

Newborn Screening Program  
 Annual Report  
 Jan 1-Dec 31 2006  
 New York State Department of Health  
 Wadsworth Center  
 Biggs Laboratory  
 Albany, NY

Specimens Received	NYS	Upstate	NYC
Initial Valid	246,140	123,815	122,325
Initial Invalid	5,874	2,433	3,441
Total Newborns	252,014	126,248	125,766
Repeat Specimens	21,107	11,251	9,856
Total Specimens	273,121	137,499	135,622

Screening Results	Actionable/Screen Positive <sup>3</sup>						Molecular Results						Confirmed Cases		
	NYS		Upstate		NYC		NYS		Upstate		NYC		NYS	Upstate	NYC
	Act.	Scr Pos	Act.	Scr Pos	Act.	Scr Pos	1Mut*	2Mut	1Mut	2Mut	1Mut	2Mut			
<b>Amino Acid Disorders</b>															
HCY•HMet	15	369	7	231	8	138							0	0	0
MSUD	9	223	5	152	4	71							3	3	0
PKU • HyperPhe	41	282	24	165	17	117							14	9	5
TYR	13	131	6	71	7	60							2	0	2
<b>Endocrine Disorders</b>															
CAH	629	950	232	352	397	598							19	10	9
CH	1,018	3,474	493	1,612	525	1,862									
Primary													182	92	90
Secondary/Tertiary													1	1	0
TBG													50	37	13
Other													6	4	2
<b>Fatty Acid Oxidation Disorders</b>															
CUD	31	584	14	267	17	317							10	7	3
CPT-I	22	89	14	62	8	27							0	0	0
CAT•CPT-II	3	44	2	18	1	26							0	0	0
2,4 Di	0	29	0	17	0	12							0	0	0
LCHAD•TFP	1	1	0	1	1	0							2	1	1
MADD•MCAD•MCKAT	34	241	18	149	16	92	5	3	3	3	2	0	8	6	2
SCAD <sup>1</sup>	13	116	7	63	6	53							11	4	7
VLCAD	7	5	2	1	5	4							2	1	1
M/SCHAD	2	24	1	13	1	11							0	0	0

\* Mutation    1 SCAD and IBCD are screened using identical markers.    3 Actionable - Specimens referred immediately for diagnosis/treatment  
 See SCAD for IBCD screen positive and confirmed data    Screen Positive - Specimens for which a repeat was requested  
 2 Krabbe testing began August 7th, 2006

	Actionable/Screen Positive						Molecular Results						Confirmed Cases		
Screening Results	NYS		Upstate		NYC		NYS		Upstate		NYC		NYS	Upstate	NYC
<b>Hemoglobin Disorders</b>	Act.	Scr Pos	Act.	Scr Pos	Act.	Scr Pos	1Mut	2Mut	1Mut	2Mut	1Mut	2Mut			
Diseases: S/S	138		23		115								130	24	106
S/C	82		14		68								76	11	65
C/C	26		5		21								25	4	21
Other	25		8		17								17	3	14
Traits: A/S		5,924		1,818		4,106									
A/C		1,718		470		1,248									
A/Other		557		227		330									
Transfusion		2,821		1,360		1,461									
<b>Infectious Diseases</b>															
HIV-I	606		117		489								Confirmed by Diagnosis Developed by the AIDS Institute		
<b>Organic Acid Disorders</b>															
BKT•MHBD	0	19	0	10	0	19							0	0	0
GA-I	18	33	12	30	6	3							5	4	1
IBCD <sup>1</sup>	**	**	**	**	**	**							11	4	7
IVA•2-MBCD	29	142	11	68	18	74							3	1	2
HMG•3-MCC•3-MGA	119	189	58	84	61	105							31	15	16
MA	0	2	0	0	0	2							0	0	0
MCD•MUT•Cbl A,B•Cbl C,D•MMA•PA	70	305	25	146	45	159							10	4	6
<b>Other Genetic Conditions</b>															
BIOT	4	6	1	1	3	5							2	0	2
CF	1,565		781		784		699	23	437	18	262	5	36	26	10
GALT	16		7		9		4	2	4	0	0	2	5	1	4
Krabbe	7		4		3		6	0	4	0	2	0	0	0	0
<b>Urea Cycle Disorders</b>															
ASA•CIT	6	93	2	60	4	33							5	3	2
ARG	2	102	1	69	1	33							0	0	0
HHH	4	114	4	68	0	46							0	0	0
<b>TOTALS</b>	4,670	18,587	1,925	7,585	2,745	11,012	714	28	448	21	266	7	655	271	384