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Pentalogy of Cantrell

Muhammad Khawar Sana

Rebecca M. Rentea

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Authors

Muhammad Khawar Sana[1]; Rebecca M. Rentea[2].

Affiliations

[1] King Edward Medical University/Mayo Hospital Lahore
[2] Children's Mercy

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Introduction

Pentalogy of Cantrell (POC) is a collection of five congenital midline birth anomalies that present a distinctive challenge for clinicians and surgeons. Those five defects are of the heart, pericardium, diaphragm, sternum, and abdominal wall. This condition has been divided into two categories, complete or partial.[1] Complete, as the name indicates, refers to the presence of all five defects, while others may present with only partial defects. It is also referred to as thoracoabdominal ectopia cordis, a condition where the heart is covered by an omphalocele-like membrane. Ectopia cordis (EC) is often found in fetuses with POC. Infants usually have multiple cardiac malformations with ventricular septal defect, and tetralogy of Fallot being the most common. POC may also carry genetic associations with trisomy 13, 18, 21, and Turner syndrome. The initial management addresses the lack of skin overlying the heart and abdominal cavity.

Following initial management, additional surgery involve covering the midline defects, separating the abdominal and pericardial compartment, and repairing the diagram. Advanced reconstructive techniques are utilized for additional closure, including the use of flaps, skin closure only, and bioprosthetic agents.[2] The intracardiac defects are often repaired at a later date. Survival depends on the associated cardiac anomalies and degree of thoracoabdominal defect.[3]

Etiology

The exact cause of this condition is unknown.[4] The cases are mostly sporadic. It has been seen in chromosomal abnormalities like trisomy 21, trisomy 18, and Turner’s Syndrome. The role of genes on the X-chromosome has been suggested, but the evidence is scarce to support this claim.[4]

Epidemiology

The condition usually presents at birth. Every 1 in 65,000 live births is affected with this condition.[5] A Baltimore-Washington population-based study of infants with congenital heart anomalies suggested a regional prevalence of 5.5 per 1 million live births.[6] A male predominance of 1.35 to 1 has been reported.[2]

Pathophysiology

The five defects of the pentalogy as described in Cantrell's initial paper are as follows:[7]

1. Supraumbilical omphalocele
2. Lower sternal cleft
3. Defect in the central tendon of the diaphragm
4. Defect in the pericardium

5. Intracardiac anomaly

It is regarded as a developmental field defect by experts due to the involvement of multiple tissues, from cardiac to ventral abdominal wall.[8] Although the condition has not been linked with genetics yet, familial cases have been reported in the literature.[8]

Cantrell's theory involved two steps:[2]

1. Defect in spetum transversum and its adjacent mesoderm
2. Failure of migration and fusion of primordial sternum

History and Physical

History can vary dramatically from patient to patient. Patients with partial defects may have an incomplete expression of the disorder. Patients with complete expression may have serious and life-threatening complications at presentation. It is pertinent to note that patients with this condition may not necessarily have all of the symptoms discussed here.

Abdominal wall defects: May present as ectopia cordis, where the heart is partially or completely displaced out of the thoracic cavity uncovered by the abdominal wall. This is frequently associated with the pentalogy of Cantrell but isn't always present. Omphalocele, where abdominal contents are protruding through the ventral abdominal wall covered by a thin membrane, is another common presentation. Abdominal diastasis (a wide separation of abdominal muscles) and gastroschisis (a protrusion of abdominal contents through the abdominal wall without any covering) are other less frequent but important presentations.

Sternal defects: May present with a complete absence of the xiphoid to complete absence of the sternum. The sternum may have a cleft or be shorter than normal.

Diaphragmatic defects: Diaphragmatic hernia where a hole in diaphragm leads to abdominal contents protrusion into the thoracic cavity. Clinically, it can present on chest X-ray as bowel contents in the thoracic cavity. Bowel sounds can also be heard on auscultation of the chest.

Pericardial defects: Pericardial effusion is found in some cases, although relatively less severe and defects in pericardium do not present an immediate threat to life. It can present with muffled heart sound on cardiac auscultation.

Cardiac defects: Atrial septal defects, ventricle septal defects, left ventricular diverticulum, dextrocardia, pulmonary stenosis, and tetralogy of Fallot have all been associated with this disease. The clinical picture may include dyspnea, cyanosis, and cardiac murmur.

Evaluation

The pentalogy of Cantrell may be diagnosed at the first-trimester ultrasound.[5] The prenatal diagnosis helps provide the families with the opportunity to make informed decisions regarding the pregnancy. Additionally, a fetal intervention can also be planned with this information. After birth, a chest X-ray can reveal diaphragmatic hernia and dextrocardia. The postnatal evaluation also includes a computed tomography scan and a cardiac magnetic resonance imaging scan to evaluate the extent of the defect and plan for further surgical correction. An echocardiography exam is necessary to evaluate the heart chambers for defects and ejection fraction. Due to the association with aneuploidy, karyotyping is also important for the counseling of the patient's families.[2]

Treatment / Management

The treatment of the pentalogy of Cantrell is tailored towards the presence of defects and their extent. Many types of reconstruction and repairs are described that can be single or multi-staged and require an interprofessional team approach. Treatment involves neonatal resuscitation to a temporary covering of contents protruding out of the ventral

abdominal wall and, finally, surgical correction. Surgical management of cardiac, diaphragmatic, and other defects is crucial and initially involves covering the defects. Assessment for risk factors for mortality should be evaluated. The surgical aim involves correcting cardiac malformations, restoring cardiac position and anatomy, and repairing the thoracoabdominal wall and diaphragmatic defects.

Initial conservative management includes prophylactic antibiotics and daily dressing changes to allow epithelialization of the omphalocele sac.[9] Despite the intervention, morbidity and mortality may remain high in severe cases. The surgery itself carries significant risk, and clinically stable patients are managed conservatively initially. Although surgeons may attempt to repair the sternum, the diaphragm, and the pericardium during the same attempt if feasible,[10] usually, a staged surgical approach is required. Ventricular diverticulum predisposes the patient to thromboembolic events, and early repair is considered to avoid complications.[4] Ventricle and atrial septal defects can be managed at a later stage in stable patients. Pericardiocentesis is indicated when the pericardial effusion is significant or symptomatic.

Avoiding high postoperative intraabdominal and intrathoracic pressure following repair of the abdominal wall and the sternal defects is crucial.[11]

Differential Diagnosis

Amniotic band syndrome presents similar to the pentalogy of Cantrell. The involvement of limbs and face favors amniotic band syndrome. Other differentials include limb body wall complex, body stalk anomaly, isolated ectopia cordis, and isolated omphalocele.[12] A detailed evaluation of the patient is required before diagnosis. In some cases, the diagnosis may not be clear until the surgery.

Prognosis

The survival rate of patients with this condition is estimated to be around 37%.[9] The prognosis is primarily poor for patients with severe disease and complete-type pentalogy of Cantrell.[13]

Complications

Complications have a wide range, from surgery to anesthesia, and lack of timely intervention can all further complicate the patient's course. Some of the complications reported in the literature include tachyarrhythmias, bradycardia, hypotension, ventricular diverticulum rupture, and heart failure.[14][15] Additionally, postoperative high intraabdominal and intrathoracic pressures following repair may lead to cardiac decompensation and injury to intraabdominal organs.[11] Vigilance from the interprofessional team is necessary to avoid these complications. Mortality overall remains at 37%.[9]

Consultations

An interprofessional team approach is essential in the successful management of patients. This team is comprised of pediatric surgeons, neonatologists, and obstetricians. All the other concerned departments shall be consulted as well if the need arises.

Deterrence and Patient Education

Parents should be counseled regarding the overall survival and prognosis of this condition, including antenatal counseling relative to mortality and morbidity risks. Although there have been case reports regarding possible familial linkage, the evidence is scarce, and more information is needed before recommending genetic counseling. The associated aneuploidy may warrant genetic testing to assess risk in future pregnancies. The aggressive prenatal interventions must be balanced with expected post-natal prognosis and expectations of the parents.

Enhancing Healthcare Team Outcomes

The pentalogy of Cantrell constitutes a variety of congenital malformations and exhibits variable presentations. Although the cause is yet unknown, the condition can often be diagnosed prenatally. While some suggest early intervention, others believe a step-wise procedure should be followed to manage this condition. A plan should be developed in a staged approach tailored to each patient. While the physical exam may reveal the apparent defects in the ventral abdominal wall, imaging is necessary to assess the full extent of the pentalogy of Cantrell. While obstetricians are usually the first to note abnormalities in first-trimester ultrasound, pediatric and family clinicians, cardiologists, and plastic surgeons are relevant to the team approach for this patient. Immediate neonatal resuscitation should be performed. Resuscitation is followed by the temporary covering of the protruding organs (heart and omphalocele). Counseling regarding the prognosis should be provided accordingly.[2] [Level 4-5]

An interprofessional team that provides a holistic and integrated approach to preoperative and postoperative care can help achieve the best possible outcomes. Before a surgical procedure, the role of the imaging cannot be underscored enough. If the patient is to be discharged home with a wound, consultation should be made with a social worker and community nurses who can monitor the patient and make referrals as needed. A nutritionist should help devise a tailored schedule for feeding and thriving.

Questions

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