



Specimens Received	NYS ¹	Upstate	NYC
Initial Valid	228478	112576	111419
Initial Invalid	9934	4092	5567
Total Newborns	238412	116668	116986
Repeat Specimens	26706	12986	12942
Total Specimens	265118	129654	129928

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	3	589	1	373	2	193							0	0	0
MSUD	6	402	2	266	3	124							3	0	3
PKU • HyperPhe	33	144	19	92	12	45							23	15	7
TYR-I	2	1	1	0	1	1							2	1	1
TYR-II, III	3	134	2	70	1	59							1	1	0
Endocrine Disorders															
CAH	243	1445	96	597	134	814							11	4	6
CH	580	2885	252	1321	316	1474									
Primary													90	46	43
Secondary/Tertiary													7	4	2
TBG													32	22	9
Other													173	80	85
Fatty Acid Oxidation Disorders															
CUD	10	885	5	436	5	425							0	0	0
CPT-I	0	34	0	20	0	13							0	0	0
CAT • CPT-II	34	6	12	2	21	3							3	0	3
2,4 Di	0	51	0	23	0	25							0	0	0
LCHAD • TFP	2	1	2	0	0	1							0	0	0
MADD (GA-II) • MCAD • MCKAT	21	137	13	68	7	63	8	6	7	4	1	2	14	11	3
VLCAD	5	0	5	0	0	0							1	1	0
SCAD ³	10	92	5	47	5	42							0	0	0
M/SCHAD	0	16	0	11	0	5							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
	Act.	Scr Pos	Act.	Scr Pos	Act.	Scr Pos	1Mut ²	2+ Mut	1Mut	2+ Mut	1Mut	2+ Mut			
Hemoglobin Disorders															
Diseases:															
S/S	114		29		85								113	29	84
S/C	67		22		45								70	25	45
C/C	25		6		19								22	5	17
β Thalassemia	5		0		5								6	1	5
H	15		1		13								16	1	14
Other	26		11		34								31	13	18
Traits:															
A/S		5288		1656		3588									
A/C		1542		471		1050									
Alpha Thalassemia		2556		838		1664									
A/Other		1891		850		955									
Transfusion		1888		849		953									
Infectious Diseases													Confirmed by Diagnosis Developed by the AIDS Institute		
HIV-I	467		110		350										
Organic Acid Disorders															
BKT • MHBD	0	5	0	5	0	0							0	0	0
GA-I	7	51	6	30	1	18							0	0	0
IBCD ³	10	92	5	47	5	42							6	2	4
IVA • 2-MBCD	6	279	4	168	2	98							0	0	0
HMG • 3-MCC • 3-MGA	29	41	14	25	13	16							13	4	8
MA	2	2	1	0	1	2							2	1	1
MCD • MUT • Cbl A,B • Cbl C,D • MMA • PA	63	303	32	135	31	161							6	0	6
Other Genetic Conditions															
BIOT	2	10	1	7	1	3							1	1	0
CF	893	0	487	0	383	0	631	29	360	24	253	5	34	26	9
SCID	233	874	84	397	144	462									
Classic SCID													8	4	4
Leaky SCID													1	0	1
Variant SCID													0	0	0
Non SCID													26	14	11
GALT	9	0	3	0	5	0	2	1	0	1	2	0	2	1	1
Krabbe ⁵	44	0	27	0	15	0	36	7	24	3	12	4	2	1	1
ALD ⁴	1	1	1	0	0	1	0	0	0	0	0	0	0	0	0
Urea Cycle Disorders															
ASA • CIT	5	25	3	19	2	6							4	2	2
ARG	2	274	2	159	0	101							0	0	0
TOTALS	2967	21852	1259	8935	1656	12365	677	43	391	32	268	11	723	315	393

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2. Mutation

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

4. ALD Testing began December 30th, 2013

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