The role of exome sequencing in newborn screening for inborn errors of metabolism


1Department of Plant and Microbial Biology, University of California Berkeley, Berkeley, CA, USA. 2Institute for Human Genetics, University of California San Francisco, San Francisco, CA, USA. 3Department of Pediatrics, University of California San Francisco, San Francisco, CA, USA. 4Program in Bioethics, University of California San Francisco, San Francisco, CA, USA. 5Innovation Labs, Tata Consultancy Services, Hyderabad, India. 6Department of Biomedical Informatics and Medical Education, University of Washington, Seattle, WA, USA. 7Invitae, San Francisco, CA, USA. 8Department of Molecular, Cellular and Developmental Biology, Center for the Molecular Biology of RNA, UC Santa Cruz Genomics Institute, University of California Santa Cruz, Santa Cruz, CA, USA. 9Genetic Disease Screening Program, California Department of Public Health, Richmond, CA, USA. 10Cardiovascular Research Institute, University of California San Francisco, San Francisco, CA, USA. 11Department of Dermatology, University of California San Francisco, San Francisco, CA, USA. 12Division of Allergy, Immunology and Blood and Marrow Transplantation, UCSF Benioff Children’s Hospital, San Francisco, CA, USA. 13Center for Computational Biology, University of California Berkeley, Berkeley, CA, USA. 14Department of Bioengineering and Therapeutic Sciences, University of California San Francisco, San Francisco, CA, USA.

These authors contributed equally and jointly supervised the work: Jennifer M. Puck, Steven E. Brenner. e-mail: anadhikari@berkeley.edu; jennifer.puck@ucsf.edu; brenner@compbio.berkeley.edu
Supplementary Fig. 1. Overlapping MS/MS analyte levels in GA-II patients, VLCADD patients, and VLCADD carriers. For five MS/MS analytes (C12, C14:1, C12:1, C14, C14:2) and two analyte ratios (C14:1/C2, C14:1/C16) that are elevated in GA-II patients (orange), the corresponding analyte and analyte ratio levels are plotted as a multiple of median reference ranges are shown for GA-II patients (orange), VLCADD patients (blue) and VLCADD carriers (purple). The reference ranges are defined by values observed in unaffected individuals (green). The underlying data are from the R4S (Region 4 Stork) consortium website (https://www.clir-r4s.org). The boxplot central line represents the median value, the bottom and top box edges represent the 25th and 75th percentiles, and the lower and upper whiskers extend to 1.5 times the interquartile range. Each boxplot is derived from the total number of cases counts provided below the corresponding boxplot. The upper and lower box edges for the reference (green) represents the 99th and the 1st percentile values for unaffected individuals.