nature medicine

SUPPLEMENTARY INFORMATION

https://doi.org/10.1038/s41591-020-0966-5

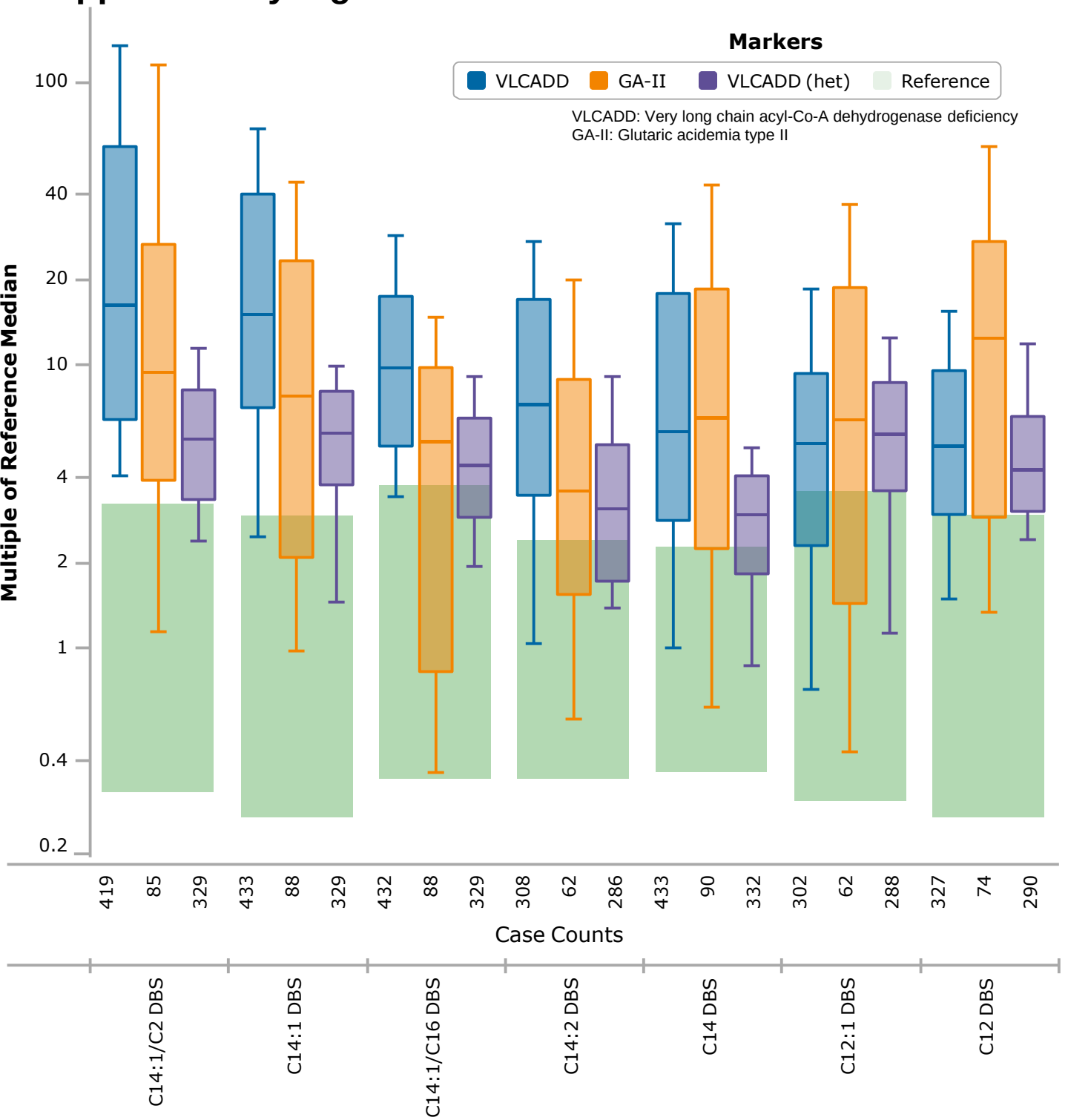
In the format provided by the authors and unedited.

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

Aashish N. Adhikari[1,2]✉, Renata C. Gallagher[2,3], Yaqiong Wang[1], Robert J. Currier[3], George Amatuni[3], Laia Bassaganyas[2], Flavia Chen[2,4], Kunal Kundu[1,5], Mark Kvale[2], Sean D. Mooney[6], Robert L. Nussbaum[2,7], Savanna S. Randi[8], Jeremy Sanford[8], Joseph T. Shieh[2,3], Rajgopal Srinivasan[5], Uma Sunderam[5], Hao Tang[9], Dedeepya Vaka[2], Yangyun Zou[1], Barbara A. Koenig[2,4], Pui-Yan Kwok[2,10,11], Neil Risch[2,12], Jennifer M. Puck[2,3,10,13,16]✉ and Steven E. Brenner[1,2,14,15,16]✉

[1]Department of Plant and Microbial Biology, University of California Berkeley, Berkeley, CA, USA. [2]Institute for Human Genetics, University of California San Francisco, San Francisco, CA, USA. [3]Department of Pediatrics, University of California San Francisco, San Francisco, CA, USA. [4]Program in Bioethics, University of California San Francisco, San Francisco, CA, USA. [5]Innovation Labs, Tata Consultancy Services, Hyderabad, India. [6]Department of Biomedical Informatics and Medical Education, University of Washington, Seattle, WA, USA. [7]Invitae, San Francisco, CA, USA. [8]Department 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. [9]Genetic Disease Screening Program, California Department of Public Health, Richmond, CA, USA. [10]Cardiovascular Research Institute, University of California San Francisco, San Francisco, CA, USA. [11]Department of Dermatology, University of California San Francisco, San Francisco, CA, USA. [12]Department of Epidemiology and Biostatistics, University of California San Francisco, San Francisco, CA, USA. [13]Division of Allergy, Immunology and Blood and Marrow Transplantation, UCSF Benioff Children's Hospital, San Francisco, CA, USA. [14]Center for Computational Biology, University of California Berkeley, Berkeley, CA, USA. [15]Department of Bioengineering and Therapeutic Sciences, University of California San Francisco, San Francisco, CA, USA. [16]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



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 perentile values for unaffected individuals.