Table 7. Prenatal whole-exome sequencing findings and postnatal outcomes of fetuses with ventriculomegaly and pathogenic or likely pathogenic variants

Case	1	2	3	4
Gene	*DCHS1*	*OTC*	*SYNGAP1*	*ANGPT2*
Inheritance	AR	X-linked	AD	AD
Zygosity	Homozygous	Hemizygous	Heterozygous	Heterozygous
Variant classification	Likely pathogenic	Pathogenic	Pathogenic	Likely pathogenic
Variant origin	Inherited from both parents	Maternal inheritance	De novo	Paternal inheritance
Prenatal ultrasound findings	Left lateral ventriculomegaly (12 mm) with poorly visualized cavum septi pellucidi	Bilateral lateral ventriculomegaly (15 mm on the left and 16 mm on the right)	Right lateral ventriculomegaly (16 mm) with nasal bone hypoplasia	Bilateral lateral ventriculomegaly (12 mm on the left and 13 mm on the right) with persistent right umbilical vein
Associated disorder	Van Maldergem syndrome type 1	Ornithine transcarbamylase deficiency	SYNGAP1-related intellectual disability syndrome	ANGPT2-related lymphatic malformation syndrome
Postnatal outcome	Pregnancy continued. At 8 months of age, Griffiths assessment showed developmental quotients ≥85 in all domains, suggesting normal neurodevelopmental outcome.	Pregnancy continued. The neonate developed lethargy, poor feeding, tachypnea, coma, seizures, and respiratory failure shortly after birth and died.	Pregnancy continued. At 12 months of age, Griffiths assessment showed global developmental delay, with DQs of 68, 71, and 65 in motor, personal-social, and language domains, respectively.	Pregnancy continued. At 24 months of age, Griffiths assessment showed DQs ≥90 in all domains, suggesting normal neurodevelopmental outcome.