

Supplementary figure 1

A



B



C



D



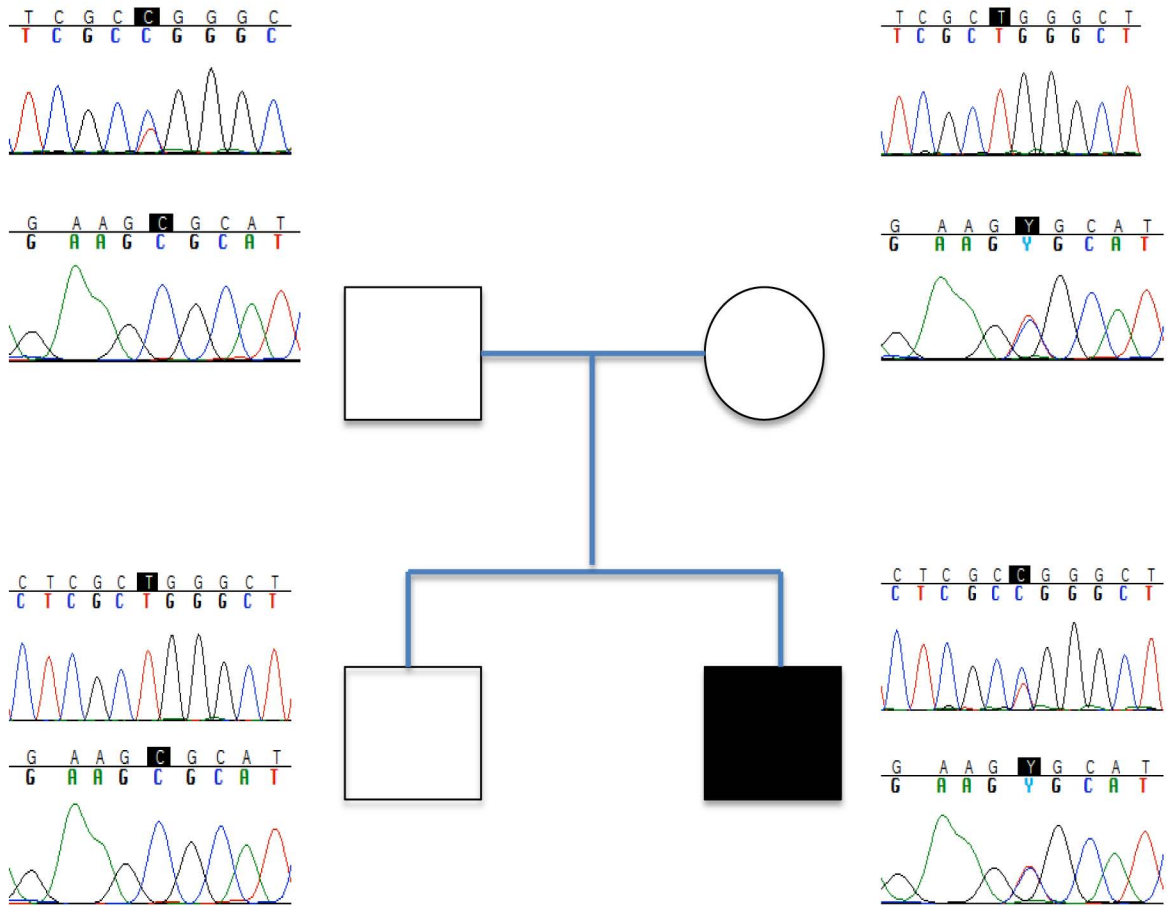
E



F



Supplementary figure 2



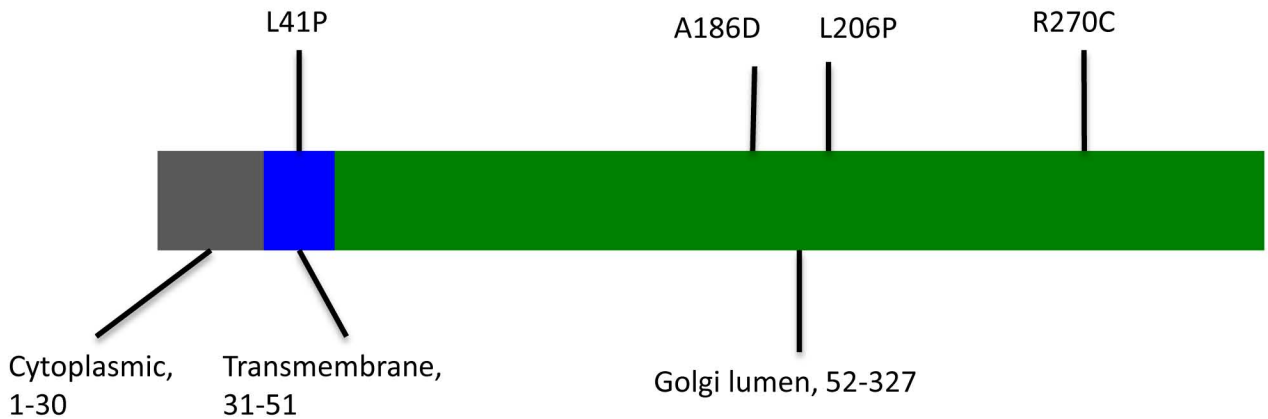
Sanger sequencing for patient and family at *B4GALT7* nucleotide positions 122 (reference allele, T) and 808 (reference allele, C). Top chromatogram for each individual is position 122 and bottom chromatogram is position 808. The c.122T>C was transmitted from the father to the patient, and the c.808C>T was transmitted from the mother to the patient, resulting in a compound heterozygous state for the patient. The unaffected sibling is homozygous wild type for both alleles.

Supplementary figure 3

A

H. sapiens	1	MFPSRRKAAQLPWEDGRSGLLSGGLPRKCSVFHLFVACL SL GGFF-----S	45
Chimpanzee	1	MFPSRRKAAQLPWEDGRSGLLSGGLPRKCSVFHLFVACL SL GGFF-----S	45
Macaque	1	MFPSRRKAAQLPWEDGRSRLLSGSLPRKCSVFHLFVACL LL GGCF-----S	45
Dog	1	-----MGTFLA-----SRLVPGGLPRKCSVFHLFVACL SL GGFL-----S	34
Mouse	1	MLPSRRKAAQLPWEDGRARLLPGGLRRKCSIFHLFIAFLL LV FFF-----S	45
Rat	1	MLPSRRKAAQLPWEDGRARLLPGGLRRKCSIFHLFIAFLL LV FFF-----S	45
Chicken	1	MGPGRRAA-LRLRGGGSPQLLGLLAGKFSIFQLFFLALL LL GFPA-----S	44
Zebrafish	1	MMYSSRRKPVLYFKDERS-----FLSRKCTVWKLFGLCM V FVFG-----S	40
D. melanogaster	1	-----MVNISTINWVFCVGL S FCLGGIIVLS	26
C. elegans	1	-----MKLKTRLILSGTILISL A ACYF----LV	24
H. Sapiens	239	GLQLFRPSGITTGY-KTFRHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	284
Chimpanzee	239	GLQLFRPSGITTGY-KTFRHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	284
Macaque	239	GLQLFRPSGITTGY-KTFRHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	284
Dog	228	GLQLFRPSGITTGY-KTFRHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	273
Mouse	239	GLQLFRPSGITTGY-QTFRHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	284
Rat	239	GLQLFRPLGITTGY-QTFRHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	284
Chicken	231	GLQVHRPSGITTGY-ETFQHLHDPAWRKR D ---Q K RIAAQKQEQFKVDRE	276
Zebrafish	229	NLELFRPTGITTGT-KTFRHIHDPAWRKR D ---Q K RIAAQKQEQFKVDPE	274
D. melanogaster	222	GLQVTRPQNIKTGTNDTFSHIHNR Y HRKR D ---T Q KCFNQKEMTRKR D HK	268
C. elegans	199	KLNLTRVSGSLSTDSSTFRHIHGPK-RKR D YTP K KN D KNQWEIKRKR D HV	247

B



- A) Amino acid sequence alignments show homology across evolution at both leucine 41 and arginine 270. Alignments were generated using NCBI HomoloGene.
- B) Sequence annotation of β 1,4-galactosyltransferase 7 protein from UniProt. The protein is predicted to be a Type II transmembrane protein in the Golgi membrane. Amino acids 1-30 are predicted to be cytoplasmic (gray), 31-51 are predicted to be a transmembrane domain (blue) and 52-327 are predicted to be a Golgi luminal domain (green). Approximate locations of mutations found in our patient (p.L41P and p.R270C), as well as other previously described pathogenic variants (p.A186D, p.L206P) are indicated above the figure.