

INCIDENCE OF NEWBORN METABOLIC SCREENING DISORDERS IN HAWAII

Disorder	PKU	CH	CAH	MSUD	GAL	BIO	HB
Incidence U.S.	1/18,000	1/4,000	1/15,000	1/250,000	1/60,000	1/70,000	1/15,000
Incidence Hawaii (7/97-01/16) # screened= 338,542	1/56,650 5 classic 1 hyperphe	1/2,634 129 primary	1/22,660 8 salt-wasters 5 virilized 1 late onset	1/84,976 4 classic (2 intermediate)	1/169,952 2 classic (34 Duarte variants-D/G)	1/37,767 2 profound 9 partial	1/19,994 7 SS Disease 6 SC Disease 1 SE Disease 3 S/Beta thal

Key: PKU Phenylketonuria
 CH Congenital Hypothyroidism
 CAH Congenital Adrenal Hyperplasia
 MSUD Maple Syrup Urine Disease
 GAL Galactosemia
 BIO Biotinidase Deficiency
 HB Hemoglobinopathies

INCIDENCE OF TANDEM MASS NEWBORN SCREENING DISORDERS IN HAWAII

Disorder	MCADD	VLCADD	3MCC	MCD	?CPT I Variant	IVA	GA-1	SCADD
Incidence U.S.	1/15,000	1/120,000	1/50,000	1/87,000	?	1/230,000	1/40,000	Rare
Incidence Hawaii (9/03-01/16) # screened= 232,482	1/58,120 4 cases	1/33,211 7 cases (2 mild, 43 variants)	1/77,494 3 cases	1/116,241 2 cases	1/17,883 13 cases	1/38,747 6 cases (2 mild)	1/116,241 2 cases	1/232,482 1 case

Disorder	ASAL	MADD	Citrullinemia	CUD
Incidence U.S.	1/70,000	Rare	1/57,000	Rare
Incidence Hawaii (9/03-01/16) # screened= 232,482	1/116,241 2 cases	1/232,482 1 case	1/232,482 1 case	1/232,482 1 case

Key:

MCADD	Medium Chain Acyl-CoA Dehydrogenase Deficiency
VLCADD	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
3 MCC DEFICIENCY	3 Methylcrotonyl CoA Carboxylase Deficiency
MCD	Multiple Carboxylase Deficiency
CPT I Variant	Carnitine Palmitoyl Transferase I Variant
IVA	Isovaleric Acidemia
GA-1	Glutaric Acidemia Type 1
SCADD	Short Chain Acyl-Co-A Dehydrogenase Deficiency
ASAL	Argininosuccinate lyase Deficiency
MADD	Multiple Acyl-CoA Dehydrogenase Deficiency
CIT	Citrullinemia
CUD	Carnitine Uptake Defect

INCIDENCE OF CYSTIC FIBROSIS IN HAWAII

Disorder	Cystic Fibrosis
Incidence U.S.	1/4,000
Incidence Hawaii (9/07-01/16) # screened= 158,505	1/14,409 11 cases, including 2 cases meconium ileus (1 CFRMS) (1 mild)

CFRMS Cystic Fibrosis Related Metabolic Syndrome

INCIDENCE OF SCID IN HAWAII

Disorder	SCID
Incidence U.S.	1/100,000
Incidence Hawaii (3/15-01/16) # screened= 16,844	1/16,844 1 case

SCID Severe Combined Immunodeficiency Disease