

Newborn Screening Program - 2007 Annual Report
 Jan 1-Dec 31 2007
 New York State Department of Health
 Wadsworth Center
 Biggs Laboratory
 Albany, NY



Specimens Received	NYS ¹	Upstate	NYC
Initial Valid	246579	122529	118981
Initial Invalid	8696	4045	4375
Total Newborns	255275	126574	123356
Repeat Specimens	25946	13337	11912
Total Specimens	281221	139911	135268

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	24	377	11	228	13	137							1	1	0
MSUD	9	208	4	148	5	50							1	0	1
PKU • HyperPhe	34	288	21	167	12	106							13	9	4
TYR-I ³	0	0	0	0	0	0							0	0	0
TYR-II, III	10	132	4	64	6	62							3	2	1
Endocrine Disorders															
CAH	380	1167	125	437	246	698							12	3	9
CH	1098	4330	451	1947	614	2234									
Primary													200	103	88
Secondary/Tertiary													3	1	2
TBG													40	26	12
Other													17	7	9
Fatty Acid Oxidation Disorders															
CUD	50	544	18	245	31	276							9	2	6
CPT-I	11	36	8	26	2	9							0	0	0
CAT • CPT-II	3	62	2	23	1	38							1	0	1
2,4 Di	0	28	0	16	0	12							0	0	0
LCHAD • TFP	0	5	0	0	0	4							0	0	0
MADD (GA-II) • MCAD •	27	218	12	114	14	99	2	2	0	2	2	0	12	9	2
MCKAT															
VLCAD	11	1	7	1	3	0							3	1	2
SCAD ⁴	23	101	8	50	13	50							11	6	5
M/SCHAD	4	35	2	27	2	8							1	0	1

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. TYR-I testing began December 5th, 2007

4. 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	125		28		96								124	22	101
S/C	88		15		73								77	17	59
C/C	23		8		14								21	9	11
Other	37		7		30								22	4	18
Traits:															
A/S		5554		1650		3842									
A/C		1723		460		1246									
A/Other		567		205		355									
Transfusion		2794		1258		1425									
Infectious Diseases													Confirmed by Diagnosis Developed by the AIDS Institute		
HIV-I	630		144		482										
Organic Acid Disorders															
BKT • MHBD	1	10	0	6	1	4							1	0	1
GA-I	7	32	5	24	1	8							1	0	1
IBCD ⁴	23	101	8	50	13	50							11	6	5
IVA • 2-MBCD	10	82	4	42	6	38							3	3	0
HMG • 3-MCC • 3-MGA	83	131	38	65	43	64							37	21	16
MA	0	3	0	3	0	0							0	0	0
MCD • MUT • Cbl A,B • Cbl C,D • MMA • PA	47	243	22	110	23	131							9	3	5
Other Genetic Conditions															
BIOT	2	4	0	1	2	3							5	2	3
CF	1651		828		780		668	30	409	24	248	6	40	27	9
GALT	12		5		7		3	2	1	0	2	2	5	2	3
Krabbe	46		25		20		32	11	20	5	11	6	4	2	2
Urea Cycle Disorders															
ASA • CIT	8	96	5	63	2	32							1	0	0
ARG	1	105	1	73	0	31							1	1	0
HHH	12	259	5	147	7	100							0	0	0
TOTALS	4467	19135	1813	7600	2549	11062	705	45	430	31	263	14	678	283	372

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