

Supplemental Data Table S1. Size distribution of CNVs found in patients

Size (Mb)	Number of CNVs (%)		
	Number of pathogenic variants (N=65)	Number of VPS (N=67)	Number of VUS (N=58)
<0.5	4 (6.2)	1 (1.5)	23 (39.7)
0.5–1	6 (9.2)	8 (11.9)	16 (27.6)
1–5	35 (53.8)	22 (32.8)	18 (31.0)
5–10	9 (13.8)	20 (29.9)	1 (1.7)
>10	11 (17.0)	16 (23.9)	0 (0.0)

Abbreviations: CNV, copy number variation; VPS, variants of possible significance; VUS, variants of unknown significance.