# Heterotaxia visceral asplenia pdf

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Although the human body appears to have relative symmetry, the same is not true at the level of the chest organs, abs or even the brain, where the general rule is asymmetry or laterality (e.g., the stomach abdominal organ is usually located on the left and the liver on the right, the right and left lungs have different anatomical characteristics,..). This normal asymmetrical development of organs occurs during the embryonic period and is determined by unequal genetic expression, which will lead to different morphogenesis on both sides of the body. Situs Solitus and Situs Inversus Normal expression of laterality in humans will be called Situs Solitus, defined as the one in which the organs are usually located. The upper left figure is a bespoke solitus situs with the liver on the right, the stomach on the left, the right lung with three lobes and the right ear. Sometimes the normal situation is completely reversed, so the bodies are usually right on the left and vice versa. This situation is known as Situs Inversus, as shown in the upper right figure, and affects approximately 1/8,500 living newborns. It is noteworthy that this anomaly is associated with congenital heart disease by only 0.6-0.8%. Although this link is therefore not common, it should be taken into account when diagnosing the reverse conditions. The ambiguous Situs, Isomerism, Heterotaxia Another very different situation arises when we talk about isomerism, heterotaxia or ambiguous Shitus. In this case (1-1,5/10000 live newborns) there is an identical genetic expression on both sides of the body (visceral symmetry), i.e. both sides of the body have exactly the same organ location and morphology. Thus, it is a failure in the normal establishment of left-right asymmetry during embryonic development. Both genetic and environmental factors have been implicated in the origins of this problem. From a genetic point of view, the most common are unique cases due to spontaneous mutations, although family inheritance patterns have also been described in the spectrum of isomerism, we have to distinguish between two types: Right Isomerism or Ivemark syndrome (Upper Left Pattern): There is a tendency towards right symmetry. For this reason it is also known as Aspnium syndrome (lack of spleen, which is typically left organ). At the heart level, it is recognizable by the presence of two morphologically right atrius. Left isomerism or Polysplenia syndrome (upper right figure): tendency to left symmetry. Although the availability of scoping, they usually do not function. At the heart level, it is recognized by the presence of two morphologically left atria. The cardiac abnormalities associated with isomerism can be multiple and varied: anomalies due to veins of cava that return with non-oxygenated blood from the body. Anomalies due to pulmonary veins that return with oxygen blood from the lungs. Defects in the septum between the atria. Anomalies in the input valves in the ventricles are common in order to be one input valve. Defects in the septum between the ventricles. See one ventricle. Lack of development of one of the ventricles of the heart. Anomalies in the connection of the vessels of the heart exit in relation to their ventricles. Obstruction of the vascular output, mainly from the pulmonary artery. Anomalies in the development of pulmonary arteries. Generally, although not always true, right isomerism is most often associated with the presence of congenital heart defects (the presence of some type of congenital heart abnormality occurs in almost 100% of cases) and with more complex forms of heart disease. There may also be heart rhythm disturbances in the form of episodes of tachycardia (abnormally high pulse) or bradycardia (abnormally slow pulse). Left isomerism can be associated with bradycardia, altering the normal function of pacemakers or altering the conductivity of the electrical impulse. Sometimes this can occur during the fetus, so it is a diagnosis that should always be taken into account in the case of fetal bradycardia. An early debut may include poor prognosis, and in some cases it could be pointed to pre-natal birth and early implantation of pacemakers after childbirth. Izomerism, heterotaxia and one ventricle. In general, the most complex forms of heart disease associated with isomerism of one functional ventricle, in which case surgical variants will be intended for single-ventricular surgical palliative pallation type Fountain, with the previous bikavopulmonar anatomous, usually in 2-3 surgical stays. See one ventricle. Cases of univenricular heart associated with isomerism have a particular risk and require particularly refined management from birth, as well as more complex surgical methods. On the other hand, the need for diagnostic or even therapeutic catheterization between different surgical stays is common and then during follow-up. Faced with a diagnosis of isomerism/heterotaxia: The significance of this type of anomaly is determined by its high association with congenital heart defects, many of which are complex, and pathology (gastrointestinal, pulmonary, genitourinary, neurological,..). Prenatal diagnosis of congenital heart defects and immediate management from birth are especially important. In the case of diagnosis of isomerism during the fetus, it is important to make a specific depisation of congenital heart disease using fetal echocardiography, trying to optimize the diagnosis of possible related abnormalities, in order to provide maximum information and prenatal advice to parents, planning an adequate follow-up during pregnancy and predicting possible problems after birth. Childbirth should be carried out at a centre specializing in the treatment of complex congenital heart defects, as there is a possibility that the newborn may need urgent intervention. Among the problems that most often require immediate treatment after birth: Extreme bradycardia. Severe blockage of pulmonary veins. Severe blockage of the flow to the pulmonary arteries or artery of the aorta. There are cases that do not require immediate intervention after birth, but may be required within the first few months of life. Extracardiacal anomalies Finally, it should be remembered that although we have focused the chapter on congenital cardiac abnormalities, this disease can affect several other organs. Although some abnormalities may not have clinical consequences, numerous problems can occur from the birth or throughout the patient's life. Here are some of them: gastrointestinal: malrota, intestinal obstruction, abdominal pain, cholestasis, pancreatic deficiency,... Lung/respiratory: bronchiectaz, sinusitis, infections,.. Genetics: urinary tract infections, infertility, impaired kidney function,.. Musmusoskel: cymocoliosis, hemi vertebrae, spina bifida,.. In short, isomerism, or heterothicy, carries a high probability of congenital involvement of the heart and other numerous organs, with very variable clinical expression after birth. Appropriate diagnosis and follow-up treatment during antenatal periods, as well as the birth of a child in a center specializing in congenital heart defects, in the conditions of immediate treatment is necessary. Situs ambiguusOther namesSitus ambiguous, heterotaxy, heterotaxiaSpecialtyCardiology Situs ambiguus is a rare birth defect in which the main visceral organs are abnormally distributed abnormally in the chest and abdomen. Heterotaxis generally refers to any defect of the left-right lateral and the location of the visceral organs. This does not include a congenital defect situs inversus, which leads to the arrangement of all organs in the abdomen and chest so the positions are the opposite of a normal placement. Situs inversus is a mirror image of situs solitus, which is a normal asymmetrical distribution of abdominal visceral organs. Situs ambiguus can also be subdivided into left isomerism and right isomerism based on defects observed in the spleen, lungs and atria of the heart. Individuals with situs inversus or situs solitus do not experience fatal dysfunction of their organ systems, as the general anatomy and morphology of the abdominal systems of organ-vessels persists. Due to the abnormal location of the organs in the situs ambiguus, orientation on the left-right axis of the body is impaired in the early stages of fetal development, leading to serious abnormalities in cardiac development and function in 50-80% of cases. They also experience complications with systemic and pulmonary blood vessels, significant morbidity and sometimes death. All patients with ambiguus situs lack side and symmetry of organs in the abdominal and thoracic cavity and are clinically considered to be a form of heterosacxia syndrome. Heertotaxis syndrome with isomerism of the atrium occurs in 1 out of every 10,000 live births and is associated with approximately 3% of cases of congenital heart defects. Additional assessment of the incidence and prevalence of isomerism is difficult because of the inability to diagnose and underestimate the disease by doctors. In addition, right isomerism is much easier to recognize than left isomerism, contributing to the inability to diagnose. Situs ambiguus is a growing field of research with findings dating back to 1973. Signs and symptoms there are various clinical manifestations of situs ambiguus. Acute symptoms can be caused by both heart and non-heart defects. Cyanosis or the coloration of blue skin, primarily affecting the lips and nails, may indicate a systemic or circulatory issue. Poor nutrition, inability to thrive, and rapid shallow breathing can also be observed due to poor circulation. When examined, arrhythmia and noise in the heart can cause further suspicion of cardiac abnormality. Non-heart symptoms include liver and gastrointestinal disorders. Biliary atresia, or inflammation and destruction of the bile ducts, can lead to jaundice. Vomiting and swelling of the abdominal cavity are features that suggest incorrect positioning of the intestines. Poor bowel positioning also makes it more prone to blockage, which can lead to numerous chronic health problems. Assplenia and polysplification are also possible features of heterotaxis syndrome. Due to abnormal heart development, patients with situs ambiguus usually develop the right atrium of isomerism, consisting of two bilateral paired right atrium, or left atrial isomerism consisting of two bilateral paired left atriums. Clinical features and may vary depending on the destination of the left compared to the atrium of isomerism. In any case, the top of the heart will be poorly positioned, which should warn the doctor about the likelihood of atrial isomeria. It is estimated that 5-10% of isometric patients have mesocardia, in which the heart is located in the center of the chest, 25-50% have a dextrocardia, in which the top of the heart is directed to the right side of the chest, and 50 - 70% have a leftcardia, in which the top of the heart is directed towards the left side of the chest. The right atrium of the right atrium isomerism, also called right atrial isomerism, is a heart defect in which the heart has a bilateral right atrium and atrium in the muscular wall, as opposed to normal right atrium and left atrium. In the right atrium, isomerism of the pulmonary oxygen tract of blood is damaged due to right-left blood bypass. In addition, there is no atrial septum, which distinguishes 2 atriums. These disorders, in addition to congestion in the pulmonary tract, allow deoxygenic blood to mix with blood oxygen, contributing to cyanosis and possible respiratory distress. Poor systemic circulation also leads to incorrect positioning of the aorta. The left atrium appendage of isomerism Left atrial isomerism, also called left atrial isomerism, is a heart malformation in which the heart has two bilateral left atrium and atrial appendages in the muscular wall. Left atrial isomerism can have various clinical manifestations, including the late onset of symptoms. Heart failure is often a concern because the lower vein kava is disturbed due to inappropriate left ventricular morphology to support the vein of the cava. Behavior of nodes and tissues Abnormal development of the heart leads to the disruption of doubling conductive nodes as well as faulty electrical fibers throughout the ventricles. Individuals with right atrium isomerism develop 2 synoactal nodes, as they have 2 mirrored right atrium, while those with left atrium isomerism are unable to develop a sinus node at all. Thus, patients with left atrium isomerism are more likely to experience atrial fibrillation, or irregular rapid heartbeat, and abnormal heart rate, known as atrial flutter. The development of the atrioventricular node and his beam largely depends on the physiological loop of the ventricles. The abnormal ventricular loop promotes arrhythmia and heart block in the fetus. Bronchial tree and mild isomerism of bronchus usually do not damage and do not represent significant clinical complications. Pulmonary valve stenosis leads to problems of blood flow to the lungs. The abdominal organs, including the liver, stomach, intestinal tract and spleen, can be randomly located throughout the left-right axis of the body. The distribution of these organs is in many ways treatment, clinical clinical and further evaluation. The liver is usually symmetrical on the left-right axis in patients with situs ambiguus, which is abnormal. Most patients with left atrium isomeric have defects throughout the bile tree, which is responsible for the production of bile, even if the gallbladder is functional and morphologically normal. This bile atresia can lead to acute problems such as nourishing malabsorption, pale stool, dark urine, and abdominal swelling. If this condition continues without proper treatment, cirrhosis of the liver and liver failure become a serious problem. Biliary atresia is usually not observed in patients with right atrial isomeria. Accidental gastric positioning is often one of the first signals of situs ambiguus when examined. Malrothation of the entire gastrointestinal tract, or improper folding and bulging of the stomach and intestines, leads to intestinal obstruction. This disorder leads to vomiting, abdominal dystension, mucus and blood in the stool. Patients may also experience abdominal pain. Intestinal malrothium is more common in patients with right atrial isomerism than in patients with left atrial isomerism. (quote is necessary) Isomerick patients often experience a developmental disorder of the spleen during embryogenesis, resulting in a general absence of spleen (assplenia) or the development of many spleen (polysplenia). Aspemia is most common in patients with right atrial isomeria. Polysplification leads to 90% of patients with left atrial isomeria. Although they have many spleen, each one is usually ineffective as a result of functional assplein. Rarely, left atrial isomeric patients have one, normal, functional spleen. Patients without a functional spleen are at risk of sepsis and should be tested. The reasons although its cause are poorly understood, the ambiguus situs was linked to the family history of malformations and maternal cocaine use, suggesting both genetic and environmental factors play a role. Several genes in the pathway of TGF-beta, which controls the left-right pattern of the visceral organ throughout the body's axis, have been shown in sporadic and family-related cases of isomerism of the atrium. The impaired function of mitochondria has also recently been associated with heterotaxis. The pathophysiological molecular and cellular mechanism Of several genes have been identified in the normal development of the right-left axis. These genes have been extensively researched. Gene mutations, which lead to atrial isomerisy, are a growing area of research. Mutations in genes encoded by proteins in the TGF-beta pathway, including NODAL, NKX2-5 and qIC3, have been linked to tetralogy and left heart hypoplastic syndrome. Mutations in the IC3 gene, which encodes the first zinc finger transcription factor, are associated with a 50% risk of atrial isomerism in families. It's also an X-related disorder, testing for IC3 IC3 highly encouraged during childbirth in men. The most common and most characteristic genetic associations of hetenotaxis include: Type OMIM Gene Locus HTX1 306955 yC3 Xq26.2 HTX2 605376 CFC1 2q21.1 HTX3 606325 PA26 6q21 HTX4 613 751 ACVR2B 992 HTX5 270100 NODAL 10q22.1 HTX6 614779 CCDC11 18q21.1 Classical Pathology of the Heart Loop: tetralogical transposition of Fallo of large ventricle vessels and septum defects. Deranged asymmetry of abdominal organs: the stomach and spleen are prone to isolated reversal of the stomach, liver and adrenal gland are sometimes in the middle line. Organ malformations: assing and polyslion often lead to sepsis, the head of the pancreas cannot form a kidneys pad to develop, which can lead to cancer, kidney stones, and/or infection. Malrotation errors cause volvulus and/or faulty peritoneal attachments, which completely hinder the intestines. Vascular anomalies: Aborted lower vein kava, bilateral superior or lower vein cavae Intrahepatic interruption of the lower vein of cava with connection to the azagos or hemazig veins of the Aberrant portal veins. The bronchial tree and lung pathophysiology in the bronchial tree can be observed using X-rays. In normal development, the bronchial tree consists of two main bronchus, which are anatomically different: the hyparterial bronchu (below the pulmonary artery): supplies blood to the double-bladed left lung Eparterial bronchus (adjacent to the artery): supplies blood to the three-bladed right lung In situs ambiguus, there is duplication or hyparterial. These features are not associated with any significant clinical complications. Mechanisms leading to duplication of bronchi are not thoroughly studied. With pulmonary valve stenosis, there is a decrease in blood flow to the lungs due to the obstruction of the heart on the pulmonary valve. This promotes cyanosis and pulmonary hypertension. It is necessary to evaluate the diagnosis for the correct diagnosis of situs-ambiguus, cardiac and non-cardiac features. Diagnostic criteria of atrial isomerism include observation of symmetry of thoracic visceral organs in echocardiogram, arrhythmia in electrocardiogram and chest x-ray to confirm the location of the heart on the left-right axis. In addition, a number of gastrointestinal tests can be carried out to monitor intestinal malrota, as well as scans of the liver and spleen for bile functions. Diagnostic Techniques for Cardiac Causes Echocardiography Electrocardiography Heart-Resonance Tomography of The Heart-Resonance Tomography of Cardiac Catheterization and Chest Angiography X-ray Diagnostic Techniques for Noncardial Causes Splenic Features Analysis Assessment of Biyal anatomy and Cholangiogram Assessment of Intestinal Malrothation Pulse Genetic testing management Each of the symptoms of situs ambiguus should be managed with appropriate treatment dependent on the system of organs involved. Intestinal malrothation is treated surgically with the Ladd procedure. This procedure extends the fold in the abdominal floor so that the intestine can be placed in a non-rotated formation. It is not possible to return the intestines to normal morphology, however, 89% of patients who undergo Ladd's surgery experience a full resolution of symptoms. After a cholangiogram, the Kasai procedure is usually performed in cases of bile atresia. In this operation, a Y-shaped shunt is used to pass bile from the liver directly into the intestines. If this is not successful, a liver transplant can be considered based on the patient's overall health. Kasai procedure is successful in about 80% of patients. After surgery, patients are advised to take fat-soluble vitamins, choleretics and anti-inflammatory drugs. Functionally assly patients have an increased lifetime risk of sepsis, as they do not have a functional spleen to fight infection. For this reason, asplenic patients are under constant surveillance for any signs of fever or infection. In the case of infection, patients are placed on controlled empirical antibiotic therapy to avoid developing antibiotic resistance. This therapy fights infection with both gram-positive and gram-negative bacteria. Right-atrial and left atrium isomerism and related pulmonary issues are addressed in a series of steps based on the severity of symptoms. Isomeric patients are first treated by inserting a shunt that will move incoming blood through the pulmonary circuit. Fontana's procedure transmits blood through one of the patient's ventricles, to the lungs and to the systemic circulation. This process is favorable in patients between the ages of 2 and 5 years. About 20-30% of patients will require a heart transplant. Left-atrium isomeric patients have less severe complications, as they tend to have 2 functional ventricles. In this case, they can undergo two-ventricular repairs to form 2 separate ventricles and functional connected valves. The prognosis for patients with situs ambiguus is very varied, given the range of clinical complications. Infants who experience severe cyanosis at birth die within hours of delivery if medical intervention is not immediate. In addition, the life expectancy of newborns with mild cardiac lesions is not affected. Ten percent of patients born with the right atrium die before the age of 5 without intervention. Improvements in therapy increased 5-year survival to 30-74% for patients with the right atrial abnormality and 65-84% for left patients with atrial ismertics based on the cause of their disease. The research there has been huge clinical studies, racial inequality, and physiological physiological heterotaxis syndrome, dating back to 1973. In November 2015, Mishra et al. published a review describing modern knowledge of cardiac and non-cardiac abnormalities associated with situs ambiguus. The author emphasizes the importance of genetic testing before deciding on a prognosis for affected patients. It also offers prenatal screening and evaluation in cases of risk of situs ambiguus. Recent studies have shown higher rates of heterotaxis syndrome among Hispanic infants of Mexican descent, as well as female infants of non-Hispanic black and white mothers. More research is needed to clarify the mechanisms of racial inequality in heterotaxis syndrome. People of Asian descent show a higher prevalence of heterotaxis syndrome in general than in the Western world. A national study on the prevention of birth defects (October 2014) attempted to link clinical presentations of situs ambiguus with demographics in an epidemiological study. This proved to be a difficult task because of the huge differences in the representation of this disorder. However, the authors hope that finding a link can help inform clinical decisions, predictive analysis, and future outcomes. 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