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Original Article

The importance and effectiveness of cardiac screening in early diagnosis of critical congenital heart diseases

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Abstract

Aim: Cardiac screening test for early diagnosis of critical congenital heart disease (CCHD) is recommended by our ministry of health in newborns. We wanted to investigate the effectiveness of the pulse-oximetry screening test recommended by the ministry of health in our neonatal intensive care unit. Our study is planned to find an answer to this question regarding the subject matter.

Materials and Methods: Our study was planned retrospectively in cases followed up in our neonatal intensive care unit and obstetrics and gynecology clinic. Our study cases were accepted to the neonatal service of our hospital starting from 01/10/2015 in the 30-month period. Patients admitted from the neonatal and obstetrics / gynecology services were included in the study. Saturation measurements of these cases were made at the earliest 6th hour after birth. The test was considered positive, if saturation was <90 % in the right hand or the saturation was 90-94 % plus the right hand and any of the lower extremities saturation difference was greater than 3 % in three measurements performed at one hour intervals. Pulse-oximetry screening test was performed in all cases included in the study. SPSS 21.0 (Chicago, Illinois) was used for statistical analysis.

Results: A total of 12,504 cases were included in our study. Considering the exclusion criteria, some of our cases were excluded from the study, and CCHD was detected in 45 of the 12,223 cases accepted by ECHO examination. 36 of these 45 cases were suspected of CCHD with a physical examination and 41 with the pulse-oximetry screening test and were referred to the pediatric cardiology outpatient clinic. Pulse-oximetry screening test was positive in all 36 cases with CCHD determined by physical examination, but physical examination was found negative in 5 of 41 cases where pulse-oximetry screening test was positive. The 4 CCHD patients in the study could not be determined either by physical examination or by the pulse-oximetry screening test.

Conclusion: Physical examination alone does not have sufficient sensitivity and specificity. Pulse-oximetry screening test is more effective than physical examination in detecting neonatal cases with CCHD. Therefore, the appropriate combination of physical examination and pulse-oximetry screening test in the detection of CCHD cases may provide an advantage to physicians in early diagnosis.

Keywords: Critical congenital heart diseases; early diagnosis newborn; pulse-oximetry screening test

INTRODUCTION

Critical Congenital heart diseases (CCHD) rank first among the congenital problems seen in the neonatal period and are responsible for 40% of deaths due to neonatal anomalies (1). The purpose of cardiac screening performed in the neonatal period is to detect newborns with structural heart defects early and start their treatment as soon as possible. The goal of these screening is to detect seven congenital heart diseases (CHD) early, consisting of hypoplastic left heart syndrome (HLHS), pulmonary atresia (PA), tetralogy of fallot (TOF), total anomalous pulmonary venous connection (TAPVC), transposition of the great arteries (TGA), interrupted aortic arch (IAA), tricuspid atresia (TA) and truncus arteriosus (TAr) (2-4). These CCHD in question were diseases that could be detected by pulse-oximetry screening test (2-5).

Although some of these diseases have obvious clinical findings, most of them may have diminished clinical findings. However, disruptions in symptoms and vital signs may not always be parallel to the severity of the pathology. In addition, 20-25 % of newborns with CHD are thought to be discharged without diagnosis. The importance of the disease increases even more from the CCHD group, which should be intervened in the first 28 days (3).

In addition, the success rate of emergency invasive surgical procedures performed after entering the cardiac shock picture is quite low in newborns (4). Therefore, clinicians are looking for a cheap and easily applicable method to help diagnose CCHD. In the studies in the literature, especially in the early stages, it is mentioned that the wrong diagnosis can be prevented by simple saturation screen in CCHD patients (5,6).

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With this study, we wanted to test the usefulness and efficacy of the pulse oximetry screening test recommended by the American Academy of Pediatrics in the early diagnosis of CCHD in our newborn population.

MATERIALS and METHODS

Newborn cases hospitalized in the neonatal intensive care unit (NICU) of our hospital in a 30-month period starting from 01/10/2015 were retrospectively analyzed and included in this study. The patient population of our study consisted of those who were with their mother in the obstetrics and gynecology service or followed up at the NICU service of our hospital. 281 of these cases were not included in the study as per the criteria below, and the remaining 12,223 cases were applied to the pulse-oximetry screening test. Ethical approval was obtained from Inonu University Scientific Research Publishing Ethics Institution (Health Sciences Non-invasive Clinical Research Ethics Committee) for our study (number of decisions: 2018/10-1).

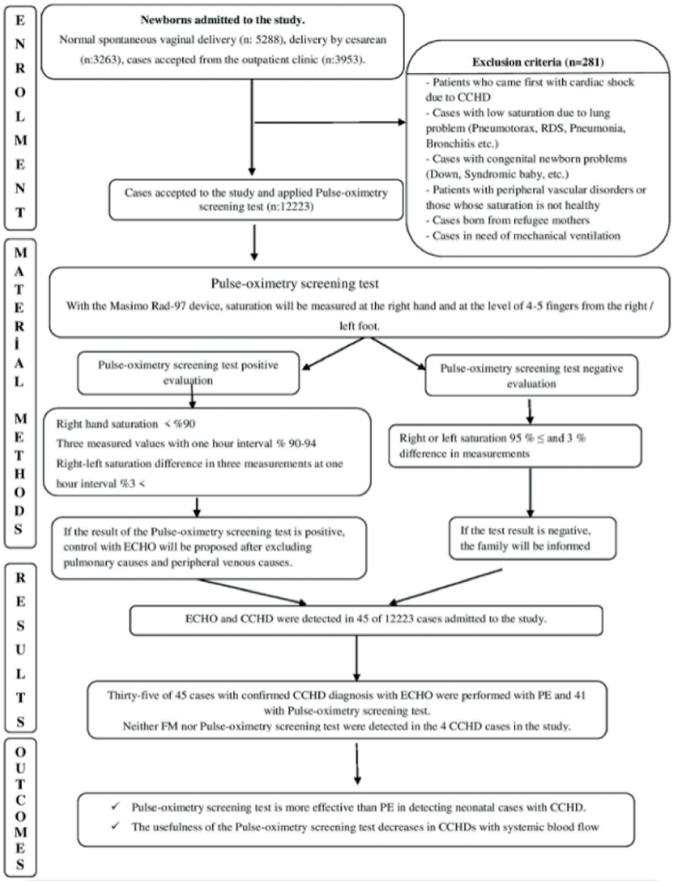
Saturation measurements of the patients included in the study were performed at the earliest 6th hour after birth, with the 4th / 5th fingers of the right hand and any of the lower extremities with the Masimo Rad-97 device. In the saturation measurements performed, the pulse-oximetry screening test was considered to be positive, if the saturation in the right hand was <90%, while the saturation difference was 90-94% in three measurements performed at one hour intervals, or the right-left (right hand and any of the lower extremities) saturation difference was above 3% in three measurements at intervals of one hour. In addition; the pulse-oximetry screening test was considered negative if the right or left saturation measurements were 95% and above in the measurements, or the saturation difference was 3% and below (5). Pulmonary (such as pneumonia, respiratory distress syndrome, transient tachypnea of the newborn) or peripheral venous causes (such as peripheral cyanosis, hypothermia) were excluded in cases with positive test results. Then, the underlying cardiac cause was investigated by performing ECHO in these cases. The cases with negative test results were evaluated as a normal condition and the families of the cases were informed about the subject (Table 1, 2) (Figure 1). Physical examination of cases with positive pulse-oximetry screening test was performed by a neonatal specialist or pediatrician. In physical examination of the cases, cyanosis, cardiac murmur, tachycardia, and inability to obtain femoral pulses were evaluated as positive findings.

Exclusion Criteria

Cases that come with a cardiac shock table, due to lung problem (pneumotorax, respiratory distress syndrome, pneumonia, bronchitis-bronchiolitis), cases with low post-saturation saturation, cases with congenital neonatal problems (Down syndrome, syndromic babies etc.), or cases whose saturation could not be measured in a healthy manner, cases of asylum-seeking mothers and cases requiring tertiary intensive care mechanical ventilation were not included in the study.

Statistical Analysis

SPSS 21.0 (SPSS Inc. Chicago, Illinois) was used for statistical analyses. Shapiro-Wilk normality test was utilized to determine whether the data had normal distribution. Data with normal distribution were compared with independent sample t test, and Mann-Whitney U test was used for inter-group comparisons of non-normally distributed data analysis. Chi-square test was used to analyze categorical variables. Statistical significance was set at $p<0.05$.



CCHD: Critical congenital heart disease; ECHO: Echocardiography; PE: Physical examination; PDA: Patent ductus arteriosus

Figure 1. Flow diagram of our study on the effectiveness of Pulse-oximetry screening test in CCHDs

RESULTS

Our study consisted of 12,504 patients born in our hospital (spontaneous vaginal / cesarean section) or hospitalized in the NICU from the newborn outpatient clinic. 281 of these cases were excluded from the study. As a result, our study was completed with 12,223 cases (Table 1) (Figure 1).

Of the cases included in the study, 21 (46.6%) of 45 cases with ECHO and CCHD were found in our hospital, which were born with normal vaginal delivery. Twelve (%26.6) of these were babies born by cesarean section in our hospital and the other 12 (%26.6) were not born in our hospital but were followed up by being hospitalized in newborn outpatient clinic. Of 21 cases born vaginally in our hospital and diagnosed with CCHD, 1 had TOF, 1 had

Ebstein's anomaly, 1 had PA, 5 had HLHS, 4 had IAA, 1 had critical aortic stenosis (AS), 4 had TA and 4 had TGA was detected. TA was detected in 4 and TGA in 4 of them. Of the 12 cases that were hospitalized in our NICU and found CCHD, there were TOF in 2 cases, Ebstein anomaly in 1, PA in 1, HLHS in 3, IAA in 3, and TGA in 2. Accordingly, TOF in 5 (11.1%) of 45 cases with CCHD, Ebstein anomaly in 3 (6.6%), PA in 3 (6.6%), HLHS in 12 (26.6%), 9 (%) Critical AS was detected in IAA (2.2%), TA in 5 (11.1%), and TGA in 7 (15.5%) (Table 1) (Figure 1).

Considering the above-mentioned exclusion criteria, a total of 12223 cases were included in the study. CCHD was detected in 45 of these cases and these cases were evaluated by ECHO. 36 (80%) of 45 cases diagnosed with ECHO were referred to the pediatric cardiology outpatient clinic with a physical examination, and 41 (91.1%) with suspected pulse-oximetry screening test method. The pulse-oximetry screening test was positive in all 36 cases in which CCHD was detected by physical examination, but physical examination was normal in 5 of 41 cases where the pulse-oximetry screening test was positive. On the other hand, in none of the cases where CCHD was detected by physical examination, the pulse-oximetry screening test was not found to be negative. The 4 (8.8%) patients with CCHD in the study could not be determined by either physical examination or pulse-oximetry screening test. On the other hand, in none of the cases where CCHD was detected by physical examination, the pulse-oximetry screening test was not found to be negative. The 4 (8.8%) patients with CCHD in the study could not be determined by either physical examination or pulse-oximetry screening. On the other hand, out of 45 cases with CCHD, TOF in 41 (80%) of 41 cases detected with pulse-oximetry screening test, Ebstein anomaly in 3/3 (100%), PA in 3/3 (100%), HLHS in 10 /12 (83.3%), IAA in 8/9 (88.8%), critical AS in 1/1 (100%), TA in 5/5 (100%), 7/7 TGA was detected in (100%). On the other hand, 5 (11.1%) cases that were not detected by physical examination, but only by the pulse-oximetry screening test were detected as 1 TOF, 1 as HLHS, 1 as IAA and 2 as TGA (Table 2) (Figure 1).

Table 1. Distribution of cases with CCHD

	TOF (n:5)	Ebstain (n:3)	PA (n:3)	HLHS (n:12)	IAA (n:9)	Critical AS (n:1)	TA (n:5)	TGA (n:7)
Outpatient Cases (n:12/3853)	2	1	1	3	3	-	-	2
Normal Vaginal Delivery Service Cases (n:21/5167)	1	1	1	5	4	1	4	4
Cesarean Section Cases (n:12/3203)	2	1	1	4	2	-	1	1

CCHD, Critical congenital heart diseases; TOF, Tetralogy of fallot; PA, Pulmonary atresia; AS, Aortic stenosis; TA, Tricuspid atresia; HLHS, hypoplastic left heart syndrome; IAA, interrupted aortic arch; TGA, Transposition of the great arteries

Table 2. Detection Methods of cases with CCHD

	TOF (n:5)	Ebstain (n:3)	PA (n:3)	HLHS (n:12)	IAA (n:9)	Critical AS (n:1)	TA (n:5)	TGA (n:7)
PE (+) Pulse-oximetry screening test (+)	3	3	3	9	7	1	5	5
PE (+) Pulse-oximetry screening test (-)	-	-	-	-	-	-	-	-
PE (-) Pulse-oximetry screening test (+)	1	-	-	1	1	-	-	2
PE (-) Pulse-oximetry screening test (-)	1	-	-	2	1	-	-	-
Cases with CCHD detected by ECHO	5	3	3	12	9	1	5	7
PE in cases with CCHD (+) (n:36/45)	3/5 (%60.0)	3/3 (%100)	3/3 (%100)	9/12 (%75.0)	7/9 (%77.7)	1/1 (%100)	5/5 (%100)	5/7 (%71.4)
Pulse-oximetry screening test in cases with CCHD (+) (n:41/45)	4/5 (%80.0)	3/3 (%100)	3/3 (%100)	10/12 (%83.3)	8/9 (%88.8)	1/1 (%100)	5/5 (%100)	7/7 (%100)

CCHD, Critical congenital heart diseases; TOF, Tetralogy of fallot; PA, Pulmonary atresia; AS, Aortic stenosis; TA, Tricuspid atresia; HLHS, hypoplastic left heart syndrome; IAA, interrupted aortic arch; TGA, Transposition of the great arteries; ECHO, Echocardiography; PE, Physical examination

DISCUSSION

CCHD is an anomaly with high morbidity and mortality from the neonatal period. In addition, CCHD children, which could not be detected early, have increased cardiac, neurological, cognitive sequelae as well as social and economic losses. In this study, 45 CCHD cases were detected with ECHO. While 36 cases were determined by physical examination of these cases, 41 were determined by pulse-oximetry screening test. With these data, it shows that physical examination alone has 80% sensitivity in determining CCHD. The sensitivity of the pulse-oximetry screening test reveals that it has a high sensitivity of 91.1 % in patients with CCHD. This means that the pulse-oximetry screening test reduces false negative rates of physical examination. However, there were 4 cases (8.8 %) that could not be detected by either a physical examination or a pulse-oximetry screening test. In these cases, if the physical examination and pulse-oximetry screening test are not used together, it shows that the sensitivity decreases. In other words, it is clearly seen that there are cases that cannot be detected even when both methods are together.

In addition, with this study, Ebstein anomaly, PA, critical AS and TA cases (100%) were detected by both physical examination and pulse-oximetry screening test. However, CCHD cases such as TOF, HLHS, IAA and TGA were detected at different sensitivity rates. It is seen that not all of the CCHD cases can be detected, but with the pulse-oximetry screening test, the detection rates of these morbidities increase and even all of the TGA cases (100%) are detected. In a study of 77,000 newborns in the literature, there were 6 false negatives, including 1 HLHS, 1 TOF, and 4 aortic coarctation (7). The false negative rate in this study was 6/70000 (0.007 %). It was remarkable that the CCHDs that caused false negativity in our study and the CCHDs that caused false negativity in the mentioned literature study in the literature were similar. In 4 (2 HLHS, 1 TOF and 1 IAA) out of 12223 cases included in our study, the screening test gave false negative results. Our false negativity rate was found to be 4/12223 (0.032 %).

Quality evaluation of 419 articles published between 2002 and 2019 was made and 5 articles were selected and evaluated. In metaanalysis conducted in 2019, it was found that CCHD was at a sensitivity of 53% only by physical examination. On the other hand, it was determined that CCHD was detected with a higher sensitivity like 92% by using physical examination and cutaneous oximetry together. When we compared our study with other studies, we found that there was a lower sensitivity with physical examination. Pulse oximetry, on the other hand, seems to have similar or almost identical sensitivity results. In the same study, it was observed that the pulse oximetry screening together with the physical examination had a specificity rate of 98% in determining CCHD, while it was found that this specificity was 99% in cases detected by physical examination alone. In the study, it was emphasized that the measurement of oxygen saturation with cutaneous oximeter is more sensitive than the physical examination alone and the most appropriate test for screening in determining CCHD. It is stated that screening in low-middle income areas where there is not enough qualified personnel and medical device-technology is a necessary and feasible test.

In another study conducted in 2017, it was emphasized that pulse oximeter is a safe, non-invasive, easily applicable and easily available screening method. In addition; it is stated that this test can detect clinically undetectable cyanosis, and can be used with prenatal ultrasound and neonatal physical examination. In addition, in another review in which 229,421 newborns were evaluated, it was stated that the pulse oximeter had high specificity (99%) and moderately high sensitivity (76.5%) in detecting CCHD (4). In this article, it is stated that pulse oximeter is cost-effective and cost-neutral in studies conducted in the USA and the UK (7-10).

It should be remembered that the usefulness of the pulse-oximetry screening test is lower in PDA-dependent CCHD than other pathologies. Patients with left ventricular outflow obstruction (such as aortic coarctation, aortic interruption) may be considered to have normal saturation screen depending on the type and weight of the lesion (3). Due to this false negativity, wide PDA, the perfusion of the lower extremities seemed to be sufficient in the first days of life and the high pressure in front of the left ventricle has a great effect on the left-right shunt. In our study, the false negativity of the pulse-oximetry screening test was found in four cases, and 3 of these cases were related to the physiopathology described above. In addition, one case included TOF (pink TOF).

With our study, it was seen that physical examination alone had a low sensitivity in detecting CCHD. However, when pulse-oximetry screening test and physical examination are used together, it is clearly seen that its sensitivity in detecting CCHD is high. This means that false negative rates were reduced with the pulse-oximetry screening test in detecting CCHD. In this way, cases with high morbidity and mortality are found to be diagnosed early and appropriate treatment options can be started early. Thus, the sequelae that may occur in the cases are reduced, as well as the opportunity to be treated with lower costs. The easy applicability, low cost, high sensitivity and reliability, and wide availability of this test make the test important and ideal for screening. Also, the fact that the Pulse-oximetry screening test can be performed in places where there is not enough qualified manpower and medical device-technology (such as Pediatric Cardiologist and ECHO) are important advantages.

LIMITATIONS

Since our study was planned retrospectively, the positivity of the pulse-oximetry screening test in morbidities other than CCHD could not be included in our study. In addition, if the timing of the test could be done later, the positive falsehood results of the test would be more ideally ruled out. It should not be forgotten that this situation is frequently encountered in all hospitals due to patient cycling in the gynecology service.

CONCLUSION

Physical examination alone does not have sufficient sensitivity and specificity in the early diagnosis of CCHD in the neonatal period. The pulse-oximetry screening test is more effective than physical examination in the detection of neonatal cases with CCHD. For this reason, combining the physical examination with the pulse-oximetry screening test appropriately in the evaluation of CCHDs may provide an advantage to clinicians in early recognition of this morbidity.

*Comp*eting Interests: The authors declare that they have no competing interest.

Financial Disclosure: There are no financial supports.

Ethical Approval: We received ethical approval for our study from the Scientific Research and Publication Ethics Committee of Inonu University (2018/10-1).

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