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Modern Strategy for Identification of Congenital Heart Defects in the Neonatal Period

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ABSTRACT

Introduction: Congenital heart defects are the most common congenital anomalies and occur with an incidence from 0.8 to 1% per 1000 live births. In recent years, the pulse oximetry has become a strong candidate for detecting cyanogen congenital heart defects and in combination with routine clinical exam can improve diagnostic of congenital heart diseases. **Objective**: To apply the modern algorithm for early detection of congenital heart defects in order to improve the diagnosis in the neonatal period. **Patients and Methods:** This was a prospective study that included children born in Bihac Cantonal Hospital during 2012. The diagnostic algorithm included a clinical examination of the newborn, measuring of transcutaneous oxygen saturation with the pulse oximeter between 24 and 48 hours of life, and, in some cases, additional tests (cardiac ultrasound). **Results:** A total of 1,865 children were examined. The application of diagnostic protocol identified the existence of congenital heart defects in 29 children. In re-evaluating the auscultator and ultrasound findings, we identified congenital heart defects in 19 children. **Conclusion:** The application of the modern algorithm for early detection of congenital heart diseases in the neonatal period can significantly improve the making of diagnosis of these anomalies. The concept is simple, inexpensive and applicable in most maternity wards.

Key words: newborn, congenital heart defects, pulse oximetry.

1. INTRODUCTION

Congenital heart defects (CHD) are the most common among all birth defects and has an incidence between 0.8 and 1% (1). In clinical practice, the most common is the classification on the basis of hemodynamic events:

Defects with dominant left-right shunt (atrial septal defect, ventricular septal defect, atrio-ventricular septal defect and persistent ductus arteriosus);

Valvular and/or obstructive defects (coarctation of the aorta, aortic and pulmonary stenosis, mitral stenosis, Ebstein anomaly);

Defects with the dominant right-left shunt or cyanogen anomalies (Fallot tetralogy, tricuspid atresia, pulmonary atresia, transposition of the large arteries, double exit right ventricle, double entrance ventricle, truncus arteriosus, hypoplastic left heart syndrome, anomalous inflow of the pulmonary veins) (2).

The etiology is multifactorial and CHD occurs in combination of genetic and environmental factors (2). Approximately one quarter of these children will have cyanogen anomaly, being critically ill and require surgical treatment or catheter intervention during the first year of life (3,4). Since the CHD is leading cause of death in infants with congenital anomalies, even in developed countries such as USA, England, Sweden (5,6,7), early detection would greatly enhance the effect of therapeutic procedures and the final outcome (8). Previous methods of CHD screening (ultrasonography in the second trimester, postnatal examination of the

newborn) have a fairly low rate of CHD detection and a significant number of children are discharged from the maternity ward with undiagnosed congenital heart disease (8, 9, 10, 11). Taking into account new trends for early discharge of the child from the Maternity ward (after 24 or 48 hours of life), the need to find an efficient diagnostic protocol that would eliminate or at least reduced to a minimum, the possibility to discharge a child with unrecognized CHD in the diagnosis is traditionally used a combination of clinical examination, electrocardiography (ECG) and radiological examinations. In this way, not all CHD can be detected (9,12). Ultrasonography is considered to be sovereign in diagnosing cardiac abnormalities, but its use for the purpose of screening is rather limiting (cost, lack of qualified staff) (13). The combination of clinical examination and pulse oximetry enables greater diagnostic sensitivity while ultrasound is focused on selected cases (14).

Pulse oximetry is a diagnostic method that can detect cyanogen anomalies in patients with mild hypoxemia (O_2 saturation 85-95%), which cannot be detected by clinical examination, and is mandatory companion of cyanogen anomalies (15). On the other hand, they do not have to be accompanied by murmur. Murmur is usually wrongly considered as a leading sign of heart disease (16). Non cyanogen anomalies are not accompanied by cyanosis and suspicion of the existence of a structural defect in the heart is generally made based on clinical examination (heart murmur, abnormal respiratory status, absence of peripheral pulsations). Additional diagnostic tests (ECG, X-ray, echocardiography) confirm or exclude the existence of CHD (cyanogen or non-cyanogen). Of crucial importance is the optimal time of diagnosis. This especially applies to cyanogen anomalies that require immediate treatment. Worse preoperative state with such children, due to late diagnosis, also leads to worse postoperative outcome (8).

2. GOALS

- Efficiency analysis of a new review methodology of the cardiovascular system;
- Application of protocol for measurement of arterial oxygen saturation in all infants between 24 and 72 hours of age;
- The proposal of algorithm for early CHD diagnosis in newborns, which is administrable in our conditions.

3. MATERIAL AND METHODS

According to a defined protocol all infants born in the Maternity ward of the Cantonal Hospital in Bihac was examined from 1st of January to 31st December 2012 (1865 children). As a clinical examination parameters were taken into account: skin appearance of the newborn, respiratory status, presence of degenerative stigmata and assessment of the general condition of the child, auscultation evaluation of clinical findings on the heart in the first and in the second day of life, and palpation of peripheral pulses.

Each born in term, warmed child, at gestational period of 37 weeks and more, was subjected to the measurement of transcutaneous saturation by pulsoxymeter between 24 and 72 hours of age, at the right arm and right leg.

Pulse oximetry was performed by functional type of pulse oximeter (GT 700), with sensors for infants. The measurement result with saturation of 95% or more at any metering point or the difference in measurement between the right arm and leg of 3% or less, is considered as normal finding. Otherwise, the measurement was repeated two more times at intervals of one hour. In the case of differing values and after repeated measurements, screening was considered as positive and the child was a candidate for a cardiology consultation and ultrasound of the heart. Based on the parameters of clinical examination and test results of transcutaneous pulsoximetry, to the children was applied one of three protocols.

Protocol 1 involved a normal parameter of clinical examination and proper values of transcutaneous PO. Such children are discharged home, with regular recommendations. **Protocol 2** implied a proper clinical examination parameter, except for the positive physical findings on the heart, and the planned evaluation in our institution, during the seventh day of life. **Protocol 3** was related to children with cyanosis, dyspnea, tachypnea, disturbances in heart rate or rhythm disorders, with or without a heart murmur, absent peripheral pulsations, pathological findings of transcutaneous PO or degenerative stigmata. This child required a transfer to the Department of neonatology for prompt further diagnostics. Ultrasound examination of the heart was made by Aloka 2000 multi frequency probe 3.5 to 5 MHz, by the pediatric cardiologist. All the children studied were monitored with one or more check-ups at the age of 3-6 months, in order to get information about the real number of children who were born with CHD. Data were statistically analyzed, and the results presented in tables and graphs.

4. RESULTS

During the tested period at the Cantonal hospital Bihac 1865 babies were born. The study included children born in term (1,745 children) (Figure 1). In the population of the premature children (115 premature infants) in total were registered four children with CHD, (0.21% of the total number of live births). For these children has not been implemented algorithm for children born in term. The diagnosis was reached by the usual diagnostic tools, during hospitalization at the Department of neonatology.

Of the total number of children included in the study, the majority was discharged from Maternity ward (1535 children). Of that number, 1290 children (84%) had normal clinical findings and proper transcutaneous PO. Murmur in the first day of life is registered in 245 infants (15.9%). Murmur during the second day with 138 of cases (8.9%). According to the protocol, the finding was evaluated during the seventh day of life. From total 34 children still had a heart murmur (2.2%) (Figure 1).

Children with maintained auscultation finding on the heart were sent to a cardiologist. After cardiology con-

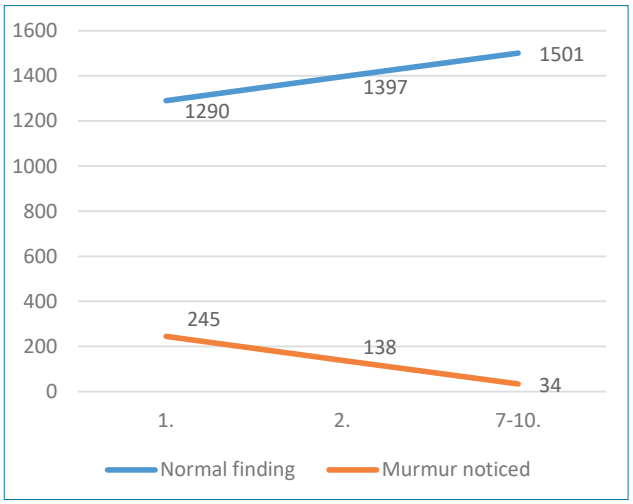

Figure 1. Chart of auscultation findings of children discharged from the Maternity ward

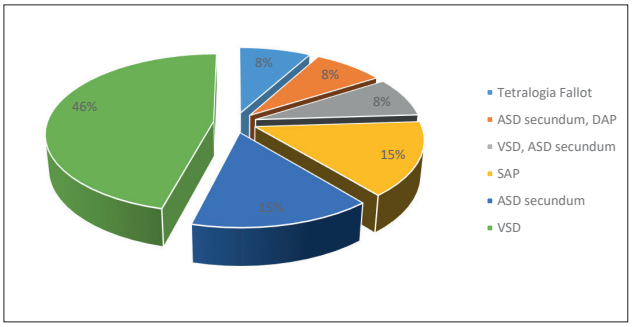

Figure 2. Percentage share of certain heart defects of children discharged from the Maternity ward

sultation, CHD is registered in 13 children (38%) (Figure 2). Of the total number of children born in term, and transferred to the Department of Neonatology (210 children), 41 children were vitally threatened. For 15 vulnerable children the cardiologist was consulted (36.5%). After ultrasound examinations structural defect in the heart had 8 children while in 7 children was a normal or insignificant findings. Of this number 9 children had perinatal asphyxia as the cause life-threatening condition, 6 children were at risk due to respiratory causes and about the same due to neurological etiology.

Of the total number of children at the Department of Neonatology, diagnosed with structural defects in the heart, majority had a ventricular septal defect (38%), and atrial septal defect (38%). Cyanogen defect had one child and one child regurgitant tricuspid valve (Figure 3).

By application of modern diagnostic algorithm for CHD in the neonatal period, there were 29 children with CHD during the study period (1.55% in the total number of live births (1865)). Comparative presentation of the data showed that from the previous (2011) year, when it was registered 1.1% CHD on the total number of live births (1899) (Table 1).

After repeated evaluation periods lasting 3-6 months, reached is the final number of CHD. A total of 19 children was registered during studied year (1.0% of the total number of live births). Comparatively, in 2011, was reg-

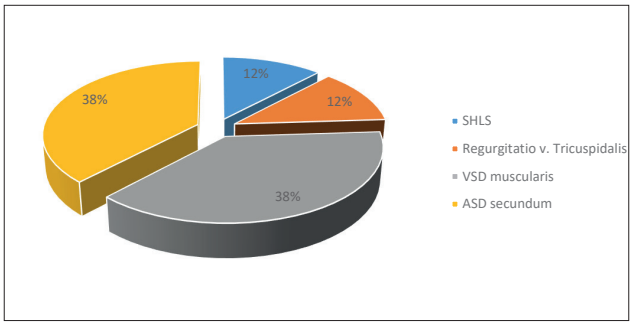

Figure 3 . Share of various heart defects in children transferred to the Department of Neonatology

istered 18 children with CHD (0.9% of the total number of live births) (Table 2).

5. DISCUSSION

In the period from January 1st until December 31st 2012 in the Maternity ward of the Cantonal Hospital Bihac was born 1865 babies. In accordance with pre-specified criteria, the study included 115 prematurely born children. Prematurely born children are by rule hospitalized at the Department of neonatology due to gestational age, follow-up and treatment. Conventional diagnostic methods provide the data on the existence of CHD in four children (3.4% of the total number of CHD in the study year). The children included in the study were screened by contemporary methodology, using the protocol for measuring arterial saturation by transcutaneous PO. Due to the predetermined goal (detection of CHD be-

Congenital heart defect	Number of children			
	2012 year		2011 year	
	N	%	N	%
VSD	10	34.48	10	47.65
ASD secundum	7	24.14	3	14.28
Cyanogen anomalies	3	10.34	2	9.52
SAP	2	6.90	1	4.76
PDA	2	6.90	0	0.00
ASD+VSD	2	6.90	1	4.76
ASD+SAP	1	3.45	0	0.00
ASD+ PDA	1	3.45	2	9.52
Regurgitatio v tricuspidalis	1	3.45	2	11.11
Total	29	100	21	100

Table 1. Comparative presentation of diagnosed defects in the study and the previous year

Congenital heart defect	Number of children			
	2012 year		2011 year	
	N	%	N	%
VSD	5	26.32	5	31.25
ASD secundum	2	10.53	3	18.75
Cyanogen anomalies	3	15.79	2	12.5
SAP	2	10.53	1	6.25
PDA	2	10.53	0	0.00
ASD+VSD	2	10.53	1	6.25
ASD+SAP	1	5.26	0	0.00
ASD+ PDA	1	5.26	2	12.5
Regurgitatio v tricuspidalis	1	5.26	2	12.5
Total	19	100	16	100

Table 2. Comparative presentation of the final number of anomalies in the study and the previous year

fore discharge from the maternity ward), was applied the so-called late screening with PO. This method provides fewer false-positive results, requires a smaller number of repeated testing, and additional, often unnecessary tests, are reduced to a minimum [17]. In our study retesting was required only for 7 children (0.4%), with the aim of excluding the healthy children with delayed transition circulation (18,19), and consequently a false positive test for transcutaneous PO measurement. Normal values of saturation in most cases can be read already at 12 or more hours of life (20). However, half of the children with cyanogen CHD symptoms develop after the second day of life (21). Screening with PO in the first hours of life has more false positive results due to other diseases of non-cardiac etiology (respiratory causes, conditions that are accompanied by seizures, wound sepsis) (18,19,22). Although also these conditions have the benefit of an earlier recognition and timely treatment (16). Combining PO screening and clinical examination enables greater sensitivity in the detection of CHD, compared to PO applied alone, which is consistent with other studies (16,23,24). As part of our research, from 1535 children discharged from the Maternity ward, murmurs were noted in the first day of life in 15.96% of the children in order that, ultimately, 2.2% of children were referred for cardiac evaluation. Of the total number of patients referred to cardiologist, 38% had a structural defect of the heart. For the most part it was a case of non-cyanogen anomalies and all had normal findings of transcutaneous PO. Although the descriptions of the positive screening at PO of non-cyanogen anomalies such as VSD, ASD, PDA, it is almost always in the first 24 hours of life, due to bidirectional shunting of blood due to early postnatal pulmonary hypertension (25). non cyanogen defects are not life threating in the first days of life, and its clinical picture usually manifest at the end of newborn age. Earlier diagnosis will enable optimum monitoring, treatment and planning of possible surgery.

In one child discharged from the maternity ward, after ultrasound examinations was diagnosed cyanogen anomalies (Tetralogia Fallot), with normal transcutaneous PO finding and with pathologic auscultatory findings. False negative findings of PO in this case, interpreting the specificity hemodynamic cardiac anomalies. In fact, it was a variant of the so-called "Pink Fallot" without significant stenosis of the output part of the right ventricle and the dominant left-right shunt at the level of the interventricular septum. In large studies, such as in Germany with the study protocol approved by the Ethics Committee in Leipzig [12] states the number of 4 children with CHD who had a false negative PO finding in relation to the total number of children with negative PO (41388). In three cases it was left-obstructive cardiac anomalies, and in one case a complex cyanogen cardiac anomaly. It has already been noted that of the total number of born, children born in term, included in the study, 210 children were transferred to the Department of neonatology for further diagnosis and treatment. Of this number 41 children were transferred due to life-threatening cyanosis or pathological PO findings. In percentages, in 36.5% of the cases we suspected the existence of cardiac causes of disease, and that sought consultative examination by a cardiologist. Of this number, more than half of the cases CHD was noted, while the other had a normal or insignificant findings. One child had a cyanogen, severe and life-threatening cardiac anomaly (hypoplastic left heart syndrome or SHLS). It is one of the most difficult heart anomalies with the worst prognosis (26). The first pathological finding was the value of transcutaneous PO. Later developed a clinical picture of circulatory arrest (in parallel with the closure of the ductus). SHLS as typical left obstructive anomalies, together with the CoA and aortic stenosis, is classified as a group of abnormalities for which by this type of screening may be missed (27). Measuring the saturation before and after ductus, limits this possibility. We had no other anomalies of the aortic arch, which would refute or confirm this information. Reich and colleagues in their program of screening used measuring saturation on any limb, not necessarily after ductus. Of the three children with cyanogen congenital heart anomaly, was not diagnosed with total anomalous of pulmonary veins (28).

The essence of successful treatment "on the ductus-dependent anomaly" is rapid diagnosis and appropriate therapy. Rehabilitation should begin immediately after diagnosis (prostaglandins) (29).

The total number of newly diagnosed cardiac anomalies and the time frame within which they are diagnosed provides insight into the development of methods for recognition, treatment success and understanding of the whole problem for care of these children.

6. CONCLUSIONS

A new methodology for the review of the cardiovascular system in a population of newborns, with application of protocol for measurements of arterial saturation, represent a modern algorithm for early detection of congenital heart anomalies.

The majority of newborn children has normal values of transcutaneous pulsoximetry. If it is measured with 24 hours of age, it significantly reduces the number of false positive results and reduce the number of repeated measurements. Application of this methodology reduce the number of children referred for cardiac evaluation.

For children referred to a cardiologist, after discharge from the Maternity ward, in majority of cases is a situation of non-cyanogen anomalies, which are not life threatening during the first days of life.

Cyanogen cardiac anomalies usually have pathological values of transcutaneous pulsoximetry. Clinical examination combined with protocol for measurements of arterial saturation, CHD are diagnosed in optimal time for adequate care and monitoring of the child.

Time frame within which the diagnosis is set was significantly shorter in the studied year compared to the previous one.

This approach to newborn, in order to search for congenital heart defects, is simple, efficient and affordable in most maternity wards and should be imperative in institutions of secondary health care level. As such, it has

already been recognized and widely applied in developed countries.

- Conflict of interest: none declared.

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