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REGULAR ARTICLE

Pulse oximetry screening as a complementary strategy to detect critical congenital heart defects

Alf Meberg (alfmeb@start.no)[1], Andreas Andreassen[2], Leif Brunvand[3], Trond Markestad[4], Dag Moster[5], Lutz Nietsch[6], Inger Elisabeth Silberg[7], Jan Einar Skålevik[8]

1.Department of Paediatrics, Vestfold Hospital, Tønsberg, Norway
2.Department of Paediatrics, Haugesund Hospital, Haugesund, Norway
3.Department of Paediatrics, Ullevål University Hospital, Oslo, Norway
4.Department of Paediatrics, Innlandet Hospital, Gjøvik, Norway
5.Department of Paediatrics, Haukeland University Hospital, Bergen, Norway
6.Department of Paediatrics, Ålesund Hospital, Ålesund, Norway
7.Department of Paediatrics, The National Hospital, Oslo, Norway
8.Department of Paediatrics, University Hospital of Northern Norway, Tromsø, Norway

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Correspondence
Alf Meberg, MD, PhD, Department of Paediatrics, Vestfold Hospital, 3103 Tønsberg, Norway.
Tel: +47 33 34 20 00 |
Fax: +47 33 34 39 63 |
Email: alfmeb@start.no

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Abstract

Objective: To compare strategies with and without first-day of life pulse oximetry screening to detect critical congenital heart defects (CCHDs).

Study design: Population based study including all live born infants in Norway in 2005 and 2006 (n = 116 057). Postductal (foot) arterial oxygen saturation (SpO_2) was measured in apparently healthy newborns after transferral to the nursery, with $SpO_2 < 95\%$ as cut-off point. Out of 57 959 live births in the hospitals performing pulse oximetry screening, 50 008 (86%) were screened.

Results: A total of 136 CCHDs (1.2 per 1000) were diagnosed, 38 (28%) of these prenatally. Of the CCHDs detected after birth, 44/50 (88%) were detected before discharge in the population offered pulse oximetry screening (25 by pulse oximetry), compared to 37/48 (77%) in the non-screened population ($p = 0.15$). Median times for diagnosing CCHDs in-hospital before discharge were 6 and 16 h after birth respectively ($p < 0.0001$). In the screened population 6/50 (12%) CCHDs were missed and recognized after discharge because of symptoms. Two of the six missed cases failed the pulse oximetry screening, but were overlooked (echocardiography not performed before discharge). If these cases had been recognized, 4/50 (8%) would have been missed compared to 11/48 (23%) in the non-screened population ($p = 0.05$). Of the cases missed, 14/17 (82%) had left-sided obstructive lesions.

Conclusion: First-day of life pulse oximetry screening provides early in-hospital detection of CCHDs and may reduce the number missed and diagnosed after discharge.

INTRODUCTION

Today, diagnostic strategies for the detection of congenital heart defects (CHDs) include prenatal ultrasound screening programmes, routine clinical examination of apparently healthy babies in the nursery and extensive clinical and laboratory investigations of infants transferred to a neonatal special or intensive care unit. For the subgroup of critical congenital heart defects (CCHDs), such as ductus dependent and cyanotic lesions, early diagnosis is a special challenge. A substantial percentage are discharged home undiagnosed and readmitted with severe heart failure or circulatory collapse (1–4). Screening apparently healthy babies with pulse oximetry has been put forth as a complementary strategy for early detection of CCHDs, because they often present with a decreased arterial oxygen saturation (SpO_2) (5–12). The aim of this study was to compare the effects of a pulse oximetry-screening programme for diagnosing CCHDs with a non-screened control population.

Abbreviations
CHD, congenital heart defect; CCHD, critical congenital heart defect; SpO_2, arterial oxygen saturation.

MATERIAL AND METHODS

Fourteen Norwegian hospitals with obstetric departments as well as paediatric services and neonatal special or intensive care units established a pulse oximetry-screening programme for infants born between 2005 and 2006 (9). Postductal (probe on the foot) SpO_2 was consecutively measured in apparently healthy babies on their first day of life when admitted to the nursery from the delivery suite. The pulse oximetry probe was attached for at least 2 min, until a stable recording was observed. A nurse or midwife examined the baby for clinical symptoms (tachypnoe, cyanosis) when $SpO_2 < 95\%$. If symptomatic, the paediatrician was contacted. If asymptomatic, the infant was retested 2–3 h later. If the infant failed the retest, the paediatrician was contacted. If $SpO_2 \geq 95\%$, the infant was referred for a routine clinical examination. Infants transferred to a special

or intensive care unit directly after delivery or from the nursery before SpO_2 was measured, or who had a heart defect detected prenatally, were not included in the screening programme.

A pulse oximeter RAD-5v (Masimo Corporation, Irvine, CA, USA) with a multisite reusable sensor (LNOP® YI) was used for the SpO_2-measurements. This new generation technology secures reliable oxygen saturation values in newborn infants, overcoming previous limitations related to low perfusion states and motion artifacts (13). The cut-off for SpO_2 (<95%) was based on measurements in 1000 consecutively admitted babies showing a SpO_2 of 95% to represent the 2.5 centile for distribution of the saturation measurements. Further investigations, such as echocardiography, radiographic studies or blood samples were performed according to local routines and at the discretion of the paediatrician.

CCHDs were defined as ductus dependent and cyanotic lesions. Tetralogy of Fallot, most often acyanotic at birth, was classified as critical only when the extreme type existed (pulmonary atresia with ventricular septal defect; classified as pulmonary atresia) (14). CCHDs in the screened population were registered prospectively, until six months after the last infants were born. In the population born at hospitals not offering pulse oximetry screening, CCHDs were retrospectively registered through the database systems of the hospitals. The database used at the Pediatric Cardiology Unit at The National Hospital, Oslo, the tertiary care referral centre for CCHDs for all of Norway, was checked to confirm diagnosis and secure complete ascertainment of cases.

Some of the data from the pulse oximetry screened population have been published as part of a project investigating the statistical accuracy of such screening in detecting CHDs (9).

The study was approved by the Regional Committee for Medical Research Ethics.

Statistical analysis

Statistical analyses were performed by the chi-square test, Fischer's exact test and the Wilcoxon signed-rank test. A probability value of $p < 0.05$ was regarded as significant.

RESULTS

A total of 116 057 infants were live born in Norway during the years 2005 and 2006. Of these 57 959 were born in the hospitals where first day of life pulse oximetry was performed (of whom 50 008 (86%) were screened). A total of 58 098 were born in hospitals without pulse oximetry screening. In the two populations, respectively, 24 233 (42%) and 29 210 (50%) were born in university hospitals, 32 773 (57%) and 19 308 (33%) in other hospitals with >500 deliveries a year and 953 (2%) and 9580 (16%) in hospitals with less than 500 deliveries a year.

A total of 136 CCHDs were diagnosed (1.2 per 1000). Of these 88 (65%) had cyanotic heart defects (transposition of the great arteries 39, pulmonary atresia 19, total anomalous pulmonary venous return 11, double outlet right ventricle 6, tricuspid atresia 5, single ventricle 4, common arterial truncus 2, Ebstein anomaly 2). Forty-eight (35%) had left-sided obstructive lesions (coarctation of the aorta/interrupted aortic arch 22, hypoplastic left heart syndrome 18, critical aortic stenosis 8).

Table 1 shows the distribution and place of diagnosis of CCHDs. Significantly more CCHDs were born in hospitals offering pulse oximetry screening. This was entirely caused by in utero transport of prenatally detected CCHDs to the tertiary care heart centre, participating in the pulse oximetry-screening project. When allocated to hospitals based on the mother's place of residence, no difference existed for the occurrence of prenatally detected CCHDs (17/67 vs. 21/69 in hospitals screening and not screening with pulse oximetry respectively; $p = 0.51$).

When prenatally detected CCHDs are excluded, 25/50 (50%) CCHDs were detected by the pulse oximetry screening in the population offered such screening. In this population a trend was found for less CCHDs missed and diagnosed after discharge (6/50; 12% vs. 11/48; 23% in the screened and non-screened populations respectively; $p = 0.15$).

Table 1 Diagnosis of critical congenital heart defects (CCHDs)

	Pulse oximetry screening		No pulse oximetry screening		Total	
	n	%	n	%	n	%
Live born infants	57 959		58 098		116 057	
CCHDs in total	81*†		55		136	
Detected prenatally	31*†	38	7	13	38	28
Detected postnatally	50	62	48	87	98	72
Cyanotic	32	64	31	65	63	64
Left-sided obstructive lesions	18	36	17	35	35	36
Pulse oximetry screening	25*‡	50	0	0	25	26
Clinical routine examination in nursery	4	8	11	23	15	15
After transfer to neonatal unit	15*	30	26	54	41	42
In nursery + neonatal unit	19*	38	37	77	56	57
After discharge	6§	12	11	23	17	17

*$p < 0.05$; statistically significant difference from no pulse oximetry screening.

†Significant difference from no pulse oximetry screening because of in utero transports of CCHDs to the tertiary care heart centre offering screening.

‡In addition two cases failed the screening, however, were diagnosed after discharge home and included in that group.

§Two failed the pulse oximetry screening, but were not referred for echocardiography (when excluded, $p = 0.05$ compared to no pulse oximetry screening).

Table 2 Time for detecting critical congenital heart defects (prenatally detected lesions excluded)

	Pulse oximetry screening			No pulse oximetry screening		
	Hours after birth			Hours after birth		
	n	Median	Range	n	Median	Range
Pulse oximetry	25	5	1–21	-	-	-
Routine in nursery	4	16	10–48	11	32	13–72
After transfer to neonatal unit	15	7	1–36	26	6	1–120
Total in-hospital before discharge	44	6*	1–48	37	16	1–120
After discharge	6	624	120–1344	11	240	144–1344

*$p < 0.05$; statistically significant difference from no pulse oximetry screening.

Two patients with CCHDs who failed the pulse oximetry screening (assessed by the paediatrician to be healthy in spite of subnormal SpO_2, and not referred for echocardiography) were not recognized before readmitted with clinical symptoms (heart failure, cyanosis). One had critical aortic stenosis/coractation of the aorta, and the other total anomalous pulmonary venous return. If these two cases had been recognized before discharge, 4/50 (8%) would have been missed compared to 11/48 (23%) in the population not offered pulse oximetry screening ($p = 0.05$). In the hospitals not screening, significantly more CCHDs were diagnosed after transferral to a neonatal special or intensive care unit.

Of the 38 prenatally detected CCHDs, 31 (82%) had $SpO_2 < 95\%$ when examined after birth.

Of the postnatally detected CCHDs, 17/98 (17%) were recognized after discharge. Of these, 14 (82%) were readmitted with circulatory collapse/severe heart failure, two due to heart murmur and one due to cyanosis. 14/17 (82%) had obstructive left heart lesions (coarctation of the aorta/interruption of the aortic arch 9, hypoplastic left heart syndrome 3 and critical aortic stenosis 2). Eight of the missed cases (1.5 per 10 000) occurred in university hospitals, six (1.2 per 10 000) in other hospitals with more than 500 deliveries a year and three (2.8 per 10 000) in hospitals with less than 500 deliveries a year ($p = 0.41$ compared to university hospitals and $p = 0.18$ compared to hospitals with more than 500 deliveries a year). No death from unrecognized CCHD was observed.

Table 2 shows the age of the babies when the CCHDs were detected. The time for in-hospital diagnosis of CCHDs was significantly shorter for the population offered pulse oximetry screening than in the non-screened population.

DISCUSSION

In this population-based study, CCHDs were diagnosed in 1.2 per 1000 live born infants. This is somewhat less than calculated in the study of Liske et al. (15) (1.7 per 1000) and may be explained by exclusion of Tetralogy of Fallot, except for its extreme type (pulmonary atresia with ventricular septal defect), in our study. Of the CCHDs 28% were diagnosed prenatally, leaving the majority for postnatal detection. In the present study the vast majority (88%) of the postnatally detected CCHDs were diagnosed before discharge home in the population offered pulse oximetry screening. The hospitals not screening, however, still detected most such lesions (77%) before discharge, most in the neonatal unit after transfer for clinical cyanosis, murmur, tachypnoea or associated extracardiac malformations. Our results are nearly identical with the findings in a recently published Swedish study, where pulse oximetry reading from both hand and foot combined with physical examination detected 87% of the critical CHDs (16). This increased the detection rate with 14%, compared to physical examination alone.

First-day of life pulse oximetry screening of apparently healthy babies admitted to the nursery alerted the staff to a considerable percentage of CCHDs very early (median 5 h after birth) and caused the time for in-hospital detection of CCHDs to be significantly reduced, compared to the population not offered screening. Valuable time was gained compared to detection by the paediatric routine examination or from symptoms recognized clinically. A trend existed for less CCHDs missed and detected after discharge home. Two cases with CCHDs failing the pulse oximetry screening, were not detected before discharge (echocardiography not performed), underscoring that scrupulous diagnostic procedures should be undertaken, including extensive use of echocardiography, in infants with persistent subnormal SpO_2. If these two cases had been diagnosed before discharge, the number of CCHDs missed would have been statistically significantly reduced (borderline) compared to the non-screened population. Of those CHDs, most were readmitted in a condition of severe heart failure or circulatory collapse. It is an important task to avoid such conditions, as mortality and results of surgery may be negatively influenced by a bad condition of the infant (17).

Reinhardt and Wren found that as many as one-third of infants with a potentially life-threathening CHD may leave the hospital undiagnosed (3). Among these, are CHDs with critical obstruction for blood flow to the lungs (e.g. atresia of the pulmonary or tricuspid valves) or systemic circulation (e.g. hypoplastic left heart syndrome, severe aortic stenosis, coarctation of the aorta or interrupted aortic arch). Such CHDs depend on a patent ductus arteriosus for pulmonary and systemic perfusion, and severe hypoxia, acidosis, circulatory collapse and possibly death will occur when the arterial duct closes (18). Early discharge may increase the risk for ductal closure outside hospital. In our study coarctation

of the aorta and interruption of the aortic arch were the defects especially at risk for being overlooked, as also found by others (1,3,5).

Among infants in need of cardiac surgery before two months of age Mellander and Sunnegårdh (19) found that 4% of CHDs with ductus dependent pulmonary circulation and 30% of those with ductus dependent systemic circulation were missed and readmitted. An even higher percentage (38%) of ductus independent severe CHDs was not detected before discharge. Although ductus dependent, as well as ductus independent CHDs (e.g. lesions with common mixing of blood), often have a decreased SpO_2, clinical cyanosis may not be visible. Measuring SpO_2 is an objective method to overcome this problem.

Fetal ultrasound screening programmes improve detection of major CHDs (20). In routine screening of large populations the detection rate may be low (21). When CCHDs are delivered unrecognized, pulse oximetry screening may represent a useful complementary strategy for early diagnosis, since a low SpO_2 will be found in the majority of cases detected prenatally, in our study 82%.

Although this is not a randomized study, and compares prospective (population screened with pulse oximetry) with retrospective data (population not screened), the data should be valid. The occurrence and distribution of CCHDs are quite similar in the two populations. Place and time for detecting the CCHDs are basic data, which should be correct, even for those registered retrospectively from the records of the patients. The profile of hospitals taking part in the study was somewhat different between the two populations (more infants born in university hospitals and in smaller hospitals in the population not offered pulse oximetry screening). However, the number of cases missed was not related to type of hospital, but increased in proportion to the number of infants born.

In conclusion, diagnostic strategies which include first-day of life pulse oximetry screening was found to detect CCHDs in-hospital earlier than in the population not offered screening, and may reduce the number of missed cases to be diagnosed after discharge home. Coarctation of the aorta and interruption of the aortic arch are the CCHDs especially at risk for being overlooked, and readmitted in circulatory collapse. Pulse oximetry screening of apparently healthy newborn babies should be implemented as part of a combined strategy for early detection of CCHDs.

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CONFLICT OF INTEREST

The authors have no conflicts of interest to declare.

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