

## Supplementary material

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Totoń-Żurańska J, Wołkow P, Kapusta M, et al. Targeted sequencing of a gene panel in patients with familial hypercholesterolemia from Southern Poland. *Pol Arch Intern Med.* 2023; 133: 16417. doi:10.20452/pamw.16417

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*Table S1* Family with homozygous children with mutation NM\_000527.4:c.100T>G

p.Cys34Gly in exon 2 of the *LDLR* gene

<b>Patient number</b>	<b>Age</b>	<b>LDL-C, mmol/l</b>
418.1 mother heFH	40	4.6
418.2 husband heFH	40	6.2
418.3 son hoFH	18	7.47
418.4 daughter hoFH	16	6.68
418.5 mother 2		1.8
418.6 brother 1	24	3.2
418.7 father 1		5.2
418.8 mother 1	56	1.57

*Table S2* Lipid characteristics of a family with a *LDLR* variant that has not been previously

linked to FH in ClinVar: NM\_000527.4:c.119dupT p.Ser41fs in exon 2

<b>Patient number</b>	<b>Age</b>	<b>LDL-C, mmol/l</b>
284.1 proband F	30	6.5
284.5 sister	39	5.7

284.6 mother	65	3.2
284.7 father	67	3.5
284.8 son	8	4.8

*Figure S1* Corneal arcus in an 18-year-old hoFH patient (published with permission of patient)



Figure S2 Family tree of family 418

418.1 mother (with heterozygous FH, heFH), 418.2 husband of 418.1, also with heFH, 418.3 son (homozygous FH, hoFH), 418.4 daughter (hoFH), 418.5 mother of 418.2, without mutation for FH, 418.6 brother of 418.1, 418.7 father of 418.1 (heFH), 418.8 mother of 418.1

