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Early versus late diagnosis of critical congenital heart disease at Sanglah Hospital Denpasar, Bali

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Abstract

Background: Critical congenital heart disease (CCHD) remains significant clinical and public health challenge. Risk of morbidity and mortality in CCHD increases when there is a delay in diagnosis and referral to a tertiary center with expertise in treating these patients. In the last few years, pulse oximetry screening for CCHD in newborns has been added to the list of recommended uniform screening panels and advocated by several health-care authorities. Early detection of CCHD by using pulse oxymetry was recommended by American Academy of Pediatrics (AAP), the American Heart Association, and the American College of Cardiology to improve early identification of infants with CCHD.

Objective: To describe early versus late diagnosis of critical congenital heart disease (CCHD) at single tertiary center.

Methods: A retrospective observational study was conducted in Sanglah Hospital, Bali. Data collected from medical records. Early diagnosis defined as diagnosis which made during prenatal examination or before birth hospital dischare. Late diagnosis defined as diagnosis which made after birth hospital discharge, after 3 days of birth, or even at death. Diagnosis of CCHD was retrieved based on echocardiography examination.

Results: From June 2016 to February2020 we found 86 CCHD cases which were tetralogy of Fallot (41 cases), pulmonary atresia (15 cases), transposition of great arteries (14 cases), total anomalous pulmonary venous return (4 cases), tricuspid atresia (3 cases), truncus arteriosus (3 cases), and hypoplastic left heart syndrome (1 case). Only 26% cases of children with CCHD were diagnosed early, mostly came with chief complaint bluish appearance. Range of oxygen saturation at diagnosis varied from 51-90%. Among cases with late diagnosis, the most common defect was tetralogy of Fallot. Most late diagnosed CCHD came because of referral from other hospitals or pediatricians.

Conclusion: The rate of delayed CCHD diagnosis still occurs in 74%. Factors that might be contribute to late CCHD diagnosis include certain CCHD types, nontertiary hospital nursery and absence of clinical findings.

Keywords: Critical congenital heart disease; Early screening; Early diagnosis; Late diagnosis

1. Introduction

Critical congenital heart disease (CCHD) affects about 10 to 31 per 10,000 live births which remains significant clinical and public health challenge [1]. Risk of morbidity and mortality in CCHD increases when there is a delay in diagnosis and referral to a tertiary center with expertise in treating these patients [2]. There are seven main CCHD screening targets are hypoplastic left heart syndrome, pulmonary atresia, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus [3]. Patients with CCHD usually

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require surgery or catheter intervention in the first year of life to ensure survival. Without intervention, the rates of mortality and survival with significant disability are extremely high. Early diagnosis can potentially improve health outcomes in newborns with CCHD [4].

In the last few years, pulse oximetry screening for CCHD in newborns has been added to the list of recommended uniform screening panels and advocated by several health-care authorities [5]. Early detection of CCHD by using pulse oxymetry was recommended by American Academy of Pediatrics (AAP), the American Heart Association, and the American College of Cardiology to improve early identification of infants with CCHD [6]. There is an increased interest in CCHD screening all over the world. It was estimated that ≥90% of infants born in the United States were screened for CCHD screening by the end of 2014 [7]. Sanglah Hospital was a tertiary hospital located in Bali, Indonesia and as a referral hospital for Bali, West Nusa Tenggara, East Nusa Tenggara, and East Timor. To our knowledge, no study to date has been conducted on screening CCHD in children in Sanglah Hospital, Denpasar. Therefore we aimed to evaluate early versus late detection of CCHD in Sanglah Hospital.

2. Methods

We conduct a retrospective and descriptive study using secondary data collected from register in Sanglah Hospital, Bali between January 2016 to February 2020. The sample of this study was pediatric patients (age 0 to 18 years) with diagnosis of CCHD. Inclusion criteria were patient aged 0 to 12 months with CCHD. Subjects with incomplete echocardiography data were excluded from this study. The sampling technique used was total sampling. We evaluated variable such as gender, types of CCHD, clinical manifestations at diagnosis, oxygen saturation at diagnosis, parity, birth weight, gestational age (premature, term, postdate), referal status, and diagnosis.

Early diagnosis defined as diagnosis which made during prenatal examination or before birth hospital discharge [8]. Late diagnosis defined as diagnosis which made after birth hospital discharge, after 3 days of birth, or even at death [8]. Critical congenital heart defects defined as structural malformations of the heart that are present at birth and require intervention in the first year of life [3]. Diagnosis of CCHD was retrieved based on echocardiography examination. Genetic syndrome suspicion defined as a combination of clinical features include dysmorphic features that are evident on physical examination; multiple anomalies in one patient; unexplained neurocognitive impairment; and a family history that is suggestive of a hereditary disease [9].

Data analysis was done with Statistical Package for Social Sciences (SPSS) software version 23. The discrete variables are expressed as counts (percentage) and continous variables as mean or median. This study was approved by the Ethics Committee of Sanglah Hospital, Denpasar number 2020.12.1.1005.

3. Results

From June 2016 to February 2020 we found 86 CCHD cases. The basic characteristics of subjects were gender, type of CCHD, clinical manifestation at diagnosis, oxygen saturation at diagnosis, and age at diagnosis. Patient demographic characteristic is listed in Table 1.

Table 1 Demographic characteristics of the study

Demographic characteristics	**Early detection (n=22)**	**Late detection (n=64)**
Gender, n (%)		
Male	15 (68.2)	36 (56.3)
Types of CCHD, n (%)		
Tetralogy of Fallot	10 (45.5)	41 (64.1)
Pulmonary atresia	5 (22.7)	15 (23.4)
Transposition of great arteries	4 (18.2)	14 (21.9)
Total anomalous pulmonary venous return	3 (13.6)	4 (6.3)
Others[a]	0 (0)	7 (10.9)

Clinical manifestation at diagnosis, n (%)		
Cyanosis	15 (68.2)	29 (45.3)
Shortness of breath	3 (13.6)	23 (35.9)
Others[b]	4 (18.2)	12 (18.8)
Oxygen saturation at diagnosis, n (%)	67 (11.2)	72 (10.4)
Age at diagnosis, median (range) days	13 (0-39)	120 (14-365)
Other characteristics, n (%)		
Uniparity	12 (54.5)	34 (53.1)
Birth weight, mean (SD) grams	2920 (125.7)	3100 (255.8)
Aterm	20 (90.1)	56 (87.5)
Referal status	5 (22.,7)	47 (73.4)
Appearance with specific syndrome, n (%)	7 (31.8)	5 (7.8)

[a)] Others CCHD include tricuspid atresia (3 cases), truncus arteriosus (3 cases), and hypoplactic left heart syndrome (1 case). [b)] Other clinical manifestations include failure to thrive, cough and recurrent fainting. Specific syndrome were Down syndrome, Pierre-Robin syndrome and Edward syndrome.

Early CCHD detection was found in 25.7% cases, mostly came with cyanosis as clinical manifestation. Range of oxygen saturation at diagnosis varied from 51-90%. Most common CCHD found in early and late diagnosis was tetralogy of Fallot. Diagnosis of tricuspid atresia mostly found in late detection.

4. Discussion

Congenital heart disease (CHD) is the most common congenital malformation, occurring at a frequency of 8-12 per 1000 live births [10]. Critical congenital heart disease (CCHD) defined as any congenital cardiac lesion that requires intervention or may cause significant morbidity or mortality in the first weeks of life.[4] Delays in diagnosis can also lead to significant morbidity and worse outcomes after interventions. Because of its frequency in the population, potential for serious and life-threatening presentation, and availability of effective interventions, CCHD is an excellent candidate for a screening examination [11].

Screening for CCHD could be performed in two ways, prenatally and postnatally. Prenatal screening usually performed by using obstetric ultrasound while postnatal screening done by using pulse oxymetry as recommended by America Heart Association (AHA) and American Academy of Pediatrics (AAP) [8]. Previous studies have examined issues related to late CCHD detection. In Indonesia, screening for CCHD using pulse oxymetry was introduced well in hospitals but there were no data available about early detection for CCHD. In our study, late detection occured in 74% patients. This study was in line with other study in Malaysia with rate of late detection was 51% [12]. The most relevant studies of late detection of CCHD in US published before the recommendation produced widely varied estimated 7.5% to 62.0% of infants with CCHD received late diagnosis [13, 14]. This varied results varied significantly by the presence of extracardiac defects, CCHD types, study site, and income countries classification. Although a more recent study has placed the missed diagnosis rate at 25%, figures vary widely, and it is reasonable to conclude that a more sensitive and uniform newborn screening is needed [15].

Factors that might be contributed to late CCHD detection include hospital nursery levels, the presence of multiple congenital anomaly, low birth weight, prematurity, intrauterine growth restriction, and CHD types [16]. Children with syndrome suspicion were most likely to have early detection of CCHD in our study. Infants with birth defects affecting multiple organ systems may receive additional medical attention prenatally or at birth, which might explain why late detection was significantly lower among such infants. In nonsyndromic infants, late CCHD detection occured in 11 to 39% cases with most common CCHD cases were truncus arteriosus followed by total anomalous pulmonary venous return and Tetralogy of Fallot [17].

Among children with early detection of CCHD, Tetralogy of Fallot, pulmonary atresia and transposition of great arteries were the most widely found. This study was in line with study in North Sumatra in which the most common CCHD found in early detection were Tetralogy of Fallot and transposition of great arteries [18]. Tetralogy of Fallot is the most

common cyanotic heart condition in children, with estimated prevalence 1 in 3,500 to 1 in 4,300 people followed by transposition of great arteries and hypoplastic left heart syndrome [19].

Our study also found Tetralogy of Fallot as the most common CCHD findings among late detection. Study in Massachussets found that most common defects among cases with delayed diagnosis were coarctation of aorta, pulmonary valve stenosis and Tetralogy of Fallot [15]. Other study found the highest rate of late diagnosis was observed in coarctation of the aorta with a rate of 74% [12]. This difference possibly because of difference in prevalence of defects in which the most common CCHD in that study were Tetralogy of Fallot, coarctation of aorta followed by complete atrioventricular septal defects. Unlike most of the primary screening target defects, which tend to present with greatly reduced pulmonary or systemic blood flow, some tetralogy of Fallot cases occur with minimal right ventricular outflow obstruction ("pink tets"), which can result in delayed diagnosis [20]. Similarly, some CCHDs such as coarctation of aorta, aortic stenosis, and pulmonary valve stenosis may not present clinically until weeks or months after birth, depending on the severity of the obstruction [15].

In this study, most patient came with chief complaint bluish appearance. The immediate postnatal period provides another opportunity for screening for CHD via the routine newborn physical examination. Unfortunately, many forms of CCHD do not present with obvious heart murmurs. Cyanosis may not be easily apparent until saturations are <80% and may be more difficult to appreciate in individuals with dark skin pigmentation [21]. A Chinese study found that 46 of 49 newborns (94%) born without symptoms suffered from congenital heart disease. Also, in 8 of 8 newborns (100%) born without symptoms, critical congenital heart disease could be detected by pulse oximetry screening and physical examination when the baby was discharged [22]. Early detection of CCHD is challenging, however, newborns screening using pulse oximetry has been strongly supported by recent literature as a valuable tool facilitating the prompt detection of infants with CCHD. Pulse oximetry is easily accessible, inexpensive, and noninvasive and can be easily performed by the nurses at the infant's bedside; however, it remains a technology that is underutilized in newborns [23].

Limitations of this study were more datas about reasons whether such late detection rate of CCHD was not evaluated. Understanding factors associated with delayed diagnosis could help to improve prenatal and postnatal screening efforts, including pulse oxymetry testing. Further study is needed with a larger sample size to assess the associated factors with late CCHD detection in children. An advantage of this study was that it is a pilot study to assess early and late detection for CCHD in children. Factors that might be contribute to late CCHD diagnosis include certain CCHD types, nontertiary hospital nursery and absence of clinical findings.

5. Conclusion

Most CCHD cases were detected late with cyanotic appearance as the most common feature at first diagnosis. Early CCHD screening by using pulse oxymetry must be done as a routine procedure in every health centre.

Compliance with ethical standards

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Disclosure of conflict of interest

There is no conflict of interests. The author reports no conflicts of interest in this work. By this statement, all authors who consist of Putu Dianisa Rosari Dewi, Eka Gunawijaya and Ni Putu Veny Kartika Yantir have no conflict of interest regarding this manuscript publication.

Statement of informed consent

Informed consent was obtained from the patient whose data mentioned in the study.

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