

## INCIDENCE OF NEWBORN METABOLIC SCREENING DISORDERS IN HAWAII

Disorder	PKU	CH	CAH	MSUD	GAL	BIO	HB
<b>Incidence U.S.</b>	1/18,000	1/4,000	1/15,000	1/250,000	1/60,000	1/70,000	1/15,000
<b>Incidence Hawaii (7/97-12/12)</b>	1/47,116 5 classic 1 hyperphe	1/2,546 111 primary	1/25,699 5 salt-wasters 5 virilized 1 late onset	1/47,116 4 classic 2 intermediate	1/141,349 2 classic 26 Duarte variants	1/31,411 2 profound 7 partial	1/20,192 7 SS Disease 6 SC Disease 1 SE Disease

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
<b>Incidence U.S.</b>	1/15,000	1/120,000	1/50,000	1/87,000	?	1/230,000	1/40,000	Rare
<b>Incidence Hawaii (9/03-12/12)</b>	1/43,528 4 cases	1/29,018 6 cases 29 variants	1/58,037 3 cases	1/87,056 2 cases	1/19,345 9 cases	1/43,528 4 cases	1/87,056 2 cases	1/174,112 1 case

Disorder	ASAL	MADD	Citrullinemia
<b>Incidence U.S.</b>	1/70,000	Rare	1/57,000
<b>Incidence Hawaii (9/03-12/12)</b>	1/174,112 1 case	1/174,112 1 case	1/174,112 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

### INCIDENCE OF CYSTIC FIBROSIS IN HAWAII

Disorder	Cystic Fibrosis
<b>Incidence U.S.</b>	1/4,000
<b>Incidence Hawaii (9/07-12/12)</b>	1/19,959 3 cases, 1 case CFTR related metabolic syndrome, 1 case meconium ileus