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Supplemental Data

Mosaicism of UDP-Galactose Transporter *SLC35A2* Causes a Congenital Disorder of Glycosylation

Bobby G. Ng, Kati J. Buckingham, Kimiyo Raymond, Martin Kircher, Emily H. Turner, Miao He, Joshua D. Smith, Alexey Eroshkin, Marta Szybowska, Marie Estelle Losfeld, Jessica X. Chong, Mariya Kozenko, Chumei Li, Marc C. Patterson, Rodney D. Gilbert, Deborah A. Nickerson, Jay Shendure, Michael J. Bamshad, University of Washington Center for Mendelian Genomics, and Hudson H. Freeze

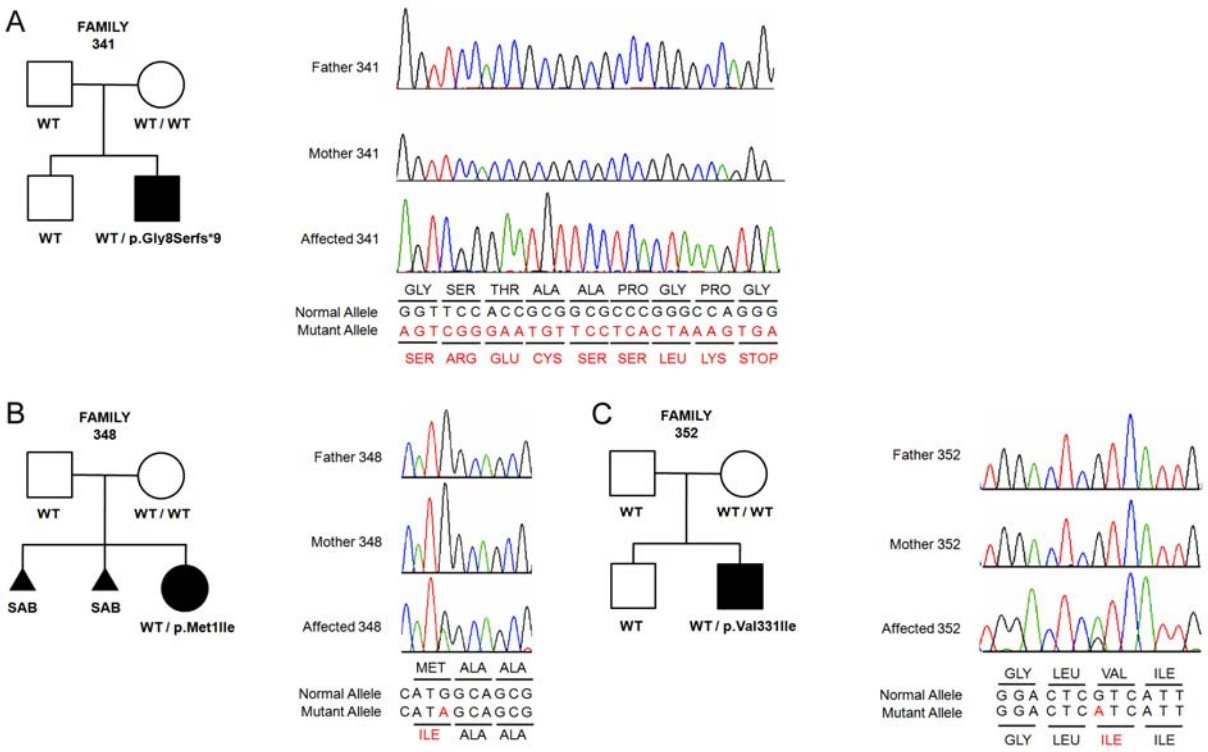


Figure S1. Confirmation of De Novo Mutations in *SLC35A2* by Sanger Sequencing within Each Familial Pedigree

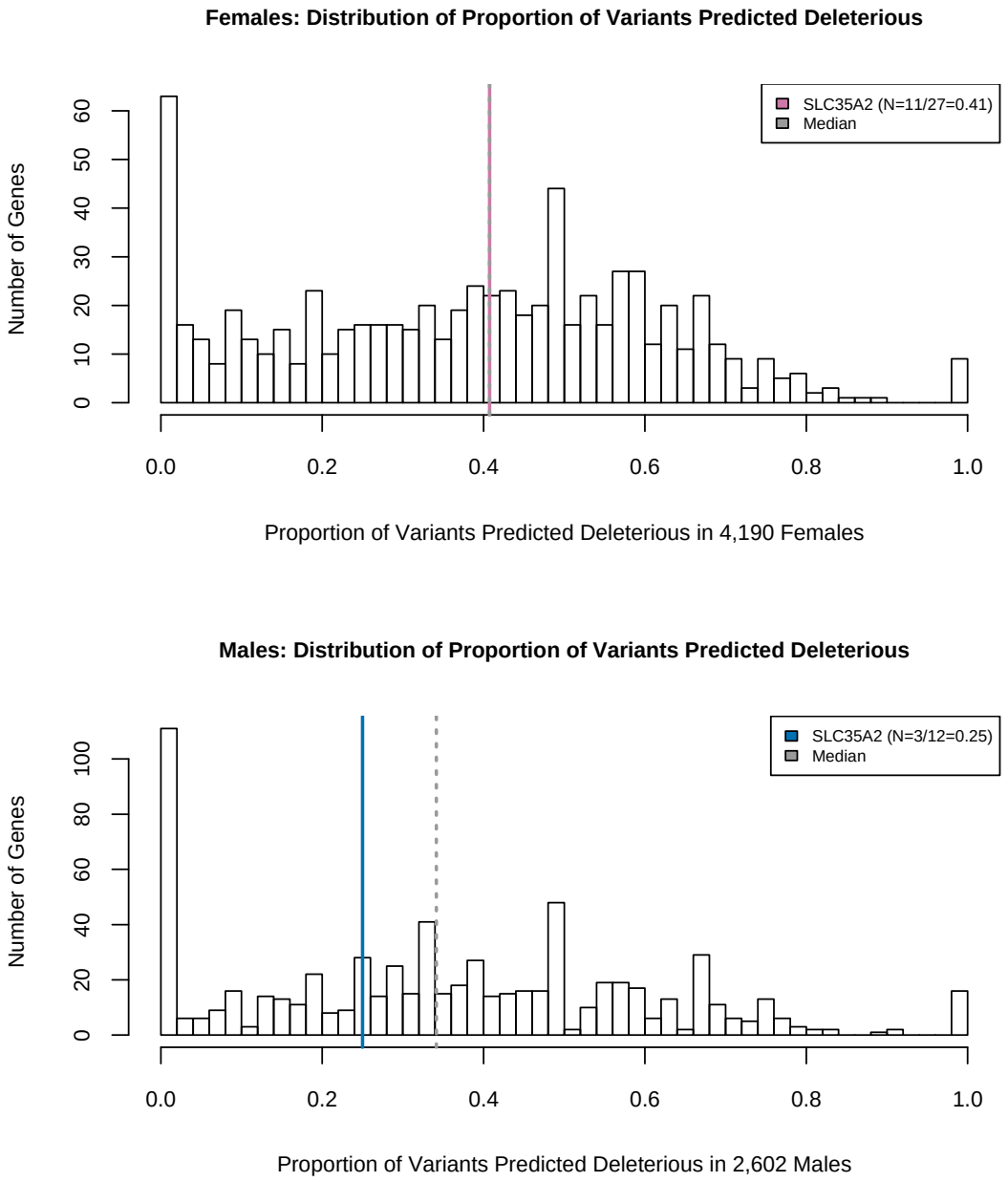


Figure S2. Population-Level Analysis of Putatively Deleterious *SLC35A2* Variants in ESP

The proportion of *SLC35A2* alleles predicted to be functionally deleterious was higher in females (0.41) compared to males (0.25) and the *SLC35A2* alleles observed in males (mean GERP 2.08) were less conserved than in females (mean GERP 2.90). A greater tolerance for deleterious SLC35A2 alleles in females versus males is also supported by the observation that compared to all X chromosome genes, *SLC35A2* is ranked in the lowest ~1/3 of genes in males for proportion of alleles that are deleterious while *SLC35A2* is ranked in the 50th percentile in females.

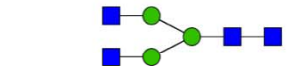
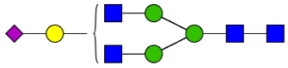
Glycan Structure	Glycan Annotation	Reference Value (Low-High)	CDG-341	CDG-348	CDG-352
	Hex3HexNAc4	0.00 – 0.22	3.28	8.16	0.79
	Neu5Ac1Hex4HexNAc4	0.40 – 1.28	4.86	3.91	2.59

Figure S3. Presence of Galactose-Deficient Glycans in Serum Glycoproteins

N-glycans from whole serum glycoproteins were analyzed by MALDI-TOF and shows accumulation of both agalactosylated and monogalactosylated glycans. Reference values are based on the percentage that glycan constitutes of the total glycan pool. Sera samples for 341 and 348 were taken before the age of 1. Sample 352 was taken at 5 years of age, when his Tf profile was nearly normal.

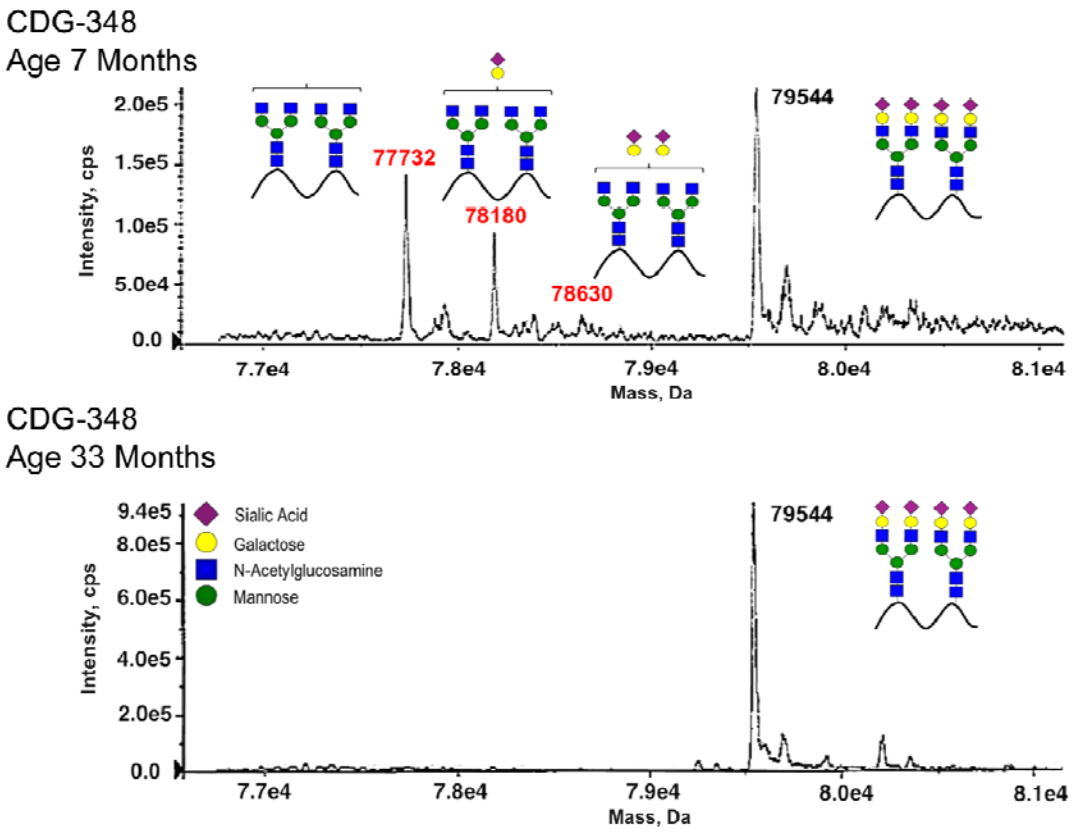


Figure S4. Improved Glycosylated Transferrin

Analysis of serum transferrin from affected individual CDG-348 showing age-dependent correction of the galactosylation deficiency.