



Specimens Received	NYS ¹	Upstate	NYC
Initial Valid	231773	112842	114015
Initial Invalid	9848	4333	5226
Total Newborns	241621	117175	119241
Repeat Specimens	26660	13303	12482
Total Specimens	268282	130478	131723

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	7	575	4	348	3	205							0	0	0
MSUD	6	441	4	281	2	145							0	0	0
PKU • HyperPhe	25	161	16	103	9	50							18	13	5
TYR-I ³	0	0	0	0	0	0							0	0	0
TYR-II, III	11	123	6	60	5	58							0	0	0
Endocrine Disorders															
CAH	191	1306	97	517	92	753							17	7	8
CH	672	3463	318	1587	329	1755									
Primary													100	43	53
Secondary/Tertiary							7	5	2						
TBG							55	31	24						
Other							178	76	100						
Fatty Acid Oxidation Disorders															
CUD	12	783	4	354	8	398							2	1	1
CPT-I	0	51	0	37	0	11							0	0	0
CAT • CPT-II	14	3	4	3	10								2	0	2
2,4 Di	0	80	0	54	0	23							0	0	0
LCHAD • TFP	3	1	1	1	1								2	0	1
MADD (GA-II) • MCAD •	18	173	14	87	4	77	7	3	6	3	1	0	13	9	4
MCKAT															
VLCAD	21	0	16	0	5	0							3	1	2
SCAD ³	17	94	7	66	10	27							9	2	7
M/SCHAD	0	16	0	13	0	3							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.

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

5. Screening for HHH ended Oct 1st, 2012

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	135		31		104								146	35	110
S/C	74		22		52								70	16	54
C/C	19		5		14								21	5	16
β Thalassemia	7		5		2								5	4	1
H	9		1		8								8	2	6
Other	24		7		16								35	14	20
Traits:															
A/S		5478		1746		3668									
A/C		1602		482		1110									
Alpha Thalassemia		5		0		5									
A/Other		566		201		353									
Transfusion		2163		983		1071									
Infectious Diseases													Confirmed by Diagnosis Developed by the AIDS Institute		
HIV-I	452		93		352										
Organic Acid Disorders															
BKT • MHBD	1	12	0	8	1	4							1	0	1
GA-I	0	24	0	18	0	6							0	0	0
IBCD ³	17	94	7	66	10	27							2	0	2
IVA • 2-MBCD	3	352	3	236	0	105							0	0	0
HMG • 3-MCC • 3-MGA	39	37	13	20	26	17							21	5	16
MA	0	4	0	4	0	0							0	0	0
MCD • MUT • Cbl A,B • Cbl C,D • MMA • PA	44	249	16	116	25	128							10	2	6
Other Genetic Conditions															
BIOT	2	17	1	10	1	7							1	1	0
CF	898		529		349		653	30	401	25	237	5	35	30	5
SCID	249	866	108	404	135	441									
Classic SCID													6	4	2
Leaky SCID													0	0	0
Variant SCID													0	0	0
Non SCID													21	10	8
GALT	18		14		4		6	3	5	3	1	0	12	10	2
Krabbe	41		23		11		30	4	21	3	9	1	1	0	1
Urea Cycle Disorders															
ASA • CIT	10	28	6	15	4	11							2	1	1
ARG	2	191	0	117	2	64							2	0	2
HHH ⁵	0	159	0	81	0	71							0	0	0
TOTALS	3024	19023	1368	7952	1584	10566	696	40	433	34	248	6	796	325	455

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