

Supplemental Data Table S2. Demographic and clinical features of patients according to CMA results

	Total N (%)	Pathogenic N (%)	VPS N (%)	VUS N (%)	Benign N (%)
Gender					
Male	372/617 (60.3)	34/65 (52.3)	33/57 (57.9)	23/51 (45.1)	282/444 (63.5)
Female	245/617 (39.7)	31/65 (47.7)	24/57 (42.1)	28/51 (54.9)	162/444 (36.5)
Age (yr)					
<2	218/617 (35.3)	22/65 (33.8)	25/57 (43.9)	20/51 (39.2)	151/444 (34.0)
2–5	254/617 (41.2)	29/65 (44.6)	18/57 (31.6)	17/51 (33.3)	190/444 (42.8)
5–10	66/617 (10.7)	7/65 (10.8)	4/57 (7.0)	6/51 (11.8)	49/444 (11.0)
>10	79/617 (12.8)	7/65 (10.8)	10/57 (17.5)	8/51 (15.7)	54/444 (12.2)
Clinical features					
Neurodevelopmental disorder	561/589 (95.2)	59/61 (96.7)	57/57 (100)	47/48 (97.9)	398/423 (94.1)
Developmental delay	536/588 (91.2)	56/61 (91.8)	57/57 (98.2)	45/48 (93.8)	378/422 (89.6)
Learning disability or behavioral or psychiatric disorder	390/535 (72.9)	44/57 (77.2)	42/51 (82.4)	33/45 (73.3)	271/382 (70.9)
Seizures	98/584 (16.8)	9/61 (14.8)	12/56 (21.4)	8/47 (17.0)	69/420 (16.4)
Intellectual disability	372/542 (68.6)	42/57 (73.7)	43/53 (81.1)	31/45 (68.9)	256/387 (66.1)
Speech delay	435/553 (78.7)	49/60 (81.7)	48/54 (88.9)	37/47 (78.7)	301/392 (76.8)
Autism spectrum disorder	95/549 (17.3)	12/58 (20.7)	8/55 (14.5)	10/46 (21.7)	65/390 (16.7)
Congenital anomalies	97/580 (16.7)	9/58 (15.5)	11/57 (19.3)	7/48 (14.6)	71/418 (17.0)
Isolated anomaly	52/580 (9.0)	8/58 (13.8)	5/57 (8.8)	2/48 (4.2)	38/418 (9.1)
Multiple anomalies	45/580 (7.8)	1/58 (1.7)	6/57 (10.5)	5/48 (10.4)	33/418 (7.9)
Dysmorphic features	107/580 (18.4)	17/59 (28.8)	19/57 (33.3)	10/47 (21.3)	61/417 (14.6)
Abnormal growth	82/563 (14.6)	9/60 (15.0)	9/56 (16.1)	6/46 (13.0)	58/401 (14.5)
Failure to thrive	66/562 (11.7)	6/60 (10.0)	7/56 (12.5)	5/46 (10.9)	48/400 (12.0)
Short stature	37/529 (7.0)	4/58 (6.9)	4/53 (7.5)	3/45 (6.7)	26/373 (7.0)
Hypotonia	136/584 (23.3)	19/61 (31.1)	21/57 (36.8)	9/48 (18.8)	87/418 (20.8)

Abbreviations: CMA, chromosomal microarray analysis; VPS, variants of possible significance; VUS, variants of unknown significance.