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Ultrasonographic diagnosis of congenital fetal anomaly - intestinal atresia

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Annotation: The article provides detailed information about congenital intestinal obstruction and intestinal atresia. It explains that various congenital anomalies arise due to disturbances in the formation, rotation, and positioning of the intestines during fetal development. Intestinal atresia is described as a condition in which a segment of the intestine is closed or absent, leading to obstruction of food passage and posing a serious risk.In addition, malrotation, malformation, and malfixation syndromes are discussed as major developmental abnormalities of the intestines. The article outlines the clinical manifestations of these conditions, including vomiting, abdominal distension, failure to pass stool, dehydration, and feeding difficulties. Furthermore, the causes, clinical features, and diagnosis of pylorospasm and congenital pyloric stenosis are also described. The conclusion emphasizes that early diagnosis and surgical treatment of these conditions are crucial for saving children's lives.

Keywords: intestinal atresia, malrotation syndrome, malformation syndrome, congenital intestinal obstruction, congenital pyloric stenosis.

Relevance of the topic: Intestinal atresia is a congenital condition in which the fetal intestine fails to develop completely or a segment of the intestine is born closed. This condition is considered dangerous because, after birth, food and intestinal contents cannot pass through the bowel.

Main risks include: Intestinal obstruction — the newborn begins to vomit, develops abdominal distension, and fails to pass stool. Dehydration and inability to feed properly. Damage to or perforation of the intestinal wall.

Risk of infection and sepsis. If left untreated, it can be life-threatening. In most cases, surgery is required after birth. With modern surgical techniques, many children recover well, especially if the condition is diagnosed in time.

Research Methods and Results: Congenital intestinal obstruction is a condition in which the passage of food (including breast milk) is impaired or completely blocked due to a congenital obstruction in the gastrointestinal tract.The causes of congenital intestinal obstruction are diverse and can be classified as follows:Malrotation syndrome — resulting from abnormal rotation of the intestines;Malformation syndrome — due to defects in the formation of the intestinal tube and its wall;Malfixation syndrome — resulting from abnormal fixation and attachment of the intestines;Other congenital anomalies of the abdominal cavity organs, including meconium ileus (associated with cystic fibrosis of the pancreas), aberrant blood vessels, annular pancreas, and intra-abdominal cysts.Formation of the Digestive Tract:Congenital intestinal obstructions are classified into upper and lower intestinal obstructions based on the level of obstruction. The anatomical landmark separating them is the ligament of Treitz. Regardless of the cause or anatomical variation, both upper and lower intestinal obstructions located above and below the ligament of Treitz present almost identical clinical and radiological features; therefore, congenital intestinal obstructions are divided into upper and lower types based on this classification.According to the clinical course, intestinal obstruction may be acute, chronic, or recurrent.Embryogenesis of the intestines: the formation of the intestinal tube and its rotation. In the early stages of embryonic development, the primitive intestinal tube is a tubular structure connected by a primary mesentery in the region of the superior mesenteric artery within the abdominal cavity. During development, it undergoes a physiological embryonic herniation stage.At the same time, processes of intestinal rotation (rotation), formation of intestinal loops and mesentery (formation), and fixation (fixation —

stabilization) begin. Disruption of these processes, either individually or in combination, due to various causes may lead to congenital malformations and anomalies of the gastrointestinal tract.

Discussion of Results: Disruption of these processes leads to the following conditions:A disorder of intestinal rotation and the resulting syndrome or disease is called malrotation syndrome;A disturbance in the formation and development of the intestinal wall and its mesentery is referred to as malformation syndrome;A disturbance in intestinal fixation (fixation refers to the process by which abdominal organs take their normal position and acquire mesentery) and the formation of mesenteries, resulting in a syndrome or disease, is called malfixation syndrome. Each of these will be discussed separately.During its embryonic development, the primary tubular intestine undergoes a cord-like stage. Due to epithelial proliferation, the primary intestinal tube becomes completely occluded, and during the subsequent vacuolization (recanalization) process, the lumen of the intestine is restored to a tubular structure. However, due to certain influences, parts of the intestine may remain closed.If the recanalization process is disrupted in a small portion of the intestinal tube, the lumen may be covered by a thin membrane, leading to membranous atresia. If recanalization begins in a small segment but is later disrupted, a small (or variably sized) opening may remain, resulting in membranous stenosis. If the recanalization process is disrupted in a large portion of the intestinal tube, atresia develops over a large segment, leading to fibrous atresia. If the disruption occurs in multiple separate areas of the intestinal tube, a sausage-shaped (segmental) atresia may develop.Sometimes, intestinal atresia is not caused by a failure of recanalization but rather by impaired mesenteric blood circulation. Intestinal atresias most commonly occur in those parts of the intestinal tube where complex embryological processes take place.Conditions resulting from disruption of embryonic processes are classified as malformation syndrome. As mentioned above, in the early stages of embryonic development, the primitive intestinal tube is a tubular structure connected by a primary (common) mesentery in the region of the superior mesenteric artery of the abdominal cavity. Later, the process of intestinal rotation begins: the “midgut” (from the duodenum to the middle of the colon) undergoes a stage of physiological embryonic herniation during development and rotates 270° counterclockwise before returning to the abdominal cavity.Initially, the small intestinal loops descend into the abdominal cavity, while the large intestine moves from the left iliac region upward under the spleen, then to the right side under the liver, and finally to the right iliac region. As a result, the duodenum remains located behind the colon. During rotation, the small and large intestines acquire separate mesenteries and take their proper positions in the abdominal cavity.Disorders of rotation (strangulation) and defects in mesentery formation may lead to the formation of peritoneal pockets (bursa), internal hernias, and intestinal obstruction. Abnormal positioning of the small and large intestines during rotation results in malrotation syndrome. Abnormalities in mesentery formation lead to malfixation syndrome.During embryonic development, if the intestines partially or completely remain in the abdominal cavity during the physiological embryonic herniation stage, various sizes of embryonic umbilical cord hernias and omphalocele may develop.Pylorospasm is a functional insufficiency of the gastric pyloric sphincter. Due to functional immaturity of the nerve fibers, innervation is impaired, resulting in persistent spasm of the pyloric sphincter. The pyloric smooth muscles are morphologically normal, but due to impaired neural regulation, vomiting is the main clinical symptom.Clinical features: vomiting begins after birth, every other day. The volume of vomitus is less than the amount of milk consumed per feeding. Stool is normal. Urination is slightly reduced. Skin color is pale. The infant is slightly irritable. Body weight is unchanged or slightly below normal for age.Differential diagnosis: mainly compared with conditions causing vomiting in newborns, especially congenital pyloric stenosis. In contrast to congenital pyloric stenosis, in pylorospasm, contrast radiography shows that barium passes freely and completely from the stomach into the intestinesTreatment: atropine therapy is effective. Nutritional correction is also recommended.Congenital Pyloric StenosisCongenital pyloric stenosis is a group of diseases in newborns characterized by vomiting syndrome and requiring surgical correction. Morphologically, it is a congenital narrowing of the pyloric part of the stomach due to hypertrophy of the circular muscles of the pylorus. Clinically, it is a condition in which breast milk cannot pass into the intestines, leading to persistent vomiting. In this condition,

connective tissue fibers infiltrate and intertwine with muscle layers. The thickened area is mainly composed of circular muscle fibers. Fibrous tissue surrounds and compresses the nerve bundles located within the muscle layer.Subsequently, nerve cells become compressed and twisted, glial elements undergo hyperplasia, and nerve fibers are deformed within the serous layer. The affected area becomes hardened with a cartilage-like consistency. Clinical features.The symptoms of the disease usually begin in newborns at 2–4 weeks of age. The main clinical sign is projectile vomiting. In affected infants, vomiting occurs shortly after breastfeeding or formula feeding, either once or multiple times. The volume of vomitus may exceed the amount of milk consumed and has an unpleasant odor.The child begins to lose weight, the skin becomes dry, urine output decreases, and constipation develops. Frequent vomiting makes the child weak and lethargic. By the age of two months, the infant's weight is lower than at birth. Subcutaneous fat decreases, the abdominal skin becomes wrinkled, the eyes appear sunken, and the fontanelle is depressed. The external appearance resembles an "aged" or emaciated old person.Another key sign of pyloric stenosis is increased gastric peristalsis and the "sand-timer" (or "olive-like") sign visible through the thin abdominal wall. In some cases, the hypertrophied pyloric sphincter can be palpated. The acute form of the disease rapidly worsens the infant's condition within one week, leading to malnutrition. In this condition, water-electrolyte and acid-base balance is quickly disturbed. Laboratory tests show hemoconcentration (increased hemoglobin and hematocrit levels, with decreased chloride and potassium levels in the blood).For accurate diagnosis, radiological and endoscopic examinations are performed. These are usually done in the morning on an empty stomach. Fibrogastroscopy reveals dilation of the gastric inlet and narrowing of the pyloric region. Initially, a plain abdominal X-ray is performed to assess gastric size. Then a barium suspension (1:1 dilution with water) is administered, followed by X-rays taken after 2–3 hours and again after 23–24 hours.Differential diagnosis .It is differentiated from pylorospasm, adrenogenital (pseudo-pyloric stenosis) syndrome, gastroesophageal reflux, stenosis above the Vater's papilla of the duodenum, congenital partial upper intestinal obstruction, postnatal brain injury–related vomiting syndrome, diarrhea, and vomiting caused by toxic infections.In comparison with adrenogenital syndrome, bile is present in vomit. Blood potassium is elevated while sodium is decreased, and contrast medium passes easily through the pyloric region on X-ray. It is also differentiated from pylorospasm.ConclusionIntestinal atresia and congenital intestinal obstructions are dangerous congenital conditions resulting from disturbances in embryonic development. In these cases, a part of the intestine is either completely closed or abnormally formed, preventing the passage of food after birth. The main symptoms include vomiting, abdominal distension, failure to pass stool, dehydration, and feeding difficulties. Intestinal atresia develops due to disruption of the recanalization process or impaired blood circulation. Malrotation, malformation, and malfixation syndromes are also associated with abnormal rotation and positioning of the intestines, leading to congenital defects. If not diagnosed in time, the condition can be life-threatening; however, with modern diagnostic and surgical techniques, many children can be successfully treated**.**

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