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Sudden Infant Death Due to Misdiagnosis of Hypoplastic Left Heart Syndrome, a Case Report

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Abstract

Heart abnormalities are the most prevalent congenital defects, with Hypoplastic Left Heart Syndrome (HLHS) being a critical type marked by the left heart's underdevelopment. Immediate diagnosis and intervention are crucial to avert dire consequences like sudden infant death. The report described the sudden death of a 4.5-month-old female infant, who showed symptoms like cyanosis, dyspnea, and distress while crying and breastfeeding. On the day of the incident, the infant experienced a seizure after crying for 10 minutes. She passed away in the Emergency Department due to resistant bradycardia. The case highlights the devastating effects of misdiagnosis and the difficulties in detecting complex HLHS, underlining the urgency for increased vigilance, and the use of diagnostic techniques.

Keywords: Congenital abnormalities, ECG, Differential Diagnosis, Pediatric Case Report

Introduction:

Sudden Infant Death Syndrome (SIDS) is a tragic event and unexplained phenomenon that leaves families devastated and medical professionals perplexed (1, 2). It is characterized by the sudden and unexpected death of an infant less than one-year-old (3). While the exact cause of SIDS remains unknown, certain have been associated with an increased risk (4-7). The most common congenital abnormality is congenital heart disease (CHD), affecting around one percent of newborns (8). Hypoplastic Left Heart Syndrome (HLHS) is a severe congenital heart defect that includes abnormalities in the left-sided heart resulting from left inflow or outflow obstruction (9, 10). The most common symptom of HLHS is dyspnea and cyanosis, also right axis deviation and right ventricular hypertrophy are reported in the electrocardiograms (11-13). Misdiagnosis or late diagnosis of HLHS can lead to catastrophic outcomes, including sudden infant death (14). This case report aims to shed light on a heartbreaking incident of sudden infant death due to the misdiagnosis of HLHS. Through this report, we hope to emphasize the importance of early and accurate diagnosis of HLHS and explore strategies for improving diagnostic accuracy and patient outcomes.

Case Report

Herein, we described the sudden death of a 4.5-month-female child. Her mother and her father are 34 and 32 years old, respectively. Her parents have 3 other children (one boy and two girls in order 7, 9 & 11 years old). they report the normal condition of the patient although sometimes blue lips, shortness of breath, and anxiety are observed while crying and breastfeeding. On the day of the accident, the patient develops a seizure, after 10 minutes of crying. She was brought to the infirmary by her parents and then transported to the Emergency Department at 07:11 in the morning. Physical examination reported gasping, cyanosis, loss of consciousness, and low oxygen saturation (73%). Normal CXR was reported and the patient was transferred to the PICU under a ventilator. The oxygen saturation level reaches 95%, but the level of consciousness is still low. Sampling performed for coronavirus. The patient was ventilated and intubated. Bradycardia was reported at 07:20 AM. PPV and Chest Compression after epinephrine injection 1 in 10,000 (1mg in 10ml) in five doses of 0.5 ml performed. Bradycardia does persist. Despite 50 minutes of incomplete resuscitation, the infant's condition deteriorated rapidly, resulting in sudden death.

In the autopsy, the body was 59 cm in high (head to heel) and 26 cm (pelvis to heel), weighted 5902 g. Head circumference is 40 cm. Abdominal circumference is 37 cm. Chest circumference 39 cm. there were no findings of putrefaction. There was cyanosis in the extreme, blue fingers (Figure1). There were no other definite medical findings in the body. heart size is 6*4*4 cm that the ventricle is not developed and is very small (Figure2). A small aortic artery with an underdeveloped mitral valve was also seen. The hypoplastic left ventricular syndrome was reported in the pathology examination of the heart. In the brain, focal hemorrhage, dilatation of ventricles, and hyperemia of the arachnoid vessels were reported. No Poison has been reported in post-mortem toxicology analysis. The cause of death was determined to be congenital heart disease.

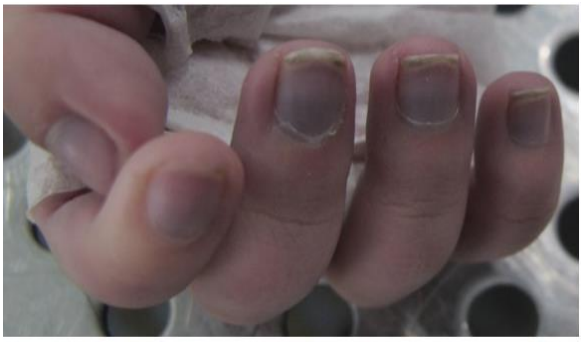

Figure 1: Infant Death due to HLHS. The hands were cyanotic.

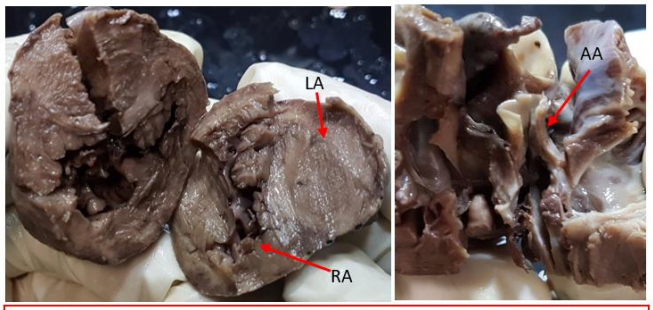

Figure 2: Hypoplasia of the left heart. AA, ascending aorta. LA, left atrium. RA, right atrium.

Discussion

The post-mortem examination revealed the presence of HLHS, which had been missed during the initial diagnosis. Cardiovascular malformations contribute the most commonly to infant mortality due to birth defects (15). HLHS is a congenital defect (about 2 in every 10,000 live births) in which the left side of the heart is underdeveloped (16). The highest mortality rate among 30 children who die each year in California from misdiagnosis or late diagnosis is related to HLHS and anomaly in aorta disease(17). Ng and Hokansan report that misdiagnosis of CHD occurred in 1: 24684 neonates (18). Misdiagnosis of HLHS was decreased in California (1990 to 2004) (15). According to studies, there are no accurate statistics on the prevalence of HLHS and its misdiagnosis in Iran and Asia.

The term "hypoplastic" describes the condition of an organ that stunted in its maturity (10). HLHS is a complex cardiac condition that often presents with non-specific symptoms such as feeding difficulties and lethargy, like many common pediatric illnesses. One of the cases that lead to untimely treatment of this disease is misdiagnosis. If untreated with cardiac

transplant, staged surgical palliation or other techniques, there are nearly no survivors more than one year of age (14, 19, 20). This underscores the importance of thorough differential diagnosis when infants present with such symptoms, especially considering the high mortality rate associated with untreated HLHS.

Hypoplastic Left Heart Syndrome (HLHS) can be challenging to diagnose accurately (14). It is often misdiagnosed due to its complex nature and variability (21). HLHS encompasses a spectrum of cardiac malformations ranging from severe underdevelopment of the left heart components to milder forms with patent valves (14, 22, 23). The prompt identification of HLHS during pregnancy is crucial for successful management through surgical interventions (21). Echocardiography allows examination of underlying anatomy for accurate diagnosis and HLHS misdiagnosis is not mentioned (24). Prenatal diagnosis improves the prognosis of hypoplastic left heart syndrome (25). HLHS diagnosis prenatally is often made by fetal echocardiogram before delivery (26). HLHS can be diagnosed by fetal echocardiography between 18-22 weeks (27). First-trimester diagnosis of hypoplastic left heart syndrome was reported (28). Advances in fetal echocardiography have improved pre-birth diagnosis rates, allowing for early intervention and improved outcomes (29). HLHS may be misdiagnosed due to left isomerism confusion due to an Azygous venous connection (30). Differential diagnosis includes critical aortic stenosis and coarctation of the aorta (27).

After delivery, HLHS has symptoms such as severe respiratory failure, severe preoperative obstruction to pulmonary venous return, restrictive atrial septum, coronary fistulae, severe tricuspid regurgitation, smaller ascending aorta diameter, and poor ventricular function (21, 31). Postnatal diagnosis is crucial due to high mortality rates after birth (22). It is often made by fetal echocardiogram before delivery (26). Also, Echocardiography is the main diagnostic tool for postpartum diagnosis (24). Echocardiography plays a vital role in diagnosing HLHS both prenatally and postnatally, enabling detailed assessment of cardiac anatomy and guiding therapeutic strategies.

Because the patient with untreated left ventricular hypoplastic syndrome dies rapidly if clinicians fail to recognize and/or misdiagnose the patient. So, focusing on the symptoms of the disease due to its high prevalence compared to other anomalies can prevent unexpected death. in this study, we report a misdiagnosis of the left ventricular hypoplastic syndrome in an infant that was caused fatally.

Conclusion:

This case highlights the tragic consequences of misdiagnosis and provides a stark reminder of the challenges faced in diagnosing complex congenital heart diseases like HLHS. It underscores the need for increased awareness, timely intervention, and the importance of advanced diagnostic tools in preventing such unfortunate incidents.

Suggestions:

Potential future research directions could encompass: enhancing prenatal and postnatal diagnostic techniques, investigating the prevalence and misdiagnosis across various regions, and examining strategies for postnatal management.

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