

Supplementary Materials

A novel homozygous variant in the *ABCG8* gene identified in a child with sitosterolemia

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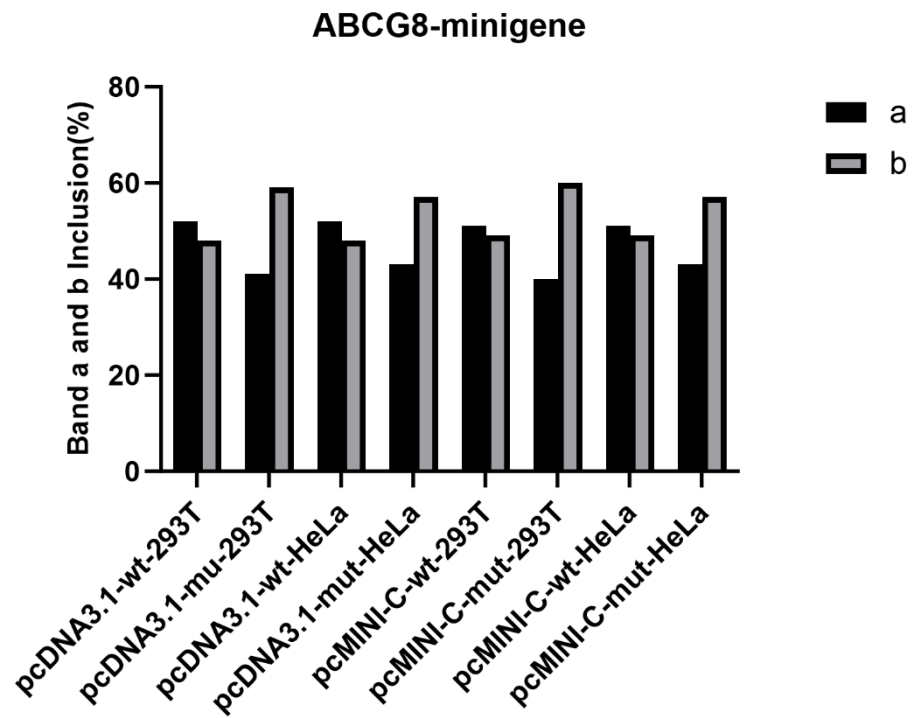


Fig. S1 Quantitative analysis of the minigene splicing assay. Band a, normal transcript; band b, aberrantly spliced product.

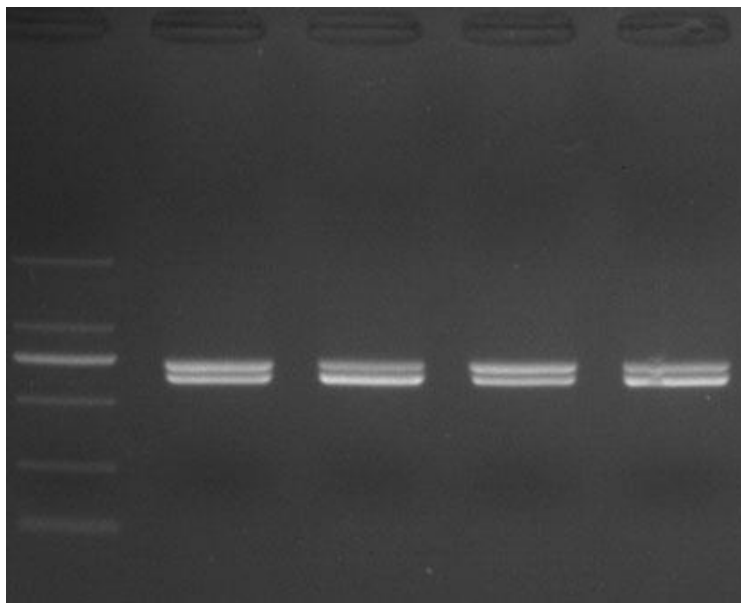


Fig.S2 The original uncropped gel images from minigene assays using the pcDNA3.1 Vector.

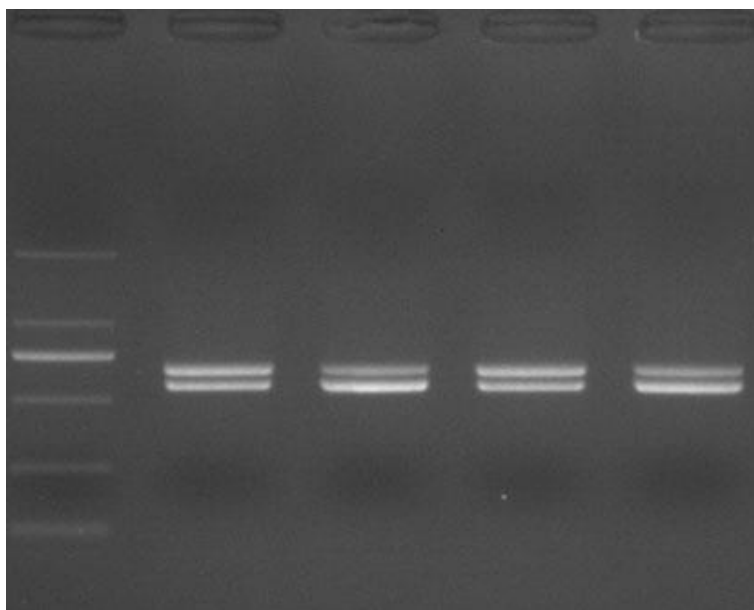


Fig.S3 The original uncropped gel images from minigene assays using the pcMINI-C Vector.

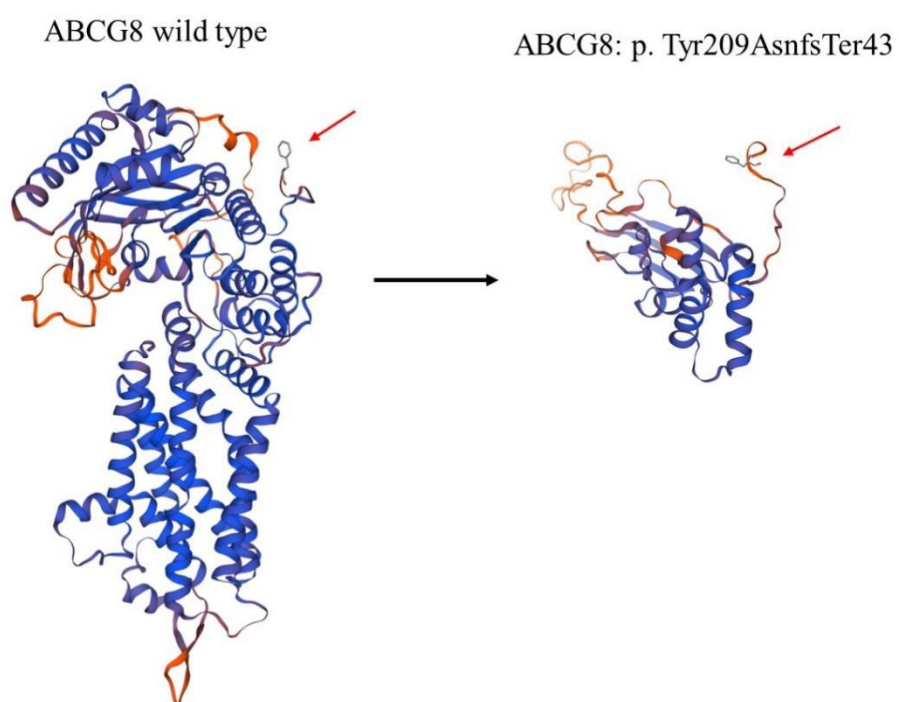


Fig. S4 The software predicts that abnormal splicing leads to truncated protein. The red arrow marks the N-terminus, with phenylalanine (Phe) at position 28.

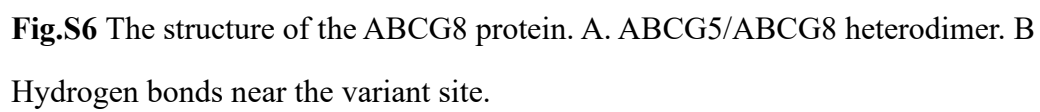


Table S1. The blood test timeline of the patient

	WBC		RBC	HCT	PLT	MPV		P-LCR	MCV		MCHC	RWD-CV
Timeline	(10^9/L)	Hb (g/L)	(10^12/L)	(%)	(10^9/L)	(fL)	PDW (%)	(%)	(fL)	MCH (pg)	(g/L)	(%)
3 years and 10 months old	6.97	76	2.85	26	105				91.2	26.7	292	16.8
3 years and 10 months old	7.23	91	3.33	29.1	103				87.4	27.3	313	17.1
4 years and 3 months old*	8.56	107	3.96	35.6	126				89.9	27	301	16.5
4 years and 4 months old	8.89	123	4.37	39.1	138				89.5	28.1	315	16.3
4 years and 6 months old	Genetic diagnosis and initiation of ezetimibe therapy											
6 years old	7.42	106	3.67	31.6	101	15.2	33.4	57.4	86.2	29	337	15.7
7 years old	7.48	122	4.41	36.3	113	17.3	28.8	55.4	82.2	27.6	336	14.8
Normal reference range	4.3-11.3	118-156	4.2-5.7	36-46	167-453	9.4-12.5	15.5-18.1	13-43	76-88	25-34	309-359	11-15

Abbreviations: Hb, hemoglobin; RBC, red blood cell count; HCT, hematocrit; PLT, blood platelet; MPV, mean platelet volume; PDW, platelet distribution width; P-LCR, platelet large cell ratio; MCV, mean corpuscular volume; MCH,mean corpuscular hemoglobin; MCHC, mean corpuscular hemoglobin concentration, RDW, red blood cell distribution width. The green numbers indicate values below the normal reference range, while the red numbers indicate values above the normal reference range. * Peripheral blood smear collection time.

Table S2 Concentration of plant sterols in the blood of the patient's family members (μmol/L)

	β-sitosterol	Stigmasterol	Campesterol	7-Enylcholestanol	Dehydrocholesterol	Dihydrocholesterol
	(3.74-14.58)	(0.09-1.08)	(5.39-20.81)	(1.71-8.51)	(2.81-8.00)	(4.44-12.53)
Pre-treatment of the proband	565.99	42.31	238.46	2.48	6.36	27.85
Post-treatment of the proband	374.43	19.34	112.11	4.22	4.1	14.14
Mother	21.73	0.67	19.9	2.94	3.13	9.35
Elder sister	17.06	0.33	15.57	1.92	3.02	7.29
Younger sister	20.65	0.47	21.17	3.7	5.08	6.93

Note: Values above the reference range are shown in red.