Cardiovascular and Metabolic Science www.ancam.org.mx

Chapter 2

Vol. 33 Supplement 5
October-December2022

doi: 10.35366/108043

Screening for congenital heart disease

Tamizaje de las cardiopatías congénitas

Iván Romero-Rivera,* Angélica Grullón,* Demián Herrera*

INTRODUCTION

Congenital heart defects (CHDs) are present in approximately 9 per 1,000 live newborns (NBs), representing 28% of all congenital malformations. It is estimated that there are 1.35 million live newborns with CHDs worldwide.

CHDs screening requires knowledge of the symptoms and the tests that can be performed for this purpose.

Most of them, like ventricular septum defect (VSD), do not have fetal and postnatal repercussions and may present heart failure (HF) later, depending on the size of the defect. Other heart diseases may have clinical expression after birth, such as atrial septal defect (ASD) or patent ductus arteriosus (PDA), which are essential for intrauterine life. Still, their persistence after birth can produce symptoms of HF. The *foramen ovale* is almost universal in the first hours of life but may remain patent in up to 25% of adults. Other heart diseases are challenging to detect in the fetus, for example, coarctation of the aorta (Co Ao). This condition can cause severe symptoms in the NB or remain latent until adulthood, with significant arterial hypertension in the upper limbs and complications, such as cerebral aneurysms or dilatation of the ascending aorta, generally in cases associated with the bicuspid aortic valve.

Some CHDs produce severe symptoms of HF in the NB, such as the total anomalous connection of the pulmonary veins with the right atrium, mainly when there is obstruction of the collecting duct. Others may present with cyanosis and metabolic acidosis, such as complete transposition of the great arteries (TGA) or pulmonary atresia. In these cases, keeping the ductus arteriosus (DA) patent is essential to allow survival. Tetralogy of Fallot (TOF) generally presents cyanosis later when the pulmonary obstruction becomes important and produces hypoxic crises.

One of the most feared CHDs, due to poor evolution and high mortality even with treatment, is the hypoplastic syndrome of the left ventricle (LVHS) and the ascending aorta. Since the flow of the lower limbs depends on the DA and has low saturation, greater cyanosis is seen in the inferior extremities than in the upper limbs (differential cyanosis). Other severe heart diseases, such as interruption of the aortic arch, also present this sign. These cases decompensate in the first week of life due to the spontaneous closure of the DA. This difference in saturation between the upper and lower limbs may indicate more severe heart diseases in NBs. This fact supports the principle for performing the pulse oximetry test in these babies.

PULSE OXIMETRY TEST

The test must be performed in the first 24 to 48 hours of life, with adequate equipment, for asymptomatic newborns born with gestational age > 35 weeks. The test consists of measuring oximetry with a pulse oximeter in the right hand (pre-ductal measurement) and one of the lower limbs (post-ductal measurement). The result can be positive, negative, or equivocal *(Figure 1)*.

* Universidad Federal de Alagoas; Hospital Santa Casa de Misericórdia de Maceió-Brasil.

How to cite: Romero-Rivera I, Grullón A, Herrera D. Screening for congenital heart disease. Cardiovasc Metab Sci. 2022; 33 (s5): s426-s428. https://dx.doi.org/10.35366/108043

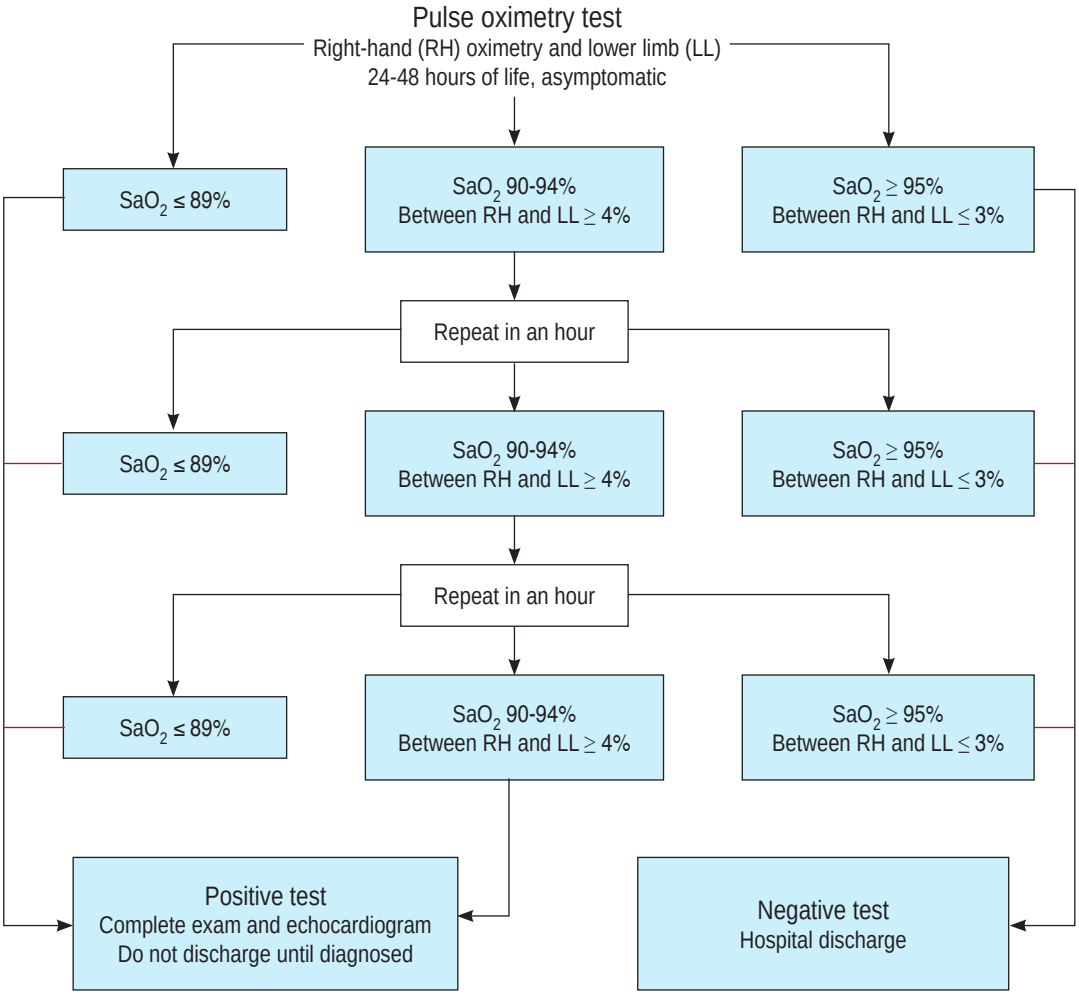

Figure 1: Pulse oximetry test, showing a positive test in the left column, negative in the right, and doubtful in the central part.
SaO_2 = oxygen saturation obtained by a pulse oximeter. RH = right hand. LL = lower limb.

SYMPTOMS AND SIGNS IN THE NEWBORN

CHDs can be acyanotic, with blood flow going from the left chambers (with greater saturation) to the right (with less saturation), as occurs in VSDs, ASDs, and PDAs. In these cases, pulmonary flow increases, and heart failure (HF) could manifest. If pulmonary stenosis (PS) is associated, and depending on the degree of obstruction, the pulmonary flow decreases, and the flow through the defect is reversed, going from the right to the left chambers, causing cyanosis and not HF, as in TOF. Single chambers (i.e., single ventricle) present a mixture of saturated and unsaturated blood with cyanosis and absence or with different degrees of HF, depending on the presence and degree of PS. Pure obstructions, such as isolated PS, are acyanotic, and left-sided obstructions can cause HF, depending on the degree of the obstructive lesion.

Hypoxemia and cyanosis are manifestations of DA-dependent heart disease, such as pulmonary atresia and TGA. As lung diseases sometimes present these symptoms, a test can be performed with 100% oxygen inhalation for 15 minutes. If the pO_2 and arterial oxygen saturation increase to > 250 mmHg or 97%,

respectively, it must be a pulmonary condition, not a cardiac one. A cardiac disease must be where there is no improvement with oxygen inhalation.

Severe heart diseases such as LVHS can cause low cardiac output and severe HF with DA closure, significant fatigue with feeding, excessive sweating, pale skin, progressive tachypnea, and low pulse amplitude. Therefore, these pictures must be differentiated from neonatal sepsis.

On physical exam, can be found tachypnea (RR > 70 bpm), hypoxemia with arterial O_2 saturation < 95%; heart rhythm disturbances, rate > 180 bpm or < 90 bpm, visible or palpable precordial impulses, hyperphonesis of cardiac bullae; low pulse amplitude (low output) or asymmetry (CoAo) and pathological heart murmurs.

CHDs can occur isolated (90%), or associated with chromosomal abnormalities (5%) and genetic syndromes (3%), with the possibility of transmission of approximately 3 to 10% in isolated defects and up to 50 to 75% in some genetic syndromes.

Some chromosomal alterations, such as trisomy 18 (Edwards syndrome) and 13 (Patau syndrome) are associated with complex cardiac malformations with short life expectancy. Trisomy 21 or Down syndrome presents heart disease in half of the cases, the most frequent being VSD, the common atrioventricular canal, and TOF less frequently, susceptible to surgical correction and long-term correction survival.

Other syndromes are caused by the absence of a chromosome, such as Turner syndrome (45X), which is associated with aortic coarctation or partial chromosome abnormalities, such as 22q11.2 deletion or DiGeorge syndrome, which may present conotruncal abnormalities severe, such as truncus arteriosus.

Autosomal genetic transmission can be dominant and recessive. Among the multiple diseases with cardiac involvement and this type of transmission, we have the Noonan, Marfan, and Holt-Oram syndromes, among others. Therefore, the presence of a syndrome with cardiac compromise can lead to the diagnosis of CHDs.

Thus, neonatal screening for CHD must consider symptoms, the pulse oximetry test already incorporated into the newborn routine in some countries, and the presence of family syndromes or diseases that involve the transmission of cardiac malformation.

BIBLIOGRAPHY

1. Van der Linde D, Konings EE, Slager MA, Witsenburg M, Helbing WA, Takkenberg JJ et al. Birth prevalence of congenital heart disease worldwide: a systematic review and meta-analysis. J Am Coll Cardiol. 2011; 58: 2241-2247.
2. Rivera IR, Silva MA, Fernandes JM, Thomaz AC, Soriano CR, Souza MG. Congenital heart diseases in the newborn: from the pediatrician's request to the cardiologist's evaluation. Arq Bras Cardiol. 2007; 89 (1): 6-10.
3. Afiune JY. Sistematizacao do atendimento ao recém-nascido com suspeita ou diagnóstico de cardiopatia congenita. Manual de orientacao. Sociedade Brasileira de Pediatria, 2022. Available in: https://www.sbp.com.br/imprensa/detalhe/nid/sistematizacao-do-atendimento-ao-recem-nascido-com-suspeita-ou-diagnostico-de-cardiopatia-congenita/
4. Wren C, Reinhardt Z, Khawaja K. Twenty-year trends in diagnosis of life-threatening neonatal cardiovascular malformations. Arch Dis Child Fetal Neonatal Ed. 2008; 93 (1): F33-35.
5. Marino BS, Bird GL, Wernovsky G. Diagnosis and management of the newborn with suspected congenital heart disease. Clin Perinatol. 2001; 28: 91-133.
6. Thangaratinam S, Brown K, Zamora J, Khan KS, Ewer AK. Pulse oximetry screening for critical congenital heart defects in asymptomatic newborn babies: A systematic review and meta-analysis. Lancet. 2012; 379: 2459-2464.
7. Khoshnood B, Lelong N, Houyel L, Bonnet D, Ballon M, Jouannic J-M, et al. Impact of prenatal diagnosis on survival of newborns with four congenital heart defects: a prospective, population-based cohort study in France (the EPICARD Study). BMJ Open. 2017; 7 (11): e018285.
8. Calcagni G, Unolt M, Digilio MC, Baban A, Versacci P, Tartaglia M et al. Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. Expert Rev Mol Diagn. 2017; 17 (9): 861-870.
9. Zeigler VL. Congenital heart disease and genetics. Crit Care Nurs Clin North Am. 2008; 20 (2): 159-169.