Omphalocele

The patient presented for a routine first-trimester screening ultrasound. An anterior abdominal wall defect was seen in the midline on both sagittal and axial views. It measured (actual measurement in mm). Through the defect a herniation sac was protruding containing (specify bowel loops/liver) covered by an intact membrane. The lump protruded from the cord insertion area, and color Doppler revealed the umbilical cord inserting into the mass, with its vessels draping over the surface before entering the abdomen.

A thorough evaluation showed an increased NT of XXX mm. No additional abnormalities could be detected. The heart was in regular position and structurally normal. The baby was active, and extremities appeared normal with a good range of movements. There was a normal-sized bladder surrounded by two umbilical arteries. The umbilical cord did not appear short. A cord cyst measuring (XXX mm) was/was not noted.???

These findings were consistent with a diagnosis of omphalocele. Fetuses with omphalocele have an increased risk for chromosomal abnormalities, commonly Trisomy 18 as well as other genetic conditions, such as Beckwith–Wiedemann syndrome. In view of the increased risk for aneuploidy, chorionic villus sampling for microarray/karyotyping was offered. The couple was understandably upset and needed more time to discuss whether they wanted to undergo invasive testing. Cell-free DNA (cfDNA) screening was also offered as a second-choice option.

After a thorough counselling about the nature of the findings, implications for the pregnancy, and long-term outcomes, the option of termination of pregnancy (TOP) was also discussed.

If the pregnancy will be continued, a detailed fetal echocardiography and subsequent regular scans for fetal growth and well-being assessment will be conducted.

Arrangements will be made for delivery at a tertiary care centre with NICU and paediatric surgery services. The optimal mode of delivery will be established later in pregnancy. While vaginal delivery is not contraindicated caesarean section may be preferred in case of a giant omphalocele.

A multidisciplinary team, including a neonatologist, a clinical geneticist, and a paediatric surgeon will be involved in the management of the newborn. Surgical correction of the defect may be performed in a single intervention or may require multiple-step procedures, depending on the size of the omphalocele and the presence of associated anomalies.

Neonates with isolated omphalocele have a good prognosis, with up to 96% survival rate. In general, if the omphalocele is isolated and spares the liver, the prognosis is excellent. On the contrary, if the liver is in the sac, the prognosis is relatively poor, with decreased survival rates.

In cases without chromosomal abnormalities, neonates with omphalocele typically exhibit normal mental development. However, motor delay at the age of two is observed more often in children with a giant omphalocele compared to those with a small omphalocele.

A neonatal alert will be raised, and consultation with a paediatric surgeon for pre and postnatal management, as well as genetic consultation, will be arranged.

The recurrence risk is dependent on the underlying condition. However, in subsequent pregnancies, targeted evaluation of the fetal abdominal wall in the first trimester is suggested.

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