Gastroschisis

On today's scan, at the level of the abdomen, a full-thickness abdominal wall defect appeared to the right of the umbilical cord insertion. The diameter of the defect was XXX mm. There are multiple loops of fetal bowel floating freely in the amniotic fluid.

No membrane was identified covering the defect nor the bowel loops, and color Doppler showed an intact umbilical cord insertion into the abdomen. The stomach and liver were located intra-abdominally. There were no signs of intra or extra-abdominal bowel dilation, and the bladder was not herniated. No other defects were noted, indicating this was an isolated finding. There were no signs of intra or extra-abdominal bowel dilation. The bladder was not herniated.

The ultrasound findings were consistent with the diagnosis of isolated gastroschisis.

The patient was XXX years old with no smoking or drug use history.

Gastroschisis is typically not associated with underlying chromosomal abnormalities. The first trimester screening was deemed low risk for aneuploidies, and therefore, no further genetic testing was suggested. An elevated maternal serum alpha fetoprotein (MS-AFP) is commonly found in cases of gastroschisis.

A detailed evaluation of fetal anatomy, especially of the fetal heart was suggested in the second trimester to rule out associated anomalies.

Further scans to assess the content of the defect, the appearance of the bowel, and fetal growth were suggested. As the risk of stillbirth is increased, fetal wellbeing regular assessment is recommended (non-stress test or biophysical profile at least weekly and starting at 32 weeks' gestation or earlier if fetal growth restriction).Fetal growth restriction presents in up to 77% of the fetuses with gastroschisis. This may be an overestimation, due to the smaller abdominal circumference measurement, as a result of the gastroschisis. Umbilical artery Doppler is recommended when a diagnosis of fetal growth restriction is made.

The aim of monitoring is to get the pregnancy to term and arrange for a planned delivery. There is, however, an increased risk of iatrogenic or spontaneous premature delivery.

Consultation with a neonatologist and a pediatric surgeon will be arranged for the couple.

Delivery should be performed in a centre with neonatal intensive care and paediatric surgery services.

Vaginal delivery is not contraindicated, and caesarean delivery is reserved for the usual obstetric indications.

We explained to the parents that gastroschisis increases the risk of stillbirth, which can be as high as 12.5%. However, iatrogenic prematurity between 34-36 weeks is associated with delay in achieving full enteral feeds, prolonged hospitalization and higher incidence of sepsis compared to delivery at term. Therefore, the timing of delivery must carefully balance these risks.

The type of surgical repair will depend upon the type of gastroschisis (simple or complex) and whether the closure of the defect will be feasible within the primary surgical procedure. Some fetuses may require consecutive staged procedures with siloing and/or reoperation for complications. We informed the parents that in 90% of cases, the outcome of an isolated gastroschisis is generally favorable following a surgical repair. However, in 10% of cases, bowel complications such as intestinal stenosis or atresia may occur. There is a small risk of vascular insufficiency leading to volvulus or bowel infarction. In some instances, resection of injured intestines may result in short bowel syndrome. Additionally, a dilated fetal stomach is associated with an increased risk of volvulus and neonatal death.

Gastroschisis has a recurrence risk of approximately 3-5%, which is believed to be multifactorial, involving both genetic and environmental factors. Avoiding smoking and drug use is crucial, as they are well-known risk factors for gastroschisis as well as other adverse health effects. In subsequent pregnancies, a first trimester ultrasound is recommended after 12 weeks,, once the normal midgut herniation has resolved.