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REVIEW ARTICLE

Prenatal consultation for foetal anomalies requiring surgery

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ABSTRACT

Background: During prenatal screening of pregnant women, foetal anomalies requiring surgery may be diagnosed. Healthcare providers should have a basic knowledge of these diseases, including their workup, comorbidities, prognosis, treatment options and any considerations that need to be made in planning for birth.
Aim: This article aims to provide this information by summarising the most recent literature for some of the most commonly diagnosed foetal anomalies requiring surgical correction.
Methods: English language studies on prenatal diagnostic modalities, abdominal wall defects, congenital diaphragmatic hernias, surgical conditions leading to airway compromise, hydrops fetalis, intestinal obstruction and abdominal cysts were retrieved from the PubMed database.
Findings: The most recent and relevant literature is summarised regarding the above listed paediatric conditions. The incidence and prevalence (when available), prognosis, workup, common comorbidities, foetal interventions and special birth considerations (when applicable), and postnatal surgical treatment options are reviewed.
Conclusions: Healthcare providers will occasionally encounter foetal anomalies which may require surgery while performing prenatal screening. They may need to provide early counselling to expectant parents to inform their expectations. When indicated, referrals should be made to a foetal care centre for prenatal consultation. For conditions which may cause danger or distress to the foetus in the immediate postnatal period, preparations should be made to ensure sufficient resources are available at the location of birth.

1. Introduction

Congenital anomalies are increasingly being diagnosed in-utero. Providers caring for mothers of foetuses diagnosed with congenital anomalies will benefit from knowledge about which anomalies may require pre- or postnatal intervention, where the birth should take place and how the baby should be born. Depending on what gestational age a congenital anomaly is identified, and with what kind of anomaly the foetus is diagnosed, the mother may be faced with several options: caesarean section vs vaginal birth, ultrasound guided intervention, foetoscopic intervention, open foetal surgery, extra uterine-intrapartum-treatment (EXIT), termination of pregnancy or observation and postnatal management, including surgery. These decisions are based on a combination of what is known about the anomaly and on the parents' wishes, keeping in mind the diagnostic limitations of the tests used and the spectrum of possible outcomes.

This article aims to provide information to help guide healthcare providers' discussions with parents by summarising the most recent literature for some of the most commonly diagnosed foetal anomalies requiring surgical correction. Armed with a basic understanding of the likely prenatal and perinatal course of each of these anomalies, healthcare providers can more effectively counsel, prepare, and equip women and their families for what to expect. A brief overview of the most commonly applied foetal diagnostic modalities is followed by a review of the more commonly diagnosed foetal anomalies requiring surgery.

2. Methods

A systematic review of manuscripts covering prenatal diagnostic modalities, abdominal wall defects, congenital diaphragmatic hernias, surgical conditions leading to airway compromise, hydrops fetalis, intestinal obstruction and abdominal cysts was conducted using the PubMed database. Only English literature

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studies were included. Care was taken to ensure inclusion of the most recent literature as well as historic landmark papers examining the aforementioned anomalies. The data are presented by incidence and prevalence (when available), prognosis, work up and common comorbidities, foetal interventions and special birth considerations (when applicable), and postnatal surgical treatment options.

3. Prenatal diagnostic modalities

Prenatal diagnostic studies frequently used to survey for disease in the developing foetus include ultrasound imaging, foetal magnetic resonance imaging (MRI) and foetal echocardiography. Testing for DNA or markers for anomalies may also be performed via maternal blood sampling, such as quad screening and cell free DNA, as well as via amniocentesis and chorionic villus sampling (CVS). A structural survey ultrasound is typically performed at 18–22 weeks, unless there is early suspicion for an anomaly. Foetal MRI is an imaging modality with a great potential for better delineating anomalies. It is an expensive test that requires an MRI scanner capable of ultrafast scanning and radiologists experienced in the interpretation of the results. Compared to ultrasound, foetal MRI is particularly helpful in the evaluation of foetal spinal cord defects, brain anomalies, congenital diaphragmatic hernia and complex urogenital anomalies, and is routinely used in high volume foetal care centres. Foetal echocardiography is used in the diagnosis of congenital heart defects and to screen for cardiac anomalies in a foetus with anomalies predictive of cardiac defects such as exomphalos.[3] Foetal echocardiography can provide very important information crucial for birth planning and counselling. Aneuploidy testing, in the setting of a high grade of suspicion, may be performed via amniocentesis (after 15 weeks) or via CVS (after 9 weeks),[1] however the risk of miscarriage with these invasive procedures is about 1 in 200–600 and 1 in 100, respectively.[2] A newer test involves isolating foetal DNA from the maternal circulation. This cell free DNA test can detect specific chromosomal anomalies and aneuploidy without the risk of miscarriage. For example, Materni T21 will test for trisomies 13, 16, 18, 21, and 22, sex aneuploidies (in singleton pregnancies), DiGeorge syndrome, Cri-du-chat syndrome, Prader Willi/Angelman syndrome and 1p36 deletion syndrome. Of note, this test has only been validated in "high-risk" pregnancies.[2]

4. Abdominal wall defects

Among congenital abdominal wall defects, gastroschisis and exomphalos are the two most common, both frequently diagnosed in-utero.[4] These will be discussed in the following sections.

4.1. Gastroschisis

Gastroschisis occurs in approximately 2.5–3 in 10,000 births and the incidence has increased over the last two decades.[4–6] The prognosis is favourable, with a 95% survival rate after birth and few foetal losses.[5] The earliest gastroschisis can be diagnosed is in the late 1st/early 2nd trimester. Gastroschisis can be diagnosed with a 90–100% sensitivity in-utero, meaning up to 10% of cases will not be detected until after birth.[5] Screening for chromosomal anomalies is not usually recommended in gastroschisis. An elevated alpha fetoprotein on the triple/quad screen will raise the suspicion for an abdominal wall defect or a neural tube defect and this is typically followed up with an ultrasound examination.[4,7] The prenatal diagnosis of gastroschisis is based on identifying intestine free-floating in the amniotic fluid (there is no sac) and an abdominal defect to the right of the umbilical cord (Fig. 1). If intra-abdominal and/or extra-abdominal bowel dilation is seen, diversely defined in the literature as a lumen greater than

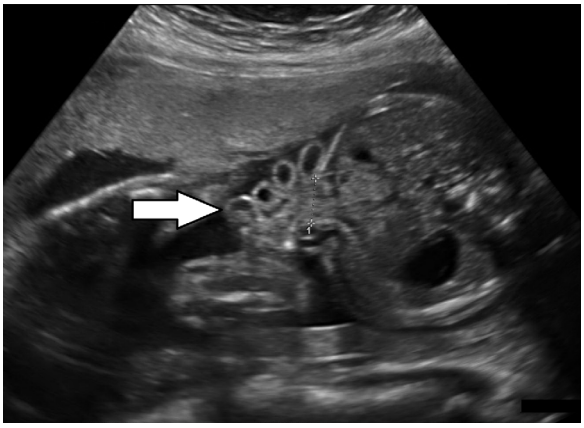

Fig. 1. An ultrasound image of a foetus with gastroschisis. The arrow indicates free floating intestine in the amniotic fluid. The markers span the diameter of the abdominal wall defect.

6–20 mm diameter, this can be an indication of intestinal atresia and "complex" gastroschisis, especially if the dilatation is present on multiple ultrasound scans.[5,8] In gastroschisis, the bowel becomes thickened, possibly due to exposure to the amniotic fluid,[9,10] or due to obstruction of the lymphatic and venous drainage to the intestine and around 15% of cases are complicated by bowel atresia, stenosis, volvulus, ischaemia, necrosis and/or perforation.[5,8,11,12] These cases are classified as "complex" and are associated with higher morbidity.[5,8,11,12] The primary morbidities of gastroschisis are intestinal dysmotility, requiring prolonged total parenteral nutrition (TPN), and the risks associated with TPN including central venous line infection and parenteral nutrition-associated liver disease.[5,8,13] While there are no validated foetal interventions, amniotic fluid exchange is currently in investigative stages.[5,8] However, foetal intervention is hard to justify considering how well overall most infants with gastroschisis do.[8] Premature birth at around 34–36 weeks is common, as is intrauterine growth restriction.[5,8] The data are mixed regarding elective preterm birth versus spontaneous birth, and there is no evidence of any benefit for elective caesarean section.[5,8,14] Postnatal treatment options include silo placement with delayed closure, versus primary closure.[5,8] Delayed closure is considered in order to reduce the risk of abdominal compartment syndrome and respiratory complications.[5] Bowel atresias are commonly repaired after 3–5 weeks,[8] when the thick intestinal peel begins to normalise. Sometimes it is necessary to bring out a stoma, especially in the setting of gastroschisis with necrotic or perforated intestine.[8] Foetuses with gastroschisis have abnormal intestinal rotation, but since the mesentery is broad-based there is a low (about 2%)[11] risk for midgut volvulus. Babies with history of gastroschisis have a life-long risk of bowel obstruction from adhesions; the exact risk is not well defined, but a considerable number of babies will eventually require a secondary operation for gastroschisis-related complications.[15]

4.2. Exomphalos

Exomphalos occurs in about 1–3 per 10,000 births.[4,5,8] However, because of the frequent association with additional birth defects, the true prenatal prevalence of exomphalos is not well defined; there is a high rate of elective termination, especially if additional anomalies are identified.[8] The morbidity and mortality of exomphali are also highly dependent upon the presence of any associated defects. Associated defects include trisomies 13, 18, and 21, cardiac anomalies (occurring in 14–47%),[8]