Table 2. Pathogenic variants identified in heterozygote carriers identified in this study as catalogued on the Leiden Open Variation Database (https://databases.lovd.nl/shared/genes/GBA)

Description at cDNA level (NM_000157.3)	Description at protein level	Type & Classification	Ref	Trial ID	gender	β-gluco-cerebrosidase [µmol/L/h]	Lyso-GB1 [ng/ml]
c.1226A>G	p.(Asn409Ser)	Missense Pathogenic (class 1)	9, 10, 11	G-PRT011	Male	2.3	3.9
				G-RHH121	Female	2.7	3.7
				G-WSH162	Female	1.8	7.2
				G-WSH032	Male	2.5	4.2
c.1504C>T	p.(Arg502Cys)	Missense Pathogenic (class 1)	10, 12, 13	G-BSUH027	Male	2.8	3.7
				G-QHB008	Male	3.6	2.7
				G-QHB014	Female	2.5	4.7
c.680A>G	p.(Asn227Ser)	Missense Pathogenic (class 1)	11, 14	G-PRT160	Female	2.9	4.8
c.1448T>C	p.(Leu483Pro)	Missense Pathogenic (class 1)	15	G-QHB028	Female	2.7	5.0

c.661C>A	p.(Pro221Thr)	Missense Likely Pathogenic (class 2)	16	G-RHH041	Female	2.8	3.5
c.1102C>T	p.(Arg368Cys)	Missense Uncertain Significance (class 3)	17	G-QHB083	Male	2.8	5.9
c.115G>C	p.(Gly39Arg)	Missense Uncertain Significance (class 3)	18	G-WSH107	Female	1.4	<1.0