

NBS CONFIRMED CASES FROM 7/1/2014-12/31/2020

Biotinidase	Profound=1; Partial=16; Compound heterozygous=6
2-Methylbutyrylglycinuria	1
3MCC/BKT/HMG	Carrier=1; Mild variant=1
Citrullinemia Type 1	Type 1=2; carriers=2; mild variants=2
Congenital Adrenal Hyperplasia	9; mild deficiency=1; salt-wasting=3
Congenital Hypothyroidism	89
CPT Type 2	1
CUD Primary carnitine deficiency	2
Cystic Fibrosis	31; heterozygous(one copy)=20; compound heterozygous=3
Classic Galactosemia	6; carrier=1
D/G Variant	25
GA1	1
Hemoglobinopathies	113; FS=50, FSC=27, FSA=11, FC=13, FCA=2, FD=1, FE=2, FSE=1, Fe/Beta Thal=2, Hydrops fet=1
Isobutyrylglycinuria(IBG)/Isobutyryl-CoA dehydrogenase (IBD) deficiency	2
MCAD	13; carrier=1; mild variant=2
MMA	2
Methionine Adenosyltransferase Deficiency (MAT)	1
PA	1 mild variant
PKU	10; Hyperlalaninemia=6
SCAD	7-all benign, no tx needed
SCID	7; Edward's Syndrome=1, DiGeorge=2, Jak3=1, Wiscott Aldrich Syndrome=1, T-cell Lymphopenia=2, Congenital hair Hypoplasia Syndrome=1
Tyrosinemia Type I	1
Tyrosinemia Type II	1; Transient=8, GALD=1
Tyrosinemia Type III	2
VLCAD	2; carriers=11