Newborn Screening Program - 2021 Annual Report
Jan 1-Dec 31 2021
New York State Department of Health
Wadsworth Center
Biggs Laboratory
Albany, NY

Specimens Received	
Initial Valid	191,788
Initial Invalid	19,481
Total Newborns	211,269
Repeat Specimens	39,993
Total Specimens	251,262

Screened Disorders	Analytes	Referrals	# Confirmed with Disease	
Endocrine Disorders				
Congenital Adrenal Hyperplasia	17-hydroxyprogesterone	119	Congenital adrenal hyperplasia - 21-Hydroxylase deficiency	16
			Congenital adrenal hyperplasia, other enzyme deficiency	0
Congenital Hypothyroidism	TSH	697	Primary Congenital Hypothyroidism	133
			Secondary Congenital Hypothyroidism	0
			Other	235
Hemoglobin Disorders				
Hemoglobin Disorders	Hemoglobin SS	118	Hemoglobin S + S (sickle cell) disease	105
	Hemoglobin SC	60	Hemoglobin S + C disease	58
	Hemoglobin CC	18	Hemoglobin C + C disease	12
	Other Hemoglobins	35	Other Hemoglobinopathies	23
Infectious Disease				
HIV	HIV Antibodies	296	Confirmed by diagnosis developed by the AIDS Institute	
Amino Acid Disorders				
Maple Syrup Urine Disease	Leucine	6	Maple Syrup Urine disease	0
			Hydroxyprolinemia	0
Homocystinuria	Methionine	4	Homocystinuria	0
			Hypermethioninemia	0
Phenylketonuria	Phenylalanine	35	Phenylketonuria (PKU)	17
			Hyperphenylalaninemia	8
Tyrosinemia Type I	Succinylacetone	3	Tyrosinemia Type 1	3
Tyrosinemia Type II, III	Tyrosine	5	Tyrosinemia Type 2	0
			Tyrosinemia Type 3	0
Fatty Acid Oxidation Disorders				
Carnitine uptake defect	Free Carnitine (C0), Total Acylcarnitines (SUM AC)	18	Carnitine uptake defect (CUD)	3
Carnitine palmitoyltransferase 1 deficiency	C0/(C16 + C18)	4	Carnitine palmitoyltransferase 1 (CPT1) deficiency	0
Carnitine palmitoyltransferase 2 deficiency/Carnitine/Acylcarnitine translocase deficiency	Hexadecanoylcarnitine (C16), Octadecenoylcarnitine (C18:1)	12	Carnitine palmitoyltransferase 2 (CPT2) deficiency	2
2,4-Dienoyl-CoA reductase deficiency	Decadienoylcarnitine (C10:2)	1	2,4-Dienoyl-CoA (2,4Di) reductase deficiency	1
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)	15	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	4
			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)	12	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	3

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Data based on specimens received before 4/4/22.

Screened Disorders	Analytes	Referrals	# Confirmed with Disease	
Short-chain acyl-CoA dehydrogenase deficiency	Butyrylcarnitine (C4)	19	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	5
			Isobutyryl-CoA dehydrogenase (IBCD) deficiency	0
Medium/short-chain hydroxyl CoA dehydrogenase deficiency	Hydroxybutyrylcarnitine (C4OH), Hydroxyhexanoylcarnitine (C6OH)	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)	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)	7	Glutaryl-CoA dehydrogenase deficiency - glutaric aciduria (GA-I)	3
Isovaleryl CoA dehydrogenase deficiency/2-methylbutyryl-CoA dehydrogenase deficiency	Isovalerylcarnitine (C5)	6	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)	38	3-Methylcrotonyl-CoA carboxylase (3MCC) deficiency	6
			3-Hydroxy-3-methylglutaryl-CoA lyase (HMG) deficiency	0
			2-Methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD) deficiency - 2-Methyl-3-hydroxybutric acidemia (2M3HB)	0
			3-Methylglutaconic aciduria (3MGA)	0
Malonyl-CoA decarboxylase deficiency	Malonylcarnitine (C3DC)	2	Malonyl-CoA decarboxylase deficiency - Malonic Aciduria (MA)	1
Propionyl-CoA carboxylase deficiency/Methylmalonyl-CoA mutase deficiency	Propionylcarnitine (C3), Methylmalonylcarnitine (C4DC)	38	Propionyl-CoA carboxylase deficiency (PA)	1
			Methylmalonyl-CoA mutase deficiency (MMA)	0
			Cobalamin A/B deficiency	1
			Cobalamin C/D/F deficiency	1
			Multiple Carboxylase deficiency	0
Urea Cycle Disorders				
Argininosuccinic aciduria/Citrullinemia	Citrulline	5	Argininosuccinic aciduria	0
			Citrullinemia	0
Argininemia	Arginine	1	Argininemia	1
Lysosomal Storage Disorders				
Krabbe Disease	Galactocerebrosidase	11	Krabbe disease possible late onset *	2
Mucopolysaccharidosis Type I	alpha-L-iduronidase	13	MPS 1	0
Pompe Disease	Alpha-glucosidase	18	Infantile-onset Pompe Disease	1
			Possible late-onset Pompe disease	8
Other Genetic Conditions				
Adrenoleukodystrophy	C26:0 Lysophosphatidylcholine (C26:0 LPC)	14	Male with X-linked Adrenoleukodystrophy (X-ALD)	4
			Female carrier of X-ALD	1
			Zellweger Syndrome	5
			Other Peroxisomal Biogenesis Disorder	0
Biotinidase Deficiency	Biotinidase	4	Biotinidase Deficiency	2
Cystic Fibrosis	Immunoreactive Trypsin	108	Cystic Fibrosis	19
Spinal Muscular Atrophy	SMN1 gene, exon 7 deletion	11	Spinal Muscular Atrophy	11
GAMT	Guanidinoacetate	3	Guanidinoacetate methyltransferase deficiency	1
Galactosemia	Galactose Transferase	7	Galactosemia	4
Severe Combined Immunodeficiency (SCID)	T-cell receptor excision circles (TRECS)	141	Classic SCID	3
			Leaky SCID	0
			Variant SCID	0

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