (GA-I)	2
Isovaleryl CoA dehydrogenase deficiency/2-methylbutyryl-CoA dehydrogenase deficiency	Isovalerylcarnitine (C5)	5	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	0
3-Methylcrotonyl-CoA carboxylase deficiency/2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency/3-Methylglutaconic aciduria	Hydroxyisovalerylcarnitine (C5OH)	36	3-Methylcrotonyl-CoA carboxylase (3MCC) deficiency	5
			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)	1	Malonyl-CoA decarboxylase deficiency - Malonic Aciduria (MA)	1
Propionyl-CoA carboxylase deficiency/Methylmalonyl-CoA mutase deficiency	Propionylcarnitine (C3), Methylmalonylcarnitine (C4DC)	33	Propionyl-CoA carboxylase deficiency (PA)	1
			Methylmalonyl-CoA mutase deficiency (MMA)	1
			Cobalamin A/B deficiency	0
			Cobalamin C/D/F deficiency	1
			Multiple Carboxylase deficiency	0
Urea Cycle Disorders				
Argininosuccinic aciduria/Citrullinemia	Citrulline	6	Argininosuccinic aciduria	0
			Citrullinemia	0
Argininemia	Arginine	0	Argininemia	1
Lysosomal Storage Disorders				
Krabbe Disease	Galactocerebrosidase	16	Krabbe disease possible late onset *	2
Mucopolysaccharidosis Type I	alpha-L-iduronidase	6	MPS 1	0
Pompe Disease	Alpha-glucosidase	17	Infantile-onset Pompe Disease	1
			Possible late-onset Pompe disease	7
Other Genetic Conditions				
Adrenoleukodystrophy	C26:0 Lysophosphatidylcholine (C26:0 LPC)	17	Male with X-linked Adrenoleukodystrophy (X-ALD)	5
			Female carrier of X-ALD	4
			Zellweger Syndrome	0
			Other Peroxisomal Biogenesis Disorder	0
Biotinidase Deficiency	Biotinidase	2	Biotinidase Deficiency	2
Cystic Fibrosis	Immunoreactive Trypsin	106	Cystic Fibrosis	28
Spinal Muscular Atrophy	SMN1 gene, exon 7 deletion	15	Spinal Muscular Atrophy	15
GAMT	Guanidinoacetate	4	Guanidinoacetate methyltransferase deficiency	0
Galactosemia	Galactose Transferase	5	Galactosemia	3
Severe Combined Immunodeficiency (SCID)	T-cell receptor excision circles (TRECS)	153	Classic SCID	3
			Leaky SCID	0
			Variant SCID	1
Total		1732		581

Data based on specimens received before 4/19/21.

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