

Prevalence as of December 2019

Disorders	Screened	Confirmed	Prevalence
Endocrine Disorders			
CH	13,798,874	5079	1:2,716
CAH	13,798,874	689	1:20,027
Amino Acid Disorders			
PKU (Classic, Mild, PTPS Def, Hyperphe)	13,798,874	107	1:128,961
MSUD	9,761,087	148	1:65,953
OTHER AMINO ACIDS (1 MET, 5 TYR)	1,859,819	6	1: 309,969
Fatty Acid Disorders (13MCADD, 3 CUD, 8 VLCAD)	1,859,819	24	1:77,492
Organic Acid Disorders (2 MMA, 14 3MCC, 19 GA1, 1 PA, 1 MCD, 1 IVA)	1,859,819	38	1:48,942
Urea Cycle Defect (4 CIT, 2 ASA)	1,859,819	6	1: 309,950
GAL (Classical, Non-Classical)	13,798,874	143	1:96,495
G6PD Def	12,835,375	225,262	1:61
Hemoglobinopathies*	1,859,819	1192	1:1,560

*based on CE confirmatory