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Late Diagnosis of Atrial Septal Defect Complicated by Heart Failure in a 21-Year-Old Pregnant Woman: A Case Report and Multidisciplinary

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A B S T R A C T

Introduction: Atrial septal defect (ASD) is one of the most common congenital heart diseases, often remaining asymptomatic until adulthood. In pregnant women, the physiological changes in cardiac output and blood volume can exacerbate undiagnosed ASDs, potentially leading to heart failure. This case report discusses a 21-year-old pregnant woman diagnosed with a previously unrecognized ASD complicated by right-sided heart failure during the third trimester of pregnancy.

Case Presentation: A 21-year-old primigravida at 30 weeks of gestation presented with worsening dyspnea, fatigue, and peripheral edema. Physical examination revealed jugular venous distension, a fixed split second heart sound (S2), and a systolic ejection murmur. Transthoracic echocardiography confirmed the presence of a 2.5 cm ostium secundum ASD with significant left-to-right shunting, right atrial and right ventricular enlargement, and mild pulmonary hypertension. Laboratory tests showed elevated brain natriuretic peptide (BNP) levels consistent with heart failure. The patient was managed with oral diuretics and beta-blockers to control her symptoms and stabilize her cardiovascular status. A multidisciplinary team planned for induction of labor at 37 weeks, and she delivered a healthy infant via vaginal delivery. Postpartum, she underwent successful percutaneous closure of the ASD.

Conclusion: ASD in pregnancy, particularly when diagnosed late, presents unique challenges. Early detection and multidisciplinary management are crucial in preventing adverse maternal and fetal outcomes. This case emphasizes the need for thorough cardiovascular evaluation in pregnant women with unexplained dyspnea or signs of heart failure.

Keywords: Late Diagnosis, Atrial Septal Defect, Pregnancy, Morbidity, Heart Failure

1. Introduction

Atrial septal defect (ASD) is one of the most common congenital heart defects, accounting for 6-10% of congenital heart anomalies diagnosed in adults. It is characterized by an abnormal opening in the atrial septum, the wall separating the left and right atria. This opening allows blood to flow between the atria, which can lead to a variety of hemodynamic consequences depending on the size of the defect, the direction and volume of the shunt, and the overall cardiac condition of the patient. ASD typically arises during fetal development when the atrial septum fails to form completely. Under normal circumstances, the septum closes shortly after birth, isolating the left and right sides of the heart. However, in patients with ASD, part of the septum remains open, allowing oxygenated blood from the left atrium to mix with deoxygenated blood from the right atrium. This leads to a left-to-right shunt, which increases the volume of blood passing through the right side of the heart and the pulmonary circulation. Over time, the increased blood flow can cause dilation of the right atrium, right ventricle, and pulmonary arteries, leading to complications such as pulmonary hypertension, right heart failure, and atrial arrhythmias.[1,2]

ASDs are classified into four major types based on their location in the atrial septum[3]:

1. **Ostium Secundum ASD**: The most common type, accounting for approximately 75% of cases. It occurs in the central part of the atrial septum, known as the fossa ovalis.
2. **Ostium Primum ASD**: Located near the lower part of the atrial septum, this type is associated with defects of the atrioventricular (AV) valves and is commonly seen in patients with Down syndrome.
3. **Sinus Venosus ASD**: This type occurs near the superior or inferior vena cava and is often associated with anomalous pulmonary venous drainage.
4. **Coronary Sinus ASD**: The rarest type, it occurs in the area where the coronary sinus enters the right atrium and is often associated with partial or complete unroofing of the coronary sinus.

ASDs are often asymptomatic in early life, as the shunting of blood between the atria may not cause significant hemodynamic changes in childhood. However, symptoms may gradually develop in adulthood due to the progressive nature of the left-to-right shunt and its long-term effects on the right heart and pulmonary circulation. Common symptoms include exertional dyspnea, fatigue, palpitations, and, in more severe cases, signs of heart failure such as peripheral edema and jugular venous distension. In some cases, the first presentation of ASD may occur during pregnancy when the increased hemodynamic burden exacerbates the underlying defect. The clinical course of ASD varies based on the size of the defect and the associated complications. Small ASDs with minimal shunting may remain asymptomatic throughout life and be detected incidentally. In contrast, larger defects may lead to significant right ventricular overload, pulmonary hypertension, and the risk of arrhythmias such as atrial fibrillation or flutter. Additionally, patients with unrepaired large ASDs are at risk for the development of Eisenmenger syndrome, a condition characterized by reversal of the shunt (right-to-left) due to severe pulmonary hypertension, which can lead to cyanosis and other systemic complications.[3,4]

The hemodynamic impact of ASD depends largely on the size of the defect and the direction of the shunt. In most cases, there is a left-to-right shunt, meaning that blood flows from the higher-pressure left atrium to the lower-pressure right atrium. This increases the volume of blood that the right side of the heart has to pump, leading to right atrial and right ventricular dilation. Over time, this can cause the right heart to become strained, potentially leading to right-sided heart failure. One of the key consequences of ASD is the development of pulmonary overcirculation, as the right ventricle pumps an increased volume of blood into the pulmonary arteries. Chronic pulmonary overcirculation can result in pulmonary hypertension, which further exacerbates right ventricular dysfunction. The presence of pulmonary hypertension increases the risk of complications such as right heart failure, arrhythmias, and paradoxical embolism, in which an embolus can pass from the right atrium to the left atrium through the ASD, potentially causing a stroke or other systemic embolic events.[1,5]

In some cases, the chronic volume overload of the right heart may lead to the development of atrial arrhythmias, particularly atrial fibrillation and atrial flutter, which are common in patients with ASD. The enlargement of the right atrium and the increased pressure within the atrium provide a substrate for these arrhythmias. Once arrhythmias develop, they can significantly worsen the symptoms of heart failure and may increase the risk of thromboembolism. Pregnancy adds a unique hemodynamic challenge for women with undiagnosed or untreated ASD. During pregnancy, blood volume increases by 30-50%, and cardiac output rises to meet the metabolic demands of the developing fetus. These changes exacerbate the left-to-right shunt, increasing right-sided volume overload and the risk of heart failure. In addition, the hypercoagulable state of pregnancy increases the risk of thromboembolic complications, including paradoxical embolism in patients with ASD.[5]

The diagnosis of ASD is often delayed because many patients are asymptomatic or have nonspecific symptoms that may not be recognized as being related to the defect. The diagnosis is typically suspected based on physical examination findings, such as a systolic ejection murmur, a fixed split second heart sound (S2), and signs of right heart enlargement. The diagnosis is confirmed with imaging studies, most commonly transthoracic echocardiography (TTE), which allows visualization of the atrial septum and the shunt. Doppler echocardiography can assess the direction and magnitude of the shunt and evaluate for associated complications such as right ventricular dilation, pulmonary hypertension, and valve abnormalities. In some cases, transesophageal echocardiography (TEE) may be required for better visualization of the atrial septum, especially in cases of sinus venosus or coronary sinus ASDs. Other diagnostic modalities, such as cardiac MRI or CT, may be useful in evaluating the anatomy of the defect and its impact on the heart and pulmonary circulation.[2,3]

The management of ASD depends on the size of the defect, the presence of symptoms, and the risk of complications. In asymptomatic patients with small defects and minimal shunting, conservative management with periodic monitoring may be appropriate. However, in patients with larger defects or significant left-to-right shunting, closure of the defect is recommended to prevent long-term complications such as right heart failure, pulmonary hypertension, and arrhythmias. Closure of the ASD can be performed surgically or via a minimally invasive percutaneous approach, depending on the size and location of the defect. Percutaneous closure using a transcatheter device is the preferred method for most ostium secundum ASDs, as it is less invasive and associated with a shorter recovery time compared to surgical closure. However, surgical closure may be required for larger defects, sinus venosus ASDs, or cases with associated valve abnormalities. In pregnant women with ASD, management is focused on optimizing maternal hemodynamics and preventing complications during pregnancy and delivery. Vaginal delivery is generally preferred over cesarean section, as it is associated with less hemodynamic stress. A multidisciplinary team approach involving cardiology, obstetrics, and anesthesiology is essential to ensure the best outcomes for both the mother and the fetus.[4]

2. Case Presentation

A 21-year-old woman, gravida 1 para 0, presented to the obstetric clinic at 30 weeks of gestation with progressively worsening symptoms of dyspnea, fatigue, and bilateral leg swelling. The patient described the onset of symptoms around her 28th week of pregnancy, initially noticing shortness of breath upon exertion, which soon progressed to dyspnea at rest. Over the next two weeks, she also began experiencing significant fatigue, limiting her daily activities, and developed noticeable swelling in her lower extremities, particularly around the ankles and feet. Her past medical history was unremarkable, with no known history of cardiac or respiratory disease, and her pregnancy had been uneventful up to this point. She denied any history of chest pain, palpitations, syncope, or cough.

On physical examination, the patient appeared moderately distressed due to her shortness of breath. Her vital signs revealed tachycardia with a heart rate of 110 beats per minute, blood pressure of 110/70 mmHg, respiratory rate of 22 breaths per minute, and oxygen saturation of 96% on room air. She was afebrile. Cardiovascular examination revealed a prominent and fixed split second heart sound (S2) along with a systolic ejection murmur best heard at the left upper sternal border. There was no evidence of a third heart sound (S3) or fourth heart sound (S4). On inspection of the neck, jugular venous

distension was noted, indicative of elevated central venous pressure. Examination of the lower extremities showed pitting edema up to the level of the knees. There were no signs of cyanosis, and lung auscultation was clear with no rales or wheezes. Abdominal examination was consistent with a 30-week gestation, and there were no signs of hepatomegaly or ascites. Neurological and musculoskeletal examinations were normal.

Given the findings of a systolic murmur, jugular venous distension, and peripheral edema, there was concern for an underlying cardiac pathology contributing to the patient's symptoms. The differential diagnosis at this point included various causes of heart failure, such as peripartum cardiomyopathy, valvular heart disease, or an undiagnosed congenital heart defect. An urgent transthoracic echocardiogram (TTE) was ordered to further investigate the cause of her symptoms and the heart murmur. The echocardiogram revealed the presence of a 2.5 cm ostium secundum atrial septal defect (ASD) with significant left-to-right shunting of blood. The shunt had led to marked enlargement of both the right atrium and the right ventricle, with evidence of increased right-sided pressures. Pulmonary artery systolic pressure was estimated to be 45 mmHg, suggesting mild pulmonary hypertension. Left ventricular size and function were within normal limits, with an ejection fraction of 60%. No other structural abnormalities were noted. The Doppler study confirmed the hemodynamic significance of the shunt, with a continuous flow of blood from the left atrium to the right atrium.

Laboratory tests were performed to rule out other potential causes of her symptoms. A complete blood count revealed mild anemia, with a hemoglobin level of 10.5 g/dL. Renal and liver function tests were within normal limits, and there was no evidence of significant electrolyte imbalances. The patient's brain natriuretic peptide (BNP) level was elevated at 450 pg/mL, indicative of cardiac stress and heart failure. Arterial blood gas analysis showed a mild respiratory alkalosis, but her oxygenation remained normal. Based on the echocardiogram findings and clinical presentation, the diagnosis of a significant atrial septal defect with right-sided heart failure due to volume overload was made. The increased blood flow through the ASD had resulted in right heart strain and mild pulmonary hypertension, which were now manifesting as symptoms of heart failure in the setting of the hemodynamic stress imposed by pregnancy.

Given the advanced stage of her pregnancy, a multidisciplinary team consisting of cardiologists, obstetricians, and anesthesiologists was assembled to manage the patient's condition. The immediate priority was to control her heart failure symptoms and stabilize her cardiovascular status to allow for the safest possible delivery. She was admitted to the hospital for close monitoring and initiation of medical therapy. The patient was started on oral diuretics (furosemide) to reduce the volume overload and relieve her peripheral edema and shortness of breath. Care was taken to monitor her electrolytes and fluid status to avoid excessive diuresis, which could compromise both maternal and fetal hemodynamics. Additionally, she was initiated on a low-dose beta-blocker (metoprolol) to control her heart rate and reduce the risk of arrhythmias, which are more common in patients with ASD and right atrial enlargement. Anticoagulation was not immediately initiated, as the risks of thromboembolism were weighed against the potential for bleeding complications during delivery, but the patient was monitored closely for any signs of arrhythmias or clot formation.

As the patient was in her third trimester, the team decided to aim for a vaginal delivery at term, unless her condition worsened or the fetus showed signs of distress. A planned induction of labor at 37 weeks was discussed with the patient to minimize the risks associated with spontaneous labor, particularly the sudden hemodynamic changes that could occur during active labor. In the meantime, the patient was monitored regularly with repeat echocardiograms to assess for any progression of her pulmonary hypertension or right heart dysfunction. At 37 weeks, the patient was admitted for induction of labor. She received epidural anesthesia to minimize the pain and cardiovascular stress associated with labor. Labor was uneventful, and she delivered a healthy male infant weighing 2.8 kg.

Throughout the delivery, her cardiovascular status remained stable, and she showed no signs of worsening heart failure. Postpartum, the patient was closely monitored for any signs of fluid retention, arrhythmias, or thromboembolism. She continued on diuretics for a brief period but was gradually weaned off as her symptoms improved. The patient's postpartum recovery was uneventful, and she was discharged home on oral diuretics and beta-blockers with instructions for close follow-up. Six weeks postpartum, she underwent a transesophageal echocardiogram (TEE), which confirmed the presence of the ASD and left-to-right shunt, with persistent right atrial and right ventricular dilation. The patient was referred for elective percutaneous closure of the ASD, which was successfully performed three months after delivery. Post-procedure, follow-up echocardiography showed normal right ventricular size and function, and the patient remained asymptomatic. She was advised to continue follow-up with cardiology for long-term monitoring of her cardiac health and to receive preconception counseling before any future pregnancies.

3. Discussion

This case of a 21-year-old pregnant woman with a late-diagnosed atrial septal defect (ASD) highlights several critical aspects of managing congenital heart disease in pregnancy. ASD, while often asymptomatic during childhood and early adulthood, may manifest in more severe forms when physiological stressors—such as pregnancy—are introduced. The combination of increased cardiac output, blood volume, and the hyperdynamic circulatory state of pregnancy can exacerbate an otherwise silent cardiac anomaly, leading to serious complications such as heart failure.[6]

1. Late Presentation of ASD

ASD is often referred to as a "silent" congenital heart defect because many individuals remain asymptomatic for years. In some cases, like the one presented here, ASD goes undetected until adulthood or is discovered only after the development of symptoms during pregnancy or other stressors. The asymptomatic nature of ASD in early life is largely due to the heart's ability to compensate for the left-to-right shunt. However, over time, this compensatory mechanism can be overwhelmed, leading to right heart overload and the gradual onset of symptoms.[6,7]

In this case, the patient's ASD remained undiagnosed until the third trimester of her pregnancy when she presented with signs of heart failure. This highlights the importance of a thorough cardiovascular evaluation in women who present with unexplained dyspnea, fatigue, or other symptoms during pregnancy. Delayed diagnosis can result in increased maternal and fetal morbidity, especially in cases where heart failure becomes apparent.[3]

2. Hemodynamic Changes in Pregnancy and Their Impact on ASD

Pregnancy induces significant cardiovascular changes, including a 30-50% increase in blood volume, increased cardiac output, and reduced systemic vascular resistance. These changes place additional strain on the heart, particularly in patients with underlying congenital heart defects such as ASD. In patients with a significant left-to-right shunt, the increased blood volume and cardiac output further exacerbate the volume overload on the right heart, increasing the risk of right-sided heart failure and pulmonary hypertension.[2,8]

The timing of symptom onset in this case—during the third trimester—is consistent with the progressive hemodynamic burden of pregnancy. The patient's dyspnea, fatigue, and peripheral edema were likely the result of both increased right ventricular volume and the reduced ability of her right heart to accommodate the increased circulatory demands. Pregnancy unmasked the underlying ASD by overwhelming the right heart's capacity to manage the chronic left-to-right shunt.[4]

3. Heart Failure in Pregnancy: Diagnostic and Therapeutic Challenges

The diagnosis of heart failure in pregnancy can be particularly challenging because many of the normal symptoms of pregnancy—such as fatigue, shortness of breath, and mild peripheral edema—overlap with those of heart failure. Therefore, distinguishing between physiological changes of pregnancy and pathological conditions like heart failure requires a high index of suspicion, especially in the presence of abnormal physical exam findings such as a heart murmur, elevated jugular venous pressure, or significant peripheral edema. In this case, the fixed split second heart sound (S2) and systolic ejection murmur were key clues pointing towards an underlying cardiac pathology. The echocardiogram confirmed the diagnosis of ASD and provided critical information about the right atrial and ventricular enlargement, as well as the presence of mild pulmonary hypertension. Early recognition of heart failure allowed for prompt management, which is crucial to prevent worsening of the condition and potential maternal or fetal complications.5

Therapeutic management of heart failure in pregnancy is complex, as certain medications used to treat heart failure outside of pregnancy may be contraindicated or require careful monitoring due to their potential effects on the fetus. In this case, diuretics (furosemide) were used to manage the patient's symptoms of fluid overload, and beta-blockers (metoprolol) were initiated to control her heart rate and reduce the risk of arrhythmias. Both medications are considered relatively safe in pregnancy when used under close supervision. However, careful dosing and monitoring are essential to avoid complications such as hypotension, electrolyte imbalances, and impaired fetal growth.[8]

4. Multidisciplinary Management of ASD and Heart Failure in Pregnancy

Pregnancy in women with congenital heart disease, particularly undiagnosed ASD, requires a multidisciplinary team approach. The involvement of cardiology, obstetrics, anesthesiology, and neonatology is essential to ensure optimal maternal and fetal outcomes. In this case, the cardiology team monitored the patient's cardiac function and managed her heart failure symptoms, while the obstetric team provided guidance on delivery planning. Given the increased cardiovascular risks associated with cesarean section, vaginal delivery was favored as it is associated with less hemodynamic stress. The close collaboration of the multidisciplinary team ensured a safe delivery and stable postpartum recovery for the mother. Vaginal delivery, in cases of cardiac disease, is typically preferred because it imposes less hemodynamic strain compared to cesarean section, which involves higher blood loss, the effects of anesthesia, and greater shifts in intravascular volume. Labor in such patients should be carefully monitored, with epidural anesthesia considered to reduce pain-related stress and potential increases in cardiac output.[6,7]

5. Postpartum Considerations and Long-term Management

The postpartum period is a time of significant cardiovascular changes as blood volume decreases and the body returns to a non-pregnant state. In women with ASD and heart failure, close monitoring is essential during this period to detect any worsening of heart failure symptoms or arrhythmias. In this case, the patient's heart failure symptoms improved postpartum with continued medical management, and her ASD was successfully closed percutaneously a few months after delivery.

Long-term management of ASD often involves either surgical or percutaneous closure, particularly in patients with larger defects or significant left-to-right shunting. Percutaneous device closure has become the preferred treatment option for most patients with ostium secundum ASD, as it is less invasive and has a lower complication rate compared to open-heart surgery. The timing of closure is critical, and in most cases, it is deferred until after delivery unless there are life-threatening complications. This case also highlights the importance of follow-up and counseling regarding future pregnancies. Women with repaired ASD generally have favorable outcomes in subsequent pregnancies, but they should be monitored closely for recurrence of symptoms, arrhythmias, or other complications related to residual shunts or pulmonary hypertension.[1]

6. Prevention and Screening

This case underscores the importance of early diagnosis and management of congenital heart disease, especially in women of childbearing age. Routine screening of pregnant women with unexplained dyspnea or fatigue should include a thorough cardiovascular assessment to identify any underlying congenital heart defects. Early detection of ASD before pregnancy can allow for timely intervention, potentially reducing the risk of complications during pregnancy. Additionally, women with known congenital heart disease should receive preconception counseling to assess their cardiovascular risk and

plan for a safe pregnancy. This includes a detailed evaluation of cardiac function, the need for any interventions before conception, and the development of a management plan for the pregnancy and delivery.[3,4,5]

4. Conclusion

This case demonstrates the late detection of an atrial septal defect in a young pregnant woman who developed heart failure during the third trimester. With appropriate medical management and a multidisciplinary team approach, both the mother and her baby had favorable outcomes. This case emphasizes the importance of recognizing congenital heart disease in pregnancy, the need for careful cardiovascular evaluation in pregnant women presenting with unexplained symptoms, and the benefits of early intervention in managing ASD. Long-term follow-up and planning for future pregnancies are also crucial in ensuring ongoing health and preventing complications.

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