ABBREVIATIONS

AV = Atrioventricular
AoV = Aortic valve
CHD = Congenital heart disease
C-CHD = Critical congenital heart disease(s)
DAo = Descending aorta
DILV = Double inlet left ventricle
DORV = Double outlet right ventricle
d-TGA = Dextro-transposition of the great arteries
ECG = Electrocardiogram
HLHS = Hypoplastic left heart syndrome
LA = Left atrium
LPA = Left pulmonary artery
LV = Left ventricle
LVOT = Left ventricular outflow tract
L-TGA = Levo-transposition of the great arteries
MPA = Main pulmonary artery
PA = Pulmonary atresia
PDA = Patent ductus arteriosus
PFO = Patent foramen ovale
PLAX = Parasternal long-axis
POS = Pulse oximetry screening
PSAX = Parasternal short-axis
PV = Pulmonary valve
RPA = Right pulmonary artery
RV = Right ventricle
RVH = Right ventricular hypertrophy
RVOT = Right ventricular outflow tract
SAX = Short-axis
SMA = Superior mesenteric artery
TAPVR = Total anomalous pulmonary venous return
TOF = Tetralogy of Fallot
TOF-PA = Tetralogy of Fallot with pulmonary atresia
TV = Tricuspid valve
VSD = Ventricular septal defect

There was also significant geographic variation in rates of prenatal detection across states with a low of only 11%, further reinforcing the need to expand the ability of all sonographers to be able to adequately screen for C-CHD. Lesions identifiable on a 4-chamber view such as atrioventricular canal defect or hypoplastic left heart syndrome have detection rates close to 67%, while those requiring outflow tract visualization such as transposition of the great arteries have considerably lower rates of prenatal detection, ~25%.[5] Prenatal detection rates remain poor for conditions such as total anomalous pulmonary venous return and aortic arch obstruction, due to fetal cardiac physiology and associated challenges with detection.[5-7]

Neonates with C-CHD may present with a variety of findings that would warrant an echocardiogram, including tachypnea, cyanosis, and heart murmurs. However, these may not manifest until after 48 hours of life and therefore may be missed during the newborn hospitalization. This delayed manifestation of symptoms is due to the profound hemodynamic changes that occur in the first few days of life as the neonate transitions from fetal circulation to postnatal circulation. In particular, closure of the ductus arteriosus plays a major role in the hemodynamic deterioration in C-CHD that are ductal dependent for systemic or pulmonary blood flow, and the ductus arteriosus may remain open for days. Delayed or missed diagnosis may result in severe cyanosis and/or cardiovascular collapse after discharge from the hospital, which in turn can result in mortality as well as morbidity from hypoxic-ischemic end organ injury, including neurodevelopmental abnormalities due to brain injury.[8-14] Wren *et al* reported from the United Kingdom that 25% of C-CHD were diagnosed after discharge from the newborn nursery.[14] A United States (U.S.)-based study estimated that 29.5% of live-born infants with non-syndromic C-CHD in the National Birth Defect Prevention Study received a diagnosis more than 3 days after birth and late detection varied by C-CHD type (range 7.5%-62%) as well as geographic site.[15] The newborn hospitalization thus represents a critical window during which screening for and detection of C-CHD could potentially result in improved outcomes for these critically ill neonates.[16] These statistics also demonstrate that a discharged newborn is not necessarily free of C-CHD and needs to be evaluated thoroughly with the development of symptoms.

The purpose of this document is to provide the adult sonographer, who does not typically screen for C-CHD, with the essential information and tools needed to detect C-CHD in newborns and aid in life-saving diagnosis.

Pulse Oximetry for Detection of C-CHD

A common feature of many forms of C-CHD is hypoxemia due to the mixing of oxygenated and deoxygenated blood. Hypoxia has to be quite significant (≥ 4-5 gm/dL of deoxyhemoglobin or an oxygen saturation of ≤ ~80%) for cyanosis to be visible to the naked eye and is particularly difficult to detect in infants with pigmented skin, such as Black or Hispanic infants. Pulse oximetry uses the difference in absorption spectra of wavelengths of light between oxygenated and deoxygenated hemoglobin to detect hypoxemia at much milder levels than those detectable by examination alone and is widely accepted as a noninvasive method to measure oxygen saturation in the blood. Multiple studies have looked into the utility of pulse oximetry screening (POS) to detect C-CHD and normal values in newborns have been reported.[17-23] The American Heart Association and American Academy of Pediatrics issued a joint statement in 2009 presenting the evidence for routine use of pulse oximetry in newborns to detect C-CHD. In an analysis of pooled studies of oximetry assessment performed after 24 hours of life, the estimated sensitivity for detecting C-CHD was 69.6% while specificity was 99%, and the positive predictive value was 47%.[24] False-positive screens that required further evaluation occurred in only 0.05% of infants screened after 24 hours. Subsequently, in 2011, a working group convened with members selected by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, the American Academy of Pediatrics, the American College of Cardiology Foundation, and the American Heart Association recommended routine use of POS in well-born and intermediate care nurseries.[25] In September 2011, the U.S. Secretary of Health and Human Services added newborn screening for C-CHD to the Recommended Uniform Screening Panel, an action that was endorsed by academic societies.[26] C-CHD screening with pulse oximetry has become nearly universal in the U.S. with 46 states and the District of Columbia having adopted it into their newborn screening program. A simple algorithm used for POS has been developed to assist the provider in management decisions.[16,27-31]

Targets for Screening

Per the Centers for Disease Control and Prevention (CDC), there are a number of types of C-CHD that are targeted for their reliability of identification by POS. (https://www.cdc.gov/ncbddd/heartdefects/hcp.html#Kemper). They collectively represent common forms of C-CHD presenting with hypoxemia.[30] (Table 1). POS will also detect cyanosis due to a non-C-CHD etiology such as noncritical CHD, sepsis, other infection, persistent pulmonary hypertension, parenchymal or anatomic pulmonary disease, transient tachypnea of the newborn, hypothermia, and hemoglobinopathies.[31] Although not C-CHD, these conditions can pose a significant health risk to the neonate and may