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Supplemental Data

**TMEM199 Deficiency Is a Disorder of Golgi Homeostasis
Characterized by Elevated Aminotransferases, Alkaline
Phosphatase, and Cholesterol and Abnormal Glycosylation**

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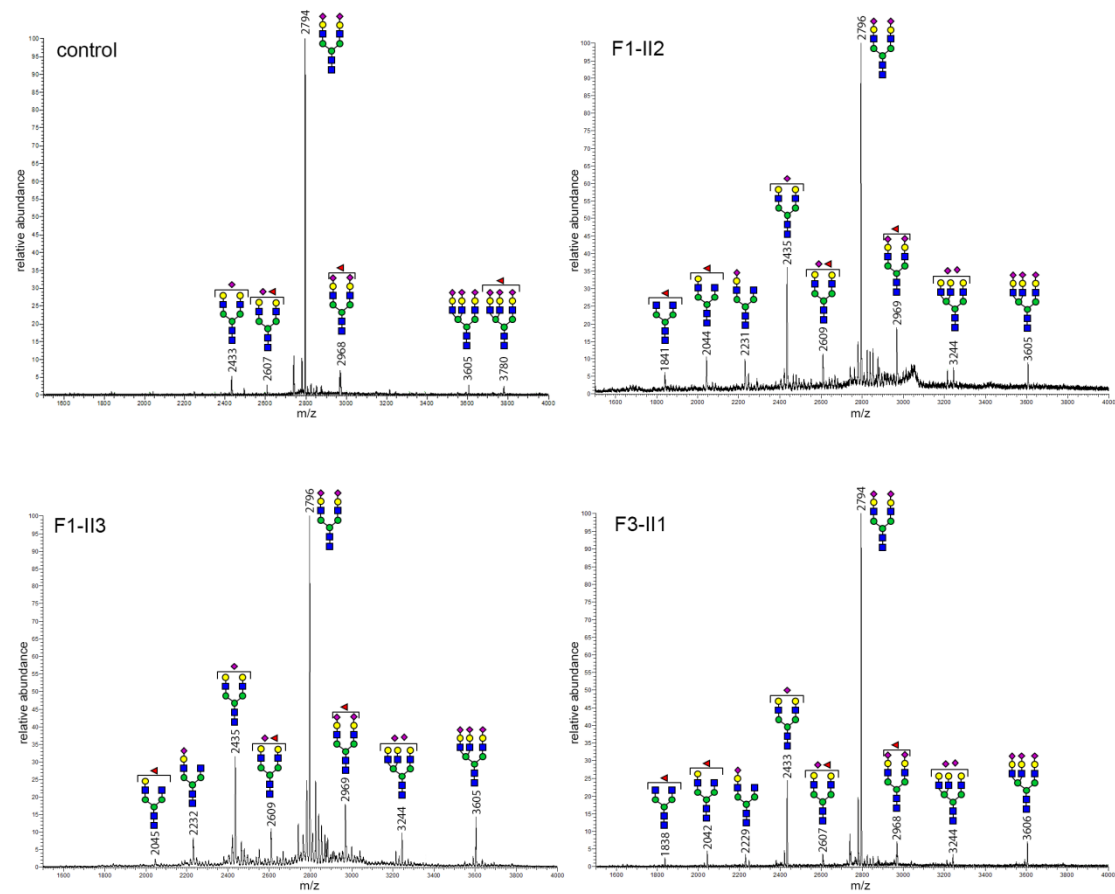
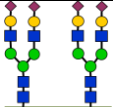
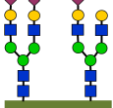
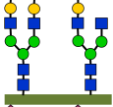
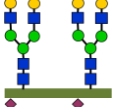
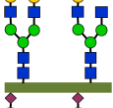
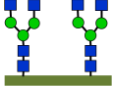


Figure S1 MALDI mass spectrometry profiles of total serum N-glycans of a healthy control and TMEM199 deficient individuals F1-II2, F1-II3 and F3-II1. An increase in hypoglycosylated glycans can be seen for all affected individuals when compared to a healthy control. Most notably the glycans with theoretical masses of 2433 m/z and 2228 m/z are increased, indicating loss of one sialic acid (2433 m/z) or one sialic acid plus one galactose (2228m/z).

Table S1 quantifications of transferrin Isoelectric focusing								
		0-sialo (%)	1-sialo (%)	2-sialo (%)	3-sialo (%)	4-sialo (%)	5-sialo (%)	6-sialo (%)
Control range (n=60)		0.0-3.2	0.0-5.0	3.3 -7.6	4.9 -10.6	47.3-62.7	18.7-31.5	3.2-7.8
Family	individual							
F1	II2	2.85	7.07 ↑	16.46 ↑	24.38 ↑	33.58 ↓	13.49 ↓	2.17 ↓
	II3	1.28	4.53	15.60 ↑	27.26 ↑	34.64 ↓	13.77 ↓	2.92 ↓
F2	II2	1.31	3.51	11.18 ↑	21.95 ↑	49.53 ↓	10.54 ↓	1.98 ↓
F3	II1	1.4	3.9	13.6 ↑	27.6 ↑	38.7 ↓	12.4 ↓	2.5

Table S2 quantifications of Apolipoprotein C-III Isoelectric focusing					
			0-sialo (%)	1-sialo (%)	2-sialo (%)
Control range	Age 0-1		0.2-4.5	42.7-69.8	26.2-56.7
	Age 1-18		1.4-9.5	48.5-75.2	21.0-45.8
	Age >18		1.8-7.6	47.4-73.6	20.2-46.5
Family	individual	Age at analysis			
F1	II2	17	5.1	83.9 ↑	11.0 ↓
	II3	9	5.9	81.7 ↑	12.6 ↓
F2	II2	>18	7.2	65.5	27.3
F3	II1	12	3.5	85.3 ↑	11.2 ↓

Table S3 Glycan structures (peak numbers associated with Figure 3C)

Peak	Structure	Predicted mass (amu)
1		79555
2		79265
3		79104
4		78973
5		78811
6		78649