

Neonatal pulse oximetry screening for congenital heart defects in Switzerland: range of pathology in screening-positive individuals

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Summary

Background: Timely recognition of critical congenital heart defects in neonates is eminent to decrease the associated morbidity and mortality. Clinical symptoms are often lacking within the first few days of life, and 30% of the patients with critical congenital heart defects leave the maternity unit without being diagnosed. Early detection can be improved by measuring the transcutaneous oxygen saturation within the first hours of life. Recent studies on pulse oximetry (pox) screening report a specificity of 95.5 to 100% and a sensitivity of 62–97% for detecting critical congenital heart defects in neonates when postductal transcutaneous saturation is below 95%. Pox screening has been recommended in Switzerland since 2005 by the Swiss Societies of Neonatology and Paediatric Cardiology. The aim of this study was to assess the identification of critical congenital heart defects (CHD) through pox screening by describing patient characteristics in screening-positive individuals.

Methods and results: From January 2009 to January 2011 all Swiss paediatric cardiologists were asked to voluntarily fill out a standardised questionnaire for each patient undergoing echocardiography on account of positive pox screening. Prenatally diagnosed CHD were excluded from this analysis. A total of 48 questionnaires were returned. 27 neonates (56%) had structural heart defects (true positive pox screening test results) including 24 neonates with critical heart defects. In 21 patients (44%), cardiac anatomy was normal (false positive pox-screening test results). In 10 out of the 21 patients, low saturation was explained by evidence of pulmonary hypertension on echocardiography, with right to left shunting through a patent foramen ovale or arterial duct. This analysis is limited by underreporting, meaning that the true number of patients detected with pox screening in Switzerland may be higher. In ad-

dition, some Swiss maternity units do not perform routine pox screening.

Conclusion: The current practice of pox screening in Switzerland made it possible to identify 24 neonates with critical CHD within two years. These patients were diagnosed in time and referred for further treatment in a stable condition. The results of this study support the evidence for the further use of pox screening for CHD in Switzerland.

Key words: pulse oximetry; congenital heart defect; newborns

Introduction

Congenital heart defects (CHD) are the most common congenital defects, with a prevalence of 6 to 8 per 1000 live births [1–2]. If there is need for early intervention, then the term critical CHD is used. In newborns, clinical symptoms of critical CHD are often lacking within the first few days of life. Up to 30% of neonates with critical CHD leave the maternity unit undiagnosed [1]. These patients are at increased risk of sudden deterioration following spontaneous closure of the arterial duct. Pulse oximetry (pox) screening consists of measuring the postductal transcutaneous oxygen saturation within the first 24 hours of life. In patients with saturations <95%, echocardiographic examination is recommended [1]. The screening is noninvasive, and appropriately simple and not very expensive [5]. It has been shown to be effective in the detection of CHD with

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a specificity of 95.5 to 100% [4, 6] and a sensitivity of 62–97% [1–2, 4, 7–8]. In 2005, it was recommended for all Swiss maternity units by the Swiss Societies of Neonatology and Paediatric Cardiology [9]. The aim of this study was to assess the identification of critical congenital heart defects (CHD) through pox screening by describing patient characteristics in screening-positive individuals in Switzerland.

Methods

The study was conducted as a voluntary prospective Swiss multi-centre survey. From January 2009 to Jan-

uary 2011 questionnaires were sent out once a month to all the Swiss paediatric and adult cardiologists involved in neonatal diagnosis of CHD. All neonates born as of 35 weeks of gestation with a positive pox screening test were included in the study. Patients with an antenatal diagnosis of CHD and patients who had obvious severe postnatal symptoms of CHD were excluded from this study, because these patients would have been identified anyway, even in the absence of pox screening. The cardiologists were asked to indicate the patient's age at the time of echocardiography, the results of the echocardiographic examination and any associated clinical findings which could explain a low oxygen saturation. Diagnoses were grouped into categories according to the intracardiac morphology and haemodynamics. Patients with isolated patent arterial duct (PDA) or patent foramen ovale (PFO) were classified as having a normal intracardiac anatomy. Congenital heart defects which require surgery or catheter intervention within the first months of life are called critical CHD. The times specified up until the need for intervention vary in recent publications between the first month and the first year of life [3–4] and has been defined for this paper at six months. Missing data were filled in by contacting the responsible physician directly or by analysing the patient's notes. The study was approved by the local hospital's ethics committee. Descriptive statistics were performed on the data using SPSS 16.0.2., German version for Windows.

Results

During the study period of two years, a total of 48 questionnaires were returned from cardiologists located in the Zurich area (29), Aargau (11), St. Gallen (4), Luzern (2), Thurgau (1) and Vaud (1). The median age at the time of echocardiography was the first day of life (range 1 to 6 days). An overview of the echocardiographic findings in the 48 screening-positive patients is given in figure 1, and the diagnoses are listed in table 1.

In 22 of all 48 screening-positive patients, there were additional clinical findings that might have prompted an echocardiographic evaluation even in the absence of a positive pox screening result: heart murmur (11), cyanosis (8), trisomy 21 (4), trisomy 13 (1), muscular hypotonia (1). A separate analysis was performed for the 24 patients with critical CHD (table 2). 13 of these 24 patients (54%) were asymptomatic at the time of pox screening.

Discussion

The present study demonstrates that, over a period of two years, pox screening in Switzerland allowed the early diagnosis of CHD in 27 neonates, including 24 neonates with critical CHD.

Figure 1
Echocardiographic and clinical findings in 48 patients with a positive pulse oximetry screening result.

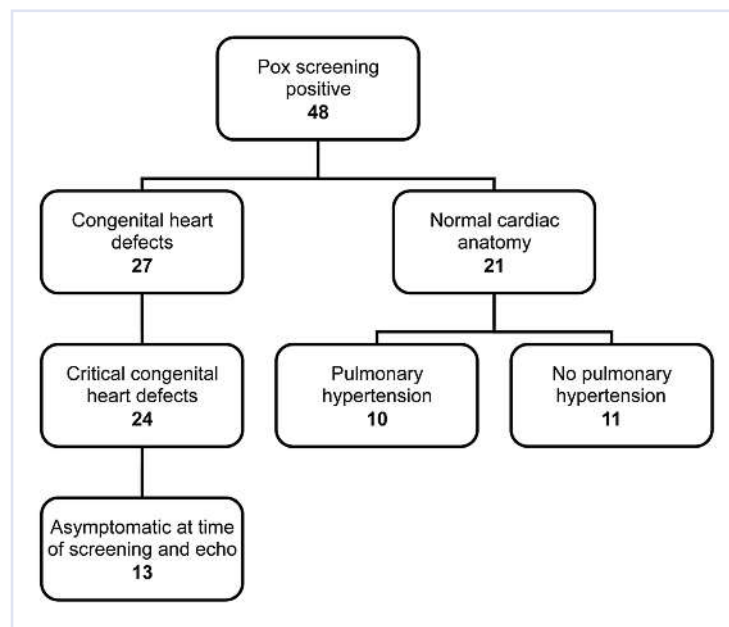


Table 1
Echocardiographic findings in 48 patients with positive pulse oximetry screening.

Group of diagnoses	N
Critical congenital heart defects	24
Transposition of great arteries	7
Total anomalous pulmonary venous return	4
Coarctation of the aorta	3
Complete atrioventricular septal defect	3
Double outlet right ventricle (with transposition of the great arteries)	3
Tetralogy of Fallot	2
Hypoplastic left heart syndrome	1
Pulmonary atresia	1
Congenital heart defects, not critical	3
Ventricular septal defect	2
Pulmonary valve stenosis	1
Normal intracardiac anatomy	21
Total	48

Table 2

Clinical findings in patients with critical congenital heart defects (n = 24). Multiple answers per patient are possible. Data are given as numbers of patients and percentage of the respective group.

		Asymptomatic	Heart murmur	Cyanosis
Coarctation of the aorta	3	1 (33%)	2	1
Transposition of great arteries	7	3 (43%)	0	4
Hypoplastic left heart syndrome	1	0	1	0
Total anomalous pulmonary venous return	4	4 (100%)	0	0
Complete atrioventricular septal defect	3	3 (100%)	0	0
Double outlet right ventricle with transposition of the great arteries	3	1 (33%)	1	1
Tetralogy of Fallot	2	1 (50%)	1	0
Pulmonary atresia	1	0	0	1
Total	24	13 (54%)		

The list of diagnosed asymptomatic critical CHD is interesting and also involves acyanotic lesions such as coarctation of the aorta and atrioventricular septal defect (table 2). Among those CHD that were positively screened, four were patients with total anomalous pulmonary venous return, and all those four neonates were asymptomatic. These patients are good examples for the benefit of the pox screening because catastrophic cardio circulatory collapse often occurs within the first days of life and it is one of the most commonly prenatal undiagnosed CHD, because recommendations for heart evaluation during the morphology exam at 20–22 weeks is directed towards visualisation of the 4-chamber view and crossing of the great vessels, and this does not allow diagnosis of total anomalous pulmonary venous return in the vast majority of fetuses.

Since pox screening was first described in Switzerland in 2005 [9], a large number of studies have been published on this subject. The results are summarised in a large meta-analysis [10] and in a scientific statement by the American Heart Association and the American Academy of Pediatrics [4]. Pox screening has continuously gained acceptance in Europe and the United States [11]. Our small study of positive screening-test results in Switzerland further supports the benefit of pox screening and, today, there is no reason not to perform the screening in Switzerland.

The normal course of postnatal oxygen saturation is well known [12]: a healthy newborn at the age of two minutes has a mean arterial oxygen saturation of 73%, which rises to 95% within one hour. This process is individual and varies considerably, which explains the high rate of false positive pox screening test results in studies with screenings very soon after birth [5, 13–14]. In our study population, 21/48 patients with a low saturation (positive pox screening) had a normal intracardiac anatomy. Pulmonary hypertension accounts for at least 48% of these false positive test results in our study. Early detection of persistent pulmonary hy-

pertension in neonates is beneficial, including in the absence of CHD, since it can draw attention to an underlying problem like a sepsis.

The number of neonates with critical CHD in Switzerland is not known but can be estimated at approximately 200–300 over a two-year period. Many of these patients did not fulfil the inclusion criteria for this study, because they were diagnosed by routine antenatal ultrasound or in the context of obvious postnatal clinical findings [15–16]. The collected sample of 48 positive screening test results still does not represent the full number of screening-positive test results expected in Switzerland during this period. This underreporting, which is due to a lack of cooperation on the part of the cardiologists involved and also to the incomplete notification of all the physicians engaged in the care and ultrasound screening of neonates, constitutes a weakness of the present study. Further insight into the topic of diagnosis and management of CHD in Switzerland will only be possible with a complete dataset of all the neonates with CHD, including the circumstances of their diagnosis and data on their follow-up.

Conclusion

Transcutaneous pulse oximetry screening is an efficient way of identifying congenital heart defects at an early stage. A significant number of newborns with critical congenital heart defects that have not been detected prenatally can be identified at an asymptomatic stage, before clinical deterioration occurs. This makes it possible to avoid cardiogenic shock in these patients. The results of this study provide further evidence of the usefulness of Switzerland's current pulse oximetry screening policy. In order to improve morbidity and mortality in patients with CHD in Switzerland, an ongoing prospective registry covering all neonates with CHD is required.

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