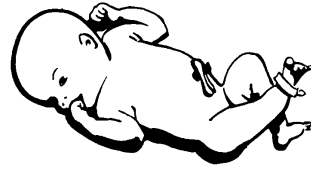


Newborn Screening Program
Annual Report
Jan 1-Dec 31 2004



New York State Department of Health
Wadsworth Center
Biggs Laboratory
Albany, New York

Specimens Received	New York State	Upstate	New York City
Initial Valid	247,240	125,334	121,906
Initial Invalid	3,691	1,360	2,331
Total Newborns	250,931	126,694	124,237
Repeat Specimens	14,100	7,729	6,371
Total Specimens	265,031	134,423	130,608

Screening Results	Screen Positive			Molecular Results						Confirmed Cases		
	NYS	Upstate	NYC	NYS		Upstate		NYC		NYS	Upstate	NYC
				1 mut*	2 mut	1 mut	2 mut	1 mut	2 mut			
Amino Acid Disorders												
HCY	381	241	140							0	0	0
MSUD	177	132	45							0	0	0
PKU	296	175	121							6	2	4
TYR	27	16	11							0	0	0
Endocrine Disorders												
CAH	901	392	509							16	10	6
CH	4,514	2,021	2,493									
Primary										168	77	91
Secondary/Tertiary										1	0	1
TBG										41	17	24
Other										13	8	5
Fatty Acid Oxidation Disorders												
CUD	119	51	68							0	0	0
CAT•CPT-II	34	17	17							0	0	0
LCHAD•TFP	1	1	0							0	0	0
MADD•MCAD	178	109	69	65	4	55	4	10	0	17	13	4
SCAD	62	34	28							0	0	0
VLCAD	10	7	3							0	0	0
Hemoglobin Disorders												
Diseases: S/S	159	30	129							134	23	111
S/C	65	13	52							61	12	49
C/C	25	9	16							23	9	14
Other	18	6	12							20	10	10
Traits: A/S	5,538	1,598	3,940									
A/C	1,636	463	1,173									
A/Other	1,813	577	1,236									
Transfusion:	2,056	1,011	1,045									

* mutation

Screening Results	Screen Positive			Molecular Results						Confirmed Cases		
	NYS	Upstate	NYC	NYS		Upstate		NYC		NYS	Upstate	NYC
Infectious Diseases				1 mut*	2 mut	1 mut	2 mut	1 mut	2 mut			
HIV-1	781	163	618							Confirmed by Diagnosis Developed by the AIDS Institute		
Organic Acid Disorders												
BKT	4	3	1							0	0	0
GA-1	3	2	1							0	0	0
IBCD	62	34	28							0	0	0
IVA•2-MBCD	26	17	9							0	0	0
HMG•3-MCC	55	27	28							0	0	0
MCD•MUT•Cbl A,B	39	17	22							0	0	0
MMA•PA												
Other Genetic Conditions												
BIOT	6	3	3							3	2	1
CF	1,348	757	591	677	32	442	29	235	3	25	21	4
GALT	13	7	6							7	2	5
Urea Cycle Disorders												
ASA•CIT	13	7	6							0	0	0
TOTALS	20,360	7,940	12,420	742	36	497	33	245	3	535	206	329

* mutation