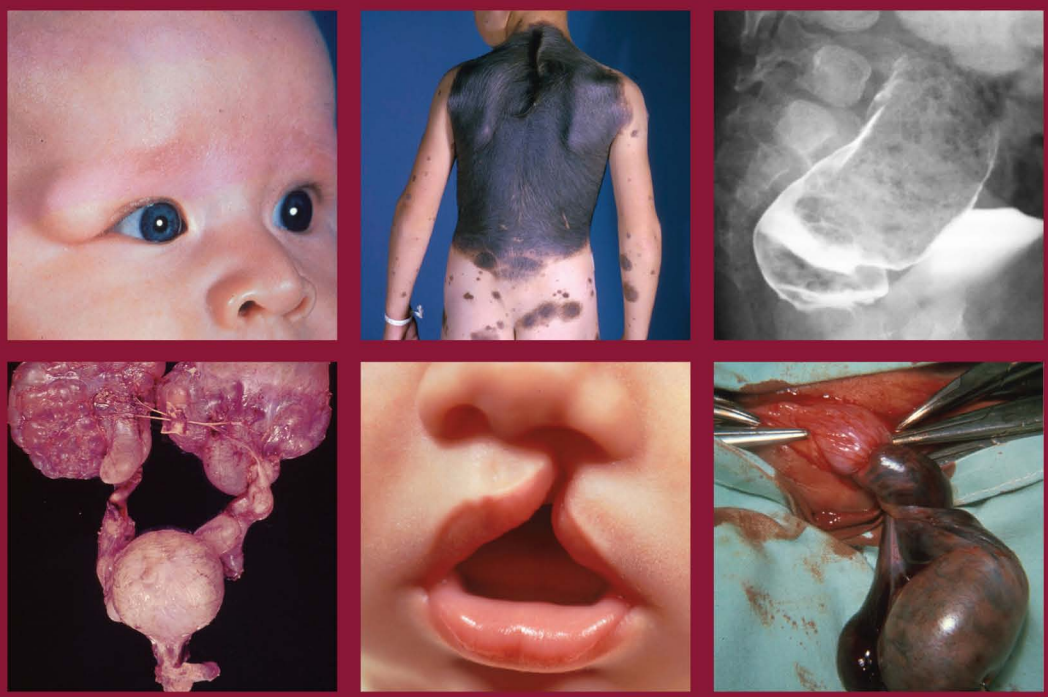
Paediatric Surgical Diagnosis

Atlas of Disorders of Surgical Significance

SECOND EDITION



Edited by
Spencer W. Beasley, John Hutson, Mark Stringer,
Sebastian K. King and Warwick J. Teague

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Foreword to the first edition

Paediatric surgery is age-related general surgery in a broad sense, involving many specialty fields. This book represents the essential elements of the multifaceted nature of the surgery of infancy and childhood.

Three very experienced individuals with well-recognised expertise in clinical care, education and research wrote this book. They have worked their entire careers in one of the most important children's centres in the world, so this book brings with it a level of authority which is unmatched.

The authors' goal was to develop an illustrated guide to diagnosis of the important paediatric surgical conditions. The common, the occasional and some rare conditions are all here. Although it would be virtually impossible to include every conceivable rare disorder, the authors come close; and certainly every condition likely to be encountered in a practitioner's professional life time is presented including entities seen mainly in third world countries. There is a great need for a book of this nature, which teaches the basics that trainees are frequently not taught today as high-tech medicine is emphasised. The emphasis is on primary care and additional management geared for medical students, trainees and practitioners in paediatrics, obstetrics and various surgical specialities, as well as nurses who might need a quick reference for diagnosis.

The particularly valuable aspect of this pictorial text is that it stresses physical diagnosis illustrated by beautiful photographs, x-rays and other imaging studies. In addition to marvellous illustrations, good descriptive legends and a concise text present a list of differential diagnoses and at least one reliable approach to confirmatory studies where appropriate.

No attempt is made to be exhaustive in the treatment of any disorder, which makes this book particularly useful for the intended audience around the world. The authors advise their readers to use this book in conjunction with a standard text for comprehensive coverage, but it would certainly stand alone as a visual guide to rapid diagnosis of almost any paediatric surgical condition likely to be seen in routine practice. This book is well organised into sections, which are comprehensively treated. In addition to conditions usually treated by general paediatric surgeons, those treated by ophthalmologists, neurosurgeons, urologists, otolaryngologists, orthopaedists and plastic surgeons are covered. Neonatal as well as acquired disorders are included. A book like this could only have come from the life's work of three such experienced paediatric surgeons.

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Foreword to the second edition

This second edition of *Atlas of Paediatric Surgery,* now titled *Paediatric Surgical Diagnosis: Atlas of Disorders of Surgical Significance*, is a superbly illustrated and well-organized guide to most common anomalies in children's surgery as well as many that are uncommon. While this text is not intended to be encyclopaedic in scope, it does present in considerable depth the broad spectrum of general and thoracic paediatric surgery, as well as offer an important perspective to any children's provider in paediatric urology, head and neck surgery, ophthalmology, orthopaedics, neurosurgery and, indeed, all of children's surgery. The emphasis is on providing photographs and illustrations to assist in the recognition, clinical understanding and multidisciplinary management of the various abnormalities. This edition builds on the first; it is not only beautifully photographed and illustrated, but it is improved in the organization and clarity of the supporting text. Taken together, the succinct narratives and lucid visual presentations make the text useful to a broad audience. Children's surgery encompasses such diverse pathology that collection of this information, and particularly these photographs, would take most individuals several careers to compile. The authors have done this for the reader with emphasis on diagnostic information including radiographic and other imaging, as well as intraoperative photographs. The presentation is uniquely informative in aggregate.

The atlas is edited by five of the most senior and well recognized children's surgeons in the world. All are experts and bring a wealth of clinical experience to this work. The authors have added new information in this edition, particularly related to antenatal diagnosis and treatment. While *in-utero* assessment and intervention were possible previously, these have become a standard aspect of care in the time interval since the first edition was published. These areas are addressed in chapter one, while the second chapter is dedicated to major congenital anomalies which would be symptomatic or apparent in the neonatal period. Together these chapters will be quite useful to any who wish to understand newborn surgical anomalies. The introductory chapters are followed by anatomical and organ system presentations, concluding with trauma and gynaecological conditions. The overall focus is on visual clarity and a pragmatic bedside clinical approach to these children with surgical needs. The atlas is designed not as a comprehensive surgical reference text or operating atlas, but as a companion or adjunct to such standard presentations, adding a depth of understanding and visual diagnostic clarity that is unique. It will be a valuable addition to the libraries of institutions and individuals who care for children with surgical needs. This includes not just general and thoracic paediatric surgeons but all children's surgeons, as well as other allied health professionals, those who are learning about these patients, including residents, fellows and students, and indeed anyone with an interest in the surgical problems of infants and children.

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Acknowledgements

ACKNOWLEDGEMENTS FOR THE FIRST EDITION

No one surgeon, not even one in the busiest clinical practice, could collect the range and variety of conditions displayed in this atlas within a lifetime. Therefore, despite fairly extensive collections of our own, we have been heavily reliant on the contributions of a number of colleagues. Many of these have gone to considerable effort to provide us with as comprehensive a coverage of the specialty as possible, and to them we are extremely grateful. They include: Alex Auldist, John Barnett, Don Cameron, Tony Catto-Smith, Bill Cole, David Croaker, Paddy Dewan, Bob Dickens, James Elder, Roger Hall, David James, Peter Jones, Anne Kosloske, Julian Keogh, Geoff Klug, Neil McMullin, Azad Najmaldin, Kevin Pringle, T.M. Ramanaujam, Barry Shandling, Errol Simpson, Arnold Smith, Durham Smith, John Solomon, Douglas Stephens, Keith Stokes, Russell Taylor, Roger Voigt, Alan and Susie Woodward. Ramanujam, in particular went to extraordinary lengths to provide us with a fine series of slides of the most bizarre and rare conditions. We are grateful to our many registrars who have had to organise the photography of many of the lesions.

Slides of interesting cases often are passed between colleagues and, with time, their origin may become obscure. It is quite possible that a number of illustrations included in this atlas have not been acknowledged appropriately. To those who have the original source of these illustrations, we are truly grateful, despite their anonymity.

An enormous contribution has been made by staff of the Educational Resource Centre of The Royal Children' Hospital, Melbourne, who have been happy to bear the burden of copying (and maintaining in order) a large number of slides, and to produce a number of line drawings. Their full cooperation in this project over many months has made the editors' task much easier.

The secretarial staff of the Department of Surgery, Elizabeth Vorrath and Judith Hayes, now well used to the rigours of medical manuscripts, have confirmed their mastery of complex, numerous and ever-changing legends. Their efficiency and good humour has obscured any frustrations they might have had.

Our ever-patient families have provided the support that has allowed us to devote consecutive long weekends to the project. We are happy to return the time to them in full now it is completed.

Finally, we are grateful to Peter Altman who encouraged us to embark on the project and who has provided helpful guidance throughout. The professional work of Chapman and Hall has been a major factor contributing to this work, which we hope will be of use to paediatric surgical aspirants and teachers for many years to come.

ACKNOWLEDGEMENTS FOR THE SECOND EDITION

Once again we are indebted to The Royal Children's Hospital Creative Studio (previously Educational Resource Centre) for their expert assistance in taking many of the clinical photographs as well as dealing with the artwork and digital file management.

We thank Shirley D'Cruz, Personal Assistant to Professor John Hutson, who worked tirelessly to prepare the base texts and image legends for revision. Shirley then shouldered the

Fig. 1 The authors (from left to right): Spencer Beasley, Mark Stringer, Warwick Teague, Sebastian King and John Hutson.

lion's share of the responsibility for ensuring version control throughout the complex task of revising the text and legends, and managing the nuances and needs of five editors and a publisher across three time zones.

Many of the original photographs provided in the first edition have been retained. In addition, we have included photographs and images provided by the listed contributors to this edition.

The staff of CRC Press throughout have been wonderfully patient and understanding of us, particularly in the final stages where the actual layout and accuracy of the figures and their legends has become important. From Cherry Allen, to Peter Beynon and Paul Bennett, the meticulous care they have taken assembling a substantial file of figures, and their detailed understanding of what we have tried to achieve, has made our work so much easier.

Creating an atlas of this magnitude takes an enormous amount of time: this time is often at the end of an already long day, and it is time that otherwise would have been spent with our wives and families. So it is to Christy, Susan, Alice, Charlotte and Kirsty, and our respective children, who have had to tolerate and forgive us our absences during the preparation of this book, that we are so grateful.

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Introduction

More than any other specialty, with the possible exception of dermatology, paediatric surgery lends itself to an illustrated guide to diagnosis. In the neonate, the dramatic appearance of exomphalos, gastroschisis, bladder exstrophy and prune belly syndrome is obvious. Anorectal malformations present with a spectrum of features, some of which are quite subtle, but which can be demonstrated with careful clinical examination. Even some internal lesions, such as volvulus, meconium ileus and bowel atresia, have external features, such as abdominal distension. The majority of orthopaedic deformities and inguinoscrotal lesions are diagnosed entirely on clinical grounds, most of which can be illustrated clearly on photography. Likewise, abnormalities and lesions of the head and neck, which are common in this age group, are usually superficial or structural, enabling easy clinical diagnosis. The relatively obscure areas of the thorax and urinary tract may have few or vague clinical features, but become apparent on appropriate radiological or other types of imaging.

This illustrated guide to the diagnosis of paediatric surgical disorders sets out to cover the broad spectrum of abnormalities encountered in this specialty. Although we have concentrated on the common lesions, we have deliberately included some extremely rare conditions to highlight the enormous variation that one may encounter in a specialty that includes bizarre congenital abnormalities.

The first chapter focuses on antenatal diagnosis, in recognition of the fact that nowadays most structural congenital abnormalities are diagnosed antenatally, and the paediatric surgeon becomes involved in the care of the unborn infant and its family well before birth. In this chapter we have deliberately included the type of ultrasound images encountered by paediatric surgeons in their everyday practice rather than concentrating on some of the recently introduced but not always widely available 'state of the art' imaging techniques. The second chapter deals with major neonatal abnormalities that are apparent at, or shortly after, birth; some will have been diagnosed several months prior to birth. Chapters 3 to 9 deal with the regions of the body sequentially, working from the head and neck, through the trunk, to the limbs. Abnormalities of the respiratory, gastrointestinal and urinary systems may be associated with a variety of external clinical manifestations but, more often than not, the definitive diagnosis is made only after specialised radiological investigation or at operation. Consequently, the operative views illustrate those conditions in which a diagnosis is made at surgery, or where the operative appearance is characteristic and clarifies the diagnosis. No attempt has been made to include details of operative technique. In the chapter on trauma, which covers all systems, special emphasis is given to non-accidental injury and in particular sexual abuse, as this is an area of considerable importance, and the clinician must be aware of the relevant features.

There are three major limitations to any pictorial guide to diagnosis. First, given the limitations of length, it is impossible to include all conditions or their variations. For example, the sections on anorectal malformations or disorders of sexual differentiation are extensive but not comprehensive: they could be vastly expanded, but for the sake of a relatively concise book this is not feasible. Second, many well-recognised conditions occur extremely rarely and, even in a large institution, certain conditions may be seen only once every decade or so. If they are not captured on film at the time of presentation, and before their operative correction, it may take some time before another similar case presents. In this regard, the editors are grateful to the many contributors who have helped 'fill the gaps'. Third, some conditions have no external clinical or radiological features that lend themselves to photography, which means that they tend to be 'down-played' in a pictorial book of this type. This is not to ignore or deny their importance, but obviously those conditions that are easily demonstrated photographically will tend to gain greater prominence. For this reason, it is important that this book should be read in conjunction with a standard paediatric surgery text if comprehensive coverage of the specialty is required.

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Antenatal diagnosis

CHAPTER 1

Antenatal diagnosis of major congenital abnormalities in the fetus has become commonplace as a result of the increasing use and sophistication of antenatal ultrasonographic equipment. Initially, it was thought that the antenatal diagnosis of fetal abnormalities would lead to better treatment and an improved outcome, but so far, this expectation has only been partly fulfilled. Nevertheless, there is little doubt that perinatal treatment of several abnormalities is improved by their foreknowledge.

Between 18 and 21 weeks of gestation is regarded as the best time for the early detection of most fetal abnormalities, although if there is a previous history of fetal abnormalities (e.g. spina bifida) an ultrasound examination earlier in pregnancy may be indicated, with either repetition of the ultrasound scan at suitable intervals throughout the pregnancy or progression to more invasive tests, such as amniocentesis, chorionic villus and fetal blood sampling. Antenatal diagnosis of fetal abnormalities has identified a group of severely affected fetuses with complex lethal abnormalities which in the past never survived the pregnancy. Those with abnormalities that are less severe and who survive long enough to reach birth – and surgical attention – are already a selected group in whom good surgical results would be expected. The range of abnormalities detectable by antenatal ultrasound scanning includes anencephaly, spina bifida, hydrocephalus, encephalocele, cardiac abnormalities, urinary tract obstruction, congenital lung malformations, congenital diaphragmatic hernia, ovarian cysts, ventral abdominal wall defects, duodenal atresia and gross skeletal abnormalities.

For a time it was hoped that early antenatal recognition of some of these conditions (e.g. congenital diaphragmatic hernia, hydrocephalus and urinary tract obstruction) would allow intrauterine fetal surgery to prevent ongoing or secondary injury to the fetus, but so far the results of fetal surgery in all but a few highly selected conditions have been disappointing. Perhaps the main value of antenatal diagnosis is that the affected infant can be delivered at a tertiary institution and the appropriate treatment initiated at birth. This may avoid some of the problems of neonatal transport and of delayed diagnosis.

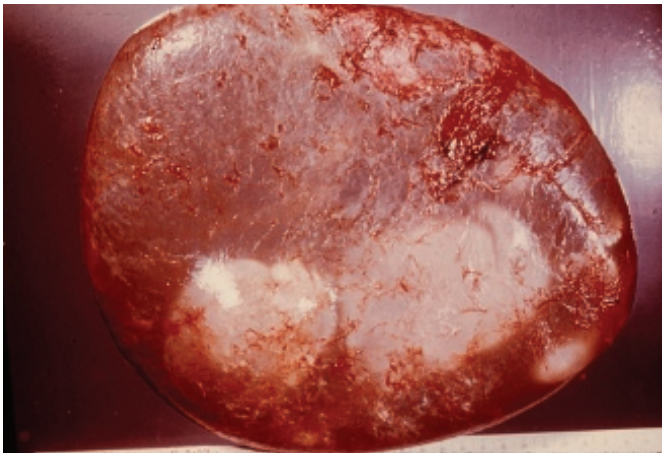


Fig. 1.1 An abortus with an intact amniotic sac containing a fetus within it. At least 10% of pregnancies abort in the embryonic stage (the first 8 weeks of gestation), mostly from chromosomal anomalies or gross malformations.

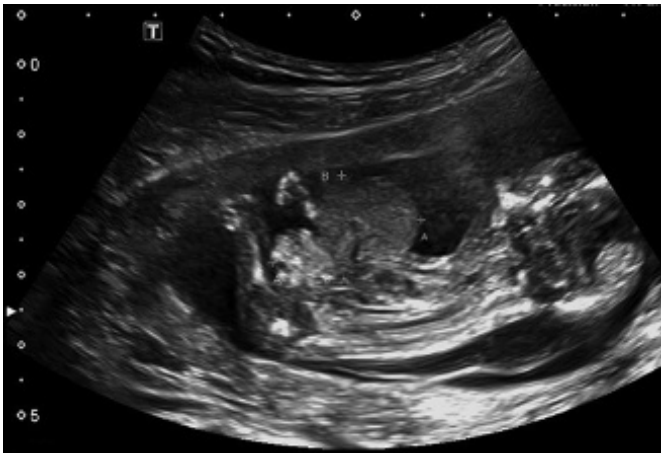


Fig. 1.2 Exomphalos in a 12-week fetus showing prolapse of the liver into the sac.

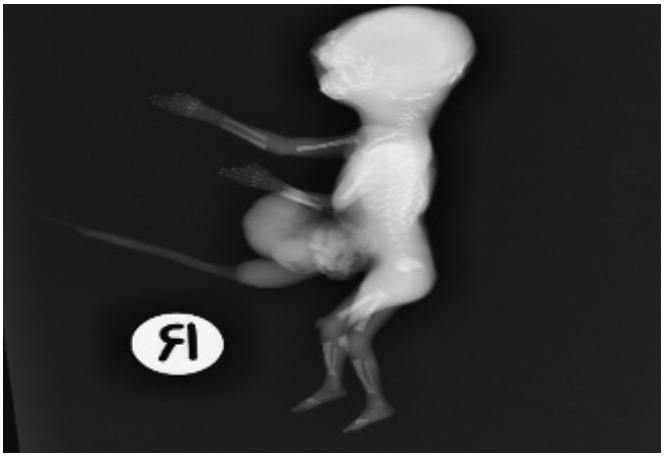


Fig. 1.3 Exomphalos in an aborted fetus demonstrated on postmortem babygram. It displays the relatively small size of the abdomen compared with the volume of bowel and liver in the sac. This illustrates why return of the sac contents into the abdominal cavity after birth can be difficult.

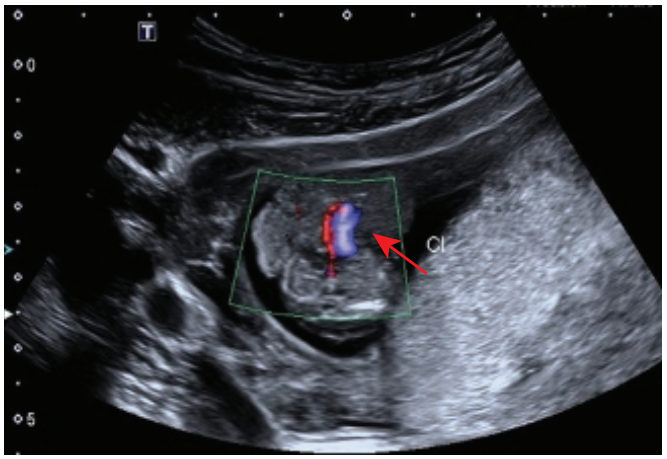


Fig. 1.4 Exomphalos in a fetus with trisomy 13. Doppler flow is seen in the umbilical cord vessels (arrow).

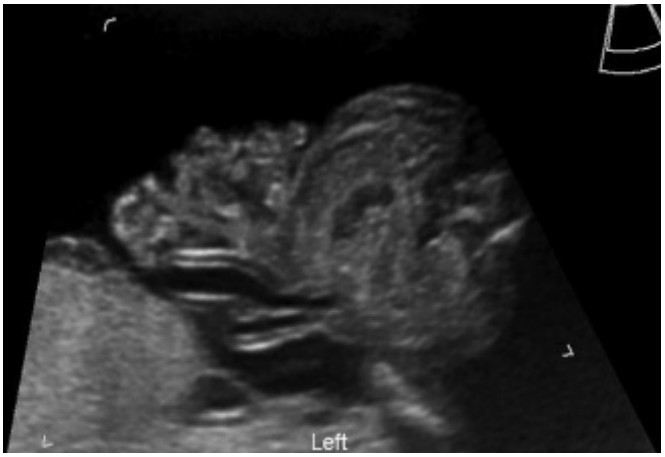


Fig. 1.5 Gastroschisis. Multiple loops of bowel have extruded through a defect in the anterior abdominal wall.

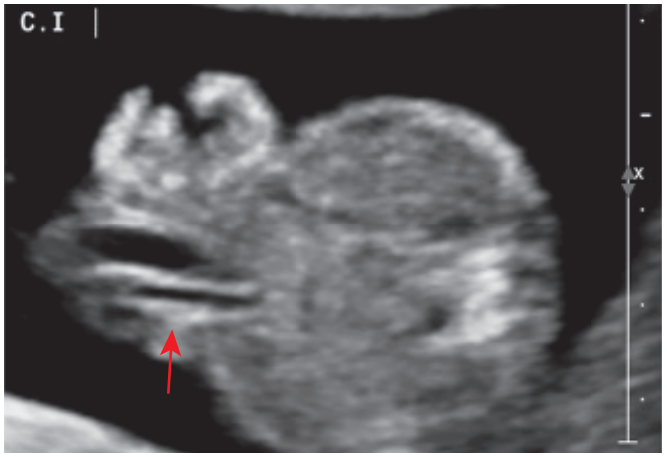


Fig. 1.6 Antenatal scan at 16 weeks' gestation showing trunk and umbilical cord (arrow) with bowel loops protruding just to the right of the attachment of the umbilical cord.

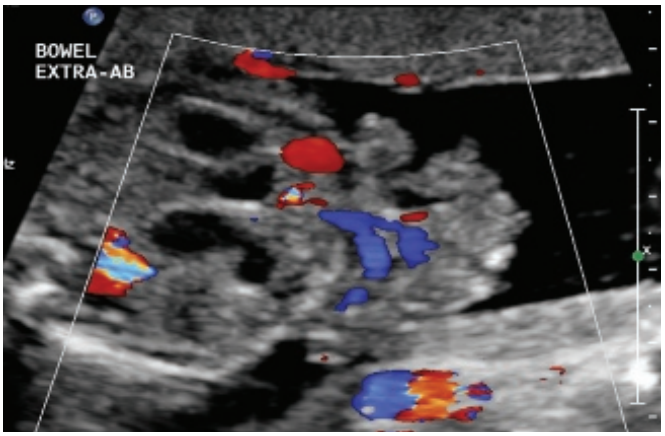


Fig. 1.7 Colour Doppler ultrasound scan in a 32-week gestation fetus with gastroschisis showing blood flow in the extra-abdominal mesenteric vessels.

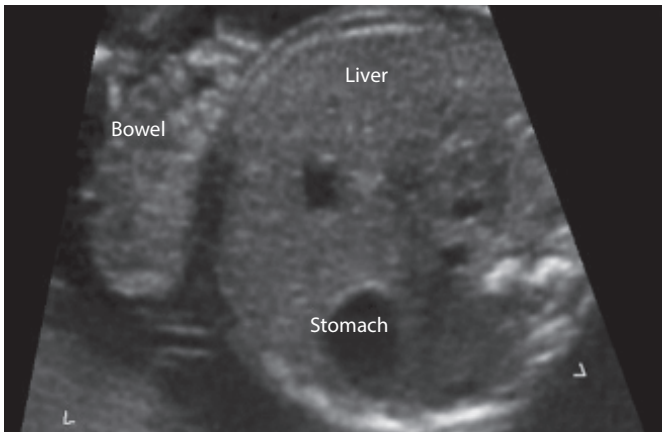


Fig. 1.8 Gastroschisis. Transverse abdominal ultrasound scan of a 19-week gestation fetus showing stomach and liver in the abdomen and extruded bowel.

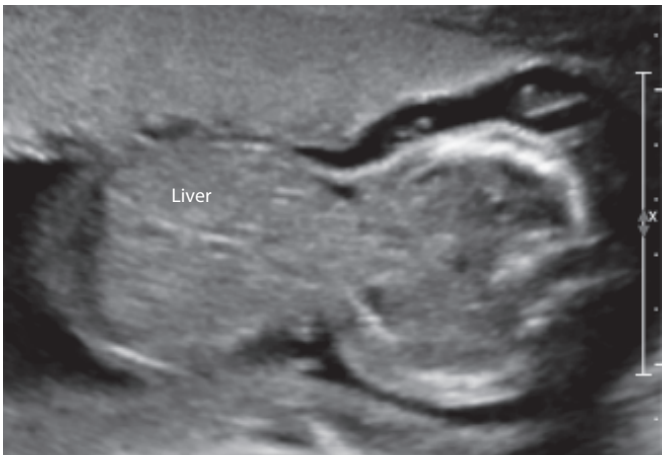


Fig. 1.9 A giant exomphalos containing the liver in a 16-week gestation fetus. Note the size of the exomphalos in comparison to the abdominal circumference.

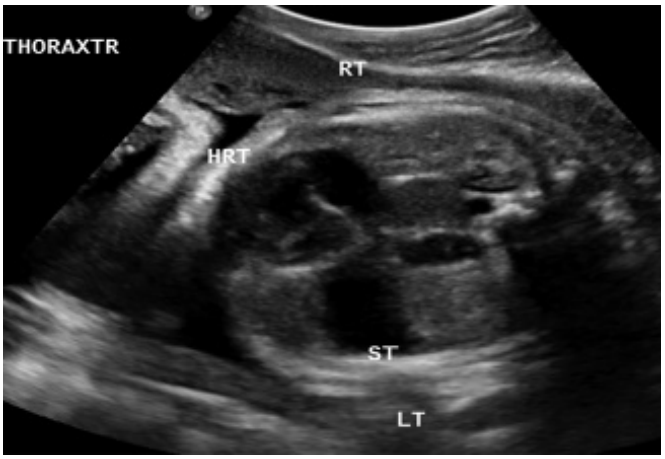


Fig. 1.10 Scan at 24 weeks' gestation showing a congenital diaphragmatic hernia with a fluid-filled cavity (stomach, ST) beside the heart (HRT).

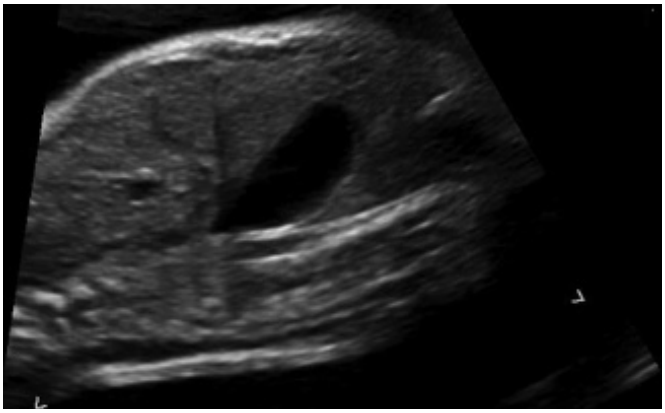


Fig. 1.11 Left-sided diaphragmatic hernia. Longitudinal scan of thorax showing a fluid-filled stomach in the thorax.

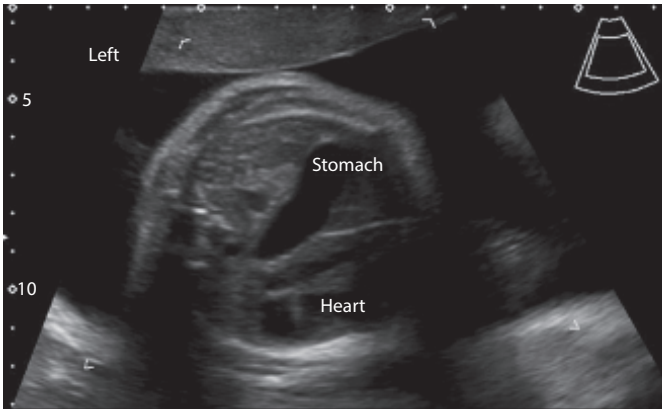


Fig. 1.12 Antenatal scan at 29 weeks' gestation showing a fluid-filled stomach beside the heart in a left-sided diaphragmatic hernia.

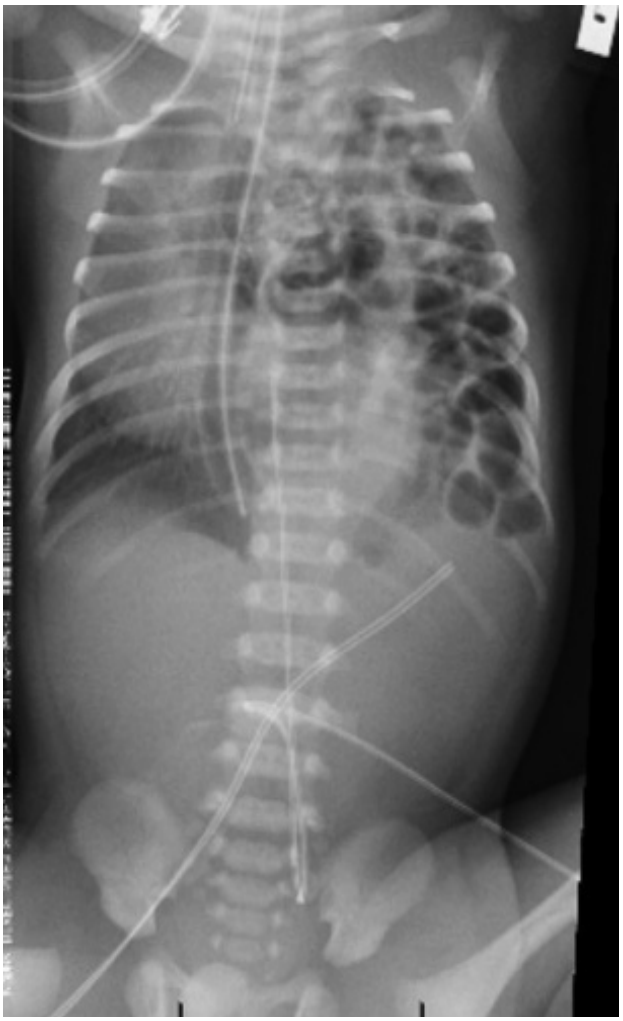


Fig. 1.13 First postnatal chest radiograph of the same infant as in **Fig. 1.12** confirming the diagnosis of a left-sided congenital diaphragmatic hernia.

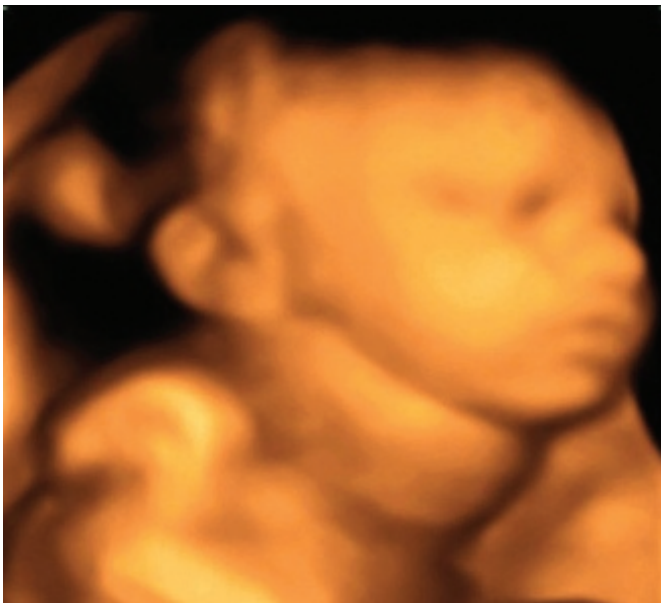


Fig. 1.14 4D ultrasound scan showing a cervical lymphatic malformation. This baby was delivered by the ex-utero intrapartum treatment (EXIT) procedure.

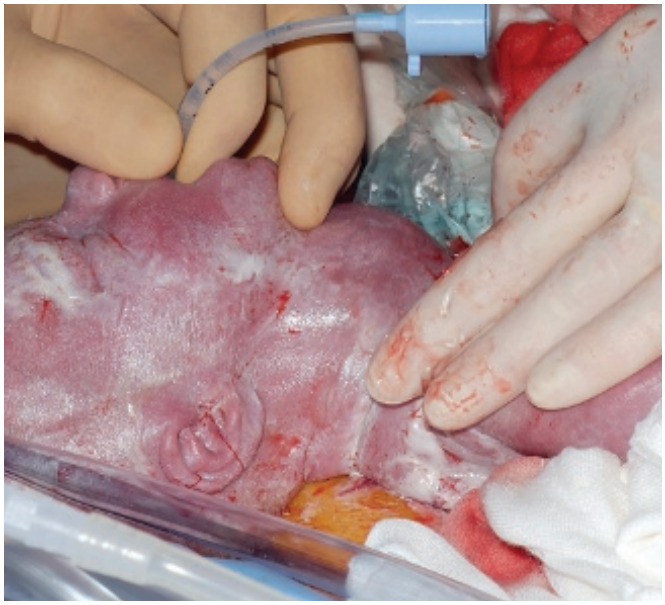


Fig. 1.15 EXIT procedure on the same infant with a cervical mass lesion.

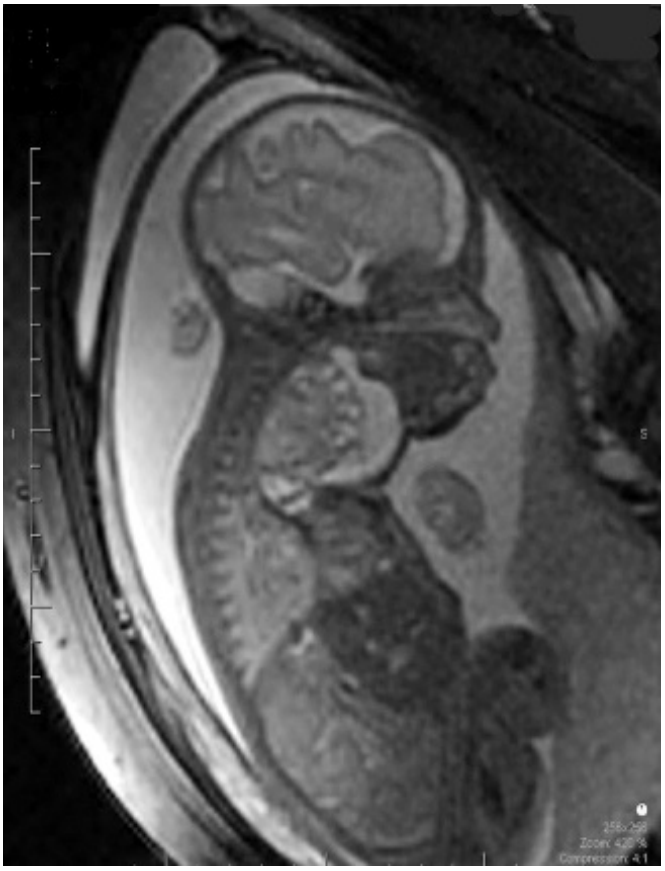


Fig. 1.16 A large cervical teratoma on antenatal MRI.

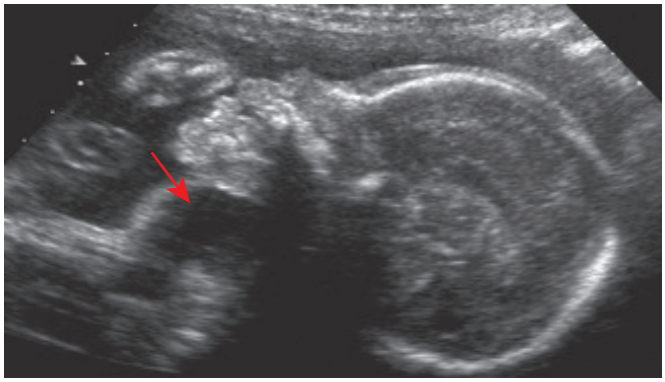


Fig. 1.17 Multicystic lymphatic malformation in the neck of a fetus (arrow) seen on ultrasound scan.

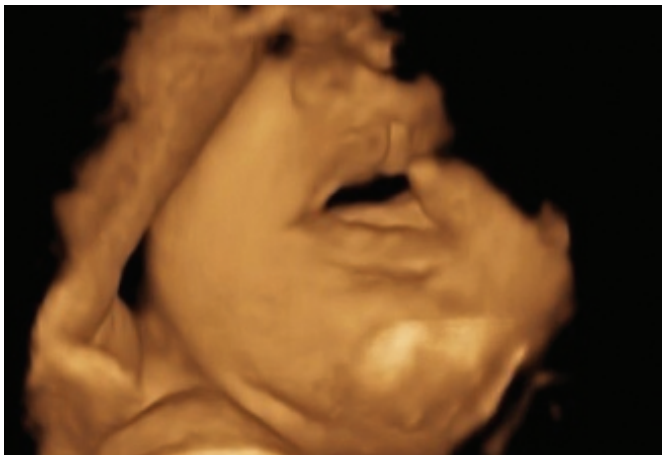


Fig. 1.18 Unilateral cleft lip and palate on 3D ultrasound scan.

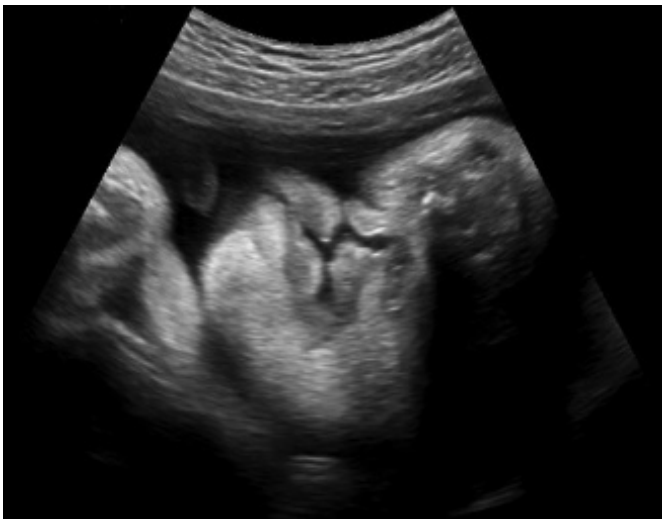


Fig. 1.19 Same fetus as in **Fig. 1.18** on 2D ultrasound scanning showing the cleft lip and palate.

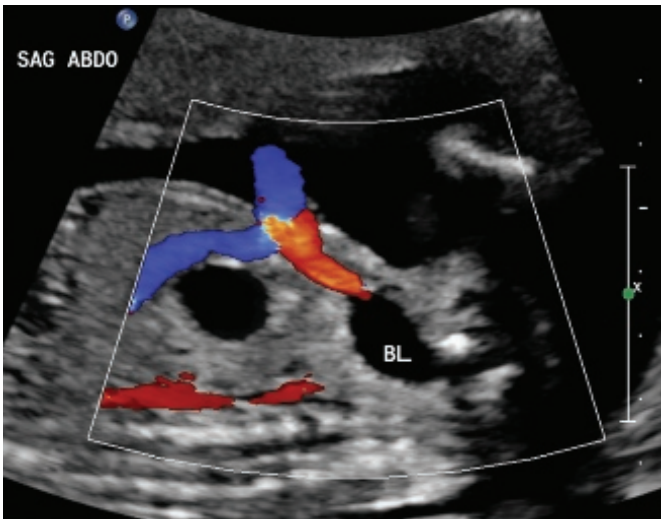


Fig. 1.20 Colour Doppler scan at 30 weeks' gestation showing an intra-abdominal fluid-filled cavity separate from the bladder, consistent with a duplication cyst.

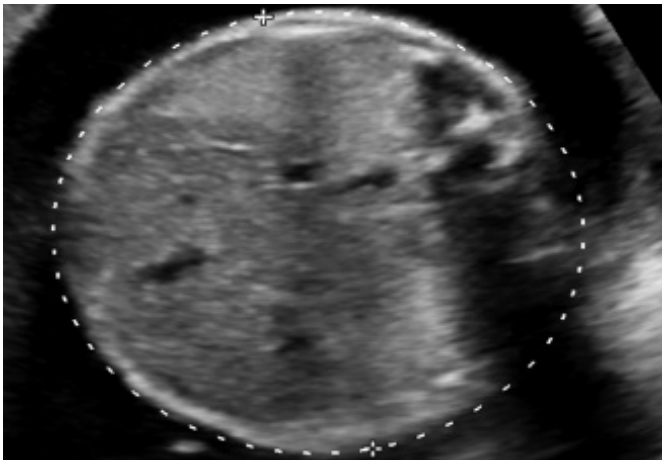
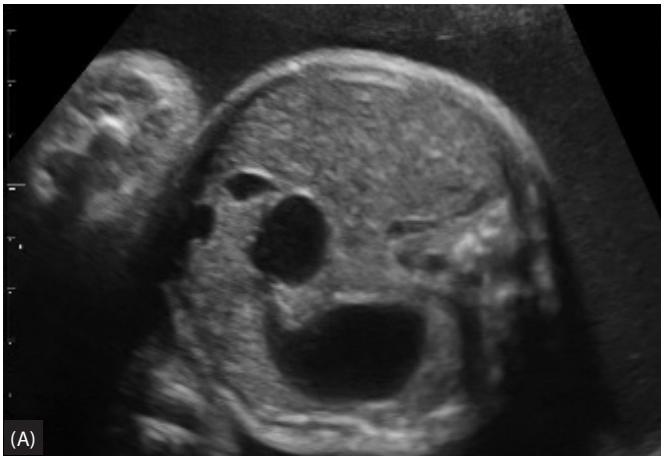
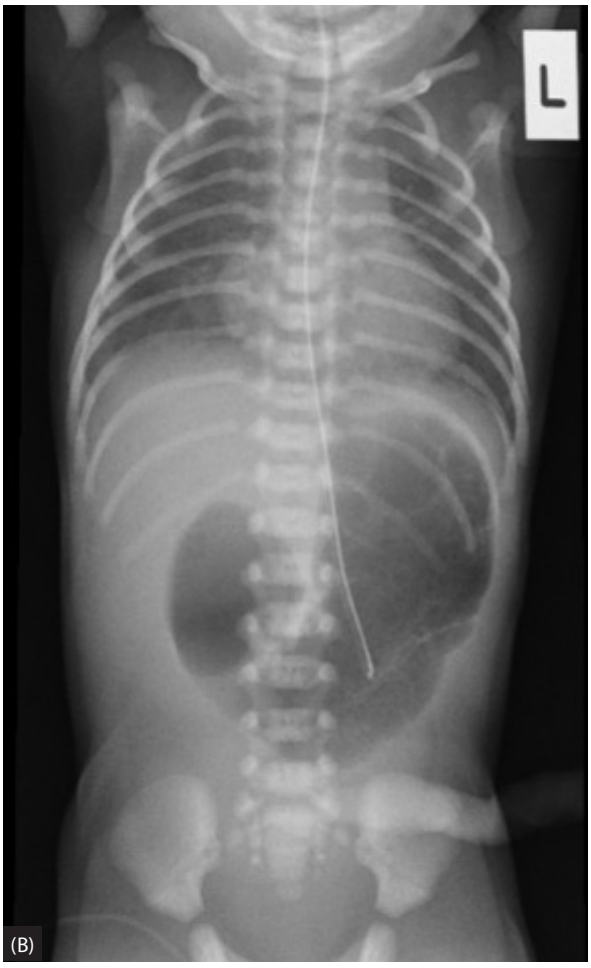


Fig. 1.21 Scan of the trunk at the level of the liver at 32 weeks' gestation, showing no stomach within the abdomen. If there is a small or absent stomach, particularly with polyhydramnios, oesophageal atresia is a possibility.





Figs. 1.22A, B (A) Antenatal ultrasound scan through the trunk showing showing two fluid-filled cavities in the upper abdomen, consistent with duodenal atresia. (B) Same baby at 3 hours after birth with classic double bubble on x-ray.

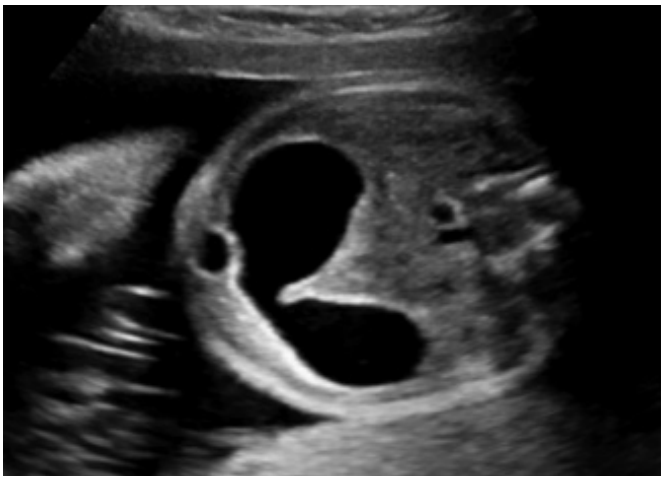


Fig. 1.23 Scan at 28 weeks' gestation showing a double-bubble sign in another fetus with duodenal atresia.

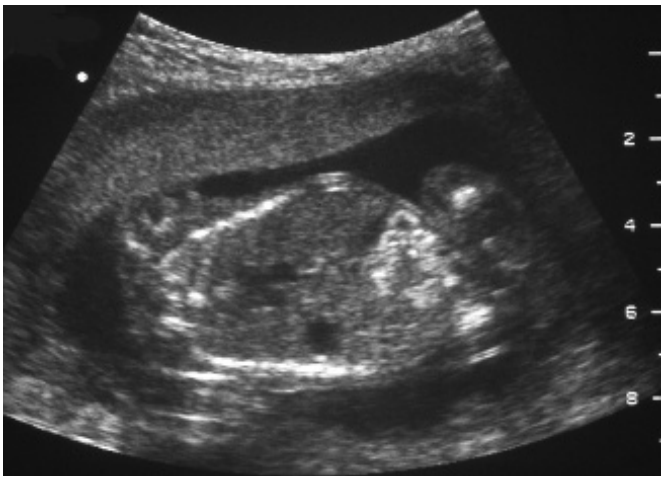


Fig. 1.24 Coronal scan of a fetus at 18 weeks showing echogenic bowel. Echogenic bowel may be seen in otherwise normal fetuses but can also be a marker for cystic fibrosis, intrauterine viral infection and aneuploidy.

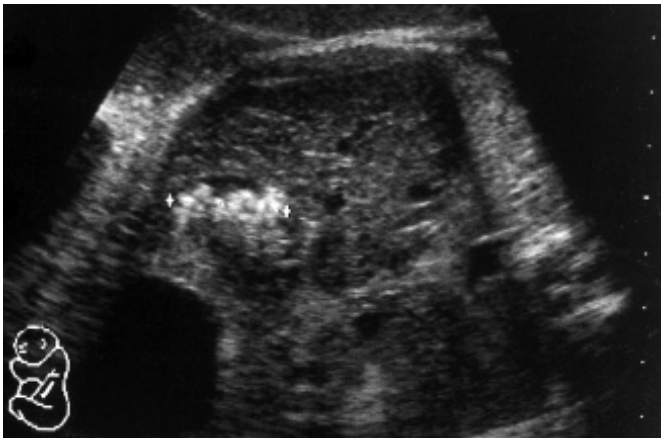


Fig. 1.25 Antenatal scan showing echogenic masses in the liver view, consistent with fetal gallstones or biliary sludge.

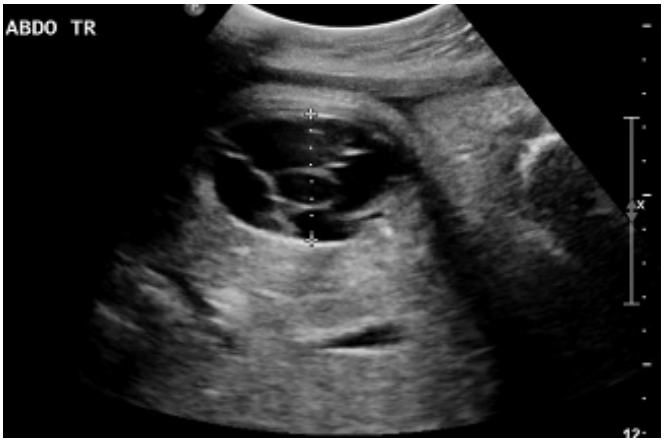


Fig. 1.26 Large multicystic ovary in a fetus at 34 weeks' gestation. Early postnatal follow-up is required.

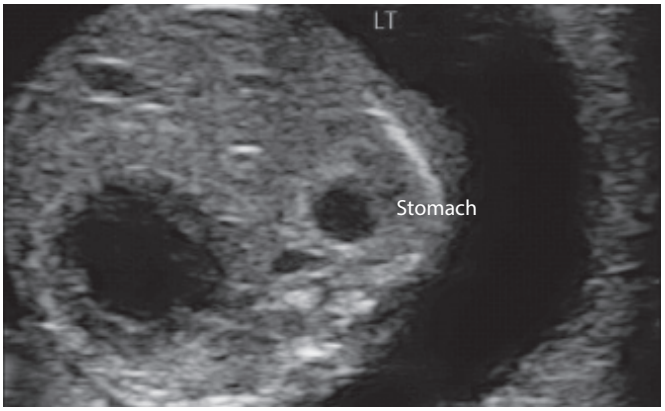


Fig. 1.27 Transverse ultrasound scan of the upper abdomen at 14 weeks' gestation showing a hepatic cyst.

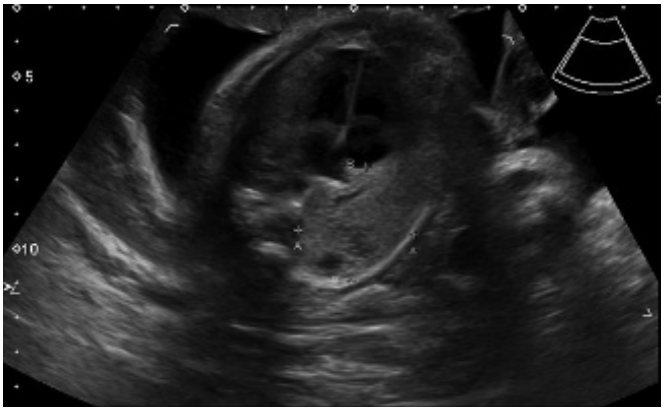


Fig. 1.28 Congenital pulmonary airway malformation (CPAM) involving the right lower lobe in a 30-week gestation fetus on transverse section.

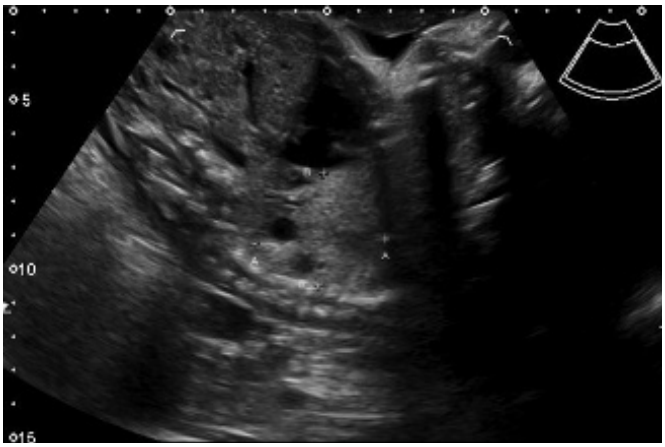


Fig. 1.29 CPAM in the right lower lobe on a longitudinal ultrasound scan of the fetus.

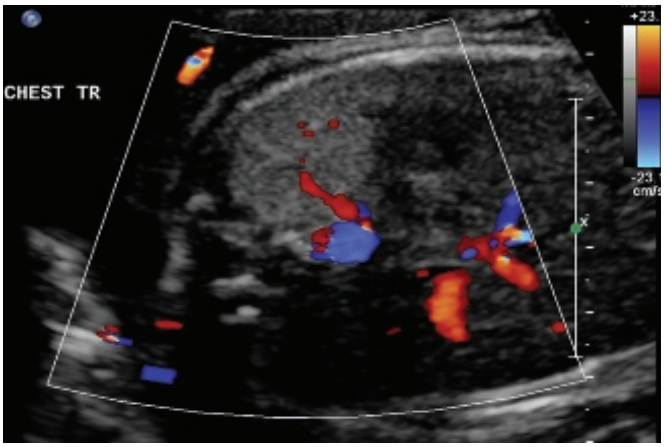


Fig. 1.30 Likely extralobar pulmonary sequestration as evident by the large vessel running directly off the aorta.

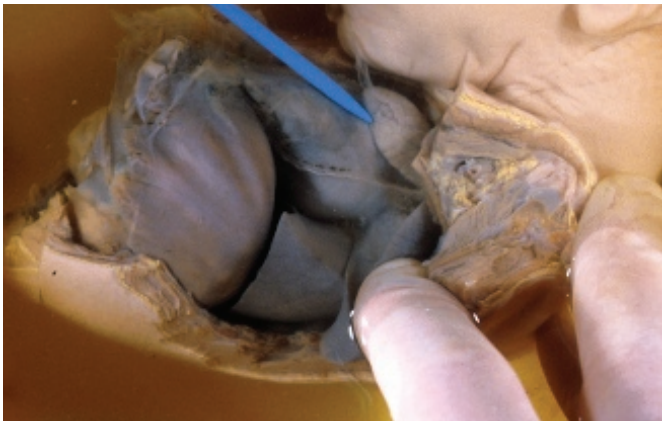


Fig. 1.31 The fetal thymus is a large structure, here exposed after removal of the left chest wall. The phrenic nerve can be seen running behind it, over the surface of the pericardium.

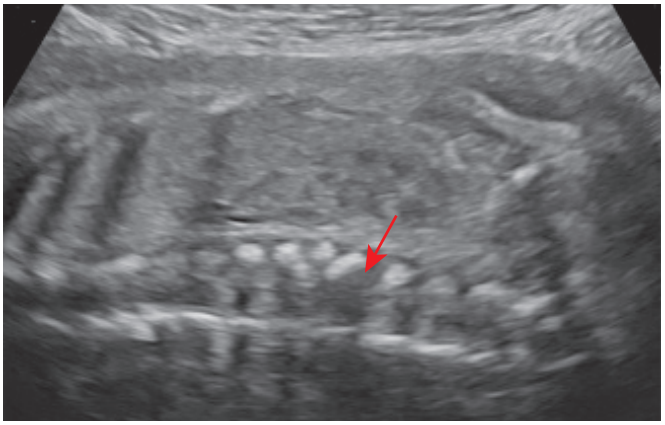
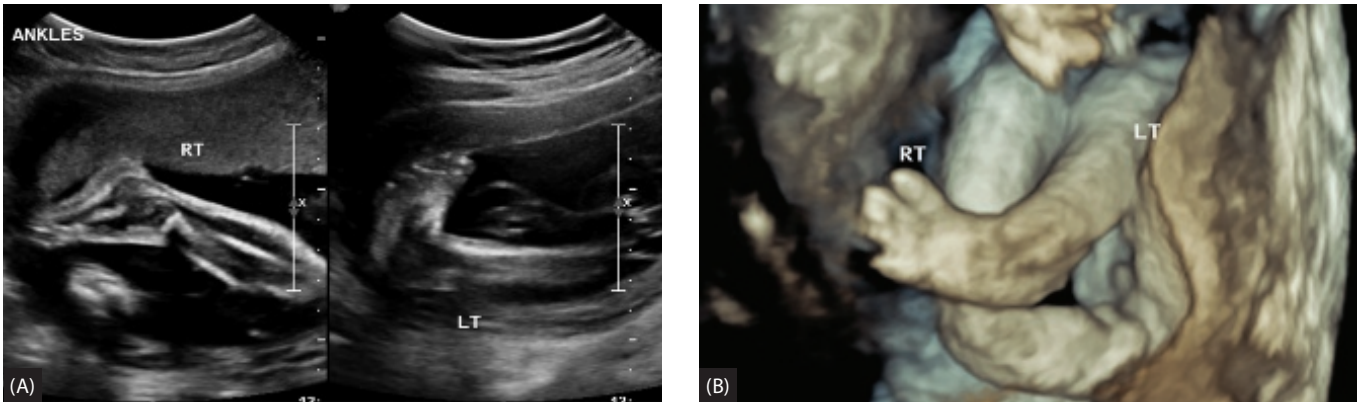


Fig. 1.32 Hemivertebrae (arrow) visible on a 28-week antenatal ultrasound scan.



Figs. 1.33A, B (A) Ultrasound scan at 20 weeks' gestation showing both feet, with a normal right foot and club foot on the left. (B) 3D reconstruction of the same fetus at 24 weeks clearly showing bilateral talipes equinovarus.

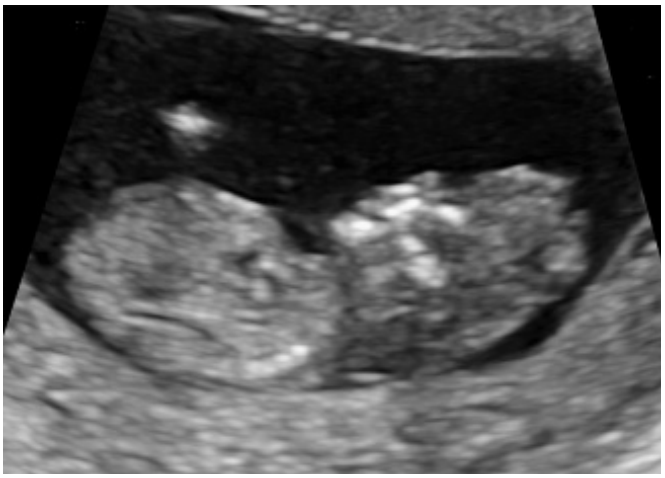


Fig. 1.34 Twelve-week' gestation ultrasound scan showing the head and face of a fetus with anencephaly.

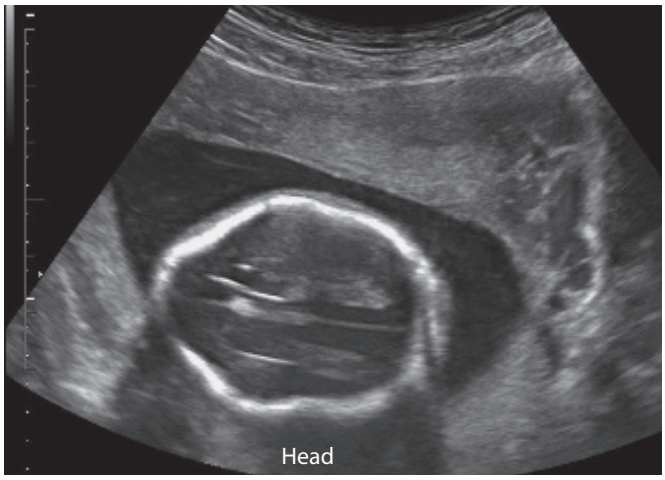


Fig. 1.35 Spina bifida was diagnosed in this fetus at 18 weeks' gestation. The lemon-shaped head is evident on ultrasonography.

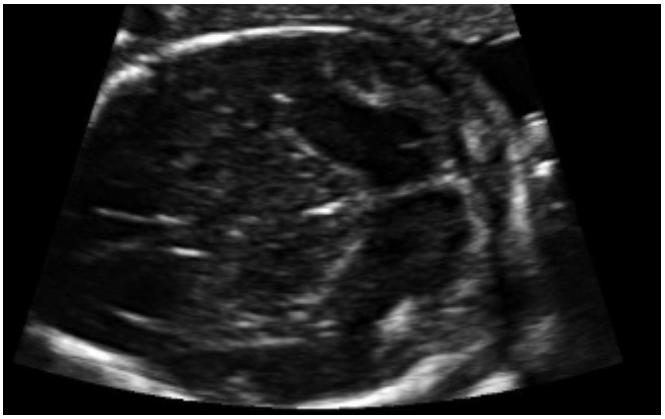


Fig. 1.36 Posterior view of the fetal skull showing dilatation of the posterior horns of the lateral ventricles in hydrocephalus.

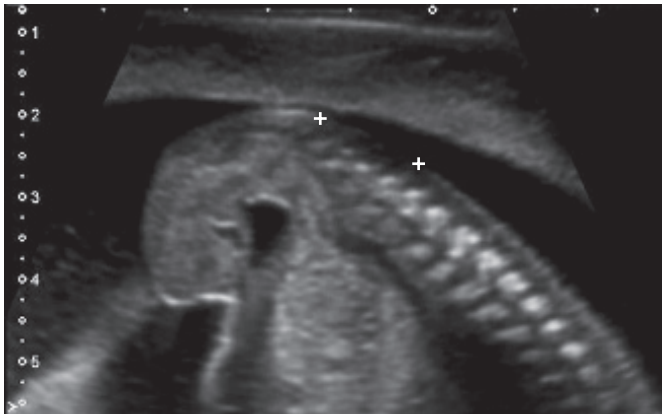


Fig. 1.37 Spina bifida of the lumbosacral spine on an antenatal scan showing an obvious gap in the dorsal arches (between the + markers).

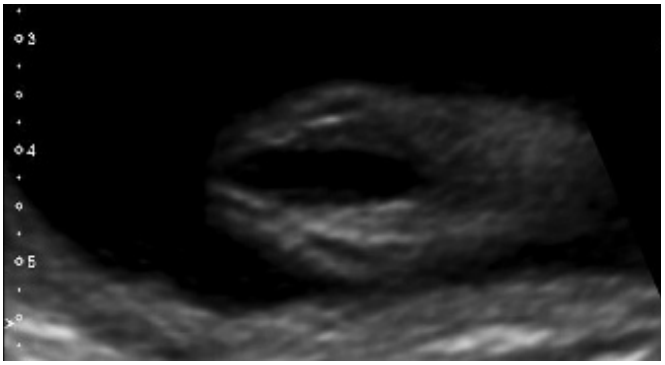


Fig. 1.38 Spina bifida showing a lumbosacral defect, which is probably myelomeningocele filled with cerebrospinal fluid (CSF).

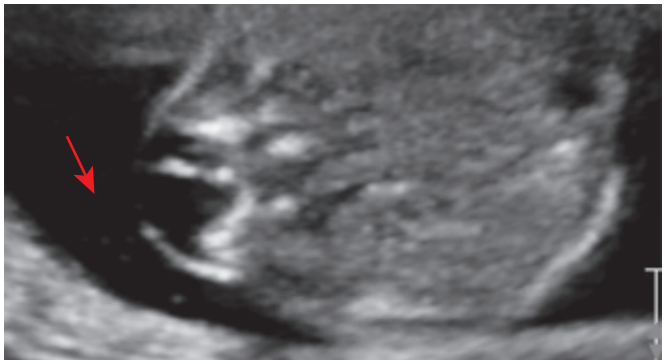


Fig. 1.39 Transverse scan of the trunk at 30 weeks' gestation showing the myelomeningocele containing CSF (arrow).

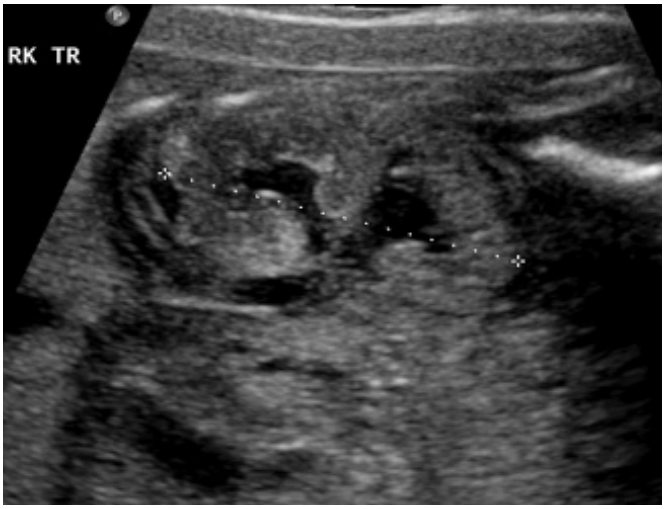


Fig. 1.40 Ultrasound scan at 28 weeks' gestation showing a duplex right kidney.

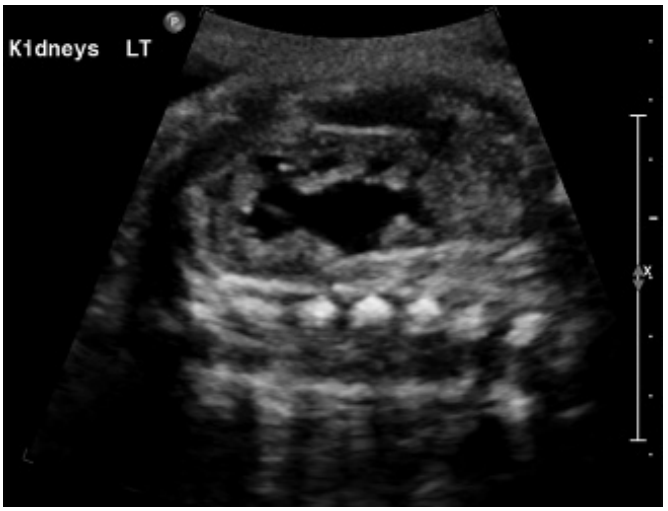


Fig. 1.41 Pelvicalyceal dilatation in the left kidney evident at 28 weeks' gestation.

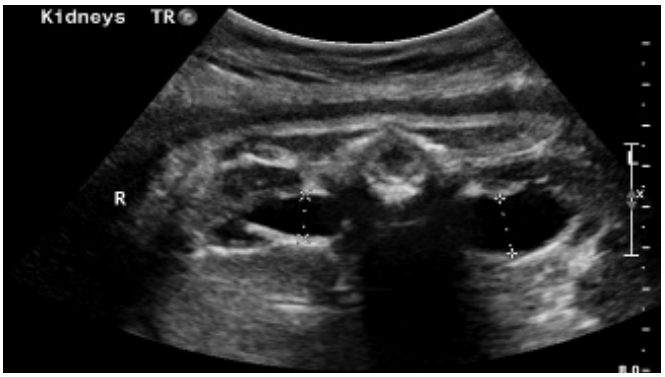


Fig. 1.42 Transverse scan of a 32-week fetus showing bilateral dilatation of the renal pelves. In this fetus posterior urethral valves must be considered and the gender, posterior urethra, thickness of the bladder wall and whether the bladder is emptying normally must also be evaluated.

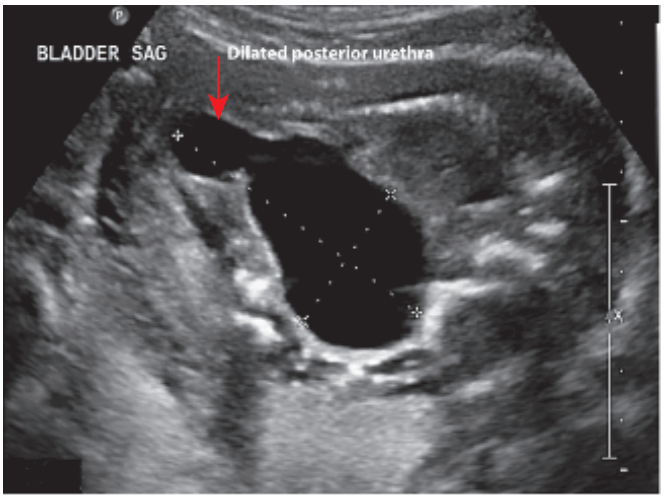


Fig. 1.43 Keyhole sign on an 18-week gestation ultrasound scan showing dilatation of both bladder and posterior urethra, consistent with a diagnosis of posterior urethral valves.

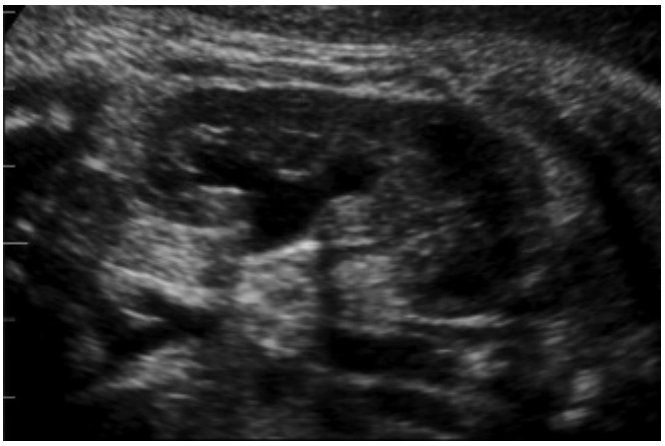


Fig. 1.44 Duplex left kidney with dilatation of the lower pole moiety.

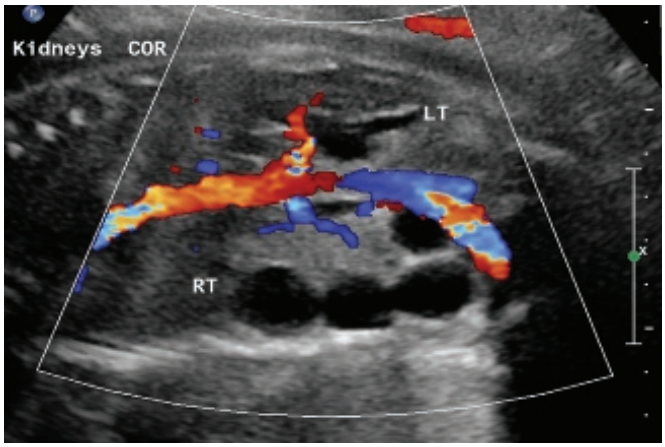


Fig. 1.45 Colour Doppler scan at 28 weeks' gestation showing both kidneys with some dilatation of the left kidney and multiple cysts in the right kidney, suggestive of a multicystic dysplastic kidney.

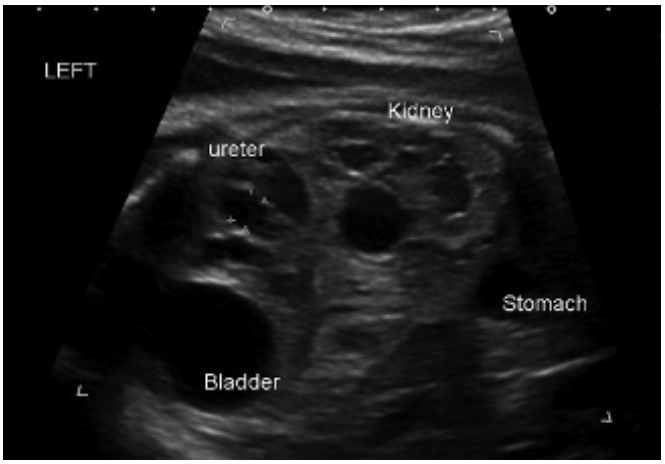


Fig. 1.46 Longitudinal view of the trunk showing hydroureteronephrosis and a full bladder in a fetus with posterior urethral valves.

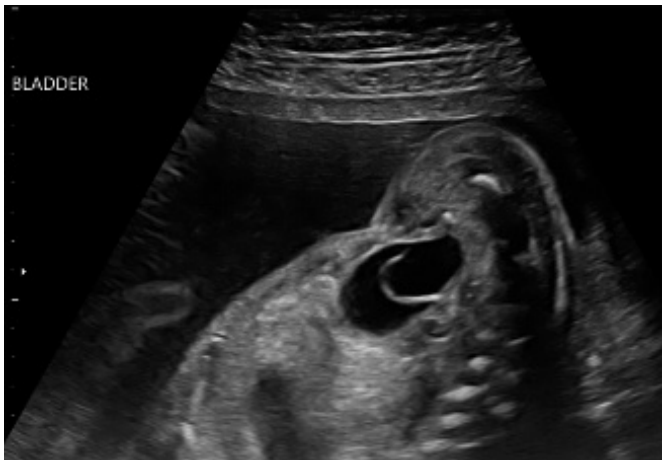


Fig. 1.47 Scan of the fetal pelvis showing a dilated bladder with a large ureterocele, which may prolapse into and block the urethra.

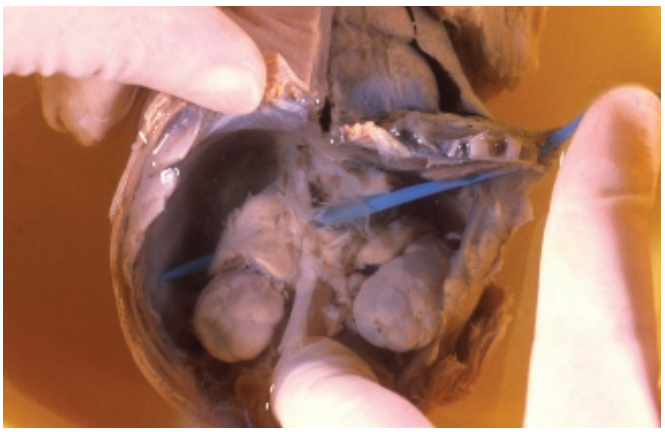


Fig. 1.48 The fetal adrenal is almost the same size as the fetal kidney. In this 16-week fetus, sectioned at the mid-abdominal level, the right adrenal dominates the right kidney on which it sits.

Major neonatal conditions

CHAPTER 2

Most major conditions in the neonate that require surgical correction are congenital, but a few (e.g. neonatal necrotising enterocolitis and pulmonary interstitial emphysema) are acquired. They often have external manifestations and are obvious at birth (e.g. gastroschisis and imperforate anus) or cause functional disturbance, which enables them to be diagnosed within the first few days (e.g. severe respiratory insufficiency in congenital diaphragmatic hernia and bilious vomiting in duodenal atresia).

VENTRAL ABDOMINAL WALL DEFECTS

The two most common major ventral abdominal wall defects are exomphalos and gastroschisis. In exomphalos, there is a large defect at the umbilicus with protrusion of abdominal viscera into a thin membranous sac, which is formed by amniotic membrane on the outside and peritoneum on the inside. In the vast majority of infants, the sac is intact irrespective of the mode of delivery (vaginal delivery versus caesarean section). At birth the contents, usually small bowel and liver, can be seen clearly through the translucent membrane but as desiccation of the membrane occurs, it becomes opaque. The size and shape of the defect vary considerably, and in very large defects (giant exomphalos), most of the intra-abdominal viscera are within the sac, and the abdomen appears scaphoid and of small volume. The skin of the ventral abdominal wall extends up the side of the sac to a variable degree. The umbilical arteries and umbilical vein can be seen traversing within the membrane, converging at the umbilical cord. Exomphalos is associated with other major congenital abnormalities, particularly cardiac, renal and chromosomal, in about 50% of cases. In Beckwith–Wiedemann syndrome, there is hemihypertrophy, macroglossia, visceromegaly and abnormal facies. It is essential to detect and treat accompanying hypoglycaemia.

In gastroschisis, the actual defect in the ventral abdominal wall is much smaller, and nearly always lies immediately to the right of the umbilicus. The umbilical vessels are unaffected and are situated to the left of the protruding bowel. There is no covering membrane. Much of the bowel and, frequently, the stomach herniates through the defect and the bowel and mesentery may be thickened, congested and oedematous with an overlying fibrin 'peel'. Gangrene of the herniated bowel may occur if the blood supply is occluded by the narrow opening or as a result of volvulus. Where this process has occurred well before birth, the necrotic bowel is resorbed, leaving an atresia. In gastroschisis, extraintestinal abnormalities are uncommon.

EXSTROPHY

Bladder exstrophy and cloacal exstrophy are major congenital abnormalities involving failure of the lower abdominal wall to close, exposing the mucosal surface of the bladder. Other features may include exomphalos, pubic diastasis, deficient urethra, unfused genitalia and rectal prolapse.

PRUNE BELLY SYNDROME

The infant, usually a boy, is born with a flat and wrinkled-looking abdomen and undescended testes. Prune belly syndrome may be due to transient intrauterine obstruction of urinary outflow, causing gross distension of the bladder and upper urinary tract, which in turn is the cause of abdominal distension. Another theory is that the condition is due to abnormal mesodermal development.

Following spontaneous relief of obstruction, the stretched and attenuated ventral abdominal wall looks shrivelled and wrinkled ('like a prune') but the dilatation of the urinary tract persists, often for many years.

ANORECTAL MALFORMATIONS

Anorectal malformations manifest as a range of appearances. In 'low' lesions in the male, a fistula communicates with the skin via an opening in the midline anterior to the normal position of the anus, either in the perineum, scrotum or ventral midline of the penis. 'High' lesions frequently have a communication with the urinary tract, most commonly through a rectourethral fistula, but occasionally directly into the bladder neck as a rectovesical fistula. 'High' anorectal malformations may occur without any fistula; this variant is most commonly seen in Down syndrome.

In the female, the number of external orifices in the perineum will give some indication of the level of the anomaly. Where there are three openings, namely urethral, vaginal and anal, the lesion is a 'low' one; when there are two obvious openings, namely urethral and vaginal, it is a high lesion; and when there is one opening, it is known as a cloacal abnormality. The exception to this general rule is in the case of a rectovestibular fistula, which may have the same external appearance as an anovestibular fistula. The clinical distinction can be made according to the direction in which a probe runs. In the former, it will tend to run deeply parallel to the vagina, whereas in the latter, it will run first in a posterior direction in a subcutaneous plane, before it turns deeply at the level of the anus.

A fistula opening on the perineal skin is easily identified when it transmits meconium or air, but the opening can be tiny and requires a careful search with magnification and good illumination. In high lesions in males, the presence of a rectourethral or rectovesical fistula can be inferred if air or meconium is passed per urethram. The fistula is usually confirmed by subsequent contrast studies (distal loopogram and cystourethrography), which show 'beaking' of the terminal rectum where the contrast enters the urinary tract.

The use of an 'invertogram' (or shoot-through lateral radiograph of the pelvis with the baby prone) at 12–24 hours of age, once gas reaches the distal bowel, may provide additional information on the level of the abnormality. The radiograph is taken in an exact lateral projection with the baby being placed prone over a padded wedge. Gas rises to the apex of the blind-ending rectum, and the level is related to various skeletal landmarks corresponding to the levator sling. There is less reliance on plain radiographs these days to determine the level of the anorectal malformation because clinical examination with or without a perineal ultrasound scan is often sufficient.

Neonates with an anorectal malformation require investigation for associated anomalies: an ultrasound scan of the urinary tract and spine and radiographs of the chest and spine. Some also need an echocardiogram and/or karyotyping. Those with 'high' lesions usually require management with a temporary diverting stoma and are further investigated at a later date by contrast studies ± endoscopy.

AMBIGUOUS GENITALIA (DISORDERS OF SEXUAL DEVELOPMENT [DSD])

Genitalia are considered ambiguous when one or more of the following features are present:

1. The phallus is too large for a clitoris or too small for a penis.
2. The urethral opening is proximal, near the labioscrotal (genital) folds.
3. The genital folds remain unfused, giving the appearance of labia or of a cleft scrotum.
4. The testes are either not descended or impalpable.

There is variation in the degree of abnormality that may be present, making gender assignment at birth hazardous. Prompt and accurate diagnosis is imperative because of the social implications for the parents of not being able to announce the gender of the baby, and the malformation may be the outward sign of the life-threatening condition congenital adrenal hyperplasia. When congenital adrenal hyperplasia occurs in females, the appearance of the external genitalia may cover a broad spectrum, suggestive more of either male or female. In the commonest autosomal recessive variety, deficiency of the adrenocortical enzyme 21-hydroxylase causes low cortisol levels and a compensatory increase in secretion of pituitary adrenocorticotrophic hormone, resulting in adrenal hyperplasia. The excessive androgens produced in females cause virilisation of the external genitalia but the internal anatomy is normal for a female. Investigations include estimation of serum electrolytes and blood glucose (to detect hyponatraemia, hyperkalaemia and hypoglycaemia), serum 17-hydroxyprogesterone and urinary pregnanetriol. Chromosomal analysis is mandatory and screening for the known genes involved in sex development is becoming more common. A urogenital sinogram or urethroscopy will show a masculinised urethra and the presence of a vagina and cervix. A pelvic ultrasound confirms the presence of a uterus, Fallopian tubes and ovaries.

Some infants are born with normal male chromosomes and testes but with insensitivity to androgens. A genetic abnormality in the androgen receptor system prevents the normal target tissues for androgens (Wolffian ducts and external genitalia) from responding to androgen stimulation. The abnormality may be partial or complete. In the complete form (complete androgen insensitivity syndrome, CAIS), the external genitalia are completely female in appearance. The clue to the gonads being testes in a phenotypic female is their discovery in inguinal herniae. The normal secretion and response to anti-Müllerian hormone (AMH) leads to regression of the Müllerian ducts, while failure to respond to androgens prevents the development of a vas deferens.

Disorders of sex development may be produced by dysplasia of one or both testes. When both testes are dysplastic, deficiency of androgen may cause incomplete virilisation, and deficiency of AMH may allow persistence of Müllerian duct structures. Usually, both testes are undescended because they are unable to secrete enough hormones to enable testicular descent. In the asymmetrical form of dysplastic testes (mixed gonadal dysgenesis), one gonad may be a testis that has descended with preservation of the ipsilateral Wolffian duct and local regression of the Müllerian duct. The more dysplastic testis usually remains in the abdomen and has failed to cause regression of the Müllerian duct or preservation of the Wolffian duct. Infants with mixed gonadal dysgenesis may appear to be males with severe hypospadias and one palpable testis. These dysplastic testes have an increased risk of malignant degeneration.

In severe hypospadias with undescended testes the phallus may be large enough to indicate that the infant is essentially male, and usually the chromosomal karyotype is normal male (46, XY). A contrast urethrogram will demonstrate a normal male urethra without a vagina or uterus. The testes may be impalpable (in the inguinal canal or abdomen) or palpable near the pubic tubercle, on one or both sides. Severe hypospadias with undescended testes has to be distinguished

from DSDs, such as severe virilising congenital adrenal hyperplasia or one of the causes of dysplastic testes.

When a newborn has ambiguous genitalia, clinical examination includes assessment of the external genitalia to determine:

1. The size of the phallus.
2. The degree of fusion of the inner genital folds to form a urethra.
3. The degree of outer genital fold fusion to form a scrotum, with its characteristically wrinkled skin.

The phallus should be lifted up to expose its ventral surface, and the genital folds spread apart to help identify the position of the urethral orifice. The skin should be carefully examined for evidence of excess pigmentation, which is common in congenital adrenal hyperplasia because of the excess melanin-stimulating hormone produced by the pituitary.

The next step is to determine the position of the gonads. Two palpable testes in the scrotum indicate that not only is the underlying sex of the child male, but also that there has been sufficient hormonal stimulation to cause testicular descent. If there are no testes palpable in the groins or labioscrotal folds, the presence of a uterus should be determined. A cervix may be palpable on rectal examination using the little finger. The presence of a uterus implies that there has been either absent or insufficient AMH.

A child with congenital adrenal hyperplasia may be vomiting, appear thin and be dehydrated, depending on the degree of salt loss and the time since birth. The nipples, skin creases and genitalia may be pigmented. Virilisation of the external genitalia is proportional to the degree of urogenital fusion. The only clue to the true sex of the child may be the absence of testes. Early and accurate diagnosis is important because:

1. Urgent medical treatment may be required if the infant has congenital adrenal hyperplasia.
2. There may be genetic implications affecting counselling.
3. The potential fertility of the infant should be established.
4. A plan for surgical management is helpful in many cases.

CONGENITAL DIAPHRAGMATIC HERNIA

Most affected children are now diagnosed from routine antenatal ultrasound scans. The defect in the diaphragm is usually left-sided and posterolateral (Bochdalek hernia). The rapidity of onset and severity of respiratory distress vary with the degree of lung hypoplasia. There is associated pulmonary hypertension from concomitant abnormal development of the pulmonary vasculature. In the most severe cases, poor peripheral perfusion and cardiovascular collapse occur within minutes of birth. A few children with a minor degree of lung hypoplasia have no symptoms for days or months and may present later with gastrointestinal symptoms or an incidental finding on imaging.

In neonatal cases, the chest is barrel-shaped and the abdomen is scaphoid because the bowel has herniated through the diaphragmatic defect into the chest. Once air is swallowed, this sign becomes less obvious. The bowel and liver in the chest displace the mediastinum to the contralateral side. In a left-sided diaphragmatic hernia, this produces apparent dextrocardia (with the heart sounds most easily audible in the right chest) and poor breath sounds on the left side. Bowel sounds may be heard in the chest on auscultation, but this sign is not particularly reliable. The diagnosis is confirmed on a plain chest radiograph. The film should include the abdomen so that the distribution of bowel gas can be determined and an oro- or nasogastric tube should be *in situ*. The features on x-ray include:

1. Loops of bowel within the chest on the side of the defect.
2. Hemidiaphragm not visible.
3. Mediastinal shift to the contralateral side.
4. Abnormal distribution of bowel gas within the abdomen.

The radiological appearances have to be distinguished from cystic lung disease, lobar emphysema, staphylococcal pneumonia with pneumatoceles and other diaphragmatic defects.

OESOPHAGEAL ATRESIA

In oesophageal atresia, the upper oesophagus ends blindly in the upper chest and there is absence of a variable length of the mid-portion of the oesophagus. In the majority of affected patients (approximately 85%), there is a distal tracheo-oesophageal fistula. Occasionally, there may be no fistula at all, or a proximal fistula with or without a distal tracheo-oesophageal fistula. An isolated tracheo-oesophageal fistula without oesophageal atresia is often included in discussion of these abnormalities, although oesophageal continuity means that the tracheo-oesophageal fistula (H-fistula) may not be recognised until beyond the neonatal period. The infant with oesophageal atresia usually presents within hours of birth with excessive drooling (the 'mucousy' baby) from accumulation in the blind upper oesophagus of saliva that cannot be swallowed.

There may be respiratory distress, which may have a variety of causes including ineffective ventilation from escape of air down the fistula, diaphragmatic splinting from gaseous abdominal distension, aspiration pneumonia and respiratory distress syndrome of prematurity. Nearly half these babies are premature and there may be a history of maternal polyhydramnios. The diagnosis is made by passing a 10Fr feeding tube through the mouth and observing that it becomes arrested at about 10 cm from the gums. A smaller calibre tube may curl up

in the blind-ending upper pouch, giving the false impression of oesophageal continuity. A plain radiograph of the chest and abdomen reveals whether there is a distal tracheo-oesophageal fistula; if present, the fistula allows gas into the stomach, which can be observed on x-ray. Associated congenital abnormalities are sought, particularly those in relation to the VACTERL association (Vertebral, Anal, Cardiac, Tracheo-Esophageal, Renal, Limb). These include imperforate anus, congenital heart disease, urinary tract abnormalities and vertebral and limb abnormalities. There is also an increased incidence of duodenal atresia. About 7% of infants born with oesophageal atresia have a significant chromosomal anomaly.

If no gas is seen below the diaphragm on x-ray, it implies there is no distal tracheo-oesophageal fistula. Most of these patients will have no fistula at all, but in 10–20% there will be a proximal tracheo-oesophageal fistula connecting the upper oesophageal pouch and the trachea. This may be identified either on tracheobronchoscopy or by performing a careful upper oesophageal contrast study, avoiding overflow of contrast into the lungs.

In isolated tracheo-oesophageal fistula, the symptoms may include respiratory distress, recurrent or repeated episodes of pneumonia, abdominal distension and choking with feeds. The fistula can be identified by a prone tube oesophageal contrast study or by tracheobronchoscopy. The fistula runs obliquely between the two structures in the lower neck, and can be divided via a cervical approach.

INTESTINAL ATRESIAS

The three cardinal symptoms of bowel obstruction in the neonate are:

1 Vomiting (particularly bilious vomiting).
2 Abdominal distension.
3 Failure to pass, or delay in the passage of, meconium.

It is important to recognise that in the neonate these symptoms represent mechanical obstruction until proved otherwise. Other causes of these symptoms include:

1 Localised or generalised sepsis.
2 Congenital heart disease.
3 Inborn errors of metabolism.

There are various causes of neonatal bowel obstruction (*Table 2.1*). Most mechanical causes present with symptoms and signs within 48 hours of birth, whereas functional obstructions (e.g. Hirschsprung disease and necrotising enterocolitis) usually present later. Bile-stained vomiting, in the absence of generalised abdominal distension, is suggestive of a high obstruction (e.g. midgut volvulus or duodenal atresia), whereas vomiting of milk, which after a period becomes bile-stained, with obvious abdominal distension, is more likely to be associated with distal small bowel or colonic obstruction. Atresia of the small bowel (ileum or jejunum) is relatively common in comparison with colonic atresia, which is rare. Small bowel atresias may be multiple. Any infant who presents with green vomitus as the initial symptom should be regarded as having intestinal malrotation with volvulus until proved otherwise, because of the potentially fatal outcome of this condition.

TABLE 2.1 Causes of neonatal bowel obstruction

Disease	Frequency
Hirschsprung disease	More common
Necrotising enterocolitis	
Small bowel atresia	Less common
Malrotation with volvulus	
Duodenal atresia/stenosis	
Imperforate anus	
Meconium ileus	
Prenatal perforation (meconium peritonitis)	Uncommon

Abdominal distension at delivery suggests distal small bowel obstruction, and is typical of meconium ileus. More distal bowel obstructions, as seen with anorectal malformations, Hirschsprung disease or necrotising enterocolitis, do not present with abdominal distension at birth. In intestinal malrotation with volvulus, abdominal distension may develop late as a result of volvulus causing obstruction of both the duodenal and colonic ends of the midgut and loss of fluid into the gut. In this situation, the abdomen may become dramatically distended and exhibit signs of peritoneal irritation. Other causes of abdominal distension/mass (e.g. massive hydronephrosis, ovarian cyst, duplication cyst, ascites or organomegaly) must be considered. Percussion of the abdomen is helpful in determining whether the distension is from dilated loops full of air or from fluid or solid structures.

Meconium is passed within 24 hours of birth in 95% of neonates born at term. One feature of Hirschsprung disease is failure to pass meconium in the first 24 hours after birth. However, in premature or very sick neonates, passage of meconium may also be delayed.

MECONIUM ILEUS

In this condition, meconium becomes excessively tenacious and sticky, causing a mechanical bowel obstruction in the ileum. The obstruction is caused by hard pellets of impacted faecal material. In most cases, meconium ileus is due to cystic fibrosis, which *in utero* causes changes to the physical properties of the meconium to make it more viscous.

The radiological appearance is variable, but classically includes air–fluid levels and a foamy pattern of air bubbles trapped around the impacted meconium. A contrast enema will demonstrate a microcolon, because the colon is unused and empty. In meconium ileus there is abdominal

distension at birth, which increases subsequently. If the heavy meconium-laden ileum twists *in utero*, the infant will be born with an intestinal atresia and signs of meconium peritonitis, including intra-abdominal calcification. A localised volvulus may also occur after birth, and cause peritonitis.

The presence of ischaemic or perforated bowel must be suspected in any neonate with marked abdominal distension. Abdominal tenderness and guarding, discolouration of the thin abdominal wall, tight and shiny skin over a distended abdomen with progressive oedema and redness of the abdominal wall, and signs of sepsis and shock are all suggestive of an underlying necrotic bowel and peritonitis. On x-ray, free gas may be evident under the diaphragm or outlining the falciform ligament (football sign) or both sides of the bowel wall (Rigler sign). It is important to take a lateral decubitus or lateral 'shoot-through' radiograph, as well as a supine film, to demonstrate small amounts of free gas in the peritoneal cavity.

HIRSCHSPRUNG DISEASE

Hirschsprung disease produces a functional obstruction secondary to absence of ganglion cells in the distal large bowel. The affected segment begins at the anorectal junction and extends proximally for a variable distance – in most cases as far as the sigmoid colon. On occasions (<10%), the affected segment can extend as far as the small bowel (total colonic aganglionosis) or, very rarely, involve the entire alimentary tract, in which situation survival is unlikely. Hirschsprung disease becomes apparent clinically in the first few days of life, when it manifests as delayed passage of meconium, followed by vomiting and abdominal distension. On rectal examination, the anus and rectum may feel tight and there may be a squirt of meconium and air on extraction of the examining little finger. The plain radiological appearance is non-specific but often shows dilated bowel with absence of rectal gas. A contrast enema may demonstrate a narrow rectal lumen with a transition zone in the proximal rectum or sigmoid colon. The definitive diagnosis is made when a suction rectal biopsy shows an absence of intrinsic ganglion cells and hypertrophic extrinsic nerve fibres. In the common type of Hirschsprung disease, males outnumber females by about 4:1, and the aganglionic segment rarely extends beyond the sigmoid colon proximally. Infants with more extensive long-segment disease have an equal sex incidence; in these there is a high degree of 'penetrance' with subsequent siblings more likely to be affected.

NEONATAL NECROTISING ENTEROCOLITIS

Neonatal necrotising enterocolitis is a potentially fatal disease in which there is ischaemia of the bowel associated with inflammation and gas forming bacteria. Premature neonates are most at risk but term infants are not immune. Abnormal gut flora, formula feeds (rather than breast milk), immature mucosal defence mechanisms, sepsis and compromised gut perfusion are all implicated in the pathogenesis of the disease. Onset is usually within 1–3 weeks of birth but is inversely related to gestational age. The infant becomes ill, lethargic and intolerant of feeds. There may be fever. Vomiting occurs early, and may become bile-stained. There is abdominal distension and the passage of loose stools containing a variable amount of blood, mucus and even necrotic tissue.

Mucosal necrosis progresses to full-thickness bowel necrosis and signs of peritonitis develop: the anterior abdominal wall becomes oedematous and red, with dilated veins. Abdominal palpation causes pain. A mass may be palpable if a localised intraperitoneal abscess has developed or if there is a persistent dilated loop of affected bowel.

The radiological features are variable. The pathognomonic finding is intramural gas ('pneumatosis intestinalis'). Gas outlining the portal vein and its branches ('portal venous gas') suggests severe disease. Free gas in the peritoneal cavity outlining the falciform ligament (football sign) or both sides of the bowel wall (Rigler sign) indicates intestinal perforation. Small amounts of free gas are best seen on a decubitus or lateral shoot-through radiograph. Other radiological features include gaseous dilatation, excessive peritoneal fluid – seen as separation of adjacent loops of bowel – and an abnormal distribution of bowel gas. At operation, the disease may be extensive and involve most or all of the bowel, or it may be localised to more discrete areas, usually within the ileum or colon.

Following medical treatment, a stricture may form, most often in the colon.

NEURAL TUBE DEFECTS

These are a group of congenital disorders in which the brain and/or spinal cord are malformed as a result of abnormal closure of the neural tube in the embryo. A range of malformations exist with variable neurological deficits. Most neural tube defects are readily detectable by antenatal screening tests including ultrasound imaging. In anencephaly, the cephalic part of the neural tube has failed not only to close, but also to develop. Deficiency of the vault of the skull exposes the brainstem and cerebellum – most of the cerebrum is missing. An encephalocele is a neural tube defect affecting the brain, and is commonest in the occipital and frontal regions.

In open spina bifida, the spinal defect is not covered by skin. There are two main variants: myelomeningocele in which neural elements are exposed (this accounts for about 90% and is associated with hydrocephalus and the Arnold Chiari malformation) and meningocele. When a baby is born with obvious open spina bifida, the vertebral level of the neural tube defect should be established as this predicts the degree of neurological deficit. Lumbosacral lesions are the most common, but fusion defects can occur at any

point along the spine. The lesion may leak cerebrospinal fluid or tissue fluid from the exposed neuroepithelium. With a meningocele, a communication with the subarachnoid space can be shown when direct pressure on the lesion causes the fontanelle to bulge. Likewise, the lesion will become more tense during crying, when intracranial pressure is increased.

The level of paralysis in the neonate with myelomeningocele may be difficult to determine exactly, but observation of spontaneous movement of the hips, knees and ankles without external stimulation is revealing. The motor deficit can be estimated by observing active movement in the hips, knees and ankles. When the lesion includes nerves from or above the third lumbar segment, the legs are totally paralysed. If L3 is functional, the posture is of flexion of the hip and extension of the knee, but there is no other limb movement. When there is preservation of L4 and L5 spinal segments, there is movement of the hip and knee and the foot can actively dorsiflex, but no plantar flexion occurs. Muscular imbalance around the ankle may cause talipes. When the first two sacral segments are intact, leg movements are essentially normal, but bowel and bladder incontinence is still likely.

The sensory deficit is usually less than the corresponding motor deficit. The pattern of dermatome distribution in the neonate is the same as that in adults. The perianal skin is innervated by the third and fourth sacral segments. The sensory level is determined by commencing examination in the anaesthetic region, and proceeding towards an area of normal sensation. Rectal prolapse and absence of a natal cleft indicate paralysis of the pelvic floor muscles and adjacent bowel and bladder sphincters.

Reflex contraction of the external anal sphincter and gluteal muscles to stimulation with a blunt-ended pin will occur if the distal sacral segments are intact. Normal perineal sensation and musculature suggests that bladder function is preserved, which can be confirmed by manual compression of the lower abdomen, which should not produce incontinence of urine.

The presence and development of hydrocephalus should be sought, as well as orthopaedic deformities of the spine, hips and feet. Secondary kyphoscoliosis is common, and if there are associated hermivertebrae, may be present at birth. The lumbosacral myelomeningocele may produce imbalance of the hip muscles, causing congenital dislocation of the hip. Therefore, Ortolani's test should be performed (Chapter 9).

In closed spina bifida, the defect is covered by skin but there is a visible abnormality of the overlying or adjacent skin such as a dermal sinus, haemangioma, hairy patch, fatty lump or other cutaneous marking. These lesions are also referred to as occult spinal dysraphism and may be associated with a tethered spinal cord, which may cause neurological signs later in childhood, including dysfunctional voiding, pes cavus and clumsiness of the legs. When a sinus or dermoid cyst of the spinal canal has an external connection, meningitis may occur. A careful inspection of the dorsal midline should be conducted to identify a possible sinus opening in any patient who develops meningitis in which a skin organism is cultured. These sinuses may not be particularly obvious. They usually lie outside the coccygeal area (above the intergluteal cleft) and should be distinguished from the common sacral dimple.

Spina bifida occulta (a skin covered posterior vertebral defect with no visible external abnormalities) is a common incidental finding on x-ray of the lower spine and is rarely associated with abnormal neurology. There is a gap in the dorsal arches of the fourth and fifth lumbar vertebrae and the first sacral vertebra.

Rare variants of neural tube fusion may occur, with the spinal cord split longitudinally by a bony ridge within the vertebral canal (diastematomyelia). Some myelomeningoceles are associated with a teratoma. In the rare Currarino syndrome there is a sacral dysgenesis (typically a hemisacrum) combined with a presacral mass (e.g. an anterior sacral meningocele or teratoma) and an anorectal malformation (usually anorectal stenosis).

CONJOINED TWINS

Conjoined twins are rare, occurring in about one in 50,000–100,000 births. They are understood to be caused by early division of the inner cell mass during embryogenesis, which forms two identical, but attached, fetuses. The classification of conjoined twins relates to the site of attachment, the commonest variety being thoraco-omphalopagus, where the attachment is along the sternum and upper abdomen. Pygopagus twins, in which the pelvis of each infant is the site of connection, occur in about 20% of instances. Conjoined twins connected by the head (craniopagus) occur in only 5% of instances. Conjoined twins require detailed and careful investigation to determine the exact anatomical and functional status of each twin. Xiphopagus twins, joined around the xiphisternum and epigastrium, often share a fused liver, but this is one of the most favourable types for separation. Surgery is best deferred until detailed imaging has been performed and an opportunity to observe function of the various organs. The ultimate aim of surgery is to separate the twins to produce two live, separate infants. Sometimes, unequal distribution of organs does not allow equal separation. In addition, in some instances of conjoined twinning, the development of each twin is not symmetrical, so that one twin is much larger than the other. In its more extreme form, this may present at birth with the appearance of a parasitic, dysmorphic twin attached to the external surface of the otherwise normal twin.

SACROCOCCYGEAL TERATOMA

This is a rare congenital germ cell tumour occurring at the caudal end of the developing vertebral column (notochord). The tumour is usually attached to the coccyx. It may be a

large (exophytic) tumour obvious at birth, or may be concealed (endophytic) growing anteriorly from the coccyx into the pelvis behind the rectum. Large external tumours diagnosed at birth are excised promptly. Endophytic tumours may not be diagnosed until later and are more likely to have become malignant. Many sacrococcygeal tumours are now detected by prenatal ultrasound scan and affected fetuses require monitoring *in utero* because of the potential of the tumour to recruit a high blood flow and cause fetal hydrops. All sacrococcygeal teratomas, irrespective of their external appearance, need prompt excision after birth because of the high risk of malignant degeneration in the first few months of life.

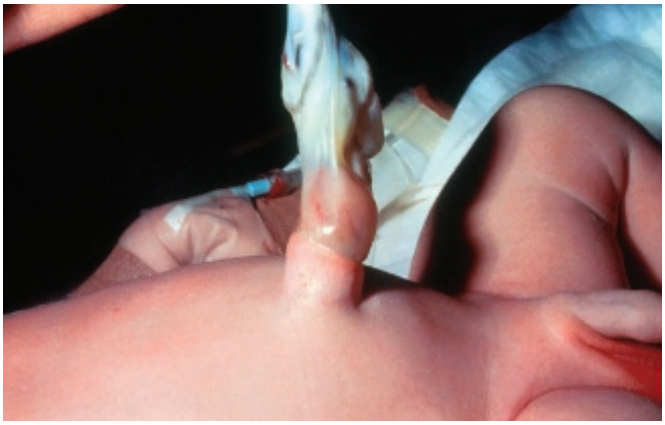


Fig. 2.1 This is the most minor form of exomphalos, essentially a hernia into the umbilical cord. Care must be taken when clamping the cord to avoid inadvertent injury to the bowel within the defect.

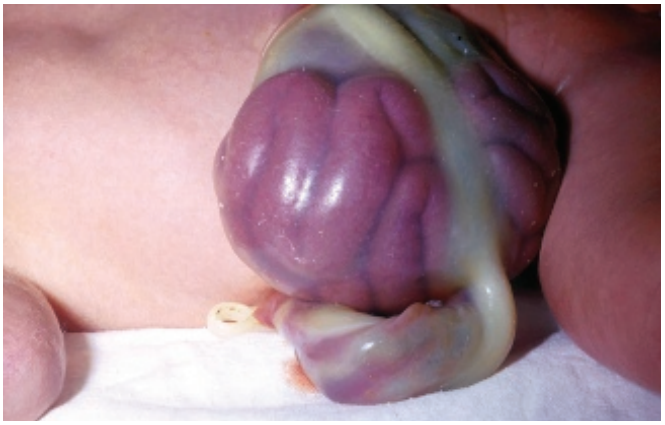


Fig. 2.2 A typical intermediate-sized exomphalos, with loops of small bowel visible through the translucent sac. The sac consists of apposed layers of amnion and peritoneum. As the sac dries it becomes opaque, such that the sac contents become more difficult to see.

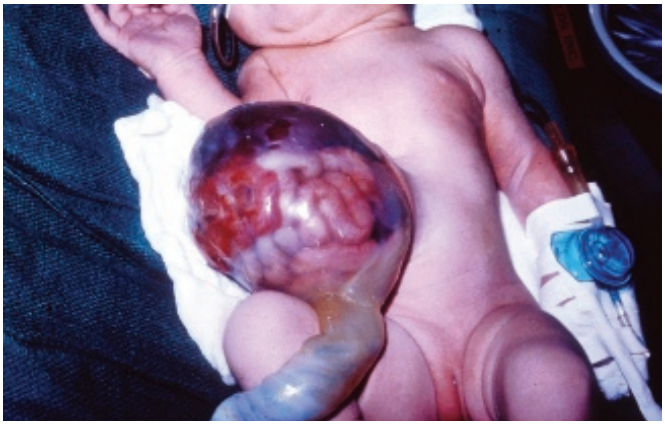


Fig. 2.3 A large exomphalos into which have herniated loops of bowel and the liver. The relatively small size of the abdominal cavity may make an exomphalos of this size difficult to close at birth with primary surgery.

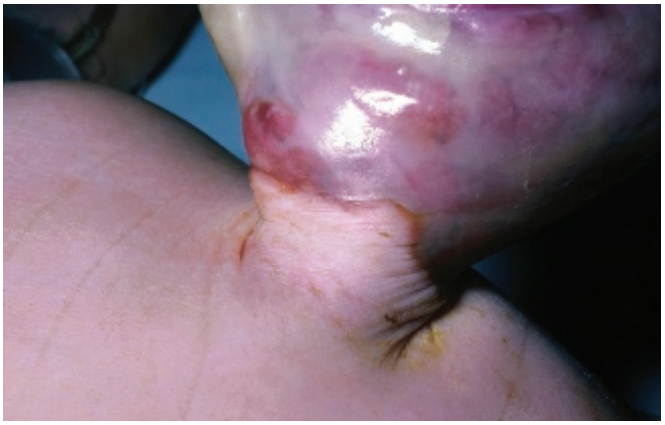


Fig. 2.4 In this moderate-sized exomphalos, the neck is relatively narrow and the skin of the abdominal wall can be seen encroaching on the sac.

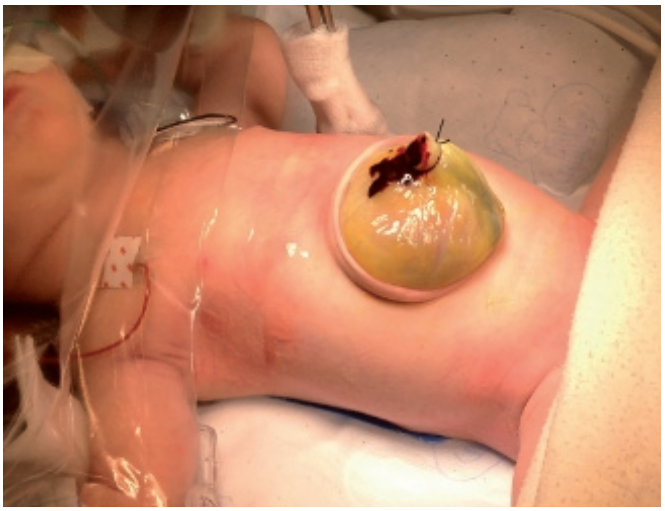


Fig. 2.5 Exomphalos major with a wide defect in the abdominal wall.

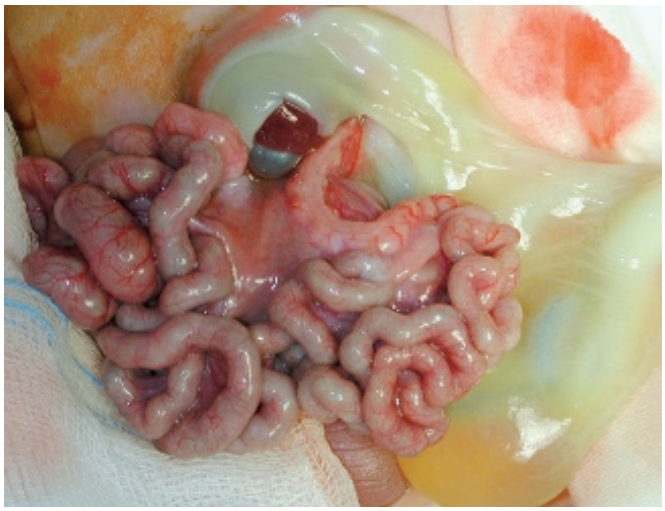


Fig. 2.6 Exomphalos with early rupture of the sac.

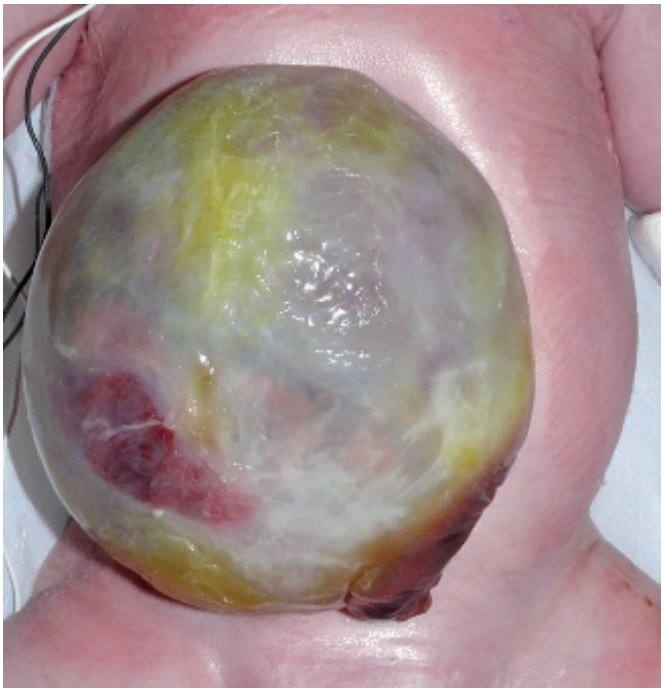


Fig. 2.7 The appearance of exomphalos major, sometimes known as 'giant exomphalos, several days after birth.

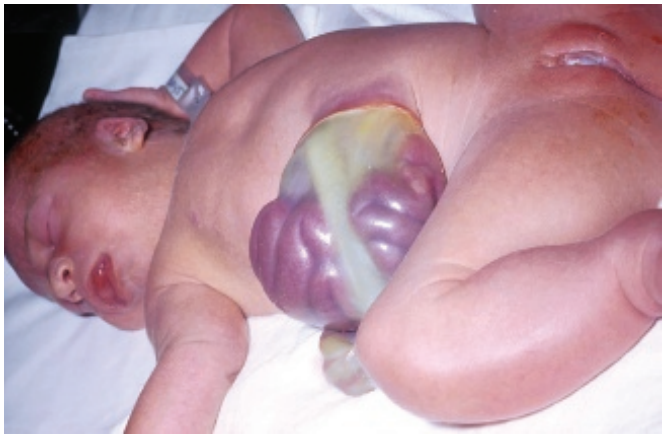


Fig. 2.8 This large baby presented with exomphalos and visceromegaly, features suggestive of Beckwith–Weidemann syndrome. The enlarged viscera may be the result of excess insulin production by the fetal pancreas. The babies are at risk of profound postnatal hypoglycaemia, which might be severe enough to cause cerebral injury. The hypoglycaemia and excess insulin production usually resolve spontaneously within a week or two of birth.