

Hydrops-related ultrasound phenotypes and chromosomal abnormalities in fetuses with omphalocele: phenotype-based risk stratification

Zhongfeng Tang¹, Xiaojuan Lin¹, Qingmei Sun¹, Ling Lv², Rui Zhang¹

- 1. Prenatal Diagnosis Center, Gansu Provincial Maternity and Child-care Hospital, Lanzhou, Gansu, China.
- 2. Department of Obstetrics, Gansu Provincial Maternity and Child-care Hospital, Lanzhou, Gansu, China.

Objectives To assess whether hydrops-related ultrasound phenotypes are associated with chromosomal or genetic abnormalities in fetuses with omphalocele and to evaluate phenotype-based risk stratification.

Methods This retrospective cohort study included 166 consecutive fetuses with prenatally diagnosed omphalocele between 2021 and 2025. Genetic results were available in 62 cases. Associations between ultrasound findings and chromosomal or genetic abnormalities were evaluated using logistic regression. Risk stratification was performed according to hydrops-related phenotypes.

Results Among 62 fetuses with genetic testing, 28 (45.2%) had chromosomal or genetic abnormalities. The most frequent diagnoses included trisomy 18, trisomy 13, Turner syndrome, and Beckwith–Wiedemann syndrome. Septated cystic lymphatic malformation (historically termed cystic hygroma) was present in 69.6% of abnormal cases compared with 17.4% of normal cases (OR 10.86, 95% CI 2.69–43.89; $p < 0.001$). Generalized skin edema was observed in 68.2% versus 13.0%, respectively (OR 14.29, 95% CI 3.16–64.61; $p < 0.001$). When stratified by phenotype, chromosomal or genetic abnormality rates were 28.6% in cases without hydrops, 50.0% in isolated cystic lymphatic malformation, and 83.3% in cases with combined cystic lymphatic malformation and generalized edema. Compared with no hydrops, the combined phenotype was associated with a 12.5-fold increase in odds of abnormality (95% CI 3.06–51.14; $p < 0.001$).

Conclusions Hydrops-related ultrasound phenotypes are strongly associated with chromosomal or genetic abnormalities in fetuses with omphalocele. The combination of cystic lymphatic malformation and generalized edema identifies a very high-risk subgroup and may help prioritize invasive genetic testing.