Table 5. Genetic findings according to ultrasound phenotype and severity of ventriculomegaly

Ultrasound phenotype	VM severity	n	Genetic abnormalities, n (%)	P/LP nvariants, (%)	VM-related P/LP variants, n (%)	VUS, n (%)
Overall	–	128	34 (26.6)	20 (15.6)	12 (9.4)	14 (10.9)
Isolated VM	Overall	80	10 (12.5)	4 (5.0)	3 (3.8)	6 (7.5)
	Mild (10–<12 mm)	58	4 (6.9)	1 (1.7)	1 (1.7)	3 (5.2)
	Moderate (12–<15 mm)	18	4 (22.2)	2 (11.1)	1 (5.6)	2 (11.1)
	Severe (≥15 mm)	4	2 (50.0)	1 (25.0)	1 (25.0)	1 (25.0)
VM with additional ultrasound abnormalities	Overall	48	24 (50.0)	16 (33.3)	9 (18.8)	8 (16.7)
	Mild (10–<12 mm)	20	7 (35.0)	3 (15.0)	2 (10.0)	4 (20.0)
	Moderate (12–<15 mm)	16	6 (37.5)	4 (25.0)	2 (12.5)	2 (12.5)
	Severe (≥15 mm)	12	11 (91.7)*/**	9 (75.0)*	5 (41.7)	2 (16.7)

*P<0.01 compared with corresponding mild VM group.

**P<0.01 compared with corresponding moderate VM group.

Abbreviations: VM, ventriculomegaly; P/LP, pathogenic/likely pathogenic; VUS, variant of uncertain significance.

Footnote: Genetic abnormalities included P/LP variants and VUS identified by WES. Percentages were calculated using the number of fetuses in each subgroup as the denominator. VM-related P/LP variants were defined as variants considered potentially associated with the fetal VM phenotype based on integrated genetic and clinical assessment.