Brief communications

Hypoplastic left heart syndrome and complete congenital heart block in a newborn, a rare association

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We present a clinical case of the association of CCHB and HLHS in a newborn. The etiological relation between these two pathologies is unclear. According to the literature data, 70–90% of isolated CCHB are caused by maternal anti-Ro and anti-La antibodies, which cross the placenta and lead to fibrosis of the AV node or occur due to genetic defects, such as mutations in the SCN5A gene. Other theories suggest that compromised coronary blood flow in late fetal life could be a cause of CCHB, as the AV-node artery is the first and longest inferior septal branch of the right (90%) or left (10%) coronary artery, arising from U- or V-shaped segment of the corresponding artery at the level of the crux cordis. In our case, the level of maternal autoantibody titers was unknown. It is possible that the heart block could be linked to the structural heart defect – HLHS, which could be the cause of hypoperfusion of AV node in fetal life. Only two similar cases of such combination are described in the literature.

Keywords: Pediatric cardiology, congenital heart defect, hypoplastic left heart syndrome, complete congenital heart block, neonates.
Introduction

We present a clinical case of association of congenital complete heart block (CCHB) and hypoplastic left heart syndrome (HLHS) in a newborn. Such association of two different congenital pathologies is extremely rare, and literature data of the possible causes of the combination of these pathologies is very limited.

Clinical case

A newborn boy was admitted to the neonatal Intensive Care Unit directly from the maternity hospital with a heart rate of 56-60 beats per minute.

The child was born from the XI th pregnancy, on the 37th week of gestation, by physiological delivery with a birthweight of 3500 gr, APGAR scores 8\8 points. The course of pregnancy was without any specific events; prenatal ultrasounds did not reveal any pathology.

On the 20th minute of life, the child’s general state worsened, heart rate was low - 58-60 bpm, and central cyanosis became evident. The child was urgently transported to the Regional Children’s Hospital.

On examination: the child appeared critically ill, poorly perfused, and presented with dyspnea; the skin appeared cyanotic, irregular heart rate and hyperdynamic precordium harsh breathing, nasal flaring, and intercostal retractions were visible. The patient’s vital signs were the following: respiratory rate (RR) – 42/min, heart rate (HR) – 54-56 b/min, and saturation (SpO2) – 86-87%. The child was put on high-flow nasal cannula oxygen.

Bronchial breathing, arrhythmic heart tones and loud single S2 were noted during auscultation. Peripheral pulses were poor; extremities appeared vasoconstricted. Prostaglandin E1 infusion was initiated.

Results

Paraclinical examination. Chest roentgenogram showed pulmonary venous congestion and edema.

An electrocardiogram (ECG) showed right axis deviation, right ventricle hypertrophy, complete atrioventricular block III stage, HR of the ventricles 56-60 bpm and the atria – 125 bpm (Figure 1).

![Figure 1. 12 lead ECG – right ventricle hypertrophy, complete atrioventricular block III stage, HR of the ventricles 56-60 bpm and the atria – 125 bpm](image)

2D Echocardiography revealed hypoplasia of the left ventricle, mitral and aortic valve atresia, and hypoplasia of ascending aorta (Figure 2).
Figure 2. 2D Echocardiography. Four-chamber view. Hypoplastic left ventricle, mitral valve atresia

Moderate tricuspid valve insufficiency (2+) with a pressure gradient of 72 mmHg, and right ventricle pressure of 80 mmHg, which corresponds with III-rd stage pulmonary hypertension, were noted.

From the suprasternal position, the hypoplastic aortic arch and ascending aorta that was filled by retrograde flow from patent ductus arteriosus – 7 mm in size were visualized. From the parasternal position, short axis view – a dilated to 1,7 cm pulmonary artery was visualized (Figure 3).

Figure 3. 2D Echocardiography. Parasternal position, short axis view. Enlarged Pulmonary Artery 1,7 cm. Patent ductus arteriosus 7 mm

The diagnosis of Hypoplastic left ventricle, mitral and aortic valve atresia, Pulmonary Hypertension III-rd stage, Patent Ductus Arteriosus were made.
Surgical correction of congenital heart defect was denied due to the presence of a complete AV block.

The child was intubated due to progressive respiratory and heart failure caused by hypoxemia and acidosis, which eventually led to his death on the third day of life; a pathological section was denied by the parents due to religious beliefs.

Discussion

Hypoplastic left heart syndrome (HLHS) is a congenital heart defect that is characterized by the abnormal development of the left-sided cardiac structures that leads to obstruction of the blood flow from the left ventricular outflow tract. HLHS usually presents with an underdeveloped or rudimentary left ventricle, mitral and aortic valve and aortic arch atresia, as was in our case [1].

The closure of the ductus arteriosus leads to hypoperfusion and hypoxemia, acidosis, shock and eventually, death of the child.

In the event of mitral and aortic atresia, a heart murmur is usually absent, and cyanosis is mild or not evident till the moment PDA begins to close, which complicates timely diagnosis. In our case, clinical symptoms were presented and exacerbated by the presence of complete heart block, which prompted for further cardiac examination.

As all congenital heart defects, HLHS is prenatally diagnosed by 2D Echocardiography between 18-22 weeks of gestation, however in this case it was missed. The etiology of HLHS is defined as multifactorial.

HLHS is usually associated with other congenital heart defects as a ventricular septal defect (VSD) in 10% of the cases and with coarctaion of the aorta (CoA) in 75% of the cases. Association with CCHB is not mentioned in the literature, and only two similar clinical cases have been published.

Narayan HK. Et al. published a clinical case, “Hypoplastic left heart syndrome with a restrictive atrial septum and advanced heart block documented with a novel fetal electrocardiographic monitor,” in 2011 [2]. The authors describe a 35-year-old woman with negative anti-Ro and anti-La antibodies, whose fetus was diagnosed with HLHS on the 27th week of gestation and 2-nd stage CHB with 2:1 atrioventricular conduction with an atrial rate of 140 bpm and ventricular rate of 70 bpm. At 34 weeks of gestation, advanced II-nd degree heart block with a ventricular rate of 48-66 bpm and intermittent periods of complete AV block were diagnosed by fetal Echo-cardiography. The final Echo on the 35th week of gestation demonstrated an irregular ventricular rate of 51 bpm with Mobitz II second-degree AV block and inconsistent 3:1 AV conduction. The baby was born on the 37th week of gestation, was active and alert, cyanotic with no signs of acute distress with irregular heart rate. The child was started on isoproterenol, underwent radiofrequency perforation of the atrial septum, and an electrophysiologic study was attempted, but a His deflection could not be identified. A temporary pacemaker was placed, and DDD pacing was initiated, followed by the Norwood procedure. However, due to persistent hypoxemia and hypertension, the decision was made to terminate the patient.

Authors argue that the CCHB developed by the fetus could either be linked with a structural congenital heart defect – HLHS or could be explained by a separate genetic basis of AV block, as intraoperatively surgeon noticed a thickened, unusual appearance of the atrial septum. However, an autopsy was denied by the parents, and therefore the cause of CCHB remains unknown.

In 2017 Al-Kubaisi M. et al. presented a case of Hypoplastic Left Heart Syndrome with Congenital Complete Heart Block in Pediatric Cardiology Journal [3]. Authors described a 30-year-old pregnant woman whose child was diagnosed with HLHS on the 23-rd week of gestation with normal sinus rhythm. On the 37th week of gestation, fetal ventricular rate was 60 bpm, and II-nd degree heart block was diagnosed. The child was delivered by a cesarian section on the 39th week of gestation.

At the age of 2 days of life, the child underwent stage I palliation and received an epicardial dual chamber pacemaker at the same time as stage I palliation. At the age of fourteen days of life, the child also underwent an atrial septostomy. However, due to the worsening of the child’s general condition, because of tricuspid valve regurgitation and worsening of the right ventricular function and decreased saturation, the child was listed for a heart transplant and did well afterward.

Authors speculated that in fetuses with HLHS with mitral and aortic atresia, the coronary blood flow is supplied by retrograde flow through PDA across the aortic arch and that the sinoatrial (SA) and atrioventricular (AV) nodes are
blood supplied by the right coronary artery and that the event of decreased coronary blood flow could happen during gestation and eventually lead to CCHB. Authors conclude that cardiac transplantation may be an ideal solution for patients both with HLHS and CCHB.

CCHB is a rare cardiac disorder that occurs in 1 of 22,000 live births [4]. According to Myung Park, it can be caused by maternal lupus erythematosus in most cases (60–90%) [5]. Brucato et al. also describe that maternal risk factors for the development of CCHB include maternal type 2 diabetes mellitus, exposures to such medications as anti-convulsants and retinoic acids, and viral infections [6]. Less frequently, in 25–33% of the cases, it may be associated with the following congenital heart defects: L-TGA, complete AV canal, in nearly 14–42% of cases with left atrial isomerism (heterotaxy syndrome) and single ventricle [7, 8]. It is argued that in LTGA, the atrioventricular node becomes elongated and malpositioned anteriorly and therefore, can lead to AV block [9]. Neonatal myocarditis and genetic disorders could also be identified as a cause. As a complication of CCHB in fetal life, fetal hydrops or myocarditis may develop.

According to Brucato, the most typical cause of isolated CCHB is fetal exposure to maternal autoimmune antibodies, which occurs in up to 91% of isolated CCHB [8]. Isolated IIIrd degree CCHB can mainly be a consequence of an immunological conflict caused by the penetration of maternal antibodies anti-Ro/La across the fetoplacental barrier. Antibodies provoke inflammation of both the conduction system and the fetal myocardium, which leads to endocardial fibroelastosis, which can cause significant blood circulation disorders and lead to the development of fetal hydrops. CCHB usually causes severe hemodynamic complications when the heart rate of the ventricles is less than 55 per 1 min and in the event of the presence of associated myocarditis.

Such genetic defects as channelopathies are also identified as a rare cause of CCHB, which can involve genetic variants of ion channel genes, such as mutations in the following genes: SCN5A, SCN1B, SCN10A, TRPM4, KCNK17 [9].

In the presented case, the auto-antibody titers of the mother were not checked, the baby was from the 11th pregnancy, and the previous children do not have reported congenital heart defects nor CCHB. We could hypothesize that the changed anatomy of the heart with HLHS is the reason for CCHB. However, it remains a hypothesis, as an autopsy was denied by the parents. The cause of CCHB could also be genetic, as it is identified as a rare cause of CCHB.

Isolated CHB has a good prognosis after pacemaker implantation; however, according to the literature data, only 15% of fetuses with a combination of CCHB and congenital heart defects survive till the end of the neonatal period. And in the case of the presence of fetal hydrops, prenatal losses amount to almost 100% [10].

In conclusions: The etiological relation between HLHS and CCHB remains unclear. According to the literature data, most of the cases of isolated CCHB are caused by maternal anti-Ro and anti-La antibodies, which cross the placenta and lead to the fibrosis of the AV node or by genetic defects, such as mutations in the SCN5A gene.

It is also possible that the CCHB could be linked to the structural heart disease – HLHS which could be the cause of hypoperfusion of AV node in fetal life.

Prenatal Echocardiography is the golden standard of the diagnosis of congenital heart defects, as well as congenital rhythm disturbances.

Isolated CCHB has a good prognosis after pacemaker implantation; however, only 15% of fetuses with a combination of CCHB and congenital heart defects survive till the end of the neonatal period.

**Informed Consent Statement:** Written informed consent was obtained from the patient’s parents for publication of this case report, including accompanying images.
References


