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Screening for Critical Congenital Heart Disease in Newborns

Christie J. Bruno, DO, Thomas Havranek, MD*

Department of Pediatrics, The Children's Hospital at Montefiore, Albert Einstein College of Medicine, 1601 Tenbroeck Avenue, 2nd Floor, Bronx, NY 10461, USA

Keywords

• Critical congenital heart disease • Newborns • Screening

Key points

- Routine newborn screening for critical congenital heart disease (CCHD) is becoming standard of care.
- Pulse oximetry screening for CCHD is reliable and cost-effective.
- Effective follow-up for screens with positive results must be implemented in order for screening programs to be successful.
- Determination of previously implemented screening programs effectiveness is preliminary and ongoing.

INTRODUCTION

Congenital heart disease affects approximately 40,000 neonates in the United States each year or about 1% of all US national births. It is the most common form of birth defect. The most common type of congenital heart disease lesion is the ventricular septal defect (VSD). A VSD is a noncyanotic heart lesion and is not considered a form of CCHD [1,2]. CCHD, on the other hand, is a heart lesion for which neonates require early surgical intervention to survive. Without intervention, the mortality and rates of survival with significant disability are extremely high. CCHD accounts for 20% of deaths in the neonatal period and is therefore a significant cause of mortality for infants in the first month of life [3].

*Corresponding author. *E-mail address*: thavrane@montefiore.org

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EPIDEMIOLOGY

There are several types of congenital heart disease that are included in CCHD. These defects are listed in Box 1. Of the defects listed in Box 1, the detection of 7 is believed to potentially contribute to an improvement in overall survival with decreased morbidity for CCHD. These defects include hypoplastic left heart syndrome (HLHS), pulmonary atresia, tetralogy of Fallot (TOF), total anomalous pulmonary venous return (TAPVR), transposition of the great arteries (TGA), tricuspid atresia, and truncus arteriosus. It is estimated that CCHD affects 2 to 3 per 1000 live births in the United States. The prevalence of CCHD varies based on geographic area as well as the type of CCHD defect. For example, the literature demonstrates that TOF was the most prevalent type of CCHD in all ethnic groups in one study out of New York State [4]. Another study, within New York State as well, reported a variety of CCHD lesions with no true predominance of one lesion. In the former study, HLHS, TAPVR, and coarctation of the aorta were reported, among others [5].

Neonates born with CCHD are often well appearing for the first 12 to 24 hours of life and may be asymptomatic for a day or two beyond that period. Infants with CCHD typically present with physiologic compromise beyond 24 hours of life related to the type of CCHD lesion. Patients with right- and left-sided obstructive heart lesions are listed in Box 1. Patients typically present with symptoms at the time of closure of the ductus arteriosus. Those with right-sided obstructive lesions lose pulmonary blood flow when the ductus closes,

Box 1: Critical congenital heart disease defects

Left-sided obstructive lesions

1. Hypoplastic left heart syndrome
2. Interrupted aortic arch
3. Critical coarctation of the aorta
4. Critical aortic stenosis

Right-sided obstructive lesions

5. Pulmonary atresia with intact septum
6. Tricuspid atresia
7. Critical pulmonary stenosis
8. Tetralogy of Fallot

Mixing lesions

9. Total anomalous pulmonary venous return
10. Transposition of the great arteries
11. Truncus arteriosus communis