Supplementary Table SIII **Exome data variant filtering. Number of variants (genes) in the filtering process per patient.**

	GL-1*	GL-2*	GL-3	GL-4	GL-5	GL-6*	GL-7*	GL-8*	GL-9*	GL-10	GL-11	GL-12	GL-13	GL-19	ARG13
All variants (genes)	120 561 (16 104)	117 926 (16 032)	116 939 (16 060)	125 504 (16 411)	122 126 (16 138)	114 545 (15 971)	116 273 (15 969)	118 482 (16 036)	125 517 (16 267)	127 436 (16 242)	127 964 (16 406)	129 222 (16 375)	124 834 (16 096)	122 653 (16116)	94 004 (14840)
Quality filter (>5 variant reads and > 15% variation)	98 839 (15 566)	96 341 (15 959)	93 555 (15 424)	102 644 (15 843)	100 440 (15 601)	90 234 (15 260)	94 018 (15 356)	95 265 (15 423)	107 070 (15 824)	110 111 (15 874)	108 950 (15 993)	110 826 (15 982)	107 164 (15704)	103 887 (15665)	73 039 (13940)
Rare (<0.5%) non-synonymous and splice variants	637 (558)	619 (541)	484 (420)	748 (646)	548 (452)	500 (414)	520 (428)	510 (419)	521 (438)	483 (422)	628 (583)	659 (554)	601 (528)	543 (433)	826 (681)
Familial filtering**	217 (188)		-	-	-	239 (216)		237 (196)		-	-	-	-	-	-
Homozygous variants	14 (14)		7 (5)	25 (24)	9 (8)	3 (3)		3 (3)		6 (5)	27 (25)	34 (21)	38 (36)	9 (6)	66 (55)
Compound heterozygous variants	42 (14)		101 (37)	168 (66)	140 (44)	65 (22)		53 (13)		97 (36)	149 (50)	155 (50)	111 (38)	163 (53	220 (75)
X-linked variants	0		9 (9)	12 (12)	2 (2)	3 (3)		0		3 (3)	9 (9)	5 (5)	5 (5)	4 (4)	7 (4)
Y-linked variants	0		0	0	0	0		0		0	0	2 (2)	0	0	1 (1)
Testis enriched	5 (3)		15 (8)	28 (14)	27 (11)	7 (4)		6 (3)		11 (4)	24 (13)	22 (11)	21 (14)	14 (6)	32 (13)
High-quality candidate variants classified as VUS or (likely) pathogenic	1 (1)		3 (2)	1 (1)	0 (0)	0 (0)		0 (0)		0 (0)	2 (2)	1 (1)	5 (4)	0 (0)	1 (1)