

OFFICE OF NEWBORN SCREENING 2008 CONFIRMED CASE REPORT
PRIMARY PANEL OF 28 BLOODSPOT DISORDERS

DISORDER	ANALYTE	PRESUMPTIVE POSITIVE	PRIMARY DISORDER	SECONDARY DISORDERS
Congenital Hypothyroidism	TSH	178	51	5-SECONDARY HYPOTHYROIDISM; 51-TRANSIENT; 9-THYROXINE BINDING
Congenital Adrenal Hyperplasia (21-hydroxylase deficiency)	17 OHP	943	5	1-NON-CLASSICAL; 2-21OH DEFICIENT
Sickle Cell Anemia			8	
Sickle Beta Thalassemia			0	1179 TRAITS; 3-OTHER HEMOGLOBIN DISEASE; 1 PRESUMED, NO FINAL DX
Sickle C Disease	Hgb	1192	1	
Biotinidase Deficiency	Biotinidase	60	1	
Galactosemia	GALT	73	2	
Phenylketonuria	Phenylalanine	64	2	6-HYPERPHENYLALANEMIA
Maple Syrup Urine Disease	Leucine	56	0	
Homocystinuria	Methionine	130	1	
Citrullinemia			1	
Argininosuccinic Acidemia	Citrulline	1	0	
Tyrosinemia Type 1	Tyrosine	52	0	
Carnitine Uptake Defect	C0	6	1	
Medium-chain Acyl-CoA Dehydrogenase Deficiency	C8	8	4	1-OTHER FATTY ACID OXIDATION; 1-CARRIER
Very Long-chain Acyl-CoA Dehydrogenase Deficiency	C14:1	1	0	
Long-chain 3-OH Acyl-CoA Dehydrogenase Deficiency			0	
Trifunctional Protein Deficiency	C16OH	0	0	
Isovaleric Acidemia	C5	10	0	
Glutaric Acidemia Type 1	C5DC	10	0	
Hydroxymethylglutaric CoA Lyase Deficiency			0	
3-Methylcrotonyl CoA Carboxylase Deficiency	C5OH	11	1	
Multiple Carboxylase Deficiency			0	
Methylmalonic Acidemia (mutase deficiency)			0	
Methylmalonic Acidemia (Cbl A,B)			0	
Propionic Acidemia	C3	6	0	2-C5OH OTHER
Beta-ketothiolase Deficiency	C5:1	0	0	
Cystic Fibrosis	IRT / DNA	176	14	
TOTALS		2977	92	

SPECIMEN DATA		
TOTAL OCCURRENT BIRTHS	100,088	PERCENTAGE
1ST SCREEN	96,963	97%
2ND SCREEN	87,503	87%
REQUESTED REPEAT	59	
DIETARY MONITORING	1,158	
1ST & 2ND UNSAT	2,297	1.2%
TOTAL SPECIMEN COUNT	187,980	