Fetal Counselling for Surgical Congenital Malformations

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2.1 Introduction

Paediatric surgeons are often called to counsel parents once a surgical abnormality is diagnosed on a prenatal scan. The referral base for a paediatric surgeon now includes the perinatal period. Favourable impact of prenatal counselling has been confirmed to influence the site of delivery in 37% of cases, change the mode of delivery in 6.8%, reverse the decision to terminate a pregnancy in 3.6% and influence the early delivery of babies in 4.5%.

Counselling parents about prenatally suspected surgically correctable anomalies should not be solely performed by obstetricians or paediatricians. Similarly, the paediatric surgeon performing these prenatal consultations must be aware of differences between the prenatal and postnatal natural history of the anomaly. There is often a lack of understanding regarding the natural history and prognosis of a condition presenting in the newborn and the same condition diagnosed prenatally.

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The diagnosis and management of complex fetal anomalies require a team effort by obstetricians, neo-natologists, geneticists, paediatricians and paediatric surgeons to deal with all the maternal and fetal complexities of a diagnosis of a structural defect. This team should be able to provide information to prospective parents on fetal outcomes, possible interventions, appropriate setting, time and route of delivery and expected postnatal outcomes. The role of the surgical consultant in this team is to present information regarding the prenatal and postnatal natural history of an anomaly, its surgical management and the long-term outcome (Lakhoo 2007).

2.2 Historical Overview

Prenatal diagnosis has remarkably improved our understanding of surgically correctable congenital malformations. It has allowed us to influence the delivery of the baby, offer prenatal surgical management and discuss the options for termination of pregnancy in the case of seriously handicapping or lethal conditions. Antenatal diagnosis has also defined an in utero mortality for some lesions, such as diaphragmatic hernia and sacrococcygeal teratoma, so that true outcomes can be measured. Prenatal ultrasound scanning has improved since its first use 50 years ago, thus providing better screening programmes and more accurate assessment of fetal anomalies. Screening

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for Down's syndrome may now be offered in the first trimester e.g. the combined test (using a combination of nuchal translucency measurement and maternal blood markers) or second trimester tests e.g. quadruple blood test. Better ultrasound resolution has led to the recognition of ultrasound soft markers that have increased the detection rate of fetal anomalies, but at the expense of higher false positive rates. Routine ultrasound screening identifies anomalies and places these pregnancies into a higher risk category. Such pregnancies may be referred to Fetal Medicine Units for further scanning and other investigations.

Parents may be offered further invasive diagnostic investigations, such as amniocentesis or chorionic villous sampling. Some structural abnormalities which are difficult to define on ultrasound, such as hindbrain lesions or in the presence of oligohydramnios, may be better imaged with magnetic resonance imaging. With the increasing range of options and sophistication of diagnostic methods, parents today are faced with more information, choice and decisions than ever before, which can create as well as help to solve dilemmas. The different tests and screening procedures commonly in use are outlined below under diagnosis.

2.3 Incidence

Congenital malformations account for one of the major causes of perinatal mortality and morbidity. Single major birth defects affect 3% of newborns and multiple defects affect 0.7% of babies. The prenatal hidden mortality is higher since the majority abort spontaneously. Despite improvements in perinatal care, serious birth defects still account for 20% of all deaths in the newborn period and an even greater percentage of serious morbidity later in infancy and childhood. The major causes of congenital malformation are chromosomal abnormalities, mutant genes, multifactorial disorders and teratogenic agents (Lakhoo 2007).

2.4 Prenatal Diagnosis

2.4.1 Screening for Fetal Anomalies

The NHS fetal anomaly screening programme (FASP) offers screening to all pregnant women in England (NHS Fetal Anomaly Screening Programme Handbook 2018). The first scan is performed at 10 to 14 weeks of gestation. It can:

- Confirm viability.
- Accurately date the pregnancy.
- Diagnose multiple pregnancy and chorionicity.
- Detect major structural anomalies, such as anencephaly.

The combined test can be used to assess the chance of the baby being born with Down's syndrome (trisomy 21), Edward's syndrome (trisomy 18) or Patau's syndrome (trisomy 13). It combines maternal age, gestational age, ultrasound measurement of the nuchal translucency (Fig. 2.1) at between 11 and 14 weeks' gestation with two biochemical markers—PAPPA and free beta hCG—to calculate the risk of the pregnancy being affected by T21, 13 or 18.

If the nuchal translucency cannot be measured, a quadruple test can be offered. This measures four biochemical markers—AFP, hCG, uE3 and inhibin-A. This test can be performed between 14 + 2 weeks and 20 + 0 weeks.

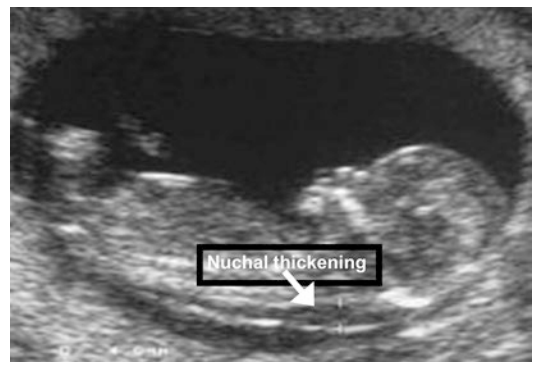

Fig. 2.1 Nuchal translucancy scan

The combined test has a detection rate of 80% for a screen positive rate of 2.5%. The quadruple test has a detection rate of 80% for a screen positive rate of up to 3.5%.

A second scan (often referred to as 'the anomaly scan' is offered at 18 + 0 to 20 + 6 weeks of pregnancy. This scan is designed to identify anomalies which indicate:

- Conditions that may benefit from treatment before or after birth.
- Conditions whose outcome may be improved by planning an appropriate place, mode and timing of birth, along with optimal postnatal management.
- That the baby may not survive the neonatal period.

As a minimum, the conditions screened for at the anomaly scan, along with their detection rates, are:

Anencephaly	98% detection rate
Open spina bifida	90%
Cleft lip	75%
Diaphragmatic hernia	60%
Gastroschisis	98%
Exomphalos	80%
Cardiac anomalies	50%
Bilateral renal agenesis	84%
Lethal skeletal dysplasia	60%

The quality of the images obtained is dependent on many factors, including the skill of the operator, maternal habitus and the position of the fetus. Scan technology is improving images, including 3-D and 4-D options.

Scanning for fetal cardiac defects remains challenging. There is an association between an increased nuchal translucency measurement and the risk of a cardiac defect, or a wide range of other syndromes, even in the context of a normal karyotype. For this reason, those fetuses with a raised NT and normal karyotype are offered fetal echocardiography.

Some anomalies may not be visible at the time of the anomaly scan, but only present later in the third trimester. Examples include duodenal atresia and other forms of bowel obstruction, and non-lethal skeletal dysplasias; in achondroplasia, the femur is of normal length at the time of the anomaly scan.

2.4.2 Invasive Diagnostic Tests

Amniocentesis and chorionic villous sampling (CVS) are the two most commonly performed invasive diagnostic tests (Fetal Medicine Foundation 2019). Since the introduction of the national screening programme for common trisomies, the second trimester scan is interpreted differently. Certain ultrasound findings, previously referred to as 'soft markers' should not be used to recalculate a risk for these trisomies. These findings include choroid plexus cysts, a dilated cisterna magna, echogenic foci in the heart and a 2-vessel umbilical cord. However, other findings, termed 'markers' are of significance and should be referred for further assessment in a Fetal Medicine Unit. These include an increased nuchal fold, cerebral ventriculomegaly, echogenic bowel, renal pelvic dilatation and a small fetus (<fifth centile).

2.4.2.1 Amniocentesis

Amniocentesis is commonly used for detecting chromosomal abnormalities and less often for molecular studies, metabolic studies and fetal infection. It is performed after 15 weeks of gestation and carries a low risk of fetal injury or loss (up to 1%). Full karyotype and microarray analysis takes approximately 2 weeks but newer RAPID techniques, using FISH (fluorescent *in situ* hybridisation) or PCR (polymerase chain reaction), can give limited (usually for trisomies 21, 18, 13) results within 2–3 days.

2.4.2.2 Chorionic Villous Sampling (CVS)

CVS is the most reliable method for first trimester diagnosis and may be performed after 11 weeks of gestation. The test involves ultrasound-guided biopsy of the chorionic villi. The added risk for fetal loss is up to 1%. The samples obtained may be subjected to a variety of tests including full karyotype, rapid karyotyping (FISH—PCR), microarray, enzyme analysis or

molecular studies. Approximate timing of chromosomal results is 1–2 weeks for karyotyping and 2–3 days for FISH and PCR.

2.4.2.3 Prenatal Maternal Serum Screening

It is possible to detect placental DNA in the mother's bloodstream. This is termed total cell free DNA (cfDNA) and in most cases will be the same as fetal DNA. This means that a maternal blood test can be used to detect conditions such as Trisomy 21. Many commercial options are currently available, as a screening test; invasive testing is still advised following a positive result.

The use of maternal blood to discover the fetal blood group for mothers with antibodies such as anti-D, and to determine fetal sex for mothers who are carriers of sex-linked conditions such as haemophilia, is now in routine clinical practice.

Invasive testing may still be advised following the finding of an abnormality on ultrasound scan because of the additional information that microarray testing may bring. ISUOG guidelines suggest that microarrays could detect clinically relevant aberrations in 6% of fetuses with normal karyotype and structural defects.

2.4.2.4 Fetal Blood Sampling (FBS)

Rapid karyotyping of CVS and amniotic fluid samples FISH and PCR have replaced fetal blood sampling for many conditions. However, FBS is still occasionally indicated for required for the diagnosis and treatment of haematological conditions, some viral infections and to investigate chromosomal mosaicism after amniocentesis. When required, it is best performed by ultrasound-guided needle sampling after 18 weeks of gestation. Fetal loss from this procedure is reported to be 1–2%.

2.4.2.5 Fetal Surgery

There is a spectrum of interventions ranging from ultrasound-guided needling procedures, such as cyst aspiration, through fetoscopic minimally invasive techniques (Flake 2017) to hysterotomy and open fetal surgery. Some have been subject to rigorous randomised controlled trials and are now incorporated into routine clinical practice; others have been abandoned and more are still being evaluated. Some remain controversial due the potential long-term effects on both mother and baby (Harrison 2006).

2.4.2.6 Genetic Diagnoses

Antenatal detection of genetic abnormalities is increasing, especially in high-risk pregnancies. Previously undiagnosed conditions such as cystic fibrosis, Beckwith-Wiedemann syndrome, Hirshsprung's disease and sickle cell disease may be detected prenatally following invasive testing and genetic counselling and assessment offered early in pregnancy.

2.4.2.7 Future Developments

The aim of prenatal diagnosis and testing is to ensure 100% accuracy without fetal loss or injury and no maternal risk. National strategies to improve Down's screening using ultrasound, biochemical combination tests and non-invasive prenatal testing (NIPT) are now in place in the UK (Fetal anomalies: screening, conditions, diagnosis, treatment in the UK, 2019).

Expansion of the use of NIPT is ongoing. Its use in Rhesus disease and sex-linked conditions is established. There are now tests available for single gene disorders, such as thanatophoric dwarfism and Apert's syndrome.

The field of genetics is also developing rapidly. WES (whole exome sequencing) and other genetic tests are becoming a clinical reality.

Imaging technology is improving. The use of 3D and 4D imaging is helping with diagnostics and in communication with parents and families. Automation and the use of artificial intelligence (AI) to develop new ways of scanning and screening is also an active area of research.

2.5 Specific Surgical Conditions

2.5.1 Congenital Diaphragmatic Hernia (CDH)

CDH accounts for 1 in 4000 live birth and challenges the neonatologist and paediatric surgeons in the management of this high-risk condition.

Lung hypoplasia and pulmonary hypertension account for most deaths in isolated CDH newborns. If isolated, survival is <15% for severe disease, 50% for moderate disease and > 90% for mild disease. Associated anomalies signify a worse prognosis with a survival rate of less than 10%; chromosomal defects mainly trisomy 13 or 18 or 12p (Pallister Killian) syndrome are found in 20% of fetuses with CDH, genetic syndromes (e.g. Fryn's syndrome) in 10% and other associated anomalies (mainly cardiac and craniofacial) in 20% of cases.

In the UK, most CDHs are diagnosed at the 20-week anomaly scan with a detection rate of around 60%, although as early as 11 weeks' gestation has been reported. 80% of CDHs involve the left side of the diaphragm, 15% are right sided and 5% are retrosternal; left sided hernias are more easily diagnosed because the stomach can be seen in the chest on routine grey scale ultrasound. Magnetic resonance imaging (MRI) can be useful in accurately differentiating CDH from cystic lung lesions. The lung head ratio (LHR) of the fetus can be calculated and has been used to predict prognosis. An LHR of <1, or an observed compared to expected ratio (O/E) of <25% along with early detection, liver in the chest and polyhydramnios are poor predictors of outcome. An O/E ratio of 26–45% suggests moderate disease and >45%, mild disease. Once a diagnosis of CDH is made, detailed scanning, including echocardiography, along with invasive testing for karyotye and microarray is offered.

For those fetuses with an isolated CDH and normal karyotype, FETO (fetoscopic endoluminal tracheal occlusion) can be offered. This involves insertion of an inflatable balloon into the fetal trachea using a fetoscope. The theory is that the balloon prevents the net flow of fluid from the lungs which occurs in fetal life, thereby promoting lung growth. The balloon remains in situ from about 26 to 34 weeks' gestation. An international randomised trial is currently underway to investigate the effectiveness of FETO in comparison to routine postnatal management; results are awaited (https://www.totaltrial.eu/). Women with a fetus with CDH should deliver in a specialist centre. CDH is not an indication for caesarean section.

Postnatal management is aimed at reducing barotrauma to the hypoplastic lung by introducing high frequency oscillatory ventilation (HFOV) or permissive hypercapnea, and treating severe pulmonary hypertension with nitric oxide. No clear benefits for CDH with ECMO (extracorporeal membrane oxygenation) have been concluded in a 2002 Cochrane ECMO study. The value of ECMO is still unclear to date (McHoney and Hammond 2018).

Surgery for CDH is no longer an emergency procedure. Delayed repair following stabilisation is employed in most paediatric surgical centres. Primary repair using the trans-abdominal route is achieved in 60–70% of patients with the rest requiring a prosthetic patch. Complications of sepsis or reherniation with prosthetic patch requiring revision are recorded in 50% of survivors. Minimally invasive techniques have been successful in repairing diaphragmatic defects in 'stable' infants. Long-term survivors of CDH are reported to develop chronic respiratory insufficiency (48%), gastro-oesophageal reflux (89%) and neurodevelopment delay (30%).

2.5.2 Cystic Lung Lesions

Congenital pulmonary airway malformations (CPAM) occur in approximately 1 in 4000 births. They can be macrocystic—made up of one or more large (>2 cm) cysts (type 1), microcystic (type 3) or mixed (type 2). The vast majority (>95%) are unilateral, involving one lobe or segment of the lung. Less common lung anomalies include bronchogenic cysts, congenital lobar emphysema and bronchial atresia. CPAMs are usually isolated. The risk of chromosomal or genetic disorders are not increased, so invasive testing is not usually offered. Fetal echocardiography is performed in cases where there is mediastinal shift, as this can make cardiac assessment more challenging.

Many CPAMs will run a benign course. Some, however, can be associated with hydrops (with a

poor prognosis) or polyhydramnios. If these complications do develop and there is a large cyst, aspiration or shunting (insertion of a pig tail catheter connecting the cyst with the amniotic fluid through the fetal chest) can improve outcome. In the case of type 3 CPAM, a course of antenatal steroids given to the mother can sometimes improve prognosis. Open fetal surgery with excision of the CPAM has been performed in a few cases.

CPAMs, particularly microcystic (type 3) ones, can be difficult to see on ultrasound in the third trimester as the fetus grows and the surrounding lung becomes more echogenic. In 80%, however, the lesion is still present and needs postnatal follow-up. A small CPAM with minimal medistainal shift should not affect the timing, place or mode of birth. A large lesion, where postnatal surgery is contemplated, should deliver in a tertiary unit. Accurate antenatal multiprofessional planning is essential. Postnatal management is dictated by clinical status at birth. Symptomatic lesions require urgent radiological evaluation with chest radiograph and ideally a CT scan (Fig. 2.2) followed by surgical excision. In asymptomatic cases, postnatal investigation consists of chest CT scan within 1 month of birth, even if regression or resolution is noted on prenatal scanning. Plain radiography should not be relied on, because it will miss and underestimate many lesions.

Surgical excision of postnatal asymptomatic lesions remains controversial, with some centres opting for conservative management. The approach to treating this asymptomatic group has evolved in some centres, whereby a CT scan is performed within 1 month post birth, followed by surgery before 6 months of age due to the inherent risk of infection and malignant transformation (Annunziata et al. 2019). Small lesions less than 1 cm may be managed expectantly, as these may not represent CCAM but artifact or end on

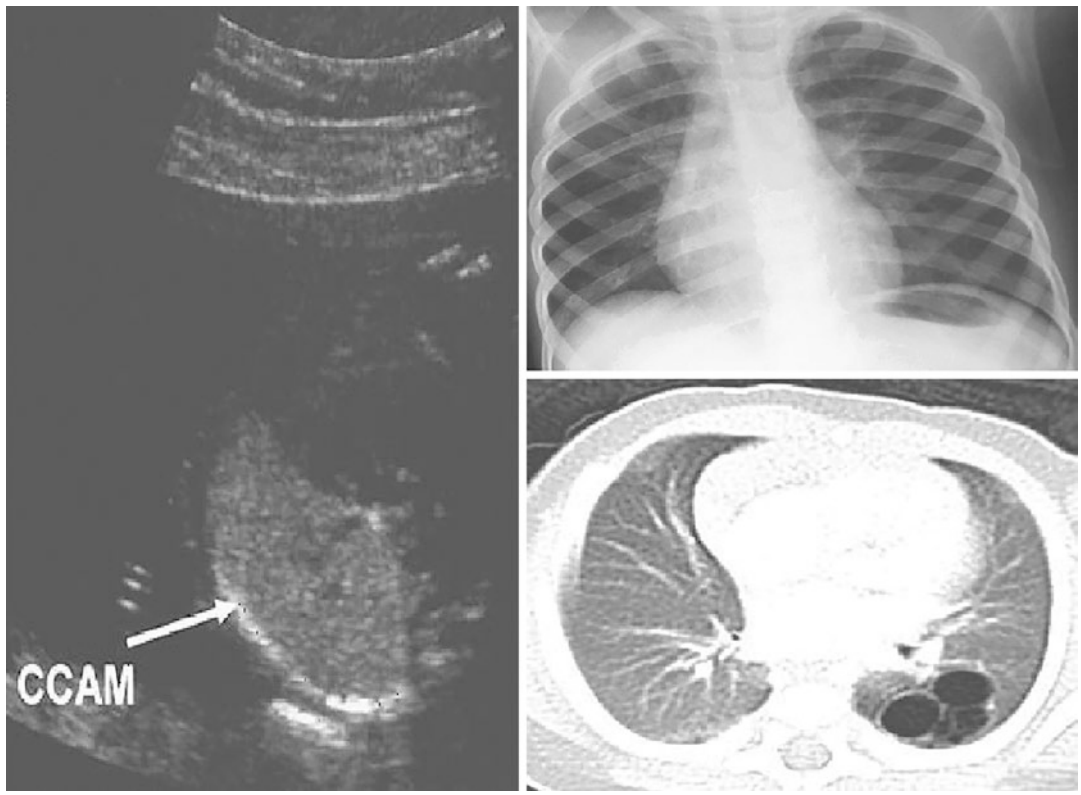

Fig. 2.2 Prenatal scan and postnatal radiological image of CPAM

view of a vessel. True resolution of these lesions is exceptional. Successful outcome of greater than 90% have been reported for these surgically managed asymptomatic lung lesions.

2.5.3 Abdominal Wall Defects

Exomphalos and gastroschisis are both common but distinct abdominal wall defects with an unclear aetiology and a controversial prognosis. Antenatal detection rates for both conditions are high. Most will be found by the time of the anomaly scan; an increasing number is being found in the first trimester.

2.5.3.1 Exomphalos

Exomphalos is characteristically a midline defect, at the insertion point of the umbilical cord, with a viable sac composed of amnion and peritoneum containing herniated abdominal contents. Incidence is known to be 1 in 4000 live births. Associated major abnormalities that include trisomy 13, 18 and 21, Beckwith-Wiedemann syndrome (macroglossia, gigantism, exomphalos), Pentology of Cantrell (sternal, pericar-dial, cardiac, abdominal wall and diaphragmatic defect), cardiac, gastrointestinal and renal abnormalities are noted in 60–70% of cases; thus, karyotyping, in addition to detailed sonographic review and fetal echocardiogram, is essential for complete prenatal screening. Fetal intervention is unlikely in this condition. If termination is not considered, normal vaginal delivery at a centre with neonatal surgical expertise is recommended and delivery by caesarean section only is reserved for large exomphalos with exteriorised liver to prevent damage.

Surgical repair includes primary closure or a staged repair with a silo for giant defects. Occasionally, in vulnerable infants with severe pulmonary hypoplasia or complex cardiac abnormalities the exomphalos may be left intact and allowed to slowly granulate and epithelialise by application of antiseptic solution. Postnatal morbidity occurs in 5–10% of cases. Malrotation and adhesive bowel obstruction does contribute to mortality in isolated exomphalos; however, the majority of these children survive to live normal lives.

2.5.3.2 Gastroschisis

Gastroschisis is an isolated lesion that usually occurs on the right side of the umbilical defect with evisceration of the abdominal contents directly into the amniotic cavity. The incidence is increasing from 1.66 per 10,000 births to 4.6 per 10,000 births affecting mainly young mothers typically less than 20 years old. Associated anomalies are noted in only 5–24% of cases with bowel atresia the most common co-existing abnormality. The incidence of chromosomal and genetic syndromes is not increased. On prenatal scan, with a detection rate approaching 100%, the bowel appears to be free floating, and the loops may appear to be thickened due to damage by amniotic fluid exposure causing a “peel” formation. Dilated loops of bowel (Fig. 2.3) may be seen from obstruction secondary to protrusion from a defect or atresia due to intestinal ischaemia.

Predicting outcome in fetuses with gastroschisis based on prenatal ultrasound finding remains a challenge. There is some evidence that internal bowel dilatation may be predictive; however, thickened matted bowel and Doppler measurements of the superior mesenteric artery are not accurate predictors of outcome. Fetal growth restriction is common (30–60% of cases) and more difficult to monitor because the abdominal

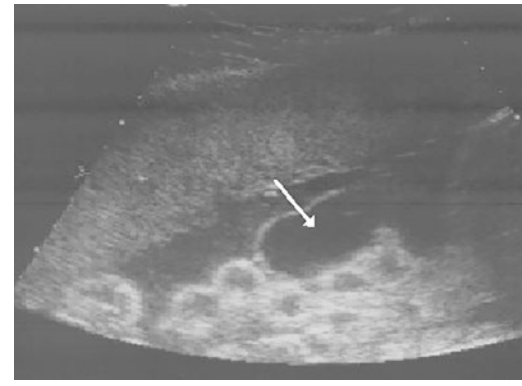

Fig. 2.3 Prenatal ultrasound of dilated bowel in gastroschisis

circumference is more difficult to measure and interpret. Some centres will opt for elective caesarean section for all, but most will offer induction of labour by around 37 weeks of gestation. Delivery needs to be at a centre with paediatric surgical facilities.

Various methods of postnatal surgical repair include the traditional primary closure, reduction of bowel without anaesthesia, reduction by preformed silo, or by means of a traditional silo. Co-existing intestinal atresia could be repaired by primary anastamosis or staged with stoma formation. Variation in achieving full enteral feeding due to prolonged gut dysmotility is expected in all cases.

The long-term outcome in gastroschisis is dependent on the condition of the bowel. In uncomplicated cases, the outcome is excellent in more than 90% of cases. The mortality of live born infants is 5%, with further 5% suffering short bowel syndrome and 10% requiring surgery for adhesive bowel obstruction. Late third trimester fetal loss should always be mentioned during fetal counselling (Gamba and Midrio 2014).

2.5.4 Tracheo-Oesophageal Fistula (TOF) and Oesophageal Atresia (OA)

Repair of TOF/OA is a condition that measures the skill of paediatric surgeons from trainees to independent surgeons. The incidence is estimated at 1 in 3000 births.

Prenatally, the condition may be suspected from maternal polyhydramnios and absence of a fetal stomach bubble at any time from the 20-week anomaly scan. However, if there is an associated tracheoesophageal fistula, the stomach may appear normal on ultrasound scan. It is therefore estimated that oesophageal atresia is suspected prenatally in only about 40% of cases (Bradshaw et al. 2016).

Additional diagnostic clues are provided by associated anomalies, such as trisomy (13, 18, 21), VACTERL sequence (vertebral, anorectal, cardiac, tracheo-oesophageal, renal, limbs) and CHARGE association (coloboma, heart defects, atresia choanae, retarded development, genital hypoplasia, ear abnormality). Associated anomalies, mainly cardiac, are present in more than 50% of cases and worsen the prognosis; fetal echocardiography and invasive testing are therefore usually offered. Duodenal atresia may co-exist with TOF/OA. Amnioreduction (draining of the amniotic fluid) can be offered, particularly for symptomatic relief for the mother, but carries a risk of preterm birth and is only a temporary measure as the fluid will reaccumulate. The risk of recurrence in subsequent pregnancies for isolated TOF/OA is less than 1%. Delivery is advised at a specialised centre with neonatal surgical input.

Postnatal surgical management is dependent on the size and condition of the baby, length of the oesophageal gap and associated anomalies. Primary repair of the oesophagus is the treatment of choice; however, if not achieved, staged repair with upper oesophageal pouch care and gastrostomy or organ replacement with stomach or large bowel are other options. Associated anomalies require evaluation and treatment. Advanced paediatric endosurgical centres may offer minimally invasive thoracoscopic approach to the repair of TOF. Early outcome of a high leak rate and oesophageal stricture requiring dilatation in 50% of cases are expected where the anastamosis of the oesophagus is created under tension.

Improved perinatal management and inherent structural and functional defects in the trachea and oesophagus indicate long-term outcome. In early life, growth of the child is reported to be below the 25th centile in 50% of cases, respiratory symptoms in two-thirds of TOF/OA and gastro-oesophageal reflux recorded in 50% of patients. Quality of life is better in the isolated group with successful primary repair compared to those with associated anomalies and delayed repair.

2.5.5 Gastrointestinal Lesions

The presence of dilated loops of bowel (>17 mm in length and 7 mm in diameter) on prenatal ultrasound scan is indicative of bowel obstruction.

Duodenal atresia has a characteristic 'double bubble' appearance on prenatal scan, resulting from the simultaneous dilatation of the stomach and proximal duodenum. This characteristic sign is, however, usually only present after 24 weeks' gestation, so does not get picked up at the time of the routine anomaly scan. Associated anomalies are present in approximately 50% of cases, most notably trisomy 21 in 30% of cases, cardiac anomalies in 20% and the presence of VACTERL association (vertebral, anorectal, cardiac, tracheo-oesophageal, renal and limbs).

The incidence of duodenal atresia is 1 in 5000 live births. The postnatal survival rate is >95% with associated anomalies, low birth weight and prematurity contributing to the <5% mortality. Temporary delay in enteral feeding occurs due to the dysmotility in the dilated stomach and duodenum.

Many bowel abnormalities may be noted on prenatal scanning (dilated bowel, ascites, cystic masses, hyperparistalsis, poyhydramnios and echogenic bowel); however, none is absolutely predictive of postnatal outcome. Patients with obstruction frequently have findings (especially in the third trimester) of bowel dilatation, polyhydramnios and hyperparistalsis, but ultrasound is much less sensitive in diagnosing anomalies in the large bowel than those in the small bowel. Since the large bowel is mostly a reservoir, with no physiologic function in utero, defects in this region such as anorectal malformations or Hirschsprung's disease are very difficult to detect. Bowel dilatation and echogenic bowel may be associated with cystic fibrosis; therefore, all such fetuses should undergo postnatal evaluation for this disease. Prenatally diagnosed small bowel atresia does not select for a group with a worse prognosis and survival rates are 95–100% (Lau et al. 2017).

2.5.6 Sacrococcygeal Teratoma

Sacrococcygeal teratoma (SCT) is the commonest neonatal teratoma with an incidence of around 1 in 20,000 births. Four types have been defined:

Type 1 external tumour with a small presacral component.

Type 2 external tumours with a large presacral component.

Type 3 predominantly presacral with a small external component.

Type 4 entirely presacral.

The latter carry the worst prognosis due to delay in diagnosis and malignant presentation. Overall perinatal mortality is around 50%. Doppler ultrasound is a useful diagnostic tool to assess tumour vascularity; fetal MRI can provide better definition of the intrapelvic component.

Most teratomas are extremely vascular and the fetus may develop high cardiac output failure, polyhydramnios, anaemia and ultimately hydrops with a mortality of almost 100%. Fetal blood transfusions and amniodrainage may be indicated. Minimally invasive techniques including fetoscopic or ultrasound-guided laser coagulation of blood vessels within the tumour have been tried with limited benefits; the rate of in utero demise or preterm birth post procedure is high (Alalfy et al. 2019).

Caesarean section may be offered to patients with large tumours to avoid the risk of bleeding during delivery. Postnatal outcomes following surgery in type 1 and 2 lesions are favourable; however, type 3 and 4 tumours may present with urological problems and less favourable outcomes. Long-term follow-up with alpha fetoprotein and serial pelvic ultrasounds are mandatory to exclude recurrence of the disease.

2.5.7 Renal Anomalies

Urogenital abnormalities are among the commonest disorders seen in the perinatal period and account for almost 20% of all prenatally diagnosed anomalies. The routine use of antenatal ultrasound scans has resulted in the early detection of these conditions, and in selected cases has led to the development of management strategies including fetal intervention aimed at preservation of renal function. Two major issues are the indications for intervention in bladder outlet obstruction and early pyeloplasty in infancy in cases with hydronephrosis.

Prenatal evaluation of a dilated urinary tract is based on serial ultrasound scans as well as measurement of urinary electrolytes. Ultrasonography provides measurements of the renal pelvis, assessment of the renal parenchyma as well as the detection of cysts in the cortex. In severe disease, lack of amniotic fluid may make ultrasound assessment of the renal tract difficult and MRI may be helpful. Oligohydramnios is indicative of poor renal function and poor prognosis owing to the associated pulmonary hypoplasia. Urogenital anomalies co-exist with many other congenital abnormalities and amniocentesis should be offered in appropriate cases. It is estimated that 3% of infants will have an abnormality of the urogenital system and half of these will require some form of surgical intervention (Yulia and Winyard 2018).

2.5.7.1 Upper Urinary Tract Obstruction

Antenatal hydronephrosis accounts for 0.6–0.65% pregnancies. The most common cause of prenatal hydronephrosis is pelvi-ureteric junction obstruction (PUJ), others being transient hydronephrosis, physiological hydronephrosis, multicystic kidney, posterior urethral valves, ureterocele, ectopic ureter, etc. The prognosis of antenatally diagnosed hydronephrosis in unilateral disease and with a renal pelvic diameter of <10 mm is excellent. Spontaneous resolution is noted in 20% of patients at birth and 80% at 3 years of age. Only around 20% of prenatally diagnosed hydronephrosis need surgical intervention.

Postnatal management of hydronephrosis requires ultrasound at birth and at 1 month of age, and further evaluation with radiology and scintigraphy if an abnormality is suspected. The non-operative treatment of antenatally detected hydro-nephrosis has been carefully monitored over a 17-year period, and from an analysis of six patient series the conclusion is that this approach is safe.

2.5.7.2 Lower Urinary Tract Obstruction

Posterior urethral valves (PUV) are the most common cause for lower urinary tract obstruction in boys with an incidence of 1 in 1500 live male births. The diagnosis of PUV is suspected with a prenatal ultrasound finding of lower urinary tract obstruction (LUTO). With LUTO, there is a thick-walled bladder with evidence of urethral obstruction (a 'keyhole' sign). Depending on the degree of obstruction, there may be back pressure causing dilatation of the ureters and renal pelvices and damage to the renal cortices. The bladder may rupture, causing urinary ascites. Poor prognostic features of PUVs include antenatal detection, damage to the renal cortex and a reduced liquor volume.

It is technically straightforward to insert a shunt between the fetal bladder and amniotic fluid in an attempt to relieve pressure on the urinary system and protect renal function. However, a randomised trial of this procedure versus standard postnatal management struggled to recruit; those babies who were involved fared badly in both arms of the trial and there were high rates of renal failure at 2 years of age in both groups.

Postnatal management includes ultrasound confirmation of the diagnosis, bladder drainage via a suprapubic or urethral route and contrast imaging of the urethra. Primary PUV ablation, vesicostomy or ureterostomy are postnatal surgical options. The overall outcome from this disease is unfavourable.

2.6 Conclusion

The boundaries of paediatric surgical practice have been extended by prenatal diagnosis. The care of patients with surgically correctable defects can now be planned prenatally with the collaborative effort of obstetricians, specialist midwives, geneticists, neonatologists and paediatric surgeons. The understanding of the specific surgical condition's prenatal natural history, the limitations of prenatal diagnosis, the detection of associated anomalies, the risks and indications of fetal intervention programmes and postnatal outcomes are essential to prenatal counselling. Prenatal counselling is an essential component of paediatric surgical practice and should be included in the training programme for future paediatric surgeons.

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