

Screening for Critical Congenital Heart Disease (CCHD): Global Implementation Efforts



October 30, 2014

Gerard R. Martin, MD, FAAP, FACC, FAHA
Lisa A. Hom, RN Esq.

2014 APHL Newborn Screening and Genetic Testing Symposium
Anaheim, California

Pulse Oximetry Examined 2002-2007

DOI: 10.1007/s

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PEDIA

Mar 2003
VOL. 111
NO. 3

Effectiveness of

Robert I. Koppel, MD*
Prabhu I

ABSTRACT. Objective.
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Conclusions. This scree
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From the *Department of Pediatric
Hyde Park, New York; §Department
New York; and §Department of Pe
Jalip, New York.
Received for publication Feb 7, 2003
Reprint requests to (R.I.K.) Division
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rkoppel@jhs.edu
PEDIATRICS (ISSN 0031-4005). Co
eny of Pediatrics.

Acta Paediatr

REVIEW

Should pulse c disease?

Pekka Valmari

Screenin oximetry

ANNE DE-
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Department of
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Abstract
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Introduction

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Received 1 December
ISSN 0803-5253 p
DOI: 10.1080/0803

Ten studies (44 969 new
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REGULAR ARTICLE

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

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Keywords
Critical congenital heart defects, Newborns, Pulse oximetry, Screening
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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 (SpO₂) was measured in apparently healthy newborns after transfer to the nursery, with SpO₂ < 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

(SpO₂) (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.

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) SpO₂ 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 (tachypnoea, cyanosis) when SpO₂ < 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 SpO₂ ≥ 95%, the infant was referred for a routine clinical examination. Infants transferred to a special

Abbreviations
CHD, congenital heart defect; CCHD, critical congenital heart defect; SpO₂, arterial oxygen saturation.

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BMJ

Impact of pulse oximetry screening on the detection of duct dependent congenital heart disease: a Swedish prospective screening study in 39 821 newborns

Table 2 | The performance of screening methods in the detection of duct dependent circulation in newborn infants in West Götaland (1 July 2004 to 31 March 2007)

Performance	Physical examination alone (n=38374)	Pulse oximetry (n=38429)	Physical examination plus pulse oximetry (n=38429)
Sensitivity (95% CI) (%)	62.50 (35.43 to 84.80)*	62.07 (42.3 to 79.31)	82.76 (64.23 to 94.15)
Specificity (95% CI) (%)	98.07 (97.93 to 98.21)	99.82 (99.77 to 99.86)	97.88 (97.73 to 98.03)
Positive predictive value (95% CI) (%)	1.35 (0.65 to 2.47)	20.69 (12.75 to 30.71)	2.92 (1.88 to 4.31)
Negative predictive value (95% CI) (%)	99.98 (99.96 to 99.99)	99.97 (99.95 to 99.99)	99.99 (99.97 to 100.00)

Table 3 | Pathology found in 69 babies with false positive results from pulse oximetry screening for duct dependent circulation in West Götaland (1 July 2004 to 31 March 2007)

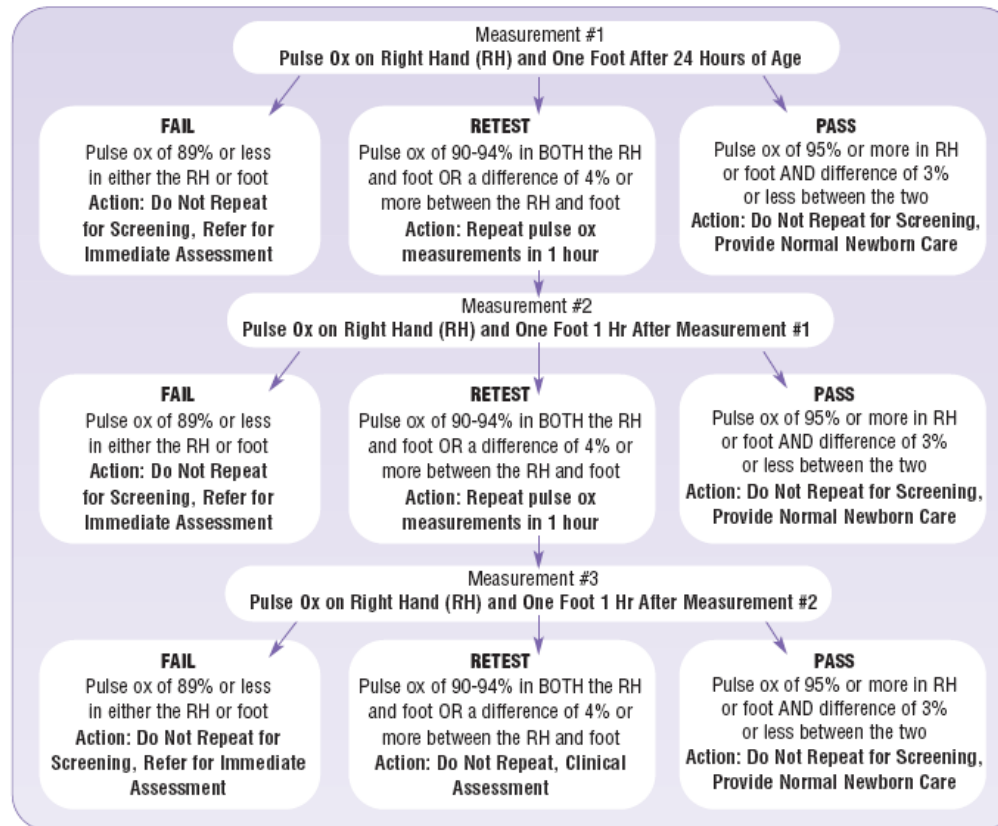
Pathology found	No (%) of babies	Subsequent management			
		Stay in neonatal intensive care		Follow-up only	Surgery
		≥5 days after screening	<5 after screening		
Other critical congenital heart disease*	4 (6)	4/4	0/4	0/4	4/4
Other milder congenital heart disease	10 (14)	4/10	1/10	5/10	4/10
Persistent pulmonary hypertension	6 (9)	3/6	0/6	3/6	N/A
Transitional circulation†	8 (12)	0/8	3/8	2/8	N/A
Infections	10 (14)	6/10	4/10	N/A	N/A
Pulmonary pathology	7 (10)	5/7	1/7	1/7	N/A
Normal (verified from hospital charts)	24 (35)	N/A	N/A	N/A	N/A

*Pulmonary atresia with multiple aorto-pulmonary collaterals (n=2), tricuspid atresia with pulmonary stenosis and ventricular septal defect (n=1), total anomalous pulmonary venous return (n=1).

†Right to left shunting across foramen ovale without pulmonary hypertension.

United States Efforts

Strategies for Implementing Screening for Critical Congenital Heart Disease



REGULAR ARTICLE

Nordic pulse oximetry screening – implementation status and proposal for uniform guidelines

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5.Department of Paediatric Cardiology, Children's Hospital Reykjavik, Landspítali University Hospital, Reykjavik, Iceland

- Norway, Sweden, Poland and Ireland have national recommendations to screen
- Identical screening protocol as used in U.S.
- **Sweden, Norway and Finland used bottom up approach - implementation near 100%**
- Significantly lower screening implementation rates
 - Denmark – claim high prenatal detection rates
 - Iceland - claim lowest infant mortality in Europe



BWH screening programme 2010-2013



(40 months)

- Total Livebirths: 25,859
- Most babies screened >12 hrs (mean age 7 hrs)
- Test positive pulse oximetry: 208
0.8% of all livebirths - Just >1 admission a week

Congenital heart defects identified: 17

- Critical CHD: 9
- Serious CHD: 3
- Significant CHD: 5

2 CCHD missed by all screening procedures

Singh, Rasiah, Ewer Arch Dis Child FN 2014;99:F297-F302.

Slide courtesy of Andrew Ewer, MD



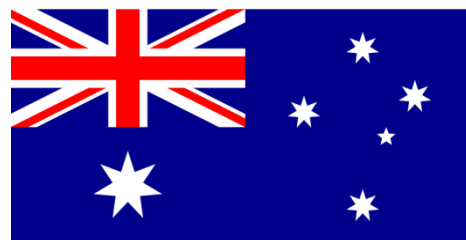
Results

- Transitional circulation/mild TTN: 43 (21%)
- No collapse in the postnatal wards
- 165/208 (79%) identified significant clinical condition
- Echos performed for test +ve pulse Ox:
61/208 (29%)
- Abnormal Echos: 29/61 (48%)

Singh, Rasiah, Ewer Arch Dis Child FN 2014;99:F297-F302.

Slide courtesy of Andrew Ewer, MD

Impact on Pediatric Cardiologist, Echocardiography and Resource Utilization



ORIGINAL ARTICLE

Post-implementation review of pulse oximetry screening of well newborns in an Australian tertiary maternity hospital

Kavita Bhola,¹ Martin Kluckow^{2,3} and Nick Evans^{1,3}

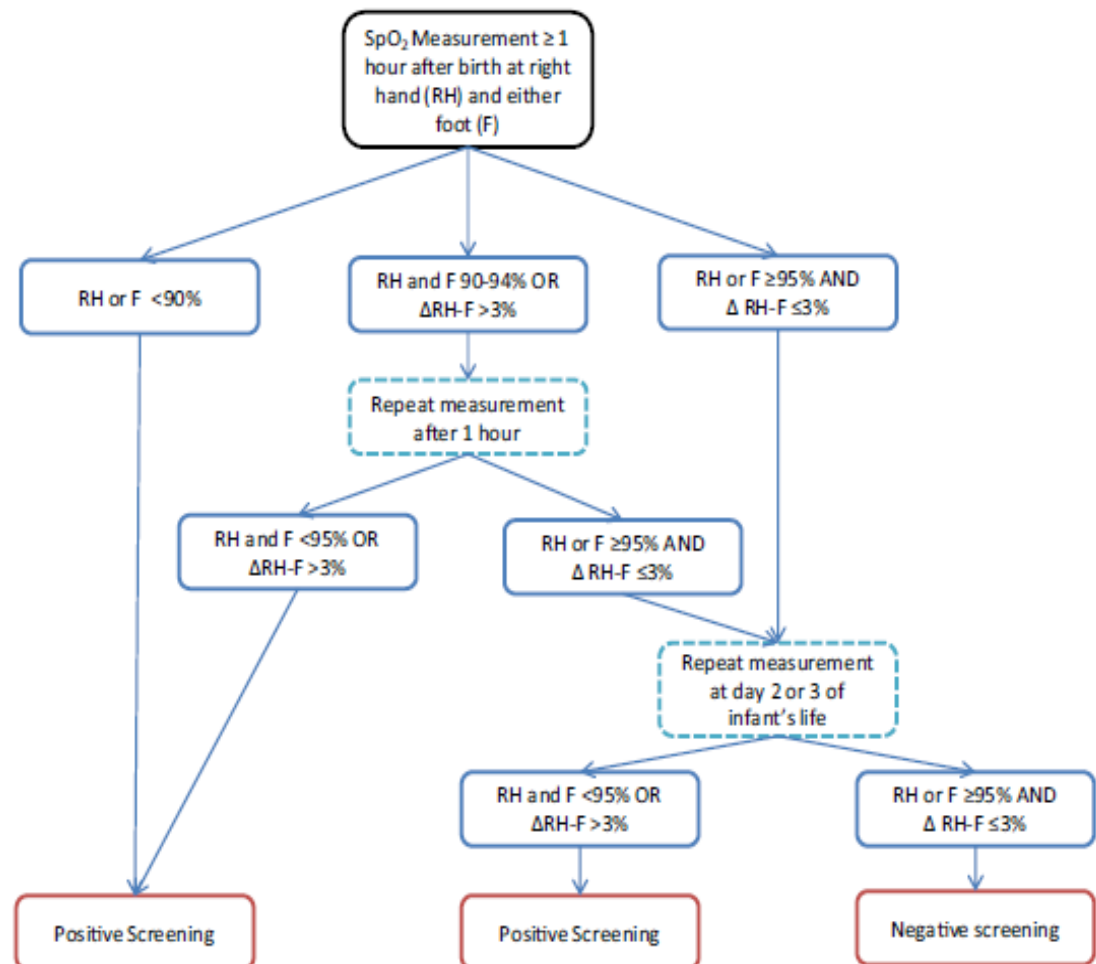
¹Department of Newborn Care, Royal Prince Alfred Hospital, ²Department of Neonatology, Royal North Shore Hospital, ³Sydney Medical School, University of Sydney, Sydney, New South Wales, Australia

- 18,801 infants screened; FP rate 0.13%; sensitivity 80%
- Only 9 echos
- No significant burden on echocardiography resources
- Equally effective tool for improving early diagnosis of other important pathologies –
 - 4 CCHD (TGA, AS, TAPVD, PS)
 - 6 with secondary targets (PPHN, PNE, congenital diaphragmatic hernia, alveolar capillary dysplasia)

Dutch Pilot Study - Home Birth Algorithm

Fig. 1 Flowchart of pulse oximetry screening. The decision tree of the protocol is shown

Narayan Eur J Pediatr 2014



China: Bigger Numbers, Similar Findings

Pulse oximetry with clinical assessment to screen for congenital heart disease in neonates in China: a prospective study

Qu-ming Zhao*, Xiao-jing Ma*, Xiao-ling Ge, Fang Liu, Wei-Ji Yan, Lin Wu, Ming Ye, Xue-cun Liang, Jing Zhang, Yan Gao, Bing Jia†, Guo-ying Huang†, and the Neonatal Congenital Heart Disease screening group‡

	N	Detection rate		
		Pulse oximetry alone	Clinical assessment alone	Pulse oximetry plus clinical assessment
Critical pulmonary stenosis	10	10 (100%)	10 (100%)	10 (100%)
Tetralogy of Fallot	9	9 (100%)	9 (100%)	9 (100%)
Truncus arteriosus	5	2 (40%)	3 (60%)	4 (80%)
Single ventricle	11	8 (73%)	9 (82%)	10 (91%)
Pulmonary atresia	30	30 (100%)	28 (93%)	30 (100%)
Transposition of great arteries	33	32 (97%)	29 (88%)	32 (97%)
Double outlet of right ventricle	9	8 (89%)	6 (67%)	9 (100%)
Hypoplastic left heart syndrome	7	3 (43%)	2 (29%)	4 (57%)
Critical coarctation of the aorta	7	3 (43%)	4 (57%)	5 (71%)
Interrupted aortic arch	5	2 (40%)	2 (40%)	4 (80%)
Critical aortic stenosis	3	1 (33%)	3 (100%)	3 (100%)
Total anomalous pulmonary venous connection	17	14 (82%)	8 (47%)	16 (94%)
Total	146	84% (122 of 146)	77% (113 of 146)	93% (136 of 146)

Table 3: Detection rate for individual critical congenital heart disease in asymptomatic newborn babies



Comment to Zhao Article

- Optimum timing of screening deserves further consideration:
- U.S. screens 24 hours - lower false positive rate (0.04%) but fewer cases of CCHD identified (NJ 2013 article)
- Need to balance lower false positive rate against likelihood of timely diagnosis (prevent collapse prior to screen)
- Less than 1/3 of positive infants need an echo (if assess for secondary targets first)

Ewer Lancet April 2014

ORIGINAL ARTICLE

Early screening for critical congenital heart defects in asymptomatic newborns in Mazovia province: experience of the POLKARD pulse oximetry programme 2006–2008 in Poland

Anna Turska-Kmieć¹, Maria Katarzyna Borszewska-Kornacka², Witold Błaż³,
Wanda Kawalec¹, Małgorzata Żuk¹

- 51,698 asymptomatic infants screened from 51 neonatal units (14.2% of total births in Poland)
- 15 cases of CCHD identified by P.O.; 4 false negatives
- Sensitivity – 78.9%; specificity – 99.9%

Research Article

DOI: 10.5455/2320-6012.ijrms20140894

Effectiveness of pulse oximetry screening for congenital heart disease in asymptomatic new-borns

Audil Mohmad Lanker¹, Javed Chowdhary¹, Nasir Jeelani¹,
Shazia Jeelani², Ashfaq Ul Hassan^{3*}, Nasir-ud-din Wani⁴

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Accepted: 20 July 2014

- Post-ductal only; all babies echocardiogram
- 1 in 600 asymptomatic newborns identified with CCHD thru P.O.

Table 1: Study group characteristics.

Characteristics	
Male	638 (53.2%)
Female	562 (46.8%)
Sex ratio	881
Rural population	60.3%
Urban population	39.7%
History of consanguinity	4.4% of new-borns.
Family history of CHD	11 new-borns (0.9%)
Median gestational age of the study group	39-40 weeks.
Mean age at screening	>24 hours
Normal cut-off value for post-ductal saturation	>95%

Table 3: Statistical parameters obtained in the study group.

Statistical parameters	
True positive	2
False positive	1
False positive rate	0.08
False negative	1
True negative	1196
Positive predictive value	66.67%
Negative predictive value	99.90%
Sensitivity	66.67%
Specificity	99.90%

Rome, Italy – Two Year Observational Cohort Study



- 5,750 asymptomatic babies; post-ductal only
- P.O screen conducted 48-72 hours
(after a physical exam at 24 hours)
- No true positives; 1 false negative CCHD
(aortic arch coarctation undetected by
both clinical assessment and P.O.)

Zuppa J Mat-Fet& Neo Med 2014



Congenital Heart Disease Screening Program: Health Authority of Abu Dhabi

New screening saves 13 babies in Abu Dhabi

Implementation Began:
January, 2011

Infants Screened:
Approx. 23,000

near 80,000

Total Detected:
13 with CCHD Detected

now 34 detected

SIMPLE TEST IS NOW MANDATORY IN ALL HOSPITALS IN THE CAPITAL

ABU DHABI

By SAMIHAH ZAMAN
Staff Reporter

A critical shortage of paediatric cardiologists in the emirate of Abu Dhabi is putting the lives of hundreds of babies at risk, especially if a mandatory test to detect serious congenital heart abnormalities is not administered, health experts have said.

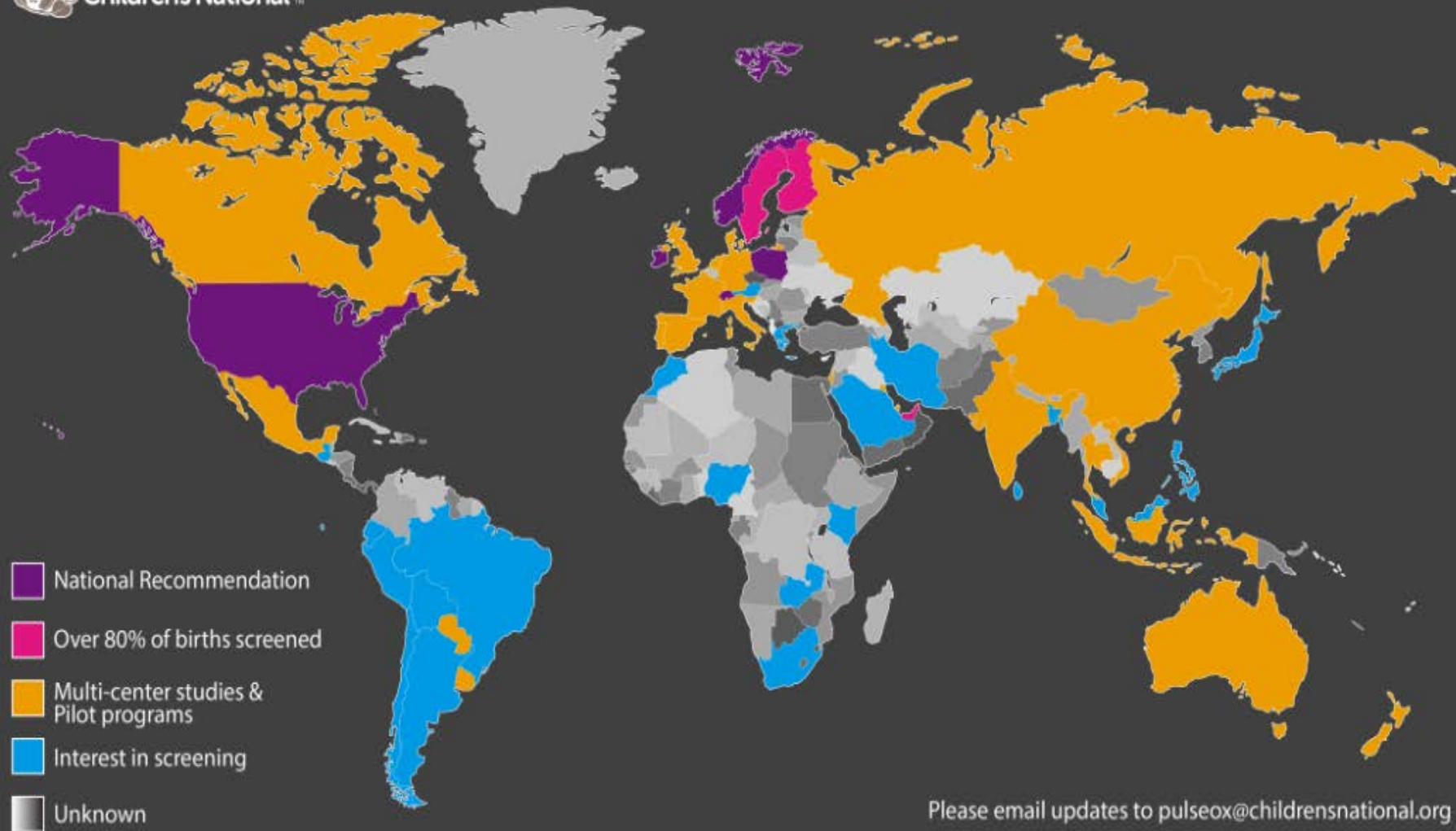
The test, which screens for critical congenital heart disease, affecting three in every 1,000 babies worldwide, is important because babies in the GCC could present certain abnormalities that can kill them within the first two months



Abdul Rahman/Gulf News

Happy and healthy

■ Dr Gerard Martin of the Children's National Medical Centre, with 8-month-old Jumana and her father Surour Khamis Abdullah.



European Efforts: Strategizing for a Uniform Recommendation Torino, Italy 2013 & 2014

THE LANCET

The Lancet, [Volume 382, Issue 9895](#), Pages 856 - 857, 7 September 2013

- France
- Germany
- Italy
- Netherlands
- Spain
- Sweden
- UK



Press release

**UK National Screening Committee
recommends new test for newborn
babies with heart disease**

European Union – Committee of Experts on Rare Diseases EUCERDS

Identified opportunity for improvements in newborn screening

- 2013 Commission Expert Group on Rare Diseases
- Centralized approach - benefits of standardization, registries, COE
- Documented wide practice variation - 2 to 29 disorders
- Prioritizing metabolic and genetic disorders
- Opportunity to consider congenital cardiac defects



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