Appendix 1

Some useful genetic websites

- ❖ ClinVar (68): https://www.ncbi.nlm.nih.gov/clinvar/
- ❖ ClinGen (69): https://clinicalgenome.org/
- ❖ db Var (70): https://www.ncbi.nlm.nih.gov/dbvar/#:~:text=dbVar%20is%20NCBI's%20database%20of,elements%2C%20translocations%2C%20and%20complex%20variants
- ❖ National Human Genome Research Institute, Genome Glossary (71): https://www.ncbi.nlm.nih.gov/projects/genome/glossary.shtml
- ❖ gnomAD: https://gnomad.broadinstitute.org/
- ❖ Human Gene Mutation Database (HGMD®) (72): https://www.hgmd.cf.ac.uk/ac/search.php
- ❖ Medline Plus (73) https://medlineplus.gov/genetics/
- ❖ Variation Viewer (74): https://www.ncbi.nlm.nih.gov/variation/view/

Table S1 Siblings' lipid profiles at the presentation (54)

Lipid profile	Reference	Female twin	Male twin	Older brother
Total cholesterol (mg/dL)	<200	250	414	381
Triglyceride (mg/dL)	<150	1,386	3,265	206
HDL-C (mg/dL)	>40 (males)	17	15	63
Direct LDL-C (mg/dL)	70–130	NA	18	36
Apolipoprotein B-100 (mg/dL)	<75	<40	<40	<40

HDL-C, high-density lipoprotein cholesterol; LDL-C, low-density lipoprotein cholesterol.

Table S2 LPL enzyme functional analysis results of the siblings (54)

LPL enzyme activity method[†]	Female twin	Male twin	Older brother
Method 1 (68) (normal: ~34.8 mmol/L/min; range, 22–47.6 mmol/L/min) (% of reference)	4.8 (13.8%)	2.5 (7.2%)	NA
Method 2 (69) (normal: ~378.11 nmol/mL/min) (% of reference)	NA	134.99 (35.7%)	146.08 (38.6%)
Method 3 (70) (% of reference)	NA	0 (0%)	0 (0%)
LPL mass, ng/mL (% of reference)	NA	112 (39.9%)	87.9 (31.3%)

[†], no standardization of LPL enzyme analysis leads to different values from different methods. LPL, lipoprotein lipase; NA, not available.