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A case of concomitant duodenal atresia and oesophageal atresia without tracheoesophageal fistula

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Objective

Concomitant duodenal atresia (DA) and oesophageal atresia (EA) without trachea-oesophageal fistula (TEF) is rare. We describe the prenatal diagnosis, perinatal management and paediatric surgical course of such a case, characterised by dilated distal oesophagus, stomach and duodenum, duodenal rupture and polyhydramnios.

Methods

This is a case report.

Results

A 37-year-old primipara was referred to our clinic at 25 weeks of gestation due to suspicion of duodenal atresia on ultrasound scan performed elsewhere. Sonographic examination showed massive dilatation of the fetal stomach bubble and duodenum ('ciled-up sign') and intermittent dilation of the distal oesophagus ('upper pouch' sign) in an otherwise structurally normal fetus and with normal amniotic fluid volume. At this time, differential diagnosis included DA associated with an intra-thoracic anomaly such as oesophageal duplication or hernia and concomitant DA and EA. An amniocentesis was performed which showed normal QF-PCR and array-CGH. Maternal viral infection and cystic fibrosis screening were negative. A follow-up ultrasound examination carried out 3 weeks later showed shrinkage of the stomach and duodenum and appearance of massive ascites, suggesting rupture of the stomach or duodenum. The couple was counselled by a multi-disciplinary team including Fetal Medicine specialists, Neonatologists and Paediatric surgeons regarding the complexity of the case and the uncertainty of the outcome with possible need for multiple-stage surgical repair. Management options including expectant management and termination of pregnancy were discussed and the parents opted for continuation of the pregnancy. On further follow-up at 29 weeks the ascites had completely subsided, both stomach and duodenum were again significantly dilated and there was moderate polyhydramnios (deepest pool 11 cm). The pregnancy was managed expectantly with weekly scans which showed stable findings, consistent growth pattern and normal biophysical parameters. The mother was asymptomatic for polyhydramnios and the cervix had remained long and closed. Spontaneous onset of late preterm labour occurred at 36 weeks of gestation and ended with emergency caesarean section due to failure to progress in second stage. A baby girl weighing 3040g was delivered in good condition. Diagnosis of concomitant DA and long-gap distal EA without TEF was confirmed on X-rays. Duodeno-duodenostomy and gastrostomy for decompression and nutritional management were carried out in the early neonatal period. In view of the long gap atresia primary repair was delayed to 12 weeks when oesophageal anastomosis was achieved albeit under some tension. Recurrent lower respiratory tract infections and congenital malacia of the mid trachea further complicated the clinical picture, the latter requiring aortopexy to stabilise the intrathoracic airway. Currently, at the age of 7 months, the infant has an on-going requirement for intermittent oesophageal dilatation of an anastomotic stricture.

Conclusion

Prenatal diagnosis of concomitant OA and DA is rare. The combination of a thoracic cystic structure, distension of the stomach and duodenum and polyhydramnios should raise the suspicion of concomitant OA and DA. Prenatal diagnosis aids the improvement of perinatal outcome. Diagnostic work-up should include detailed anatomy survey to exclude associated defects and genetic testing for exclusion of major chromosomal/genetic abnormalities. It is imperative to counsel the parents with regards to the significant perinatal morbidity and risk of short and long term complications.