

Figure S1 CLIR 'Plot by Condition' chart showing disease ranges (1%, 10%, 50%, 90%, and 99%iles, respectively) in neonatal blood spots of patients with MSUD (N=275).

The reference population (corresponding to the 1-99% ile range; Xle N=2.45 million; Val N=2.31 million; AlloIle N=635; Val/Phe N=2.18 million; Xle/Phe N=2.39 million; Xle/Tyr N=2.26 million;

(Xle+Val)/(Phe+Tyr) N=1.92 million) is depicted in green. Abbreviations as follows: Allo-Ile: allo-isoleucine, CLIR: Collaborative Laboratory Integrated Reports, MSUD: Maple Syrup Urine Disease, Phe: phenylalanine, Tyr: tyrosine, Val: valine, Xle: isoleucine + leucine.