Giant isolated exomphalos: role of prenatal diagnosis in prognosis and after birth treatment coordination

Cubo Nava A, Gastaca Abásolo I, Lapresa Alcalde MV, Goenaga Sánchez FJ, Cebrián Muñios C, Ayuso Velasco R
COMPLEJO ASISTENCIAL UNIVERSITARIO DE SALAMANCA, SALAMANCA, Spain

Objective
Exomphalos is a congenital defect of the abdominal wall at the level of the umbilical cord, which involves the herniation of the abdominal contents (intestine and/or liver), with an estimated postnatal prevalence of 1/4000 newborns. Its presence is physiological until the 10th week of gestation but persistence beyond 12 weeks is associated with an increase in chromosomal abnormalities, including trisomies 18 and 13 and Beckwith Wiedemann syndrome. The presence of isolated exomphalos without association to a genetic defect or associated structural anomaly is estimated in 3-6.5% of all cases of exomphalos diagnosed prenatally. Prenatal diagnosis has a key role, as these newborns are a selected population that can benefit from adequate prenatal care and individualized surgical treatment, improving their prognosis. We present a case report of an isolated exomphalos successfully treated in which prenatal diagnosis played a key role to coordinate multidisciplinary teams involved.

Methods
We present the case of a 34-year-old woman with a singleton pregnancy. During the 12-week scan, an exomphalos with mixed intestinal and hepatic content was detected. Informed consent was obtained to perform genetic test. The genetic study was negative for trisomies and Beckwith-Wiedemann syndrome. At 24 weeks, the content of the exomphalos became only hepatic. At 36 weeks, the estimated size of the sac was 50x40 mm. It was decided to perform a caesarean section at 38 weeks, with the coordination of the obstetrics, neonatology and paediatric surgery teams. A live male baby was born weighing 3540 g, Apgar scores were 9 and 10, pH: 7.32. He was admitted in newborn intensive care unit. At 48 hours of life, primary closure of the hernia was performed. The newborn evolved favourably and was discharged two weeks after surgery.

Results
Prenatal diagnosis plays a key role in the management of exomphalos, as it allows the coordination of a multidisciplinary team to treat the newborn. Although there are different approaches for this pathology, early surgical closure is proposed as the most appropriate. This may be primary or staged repair depending on intra-abdominal pressure, which must be monitored during the intervention to avoid abdominal compartment syndrome.

Conclusion
The presence of exomphalos not associated to genetic defect or structural anomaly is unusual, with an estimated prevalence of 3-6.5% of all cases of exomphalos prenatally diagnosed. Prenatal diagnosis is crucial concerning approach and treatment. Adequate and early postnatal surgical management significantly improves neonatal outcome. Early surgical closure is the treatment of choice at present. Intra-abdominal pressure must be monitored during surgery to avoid abdominal compartment syndrome. A coordinated multidisciplinary team is a key factor for successful management of these patients.