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Diagnosis and management of life-threatening cardiac malformations in the newborn

Mats Mellander*

Department of Paediatric Cardiology, Queen Silvia Children's Hospital, Sahlgrenska University Hospital, Göteborg, Sweden

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SUMMARY

Approximately 1–2 per 1000 newborn babies have a cardiac defect that is potentially life-threatening usually because either the systemic or the pulmonary blood flow is dependent on a patent ductus arteriosus. A significant proportion of newborns with such cardiac defects are being discharged from well-baby nurseries without a diagnosis and therefore risk circulatory collapse and death. This risk is greatest for defects with duct-dependent systemic circulation, notably aortic arch obstruction, but is also significant in transposition of the great arteries, for example. The solution to this problem, apart from improving prenatal detection rates, is to introduce effective neonatal screening including routine pulse oximetry.

1. Introduction

Cardiac malformations are the most frequently occurring of all congenital malformations with a prevalence at live birth of around 0.8%.[1–3] Most of these defects are not directly life-threatening for the newborn. However, 10–20% of all neonates with cardiac malformations, corresponding to ~1–2 per 1000 newborns, have a cardiac defect that if undetected may cause circulatory collapse and death during the neonatal period.[4–9] Abu-Harb et al.[4] found that cardiovascular malformations accounted for 9% of all infant deaths and 43% of all infant deaths due to congenital malformations. In that study 30% of the cases were not diagnosed before death. Most of these cardiac defects are life-threatening because they have a duct-dependent systemic or pulmonary circulation. A smaller proportion are life-threatening because they cause severe cardiac failure or arterial desaturation that develop early for other reasons than ductal closure.

The purpose of this article is to review the diagnosis and management of cardiac malformations that are life-threatening to the newborn with emphasis on how to avoid circulatory collapse and death through early postnatal diagnosis when the lesion has not been diagnosed prenatally.

Primary heart disease other than cardiac malformations may also be life-threatening to the neonate, although more rarely, for example cardiac arrhythmias, cardiomyopathies, cardiac tumours and myocarditis. These are beyond the scope of this review.

2. Cardiac malformations that are potentially life-threatening to the newborn

The most widespread cardiac malformations with duct-dependent systemic circulation are coarctation of the aorta (CoA) and interrupted aortic arch (IAA), critical aortic valve stenosis (AoS) and hypoplastic left heart syndrome (HLHS). In CoA, the lower body is supplied by flow through the ductus arteriosus. In IAA type B, also the left subclavian artery and in type C the left carotid are supplied by the duct. In HLHS and AoS the circulation to both the upper and lower part of the body is dependent on flow from the right ventricle through the ductus arteriosus. Sometimes obstruction to systemic flow occurs at several levels simultaneously, for example CoA and aortic valve stenosis or a supravalvular mitral membrane, parachute mitral valve, subaortic stenosis and CoA (Shone's complex).

Duct-dependent pulmonary blood flow occurs in pulmonary atresia with intact ventricular septum (PA/IVS), critical pulmonary valve stenosis (CPS), tetralogy of Fallot (ToF) with critical subvalvar and/or valvar pulmonary stenosis, pulmonary atresia with ventricular septal defect without collateral flow to the lungs (PA/VSD), tricuspid atresia (TA) with a restrictive VSD and in some forms of severe Ebstein's anomaly. In simple transposition of the great arteries (TGA) the mixing of systemic and pulmonary venous blood is dependent mainly on a patent foramen ovale but a patent ductus arteriosus is often also necessary to optimize mixing. For the

* Tel.: +46 313434659; fax: +46 31845029.
E-mail address: mats.mellander@vgregion.se.

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purpose of this review, TGA is considered among the defects with duct-dependent pulmonary blood flow.

Some cardiac defects can be life-threatening to the newborn even if neither the systemic nor the pulmonary blood flow is duct-dependent. One such cardiac defect is total anomalous pulmonary venous return (TAPVR) with obstruction to pulmonary venous flow.

3. Missed diagnoses

A significant proportion of newborns with life-threatening cardiac defects are being discharged from well-baby nurseries without a diagnosis.[4–8,10–12] In a study based on 351 843 births in Sweden, 259 newborns had critical cardiac defects defined as those requiring surgical or catheter-based intervention before 2 months of age. The most numerous defects were CoA ($n = 64$), HLHS ($n = 51$) and TGA ($n = 64$). Out of 129 with duct-dependent systemic circulation, 38 (30%) were discharged home without suspicion of heart disease.[7] The corresponding proportion for those with duct-dependent pulmonary circulation was lower at 4 of 106. More than 40% of those discharged undiagnosed were in circulatory shock at admission. The most common diagnosis among those discharged was CoA. This was in concordance with other studies showing that aortic arch obstruction is the most frequently missed defect.[5,12] It was also shown that the proportion of newborns with a life-threatening cardiac malformation who were discharged undiagnosed increased in parallel with the increasing proportion of mothers discharged early from maternity units. One explanation of this observation is that early discharge allows less time for the baby to develop detectable signs and symptoms of heart disease while still in hospital.[7] In a later study 44% of newborns with TGA left hospital undiagnosed when pulse oximetry screening was not used, presumably as a result of even earlier discharge.[9] Thus it is clear that one important area for improvement, apart from prenatal detection rates, is the newborn screening routines.

4. Newborn screening for congenital heart defects

Early diagnosis of severe heart disease in the newborn is improved if the examiner has a high index of suspicion and an awareness of the important symptoms and physical findings.

4.1. Symptoms and signs

Increased respiratory rate and/or cyanosis are the most common presenting symptoms. On physical examination a murmur, increased precordial activity, weak or absent brachial and/or femoral pulses and poor peripheral perfusion are signs that should raise the suspicion of a cardiac defect. However, less than half of all newborns with congenital cardiac malformations have a murmur and heart murmurs do not occur more frequently in more severe heart defects.[11] Importantly, a newborn baby with a duct-dependent cardiac defect may not have any symptoms at all as long as the duct is still widely patent, and physical examination may be normal. This is especially true for defects with duct-dependent systemic circulation. In CoA and IAA, for example, femoral pulses may be normal while the duct is patent because the right ventricle supplies the descending aorta through the duct. Respiratory rate and peripheral perfusion may also be normal initially. Oxygen saturations can be normal also in the lower body because of a large left-to-right shunt at the atrial level. When the duct begins to constrict, symptoms develop gradually or rapidly depending on the rate of constriction. Respiratory rate increases, peripheral perfusion decreases and pulses become weaker or disappear. Diuresis decreases and there is increasing metabolic acidosis and if undetected eventually circulatory collapse and death.

In defects with duct-dependent pulmonary blood flow, arterial desaturation is more pronounced because of restriction to pulmonary blood flow and cyanosis is therefore more readily apparent.

4.2. Pulse oximetry screening

Pulse oximetry screening has been evaluated in a few large studies.[9,13–17] In a study in Sweden the criteria for a positive screen result was a preductal and postductal oxygen saturation <95% or a difference between the two measurements >3% on three repeated measurements.[9] The screening region (46 963 births) was compared with the rest of the referral area for pediatric cardiac surgery not using pulse oximetry screening (108 604 births). The risk of leaving hospital with undiagnosed duct-dependent circulation was 5/60 in the screening region compared with 28/100 in the other referring regions and the mortality was significantly higher among those discharged undiagnosed.[9] Importantly the false-positive rate was higher with physical examination than with pulse oximetry (1.90% vs 0.17%). Five cases were missed by combined physical examination and pulse oximetry[9] and all had aortic arch obstruction (CoA or IAA). Furthermore, 8 out of 11 cases with CoA or IAA fulfilled the criteria for a normal pulse oximetry screening test.

Thus aortic arch obstruction is not only one of the most frequent duct-dependent lesions (together with HLHS and TGA) but also the one that is most frequently missed on routine neonatal examination and unfortunately the defect most often missed by pulse oximetry screening. In addition, CoA is one of the most difficult lesions to detect on antenatal ultrasound screening.[18] Postnatally there is some promise in the so-called peripheral perfusion index (PPI).[19] PPI reflects the real-time changes in peripheral blood flow. The ratio of the pulsatile to non-pulsatile components of the infrared signal (PPI) corresponds to the relationship between pulsatile and non-pulsatile flow at the site.[19] It remains to be determined in a prospective study whether the use of this index will increase the timely postnatal detection of CoA.

TGA is also a challenge; as with CoA it is difficult to diagnose prenatally[18] unless the screening protocol includes outflow views and the three vessel and tracheal view. An improvement in prenatal detection rates has been reported from some areas.[20] In maternity wards not using pulse oximetry screening, a significant proportion will also be overlooked postnatally because the mixing at the atrial and ductal levels in some cases is so good that there will be no visible cyanosis during the first few days. In the Swedish pulse oximetry study 44% of the newborns with TGA (11/25) left the hospital undiagnosed if pulse oximetry was not used versus 0% (0/18) in the screening region.[9] One of those 11 died undiagnosed and three suffered preoperative cerebral hemorrhage or seizures. Even with neonatal pulse oximetry screening newborns with TGA are at risk if born in a hospital without specialized pediatric cardiac care. Some of these infants may have profound hypoxia from birth because of a severely restrictive foramen ovale and will need an emergency Rashkind septostomy. A prenatal diagnosis would eliminate this problem since delivery can then be planned in a specialist centre. TGA was the first cardiac defect for which it was clearly shown that a prenatal diagnosis was associated with a significantly better survival[21] and neurocognitive outcome.[20]

Pulse oximetry screening is now becoming standard care in all maternity wards in Sweden and in other countries as well, including the USA after it was endorsed by the Department of Health and Human Services in September 2011.[22,23] This initiative, together with increasing antenatal detection rates of critical cardiac malformations, should result in a decrease in the proportion of undiagnosed newborns who are discharged from the maternity wards.