



Specimens Received	NYS ¹	Upstate	NYC
Initial Valid	227,775	112,076	111,550
Initial Invalid	11,243	4,973	5,987
Total Newborns	239,018	117,049	117,537
Repeat Specimens	28,916	14,571	13,606
Total Specimens	267,934	131,620	131,143

Screening Results	Actionable/Screen Positive						Molecular Results						Confirmed Cases		
	NYS ¹		Upstate		NYC		NYS		Upstate		NYC		NYS	Upstate	NYC
	Act.	Scr Pos	Act.	Scr Pos	Act.	Scr Pos	1Mut ²	2+ Mut	1Mut	2+ Mut	1Mut	2+ Mut			
Amino Acid Disorders															
HCY • HMet	5	647	1	369	4	256							0	0	0
MSUD	5	423	3	265	2	147							1	0	1
PKU • HyperPhe	21	144	14	84	7	56							11	6	5
TYR-I	0	0	0	0	0	0							0	0	0
TYR-II, III	5	121	2	59	3	56							0	0	0
Endocrine Disorders															
CAH	290	2015	122	813	158	1,152							15	11	4
CH	624	3,215	252	1,471	357	1,658									
Primary													85	39	45
Secondary/Tertiary													5	3	2
TBG													24	11	13
Other													171	73	97
Fatty Acid Oxidation Disorders															
CUD	11	722	3	362	8	349							4	2	2
CPT-I	2	35	2	18	0	16							0	0	0
CAT • CPT-II	20	7	11	4	9	3							2	0	2
2,4 Di	0	33	0	11	0	22							0	0	0
LCHAD • TFP	5	0	5	0	0	0							0	0	0
MADD (GA-II) • MCAD • MCKAT	20	167	10	86	10	76	5	3	3	2	2	1	9	7	2
VLCAD	6	0	3	0	3	0							2	1	1
SCAD ³	21	71	10	27	11	39							11	3	8
M/SCHAD	1	46	0	28	1	18							0	0	0

1. NYS contains all specimens received by NYS, including those from out-of-state and whose residence is unknown. Specimen totals may not equal the sum of the Upstate and NYC columns.

2. Mutation

3. SCAD and IBCD are screened using identical markers. Screen positive totals are counted only once.

Screening Results	Actionable/Screen Positive						Molecular Results						Confirmed Cases		
	NYS ¹		Upstate		NYC		NYS		Upstate		NYC		NYS	Upstate	NYC
Hemoglobin Disorders	Act.	Scr Pos	Act.	Scr Pos	Act.	Scr Pos	1Mut ²	2+ Mut	1Mut	2+ Mut	1Mut	2+ Mut			
Diseases:															
S/S	145		42		103								132	41	91
S/C	65		13		52								66	17	49
C/C	20		8		12								19	8	11
β Thalassemia	17		7		10								4	2	2
H	10		4		5								3	2	0
Other	23		5		18								40	13	27
Traits:															
A/S		5,296		1,698		3,538									
A/C		1,661		521		1,128									
A/E		305		99		202									
A/D		97		37		60									
A/Other		179		71		106									
Transfusion		306		194		93									
Infectious Diseases													Confirmed by Diagnosis Developed by the AIDS Institute		
HIV-I	435	0	107	0	323	0									
Organic Acid Disorders															
BKT • MHBBD	0	3	0	1	0	1							0	0	0
GA-I	1	24	0	13	1	11							1	1	0
IBCD ³	21	71	10	27	11	39							11	3	8
IVA • 2-MBCD	6	240	5	155	1	82							3	2	1
HMG • 3-MCC • 3-MGA	22	36	12	23	10	13							11	7	4
MA	1	6	0	3	1	3							1	0	1
MCD • MUT • Cbl A,B • Cbl C,D • MMA • PA	58	383	27	180	31	194							8	4	4
Other Genetic Conditions															
BIOT	2	25	1	11	1	14							1	1	0
CF	868	0	475	0	370	0	616	32	371	28	237	3	30	26	4
SCID	109	532	55	258	50	263									
Classic SCID													3	1	2
Leaky SCID													0	0	0
Variant SCID													2	2	0
Non SCID													18	12	6
GALT	3	0	3	0	0	0	0	1	0	1	0	0	2	1	1
Krabbe ⁴	44	0	31	0	13	0	36	6	26	4	10	2	1	0	1
Pompe ⁵	9	0	3	0	6	0	4	4	1	2	3	2	1	0	1
ALD	19	23	12	12	7	10							8	7	1
Urea Cycle Disorders															
ASA • CIT	0	20	0	11	0	9							1	0	1
ARG	1	238	1	133	0	96							0	0	0
TOTALS	2894	17020	1249	7017	1587	9671	661	46	401	37	252	8	695	303	389

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2. Mutation

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

5. Pompe testing began on October 1, 2014