MINISTRY OF HEALTH OF UKRAINE
ZAPORIZHZHIA STATE MEDICAL-PHARMACEUTICAL UNIVERSITY
DEPARTMENT OF PEDIATRIC SURGERY

O. V. Spakhi, O. P. Paholchuk, M. O. Makarova, O.D. Kokorkin

CONGENITAL MALFORMATIONS IN CHILDREN. DIAGNOSIS AND TREATMENT

Tutorial

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The tutorial provides data on the main diagnostic criteria and treatment tactics for acute surgical conditions and congenital malformations that require early surgical correction. The tutorial contains information necessary for independent training of students of 5-6 years, interns of children's surgeons for practical classes in children's surgery. The tutorial was developed taking into account modern views on the clinic, diagnostics, additional methods of examination, tactics of treatment of children with surgical diseases and congenital malformations.

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Preface

Accurate and timely detection is of great importance for the successful treatment of any disease, especially a disease that requires surgical treatment in childhood. Therefore, in surgical practice, the widespread use of various special research methods is relevant, which differ from the generally accepted physical research methods (inspection, palpation, percussion and auscultation) in their objectivity and allow for a more accurate establishment of the nature of the pathological process affecting the organ and the zone of its spread to other organs. Congenital malformations are quite rare, but mortality in these children remains high. In the practice of a doctor, what rarely happens is rarely diagnosed in a timely manner. An increase in antenatal pathology, including malformations associated with untimely diagnosed malformations of the genital organs in boys, has been noted, which leads to disorders of the reproductive function in men at the age of puberty. The problem of timely diagnosis and treatment of purulent-destructive diseases in children, hernias of the anterior abdominal wall, acute surgical diseases of the abdominal organs, diseases of the chest and thoracic organs, organic diseases of the intestine, among which the most common are intestinal intussusception and ileal intestinal obstruction, obstruction of the small and large intestines against the background of Meckel's diverticulum, volvulus and knotting of the small and large intestines, pinched internal hernias, remains relevant in pediatric surgery today.

With the development of medicine, new knowledge about the course of the pathological process and means of their treatment appear. Therefore, knowledge of diseases will allow to timely identify the leading syndrome of the disease, intrasyndromic diagnostics, correctly interpret the results of additional examination methods, correctly diagnose the child and choose the right treatment tactics. Diagnostic and tactical errors can be fatal in treatment, which determines the relevance of this manual and has a general goal - the ability to conduct treatment and diagnostic measures at a modern level for children with these diseases and apply the basic principles of rehabilitation in the interests of providing timely assistance to patients.

I. Relevance

The surgeon should always thoroughly explain the child's problem. Reviewing the results of imaging studies with the parents and patient is helpful. Freehand drawings and diagrams from books can also be used to aid the surgeon in illustrating anatomy and explaining the problem. Parents often gain a better understanding of their child's problem if the surgeon takes the time to explain how or why the problem arose. Be prepared to explain embryology in layperson's terms when talking to parents of patients with congenital lesions and/or defects. Also be familiar with basic genetics and modes of inheritance when counseling parents of a child with a genetic defect. Knowledge in oncology is useful when discussing tumors; be prepared to answer general questions regarding chemotherapy and radiation regimens for tumors commonly encountered in pediatric surgery. Notify parents that the oncology staff is part of the team involved in their child's care.

The explanation of the proposed surgery in layperson's terms includes describing where the incision will be made, the steps of the surgery, how the incision will be closed, and the size of the scar. At this time, basic postoperative issues can also be addressed, including the anticipated length of hospital stay, the activity and dietary restrictions in the postoperative period, and the time the child will likely be away from school. Explicitly explaining why the surgery should be performed and what it should accomplish is important. This is also the time to discuss the risks of surgery. In addition, discussing options and alternative treatment plans is important. The consequences of not performing surgery should be addressed as well.

Pause after providing important information so that parents have the opportunity to take in all the information. Leave time for questions at the end of the encounter, and give parents a means by which to contact you with questions. Refer parents to other resources, such as support groups, the hospital's family resource center, and reliable sources on the Internet. Caution parents with regard to the quality of information they might find on the Internet because the accuracy of information varies widely.

Physical Examination

The goal of the physical examination is to identify the current surgical issues and to ensure that the organ systems other than the one being treated are healthy. Unlike the adult physical examination in which one can often follow the same routine every time, the pediatric examination must be modified for each patient. Interacting with children of different ages and temperaments in different settings can be challenging.

Hand washing before and after performing the physical examination is essential. Hand washing serves purposes more than infection control. On a psychological level, it conveys a reassuring message to the parent that hygiene is important to the surgeon. On a practical level, it warms the surgeon's hands before he or she touches the child. In an older and cooperative child, physical examination can be performed by using a standard routine. However, this routine may need to be modified in young children or infants who do not cooperate.

Infants should be positioned on the examination table for the entire examination. Toddlers and small children may sit in their parent's lap for the initial part of the examination, and they can be moved to the examination table, and for the abdominal, inguinal, genital, and rectal examinations, when required. Having the parent by the examination table reduces the child's anxiety and should be encouraged.

II. Educational goals

To learn how to collect anamnesis in patients with acute pathology, to examine a patient with this pathology for the purpose of clinical diagnosis of the disease, to define diagnostic criteria, forms and severity of the disease, to identify and characterize complications, to make differential diagnosis by place and type of lesions, to appoint differential treatment , prescribe treatment, organize preventive measures.

Be able to make a preliminary diagnosis in newborns and young children with acquired pathology, make a differential diagnosis and determine the tactics of their treatment.

student should know:

- anatomical and physiological features of the chest, abdominal cavity in children;

- clinical methods of examination of a child with abdominal pain;

- classification of intestinal obstruction in children;

- etiology and pathogenesis of various types of intestinal obstruction in children;

- radiological signs of various types of intestinal obstruction;

- laboratory criteria for the diagnosis of bowel obstruction in children;

- ultrasound data of intestinal invagination in children, depending on the stage of the disease;

- to know the methods of conservative and surgical treatment of children with various types of intestinal obstruction, bleeding, trauma, inflammatory diseases;

- to know the rules of dispensary supervision of children operated on bowel obstruction, bleeding, trauma, inflammatory diseases.

- to evaluate the presence of pathological process, intoxication, complications of diseases that cause mechanical intestinal obstruction and to interpret them and to make the scheme of diagnostic and differential diagnostic search.

- have the skills of clinical investigation of the patient, be able to identify the necessary additional examinations.

- to determine the tactics and pathogenetic principle of treatment of patients.

- to know the moral and ontological principles of a medical specialist and the principles of professional subordination

- be able to interpret radiological examination data;

- interpret ultrasound and laboratory test methods;

- Based on the survey data, to diagnose the stage of intestinal invagination;

- to form indications for the operative method of treatment, depending on the variant of obstruction of intestines;

- to make an individual scheme of diagnostic search for diseases.

- to make a preliminary diagnosis for diseases.

- to carry out differential diagnostics of diseases

- determine treatment tactics and principles of disease rehabilitation

- to make prognosis of life and health in diseases.

- Demonstrate mastery of morally-deontological principles of medical specialist and principles of professional subordination in pediatric surgery.

III. Congenital malformations of the lungs and pleura. Congenital partial emphysema.

Congenital cystic diseases of the lung consist of cystic adenomatoid malformation (CAM), bronchogenic cyst, pulmonary sequestration (PS), and congenital lobar emphysema (CLE). These show close relationship in terms of embryology and clinical presentation. Some cause life threatening respiratory distress at birth while others appear late in life as incidental finding on radiograph. Majority of symptomatic patients present with progressive respiratory distress or repeated chest infections with cystic changes in the chest X-ray.

1. Congenital cystic adenomatoid malformation

Congenital cystic adenomatoid malformations (CCAMs) present as cystic or solid lung masses restricted to part of one lung. Most CAMs present with respiratory distress or compromise during infancy or recurrent pneumonias in later years, including adulthood. The pathological feature of CAMs is adenomatoid proliferation of the bronchioles that forms a cyst at the expense of alveoli. Three types of CAMs are recognized based on cyst size, number, and pathology.

Type 1 consists of multiple large cyst (2-10 cm), at least one cyst will be dominant with smaller cysts seen along its periphery. The cyst wall is lined with ciliated pseudostratified columnar epithelium, contains elastic tissue beneath the epithelium, smooth muscle, and fibrovascular connective tissue, including cartilage.

Type 2 consists of small and more uniform size cysts (0.5-2 cm). They are lined with cuboidal to columnar epithelium and have only a thin fibromuscular wall. This type has associated anomalies – renal, cardiac, skeletal, intestinal, extra-lobar sequestration.

Type 3 lesions are bulky solid lesions that usually involve entire lobe of the lung. There is slight modification of the classification of CCAM with now five types being recognized out of which only three types are identified on imaging as mentioned above. It is now called as congenital pulmonary airway malformations

since all the types are not cystic. Additional types are, Type 0, which has either no cyst or very small ones (<0.5 cm), and is incompatible with life and

Type 4 CCAM which consists of large cysts up to 10 cm in size.

The radiological features depend on the content, size, and number of cysts. Chest radiograph reveals multiple air-filled thin-walled cysts of varying sizes. At birth, these cysts are filled with fluid, later air enters into the cysts as they communicate with tracheobrochial tree and with each other. Thus may contain only air or air-fluid levels [Figure 1].

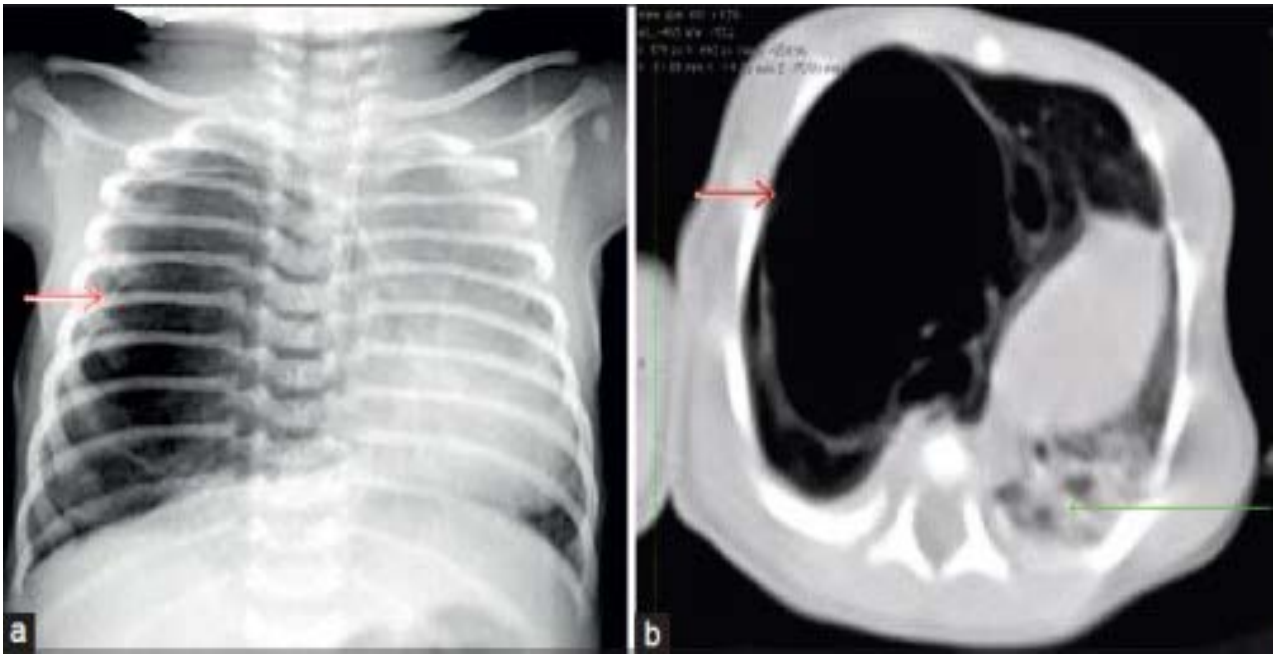


Figure 1. Chest radiograph shows a large air filled thin walled cyst (red arrow) in the right lung with herniation of the lung to the contralateral side, Computed tomography chest shows multilocular cysts (red arrow) of variable sizes in the right upper and middle lobe, largest cyst measuring 64 × 42 mm with herniation of the lung and mediastinum shift to the contralateral side. (Jain A, Anand K, Singla S, Kumar A. Congenital cystic lung diseases. J Clin Imaging Sci. 2013 Jan 30;3:5. doi: 10.4103/2156-7514.106620. PMID: 23607074; PMCID: PMC3625881.) [10]

Conversely only fluid filled cysts may be seen due to retained secretions, hemorrhage, or secondary to infection (Figure 2).

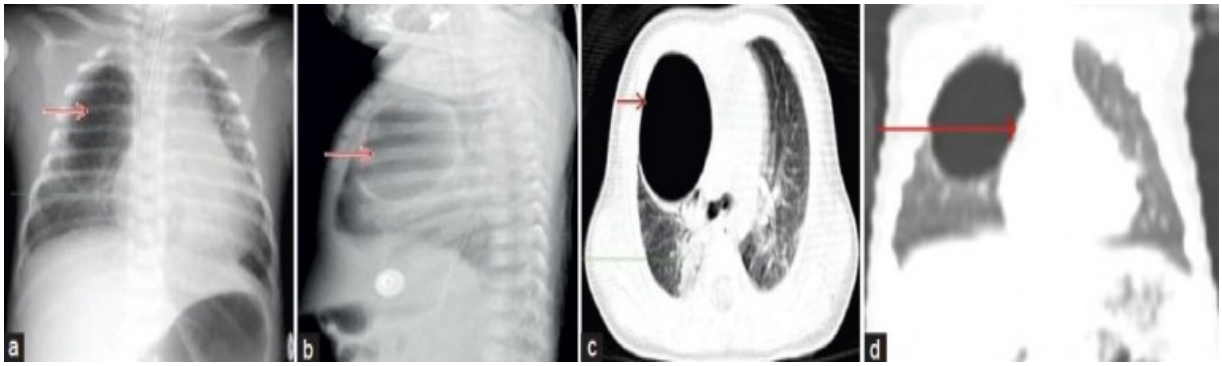


Figure 2. (a) Chest radiograph anteroposterior view shows large thick walled air filled unilocular cyst (red arrow) in the right lung upper and mid zones. Consolidation is seen adjacent to the cyst (green arrow), (b) Lateral view of chest radiograph shows large thick walled unilocular cyst (red arrow) in the right upper and mid zone, (c) Computed tomography chest (lung window) reveals a large unilocular thick walled air filled cavity (red arrow) in the right upper lobe with compression of the adjacent lung parenchyma. Consolidation is noted in the right lower lobe (green arrow). No fluid is seen within the cyst. No obvious communication of the cyst with bronchial tree is visible, (d) Computed tomography coronal reconstruction scan shows no obvious communication with bronchial tree (red arrow).

(Jain A, Anand K, Singla S, Kumar A. Congenital cystic lung diseases. J Clin Imaging Sci. 2013 Jan 30;3:5. doi: 10.4103/2156-7514.106620. PMID: 23607074; PMCID: PMC3625881.) [10]

Computed tomography (CT) also shows similar findings as radiographs. In another case, the chest radiograph showed well-defined homogenous opacity [Figure 3]. CT images also show fluid filled lesion. The main role of CT is to differentiate these cysts from bronchogenic cysts, lobar emphysema, and from sequestration, to accurately localize the site of lesion within the lung segments for surgical planning. Treatment consists of simple resection of the involved tissue or lobectomy in CCAM.

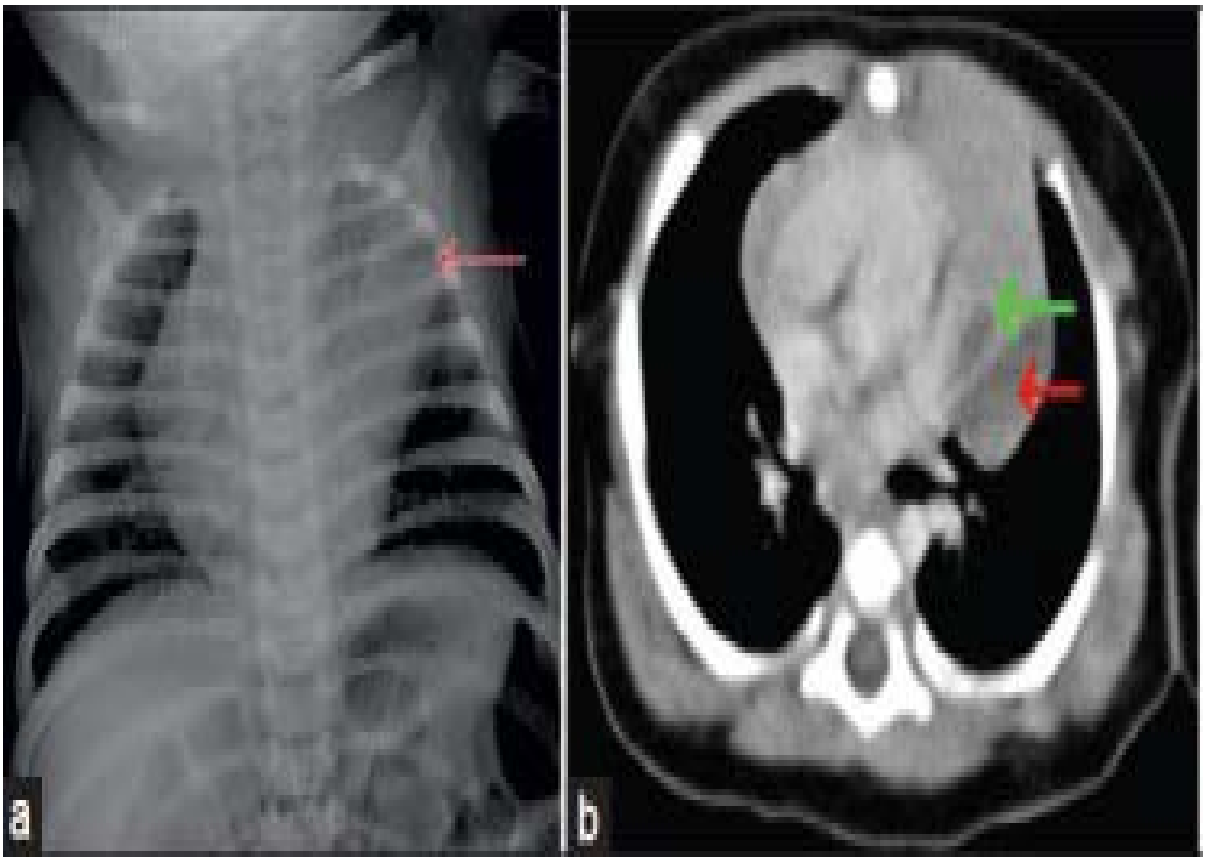


Figure 3. Chest radiograph anteroposterior view reveals well defined homogenous opacity in the left upper zone (red arrow) which could not be differentiated from normal thymic shadow, (b) Computed tomography mediastinal window reveals a well-defined fluid attenuating cyst (red arrow) with septations in the middle mediastinum posterior to the thymus. The fat planes with the thymus (green arrow) were not well delineated, (Jain A, Anand K, Singla S, Kumar A. Congenital cystic lung diseases. J Clin Imaging Sci. 2013 Jan 30;3:5. doi: 10.4103/2156-7514.106620. PMID: 23607074; PMCID: PMC3625881.) [10]

1. Bronchogenic cyst

Bronchogenic cysts arise from abnormal budding of the tracheobronchial tree during airway development. This abnormal bud subsequently differentiates into a fluid-filled blind-end pouch. More often present in the mediastinum, one third can occur in the lung parenchyma, usually within the lower lobes. These cysts may

contain air, fluid, or both. Clinical manifestations are related to various mass effects or secondary infection of the cyst. Lined by ciliated columnar or cuboidal epithelium and are surrounded by tissues similar to those of the normal bronchus; including cartilage, smooth muscle, elastic tissue, and mucous glands.

Intrapulmonary bronchogenic cyst are rare (14%), most of them are fluid attenuating. Only 36% of intrapulmonary bronchogenic cyst contains air, which occurs when the cyst communicates with the bronchial tree. Rogers and Osmer described their radiographic findings as a sharply defined, solitary, uncalcified round or oval density presenting as one of three categories: A cyst with a homogeneous density filled with fluid, an air-filled cyst, or a cyst containing an air and fluid. CT images shows the cyst as a well-defined spherical mass with or without mass effect and with attenuation of water or soft-tissue. In one case ,the radiograph shows thick walled cyst with no communication with bronchial tree. A fluid filled cyst is seen in the radiograph. CT images show fluid cyst. The CT attenuation values of the intrapulmonary bronchogenic cysts are same as of mediastinal bronchogenic cysts.

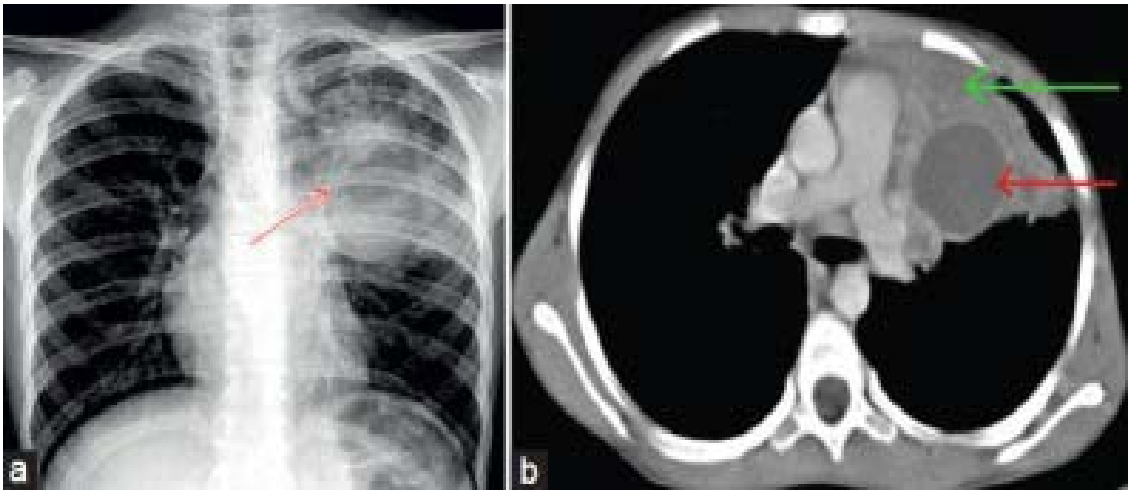


Figure 4. Chest radiograph shows inhomogeneous opacity (red arrow) in the left upper and mid zone with well-defined inferior margin, (b) Computed tomography chest shows large well defined unilocular fluid containing cyst (red arrow) in the left upper lobe with thick enhancing walls. Surrounding lung shows consolidation (green arrow) (Jain A, Anand K, Singla S, Kumar A. Congenital cystic lung diseases. J Clin Imaging Sci. 2013 Jan 30;3:5. doi: 10.4103/2156-7514.106620. PMID: 23607074; PMCID: PMC3625881.) [10]

The high attenuation on unenhanced CT scans is caused by hemorrhage, proteinaceous content, or calcium. Complications include infection; compressive symptoms, such as dysphagia or arrhythmia; malignant transformation; and the rare but fatal air embolism. The appropriate treatment is surgery, especially in patients with repeated infections. Occurrence of both mediastinal and parenchymal cysts together is very rare.

Congenital lobar emphysema

Congenital lobar emphysema (CLE) is a disease that causes breathing difficulties in newborn or infants. Furthermore, called as infantile lobar emphysema. The incidence of CLE is 1 in 20,000 to 1 in 30,000. Hence, early recognition and treatment is essential. It is reported predominantly in male children affecting left upper lobe in 43%, followed by right middle lobe in 32% of the cases.

There is over inflation of the diseased lobe resulting in shift of mediastinum to contralateral side and compression of ipsilateral lung with attenuation of vascular markings on chest X-ray [Figure 5]. CT findings help to confirm these findings [Figure 5].

CLE is commonly associated with congenital heart disease such as ventricular septal defect and patent ductus arterirous. Differential diagnosis includes CCAM, pulmonary hypoplasia, and pnuemothorax. Treatment includes lobectomy of affected lung.

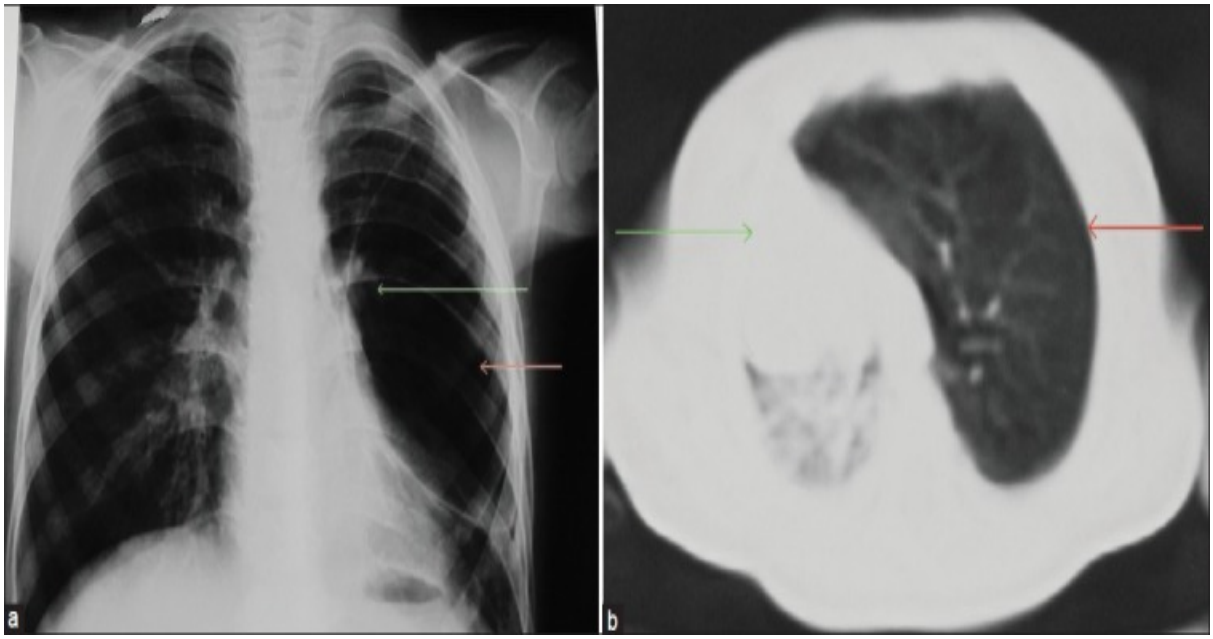


Figure 5. (a) Chest X-ray posteroanterior view shows a large hyperlucent area (red arrow) in the left upper, mid, and lower zone with attenuated vascular markings within the lucency (green arrow), (b) Computed tomography lung window (axial section) shows hyperinflated left upper lobe with attenuated vascular marking (red arrow). There is hernination of left lung field, mediastinal shift to the contralateral side with collapse of right lung (green arrow).

(Jain A, Anand K, Singla S, Kumar A. Congenital cystic lung diseases. J Clin Imaging Sci. 2013 Jan 30;3:5. doi: 10.4103/2156-7514.106620. PMID: 23607074; PMCID: PMC3625881.) [10]

Pulmonary sequestration

Pulmonary sequestration (PS) is a disorder characterized by a non-functioning lung parenchyma, which does not communicate with tracheo-bronchial tree and receives blood supply from systemic artery. Sequestration is believed to be due to abnormal budding of primitive foregut. Anatomically it can be classified as intra-lobar and extra-lobar sequestration. Clinically, sequestration present as recurrent pneumonia in adults and adolescents, in case of intra-lobar and respiratory distress, chronic cough in infancy, in case of extra-lobar sequestration. Intra-lobar sequestration lies within visceral pleura and usually located in posterobasal segment of the lung. Arterial supply is from abdominal aorta or thoracic aorta and venous drainage through pulmonary veins into left atrium. It communicates with adjacent lung parenchyma through pore of Kohn, which allows infection to occur and resolution is incomplete or slow due to inadequate bronchial drainage. Extra-lobar

sequestration is surrounded by its own pleura, usually located on the left side of the lower chest. Extra-lobar sequestration is usually associated with other congenital anomalies like diaphragmatic hernia, congenital heart disease, and CCAM. Radiologically sequestration on a chest radiograph demonstrated by a soft-tissue opacity in chest base [Figure 6]. Bronchi ecstasies, cystic areas, sub-segmental atelectasis, mediastinal shift, and prominence of ipsilateral hila are other radiological findings that arise due to recurrent infection. CT scan show cystic areas containing air or fluid, focal emphysema, and atelectasis. The arterial supply and venous drainage, which are pathognomic features of sequestration are better demonstrated by contrast enhanced CT scan. Magnetic resonance scan can be used for better characterization of the lesion by differentiating the content as solid, cystic, hemorrhagic, or containing mucus components.

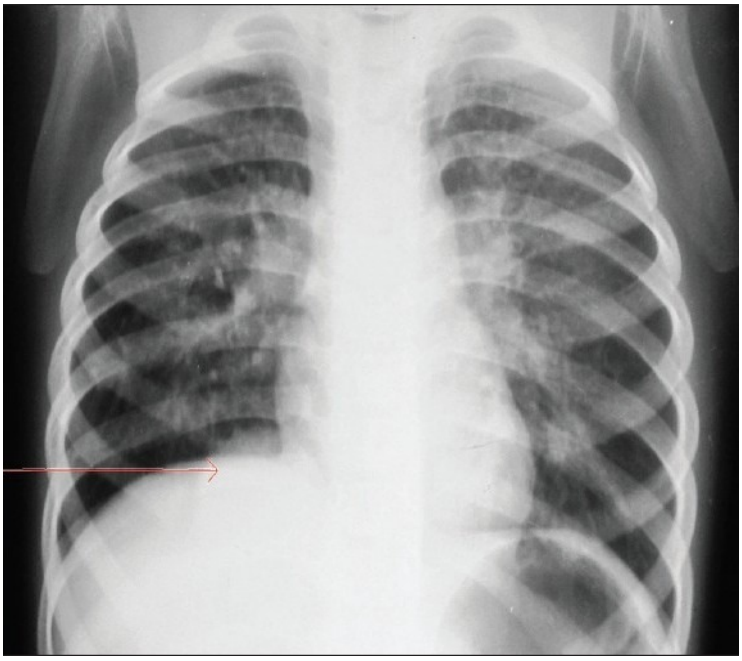


Figure 6. Chest X-ray anteroposterior view shows an homogenous dense opacity (red arrow) in right cardiophernic angle.

Radiological differential diagnosis that need to be considered with this condition are CCAM, diaphragmatic hernia, terotoma, and accessory spleen.

IV. Oesophageal atresia. Diaphragmatic hernia

1. Esophageal atresia

Esophageal atresia (EA) is a rare birth defect in which a baby is born without part of the esophagus (the tube that connects the mouth to the stomach). Instead of forming a tube between the mouth and the stomach, the esophagus grows in two separate segments that do not connect. In some children, so much of the esophagus is missing that the ends can't be easily connected with surgery. This is known as long-gap EA.

EA frequently occurs along with tracheoesophageal fistula (TEF), and as many as half of all babies with EA/TEF have another birth defect, as well. Without a working esophagus, it's impossible to receive enough nutrition by mouth. Babies with EA are also more prone to infections like pneumonia and conditions such as acid reflux. Luckily, EA is usually treatable.

Epidemiology

Oesophageal atresia is a relatively common congenital malformation occurring in one in 2500–3000 live births. The overwhelming majority of cases of oesophageal atresia are sporadic/non-syndromic, although a small number within this non-familial group are associated with chromosomal abnormalities. Familial/syndromic cases of oesophageal atresia are extremely rare, representing less than 1% of the total. Oesophageal atresia is 2 to 3 times more common in twins.

Classification

The original classification by Vogt in 1929 is still used today. Ladd (1944) and Gross (1953) modified the classification, while Kluth (1976) published an extensive "*Atlas of Esophageal Atresia"* which comprised 10 major types, each with numerous subtypes which is based on the original Vogt classification. It would appear to be more valuable to describe the anatomical anomaly rather than assign a label which may not be widely recognisable (Fig 7).

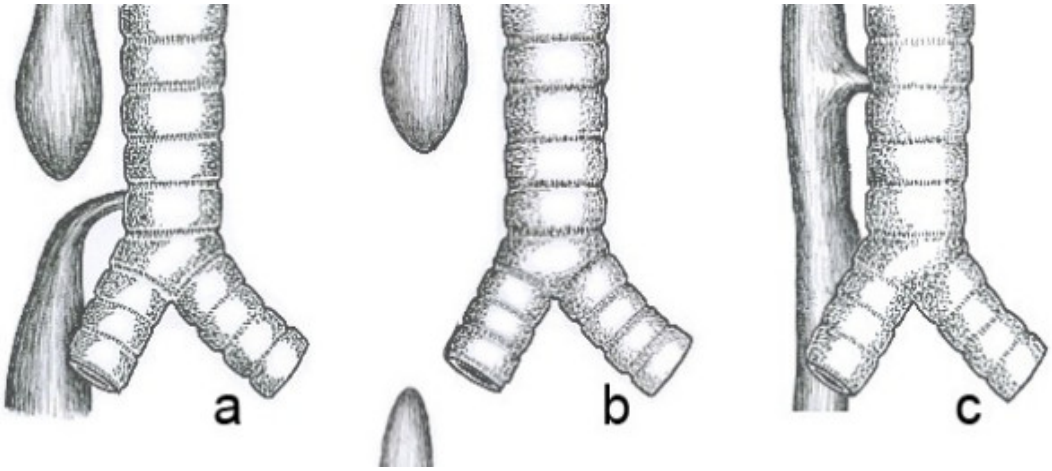


Figure 7. Common anatomical types of oesophageal atresia. a) Oesophageal atresia with distal tracheooesophageal fistula (86%). b) Isolated esophageal atresia without tracheooesophageal fistula (7%). c) H-type tracheooesophageal fistula (4%) (Spitz L. Oesophageal atresia. Orphanet J Rare Dis. 2007 May 11;2:24. doi: 10.1186/1750-1172-2-24. PMID: 17498283; PMCID: PMC1884133.) [33]

1. Oesophageal atresia with distal tracheooesophageal fistula (86% Vogt 111b. Gross C)

This is the most common variety in which the proximal oesophagus, which is dilated, and the muscular wall thickened ends blindly in the superior mediastinum at about the level of the third or fourth thoracic vertebra. The distal oesophagus, which is thinner and narrower, enters the posterior wall of the trachea at the carina or more commonly one to two centimetres more proximally in the trachea. The distance between the blind proximal oesophagus and the distal tracheooesophageal fistula varies from overlapping segments to a wide-gap. Very rarely the distal fistula may be occluded or obliterated leading to the misdiagnosis preoperatively of an isolated atresia.

2. Isolated oesophageal atresia without fistula (7%, Vogt 11, Gross A)

The proximal and distal oesophagus end blindly without any connection to the trachea. The proximal oesophageal segment is dilated and thick-walled and usually ends higher in the posterior mediastinum at around the second thoracic vertebra. The distal oesophagus is short and ends a variable distance above the diaphragm. The

distance between the two ends will determine whether a primary repair is feasible (rarely) or a whether a delayed primary anastomosis or an oesophageal replacement should be performed. It is important to exclude a proximal tracheooesophageal fistula in these cases.

3. Tracheooesophageal fistula without atresia (4%, Gross E)

There is a fistulous connection between an anatomically intact oesophagus and trachea. The fistulous tract may be very narrow or 3–5 mm in diameter and is commonly located in the lower cervical region. They are usually single but two and even three fistulas have been described.

4. Oesophageal atresia with proximal tracheooesophageal fistula (2%, Vogt III and Gross B)

This rare anomaly needs to be distinguished from the isolated variety. The fistula is not at the distal end of the upper pouch but is sited 1–2 cm above the end on the anterior wall of the oesophagus.

5. Oesophageal atresia with proximal end distal tracheooesophageal fistula (<1%m Vogt IIIa, Gross D)

In many of these infants the anomaly was misdiagnosed and managed as proximal atresia and distal fistula. As a result of recurrent respiratory infections, investigations carried out revealed a tracheooesophageal fistula, previously mistaken for a recurrent fistula. With the increasing use of preoperative endoscopy (bronchoscopy and/or oesophagoscopy) early recognition of the "double" fistula is made and total repair performed at the initial procedure. If the proximal fistula is not identified preoperatively, the diagnosis should be suspected by a large gas leak emanating from the upper pouch during the fashioning of the anastomosis.

Clinical description and diagnosis

Delaying the diagnosis until the infant presents with coughing and choking during the first feed is no longer acceptable in modern paediatric practice.

The diagnosis of oesophageal atresia may be suspected prenatally by the finding of a small or absent fetal stomach bubble on ultrasound scan performed after

the 18th week of gestation. Overall the sensitivity of ultrasonography is 42% but in combination with polyhydraminos the positive predictive value is 56%. Polyhydraminos alone is a poor indication of oesophageal atresia (1% incidence). Available methods of improving the prenatal diagnostic rate include ultrasound examination of the fetal neck to view the blind-ending upper pouch and to observe fetal swallowing and magnetic resonance imaging.

The newborn infant of a mother with polyhydramnios should always have a nasogastric tube passed soon after delivery to exclude oesophageal atresia. Infants with oesophageal atresia are unable to swallow saliva and are noted to have excessive salivation requiring repeated suctioning. At this stage, and certainly before the first feed, a stiff wide-bore (10–12 French gauge) catheter should be passed through the mouth into the oesophagus. In oesophageal atresia the catheter will not pass beyond 9–10 cm from the lower alveolar ridge. A plain X-ray of the chest and abdomen will show the tip of the catheter arrested in the superior mediastinum (T 2–4) while gas in the stomach and intestine signifies the presence of a distal tracheooesophageal fistula (Fig 8). The absence of gastrointestinal gas is indicative of an isolated atresia. A fine bore catheter may curl up in the upper pouch giving the false impression of an intact oesophagus or rarely it may pass through the trachea and proceed distally into the oesophagus through the fistula. The X-ray may reveal additional anomalies such as a "double bubble" appearance of duodenal atresia, vertebral or rib abnormalities.



Figure 8. Plain X-ray of the chest and abdomen showing the radio-opaque tube in the blind upper oesophageal pouch. Air in the stomach indicates the presence of a distal tracheooesophageal fistula. (Spitz L. Oesophageal atresia. Orphanet J Rare Dis. 2007 May 11;2:24. doi: 10.1186/1750-1172-2-24. PMID: 17498283; PMCID: PMC1884133.) [33]

Associated anomalies

Over 50% of infants with oesophageal atresia have one or more additional anomalies. The systems affected are as follows:

Cardiovascular 29%

Anorectal anomalies 14%

Genitourinary 14%

Gastrointestinal 13%

Vertebral/skeletal 10%

Respiratory 6%

Genetic 4%

Other 11%

There is an increased incidence of associated anomalies in pure atresia (65%) and a lower incidence in H-type fistula (10%).

The VATER association first described by Quan and Smith in 1973 consists of a combination of anomalies including **v**ertebral, **a**norectal, **t**racheoo**e**sophageal and **r**enal or radial abnormalities. This association was later expanded as the VACTERL association to include **c**ardiac and **l**imb defects.

Other associations which may include oesophageal atresia are the CHARGE association (coloboma, heart defects, atresia choanal, retarded growth and development, genital hypoplasia and ear deformities), POTTER'S syndrome (renal agenesis, pulmonary hypoplasia, typical dysmorphic facies) and SCHISIS association (omphalocoele, cleft lip and/or palate, genital hypoplasia). Genetic defects associated with oesophageal atresia include Trisomy 21 and 18, and 13q deletion. Of the cardiac anomalies, the most common are ventricular septal defect and tetralogy of Fallot. Major cardiac malformations are one of the main causes of mortality in infants with oesophageal atresia.

The vertebralanomalies in oesophageal atresia are mainly confined to the thoracic region and are responsible for later development of scoliosis. The claim that the presence of 13 ribs is associated with long-gap atresia has not been substantiated. Of the gastrointestinal anomalies, the most frequently encountered are duodenal atresia and malrotation, while there is an increased incidence of pyloric stenosis. Miscellaneous anomalies include cleft lip and palate, omphalocoele, lung abnormalities, choanal atresia and hypospadias.

Aetiology

The aetiology of oesophageal atresia is likely to be multifactorial and remains unknown.

Embryology

The mechanism that underlie tracheooesophageal malformations are still unclear, however, the development of reproducible animal models of these anomalies has allowed detailed analysis of the various stages of faulty organogenesis. By contrasting these stages with normal development, it has been

possible to identify key developmental processes that may be disturbed during embyogenesis.

It is generally accepted that the respiratory primordium appears as a ventral evagination on the floor of the post-pharyngeal foregut at the beginning of the fourth week of gestation and that the primitive lung buds are located at the caudal end of this evagination. During a period of rapid growth, the ventrally placed trachea becomes separated from the dorsally placed oesophagus. One theory postulates that the trachea becomes a separate organ as a result of rapid longitudinal growth of the respiratory primordium away from the foregut. An alternative theory is that the trachea initially grows as part of an undivided foregut and then becomes a separate structure as a result of a separation process that starts at the level of the lung buds and proceeds in a cranial direction. This process is associated with a precise temporospatial pattern of expression of the key developmental gene *Sonic hedgehog* (*Shh*) and members of its signalling cascade. A precise ventral-to-dorsal switch in foregut *Shh* expression is itself propagated cranially, ahead of tracheooesophageal separation. The separating foregut epithelium is marked by increased numbers of cells undergoing programmed cell death. Theories of abnormal organogenesis reflect the theories of normal development and are largely based on evidence from the Adriamycin rat model of oesophageal atresia (OA) and tracheooesophageal fistula (TOF) and a more recently described mouse model. In sporadic cases of oesophageal atresia, the likely cause is an insult that occurs during the narrow gestational window of tracheooesophageal organogenesis; this is the basis of the Adriamycin animal models. Most studies suggest that the primary defect is the persistence of an undivided foregut, either as a result of failure of tracheal growth or failure of the already specified trachea to physically separate from the oesophagus. An alternative theory suggests that atresia of the proximal oesophagus is the primary event and that the malformation results secondarily from the establishment of continuity between the trachea and the stomach/distal esophagus (tracheooesophageal fistula).

The presence of associated malformations could provide clues as to the possible aetiology of oesophageal atresia. Such malformations are present in 50% of cases and can occur in distinct patterns. These are non-random associations rather than syndromes because the presence of anomalies in one system makes it more likely that defects exist in another. One of the best-described associations is the **VACTERL** association, which comprises **v**ertebral, **a**norectal**, c**ardiac, **t**racheo-o**e**sophageal, **r**enal and **l**imb abnormalities. The pattern of these associations is likely to be dictated by the timing of a possible insult that affects multiple morphogenetic events. In the rat model, Adriamycin exposure affects a number of systems broadly consistent with the VACTERL association. The insult could act by transiently disturbing a specific developmental signalling pathway. In the mouse, loss of function mutations for genes of the *Shh* pathway lead to a spectrum of anomalies that is very similar to those of VACTERL, implicating the pathway in the embryogenesis of the malformations. In the human, the evidence is less conclusive, with the *Shh* mutation leading to holoprosencephaly, a malformation not associated with oesophageal atresia. Tracheooesophageal fistula and other features of VACTERL have however been described in patients with a mutation for *GL13*.

Chromosomal abnormalities such as trisomies (18 and 21) and deletions (22q11 and 17q22q23.3) are known to be associated with oesophageal atresia and have been reported in up to 6% of patients who have associated malformations in other systems.

Pathophysiology

The motility of the oesophagus is always affected in oesophageal atresia. The disordered peristalsis more commonly involves the distal oesophageal segment. Whether the motility disorder is primarily due to abnormal innervation as evidenced by an abnormality in neuropeptide distribution or secondary to vagal nerve damage occurring during the surgical repair remains uncertain. The resting pressure in the whole oesophagus is significantly higher than in normal patients and the closing pressure of the lower oesophageal sphincter is reduced.

The trachea is also abnormal in oesophageal atresia. The abnormality consists of an absolute deficiency of tracheal cartilage and an increase in the length of the transverse muscle in the posterior tracheal wall. When severe, these abnormalities result in tracheomalacia with collapse of the trachea over a 1–2 cm segment in the vicinity of the fistula.

Genetic counselling

The overwhelming majority of cases of oesophageal atresia are sporadic/non-syndromic, although a small number within this non-familial group are associated with chromosomal abnormalities. Familial/syndromic cases of oesophageal atresia are extremely rare. Oesophageal atresia is 2 to 3 times more common in twins. The overall risk of oesophageal atresia recurrence in a sibling of an affected child is about 1%.

Management

A. Preoperative

Once the diagnosis of oesophageal atresia has been established, the infant will need to be transferred from the place of birth to a regional paediatric surgical centre. A suction catheter, preferably of the double lumen type (Replogle catheter No.10 French gauge), is placed in the upper oesophageal pouch to suction secretions and prevent aspiration occurring during transfer. The infant is placed on its side in the portable incubator while monitoring the usual vital signs. Vascular access should be provided as a precautionary measure but intravenous fluid administration is not usually necessary if the condition has been diagnosed within a short period after birth and transfer is carried out within the first day of life.

The preterm infant with respiratory distress requires special attention. Clearly there is a need for endotracheal intubation and mechanical ventilation. In addition, there is the added risk of gastric over-distension and rupture of the stomach due to escape of respiratory gases down through the distal fistula into the stomach due to the increased pulmonary resistance. This sequence of events can be minimised by positioning the end of the endotracheal tube distal to entry of the tracheooesophageal fistula and by applying gentle low pressure ventilation.

Upon arrival at the Neonatal Surgical Centre, the diagnosis of oesophageal atresia must be confirmed.

All infants with oesophageal atresia should have an echocardiogram prior to surgery. The ECHO will define any structural anomaly of the heart or great blood vessels and occasionally may indicate a right-sided aortic arch which occurs in 2.5% of cases. In these cases, a magnetic resonance imaging study is the method of choice for accurate confirmation of the diagnosis of the right aortic arch and will determine the side of approach for the operative repair. In around 25% of infants with tetralogy of Fallot, the aortic arch will be on the right side. The infant with a major cardiac anomaly resulting in severe cyanotic episodes will need to undergo a shunting procedure prior to correction of the oesophageal atresia. For example, in complicated tetralogy of Fallot, a Blalock-Taussig systemic-to-pulmonary artery shunt will alleviate the cyanotic attacks and permit repair of the oesophageal atresia to take place a day or two later. Congestive cardiac failure will necessitate medical management in the first instance.

B. Risk categorisation and prognosis (outcome)

In 1962, Waterston *et al*. proposed a classification of infants born with oesophageal atresia into three groups "with different chances of survival". The classification based on birth weight, associated anomalies and pneumonia comprised:

Group A Over 5 1/2 lb (2500 g) birth weight and well

Group B 1. Birth weight 4–5 1/2 lb (1800–2500 g) and well

2. Higher birthweight, moderate pneumonia and congenital anomaly.

Group C 1. Birth weight under 4 lb (1800 g).

2. Higher birth weight and severe pneumonia and severe congenital anomaly.

This classification was applied to 113 cases treated at Great Ormond Street Hospital from 1951 to 1959. Of 38 infants in Group A, all but two survived (95%), of 43 in Group B 29 (68%) survived while only two (6%) of the 32 in Group C survived.

During the subsequent 40 years there has been a steady improvement in the overall survival rate due to early diagnosis and prompt referral, improvements in preoperative care and diagnosis and treatment of associated anomalies, advances in anaesthetic techniques and sophisticated neonatal intensive care. Applying the Waterston classification to a series of 357 infants with oesophageal atresia treated at Great Ormond Street from 1980 to 1992, the results were Group A, 153 of 154 survived (99%), 72 of 76 in Group B survived (95%) and 101 of 142 in Group C survived (71%). It became obvious that a new risk classification system was needed which was more relevant to the modern era. The new risk classification concerned birth weight and associated cardiac malformations which were previously identified as being responsible for most of the mortality.

The Spitz classification for survival in oesophageal atresia is:

Group I Birth weight over 1500 g **with no** major cardiac anomaly.

Group II Birth weight less than 1500 g **or** major cardiac anomaly

Group III Birth weight less than 1500 g **PLUS** major cardiac anomaly.

Major cardiac anomaly was defined as either cyanotic congenital heart disease that required palliative or corrective surgery or non-cyanotic heart anomaly that required medical or surgical treatment for cardiac failure.

C. Selection for non-treatment

Infants with Potter's syndrome (bilateral renal agenesis) and trisomy 18 which is fatal in the first year of life in over 90% of affected infants should be offered the option of no active treatment. Similarly, infants with totally uncorrectable major cardiac defects or with Grade IV intraventricular haemorrhage should be considered for non-operative management.

D. Emergency ligation of the distal tracheooesophageal fistula

Generally, the operative correction of an oesophageal atresia is **not** regarded as an emergency procedure. The one exception is the preterm infant with severe respiratory distress syndrome requiring ventilatory support. Ventilatory gases escaping down the distal fistula result in gastric distension which further impedes

respiratory function. With progressively increasing gastric distension, the stomach may eventually rupture causing a tension pneumoperitoneum which renders ventilatory support even more difficult.

The traditional method of dealing with this sequence of events was to perform an emergency gastrostomy. Unfortunately, most of these infants died because of further dramatic deterioration of ventilation due to the sudden reduction in intragastric pressure allowing free flow of respiratory gases through the tracheooesophageal fistula.

Many manoeuvres have been advocated to alleviate the problem including positioning of the endotracheal tube distal to the fistula. However, if the fistula is sited at the level of the carina, this manoeuvre impossible to achieve. Others have advocated blocking of the fistula by a Fogarty catheter passed at bronchoscopy. The affected infants are usually preterm and in critical respiratory status. The smallest calibre bronchoscope will not permit ventilation while manoeuvring a Fogarty catheter into the distal oesophagus and in a cyanotic infant this will aggravate the condition and further exacerbate the hypoxia.

The operation is performed under general endotracheal anaesthesia with dependable vascular access and employing gentle ventilatory pressure so as not to produce gastric distension.

1. ***Intraoperative endoscopy*** – Preliminary bronchoscopy may be carried out to define the site of entry of the distal tracheooesophageal fistula and to assess the presence of tracheomalacia. An alternative is to carry out an oesophagoscopy to define the length of the upper oesophagus and to exclude an upper pouch fistula which is more common with isolated oesophageal atresia.

2. ***Position*** – The infant is placed on the left side with the right arm across the front of the chest for a right postero-lateral thoracotomy.

3. ***Incision*** – A curved incision centred 1 cm below the inferior angle of the scapula approximately 5–6 cm long is made. The muscle of the chest wall may either be split or divided with electrocautery taking care to preserve the long thoracic nerve

supply to serratus anterior. The thorax is opened through the 4th or 5th intercostal space by dividing the intercostal muscles or by entry through the bed of the unresected rib.

4. ***Extrapleural approach*** – has the advantage of conferring protection of the pleural space in the event of an anastomotic leak. Commencing posteriorly, the pleura is gently freed off the chest wall using blunt dissection. The dissection proceeds into the mediastinum to provide good access to the oesophagus. The extrapleural approach is slightly more time-consuming and has theoretically advantages over the transpleural approach still used by many surgeons.

5. ***Exposure of the esophageal segments*** – The azygos vein is the first structure encountered on entering the mediastinum. The azygos vein is gently mobilised and divided between ligatures to expose the esophagus. The distal oesophagus usually lies directly deep to the azygos vein and is identified by the vagus nerve coursing over its anterior aspect. The distal oesophagus can be seen to distend with each inspiration but it is still advisable to gently compress the lumen of the distal oesophagus while the anaesthetist applies increased respiratory pressure. This manoeuvre allows the right lung to expand thus ensuring that the structure compressed is not the right main bronchus. The blind upper oesophageal pouch is identified high up in the mediastinum aided by the anaesthetist applying pressure on the oro- or naso-oesophageal tube.

6. ***Repair of the anomaly*** – The distal oesophagus is mobilised circumferentially just distal to the entry of the tracheooesophageal fistula into the trachea and a soft rubber sling (neoloop) is placed around the oesophagus. A marking seromuscular suture is placed in the lateral wall of the distal oesophagus to assist with orientation. The distal oesophagus is dissected to the level of the fistula and the upper and lower extent of the fistula is marked with fine non-absorbable sutures before dividing the oesophagus just distal to the fistula. The tracheal side of the fistula is closed with interrupted 5.0 sutures to achieve an air-tight closure. The tracheal closure may be tested by instilling warm saline over the suture line while the anaesthesiologist expands the lungs. Having identified the proximal blind

oesophageal pouch a figure of eight suture is inserted at its tip to aid in its mobilisation. The proximal oesophagus should be mobilized sufficiently to produce a tension-free anastomosis. Laterally, anteriorly and superiorly the mobilisation is easily achieved, but medially there are fibrous adhesions to the trachea which require sharp dissection. It is important to remain close to the oesophageal wall and avoid entering the trachea during the mobilisation. The end of the upper pouch is now opened to reveal the mucosal surface. An end-to-end anastomosis between the proximal and distal oesophagus is fashioned using interrupted full-thickness fine sutures. The posterior half of the anastomosis is completed first with sutures tied on the mucosal surface. If there is a wide gap, the distal oesophagus can be mobilised safely well down towards the diaphragm. The sutures in wide gap cases should be placed untied in the posterior half of the anastomosis and gently but firmly brought together so as to distribute the tension equally over a larger area. The sutures are then tied sequentially while the tension is maintained. It is almost always possible to achieve an anastomosis when there is a distal fistula present. The anterior half of the anastomosis is completed with interrupted full-thickness sutures. Just prior to the final suture being tied a transanastomotic fine-calibre naso-gastric tube may be passed. This allows gastric decompression in the early postoperative course and provides a route for early enteral feeding.

7. ***Methods to overcome a wide gap*** – Various manoeuvres have been proposed to overcome a wide gap but in our experience a very tense anastomosis can be achieved in most cases and if the infant is subsequently electively paralyzed and mechanically ventilated for approximately 5 days postoperatively, the anastomosis will heal without leakage. Others have proposed tubularisation of the upper pouch after creating a flap, circular myotomy of the upper pouch or abandoning any attempt at initial primary anastomosis awaiting delayed primary anastomosis 6–12 weeks later.

8. ***The thoracotomy incision*** is now closed with or preferably without intercostal drainage especially if the procedure has been totally extrapleural and a technically satisfactory anastomosis has been performed.

The procedure can be carried out thoracoscopically but this requires advanced skills in minimal invasive surgery. The accumulated results in a multi-institutional review of 104 infants showed comparable outcomes compared with the open thoracotomy procedure.

Management of isolated oesophageal atresia

The diagnosis of isolated oesophageal atresia without a fistula should be suspected when on the initial radiograph there is **no** gas in the abdomen ("gasless abdomen"). There is the remote possibility of there being an occluded fistula either by mucus or a complete obstruction. A preoperative bronchoscopy should always be performed in this situation to exclude an upper pouch fistula. The overall incidence of upper pouch fistula in oesophageal atresia is 2.5% but it is 3–4 times more common in isolated atresia.

Once the diagnosis of isolated atresia is made, the next step is to perform a feeding gastrostomy and to estimate the extent of the gap between the proximal and distal oesophagus. The stomach in isolated atresia is very small and it may not be possible to anchor the stomach to the anterior abdominal wall. To avoid tension and necrosis of the stomach it is preferable to leave the stomach at the back of the abdomen and to allow the gastrostomy tube to traverse the peritoneal cavity. The gap between the oesophageal ends is measured by injecting sufficient radioopaque contrast into the stomach to allow it to enter the distal pouch or by passing a bougie (Hegar dilator or urethral sound) through the gastrostomy site into the distal oesophagus. With a radio-opaque catheter in the proximal oesophagus it is now possible to measure, in terms of vertebral body heights, the gap between the two ends. Where the gap is less than two vertebrae, an attempt should be made at immediate primary anastomosis. For a gap of three to six vertebra, delayed primary repair should be planned. Over a period of up to 12 weeks while maintaining suction to the upper pouch and feeding by gastrostomy, the gap gradually narrows. Regular monitoring of the gap using metal sounds in the distal oesophagus is undertaken. When the two ends can be approximated or overlapped, an attempt at delayed primary anastomosis is undertaken.

When the gap between the two ends is greater than six vertebrae, the options are to proceed as above and attempt delayed primary repair, to apply graduated tension on the oesophageal ends over a period of 6–10 days and then perform a primary anastomosis, or to abandon any attempt to retain the esophagus and to perform a cervical oesophagostomy and replace the oesophagus at a later date.

The incidence of significant gastrooesophageal reflux and the subsequent need for an antireflux procedure is much higher following anastomosis under tension.

Management of the H-type fistula

An H-type tracheooesophageal fistula is suspected when the infant experiences coughing with feeds or suffers from recurrent respiratory infections. The diagnosis is established on contrast oesophagogram (ideally a tube oesophagogram) and confirmed at bronchoscopy/oesophagoscopy. It is extremely valuable to have a ureteric catheter passed across the fistula at preliminary bronchoscopy immediately prior to surgery. The operative division of the H fistula is most frequently performed via a low cervical approach.

Postoperative management

When the oesophageal anastomosis has been performed under tension, the infant is electively paralysed and mechanically ventilated for five days postoperatively. With this regimen there have been no major disruptions and only a few minor leakages which have healed spontaneously [60]. In all other instances, regular pharyngeal suction is necessary for the first few postoperative days. The suction catheter should be clearly marked to prevent the tube being passed to the site of the anastomosis and causing damage. Transanastomotic nasogastric feeds may be commenced on the second or third postoperative day and when the infant is swallowing saliva, oral feeds may be started. We do not regularly perform a follow-up contrast study but if there is any doubt as to the integrity of the anastomosis a water-soluble contrast study is carried out.

Complications

Early: anastomotic leakage

anastomotic stricture

recurrent fistula

Late: gastrooesophageal reflux

tracheomalacia

dysmotility

Anastomotic leaks occur in 15–20% of patients but in only one-third or less is there a major disruption. Major leaks occur in the early postoperative period (<48 hours) and present with life-threatening tension pneumothorax. Emergency treatment consists of tube thoracostomy following which either suction is applied to the intercostal drain while awaiting healing of the anastomosis or an early thoracotomy is carried out with the intention of repairing the anastomosis or, if there has been a complete disruption, abandoning any attempt at re-anastomosis and performing a cervical oesophagostomy and closing the distal oesophagus pending oesophageal replacement.

Minor leaks may be detected on the "routine" contrast study usually performed on the 5–7th day postoperatively. These will all seal spontaneously but there is an increased incidence of later stricture formation.

Anastomotic strictures develop in 30–40% of cases most of which will respond to one or two dilatations. Risk factors which have been implicated in stricture formation include anastomotic tension, anastomotic leakage and gastrooesophageal reflux. With meticulous handling of the oesophageal ends, preservation of the blood supply and careful inclusion of mucosa in each and every suture of the anastomosis, strictures can be kept to a minimum.

Endoscopic dilatation of the stricture can be carried out either at rigid oesophagoscopy using semi-rigid bougies (Savary-Gillard) of progressively larger calibre or by balloon dilatation introduced either at fluoroscopy or during flexible endoscopy. In either event, the balloon is carefully positioned under fluoroscopy at the centre of the stricture and contrast is gradually instilled under pressure until the

"waisting" at the stricture is abolished. At the end of the procedure, contrast is introduced into the oesophagus to ensure that there has been no perforation and to establish the effectiveness of the dilatation.

Only rarely is it necessary to resort to resection of an intractable anastomotic stricture.

Recurrent tracheooesophageal fistula

The incidence of recurrent tracheooesophageal fistula is between 5–14%. A recurrent fistula should be suspected if the infant manifests respiratory symptoms (coughing during feeds, apnoeic or cyanotic episodes) or has recurrent respiratory infections after "successful" repair of the oesophageal atresia. Urgent investigations must be undertaken to positively diagnose or exclude the possibility of a recurrent fistula.

The diagnosis may be suspected on the plain chest radiograph which shows an air oesophagogram. Routine contrast oesophagogram has a low yield and the most useful investigation is a prone tube cine-oesophagogram when water-soluble contrast is slowly instilled into the oesophagus while the nasogastric tube is gradually withdrawn from the stomach to the level of the pharynx.

Bronchoscopic examination will reveal the recurrent fistula at the site of the original tracheooesophageal fistula. It is essential to pass a fine ureteric catheter across the fistula into the oesophagus and to view the catheter in the oesophagus at endoscopy. The passage of a catheter across the fistula is also an essential preliminary step in the operative correction of the recurrent fistula. At surgery it is useful to define the oesophagus above and below the site of the fistula and to insert stay sutures at both the oesophageal and tracheal ends of the fistula before it is divided. The resulting defects in the trachea and oesophagus are closed with firm non-absorbable interrupted full-thickness sutures.

Gastrooesophageal reflux (GOR)

GOR is common in all infants following repair of oesophageal atresia – significant reflux occurs in around 40% of cases, about half of whom will require surgical management. GOR is more common following anastomosis under tension

and after delayed primary repair. It has been implicated in the pathogenesis of anastomotic stricture. However, reflux can still occur following uncomplicated, tension-free anastomosis where the distal oesophagus has remained virtually undisturbed. Incompetence of the lower oesophageal sphincter mechanism may be due to a primary neurgenic disturbance inherent in the development of the oesophageal atresia or to technical factors involved in the repair or it may be due to shortening of the intraabdominal oesophagus and/or abolishment of the gastrooesophageal angle of His.

Symptoms of GOR are similar to those of recurrent fistula with acute or chronic respiratory problems but also include recurrent vomiting and stricture formation. The diagnosis can be established on contrast swallow, pH monitoring and endoscopy and biopsy of the distal oesophagus. Stricture formation at the anastomosis which is resistant to repeated dilatations often resolves spontaneously once the GOR is corrected.

Antireflux medication including gastric acid suppression is only successful in about half the cases. Surgical treatment is problematic given the inherent dysmotility present in the distal oesophagus. A fundoplication may in itself produce a functional obstruction at the gastrooesophageal junction. The failure rate of fundoplication carried out in the first three months of life is excessively high. With regard to the nature of the wrap, there are advocates for partial (Thal) as well as for a short, floppy wrap (Nissen type).

Tracheomalacia

Tracheomalacia may be defined as a structural and function weakness of the trachea resulting in partial and occasionally complete respiratory obstruction. The structural abnormality comprises a deficiency in the cartilage in the tracheal rings and an increase in the length of the transverse muscle. The result is that the airway collapses during expiration causing expiratory stridor which varies in severity from a hoarse barking type of cough, to recurrent respiratory infection to acute life-threatening episodes of cyanosis or apnoea. The incidence of tracheomalacia is around 10% and about half will require surgical correction. The area of collapse seen

at bronchoscopy is usually restricted to the trachea at the level of the entry of the distal tracheooesophageal fistula, but it can be more extensive. It usually presents within the first few months of life and generally coincides with a period of rapid weight gain. The diagnosis is made at bronchoscopy or cine-bronchography.

If the respiratory obstruction is severe enough to present with acute life-threatening events, treatment needs to be carried out promptly and certainly before the infant leaves hospital. The definitive treatment consists of aortopexy in which the ascending and arch of the aorta are elevated anteriorly towards the sternum. Via a left anterolateral thoracotomy or a median sternotomy, the arch of the aorta is exposed after excising the lateral lobe of the thymus. Three sutures of non-absorbable material are passed several times through the adventitia of the arch and ascending aorta and then through the sternum. The sutures are left untied until all three are in position and then with the assistant depressing the sternum, all the sutures are tied. On releasing the sternum, the aortic arch is pulled forward and in doing so, allows space for the trachea to expand.

The result is usually dramatic with immediate resolution of the obstruction to the air passages.

Dysmotility

Dysmotility affects the distal oesophagus particularly in relation to abnormal coordination of contractions which in fact can be seen on contrast studies of the oesophagus. The intrinsic innervation of the distal oesophagus has been shown to be abnormal in the fetal rate model and affects both excitatory (SP-labelled) and inhibitory (VIP-labelled) intramural nerves. The dysmotility is a major factor in the long-term swallowing problems encountered in these children. The patients are advised to take fluids liberally with meals and to avoid foodstuffs which exacerbate the problems especially doughy white bread and cakes.

Respiratory function

During infancy and for the first three years of life, patients with oesophageal atresia suffer increased frequency of respiratory infections. The respiratory problems tend to resolve with time. The tendency to respiratory infections has variously been

attributed to oesophageal dysmotility and/or gastrooesophageal reflux with recurrent aspiration or to a primary respiratory abnormality. Emery and Haddadin suggested that the presence of nonciliated squamous epithelium found in the trachea of 80% of infants with oesophageal atresia could seriously impair the anti-infection mechanism and contribute to repeated attacks of bronchitis. Respiratory function tests carried out in infants soon after repair of the oesophageal atresia have shown abnormal patterns of air flow and also increased airway resistance.

Congenital oesophageal stenosis due to tracheobronchial remnants in the distal oesophagus in an infant with esophageal atresia is a rare but well documented phenomenon [74,75]. It is thought to arise as a result of defective separation of the trachea from the oesophagus. It may be recognised at the time of the initial repair of the oesophageal atresia when passage of a catheter into the stomach is impeded. Alternatively, symptoms develop quite early with dysphagia and regurgitation of solid food. Contrast radiography shows a short well-defined narrowing in the distal oesophagus which needs to be differentiated from a reflux stricture. Balloon dilatation confirms the short defined narrowing which fails to widen on inflation of the balloon. Endoscopy reveals normal mucosa to the site of the stenosis which, if composed of a complete cartilaginous ring, fails attempts at dilatation. Treatment consists of resection and end-to-end anastomosis. A useful step at the time of surgery is to pass a flexible gastroscope to the level of the stenosis to highlight the precise level of resection as the stenosis is frequently ill-defined from the external surface.

2.Diaphragmatic Hernia

Diaphragmatic hernia is a birth defect where there is a hole in the diaphragm (the large muscle that separates the chest from the abdomen). Organs in the abdomen (such as intestines, stomach, and liver) can move through the hole in the diaphragm and upwards into a baby's chest. When an organ pushes through the hole, it is called a hernia. A diaphragmatic hernia can prevent the baby's lungs from developing completely, causing breathing difficulties for the baby at birth.

Researchers estimate that about 1 in every 3,600 babies is born with diaphragmatic hernia in the United States. About half of all newborns who have diaphragmatic hernia also have other conditions, including birth defects of the brain, heart, and intestines.[2]

While the causes of diaphragmatic hernia in most infants are unknown, researchers believe that some instances of diaphragmatic hernia may be caused by abnormalities in the baby's genes.

Clinical Description

Congenital diaphragmatic hernia (CDH) refers to a developmental defect of the formation of the diaphragm that, in most individuals, is evident at birth. CDH is characterized by: (1) incomplete formation/muscularization of the diaphragm resulting in absence or deficiency of the diaphragm, or (2) eventration resulting in elevation of a portion of the diaphragm that is thinned as a result of incomplete muscularization. The prevalence of CDH is estimated at 3-3.6/10,000 live births.

Infants with CDH often present in the neonatal period with severe respiratory distress.

Presentation after infancy, occurring in 5%-10% of affected individuals, includes respiratory distress, such as from pleural effusion due to entrapment of the bowel in the chest, or gastrointestinal distress, such as abdominal pain from chronic or intermittent intestinal obstruction. About 1% of individuals are completely asymptomatic and the defect is discovered incidentally on imaging studies.

At least 10% of individuals reherniate following initial surgical repair; the risk is considerably greater among those whose hernia repair required a prosthetic patch.

Although CDH is classified into several types, distinction among hernias can be problematic. An anatomic depiction of the normal diaphragm is presented in Figure 9 and anatomic descriptions of diaphragmatic defects are presented in Figure 10.

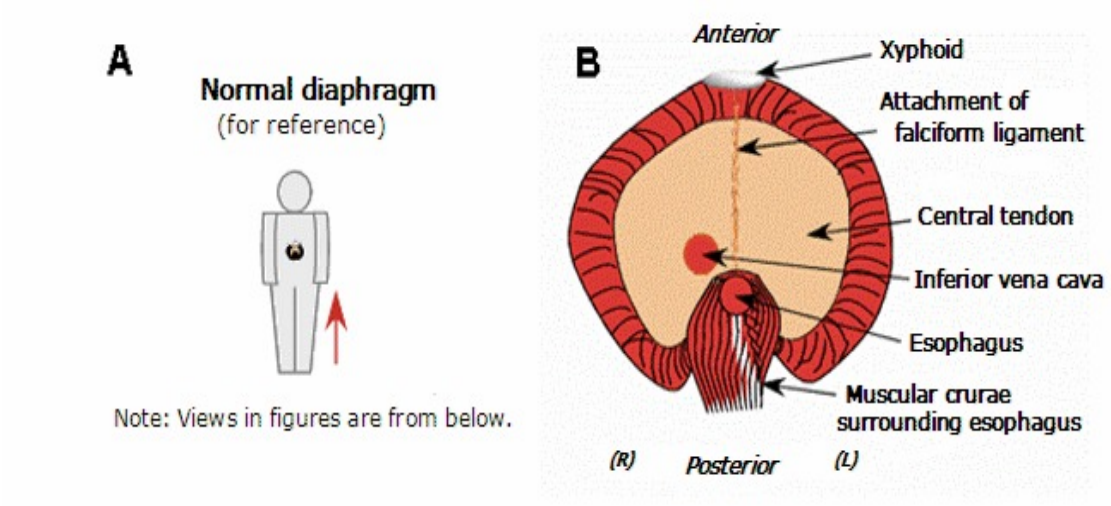


Figure 9 A. Normal diaphragm (for reference) B. View of normal diaphragm from below

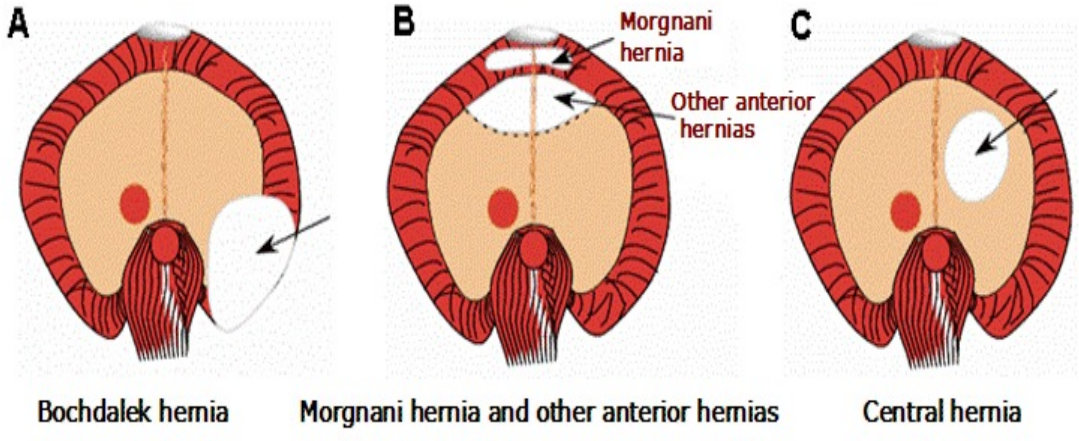


Figure 10. View of diaphragmatic defects from below: A. Bochdalek hernia

B. Morgnani hernia and other anterior hernias C. Central hernia

(Longoni M, Pober BR, High FA. Congenital Diaphragmatic Hernia Overview. 2006 Feb 1 [Updated 2020 Nov 5]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1359/)[36]

Posterolateral (Bochdalek) hernia of the diaphragm is often accompanied by herniation of the stomach, intestines, liver, and/or spleen into the chest cavity. An

extremely large defect, or apparent absence of the hemidiaphragm, is called agenesis of the diaphragm; this defect probably represents the severe end of the Bochdalek hernia spectrum.

Posterolateral hernias comprise approximately 80%-90% of all CDH and appear to fall into two types:

A diaphragmatic defect accompanied by an absent or extremely deficient rim of posterior and lateral musculature.

A diaphragmatic defect with an intact rim of posterior and lateral musculature

About 85% of Bochdalek hernias occur on the left side, about 10% on the right, and approximately 5% are bilateral.

Non-posterolateral (non-Bochdalek) hernia. Anterior defects of the diaphragm can occur in the midline, on the left side, or the right side. Several distinct subtypes are described, with considerable overlap in the anatomic location.

Morgagni (Morgagni-Larrey) hernia is an anterior retrosternal or parasternal hernia that can result in the herniation of liver or intestines into the chest cavity. Morgagni hernias comprise approximately 2% of all CDH, are generally accompanied by a hernia sac, and often do not cause symptoms in the newborn period.

Other anterior hernias associated with pentalogy of Cantrell. These rare and severe types of hernia are found in individuals with pentalogy of Cantrell (which also includes defects in the supraumbilical midline abdominal wall, lower sternum, diaphragmatic pericardium, and heart).

Central hernia. This hernia is a rare diaphragm defect involving the central tendinous (i.e., amuscular) portion of the diaphragm. The entire rim of diaphragmatic musculature is present.

Diaphragmatic eventration is incomplete muscularization of the diaphragm resulting in a thin membranous sheet of tissue. It is difficult to estimate the frequency of eventration because it may coexist with and/or be misdiagnosed as a Bochdalek hernia. Severe diaphragmatic eventration is associated with pulmonary hypoplasia and respiratory distress during infancy. Milder degrees of diaphragmatic eventration

can present later in life with respiratory symptoms such as cough and pneumonias, or without symptoms, so that the diagnosis is made incidentally on chest x-ray. Increasingly, it is observed that eventration of the diaphragm and CDH can occur in the same individual and in different individuals with pathogenic variants in the same gene, suggesting that in some instances they share a common etiology.

Sac hernia. All hernia types can present with a sac (e.g., membranous sheet of tissue) covering the herniated abdominal contents. Animal models suggest that abnormal connective tissue, resulting in a localized amuscular region, rather than a primary defect in myogenic cells, is the embryogenetic basis of the hernia sac. The presence of a sac is often reported to be associated with a better prognosis. However, because a thin and redundant membranous diaphragm resulting from an eventration defect may represent a "sac," it is probable that eventration and "sac type" CDH diagnoses are often interchanged. For this reason, it is inaccurate to assume that the prognosis is better when a sac is visualized.

CDH can occur as either an isolated or complex anomaly:

Isolated. Observed in 50%-60% of probands; not associated with other major congenital anomalies

Complex. Observed in 40%-50% of probands and associated with other congenital anomalies:

Syndromic. Some individuals with complex CDH have a readily identifiable genetic syndrome or chromosome anomaly. Individuals with syndromic CDH are more likely to be diagnosed prenatally, have an affected first-degree relative (10%), and have a worse prognosis. More than 50% of individuals with syndromic CDH are born preterm and have low birth weight.

Complex CDH not attributed to a recognized syndrome. The remaining probands with complex CDH present with associated additional malformations (e.g., craniofacial, ocular, cardiovascular, central nervous system, limb, genitourinary) but do not have a recognizable syndromic phenotype.

Establishing the Diagnosis of CDH

Congenital diaphragmatic hernias are increasingly diagnosed prenatally; however, they are occasionally diagnosed in symptomatic neonates and even asymptomatic or mildly symptomatic children, teens, and adults.

Prenatal Diagnosis

Prenatal investigations by second-trimester two-dimensional (2D) ultrasound and/or MRI detect more than 60% of affected fetuses with right-sided defects and more than 80% of those with left-sided CDH (Canadian Pediatric Surgery Network [CAPSNet] database). Ultrasound demonstrates herniated viscera with or without liver in the fetal thorax, absence of the normal position of the stomach bubble below the diaphragm, and mediastinal shift. MRI is especially useful for the prenatal diagnosis of thoracic lesions that are atypical or complicated by multiple abnormalities and for assessing lung volumes. Estimates of fetal lung volume and descriptions of the amount of herniated viscera are often used as prognostic indicators.

Color flow Doppler can be used to:

Demonstrate abnormal positioning of the umbilical and portal veins, which are indicative of liver herniation;

Identify right-sided hernias, which can be difficult to detect on ultrasound examination because of the similar echogenicity of lung and liver.

Fetal MRI is increasingly being used to confirm the diagnosis of CDH during the second and third trimester as a complementary tool to ultrasound studies to better define fetal anatomy. The primary advantage of fetal MRI is the ability to measure total lung volume and allow a quantitative assessment of the liver mass herniated into the chest. A systematic review and meta analysis of observational studies suggests that fetal MRI can be used to predict neonatal survival with some caveats:

MRI has limited benefit early in gestation.

Lung measurements in individuals with CDH are not standardized.

Survival rates and treatment protocols from different centers are not comparable.

Postnatal Diagnosis

CDH can be detected in a neonate by:

Presence of a scaphoid abdomen, diminished breath sounds ipsilateral to the side of the hernia, and displacement of the heart sounds contralateral to the hernia.

Chest x-ray shows visible bowel gas above the diaphragm accompanied by a mediastinal shift.

Rarely, older children or adults with inconspicuous congenital diaphragmatic defects can be suspected because of respiratory or gastrointestinal symptoms including chronic cough, recurrent pulmonary infections, pleural effusions, pneumonia, or dysphagia. Intestinal obstruction and volvulus may be presenting symptoms, as abnormalities in the usual intestinal rotation during fetal development are common. Finally, some individuals may be diagnosed incidentally by plain chest radiographs or other imaging procedures. In these individuals, the degree of respiratory symptoms is correlated with the degree of pulmonary hypoplasia and can be limited.

Clinical Manifestations

Respiratory Compromise

Infants with CDH often present in the neonatal period with severe respiratory distress, occasionally after a stable period of 24-48 hours followed by acute respiratory distress. Breath sounds are diminished ipsilateral to the hernia. Almost all individuals with CDH have some degree of pulmonary hypoplasia. The pathogenesis of the pulmonary hypoplasia associated with CDH appears to have both a primary component (i.e., the hypoplasia occurs independent of the diaphragm defect) and a secondary component (i.e., arising from competition for thoracic space particularly in the lung ipsilateral to the hernia). Evidence for the presence of a primary defect in lung development arises mostly from studies in animal models, some of which show that the lung hypoplasia precedes the herniation of abdominal viscera.

Infants with CDH typically require mechanical ventilation and sometimes extracorporeal membrane oxygenation (ECMO) in the newborn period. Major respiratory complications include tracheobronchomalacia, pneumothorax, and

secondary lung infection (especially viral pneumonia) that could precipitate terminal respiratory failure even months after surgery. Many infants require ongoing oxygen supplementation and diuretics following surgical correction of CDH. Given the remarkable growth and recuperative capacity of the lung, these treatments can usually be discontinued within the first two years of life.

By early childhood, few children have respiratory symptoms at rest; however, formal testing in older children shows small airway obstruction and diminished blood flow on ventilation-perfusion (V-Q) scan, especially to the lung ipsilateral to the hernia. Reduced exercise tolerance can be a lifelong problem. Intermittent wheezing requiring bronchodilator use is common in people with CDH, and they are at risk for respiratory decompensation with intercurrent illness.

Pulmonary Hypertension

Abnormal pulmonary vascular development and function is a significant problem in infants with CDH. Guidelines for the diagnosis of pediatric pulmonary hypertension were set forth by the American Heart Association and American Thoracic Society, however, individuals with CDH present a unique challenge due to very young age at the onset of PH. The mechanism of pulmonary hypertension in CDH is not completely understood. The size of the pulmonary vascular bed is decreased in the hypoplastic lungs and the vasculature displays medial and adventitial thickening. Notable is the premature differentiation of smooth muscle cells into a contractile phenotype.

Failure of relaxation of the pulmonary vasculature at birth is a critical component leading to pulmonary hypertension in CDH and it is the consequence of abnormal expression of both vasoconstrictors and vasodilators in distinct but converging pathways.

Gastrointestinal

"Failure to thrive" with growth parameters less than the third centile of normal is common among infants with more significant pulmonary hypoplasia and/or a more prolonged hospitalization following surgical repair of CDH. Growth failure is caused, in large part, by oral aversion and feeding difficulties (often requiring

gastrostomy tube insertion for the first few years of life) and gastroesophageal reflux (frequently requiring pharmacotherapy and/or surgical fundoplication). Some infants and children require long-term high-calorie nutritional supplements.

Neurologic/Developmental

Reporting of neurodevelopmental outcomes is complicated by lack of standardization in terms of outcomes assessed, age of assessment, and metrics used. The main independent predictor of intellectual disability is ECMO treatment. Mild (44%) or severe (13%) delay in at least one domain at age one year has been reported. Later, intelligence is reported in the normal range for the majority of individuals with isolated CDH, but educational support in school is often warranted and behavioral issues such as struggling with concentration and attention are common. Preschool motor development scores in individuals with CDH are normal or mildly reduced, and improve with age; however, nonfocal neurologic abnormalities such as hypotonia are common, as are motor problems, especially in ECMO survivors. Nonspecific findings such as cortical atrophy, ventriculomegaly, and intracranial hemorrhage can be seen on neuroimaging studies.

Musculoskeletal

Chest asymmetry is found in as many as half of individuals with CDH. Pectus deformity, most often of the excavatum type, and scoliosis (≥10° Cobb's angle) are found in approximately 25% of individuals. These musculoskeletal abnormalities occur more often following repair of large diaphragmatic defects, possibly as a result of the extra tension exerted on the chest wall during surgical repair.

Sensorineural Hearing Loss

Sensorineural hearing loss (SNHL) has been found in 25% of individuals with CDH and as many as 100% of individuals treated with ECMO in some series. The fact that SNHL is late-onset and progressive, and therefore is not present on neonatal hearing screening, makes it difficult to compare studies and provide a more precise frequency. One study of ECMO survivors showed that SNHL was 2.5 times more common among those requiring ECMO for CDH than for other indications; however, other studies report similar frequencies after ECMO independent of the

underlying diagnosis. Other factors such as prolonged treatment with aminoglycosides, use of nitric oxide, prolonged or high-frequency mechanical ventilation, and/or metabolic alkalosis could also contribute to the development of SNHL.

Prognosis

Mortality. Mortality estimates range from 20% to 60% due to variation in patient populations and data collection techniques. Data from neonatal or referral centers,primarily including those with isolated left-sided Bochdalek hernia, report 80%-90% survival. However, population-based studies of outcome for all prenatally diagnosed individuals with CDH report mortality of at least 50%, if pregnancy terminations are included . In a meta-analysis, Stege et al [2003] observed that approximately one quarter of all prenatally diagnosed fetuses were electively terminated, 3% spontaneously miscarried, and 3% were stillborn; 31% of the live-born infants died, the majority within the first 24 hours of life.

Differential Diagnosis

Bronchogenic (foregut duplication) cysts result from abnormal budding of the ventral foregut. They contain several components of the bronchi, including respiratory epithelia, mucous glands, and cartilage and may occur anywhere along the length of the trachea or esophagus. Most are diagnosed incidentally, although cysts can become infected or if large enough, can compress the esophagus and/or trachea.

Congenital cystic adenomatoid malformation (CCAM) is a developmental abnormality of the lung resulting from abnormal cell proliferation and decreased programmed cell death of lung tissue. Abnormally formed bronchi connect to the CCAM. Type I CCAM is most common and is distinguished by relatively large cysts and mucin production. Symptoms can result when CCAMs grow in size and compress structures in the mediastinum.

Cystic teratomas are benign tumors most often found in the anterior mediastinum. They consist of several differentiated cell types derived from

endoderm, ectoderm, and/or mesoderm. Cystic teratomas of the mediastinum are uncommon, comprising fewer than 10% of all tumors in that region.

Neurogenic tumors are the most common lesion found in the posterior mediastinum. They are likely to be of neural crest origin; the majority are benign. Examples include: neurilemoma, neurofibroma, ganglioneuroma, pheochromocytoma, and neuroblastoma. CT and MRI are helpful in establishing the diagnosis.

Paraesophageal hernia occurs when a portion of the stomach and sometimes part of the peritoneal sac containing the spleen or colon move into the chest cavity through the (normally occurring but) generally enlarged or dilated esophageal hiatus. More accurately, paraesophageal hernias are a type of hiatal hernia, in which the stomach gets "stuck" in the chest, rather than sliding back and forth between the thorax and abdomen. Approximately 5%-10% of (acquired) hernias are paraesophageal. They are rare in infancy and most commonly present in older adults.

Pulmonary agenesis refers to partial or complete absence of lung tissue that is caused by failure of lung bud development. It is often associated with additional congenital malformations.

Pulmonary sequestration results from primitive lung tissue that is not connected to the tracheobronchial tree. Sequestration may be intrapulmonary, occurring within the pleura of the normal lung, or extrapulmonary, occurring outside the normal lung within its own pleural sac. Extrapulmonary sequestration appears to arise from an accessory lung bud and often has associated anomalies, such as CDH. The most common presenting symptom is recurrent chest infections.

2. Genetic Causes of Congenital Diaphragmatic Hernia

Older data using standard cytogenetic and molecular cytogenetic techniques showed that approximately 15%-20% of individuals with CDH were identified as having a genetic cause for their diaphragm defect. These frequencies are likely an underestimate and are evolving with the availability of next-generation sequencing and the identification of new CDH-associated genes. Though monogenic CDH can be isolated, it is more commonly syndromic.

Chromosomal Causes of Congenital Diaphragmatic Hernia

Historically, cytogenetically detectable chromosome anomalies have been reported in approximately 10% of individuals with CDH (see Table 3). More recently with the advent of higher-resolution molecular cytogenetic technologies, additional copy number variations have been described in individuals with CDH.

. Evaluation Strategies to Identify the Genetic Cause of Congenital Diaphragmatic Hernia in a Proband

Establishing a specific genetic cause of congenital diaphragmatic hernia:

Is complicated by the extreme genetic heterogeneity of the condition, which requires a tiered approach to genetic testing.

Prenatal Evaluation Strategy

Medical history. CDH is commonly diagnosed prenatally by detecting the abnormal positioning of the stomach, liver, or other viscera in the fetal thorax. Polyhydramnios is frequently present. Additional congenital anomalies identified on the fetal ultrasound may direct the diagnosis to a specific cause of congenital diaphragmatic hernia. Posterolateral (Bochdalek) hernias most commonly present within hours after birth with signs of pulmonary distress. Specific diagnostic investigations may be warranted when one of these conditions is suspected.

Physical examination. In addition to viable pregnancies, possible outcomes of pregnancies with CDH include miscarriage, fetal death, and termination of pregnancy. If the cause of the diaphragmatic defect is unknown, a fetopsy or autopsy examination that includes photographs and skeletal x-rays may identify co-occurring anomalies. At the time of autopsy, a sample for genomic/genetic testing may be obtained.

Family history. A three-generation family history should be taken, with attention to relatives with manifestations of congenital diaphragmatic hernia and/or infants who died in the perinatal period. Documentation of relevant findings in relatives should be obtained through direct examination or review of medical records, including results of molecular genetic testing.

Testing

Cytogenetic testing. Chorionic villus sampling (CVS) or amniocentesis for chromosome analysis (G-banded or Q-banded karyotype) are offered in many institutions as a first-line test in all fetuses prenatally diagnosed with CDH. Special attention must be paid in certain circumstances when mosaicism is suspected. Congenital diaphragmatic hernia, congenital heart defects, and rhizomelic limb shortening identified on ultrasound raise the suspicion for Pallister-Killian syndrome; however, the most frequent ultrasound finding in this condition is the combination of polyhydramnios, macrosomia, and limb shortening. The isochromosome 12p characteristic of PKS may be missed in 50% of CVS samples, while the detection rate of amniocentesis is close to 90%.

Chromosomal microarray analysis (CMA) using oligonucleotide or SNP arrays to detect microdeletions and microduplications is indicated in a fetus with CDH and additional multiple anomalies and/or a suggestive family history, if the karyotype has not identified a diagnosis. Molecular cytogenetic testing may also be offered even in individuals with apparently isolated CDH, since complex or syndromic presentations can be difficult to appreciate on prenatal imaging.

Exome sequencing. To date, exome sequencing in the prenatal period is performed exclusively on a research basis in fetuses with abnormal ultrasound scans and normal microarray analysis of chorionic villi or amniocytes. Family trio exome sequencing is preferred in this situation, because of the reported high rate of *de novo* sequence variants in both isolated and complex CDH.

For an introduction to exome sequencing click here. More detailed information for clinicians ordering genomic testing can be found here.

Targeted prenatal testing may be available in those situations in which the causative variant is known in an affected family member.

Postnatal Evaluation Strategy

Genetic testing should be directed by the clinical evidence garnered by the interpretation of medical history, physical examination, and family history.

Medical history. Respiratory distress is the most prominent finding in a newborn with CDH. The degree of pulmonary hypoplasia and pulmonary

hypertension is not directly correlated with the genetic etiology of the condition; however, individuals with syndromic CDH and multiple anomalies usually have worse outcomes. Prenatal exposure to pharmacologic agents and environmental hazards (e.g., phenmetrazine, thalidomide, quinine, cadmium, lead, nitrofen) must be investigated and excluded. Newborns with CDH have an increased incidence of additional malformations that could assist in the differential diagnosis.

Physical examination. Infants with CDH should be evaluated by a clinical geneticist for other structural birth defects (e.g., craniofacial, ocular, cardiovascular, central nervous system, limb, genitourinary), as well as minor anomalies or dysmorphic features, that may suggest a specific genetic cause of CDH. In the absence of a specific diagnosis, permission should be sought to perform an autopsy on deceased infants with CDH, especially in the perinatal period. The autopsy should include photographs, skeletal x-rays, and a sample for molecular genetic testing. A skin biopsy for cell line development and banking in a repository could provide additional genetic material should new tests be developed in the future; however, this option may only be available within a research study.

Family history. A three-generation family history should be taken, with attention to relatives with manifestations of congenital diaphragmatic hernia and/or infants who died in the perinatal period and documentation of relevant findings through direct examination or review of medical records, including results of molecular genetic testing.

Testing

Molecular genetic testing approaches can also include a combination of comprehensive genomic testing (chromosomal microarray analysis, exome sequencing, genome sequencing), karyotype, and gene-targeted testing (single-gene testing, multigene panel).

Note:

1.In an otherwise healthy individual with isolated CHD and a negative family history, currently available clinical genetic testing has a relatively low yield. However, the decision to proceed with testing should be made by an experienced

geneticist on a case-by-case basis and should be regularly reevaluated in light of the individual's developmental course, as well as advances in identifying genetic contributions to CDH.

2.Special attention must be paid when considering the diagnosis of isochromosome 12p (Pallister-Killian syndrome) since peripheral blood chromosome studies are almost invariably normal in the postnatal period. A recent report, however, demonstrates presence of iso 12p mosaicism detected by CMA performed on DNA extracted from peripheral blood.

Chromosomal microarray analysis (CMA) using oligonucleotide or SNP arrays to detect microdeletions and microduplications should be considered as first-line testing in a child who has CDH with additional multiple anomalies and/or a suggestive family history (see Table 3). CMA is particularly useful in individuals with multiple anomalies whose constellation is not suggestive of a specific disorder.

Serial single-gene testing can be considered if clinical findings and/or family history indicate that pathogenic variants in a particular gene are most likely (see Table 1). Because it is laborious and frequently inconclusive, serial single-gene testing is not frequently employed and comprehensive genomic testing is preferred in its stead in the absence of a strong clinical suspicion.

A multigene panel that includes some or all of the genes listed in the Table 1 is most likely to identify the genetic cause of the condition at a reasonable cost while limiting identification of variants of uncertain significance and pathogenic variants in genes that do not explain the underlying phenotype. No laboratories listed in the NCBI Genetic Testing Registry (GTR) (last accession: February 1, 2019) offer multigene panels specifically designed around diaphragmatic defects, although some CDH-associated genes are frequently included in congenital heart defect panels. However, these multigene panels are of limited utility for nonsyndromic patients and narrow utility in syndromic ones. Clinical exome sequencing should be considered if comprehensive testing is clinically indicated.

Comprehensive genomic testing (which does not require the clinician to a priori hypothesize which gene[s] are likely involved) may be considered in patients

with negative prior testing, or as first-line testing in individuals for whom a hypothesis for a single-gene disorder cannot be made readily. **Exome sequencing** is most commonly used; **genome sequencing** is also possible.

Exome sequencing. Clinical exome sequencing in the postnatal period is recommended as second- or third-line testing due to considerations of cost and reimbursement (exome sequencing may also be offered as part of certain research studies if clinically unavailable). Its utility is presently confined to patients with multiple anomalies and negative CMA and karyotype results. When exome sequencing is performed, concurrent or sequential trio testing is preferred to proband-only testing because of the reported high rate of *de novo* sequence variants in both isolated and complex CDH.

For an introduction to comprehensive genomic testing click here. More detailed information for clinicians ordering genomic testing can be found here.

Treatment of Manifestations

Proper management of newborns with CDH must start in the delivery room; recently, investigative therapies may start in the prenatal period. A standardized protocol for neonatal management and treatment of infants with CDH was proposed by the CDH EURO Consortium Consensus, and more recently updated by Snoek et al [2016].

Respiratory support. Emphasis must be on preventing secondary lung injury. Newborns should be intubated immediately in the delivery room to avoid bag-mask ventilation and inflation of bowel that has herniated into the chest. Care should be taken not to induce barotrauma from bag ventilation before the neonate can be transitioned to an appropriate ventilator. In the 1990s, it was recognized that minimizing aggressive ventilation by limiting ventilator settings and/or allowing the neonate to do some of the breathing work resulted in improved outcomes. It was also recognized that infants did not need to be rushed to surgery and benefited from stabilization of respiratory and cardiovascular status prior to diaphragmatic repair. Assuring end-organ perfusion and stability instead of perfect physiologic values or immediate anatomic correction is now the standard of care.

Extracorporeal membrane oxygenation (ECMO) continues to be used frequently in tertiary centers as a rescue therapy for neonates with critical cardiopulmonary deterioration. Critical factors to consider before initiating ECMO are the presence of coexisting anomalies (lethal conditions in particular), gestational age, birth weight, uncontrollable bleeding or uncorrectable bleeding diathesis, intracranial hemorrhage, and outlook of medical management. The clinical utility of ECMO in improving overall survival hinges on the selection of the population most likely to benefit.

Complications of ECMO treatment include air embolism, neurologic complications (e.g., intracranial hemorrhage, infarct, and seizures), cannulation site bleeding, coagulation abnormalities including disseminated intravascular coagulation, left-to-right shunting through the patent ductus arteriosus due to rapid and dramatic decrease in pulmonary hypertension, renal failure, systemic hypertension, and infection. Due to the complex nature of treatment and the severity of possible complications, significant experience and a multidisciplinary team are required to manage individuals on ECMO.

Pulmonary hypertension. Unfortunately, therapies that have been extremely successful in treating the usual persistent pulmonary hypertension of the newborn (PPHN) have not been widely successful in attenuating CDH-associated pulmonary hypertension and right ventricular failure. Evidence and recommendations for the diagnosis, monitoring, and management of pulmonary hypertension are reviewed in the CDH EURO Consortium Consensus – 2015 update. Since some infants respond to nitric oxide (NO) [Okuyama et al 2002], a trial of NO with careful documentation of response with echocardiography prior to continuation is warranted. Phosphodiesterase inhibitors have been used in some individuals. Other therapies that have been introduced in the acute neonatal treatment phase for CDH but are controversial include the use of surfactant and perflubron. Unfortunately, the lack of large randomized controlled trials makes it difficult to determine which of these treatments may be beneficial. The presence of surfactant deficiency in those with

CDH may depend on the sub-population, thus making it difficult to determine whether exogenous surfactant would be efficacious in large trials.

Surveillance

Since both pre- and postnatal advances in treatment have increased survival of high-risk individuals, it is important to provide close follow up and support for major complications and potential long-term morbidities. Long-term follow up for infants with CDH is ideally provided at a specialized center by a multidisciplinary team consisting of a pediatric surgeon, surgical nurse specialist, cardiologist, nutritionist, pulmonologist, and developmental pediatrician. This type of team can recognize, treat, and coordinate care for the many medical complications frequently found in long-term survivors with CDH.

Therapies Under Investigation

Prenatal treatment. Treatment of severely affected infants has shifted to the prenatal period, as rescue of severe pulmonary hypoplasia is possible if done at the correct time. The discovery that laryngeal obstruction leads to lung distension from retained fluid prompted tracheal occlusion studies in animal models and in humans. In one US randomized trial of fetal endoscopic tracheal occlusion, the treatment group experienced a high rate of preterm delivery and did not have improved morbidity or mortality rates. However, since that time, advances have been made in tracheal occlusion techniques and in predicting which fetuses would most likely die without any intervention. Currently, high-risk fetuses in Europe or in a few US centers may receive tracheal occlusion by fetal endoscopic balloon placement. The procedure is performed in the second trimester when the observed/expected lung-area to head-circumference ratios are ≤ 1.0. Generally, fetuses selected for this procedure have isolated CDH (as well as can be determined by prenatal imaging) without chromosome aberrations. Although the treatment remains investigational, initial survival rates in high-risk fetuses are increased..

V. CONGENITAL MALFORMATIONS OF BOWEL

Most congenital gastrointestinal (GI) anomalies result in some type of intestinal obstruction, frequently manifesting with feeding difficulties, distention, and emesis at birth or within 1 or 2 days. Some congenital GI malformations, such as malrotation, have a very good outcome, whereas others, such as congenital diaphragmatic hernia, have a poor outcome, with a relatively high mortality rate of 10 to 30%.

A common type of anomaly is atresia, in which a segment of the GI tract fails to form or develop normally. The most common type is esophageal atresia, followed by atresia in the jejunoileal region and in the duodenum.

Immediate management includes bowel decompression (by continuous nasogastric suction to prevent emesis, which can lead to aspiration pneumonia or further abdominal distention with respiratory embarrassment) and referral to a center for neonatal surgery. Also vital are maintenance of body temperature, prevention of hypoglycemia with IV 10% dextrose and electrolytes, and prevention or treatment of acidosis and infections so that the infant is in optimal condition for surgery.

Because about one third of infants with a GI malformation have another congenital anomaly (up to 50% in those with congenital diaphragmatic hernia and up to 70% in those with omphalocele), they should be evaluated for malformations of other organ systems, especially of the central nervous system, heart, and kidneys.

High Alimentary Tract Obstruction

Esophageal, gastric, duodenal, and sometimes jejunal obstruction should be considered when excess amniotic fluid (polyhydramnios) is diagnosed, because such obstructions prevent the fetus from swallowing and absorbing amniotic fluid.

A nasogastric tube should be passed into the neonate’s stomach immediately after cardiovascular stability has been attained after delivery. Finding large amounts of fluid in the stomach, especially if bile-stained, supports the diagnosis of upper GI obstruction, whereas inability to pass the tube into the stomach suggests esophageal atresia (or nasal obstruction [eg, choanal atresia]).

Cause

The most common cause of non-duodenal intestinal atresia is a vascular accident in utero that leads to decreased intestinal perfusion and ischemia of the respective segment of bowel. This leads to narrowing, or in the most severe cases, complete obliteration of the intestinal lumen.

In the case that the superior mesenteric artery, or another major intestinal artery, is occluded, large segments of bowel can be entirely underdeveloped (Type III). Classically, the affected area of bowel assumes a spiral configuration and is described to have an "apple peel" like appearance; this is accompanied by lack of a dorsal mesentery (Type IIIb).

An inherited form – familial multiple intestinal atresia – has also been described. This disorder was first reported in 1971. It is due to a mutation in the gene TTC7A on short arm of chromosome 2 (2p16). It is inherited as an autosomal recessive gene and is usually fatal in infancy.

Ileal atresia can also result as a complication of meconium ileus.

A third of infants with intestinal atresia are born prematurely or with low birth weight.

Intestinal atresia occurs in around 1 in 3,000 births in the United States. The most common form of intestinal atresia is duodenal atresia. It has a strong association with Down syndrome. The second-most common type is ileal atresia. 95% of congenital jejunoileal obstructions are atresias; only 5% are stenoses.

Prevalence of jejunoileal atresia is 1 to 3 in 10,000 live births. It is weakly associated with cystic fibrosis, intestinal malrotation, and gastroschisis.

The frequencies of each type from Louw and Barnard's classification are as follows:

Type I: 19–23% of cases (mean: 20.6%)

Type II:10–35% of cases (mean: 25.3%)

Type IIIa: 15%–46% of cases (mean: 30%)

Type IIIb ("apple-peel" type): 4–19% of cases (mean: 8.8%)

Type IV: 6–32% of cases (mean: 15.9%). (Note: the mean percentages total higher than 100% due to rounding.)

Classification

By location

Intestinal atresia may be classified by its location. Patients may have intestinal atresia in multiple locations.

Duodenal atresia – malformation of the duodenum, part of the intestine that empties from the stomach, and first section of the small intestine

Jejunal atresia – malformation of the jejunum, the second part of the small intestine extending from the duodenum to the ileum, that causes the jejunum to block blood flow to the colon.

Ileal atresia – malformation of the ileum, the lower part of the small intestine

Colon atresia – malformation of the colon

Malformations may also occur along multiple portions of the intestinal tract; for instance a malformation that occurs along or spans the length of the jejunum and the ilieum is termed jejunoileal atresia.

By malformation

Intestinal atresia can also by classified by the type of malformation. The classification system by Bland-Sutton and Louw and Barnard (1955) initially divided them into three types. This was later expanded to five by Zerella and Grosfeld et al.

Type I

In type I, there is a wall (septum) or membrane at some point in the bowel, leading to dilation of the bowel on the nearer side and a collapse of the bowel on the latter side. Bowel length is not usually affected in this type.



Fig 11. Type I jejunal atresia. Membrane occlusion without mesenteric defect or loss of intestinal length.

Type II

In type II, there is a gap in the bowel, and either end of the remaining intestine is closed off and connected to the other by a fibrous cord that runs along the edge of the mesentery. The mesentery remains intact.

Type IIIa

Type IIIa is similar to type II, but the mesentery is defective (there is a V-shaped gap) and the bowel length may be shortened.

Type IIIb

In type IIIb, also known as the "apple peel" or "Christmas tree" deformity, the atresia affects the jejunum, and the intestine is often malrotated with most of the mesenteric arteries absent. The remaining ileum, which is of varying length, survives on a single mesenteric artery, which it is twisted around in a spiral form. The term apple-peel intestinal atresia is generally reserved for when it affects the jejunum, while Christmas tree intestinal atresia is used if it affects the duodenum. It may affect both, however.



Fig 12. Type IIIb atresia (apple peel deformity). Note proximal jejunal atresia, malrotation, and mesenteric defect with a single artery of supply from the middle and right colic vessels with significant loss of intestinal length.

Type IV

Type IV involves a combination of all the other types and takes the appearance of a string of sausages. The length of the bowel is always shortened, but the last part of the ileum is usually not affected, as in type III. This type usually affects the nearest end of the jejunum, but the far end of the ileum may instead be affected.

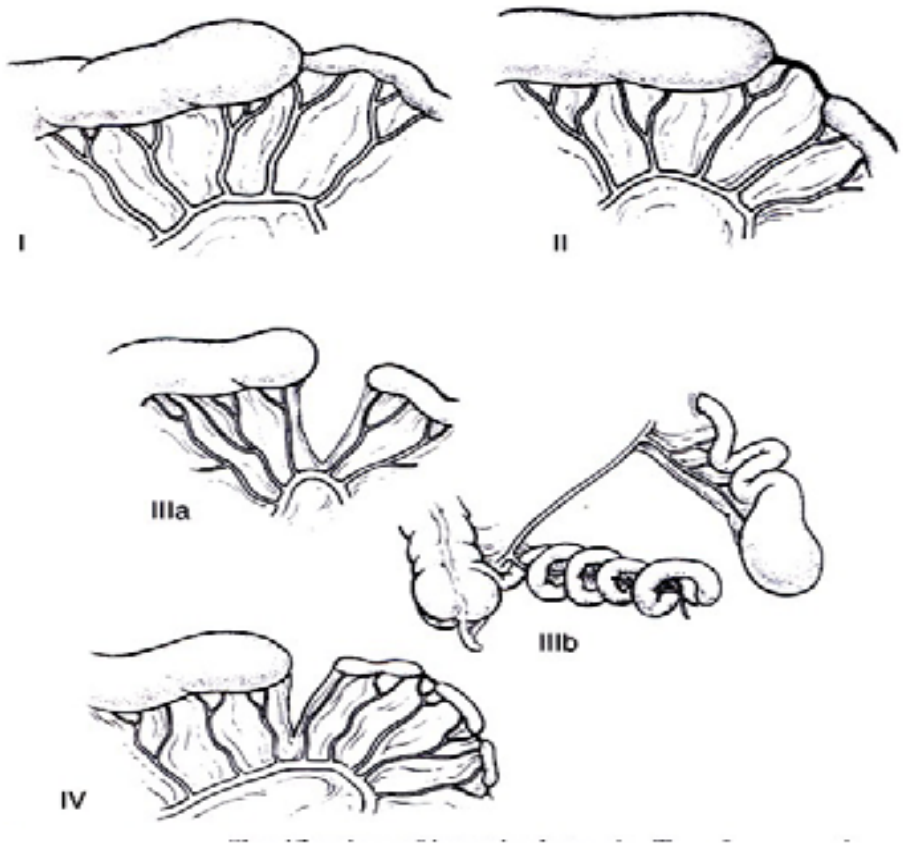


Fig 13. Type of atresia.

(Miguel Guelfand and Constanza Harding.Laparoscopic Management of Congenital Intestinal Obstruction: Duodenal Atresia and Small Bowel Atresia.Journal of Laparoendoscopic & Advanced Surgical Techniques.Oct 2021.1185-1194.http://doi.org/10.1089/lap.2021.0395**) [41]**

Diagnosis

Intestinal atresias are often discovered before birth; either during a routine sonogram which shows a dilated intestinal segment due to the blockage, or by the development of polyhydramnios (the buildup of too much amniotic fluid in the uterus). These abnormalities are indications that the fetus may have a bowel obstruction which a more detailed ultrasound study can confirm. Infants with stenosis instead of atresia are often not discovered until several days after birth.

Some fetuses with bowel obstruction have abnormal chromosomes. An amniocentesis is recommended because it can determine not only the sex of the baby, but whether or not there is a problem with the chromosomes.

If not diagnosed *in utero*, infants with intestinal atresia are typically diagnosed at day 1 or day 2 after presenting with eating problems, vomiting, and/or

failure to have a bowel movement. Diagnosis can be confirmed with an X-ray, and typically followed with an upper gastrointestinal series, lower gastrointestinal series, and ultrasound.



Fig 14. Radiograph with double-bubble sign from duodenal atresia



Figure 15: Abdominal radiograph showing several dilated gas-filled bowel loops in a jejunal atresia.



Figure 16: Contrast enema showing normal colon with dilated proximal small bowel in an infant with jejunal atresia.

Treatment

Fetal and neonatal intestinal atresia are treated using laparotomy after birth. If the area affected is small, the surgeon may be able to remove the damaged portion and join the intestine back together. In instances where the narrowing is longer, or the area is damaged and cannot be used for period of time, a temporary stoma may be placed.

The infant is usually given intravenous fluid hydration, and a nasogastric or orogastric tube may be used to aspirate the contents of the stomach. The nutritional administration is maintained after surgery until the bowel can resume normal function.

Meconium ileus

Meconium ileus is obstruction of the terminal ileum by abnormally tenacious meconium; it most often occurs in neonates with cystic fibrosis. Meconium ileus accounts for up to 33% of neonatal small-bowel obstructions. Symptoms include emesis that may be bilious, abdominal distention, and failure to pass meconium in the first several days of life. Diagnosis is based on clinical presentation and x-rays.

Treatment is enemas with dilute contrast under fluoroscopy and surgery if enemas fail.

Meconium ileus is most often an early manifestation of cystic fibrosis, which causes GI secretions to be extremely viscid and adherent to the intestinal mucosa. Meconium ileus is the presenting clinical manifestation of cystic fibrosis in 10 to 20% of cases. Of infants with meconium ileus, 80 to 90% have cystic fibrosis.

Obstruction occurs at the level of the terminal ileum (unlike the colonic obstruction caused by meconium plug syndrome) and may be diagnosed by prenatal ultrasonography. Distal to the obstruction, the colon is narrow and empty or contains small amounts of desiccated meconium pellets. The relatively empty, small-caliber colon is called a microcolon and is secondary to disuse.

Complications of meconium ileus

About 50% of cases are complicated by malrotation, intestinal atresia, or perforation. The distended loops of small bowel may twist to form a volvulus in utero. If the intestine loses its vascular supply and infarcts, sterile meconium peritonitis can result. The infarcted intestinal loop may be resorbed, leaving an area or areas of intestinal atresia. Infants with meconium ileus are also at increased risk of developing cholestasis.

Symptoms and Signs

After birth, unlike normal neonates, infants with meconium ileus fail to pass meconium in the first 12 to 24 h. They have signs of intestinal obstruction, including emesis that may be bilious and abdominal distention. Loops of distended small bowel sometimes can be palpated through the abdominal wall and may feel characteristically doughy. Meconium peritonitis with respiratory distress and ascites can occur secondary to perforation.

Diagnosis

Plain x-rays

If positive, tests for cystic fibrosis

Prenatal ultrasonography can detect changes in utero suggestive of cystic fibrosis and meconium ileus (eg, dilated bowel, polyhydramnios), but these changes are not specific.

Diagnosis of meconium ileus is suspected in a neonate with signs of intestinal obstruction, particularly if a family history of cystic fibrosis exists. Patients should undergo abdominal x-rays, which show dilated intestinal loops; however, fluid levels may be absent. A “soap bubble” or “ground glass” appearance due to small air bubbles mixed with the meconium is diagnostic of meconium ileus. If meconium peritonitis is present, calcified meconium flecks may line the peritoneal surfaces and even the scrotum. A water-soluble contrast enema reveals a microcolon with an obstruction in the terminal ileum.

Patients diagnosed with meconium ileus should be tested for cystic fibrosis.

Treatment

Radiographic contrast enema

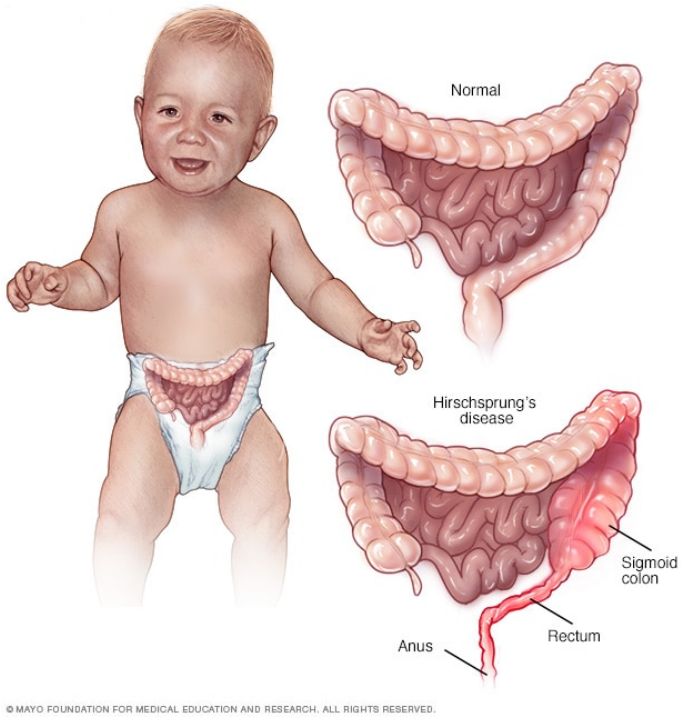
Sometimes surgery

Obstruction may be relieved in uncomplicated cases (ie, without perforation, volvulus, or atresia) by giving ≥ 1 enema with a dilute radiographic contrast medium plus *N*-acetylcysteine under fluoroscopy; hypertonic contrast material may cause large GI water losses requiring IV rehydration.

If the enema does not relieve the obstruction, laparotomy is required. A double-barreled ileostomy with repeated *N*-acetylcysteine lavage of the proximal and distal loops is usually required to liquefy and remove the abnormal meconium.

VI. Hirschsprung's disease. Rectal atresia

Hirschsprung's disease



[https://www.mayoclinic.org/diseases-conditions/hirschsprungs-disease/symptoms-causes/syc-20351556]

Hirschsprung's (HIRSH-sproongz) disease is a condition that affects the large intestine (colon) and causes problems with passing stool. The condition is present at birth (congenital) as a result of missing nerve cells in the muscles of the baby's colon.

A newborn who has Hirschsprung's disease usually can't have a bowel movement in the days after birth. In mild cases, the condition might not be detected until later in childhood. Uncommonly, Hirschsprung's disease is first diagnosed in adults.

Synonyms of Hirschsprung Disease

- colonic aganglionosis
- congenital megacolon
- HAEC
- Hirschsprung-associated enterocolitis
- Hirschsprung’s disease
- HSCR
- intestinal aganglionosis
- megacolon, aganglionic

General Discussion

Hirschsprung disease (HSCR) is a birth defect. This disorder is characterized by the absence of particular nerve cells (ganglions) in a segment of the bowel in an infant. The absence of ganglion cells causes the muscles in the bowels to lose their ability to move stool through the intestine (peristalsis). Peristalsis is a normal process of the body. Peristalsis creates wave-like contractions from the muscles lining the intestines. These contractions propel stool and other waste material through the digestive system. Ineffective peristalsis leads to stool backing up in the intestines. Affected individuals can develop constipation and partial or total obstruction of the bowels. Pain and discomfort can result. If not treated, a potentially serious bacterial infection may develop. The specific symptoms can vary from one person to another. HSCR can occur as an isolated problem or as part of a broader disorder that affects multiple organ systems.

Signs & Symptoms

Symptoms in the newborn period include failure to pass the meconium within a short time after birth. Meconium is the dark sticky substance that is normally present in the intestine at birth and is passed as an infant's first bowel movement after birth. Failure to pass a first stool for 24-48 hours is suggestive of HSCR.

Infants with HSCR will very often have abdominal swelling (distention), abdominal pain, and vomiting. Affected infants have constipation and often exhibit poor weight gain, and slow growth.

HSCR can sometimes lead to a condition called enterocolitis, which is inflammation of the small intestines and colon. This is often referred to as Hirschsprung-associated enterocolitis. Hirschsprung-associated enterocolitis is the most frequent complication of HSCR occurring in 30-40% of individuals with HSCR and can be mild to severe in nature. Hirschsprung-associated enterocolitis often presents with fever, explosive diarrhea, abdominal swelling, lethargy, and vomiting. Some individuals with either severe or untreated Hirschsprung-associated enterocolitis may develop sepsis, which is a widespread bacterial infection of the bloodstream and is potentially life-threatening. Severe or untreated enterocolitis can also lead to toxic megacolon, another life-threatening complication. It cannot be overemphasized that an individual with HSCR who develops these symptoms should urgently seek medical attention.

Approximately 90% of initial HSCR diagnoses in the United States are made within the first year of life. Most of the remaining 10% are made in early childhood, with less than 1% being made in teenage years or adulthood. Not surprisingly, these individuals often report a lifelong history of constipation.

Causes

Hirschsprung disease that occurs as an isolated problem has been associated with mutations in several different genes. Approximately 50% of affected individuals have one of these gene abnormalities. This gene changes cause people to be susceptible or predisposed to developing the disorder. A person who is genetically predisposed to a disorder carries a gene (or genes) for the disease, but it may not be expressed unless it is triggered or "activated" under certain circumstances, such as due to particular environmental factors (multifactorial inheritance).

Inheritance of these gene changes can be dominant or recessive depending on the gene involved, but it is probably necessary for multiple abnormal genes to be present for the disorder to occur. The abnormal genes involved in HSCR can have different effects in members of the same family. If parents have an affected child, their chance of having another child with the disorder is increased. A parent who has HSCR also has an increased chance of having a child with the disorder.

The genes associated with HSCR are in two major groups called *RET* genes and *EDNRB* genes. When the disorder involves a short segment of the colon, the major gene involved is the *RET* gene located on chromosome 10q11.2.

When HSCR occurs along with other abnormalities, the cause is frequently a chromosome abnormality or genetic syndrome. People with Down syndrome are at a greater risk of developing HSCR than people in the general population. Genetic syndromes that can be associated with HSCR include Mowat-Wilson syndrome, Waardenburg syndrome, Bardet-Biedel syndrome, Cartilage-Hair hypoplasia, congenital central hypoventilation syndrome, Fryns syndrome, multiple endocrine neoplasia type 2, Smith-Lemli-Optiz syndrome, L1 syndrome, and Pitt-Hopkins syndrome. (For more information on these disorders, choose the specific disorder name as your search term in the Rare Disease Database.)

Signs and symptoms of HSCR occur because of the failure of specific nerve cells called ganglions to develop in a part of the large intestine of an infant. HSCR is sometimes called a neurocristopathy, meaning that the disorder results from abnormalities in cells and tissues that arise from the neural crest. The neural crest is a temporary group of cells found in the developing embryo. The neural crest gives to various types of cells in the body. In HSCR, ganglions do not develop properly from the neural crest. Since ganglions are missing from the intestine, stool cannot be pushed through the intestine and out of the body via peristalsis.

The length of the intestines that is affected in HSCR can vary. In approximately 80% of affected infants, it is the large intestine, commonly referred to as the colon and rectum, that are affected. The rectum is the last portion of the large intestine and connects the anus to the sigmoid colon. Infants with an absence of ganglion cells in the rectum and sigmoid colon are said to have "short-segment"

Hirschsprung disease. While approximately 12% of infants will have ganglion cells missing from most of the large intestine and are referred to as “long-segment” Hirschsprung disease, and approximately 7% will have ganglion cells missing in the entire colon and possibly part of the small intestine, these are called “total colonic” Hirschsprung disease. In rare instances, ganglionic nerve cells are missing from the entire length of the large and small intestine. This is referred to as total intestinal aganglionosis.

Affected Populations

Hirschsprung disease affects males 3 to 4 times more often than females, although long-segment HSCR has a gender ratio of 1:1. The disorder occurs in approximately one in 5,000 live births. It is usually apparent shortly after birth, but may present in older children and adults. Hirschsprung disease should be considered in people with a history of severe constipation.

Related Disorders

Symptoms of the following disorders can be similar to those of Hirschsprung disease. Comparisons may be useful for a differential diagnosis:

Chronic intestinal pseudo-obstruction (CIP) is a rare, potentially disabling gastrointestinal disorder characterized by abnormalities affecting the involuntary, coordinated muscular contractions (a process called peristalsis) of the gastrointestinal (GI) tract. Peristalsis propels food and other material through the digestive system under the control of nerves, pacemaker cells and hormones. CIP usually results from abnormalities affecting the muscles or nerves that are involved in peristalsis. Consequently, peristalsis becomes altered and inefficient. The symptoms of CIP resemble those caused by mechanical obstruction of the small bowel. Mechanical obstruction refers to something (such as a tumor, scar tissue, etc.) physically blocking the passage of food and other material through the GI tract. In individuals with CIP no such physical obstruction is present, hence the term pseudo-obstruction. Common symptoms include nausea, vomiting, abdominal pain, abdominal swelling (distention) and constipation. Ultimately, normal nutritional requirements cannot be met leading to unintended weight loss and malnourishment. CIP can potentially cause severe, even life-threatening complications. (For more information on this disorder, choose “Intestinal pseudoobstruction” as your search term in the Rare Disease Database.)

In the newborn period, several other conditions can have signs or symptoms that are similar to those seen in HSCR. These conditions include narrowing (atresia) of the colon causing intestinal blockage; a temporary condition called meconium plug syndrome; meconium ileus, a condition often seen in cystic fibrosis and in which the meconium in a child’s intestine is abnormally thick and sticky causing

intestinal blockage; narrow of the portion of the small intestine called the ileum (ileal atresia). Sometimes, necrotizing enterocolitis can cause symptoms similar to HSCR.

Diagnosis

A diagnosis of HSCR can be suspected based on a physical examination, a complete patient and family history, identification of characteristic symptoms and a variety of specialized tests. Most people (85-90%) are diagnosed in early infancy. The first symptom is usually failure to pass the first bowel movement (meconium). The preferred diagnostic test for HSCR is a suction biopsy of the rectum. A biopsy involves surgically cutting out a small sample of affected tissue and studying it under a microscope. Absence of ganglion cells confirms the diagnosis.

Additional tests that can be used include an abdominal x-ray, which can reveal the presence of an intestinal blockage, an anorectal manometry, which involves using balloons and pressure sensors to assess the health and function of the rectum, and a contrast or barium enema, which involves using a contrast agent in the rectum. A contrast agent is a substance that is used to enhance how a structure or part of the body appears on an x-ray. After using a contrast enema in the rectum, x-rays are taken to assess the health and function of the large intestines.

When other abnormalities are present in addition to HSCR, it is possible that the HSCR is due to a chromosomal abnormality or genetic syndrome. Individuals with multiple anomalies should be evaluated by a clinical geneticist in order to attempt to establish an underlying diagnosis.

Standard Therapies

In almost all cases, treatment of HSCR requires surgery to remove the part of the colon and/or rectum that lacks the normal nerve development, and to join the two healthy ends together. There are three standard surgical procedures designed to correct this disorder. The choice of procedure is a function of the training and experience of the surgeon. Each procedure removes the affected part and attaches the healthy part of the bowel to the rectum completing what is known as a “pull-through” procedure. Currently most procedures are performed in a single stage.

If the child is born prematurely, has low-birth weight, or if he/she is critically ill the surgeon may counsel the parents that a safer approach is a multi-stage strategy. The first stage is to create a temporary colostomy in which the healthy end intestine, upstream of the affected intestine, is brought to the surface of the abdomen creating stoma. Through this opening or ‘stoma’, the contents of the bowel are eliminated into a special bag and removed. After a time, the second stage “pull through” operation is carried out at which time the stoma may be closed. Most children with HSCR do not need a colostomy or an ileostomy.

According to the medical literature, most children enjoy a good to excellent quality of life following successful surgery. Some children may require bowel

management after surgery. In rare instances, some children may require revisional or repeat “pull-through” surgery.

Genetic counseling may be of benefit for affected individuals and their families.

Anorectal malformations

Anorectal malformations such as imperforate anus cause abnormalities in the way a baby has a bowel movement. These problems vary depending on the type of malformation:

- If the anal passage is narrow, your baby may have a difficult time passing a stool, causing constipation and possibly discomfort.
- If there is a membrane over the anal opening, your baby may be unable to have a bowel movement.
- If the rectum is not connected to the anus but there is a fistula present, stool will leave your baby's body through the fistula and enter the urinary tract instead of the anus. This can cause urinary infection.
- If the rectum is not connected to the anus and there is not a fistula present, there is no way for the stool to leave the intestine. Then, your baby will be unable to have a bowel movement and will have a blockage of the intestine as a newborn.

As a fetus is growing in its mother's uterus, all organ systems are developing and maturing at different rates. Certain steps have to take place in the seventh to tenth weeks of gestation for the rectum and anus to separate from the urinary tract and form properly. Sometimes these steps do not occur as they should, and the rectum, anus or both may not develop normally.

Most of the time, the cause for an anorectal malformation is unknown, but in rare cases, it appears to run in families. Boys are at a slightly higher risk for this abnormality than girls.

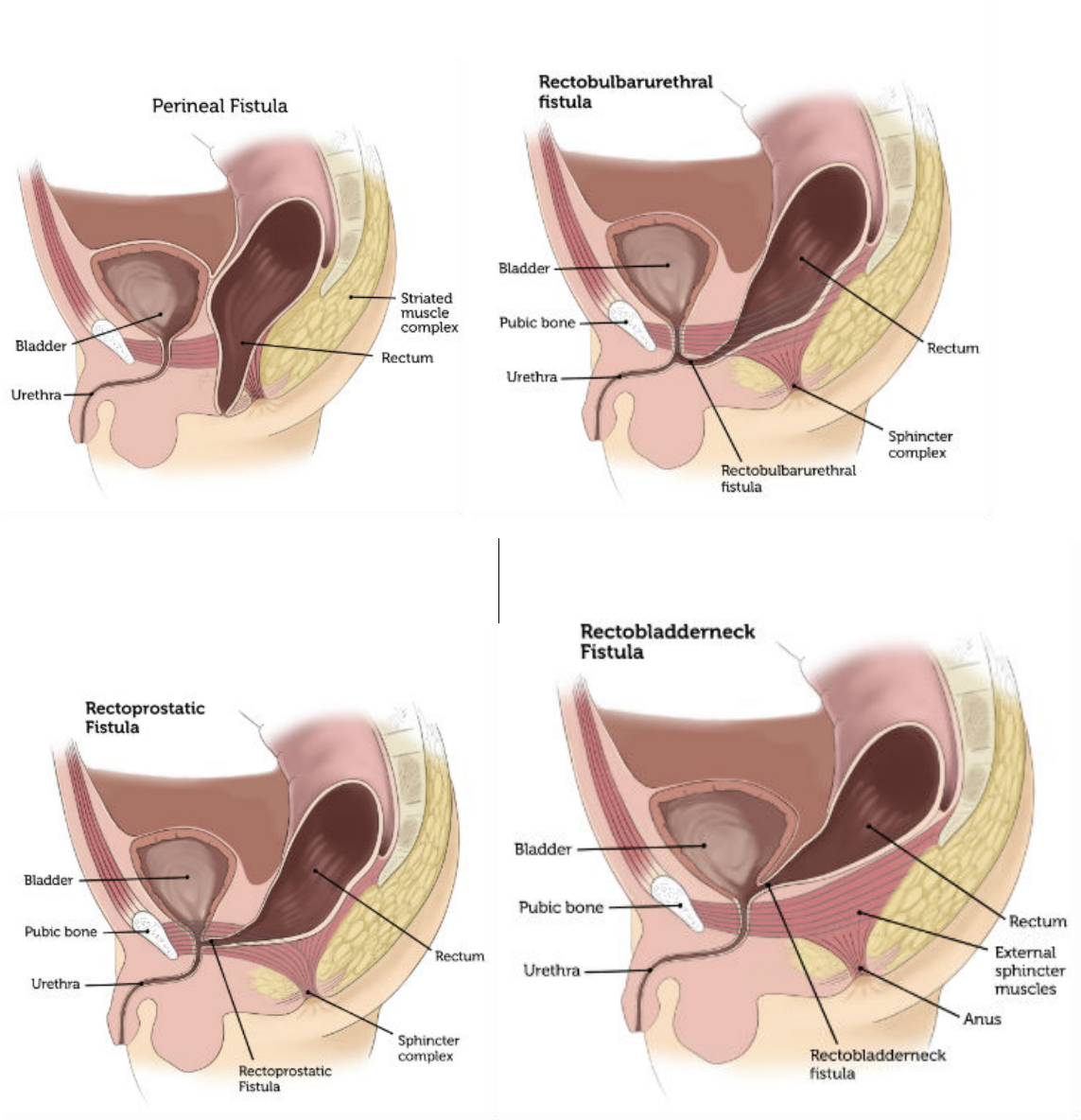
Up to one third of babies who have genetic syndromes, chromosomal abnormalities or other congenital defects also have imperforate anus or other anorectal malformations. These genetic syndromes and other congenital problems include:

- VACTERL association (a syndrome in which there are vertebral, anal, cardiac, tracheal, esophageal, renal and limb abnormalities)
- digestive system abnormalities
- urinary tract abnormalities
- abnormalities of the spine

Types of Anorectal Malformations

Anorectal malformations, including imperforate anus, can affect male and female babies in different ways.

In boys, the main anorectal malformations are perineal fistula, rectobulbarurethral fistula, rectoprostatic fistula and rectobladderneck fistula.



[https://healthjade.net/anorectal-anomalies/]

CLINICAL PRESENTATION

At birth, a general examination in any newborn should include the perineum. The absence or abnormal location of the anus is generally apparent. In the male, besides the absent anus, a note must be made of the anal pit. The fistula may be of small caliber; hence, it may take up to 24 h for it to be apparent. The reason for this is that it takes this long for ingested gas to travel down the gut to the blind rectum.

In the female child, the diagnosis rests on the appearance of the perineum. Normally, there are three visible openings — the most anterior being the urethra, followed by the vagina, both of these being within the vestibule. Behind the perineal body is the anus. The presence of three openings, with the anus not being at its normal site is indicative of either a perineal fistula, formerly called the anterior perineal anus. If the third opening is seen within the vestibule, it is a vestibular fistula. Two openings only, indicate two extremely rare clinical entities, namely a recto vaginal fistula, or a blind ending rectum with no fistula. A single opening indicates the persistent cloaca.

INVESTIGATIONS

Invertogram

Wangensteen and Rice first described the use of inversion radiography in 1930 to indicate the distance between the gas bubble within the terminal colon and the perineal skin. Direct measurement between the gas-filled gut and the skin of the anus by placing a radiopaque marker on the skin is made. Subsequently, the P–C line and the I line are determined by invertography. If the rectum ends below the P–C line, but not below the I line, it is “intermediate”. When a rectal pouch that is clearly below the I line, it may be referred to as “low,” whereas when pouch ends above the P–C line is called as high type.

Prone cross-table lateral view

The babies are kept in the genupectoral position for 3 min by holding their facedown with hips flexed. Prone lateral radiographs is obtained, which is centered over the greater trochanters like in the invertogram. The prone cross-lateral view’ has few advantages like the baby is comfortable, whereas in invertogram requires splints and adhesive tapes and the baby keeps crying due to which puborectalis sling contracts and hence there is deceptive obliteration of the lower rectum.

Ultrasonic examination

Ultrasonographic examination has been used to know the pouch perineal distance. It can be performed through a transperineal or infracoccygeal route.

Infracoccygeal route can directly demonstrate the puborectalis as a hypoechoic U-shaped band. The noninvasive nature and no radiation exposure are the main advantages, but it is highly observer dependent.

Computer tomography and magnetic resonance imaging

Computer tomography (CT) and magnetic resonance imaging (MRI) of pelvis have been utilized for the direct visualization of the sphincteric muscles. These have been used for the structural evaluation of pelvic floor muscle and relation to the pouch, for both the pre- and post-operative evaluation. The exact location of fistula and relation to the pelvic floor muscle provides crucial information regarding the approach, whether a sagittal approach or an approach through abdominal route is required. MRI and CT scans are also utilized for the assessment of structural development following different procedures for ARM and can help in comparison of outcome between different procedures. MRI is considered superior to CT scan because of excellent soft tissue characterization, multiplanar imaging, and lack of ionizing radiation.

SURGICAL MANAGEMENT

Initial evaluation and decision-making

The early management of a newborn with an anorectal anomaly is crucial. In developing countries, presentation can be delayed. This is with associated abdominal distension, dehydration and sepsis. Initial resuscitation with intravenous fluid and broad spectrum antibiotics holds the key for the final outcome in such cases. After assessment of associated anomalies the child can be taken for a protective colostomy, followed by delayed repair later or a single staged definitive procedure can also be performed in selected cases. Management of male and females cases differ.

Male newborn

Male newborns with rectoperineal fistula do not need a colostomy. They can undergo a posterior sagittal anoplasty or limited PSARP. The male babies with evidence of a recto-urinary tract communication should undergo fecal diversion with

a colostomy. Patients with rectobulbar, rectoprostatic fistula can be managed by primary PSARP. Abdominal approach is required to get access to fistula in cases with recto-bladder neck fistula. Pouch colon also requires abdominal approach. In cases with sepsis, perforation a protective colostomy is performed in the neonatal period followed by a definitive pull-through procedure at 3-6 months of age. Before the definitive operation, a pressured distal colonogram and voiding cystourethrogram should be performed to reveal the site of rectourethral fistulas.

Female newborn

General condition, duration of presentation and number of openings in the vestibule decide the management. Gross abdominal distension, sepsis, or single opening (persistent cloaca) warrants a diversion colostomy. The most common anomaly in females is a rectovestibular and shows a normal urethra, normal vagina, and another orifice, which is the rectal fistula in the vestibule. Such cases can be managed by a diversion colostomy and delayed definitive repair by PSARP. In selected cases, a primary PSARP or a primary anterior sagittal anorectoplasty can also be performed. Definitive repair of persistent cloaca is performed through PSARP. In cases with common channels longer than 3 cm, it is difficult to mobilize the vagina through PSARP, and an abdominoperineal approach is required. In cases with a common channel of <3 cm, total urogenital mobilization is possible, in which both the vagina and urethra are mobilized as a unit, without separation. If the distance from the vagina to the perineum is long, a bowel segment can be used to bridge the gap, preferably a segment of the colon or a vaginal switch procedure can be done in cases with bicornuate uterus.

Single stage versus staged procedure

There has been debate over single stage versus staged repair of ARM. Single stage procedure has less morbidity, mortality at low cost. The better continence may be attributed to the better development of cerebral fibers. These cerebral cortical fibers develop in the 1st year of life and sensations of rectal fullness are essential for these fibers to develop fully so that continence can be achieved to its maximal potential. The other advantages of single stage repair are easier dissection in the

neonatal period due to virgin tissue planes with no fibrosis due to pouchitis, no need of bowel preparation and colostomy is not usually accepted in our society. Physical and psychological stress to the parents, child and surgeon are less. Absolute contraindication for single stage repair is severe life-threatening other congenital malformations, sepsis or necrotizing enterocolitis with pneumoperitoneum, extreme prematurity and common cloacae.

The single staged repair carries a considerable risk to the urinary tract with this practice because the surgeon does not know the precise anorectal defect. The only way to definitively determine the patient's anorectal defect is to perform a distal colostogram, which of course requires the presence of a colostomy. Without this information, an operation in the newborn period is essentially a blind perineal exploration. The surgeon may not be able to find the rectum and may find and damage other, unexpected, structures, such as the posterior urethra, seminal vesicles, vas deferens, and ectopic ureter during the search for the rectum. Finally, without fecal diversion, there is the risk of dehiscence and infection. These complications may compromise the ultimate functional prognosis.

Newer imaging modalities and advances in radiological technique have made it possible to exactly delineate the site of rectourethral fistula. This has tilted the balance in favor of single stage repair of appropriately selected cases, as most feared complication of single staged repair like trauma to urinary tract can be avoided.

Surgical procedure

Understanding of anatomy and thus the principal of surgical repair of ARM has evolved continuously over the past century. Prof. Douglas Stephens puborectalis concept introduced in 1953 remained the basis of all surgical procedures for intermediate and high ARM.[15] Classically abdominoperineal pull-through approach is used for high-or intermediate-type ARMs. A combination of perineal and abdominal approach was done by Rhoads *et al.*, and a pull-through route was made blindly with the fingers.[16] There was fear of damage to pelvis innervations with this approach. So Rehbein performed endorectal pull-through to avoid any damage to the pelvic innervations.[17] A modified abdominoperineal pull-through

procedure was proposed by Iwai *et al*. in which the rectum was dissected carefully along the rectal wall to avoid damage to the pelvic nerves, using an electric stimulator.[18] Stephens's procedure involved the identification of the proximal portion of the levator ani through a posterior approach through the sacrococcygeal junction. In cases with rectal pouch distal to the pubococcygeal line, it was possible to divide the fistula and mobilize the rectum via the sacrococcygeal route alone without abdominal exploration.[19] Stephens's sacral dissection with Rehbein's endorectal dissection was combined by Kiesewetter to developed a sacroabdominoperineal pull-through approach that preserved the puborectalis muscle.[10] In spite of the vast number of procedure and their modification, the postoperative outcome of ARM remained dismal and variable throughout the world. Peña introduced the posterior sagittal approach (PSARP).[6,20] Soon after its introduction, PSARP became the gold standard procedure worldwide. This approach allowed surgeons to view the anatomy of these defects clearly, to repair them under direct vision, and to learn about the complex anatomic arrangement of the junction of the rectum and genitourinary tract.[21] It has become the predominant surgical method for anorectal anomalies. In cases, when the rectum is very high an abdominal approach is needed Abdomino-PSARP. Posterior sagittal anorectoplasty has become the standard of care for dealing with ARM. Despite the excellent exposure of anatomy and exact placement of the distal rectum within the muscle complex, the technique is not perfect, especially in patients with “high” ARM defects. Georgeson *et al*. described a new laparoscopically assisted anorectal pull-through (LAARP) for the repair of high-type ARMs, which utilizes a laparoscopic vantage point to reduce the amount of posterior dissection required for accurate placement of the bowel into the muscle complex.[22] Sharp dissection and cautery are used laparoscopically to expose the rectal pouch down to the urethral or vaginal fistula. LAARP is not recommended for intermediate-type ARMs, because this method requires wide dissection of the rectum and may cause damage to the rectal nerves and pelvic plexus, resulting in poor bowel function. Bischoff *et al*. suggested that laparoscopy for the ARM is a less invasive procedure when compared with those

operations that would have previously required a laparotomy such as a rectobladder neck fistula and rectoprostatic fistula.[23]

Congenital pouch colon

Congenital pouch colon (CPC) is a congenital anomaly in which whole or part of the colon is replaced by a pouch-like dilatation that communicates distally with the urogenital tract by means of fistula and is uniformly associated with ARMs. The incidence of CPC among all cases of ARM has been reported to occur from 2% to 18% with a high incidence (30-40%) reported from Indian sub-continent. Various classifications have been proposed for CPC, but classification proposed by Rao *et al*. is most acceptable classification.[24] The standard procedure for the management of the CPC cases is a three-staged procedure. In cases with complete pouch colon (types I and II), as adequate length of the normal colon is absent, construction of distal colon by tabularizing (coloplasty) of the remaining colon has been proposed. Single staged management of pouch colon has also shown good results, though few authors have found increased mortality with this approach.

Spinal cord tethering

Intravertebral fixation of the filum terminale results in the tethering of the spinal cord In recent years, high resolution diagnostic equipment such as spinal MRI and three-dimensional CT have demonstrated a higher frequency of tethered cord in the setting of ARM than previously estimated. The prevalence in the literature is variable (10-52%). A conservative approach has been advocated for ARM patients with asymptomatic tethered cord. Prophylactic surgery appears to have a minimal benefits and release of tethered cord should be done in symptomatic cases only.[25]

Secondary reconstruction/redo posterior sagittal anorectoplasty

Secondary reconstructions to improve fecal continence have been used extensively in patients with ARMs. In most reports, the long-term functional outcome is not better in patients who had secondary surgery and may be worse than in those with only primary repair. However, Peña *et al*. produced good results with secondary penicillin-resistant streptococcus pneumonia. Rerouting of the pulled-through bowel has been advocated for patients who have a misplaced anal canal

following primary operation. The bowel may traverse the levator and not lay anterior to it.[26]

Long-term results

The main objective of any surgery for ARMs is to achieve a good quality of life in adults patients with ARMs.

Bowel function

Most patients who undergo repair of an ARM suffer from variable degrees of fecal incontinence, depending upon the type of anomaly, associated anomalies and the effectiveness of corrective procedure. The functional results after surgical correction of ARMs are often assessed by a manometric study, neural electrophysiological studies, magnetic spinal stimulation, and fecoflowmetry. Most cases with low ARM have good bowel function and enjoy social activities. One-third of patients with high- or intermediate-type anomalies occasionally complained of fecal soiling. Bowel management is an artificial way to keep patients who have fecal incontinence clean. It consists of enemas, colonic irrigation, or daily suppositories to keep the rectum vacant. Biofeedback therapy was reported to be a simple and safe method for treating children. In resistant cases, The Malone antigrade continence enema procedure using an Appendicectomy represents a useful alternative. Secondary reconstructions to improve fecal continence have been used extensively in patients with ARM. Historically, a gracilis muscle or gluteus maximus muscle transplant to strengthen the striated anal sphincter has been used.[27]

Urinary function

The patients with ARM are frequently complicated with urinary tract anomalies or sacral anomalies. One-third of patients with traditional abdominoperineal methods, have urinary complaints compared to about 10% of those who underwent PSARP.[28]

Sexual function

In male patients with urinary tract or sacral anomalies, erection and ejaculation problems are often seen. Sexual problems of pubescent males with high- and intermediate-type ARM such as erectile and ejaculatory dysfunction is about

40%. Adolescent females with low ARM are limited in terms of sociosexual activities. Other common problems in females are vaginal and uterine septation anomalies and vaginal agenesis. In sexually active females, these anomalies often cause infertility and sexual problems.[29]

Long-term growth and development

Very few long-term prospective studies have been performed to know the overall growth and development of ARM patients. Newborns with severe anatomical malformations with associated anomalies, long duration hospital stay and multiple surgical interventions may negatively influence growth and development within the first 2 years of life. Children with an ARM are at risk for gross motor function problems so the nutritional status and bowel management should be optimized individually to prevent stunting and comprehensive physical and mental development.

VII. Congenital defects of the kidneys and urinary tract

Introduction

The congenital anomalies of the urinary tract include a large number of diseases caused by anomalies in the morphogenesis of the urinary system. These anomalies include obstructive and nonobstructive dilatation of the urinary tract that can be associated with alterations in the number, size and/or position of the kidneys.

We will discuss the congenital anomalies of the urinary and genital tract, with a short review of kidney abnormalities. These malformations may coexist within the same case, and this is due to their common embryonic origin .

The reno-urinary anomalies occur more frequently in males than in females, the ratio being 2.5:1 (M-F), and there are many cases with family aggregation. The incidence is 3–4 at 1000 lives or 1–5% of all pregnancies . If including all cases detected at post-mortem fetal autopsies, the prevalence of these malformations is much higher

Congenital anomalies of urinary tract

2.1. Relevant embryology

The urogenital system is represented by two major components: the urinary system and the genital system. Embryological and fetal kidneys, genital tract and the urinary system develop from intermediate mesoderm. The rhythm of growth and development of the collector tubes differs greatly during pregnancy. Until 15 weeks of gestation, the rhythm of development is very rapid and later decreases.

The urinary tract is almost entirely developed from the intermediate mesoderm. The evolution implies different stages in development: pronephros, mesonephros and metanephros. These stages will appear successively in the craniocaudal direction and coexist over time. Intermediate mesoderm is divided in the upper cervical and thoracic region, resulting in nephrotomes.

The mesonephric duct will give rise to the ureteral bud after the formation of the pronephros. This, after 15 generations of divisions, will lead to the formation of the ureter, the calves, the collector tubes and the kidney pelvis. The lack of formation or agenesis of the pronephros or of the mesonephric duct can lead to the total or partial absence of the kidney or to other anomalies of the reno-urinary tract.

CAKUT (congenital anomalies of kidney and urinary tract) may be part of multi-organ processes in single-gene disorders, with dominant or recessive inheritance, as we can find in Fraser syndrome, the branchiootorenal syndrome, Kallmann syndrome, Ehlers-Danlos syndrome and others.

The genitalia differentiation, which leads to female or male gender, starts at 7–8 weeks of pregnancy and finalizes at 12–13 weeks. Every fetus will develop female characteristics, because at the beginning of gestational period, the Mullerian and Wolffian structures coexist. If the Mullerian structure will suffer atrophy, due to testosterone and anti-Mullerian hormone effects, male sex structures will develop from the Wolffian structure, and the fetus will become a male. In the opposite circumstance, the fetus will become a female. In conclusion, from the embryological point of view, an individual could be a female if the masculine features will not develop.

2.2. Classification

Congenital urinary abnormalities are often associated with the kidney anomalies, and there is a wide range of malformations resulting from disorders in the normal development process.

Malformations of renal parenchyma may occur due to the abnormal nephron development—in cases of renal dysplasia, renal agenesis and renal polycystic disease. The migration abnormalities of kidney embryo buds are found in renal ectopy and in the mismatched malformations.

Abnormalities in the development of the urinary tract system cause duplicated collective systems, posterior urethral valves and obstructions of the pyeloureteral junction. Defects may be unilateral or bilateral, and several types of defects may be associated.

The anomalies of the urinary system can be divided into nephropathies and uropathies:

Nephropathy involves the renal parenchyma and refers to multicystic kidney disease (MCDK), renal dysplasia disease, renal agenesis defect, congenital polycystic kidney disease and other anomalies (nephromegaly, trisomy 13, Meckel syndrome, Beckwith-Wiedemann syndrome).

Uropathies represent the pathology of the urinary tract, and by the site of the defect, they can be pyeloureteral and ureterovesical, or they can refer to the vesicoureteral reflux and to the posterior urethral valve. Many authors also consider here the urachal fistula, the urachal cyst and the exstrophy of the urinary bladder.

For a better understanding of urinary tract abnormalities, we will summarize the classification of kidney malformations that may accompany these anomalies:

Anomalies in number

Complete bilateral renal agenesis is a rare condition, not compatible with extrauterine life. The physical appearance of the babies is very characteristic (the so-called Potter syndrome). The absence of the kidneys may be suspected before conventional postmortem autopsy. Fetal ultrasound examination is very difficult, due to the absence of amniotic fluid.

Unilateral renal agenesis or the 'congenital solitary kidney' is probably the most difficult diagnosis of renal malformations. There is a consensus in terms of a definitive diagnosis, autopsy being the only conclusive method.

Renal aplasia—there is a fetal bud, but it does not develop into a normal functioning organ. The pelvis and ureter are usually absent or are rudimentary.

Supernumerary kidney is the rarest anomaly and consists of a third kidney with excretory cavities and its own vascularity, completely separated from the other kidney.

Renal size abnormalities

Renal hypoplasia. There are small congenital kidneys, located next to the median line and excretory cavities. The renal function is normal or low.

Kidney hyperplasia. There is a large kidney, after ruling out any other cause.

Kidney's shape abnormalities

Lobular kidney. There is a persistence of fetal lobulation.

Kidney fusion. There are bilateral symmetrical or asymmetrical mergers or unilateral asymmetry.

The kidney in the horseshoe. In 90% of the cases, kidneys join at the lower pole through a parenchymal or fibrous bridge.

Bilateral asymmetric mergers. One of the kidneys is smaller than the other.

Unilateral asymmetric fusions. There is a single kidney mass with a crossed ectopic kidney, known as 'the sigmoid kidney', usually located in the pelvis.

Renal position abnormalities

Ectopic kidney. A birth defect given by an abnormal (unilateral or bilateral) kidney position. The ectopic kidney is more often located on the left side. There are several ectopic kidney types: the caudal (lumbar, iliac, sacral) kidney, the cranial kidney and the crossed kidney.

Renal dystopia. Initially, in embryonic life, the hill and pelvis are located in the anterior part, and then they undergo a rotation around the longitudinal axis, until they reach the median side.

Multicystic renal dysplasia

These are large and hyperechogenic kidneys, including renal ciliopathies. These conditions may be divided into autosomal dominant diseases and autosomal recessive disorders, including the polycystic kidney disease, renal dysplasia, glomerulocystic kidney disease (trisomy 13 and trisomy 18, Beemer syndrome), multicystic dysplastic kidneys (MCDK), medullary cystic dysplasia (Meckel-Gruber and Beckwith-Wiedemann syndromes, as well as congenital infections)

2.3. Diagnosis

Nowadays, the fetal bladder and kidneys may be visualized starting with 11 weeks (by transvaginal echography) or 12 weeks (by means of transabdominal echography). The fetal bladder and kidneys are located near the spine, having an elliptical shape.

In the first trimester of pregnancy, the kidneys appear as an ovoid structure located on both sides of the spine (Figure 11).



Figure 17 . Fetal kidneys in the first trimester of pregnancy.

Uropathies are the most common reno-urinary abnormalities diagnosed during the prenatal period and may be caused by obstructive or nonobstructive factors. The most common urinary malformations encountered in children are vesicoureteral reflux (VUR), obstructive megaureter, posterior urethral valve and megacystis.

Fetal hydronephrosis may result in a number of conditions, like pelvic or vesicoureteral junction obstruction, posterior urethral valves, vesicoureteral reflux, pelvic-ureteric junction obstruction and other rare congenital anomalies. A clear definition of hydronephrosis would be that it is a dilatation of the calyces and the renal pelvis of over 10 mm or more than 50% of the anteroposterior diameter of the kidney. This dilatation of the urinary system may occur at the upper urethral segment, ureters, bladder or kidney pelvis. Pyelectasis is the dilatation of the renal pelvis only

There is no clear consensus on the follow-up and management of mild or moderate hydronephrosis observed on antenatal ultrasound, although it is generally

accepted that a postnatal assessment should be performed if the AP diameter of the renal pelvis exceeds 10 mm at any point in gestation [17].

Many studies show that a right or left prenatal AP renal pelvic diameter > 4 mm is associated with a higher risk of postnatal hydronephrosis compared with a right and left prenatal AP renal pelvic diameter ≤ 4 mm [15, 16]. Male neonates have a higher risk of postnatal hydronephrosis than females. These results can assist in establishing the appropriate follow-up method and evaluation of fetuses with renal pelvic dilatation.

2.5. Ureteral malformations

In medical practice many abnormalities can occur, like:

Number abnormalities

Structure abnormalities

Calibration abnormalities

Vesicoureteral reflux (VUR)

Abnormalities of the pyeloureteral junction

Position and opening abnormalities

Ureteral abnormalities

Ureteral agenesis is accompanied by the absence of the ipsilateral kidney. It may be uni- or bilateral, the latter being incompatible with life.

Incomplete ureteral duplication (ureter fissus or ureter bifid)—a double-sided pyeloureteral system: the ureters bend before opening into the bladder through a single orifice.

Complete ureteral duplication (duplex ureter)—a double-sided pyeloureteral collection system: the ureters open through separate boreholes in the bladder, one in orthoposition and the other in the ectopic position.

The ureteropelvic junction obstruction

Obstruction of the ureterobladder junction (obstructive megaureter) or obstruction on pyeloureteral junction is one of the most common causes of hydronephrosis found in children, its incidence being 1 case in 1000–2000 newborns. In this situation, hydronephrosis may be caused by an abnormal shape of

the ureteral junction—the existence of the ureteral valves and the mucosal fold at this level. Like many other abnormalities, this pathology occurs much more frequently in males, usually unilaterally and especially on the left side [10, 14].

The ultrasound diagnosis is based on the observation of the increase of the anteroposterior diameter of the kidney, a degree of kidney pyelectasis. If the obstruction is unilateral and the filling of the bladder is normal, the normal amount of amniotic fluid will be preserved. When the damage is bilateral, the oligoamnios/oligohydramnios may appear.

In most cases this condition does not produce symptoms after birth; in 10–15% of the cases, it regresses spontaneously, but sometimes repeated evaluation is needed.

Obstruction of the ureterobladder junction

This condition is the cause of approximately 5–10% of all cases of dilatation of the urinary tract. Ultrasound diagnosis is based on dilated urethra, with renal pelvis dilatation and normal bladder image. The cause of the disease is the dysfunction of the lower ureter, the outcome is favorable and the pathologic aspects disappear postnatally in many cases.

The normal ureter size in children is up to 5 mm. Normal ureter cannot be detected during prenatal ultrasound examination. It can only be detected if it is dilated to a size greater than 7 mm and is observed like a translucent structure. The dilated ureter should be differentiated from full intestinal loops. Pathological dilatation of the ureter is more common in male patients, usually appearing to be unilateral.

The dilatation of the ureter may be partial or total, and this pathology can be classified into the following conditions:

Obstructive-type megaureter

Reflux-type megaureter

Megaureter without reflux and without obstruction

Obstructive- and reflux-type megaureter

Vesicoureteral reflux

Vesicoureteral reflux (VUR) is characterized by the return of urine from the bladder to the kidney which often causes hydronephrosis and sometimes even abnormalities in kidney development (renal dysplasia). Causes include transient obstruction of bladder discharge, delayed maturation of the vesicoureteral junction and high bladder evacuation pressure. Patients with vesicoureteral reflux have a high risk of pyelonephritis, hypertension and progressive renal failure. Vesicoureteral reflux is represented by the retrograde passage of urine from the bladder to the ureter and subsequently to the kidneys, due to the incompetence of antireflux mechanisms.

This condition is usually easy to diagnose prenatally, when visualizing renal pelvis and calyces dilatations that vary in size, associated with unilateral or bilateral ureteral dilatations. RVU reveals a nonprogressive hydronephrosis with normal amniotic fluid index.

VUR classification:

Grade I reflux: reflux is only present in the ureter that has various degrees of dilatation.

Grade II reflux: reflux reaches the renal pelvis, without dilatation of the collector system, the papillae being normal.

Grade III reflux: moderate dilatation of the ureter, with or without sinusitis; moderate dilatation of the collector system.

Grade IV reflux: moderate dilatation of the ureter with or without sinuosity; moderate dilatation of the collector system; calyces are flattened but with the impression of the papilla still visible.

Grade V reflux: severe dilatation with ureter sinuosity and marked dilatation of the collector system; reflux in the parenchyma, with parenchymal thinning.

The prognosis is good. The prenatal diagnosis is important. Forty percent of the newborns develop severe kidney damage [2].

Ureterocele

As many other kidney abnormalities, ureterocele is more common in male patients. This pathology appears as a thin wall cyst or sept on the bladder wall. In many cases the diagnosis may be omitted when the bladder is full, the filling

masking the cyst. There have been described cases of ureterocele shed in the urethra. This phenomenon may lead to acute obstruction of the lower urinary tract. The prognosis is favorable, but its evolution must be assessed postnatally, by residual renal function testing and reflux tests

3. Bladder anomalies

The bladder is a very important element in antenatal fetal examination. Clinicians may suspect anomalies of the bladder when not visualized on prenatal examination or when enlarged. From the pathological point of view, we may encounter various anomalies as well:

Megacystis is a neurodysplastic disease, due to the anomaly of innervation. This leads to bladder dilatation (over 7 cm in diameter). It may be associated with small, short and dilated bowels, as well as a low-caliber colon. At ultrasound, the striking signs are polyhydramnios, enlarged ureters and kidneys [17].

Congenital bladder diverticulitis. A septum can be observed.

Bladder exstrophy. It is commonly associated with other urogenital malformations and is constantly accompanied by epispadias. This pathology is characterized by many anomalies and lesions, such as:

Abdominal anterior wall injuries: inguinal hernia is common.

Lesion of pelvic bones: shortening and defects of rotation of pubic bones.

Lesions of the genitalia: micropenis and cryptorchidism appear in boys; in girls, the vagina is short and has various degrees of stenosis, the clitoris is bifid and the small labia are divergent.

The anterior bladder wall is absent, and the posterior wall is the submuscular segment of the anterior abdominal wall.

The bladder mucosa is exposed to the external environment, with the ureteric holes and the inner urethra opening visible.

Vesicoureteral reflux is present in all cases.

Some patients have rectal prolapse and anal incontinence; this phenomenon is due to the malposition of sphincter structures.

4. Urethral malformations

The most common obstructive cause of the lower urinary tract is the posterior urethral valve (PUV). PUV is due to the existence of membranes in the posterior urethra. Echography shows dilated bladder and urethral dilatation, giving a classic echographic sign—'the keyhole sign'. Dystriation of ureters and troughs also occurs. In advanced forms, oligohydramnios-associated pulmonary hypoplasia and urinary ascites may also appear. These fetuses require prenatal intervention to avoid kidney damage. Some authors tried to demonstrate that creating a vesicoamniotic passage may lead to a better outcome, but this has not yet been fully demonstrated.

In the prenatal life, other malformations may be seen:

Urethral agenesis (absence of urethra) is a rare malformation, incompatible with life unless an alternative communication between the bladder and amniotic sac exists. The urine is discharged through abnormal communication with other organs (between bladder and bowels or by umbilical fistula).

Congenital structures.

Rear urethral valves: mucosal lining located in the posterior urethra originating from the seminal colonic and insertion on the anterior wall of the urethra.

Epispadias: abnormality characterized by opening the urethra on the dorsal face of the penis at variable distance of the gland, dorsal curvature of the penis and foreskin abnormalities. There are many forms of epispadias:

Balanic: distal incomplete defect.

Penian: incomplete proximal defect.

Penopubian: a complete defect, often associated with bladder exstrophy. In this situation, a distinct pathological condition appears: the exstrophy-epispadias complex [19].

Hypospadias: the anomaly is characterized by opening the urethra on the ventral side of the penis, at any level between the gland and the perineum, causing a ventral penis (chord). Patients with hypospadias have often abnormalities of the foreskin and cryptorchidic scrotum, inguinal hernia and hydrocele. Studies have shown a higher presence of hypospadias in patients with fetal growth restriction

Treatment

The pediatric nephrologists with Norton Children's Nephrology can work with you and your child to create a unique treatment plan. Depending on your child's type of anomaly, treatment may include:

Antibiotics to prevent urinary tract infections.

Observation: Children can outgrow some issues with urine flow. Children also can live healthy lives with one kidney missing or damaged.

Surgery to address vesicoureteral reflux or urethra or ureters blockages.

When these kinds of congenital anomalies are not detected early or treatment is unable to prevent kidney damage, a child may experience chronic kidney disease or kidney failure that requires dialysis and kidney transplantation.

VIII. CONGENITAL MALFORMATIONS OF THE ANTERIOR ABDOMINAL WALL

Anterior Abdominal Wall Hernias

Anterior abdominal wall hernias, also known as ventral hernias, are a leading cause of abdominal surgery in the United States (,1). These hernias involve the protrusion of part of the peritoneal sac through a defect in the muscle layers of the anterior abdominal wall. They are classified on the basis of their location or cause. The major types include incisional hernias, parastomal hernias, umbilical or periumbilical hernias, and spigelian hernias. With the increasing frequency of abdominal surgery, incisional hernias have become the most common type of anterior abdominal wall hernia. Incisional hernias occur at sites of weakening along postoperative incisions, usually at the midline (,2,,3). In our experience, parastomal hernias also are common, as ileostomies and colostomies are frequently performed by the surgical service at our hospital.

Anterior abdominal wall hernias may contain a variety of intraperitoneal structures, including fat, omentum, and bowel. Bowel-containing hernias are particularly important because of potential complications related to incarceration, obstruction, or strangulation of the bowel, which lead to ischemia and infarction (,1). The features of anterior abdominal wall hernias at computed tomography (CT) and magnetic resonance imaging are well documented. However, little information is available about the detection and characterization of such hernias in barium studies, primarily in small-bowel follow-through examinations. The data from a recent survey show that nearly two-thirds of radiology groups perform at least as many barium studies of the small bowel as were performed 5 years ago, and some perform more. Radiologists therefore should be familiar with the various features of anterior abdominal wall hernias in barium studies.

Clinical Findings

Anterior abdominal wall hernias can be diagnosed on clinical grounds when a physical examination reveals a bulge in the abdominal wall that is visualized or palpated with or without a Valsalva maneuver to increase intra-abdominal pressure. Some patients present with abdominal pain or, if the hernia is incarcerated (ie, nonreducible) or strangulated, with signs and symptoms of intestinal obstruction or ischemia, but others have no symptoms that are attributable to a hernia of this type. Anterior abdominal wall hernias also can be difficult to detect at physical examination because of obesity or abdominal pain that limits the physician's ability to palpate the abdomen.

As a result, anterior abdominal wall hernias, particularly small hernias in asymptomatic patients, often are not detected because the clinical findings do not evoke suspicion.

Types of Abdominal Wall Hernias

Epigastric Hernia

An epigastric hernia occurs when a weakened area in the abdominal wall allows a bit of fat to push through. Epigastric hernias are typically small. They occur in the middle of the belly, in the area between the belly button and the breastbone.

Some patients develop more than one epigastric hernia at a time. These hernias typically don't cause symptoms, but you may experience pain in your upper belly.

Treatment of epigastric hernias typically involves surgery, but your doctor will discuss all your options with you in detail.

Incisional Hernia

A hernia that occurs in the area of a previous surgery is known as an incisional hernia. These hernias may occur when the abdominal wall has been weakened by surgery, or when a surgical incision becomes infected, further weakening the area.

Incisional hernias are relatively common because surgical incisions weaken the abdominal area. That weakness makes it easier for a part of the intestine or other tissue to protrude.

Incisional hernias can develop soon after surgery, or they can develop slowly, over months or even years. They typically occur alongside vertical incisions. Incisional hernias tend to be large and rather painful.

Incisional hernias will not heal on their own. Talk to your doctor for more details about your treatment options.

Spigelian Hernia

Spigelian hernias occur throughout the spigelian fascia. Unlike other types of hernias, which develop immediately below layers of fat, spigelian hernias occur in the midst of abdominal muscles. This means that spigelian hernias may not be immediately visible as a bulge or lump. They can go undetected for longer periods of time.

Because spigelian hernias tend to be small, the risk of developing a strangulated hernia is higher.

Spigelian hernias tend to occur more rarely than other types of hernias.

Umbilical Hernia

An umbilical hernia occurs when a weak spot in the belly allows a bit of fat, fluid, or intestine to push through, creating a lump or bulge near the belly button.

Umbilical hernias frequently occur in infants. In most cases, these hernias will heal on their own. Occasionally, however, surgery may be required.

Umbilical hernias also occur in adults, particularly those with health issues that cause increased pressure in the belly. Those issues may include obesity, pregnancy, chronic coughing or constipation, and difficulty urinating.

Because they tend to grow bigger over time, umbilical hernias require treatment. Treatment typically involves surgery, but your doctor will discuss all your options with you in detail. Without treatment, you are at risk of developing a strangulated hernia, which means that blood supply to the tissue has been cut off. **Strangulation is life threatening and requires emergency surgery.**

Symptoms

Abdominal wall hernias are generally visible: they will look like a lump or bulge beneath the skin. These hernias don’t usually cause any other symptoms

except for mild pain or discomfort, usually when you are straining (for instance, lifting something heavy).

If the hernia becomes strangulated, however, you may experience more serious symptoms, such as severe pain, nausea, vomiting, and/or redness in the area of the hernia. Contact your doctor immediately if you begin to experience these symptoms. **A strangulated hernia is life-threatening.**

Causes

Abdominal wall hernias can occur in people of any age, including infants. However, the risk of developing a hernia tends to increase as you age. Most abdominal wall hernias are caused by an area of weakness in the abdominal walls. A number of different factors can contribute to the development of that weakness. These factors include:

- Aging
- Chronic coughing
- Collagen vascular disease
- Frequent heavy lifting
- Genetic defects
- History of previous hernias
- Infection (especially following surgery)
- Injuries to the abdominal area
- Obesity
- Pregnancy
- Straining during bowel movements or urination
- Surgical openings

Diagnosis

In most cases, your doctor will be able to determine whether you are in fact suffering from a hernia simply by looking and by gently palpating the affected area.

If for some reason a diagnosis isn’t immediately apparent, your doctor may decide to order an imaging test, such as an abdominal ultrasound, CT scan, or MRI.

These imaging tests can help to show the hole in the muscle wall, along with the tissue protruding from it.

Treatment

Abdominal wall hernias that have no associated symptoms may not require any treatment at all. Your doctor will discuss your options with you which may include surgery or watchful waiting.

Larger hernias, however, or hernias that are causing pain, may require surgical repair to relieve pain as well as to prevent complications.

There are two types of surgical hernia repair: open and minimally invasive surgery. The type of surgery chosen will depend on the severity and type of hernia you've developed, the anticipated recovery time, your past medical and surgical history, and your surgeon's expertise.

Open Surgery: During this procedure, your surgeon will make a small incision into your groin, and then push the protruding tissue back into your abdomen. Your surgeon will then sew up the weakened area. In some cases, your surgeon will use a mesh to reinforce that weakened area.

Open surgery can be performed either with general anesthesia or with sedation or local anesthesia.

After your surgery, it might be several weeks before you're able to fully resume your normal activities. However, it's still important that you begin moving about again as soon as possible for a healthier recovery.

Minimally Invasive Surgery: Minimally invasive surgery is typically performed under general anesthesia.

During this procedure, your surgeon will make a few small incisions in your abdomen. Your surgeon will then inflate your abdomen, using a special gas, in order to make your internal organs easier to see.

Your surgeon will then insert a small, narrow tube into one of the incisions in your abdomen. This tube has a tiny camera, or laparoscope, at the end of it. That camera serves as a kind of guide for your surgeon, who is then able to insert surgical

instruments through the other incisions in your abdomen. Your surgeon will repair the hernia using and may use mesh.

Patients who are candidates for minimally invasive surgery may experience less scarring and discomfort following surgery than those who undergo open surgery. Patients may also be able to return more quickly to their normal activities.

Your doctors will speak with you in detail about all of your treatment options and will recommend a course of action best suited to your individual needs.

IX. Materials for independent work

What is the annual incidence of testicular torsion in young males?

a. 1: 500
b. 1: 4000
c. 1: 10 000
d. 1: 20 000
e. 1: 100 000

In the history of a patient with an acute scrotum, what sign should be asked about in particular?

a. petechiae
b. nevus lenticularis
c. erythema
d. milia
e. comedones

Question 3

What reflex should be tested as part of the diagnostic evaluation of the acute scrotum?

a. the anal reflex
b. the pupillary light reflex
c. the abdominal wall reflex
d. the knee-jerk reflex
e. the cremasteric reflex

Question 4

Which of the following is essential in the ultrasonographic evaluation of the acute scrotum, aside from the expertise of the examiner?

a. a 1.5–3.5 MHz linear transducer with Doppler or power-Doppler function
b. a 2–4 MHz linear transducer with Doppler or power-Doppler function
c. a 2–5 MHz linear transducer with Doppler or power-Doppler function
d. a 4–7 MHz linear transducer with Doppler or power-Doppler function
e. a 7–13 MHz linear transducer with Doppler or power-Doppler function

Question 5

A six-year-old boy presents with a painful acute scrotum but feels better rapidly thereafter. Doppler ultrasonography reveals a hyperperfused testis. What is your suspected diagnosis?

a. blunt trauma
b. testicular tumor

c. intermittent testicular torsion

d. appendicitis

e. incarcerated inguinal hernia

Question 6

Palpation of the acute scrotum of a 10-year-old boy reveals maximum tenderness above the testis. Transillumination reveals a bluish structure. Ultrasonography reveals a round, hyperechogenic, non-perfused structure next to the upper pole of the testis. What is the most likely diagnosis?

a. scrotal edema

b. hydatid torsion

c. epididymitis

d. intravaginal testicular torsion

e. neonatal testicular torsion

Question 7

A standardized diagnostic evaluation including Doppler ultrasonography in a patient with an acute scrotum fails to demonstrate adequate central perfusion of the testis with certainty. In this situation, what is the treatment of choice?

a. emergency surgical exploration of the testis

b. intravenous antibiotics

c. perfusion-promoting drugs

d. bed rest

e. measurement of tumor markers

Question 8

Doppler ultrasonography reveals a scrotal wall that is expanded by edema. Individual layers can no longer be distinguished. The central perfusion of the testis is within normal limits. What is the most likely diagnosis?

a. hematocele

b. hydatid torsion

c. idiopathic scrotal edema

d. intermittent torsion

e. orchitis

Question 9

Which of the following can be used to treat hydatid torsion?

a. anti-inflammatory drugs

b. antibiotics

c. anti-diuretic drugs
d. anti-arrhythmic drugs
e. anti-emetic drugs

Question 10

What disease can present with scrotal involvement as its initial manifestation?

a. diabetes
b. leukemia
c. hypertension
d. plasmocytoma
e. rheumatoid arthritis

X. Educational tasks at the final stage of study

1. A family doctor examines the home of a 6-month-old baby, which, according to her mother, 8:00 ago became very restless, screaming, pushing, banging her legs. At first, the attacks were replaced by “bright intervals”, repeated vomiting appeared, and dark blood appeared on the diapers after the stool. From the anamnesis, the doctor found out that the mother first gave the child vegetable puree. On examination, palpation of the abdomen is painless, there is no muscle tension, an oval form of formation is determined in the right hypochondrium, desolation in the right iliac region.

1. Make a preliminary diagnosis.

2. Determine the tactics of the doctor

2. A child of 8 years after a diet violation has paroxysmal abdominal pain, repeated vomiting with an admixture of bile, and lack of stool during the day. A child 6 months ago underwent surgery for gangrenous appendicitis. Upon examination, the condition of the child of moderate severity, is in the knee-elbow position, the tongue is dry, coated. The abdomen is asymmetric, swollen in the upper half, pain in the postoperative scar, auscultation - peristalsis is enhanced. On the survey radiograph of the abdominal cavity there are single Kloiber cups.

1.Provide a preliminary diagnosis.

2. Define the tactics of treatment.

3. A child of 8 years suddenly had massive bloody vomiting. An hour later, blood appeared in the feces. From the anamnesis, it is known that 2 days before bleeding, the patient had weakness, pain in the epigastric region, fever up to 38 ° C, which was regarded as the onset of SARS. According to the mother, after the birth, the child was in the neonatal intensive care unit, where the umbilical vein was catheterized. On examination, there is weakness, lethargy, pallor of the skin, the patient complains

of thirst, there is an expansion of the veins of the anterior abdominal wall. Palpation is determined by splenomegaly.

1. Make a preliminary diagnosis.
2. Determine the doctor’s tactics.

4. A child of 3 years old fell ill unexpectedly, a significant amount of blood change appeared in the stool. No vomiting. General weakness. The abdomen is moderately painful on palpation of the navel. No medical history has been identified.

1. Establish a diagnosis.
2. Therapeutic tactics

5. A 2-year-old child has a sharp pain during bowel movements, the last portion of feces with streaks of red blood. A history of persistent constipation.

1. Make a preliminary diagnosis.
2. Determine the doctor’s tactics.

6. In a 6-year-old child, feces periodically marked with an admixture of red blood and mucus, blood in the last portion of feces. The general condition of the child is not broken. With rectal digital examination at a depth of 4 cm, a tumor-like formation, 1.5 - 1.5 cm, of a round shape on a narrow thin leg is determined.

1. Make a preliminary diagnosis.
2. Determine the doctor’s tactics.

7. A 9-year-old child was delivered to the front desk by ambulance. The boy was hit by a car 15 minutes ago. The child is conscious, does not answer questions, languid, dynamic, pale skin, pulse on the peripheral vessels filiform, intense, heart rate 95 beats. in minutes AO 70/0 mm RT. Art. When viewed in the right hypochondrium is abrasion.

1. What is your suspected diagnosis?
2. Define further tactics.

8. A boy of 10 years old was admitted to the hospital 40 minutes after falling from a tree complaining of pain in his left thigh, left forearm, chest and abdominal cavity. Examination revealed edema, deformation of the thigh and forearm, limitation of limb movements, abrasions in the left half of the chest, pulse - 110 beats per minute, blood pressure 90/50 mm

1.Provide a preliminary diagnosis.

2. Create a survey plan.

9. A girl of 13 years old was hospitalized in an emergency department. When examining the victim after a road accident after 6:00, cyanosis is observed, shortness of breath. The patient's condition is serious, the right half of the chest lags behind in the act of breathing, the intercostal spaces are widened, the case with percussion is box sound, there is no breathing during auscultation.

1. Repeat the methods that will help in the diagnosis?

2. What is your diagnosis?

3. What is emergency care?

10. A 5-year-old child was injured in a car accident. Arriving after 20 minutes at the scene, the ambulance doctor established the following. The child is fully conscious, sharply excited, pale, the body is covered with sweat. Breathing, central nervous system, head, chest, spine, abdomen - without clinical signs of damage. Pulse 140ud. / Min., Blood pressure - 90/45 mm RT. Art. The reaction of filling the subungual capillaries - C sec. The left thigh is deformed in the upper part, palpation of it causes a child's excitement and cry, there are no active movements in the joints of the left limb. On the left side of the pelvis are abrasions and a subcutaneous hematoma is common. Palpation of the wing of the left iliac bone causes a sharp pain and a protective reaction of the child (rejects the doctor’s hand). Diagnosis of an ambulance doctor: multiple trauma - a fracture of the left femur with displacement

of fragments, a fracture of the left iliac bone, soft tissue injury. Traumatic shock (severity not specified).

1. What are early signs of traumatic shock in children?
2. In the presence of traumatic shock, where should treatment begin?
3. What complications may arise in this patient?

11. A child aged 3 years who fell ill with a 12:00 volume aimed at hospitalization with a preliminary diagnosis of acute appendicitis. The child had vomiting, stool twice, loose, without impurities. When viewed at the reception, there is no sufficient data for acute appendicitis.

1. What will be the tactics of the doctor of the admission department?
2. What research methods should be prescribed to the child?

12. A 2-year-old child fell ill 20 hours ago: anxiety appeared, the child began to clutch at his stomach, twice had vomiting, and then frequent loose stools appeared. Body temperature - 38.7 ° C. The tongue is coated with a white coating, dry. The abdomen is inactive in breathing, palpation is not possible due to the expressed anxiety of the child and the tension of the anterior abdominal wall. With digital rectal examination, an overhang of the anterior rectal wall was detected.

1. What preliminary diagnosis can be made to the child.
2. Assign preoperative preparation to the child.
3. What treatment method to use in this case?

13. A child of 6 years old experienced acute abdominal pain, vomiting, febrile body temperature, accompanied by frequent urges to defecate, and liquid bowel movements with mucus. The condition of the child worsens, significant manifestations of intoxication increase.

1. Which of the atypical localizations of the appendix during its inflammation can give a similar picture?
2. What research methods should be assigned to the child.

3. Perform differential diagnostics.

4. What treatment method should be chosen.

14. A girl of 6 years old was transferred to the children's surgical department with complaints of high body temperature, general weakness, cough, catarrhal phenomena. The severity of the child's condition due to respiratory failure, is sick for 5 days. During auscultation, the matter is hard breathing, on the left - breathing is not audible on the left in the upper sections - tympanitis, in the lower sections - blunting. In the survey roentgenogram, upper left - enlightenment, homogeneous dimming with the liquid level below the IV rib.

What is your diagnosis?

1. With what diseases will you conduct a differential diagnosis?

2. What diagnostic methods need to be carried out?

3. The basic principles of treatment of this patient

4. First aid, surgical treatment.

15. A 5-year-old boy is undergoing treatment in the surgical department in connection with destructive pneumonia. The child complains of coughing, high body temperature, weakness, and the child’s dynamics have become easier. When examining a child - respiratory failure decreased. Above the lungs on the left - hard breathing, on the right - hard breathing, inhomogeneous over the entire surface, with percussion - tympanitis. On the survey roentgenogram, thin-walled formations from 3 to 4 cm in diameter, filled with air, are a matter.

What is your diagnosis?

1. What diagnostic methods will help establish a diagnosis and determine treatment tactics?

2. What diseases should be diagnosed with?

3. The basic principles of the treatment of pulmonary forms of HDH.

16. A 7-year-old boy is undergoing treatment in the intensive care unit with a septic state, bilateral pneumonia. Intensive therapy, continuous rehabilitation of the

tracheobronchial tree. Suddenly, the child's condition worsened, and respiratory failure began to increase. During auscultation, breathing is not heard on the left, dullness of the pulmonary sound is percussion, displacement of the mediastinum and cardiac impulse to the left. The thing - breathing is weakened, percussion - in the lower sections, the dullness of the pulmonary sound.

What is your diagnosis?

1. What complication arose in the patient?
2. What are the diagnostic methods.
3. What are your immediate actions?
4. What is the tactics of patient management.

17. A two-month-old child is hospitalized in the surgical department. The mother complains of an increase in body temperature to 38.5 ° C, edema, hyperemia and lack of movement in the region of the left shoulder joint. In the anamnesis: omphalitis, pseudofurunculosis.

1. What is the most likely diagnosis?
2. The doctor’s tactics in identifying the disease.
3. The etiology and pathogenesis of the disease.
4. The main directions of treatment.
5. What are the features of the follow-up of a child after recovery?

19. A child, 12 years old, is sick for 2 days. Complains of hypertension, pain in the lower third of the right thigh and in the knee joint. History: 3 days ago there was an injury. Objectively: a moderate increase in local skin temperature, slight soft tissue infiltration. During percussion of this site, local pain intensifies. A preliminary diagnosis was made: acute hematogenous osteomyelitis of the lower third of the right thigh. What is the cause of pain with osteomyelitis in the early days of the disease?

1. Your diagnosis.
2. What measures should be taken to prevent the disease?
3. What measures should the surgeon take first.
4. What is the cause of pain during percussion of the femur?

5. What conservative measures should be used in the treatment of the disease?

20. A child of 2 years old was admitted 2 days after the onset of the disease. The condition worsened, hyperthermia, anxiety appeared. In the region of the middle third of the shoulder, edema, hyperemia of the skin with clear contours is determined. On palpation, sharp pain and softening of the skin in the center of formation, fluctuation are determined.

1. Your diagnosis.
2. The doctor's tactics in identifying the disease.
3. The etiology and pathogenesis of the disease.
4. The main directions of treatment.
5. What preventive measures should be taken after recovery?

21. A 13-year-old boy began to worry about pains in the upper third of his left thigh, his body temperature increased to 39 ° C. Hip swelling in the upper third and smoothness of the inguinal fold, hyperemia of the skin, and an increase in their density were noted. The ending is in a bent position. Active and passive movements are not possible due to sharp pain.

1. What is the most likely diagnosis?
2. What measures should be taken to prevent the disease?
3. What measures should the surgeon take first.
4. What is the cause of pain during percussion of the femur?
5. What conservative measures should be used in the treatment of the disease?

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