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Literature Review

Early Detection and Management of Critical Congenital Heart Disease

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ABSTRACT

Critical congenital heart disease (CCHD) represents a severe subset of congenital heart defects (CHD) with high neonatal mortality rates if undiagnosed or untreated. This review highlights the prevalence, diagnosis, and management strategies for CCHD. In Asia, CHD prevalence is among the highest globally, contributing significantly to neonatal mortality. Early detection, such as prenatal ultrasound and pulse oximetry screening, plays a crucial role in improving patient outcomes. While fetal echocardiography provides diagnostic insights, it has limitations, emphasizing the value of postnatal pulse oximetry as a cost-effective, non-invasive screening tool. Management involves early interventions, including hemodynamic stabilization, pharmacological therapies like prostaglandin infusion, and surgical or catheter-based procedures. Definitive corrective surgeries, although complex, are increasingly performed in neonates to minimize long-term complications. Early diagnosis and timely referral to advanced facilities remain vital to reducing mortality and enhancing the quality of life for affected neonates.

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INTRODUCTION

Various studies show that Asian countries have the highest prevalence of congenital heart disease (CHD) in the world, with a proportion of 1 per 100 live births. CHD contributes to 81 cases per 100,000 live births (Djer, M.M. & Madiyono, B, 2016). Furthermore, CHD has an impact on 70% of the decrease in life expectancy in the perinatal period (infants aged up to 28 days of life). Critical congenital heart disease (CCHD) is considered a severe manifestation of CHD, with life-threatening symptoms that require intervention within the first year of life. It contributes to 64.7% of infant deaths due to CHD (Zeng et al., 2016). Although Indonesia lacks specific data on CCHD prevalence and mortality, 2017 data shows that congenital abnormalities (including CHD) are the fourth leading cause of death in infants aged 0–7 days and the second leading cause in infants aged 8–28 days. It is estimated that approximately 50,000 babies are born with CHD each year, with about 75% of them having non-critical CHD and around 25% (12,500 babies) being affected by CCHD (Murni et al., 2022). These numbers suggest that the majority of deaths due to congenital abnormalities in infants aged 0–28 days are likely attributed to CHD and CCHD (Chamsi-Pasha et al., 2016). Early diagnosis greatly influences the prognosis and quality of life. Prenatal detection using ultrasound is beneficial but underutilized due to the need for specialized skills. Physical examinations—such as identifying heart murmurs—can only detect approximately half of CCHD cases (Engel et al., 2016). Although echocardiography remains the gold standard, primary health services can utilize pulse oximetry as a cost-effective, painless, and easy alternative (Khamkar et al., 2022). In the United States, routine CCHD screening with pulse oximetry has led to a 33% reduction in CCHD-related deaths over six years, potentially preventing 120 infant deaths annually (Wang et al., 2021).

Critical congenital heart disease (CCHD) is the leading cause of death in infants less than 1 month of age. Prenatal echocardiography is an important examination in order to detect CCHD during the prenatal period. However, sensitivity varies greatly, depending on the skill and experience of the operator, gestational age, fetal position, and type and degree of heart defect (Murni et al., 2022). Therefore, if relying only on fetal echocardiography, it will miss the early diagnosis of CCHD in neonates; examination with “normal” results cannot rule out CCHD (Kondo et al., 2018)

Pulse oximetry is a simple, non-invasive, inexpensive, simple, and accurate test to measure the percentage of oxygen saturation in the blood (Nastiti et al., 2023). It has been widely used since it was first discovered in 1970, and it is considered a vital symptom examination so that it can be detected early based on the percentage of hemoglobin in the blood that is saturated with oxygen, while checking the pulse rate. Thus, the results of fetal echocardiography, physical examination at birth, and pulse oximetry examination results can be used as a standard for early detection of CCHD in neonates, but they cannot replace them as the most important examination (Kondo et al., 2018; Nastiti et al., 2023).

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Critical Congenital Heart Disease

Table 1. Clinical features of CCHD are dependent on the ductus arteriosus

Systemic Circulation Depends on the Ductus Arteriosus	Circulatory Collapse + Cyanosis
Hypoplastic left heart syndrome (HLHS)	TOF with pulmonary atresia
Critical aortic stenosis (AS)	Pulmonary atresia
Coarctation aorta (CoA)	Pulmonary atresia with intact ventricular septum
Interrupted aortic arch (IAA)	Critical pulmonary stenosis
	Tricuspid atresia, with PS/PA (with/without VSD)
	Univentricular heart with PS/PA
	Severe Ebstein anomaly
	Complete transposition of great arteries with IVS (TGA/IVS)

Sources : (Cucerea et al., 2016)

History Taking and Physical Examination

Not all CCHD show symptoms. Newborns with CCHD appear healthy and asymptomatic at birth. Symptoms and signs of CCHD appear when the ductus arteriosus closes, usually when the baby has been discharged from the birthplace. (Cucerea et al., 2016)

The main symptoms commonly observed in CCHD include central cyanosis, which manifests as a bluish discoloration of the tongue, gums, and buccal mucosa, especially when the pulse oximeter reading is below 80%. Affected neonates may also experience shortness of breath, which is related to low oxygen saturation and may resemble respiratory conditions such as pneumonia or asthma. Another important clinical sign is cyanosis of the skin or mucous membranes—including the lips and nails—indicating inadequate oxygenation or circulatory compromise.

Other important clinical indicators include irregular heartbeat, where low oxygen levels may result in tachycardia or arrhythmias that require prompt attention. Additionally, decreased systemic perfusion may be suspected when the pulse pressure in the lower extremities is weaker than in the right arm or when the leg blood pressure is lower than that in the right arm.

A reliable diagnosis of CCHD can be established through a combination of three approaches: pulse oximeter screening, physical examination, and echocardiography. The combination of these three things is the best approach in order to prevent delays in diagnosis.6 Health workers at the health center need to do a physical examination and screening using a pulse oximeter first (Kondo et al., 2018; Nastiti et al., 2023). The results

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of the pulse oximetry examination are divided into 3, namely pass (negative), repeat, and fail (positive), meaning that health workers need to refer. If the baby is suspected of having symptoms of CHD, the baby is referred for echocardiography. The following is a pulse oximeter screening examination algorithm at the health center (Murni et al., 2022; Plana et al., 2018)

Early Detection of Congenital Heart Disease

Examination of oxygen saturation in newborns with pulse oximetry is a non-invasive, relatively inexpensive, and easy-to-do method for early detection of CCHD. Every baby born is examined with pulse oximetry on the right hand and one leg. If the clinical condition is stable, the examination is conducted after the baby is 24 hours old or more, to avoid false positive results (Murni et al., 2022). The examination results are called negative if the saturation (SpO2) of the right hand and one leg is ≥95% or there is a difference of ≤3%. If the results are negative, no further examination is needed. The examination results are positive if there is a difference in SpO2 of 3% between the upper and lower extremities. Neonates with positive pulse oximetry examination results should be further examined with echocardiography to confirm or rule out the possibility of CCHD (Nastiti et al., 2023)

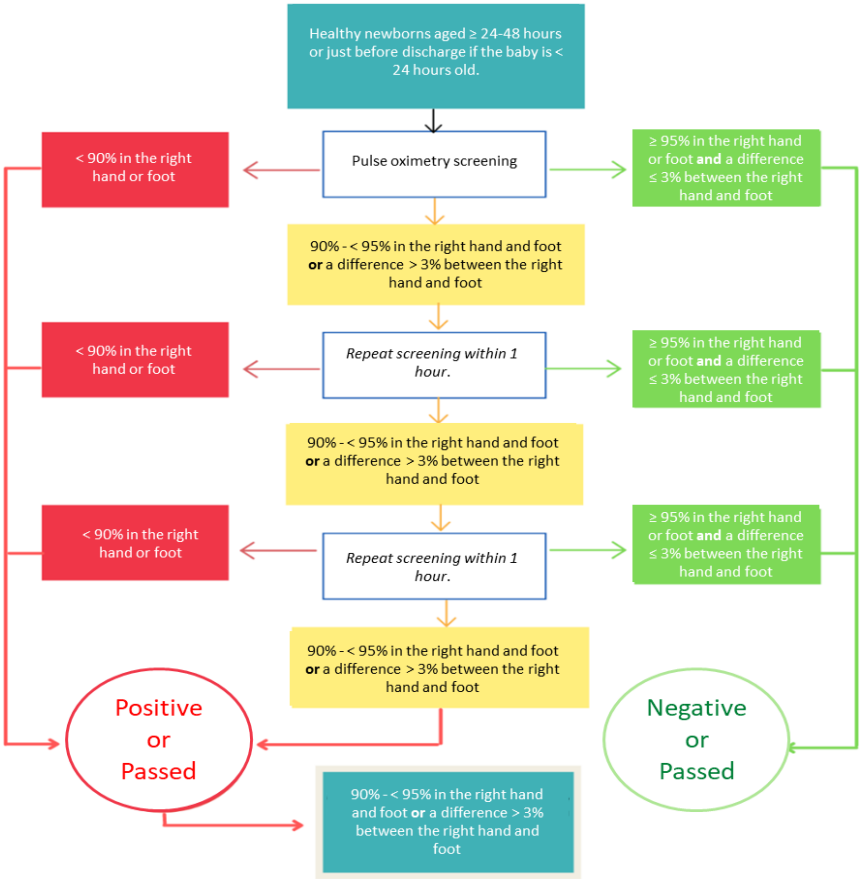

Figure 1. CCHD Screening Algorithm *(Kemenkes RI,2023)*

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DIAGNOSIS

The initial diagnosis of CCHD can be established based on anamnesis, physical examination, and supporting examinations. Important basic supporting examinations are chest x-ray and electrocardiography (van Vliet et al., 2024). Supporting examinations should not delay the management process and referral to more adequate facilities in emergency conditions.

1) Chest x-ray
   Chest x-ray can be used in order to evaluate other congenital causes of respiratory distress in neonates, such as diaphragmatic hernia or congenital cystic adenomatous malformation (CCAM). The presence of cardiomegaly, a typical heart shape, increased lung pattern (plethora) or decreased (oligemia) can direct the diagnosis to certain CHD. Cardiomegaly is one of the symptoms of heart failure. Plethora is seen in CHD with increased pulmonary flow, such as truncus arteriosus or TAPVR. Oligemia is seen in CHD with decreased pulmonary flow, such as, TOF or pulmonary atresia. The boot-shaped heart corresponds to TOF/PA and its variants. The "egg on string" heart shape is seen on TGA. A very large right atrial shadow corresponds to Ebstein anomaly (Djer, M.M. et.al, 2016).
2) Electrocardiography (ECG)
   Electrocardiography can help establish the diagnosis of CHD. Left axis deviation can be found in tricuspid atresia and atrioventricular septal defect (AVSD). The presence of a large pulmonary P wave corresponds to the picture of Ebstein anomaly. Right axis deviation accompanied by a picture of right ventricular hypertrophy is a manifestation of TOF and its variants (Lantin-Hermoso et.al, 2017).

Initial Management of Critical Congenital Heart Disease

Initial management of critical congenital heart disease (CCHD) aims to stabilize the hemodynamic and respiratory conditions of the neonate as quickly and effectively as possible. One of the first steps involves maintaining a warm environmental temperature, either by swaddling the infant or placing them in an incubator. This measure reduces the neonate's oxygen consumption and is especially important in critically ill newborns. Ensuring airway patency is another essential aspect of early management. This can be achieved by adjusting the head position appropriately and, when necessary, performing early endotracheal intubation followed by mechanical ventilation. Oxygenation should be administered with caution to prevent premature closure of the ductus arteriosus. Oxygen saturation is ideally maintained between 75% and 85% to balance adequate oxygen delivery without suppressing prostaglandin-mediated ductal patency (Kritzmire SM, 2023).

Perfusion status must be carefully evaluated, including assessments of consciousness, central and peripheral pulses, capillary refill time, and urine output. Poor perfusion findings—such as weak lower limb pulses or delayed capillary refill—can indicate circulatory collapse or impending shock. Venous access should be established promptly, allowing for the administration of parenteral fluids and correction of acid-base imbalances. In neonates presenting with severe metabolic acidosis, 4.2% sodium bicarbonate (2 mEq/kg/dose) should be given intravenously at a slow rate, corresponding to 2–4 mL/kg/dose (Djer, M.M. et al., 2016).

Hemoglobin levels should be maintained above 15 g/dL to ensure sufficient oxygen-carrying capacity in the bloodstream. If signs of heart failure are evident, treatment

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with inotropic agents and diuretics should be initiated. Rhythm abnormalities require specific pharmacological interventions. For instance, bradyarrhythmias may be corrected using atropine (0.02–0.03 mg/kg), while tachyarrhythmias can be managed using lidocaine, starting with a bolus of 0.5–1 mg/kg, followed by a continuous infusion of 0.02–0.3 mg/kg/min.

Prostaglandin E (PGE) plays a vital role in ductal-dependent cardiac lesions. It should be infused immediately to reopen and maintain the patency of the ductus arteriosus, thereby enhancing cardiac output and correcting metabolic acidosis resulting from hypoxemia or circulatory failure (Akkinapally et al., 2018). Prostaglandin E (PGE) is commonly administered in the form of alprostadil (PGE1) via intravenous infusion. The initial dose is 5 nanograms/kg/minute, and this can be titrated up to a maximum of 50 nanograms/kg/minute as clinically indicated. The primary goal of this intervention is to maintain ductal patency in neonates with ductal-dependent circulation. Apnea is the most common side effect and should be closely monitored. Other adverse effects—such as jitteriness, seizures, fever, flushing, and diarrhea—may also occur but are typically manageable by reducing the dosage (Akkinapally et al., 2018).

In cases where intravenous preparations are unavailable, oral prostaglandin preparations can be considered as an alternative. These are administered at a dose of 10–65 micrograms/kg every 2 hours, but they require extremely close monitoring due to the higher variability in absorption and the risk of systemic side effects. PGE should be initiated in neonates presenting with worsening cyanosis suggestive of ductal-dependent pulmonary circulation, as well as in those experiencing shock or metabolic acidosis likely caused by ductal-dependent systemic circulation. The timely initiation of this therapy is crucial for stabilizing these neonates before referral for definitive care.

Communication, information, and education about the patient's condition and subsequent management to parents are very important. Patients can be referred after initial management, the transport team is ready to accompany, and the referral hospital is ready to receive. The patient's condition at the time of referral must be communicated to the doctor at the referral hospital.

Surgical Therapy

Surgical intervention in congenital heart disease should ideally be conducted as early as possible. Early definitive correction helps prevent structural distortion of the heart and reduces the risk of pulmonary hypertension. Although palliative procedures are still widely practiced, they are primarily aimed at stabilizing the patient's condition while awaiting the appropriate time for definitive surgery. However, such palliative approaches carry risks, including potential heart distortion and the burden of undergoing multiple surgeries with higher associated costs (Djer, M.M. et al., 2016). Ongoing research continues to improve the safety and feasibility of neonatal heart surgeries, with current trends moving toward definitive correction during the neonatal period.

Several types of palliative procedures are commonly performed. One such procedure is pulmonary artery banding, which involves placing a Dacron ribbon to narrow the pulmonary artery's diameter. This technique is typically applied in conditions involving excessive pulmonary flow due to left-to-right shunts, such as large ventricular septal defects, double-outlet right ventricle without pulmonary stenosis, atrioventricular septal defects, and transposition of the great arteries (Djer, M.M. et al., 2016).

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Another form of palliative intervention involves shunting between systemic and pulmonary circulations, especially for cases with significantly reduced pulmonary blood flow leading to cyanosis and acidosis. Common types of shunt procedures include the Classic Blalock-Taussig, which reroutes the subclavian artery to the pulmonary artery; the Modified Blalock-Taussig, which utilizes a Gore-Tex tube between these vessels; the central shunt, which connects the aorta and pulmonary artery using grafts such as Waterson or Potts; and the Glenn shunt (also known as the bidirectional cavo-pulmonary shunt), which establishes a connection between the superior vena cava and the pulmonary artery (Djer, M.M. et al., 2016).

A third type of palliative measure is atrial septostomy, generally performed in infants under three months old. This procedure is carried out using a balloon catheter inserted via the femoral vein and guided by echocardiography, either in an intensive care unit or cardiac catheterization lab. In older children, this procedure follows the Blalock-Hanlon method. Atrial septostomy is indicated in transposition of the great arteries to enhance blood mixing, in partial anomalous pulmonary venous return to reduce congestion, and in tricuspid atresia to alleviate systemic venous congestion (Djer, M.M. et al., 2016).

Advancements in surgical techniques have made corrective surgeries more feasible in the neonatal period, particularly for conditions like total anomalous pulmonary venous return with obstruction, transposition of the great arteries without ventricular septal defect, and truncus arteriosus associated with heart failure. Some congenital heart defects, however, still require delayed correction or initial palliative intervention until the neonate reaches an appropriate age or weight for surgery (Djer, M.M. et al., 2016).

Interventional cardiology therapy

One of the highly anticipated elective procedures in the field of pediatric cardiology is non-surgical interventional cardiology through catheterization in patients with congenital heart disease. CCHD can be repaired by using a thin, flexible tube (catheter) without the need for open-heart surgery; for example, cardiac catheterization can be used to repair a hole in the heart or a narrowed area (Kim, 2017). During cardiac catheterization, the procedure involves inserting one or more catheters into a blood vessel. A small instrument is passed through the catheter to the heart to repair the disease. Some catheter procedures should be conducted in stages over several years. This procedure is not only less traumatic and less likely to cause scarring, but it is also expected to be more cost-effective (Manda YR et.al, 2023).

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Table 2. Types of Cardiology Interventions in CCHD

CCHD Types	Cardiology Interventions
Tetralogy of Fallot	BT *shunt* cito
Pulmonary Atresia	PTBV/PDA *stent*/BT *shunt* cito
Tricuspid Atresia	*Balloon Atrial Septostomy* (BAS)/PDA *stent*/BT *shunt* cito
Transposition of Great Arteries	*Balloon Atrial Septostomy* (BAS)
Total Anomalous Pulmonary Venous Return	*Balloon Atrial Septostomy* (BAS)
Single Ventricle	Diagnostic catheterization BT *shunt*, PA appeal < 3 months
Hypoplastic Left Heart Syndrome	*Balloon Atrial Septostomy* (BAS)/PDA *stenting* cito

Sources : (Cucerea et al., 2016)

CONCLUSION

Critical congenital heart disease (CHD) is considered one of the groups of diseases that have high mortality in newborns and children if not diagnosed immediately and receive palliative or definitive therapy. For therapy in the form of catheterization intervention and/ or surgery, referral to a higher health facility is often required. Initial management actions, availability of drugs, such as prostaglandins, and good referral preparation are very important in ensuring the safety of CCHD patients until they reach the referral hospital.

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