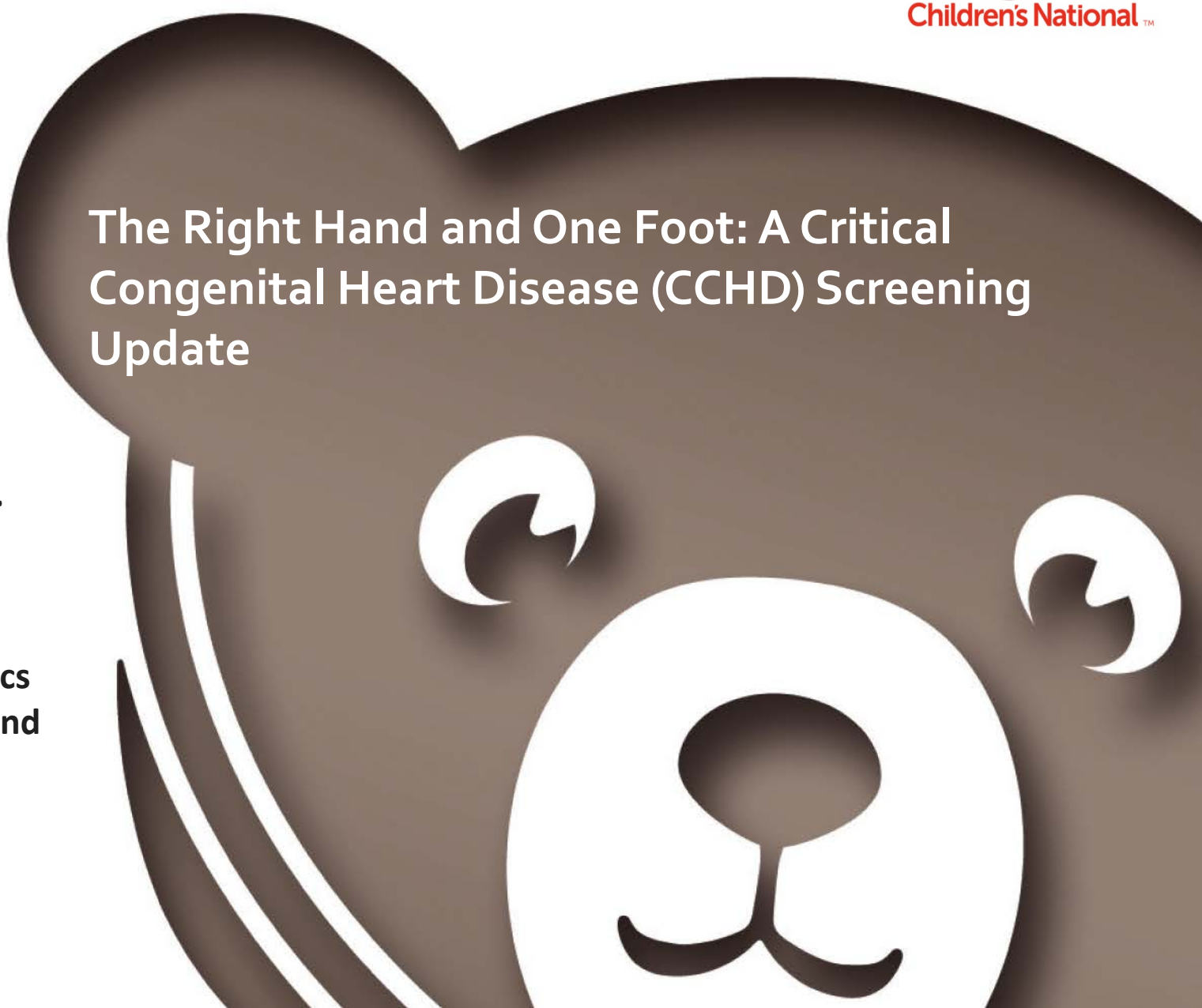
The Right Hand and One Foot: A Critical Congenital Heart Disease (CCHD) Screening Update

Lisa Hom, RN Esq.

Lowell Frank, MD

Future of Pediatrics
Bethesda, Maryland
June 4, 2014



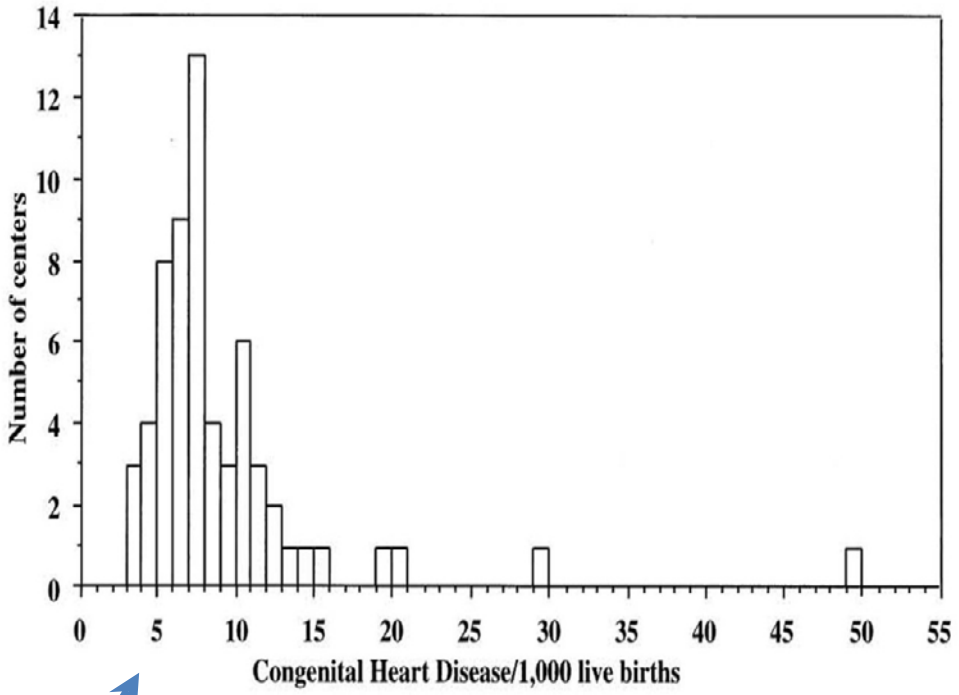
Children's National

Heart Institute

Part of the Children's National Health System

Congenital Heart Disease

- ♥ Most common birth defect
- ♥ 8:1,000 with CHD
  3:1,000 with CCHD
- ♥ Accounts for ~ 40% deaths from congenital anomalies
- ♥ Majority of deaths due to CHD occur in first year of life



Hoffman JACC 39:2002

Children's National
Heart Institute
Part of the Children's National Health System

Perspective on Importance

Missed Diagnosis of Critical Congenital Heart Disease

Ruey-Kang R. Chang, MD, MPH; Michelle Gurvitz, MD; Sandra Rodriguez, MS

- 15 year retrospective study
- 898 infants died of CCHD in infancy
  152 with missed diagnosis
  299 with late diagnosis
- >50% of CCHD deaths were attributed to late/missed diagnosis
- 30 babies died each year secondary to late diagnosis

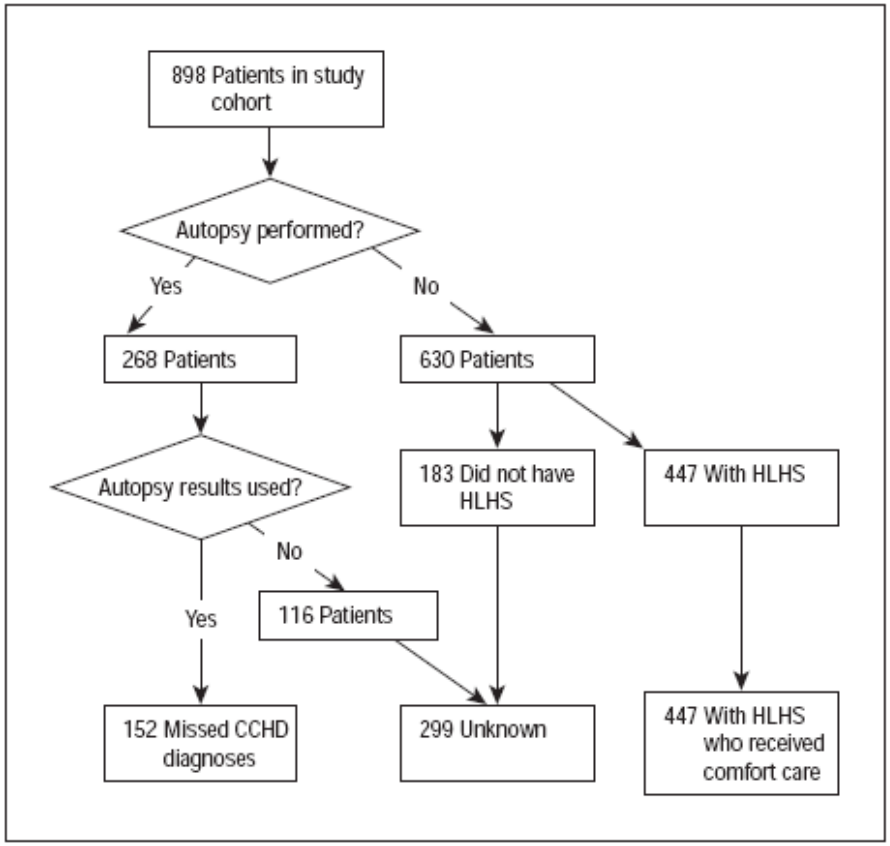
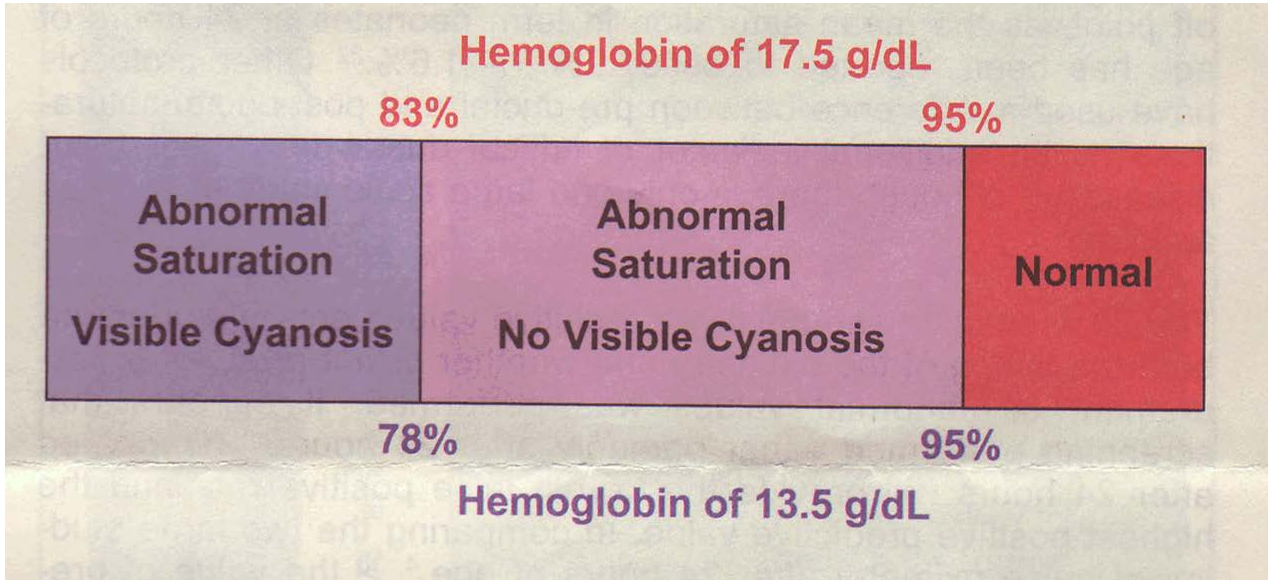


Figure 1. Selection and identification of patients with missed and unknown critical congenital heart disease (CCHD) diagnoses. HLHS indicates hypoplastic left heart syndrome.

Children's National
Heart Institute
Part of the Children's National Health System

Why is Detecting Newborns with CCHD Difficult?

- Complex changes from fetal to normal circulation after birth
- Fetal Ultrasound
- Detection through physical examination may be < 50%

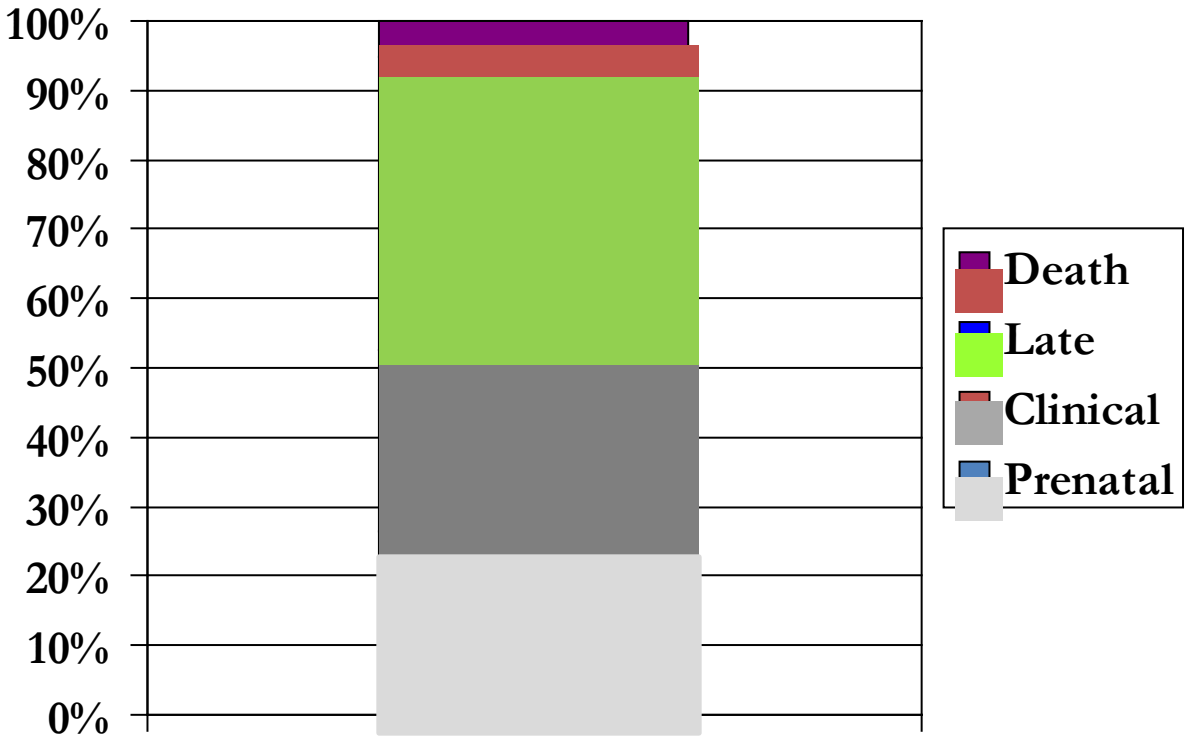


Cyanotic "Blind Spot"

Mean threshold for detection 69%!

Children's National
Heart Institute
Part of the Children's National Health System

Diagnostic Gap



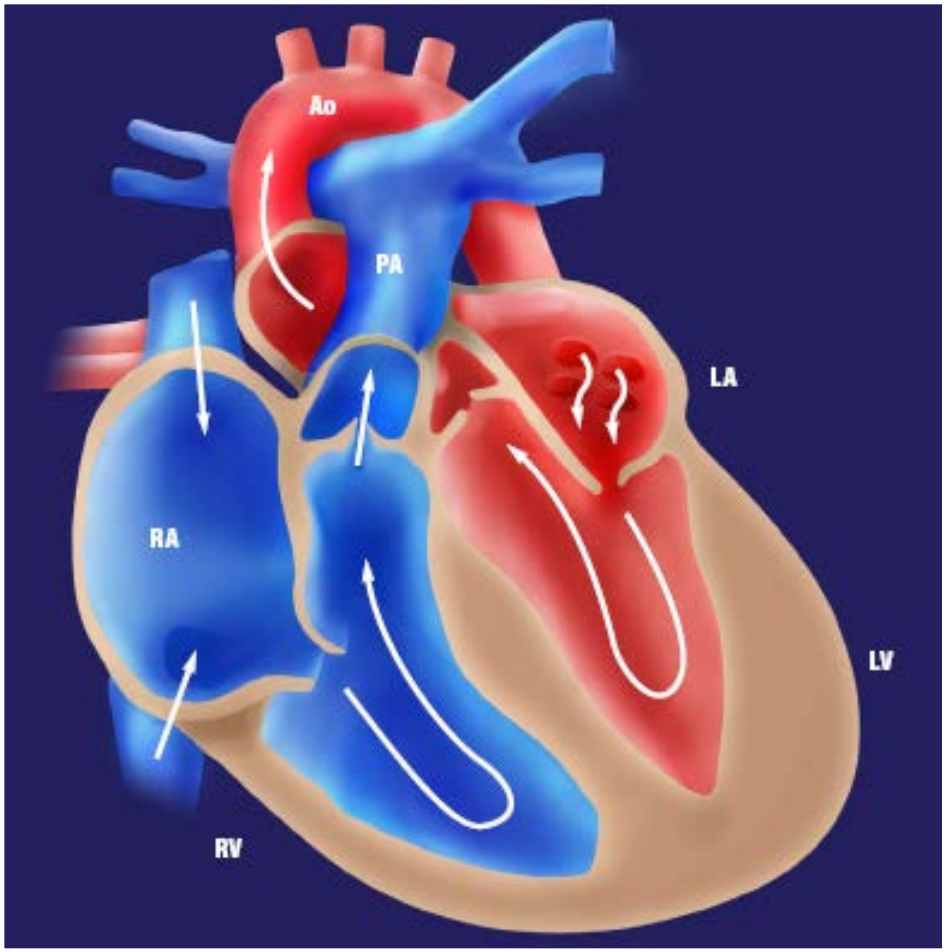
Children's National
Heart Institute
Part of the Children's National Health System

Pulse Oximetry as a Screening Method

- ♥ Pulse oximetry measures oxygen saturation of hemoglobin in arterial blood
- ♥ Non-invasive and painless test

Children's National
Heart Institute
Part of the Children's National Health System

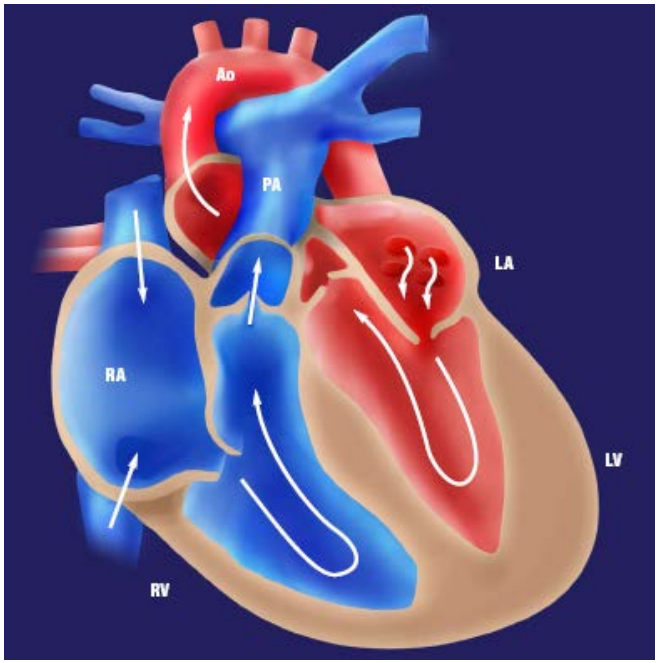
Normal Newborn Circulation

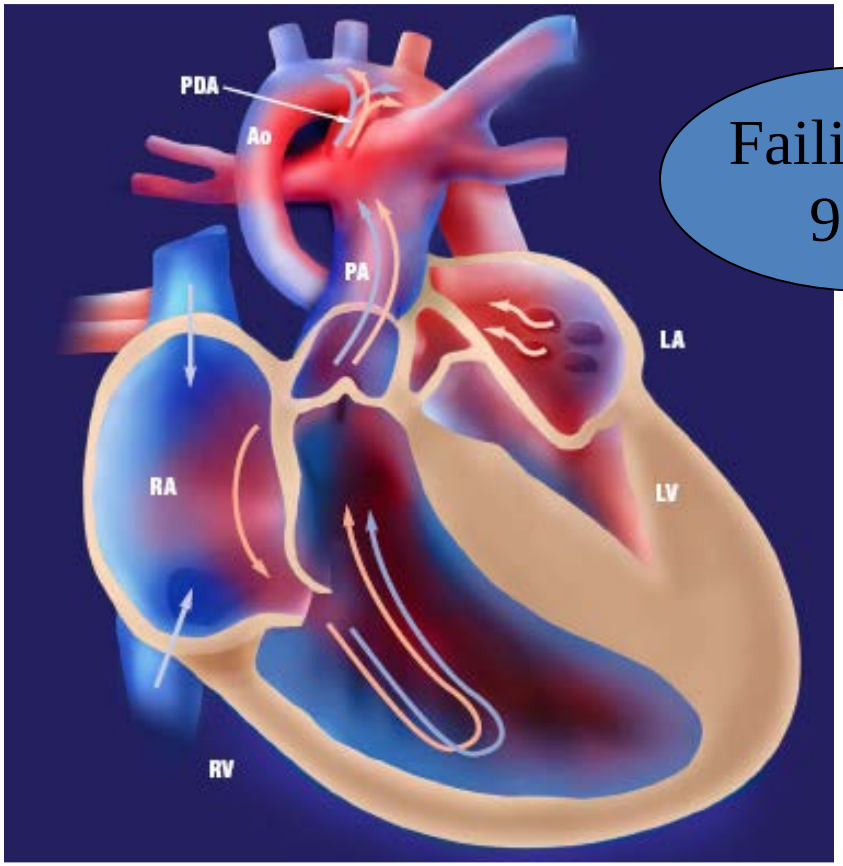


Passing Sat 100%

Children's National
Heart Institute
Part of the Children's National Health System

Hypoplastic Left Heart Syndrome





Failing Sat 90%

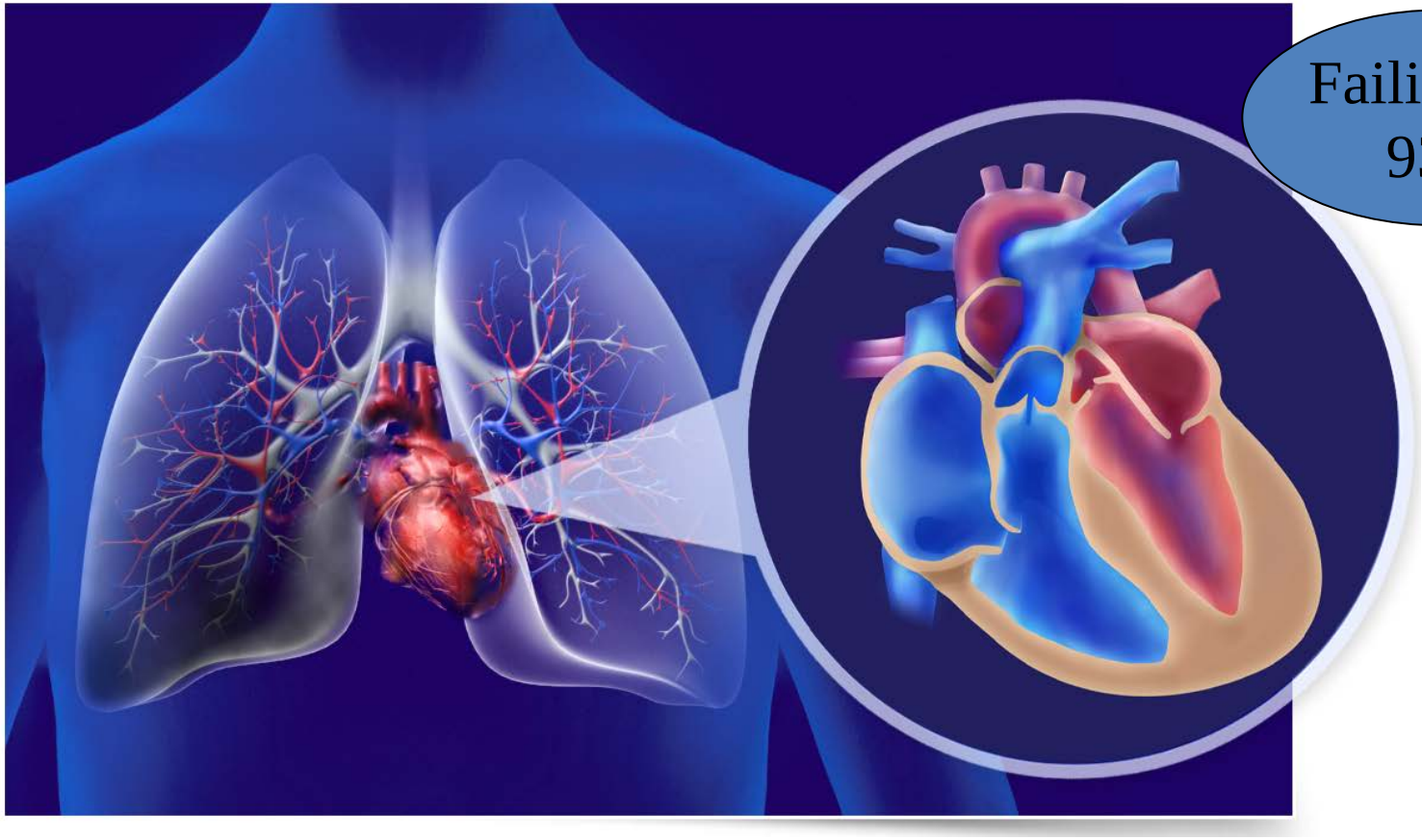
Children's National
Heart Institute
Part of the Children's National Health System

CCHD Screening Primary Targets

1. Hypoplastic Left Heart Syndrome
2. Pulmonary Atresia (with intact septum)
3. Tetralogy of Fallot
4. Total Anomalous Pulmonary Venous Return
5. Transposition of the Great Arteries
6. Tricuspid Atresia
7. Truncus Arteriosus

Children's National
Heart Institute
Part of the Children's National Health System

Secondary Target: Pneumonia



Children's National
Heart Institute
Part of the Children's National Health System

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)

		Subsequent management			
		Stay in neonatal intensive care			
Pathology found	No (%) of babies	≥5 days after screening	<5 after screening	Follow-up only	Surgery
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.

Granelli BMJ 338:2009

Children's National
Heart Institute
Part of the Children's National Health System

Is Pulse Oximetry Effective in Detecting CCHD?

-13 primary studies

- 229,421 infants screened

Pulse oximetry screening for critical congenital heart defects in asymptomatic newborn babies: a systematic review and meta-analysis

Shakila Thangaratinam, Kiritrea Brown, Javier Zamora, Khalid S Khan, Andrew K Ewer

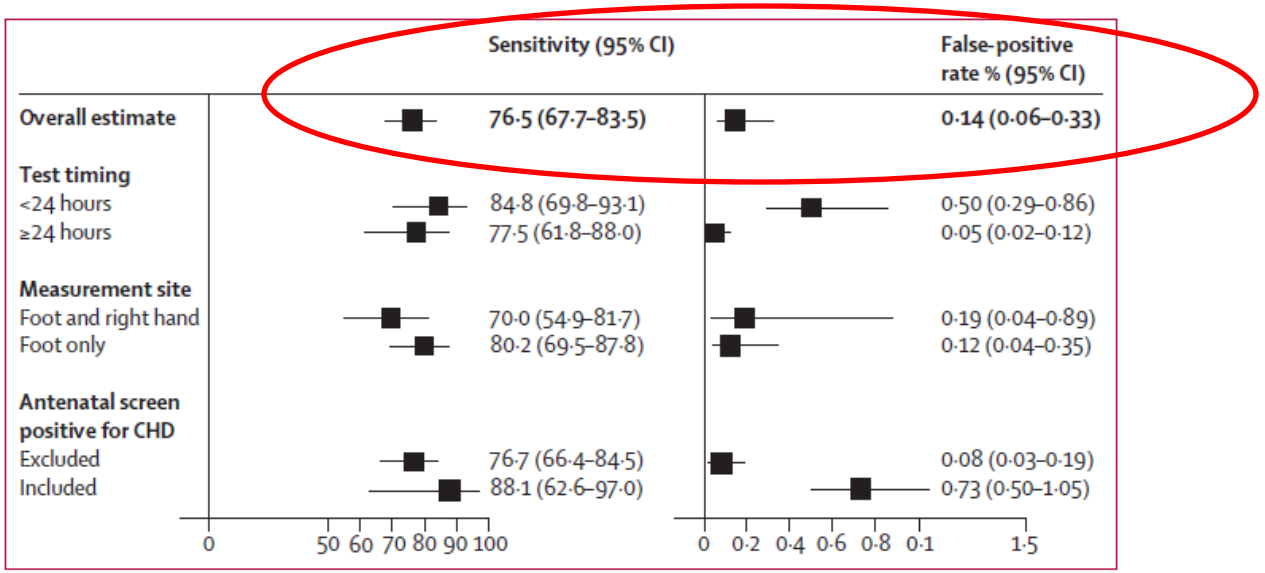
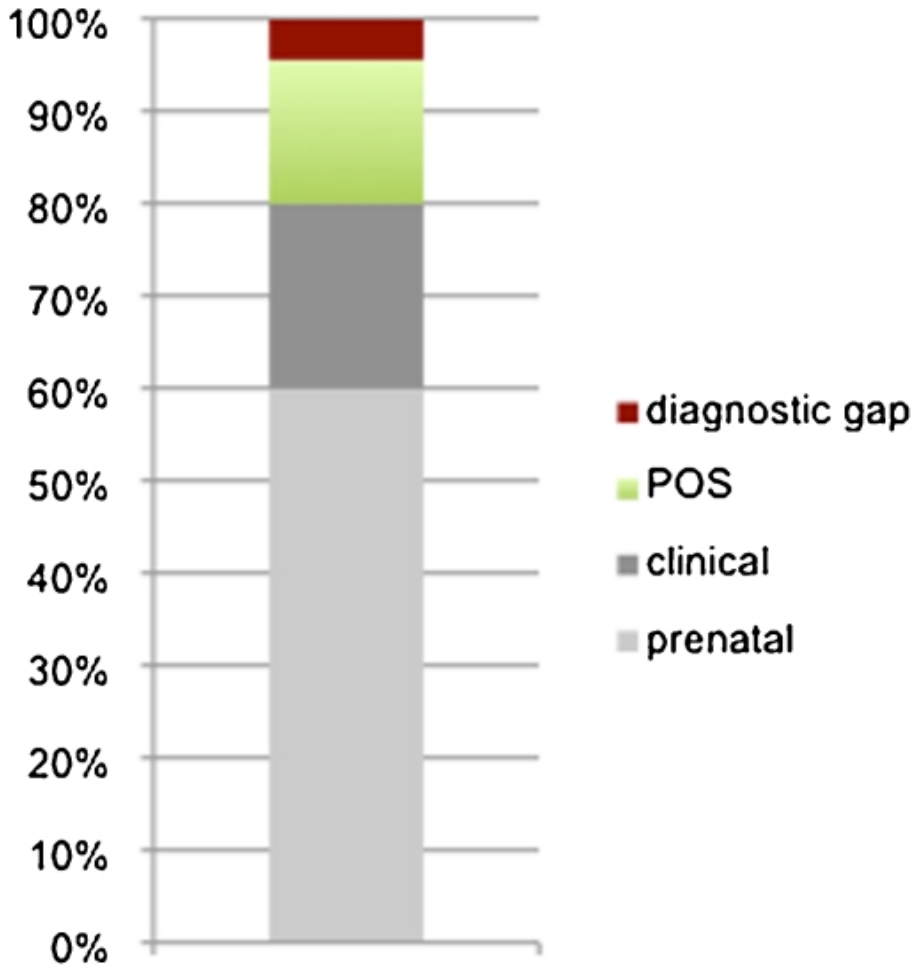


Figure 3: Accuracy estimates based on clinical and test characteristics of pulse oximetry in detection of critical congenital heart defects in newborn babies
CHD=congenital heart defects.

Interpretation Pulse oximetry is highly specific for detection of critical congenital heart defects with moderate sensitivity, that meets criteria for universal screening.

Children's National
Heart Institute
Part of the Children's National Health System

Research on Populations



- 42,240 infants in 34 German hospitals
- Sensitivity 78%
- Specificity 99%
- PPV 26% NPV 99%

Riede Eur J Peds 169:2010

Children's National
Heart Institute
Part of the Children's National Health System

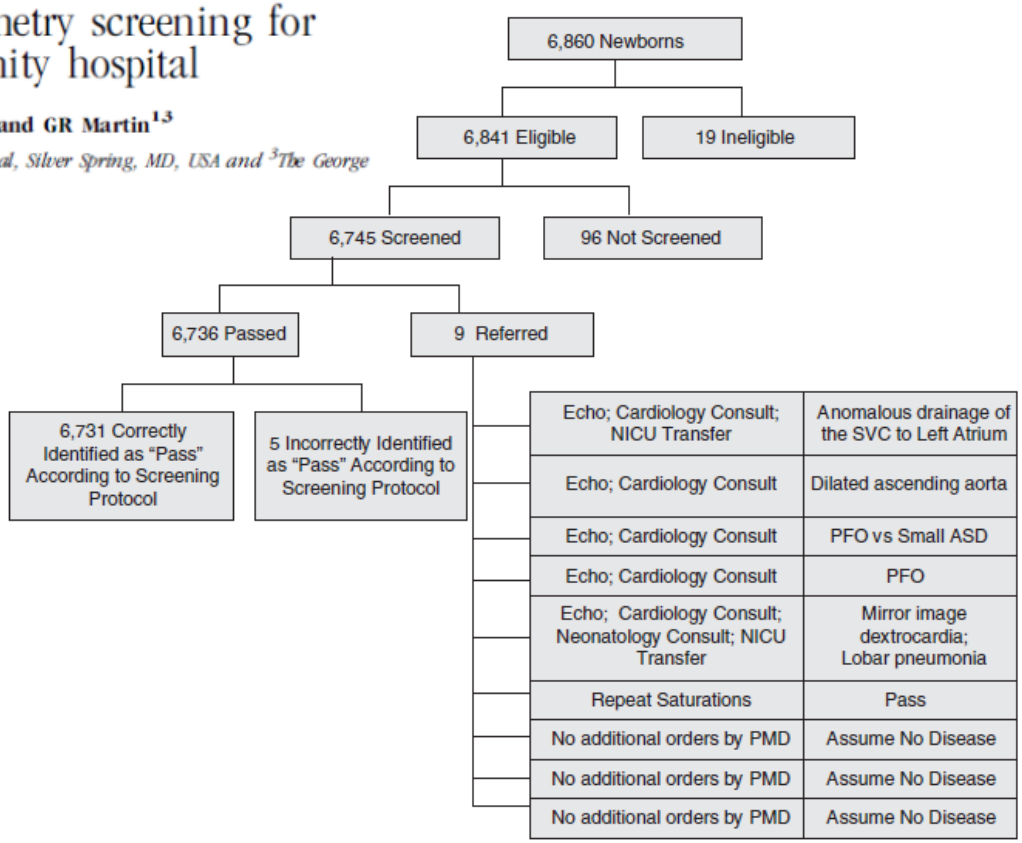
CCHD Screening Feasibility in Community Hospitals

ORIGINAL ARTICLE

Feasibility of implementing pulse oximetry screening for congenital heart disease in a community hospital

EA Bradshaw[1], S Cuzzi[1,2,3], SC Kiernan[2], N Nagel[2], JA Becker[1,3] and GR Martin[1,3]

[1]Children's National Medical Center, Washington, DC, USA; [2]Holy Cross Hospital, Silver Spring, MD, USA and [3]The George Washington University School of Medicine, Washington, DC, USA



- Avg. Pox Sat

  Rt Hand/Foot
  100% (90-100%)

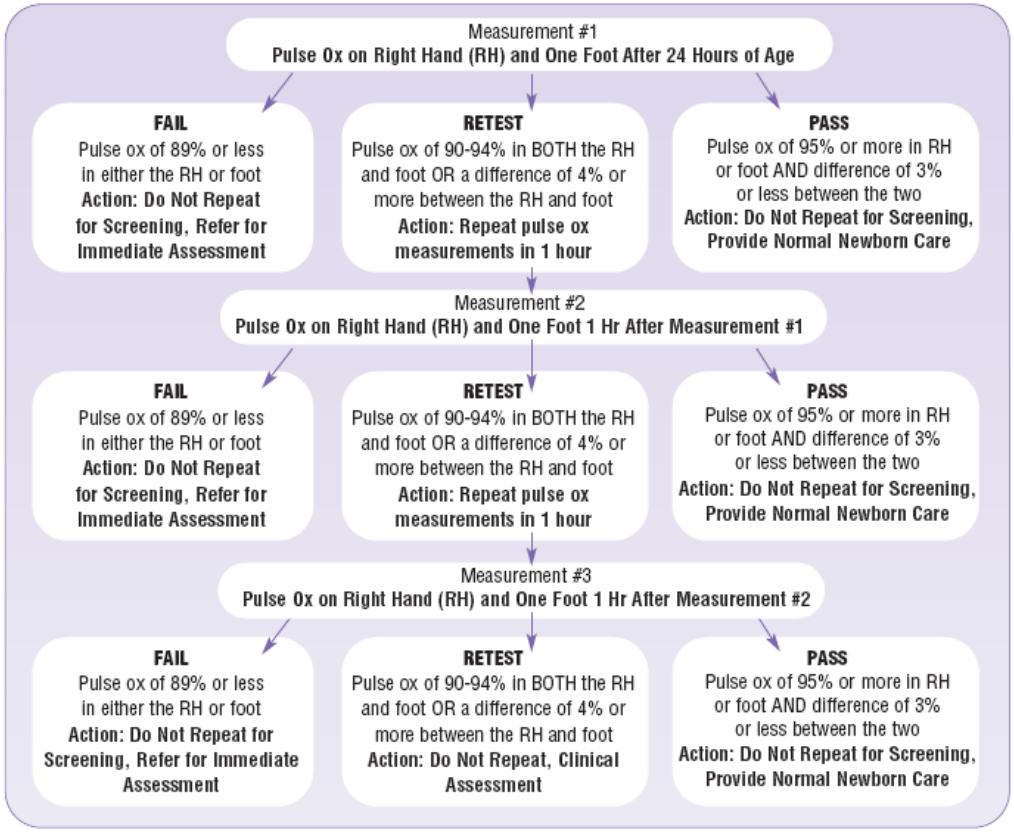
  Difference
  0.2% (0-6%)

-CCHD screening did not lead to a significant increase in echos

Children's National
Heart Institute
Part of the Children's National Health System

United States Efforts

Strategies for Implementing Screening for Critical Congenital Heart Disease



Children's National
Heart Institute
Part of the Children's National Health System

RH Application Sites

RH Application Sites

REMINDER ALGORITHM FOR SCREENERS

- **C**onfirm that the infant is at least 24 hours of age and eligible for screening.
- **H**elp the parent to warm and calm the infant in a quiet and peaceful environment.
- **D**escribe the pulse ox test to the parent.
- **S**elect a site on the right hand and one foot that is clean and dry.
- **P**lace the pulse ox sensor and perform the pulse ox test.

Children's National™

Heart Institute

Part of the Children's National Health System

Management of Failed CCHD Screen

Cardiac Evaluation:

Physical Examination

Pulse Ox

Perfusion Check

- BP and Pulses x 4 extremities

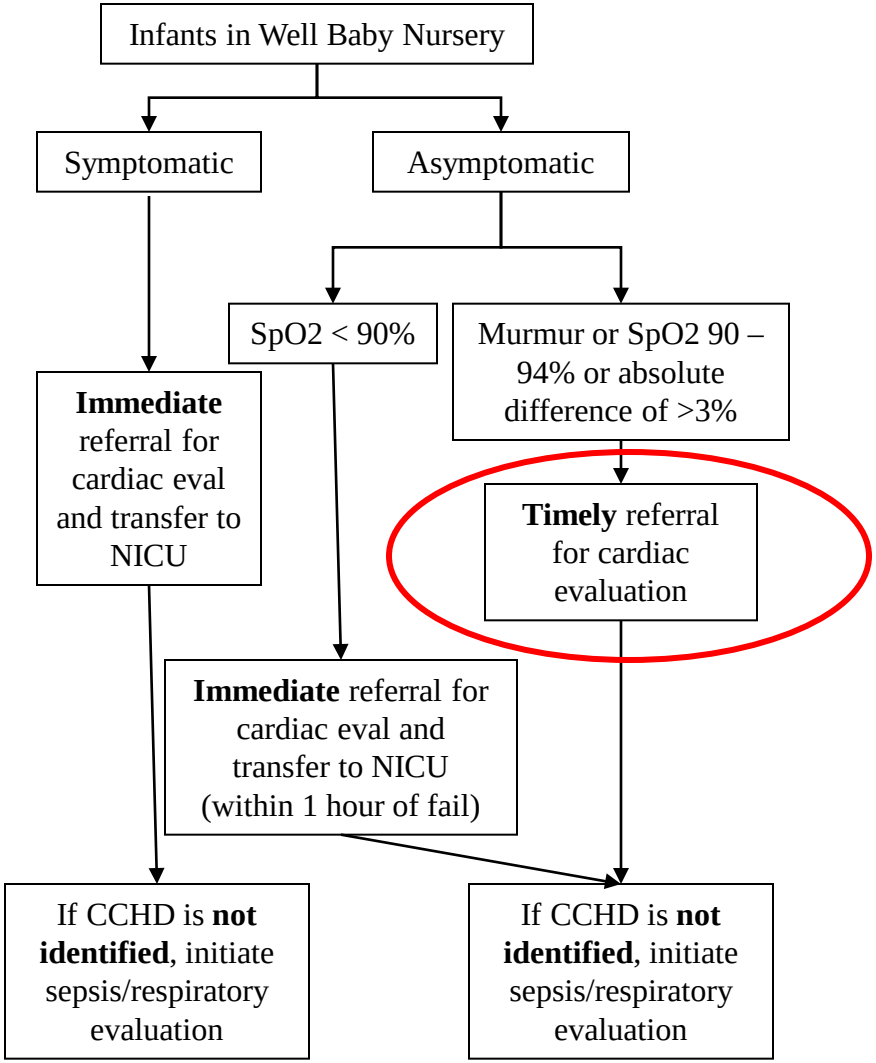
ABG

ECHO

EKG

If CCHD is Identified:

Initiate appropriate therapy and, if necessary, arrange transfer.



Sepsis and Respiratory Evaluation:

Physical Examination

Rectal Temperature

Blood Culture

CBC with Differential

Chest X-Ray

C-Reactive Protein

Blood Glucose

Lumbar Puncture

Children's National

Heart Institute

Part of the Children's National Health System

CCHD Screening Using Pulse Oximetry: National and Global Implementation

Children's National

Heart Institute

Part of the Children's National Health System

United States Efforts

THE SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

September 21, 2011

R. Rodney Howell, M.D.
Committee Chairperson
Secretary's Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane, Room 18A19
Rockville, MD 20857

Dear Dr. Howell:

As indicated in my letter to you on April 20, 2011, I determined that the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children's (SACHDNC) recommendations pertaining to the addition of Critical Congenital Heart Disease (CCHD) screening to the Recommended Uniform Screening Panel (RUSP) were not yet ready for adoption. Consequently, I referred the SACHDNC's recommendations to the Interagency Coordinating Committee on Screening in Newborns and Children (ICC) for additional review and input regarding implementation. I asked the ICC to review the evidence gaps described by the SACHDNC and propose a plan of action to address: identification of effective screening technologies, development of diagnostic processes and protocols, education of providers and the public, and strengthening service infrastructure needs for follow-up and surveillance. I have received and reviewed the requested ICC Plan of Action.

As you know, congenital heart disease causes up to 3% of all infant deaths in the first year of life. Heart defects affect about 7 to 9 of every 1000 live births, one quarter of which could be detected and potentially treated by measuring blood oxygen saturation. Given this reality and the available information on the effectiveness of screening, I have decided to adopt the SACHDNC's first recommendation to add CCHD to the RUSP. In addition, I am requesting that the SACHDNC collaborate with the Health Resources and Services Administration (HRSA) to complete a thorough evaluation of the potential public health impact of universal screening for CCHD, as required by the authorizing statute, section 1111 of the Public Health Service Act (42 U.S.C. § 300b-10(b)(4)).

Children's National
Heart Institute
Part of the Children's National Health System

United States Efforts

Maryland Bill Signing May 19, 2011

- 2011: Indiana and Maryland first states to pass CCHD screening legislation. New Jersey first state to implement universal CCHD screening.

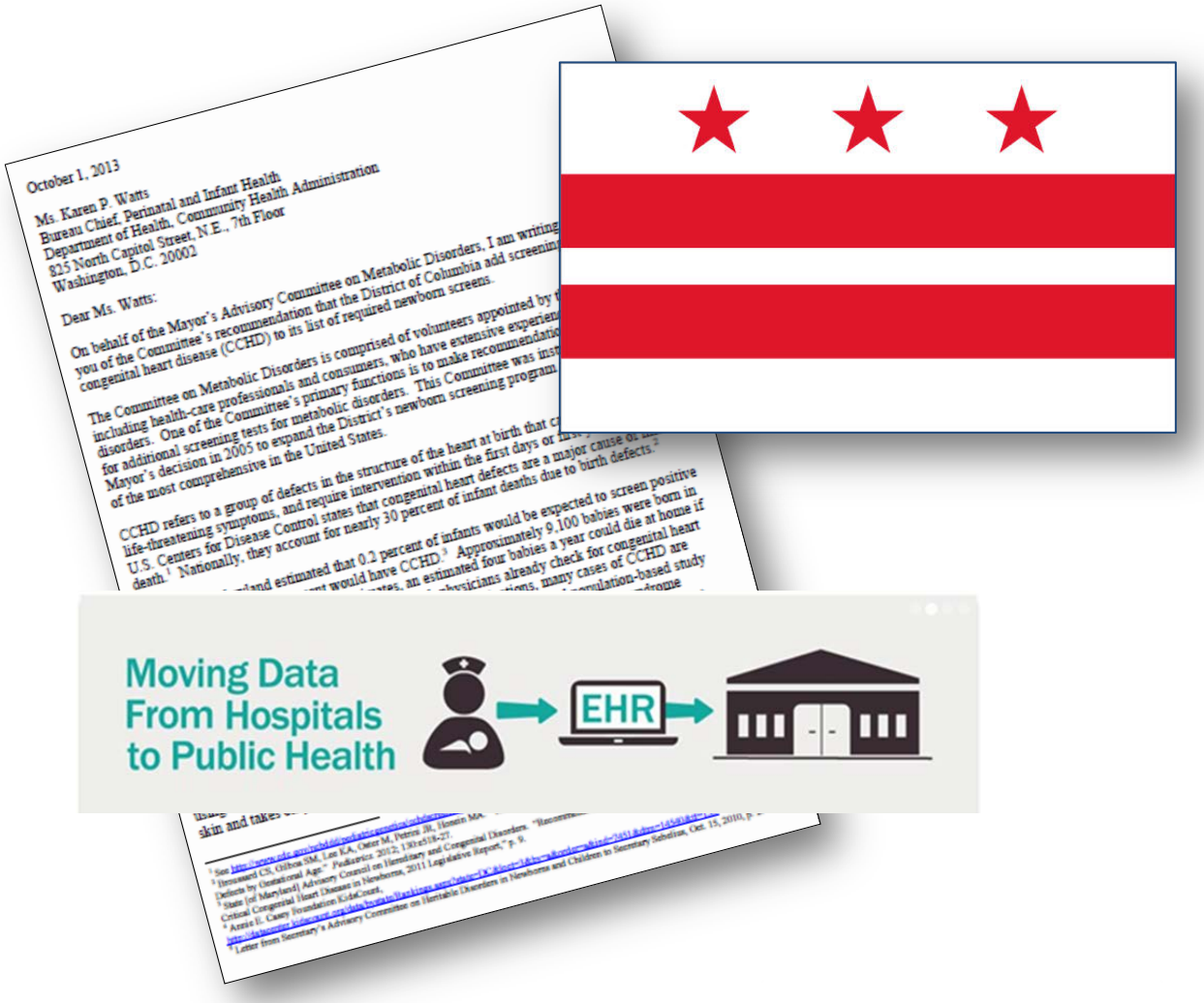
- 20 states enacted legislation in the first half of 2013 (rolling implementation dates during 2014 – over 80% of births screened by end of 2014)

- 36 states total with legislation pending in many more

Children's National
Heart Institute
Part of the Children's National Health System

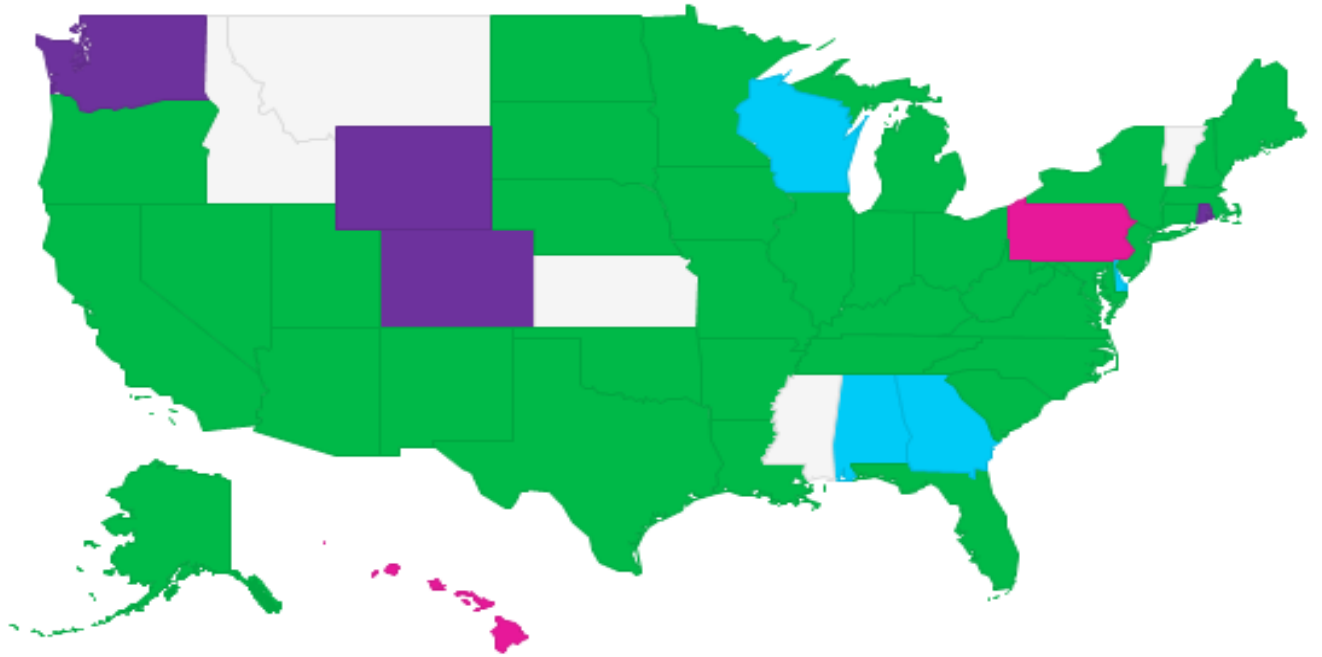
Working Towards a Mandate for the District of Columbia

- Recommendation filed by the Mayor's Advisory Committee, October 2013
- Support letters from all 7 hospitals, a parent advocate, March of Dimes and Children's National
- Follow up with Department of Health through rulemaking process



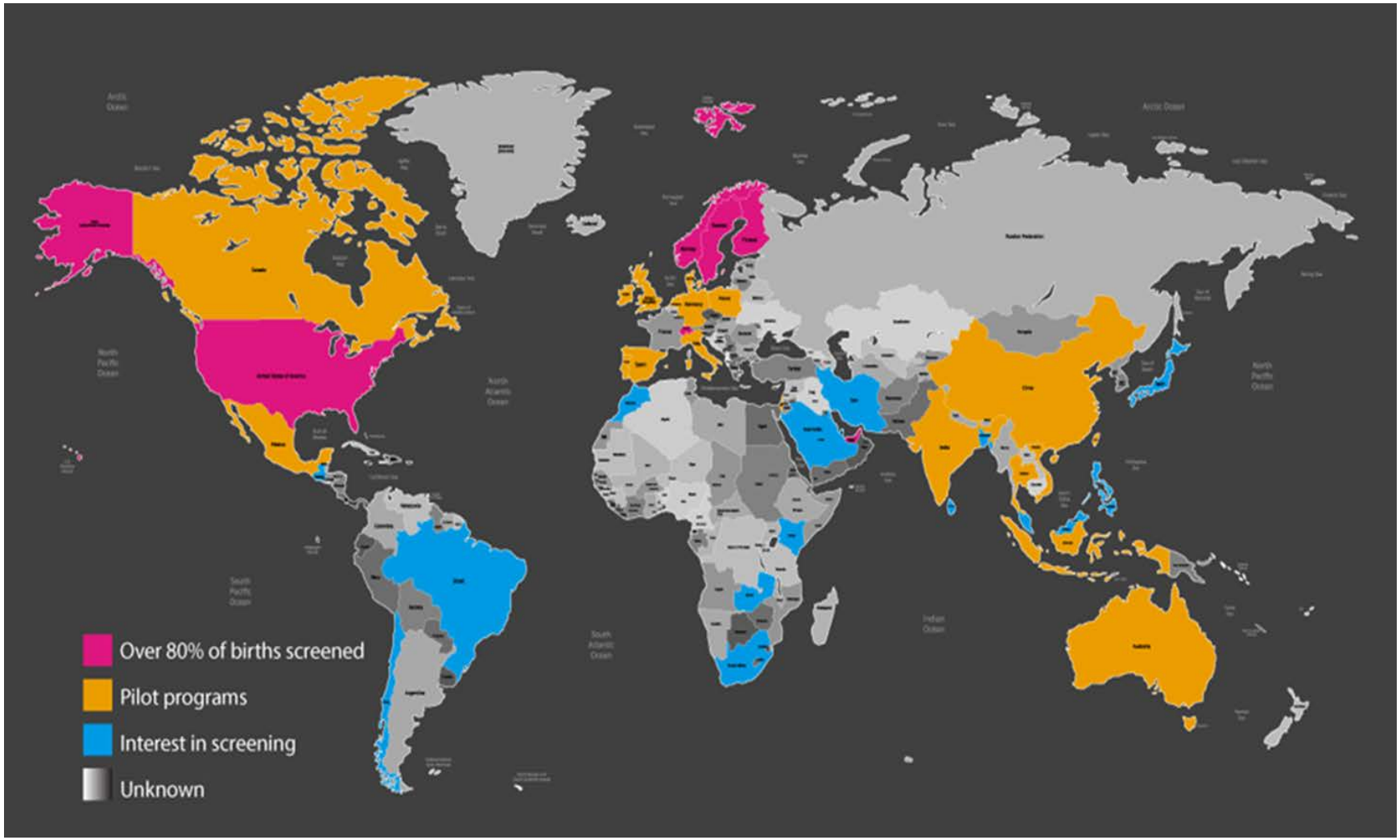
Children's National
Heart Institute
Part of the Children's National Health System

United States Efforts 2014



Active Legislation | Legislation Enacted | Regulatory Addition to NBS Panel | Multi-Center Screening or Pilot Project

Children's National
Heart Institute
Part of the Children's National Health System



Children's National

Heart Institute

Part of the Children's National Health System

European Efforts: Strategizing for a Uniform Recommendation

THE LANCET

The Lancet, Volume 382, Issue 9895, Pages 856 - 857, 7 September 2013

Pulse oximetry screening for congenital heart defects

*Andrew K Ewer, Anne De-Wahl Granelli, Paolo Manzoni, Manuel Sánchez Luna, Gerard R Martin

a.k.ewer@bham.ac.uk

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

Children's National

Heart Institute

Part of the Children's National Health System

Right is Right.

Children's National
Heart Institute
Part of the Children's National Health System

CCHD Screening Using Pulse Oximetry: Next Hurdle - False Negatives

Children's National

Heart Institute

Part of the Children's National Health System

Regional False Negative Surveillance

Maryland CCHD Advisory Council Initiative

Working with:

- Children's National
- INOVA Health System
- Georgetown University Medical Center

To evaluate why the infant was not identified on birth screening

STATE OF MARYLAND

DHMH

Maryland Department of Health and Mental Hygiene

Martin O'Malley, Governor – Anthony G. Brown, Lt. Governor – Joshua M. Sharfstein M.D., Secretary

Prevention and Health Promotion Administration

Michelle Spencer, MS, Director
Donna Gugel, MHS, Deputy Director

Ilise D. Marrazzo, RN, BSN, MPH, Director, Maternal and Child Health Bureau
Deborah B. McGruder, MPH, PMP, Director, Infectious Disease Bureau
Clifford S. Mitchell, MS, MD, MPH, Director, Environmental Health Bureau
Donald Shell, MD, MA, Director, Cancer and Chronic Disease Bureau

August 23, 2013

Dr. Gerard Martin
Pediatric Cardiology, Children's National Medical Center
111 Michigan Avenue
NW Washington, DC 20010

Dear Dr. Martin,

The Maryland Critical Congenital Heart Disease (CCHD) Newborn Screening Follow Up Program is charged with providing surveillance and quality assurance for CCHD screening. As part of that function, this program needs to be informed of babies who are diagnosed with CCHD after their newborn care is completed. This allows us to evaluate why the infant was not identified on birth screening. Children's National Medical Center is a major referral center for pediatric cardiology patients from Maryland, and as such, I would like to request that your institution provide us with the following information on infants seen for a new diagnosis of CCHD:

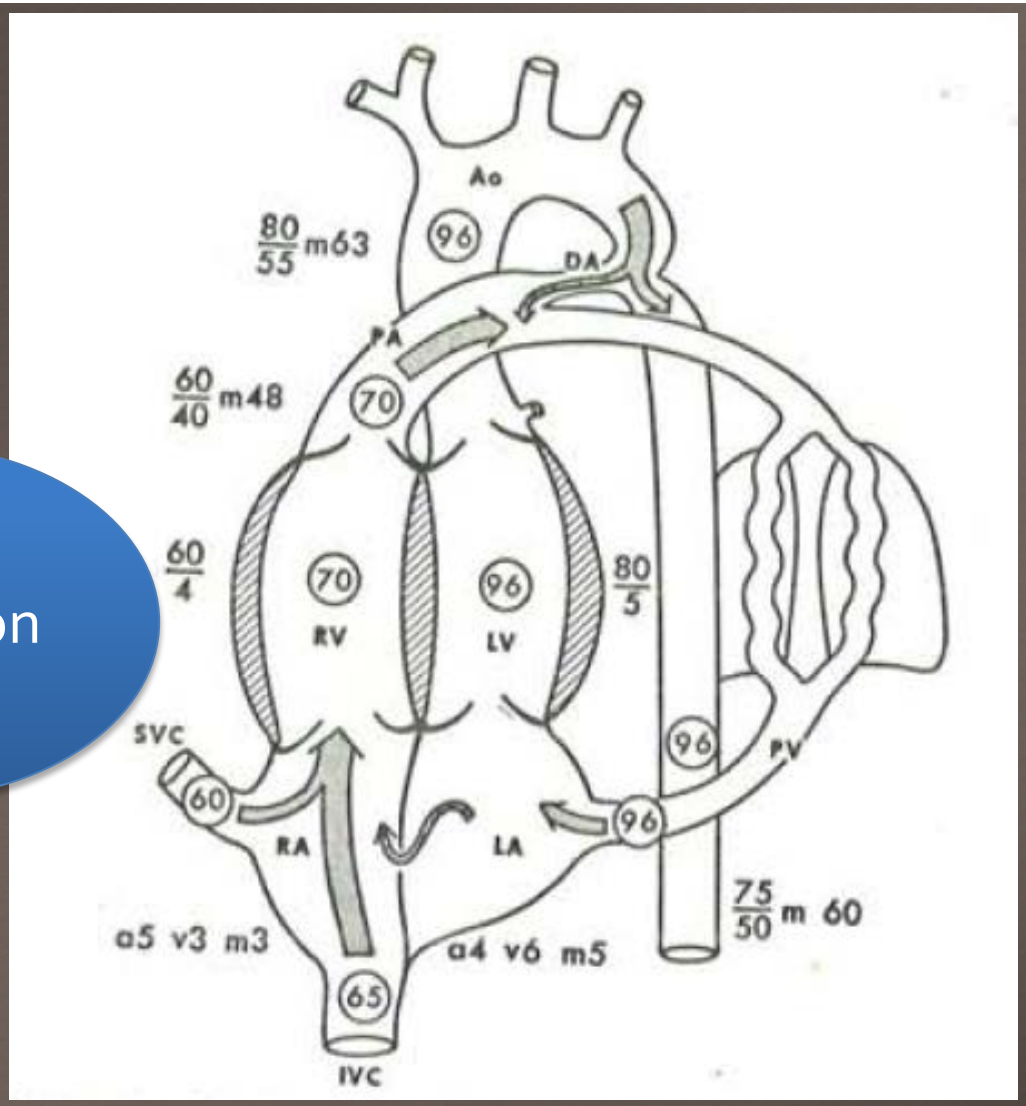
Children's National
Heart Institute
Part of the Children's National Health System

Regional False Negatives

Diagnosis	Birth Hospital	Screened	Pulse Ox Screen Results pre-ductal/post-ductal (%)	Pulse Ox Value Diagnosis (%)
Coarctation of Aorta	A	Yes	98/97	99
TOF	B	Yes	99/97	97
TOF/AV Canal	C	No	Not screened at birth hospital	85
TAPVD	D	Yes	95/95	84
Coarctation of Aorta	E	Yes	98/96	94/84

Coarctation

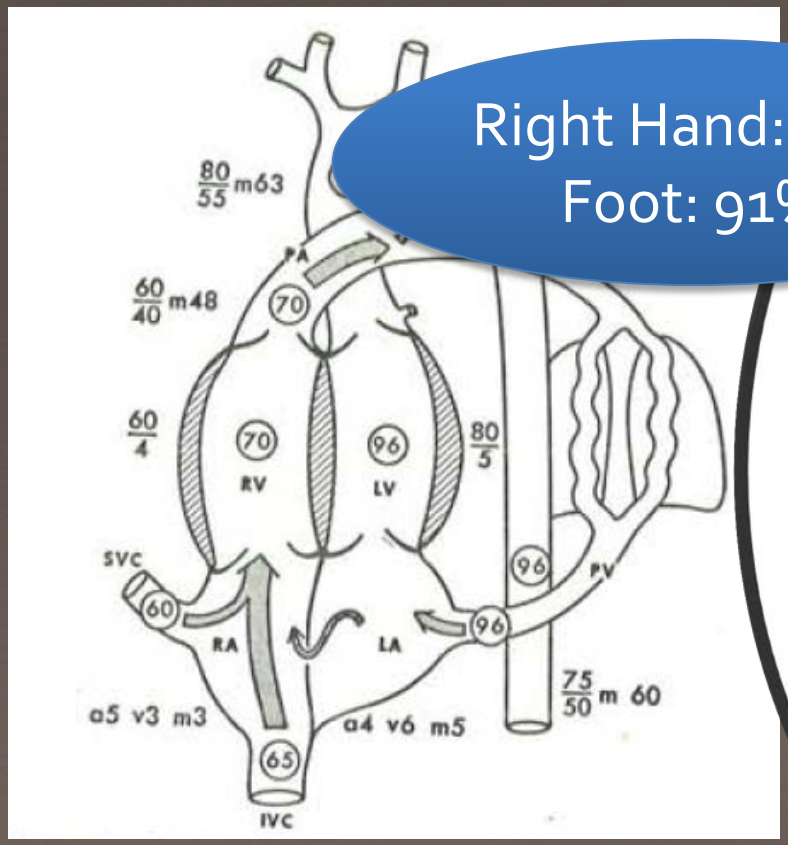
Passing Saturation 96%



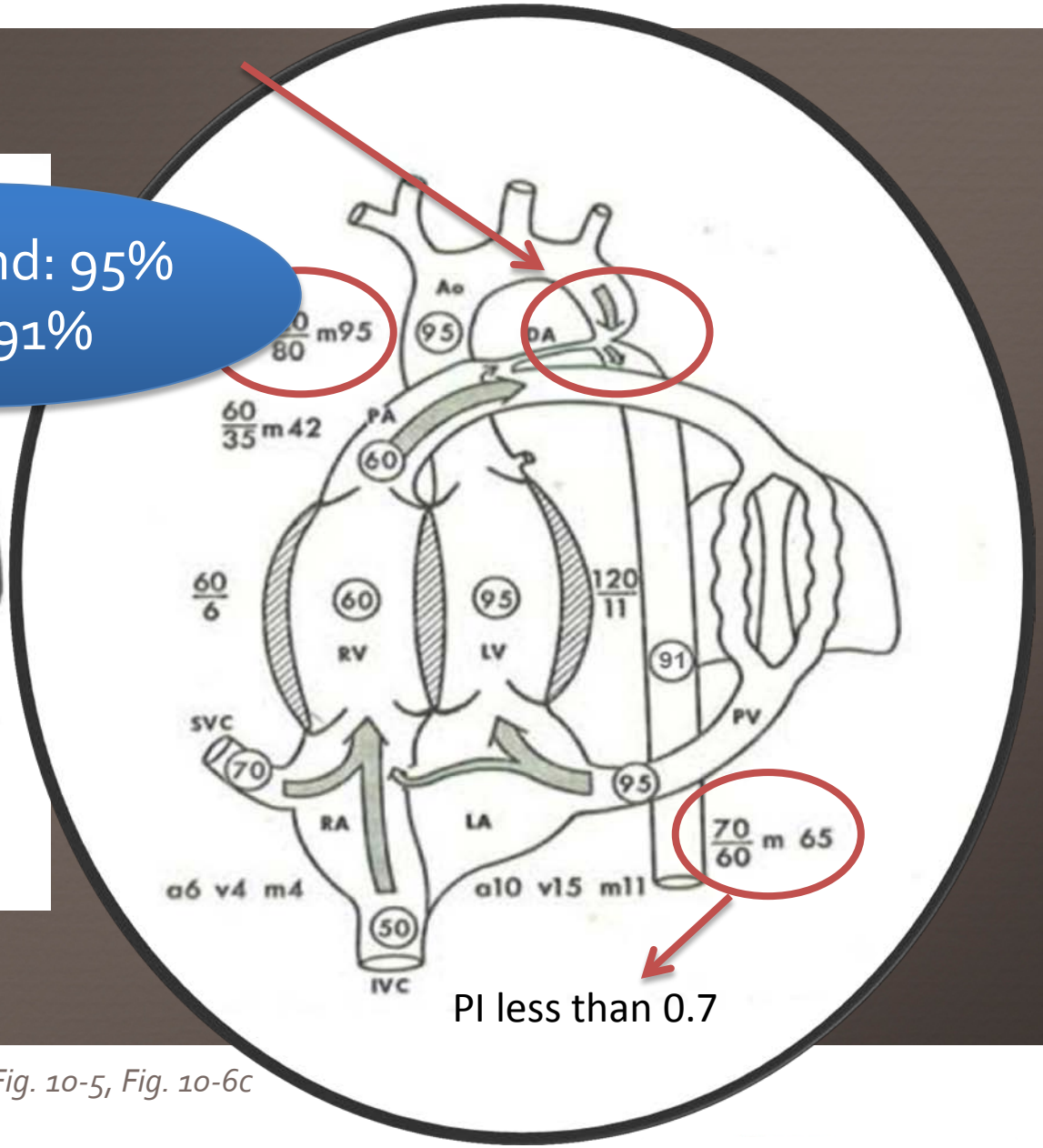
Modified from *Rudolph, 1974, Fig. 10-5*

Children's National Heart Institute
Part of the Children's National Health System

Coarctation



Right Hand: 95%
Foot: 91%



PI less than 0.7

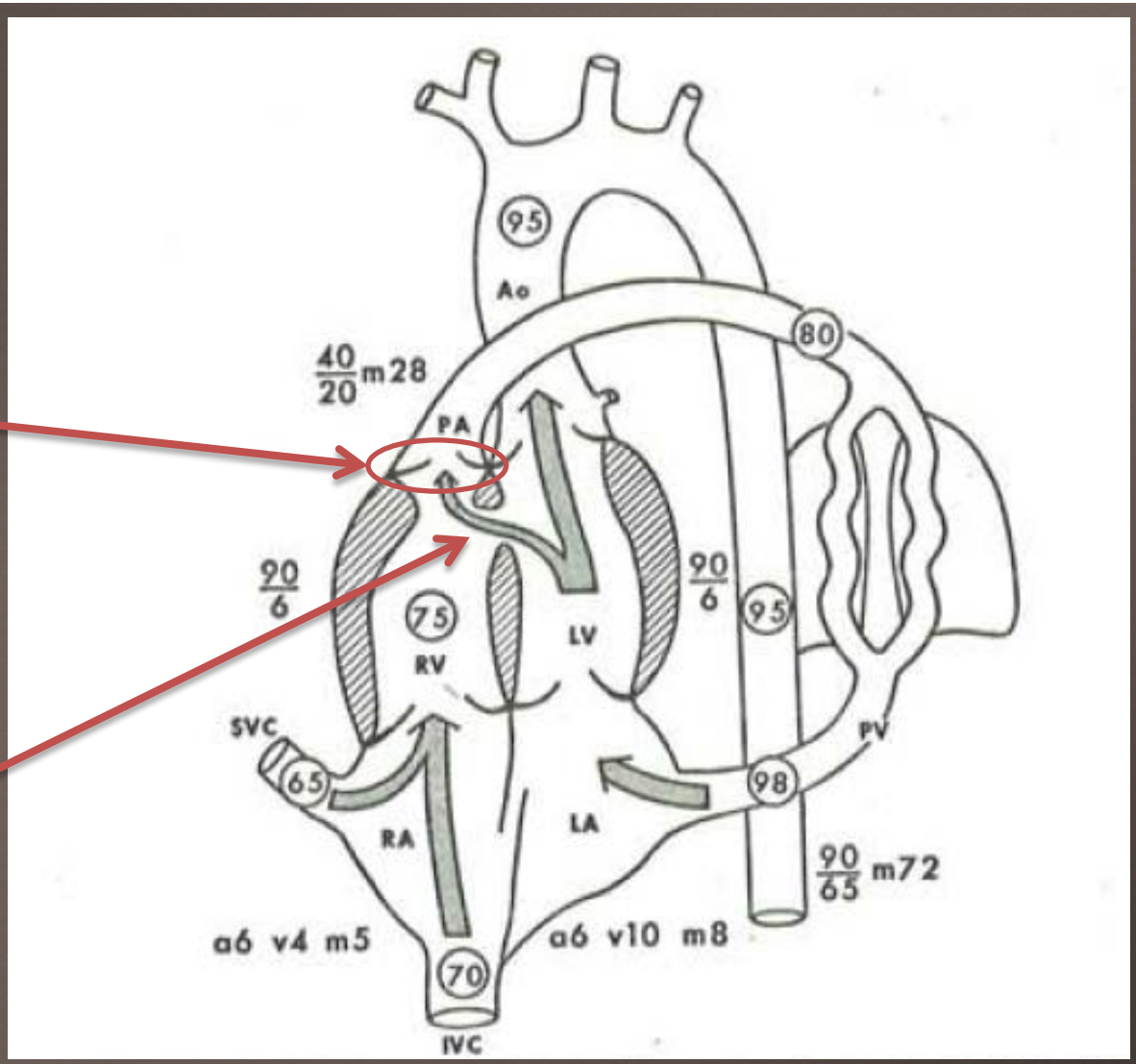
Modified from *Rudolph, 1974, Fig. 10-5, Fig. 10-6c*

Children's National
Heart Institute
Part of the Children's National Health System

Tetralogy of Fallot

Right Ventricular Outflow Obstruction

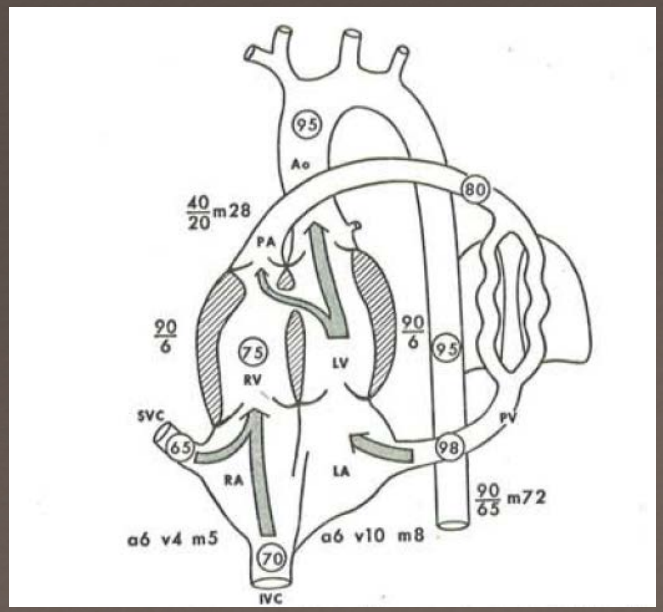
Passing Saturation: 95%

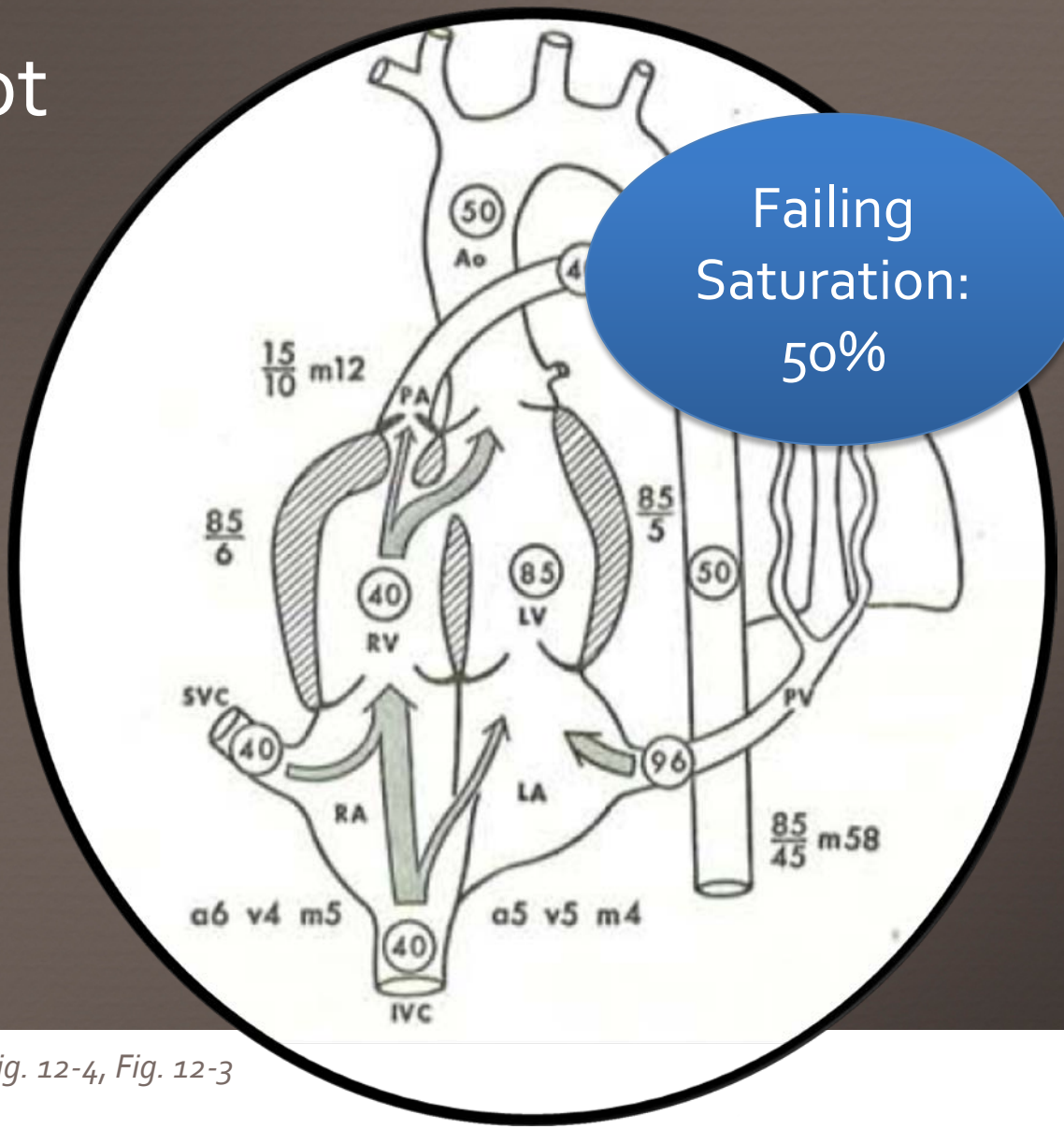


Modified from *Rudolph, 1974, Fig. 12-4*

Children's National Heart Institute
Part of the Children's National Health System

Tetralogy of Fallot





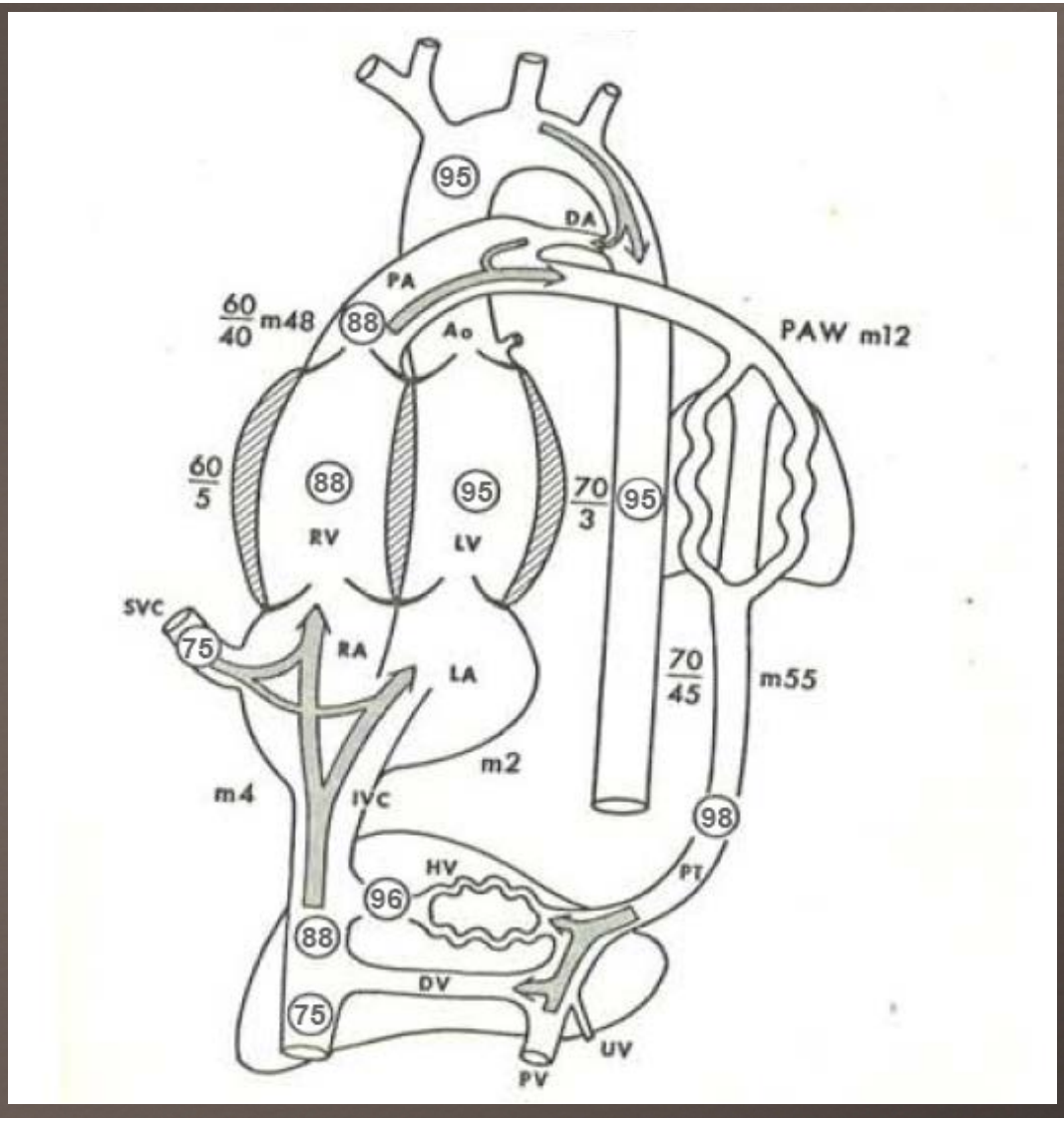
Failing Saturation: 50%

Modified from *Rudolph, 1974, Fig. 12-4, Fig. 12-3*

Children's National
Heart Institute
Part of the Children's National Health System

Total Anomalous Pulmonary Venous Drainage (TAPVD)

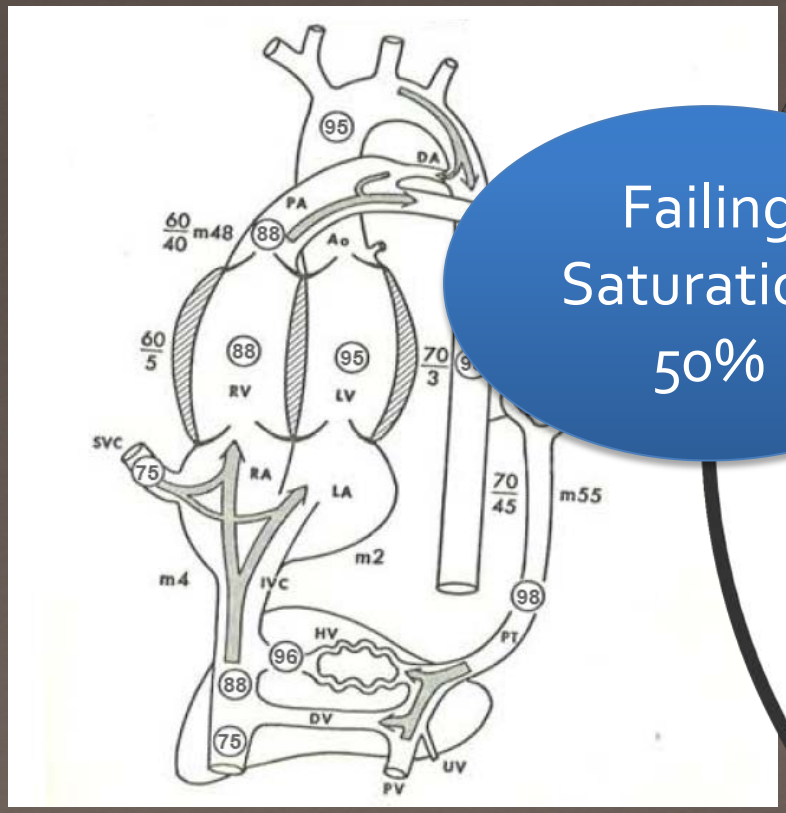
Passing Saturation: 95%



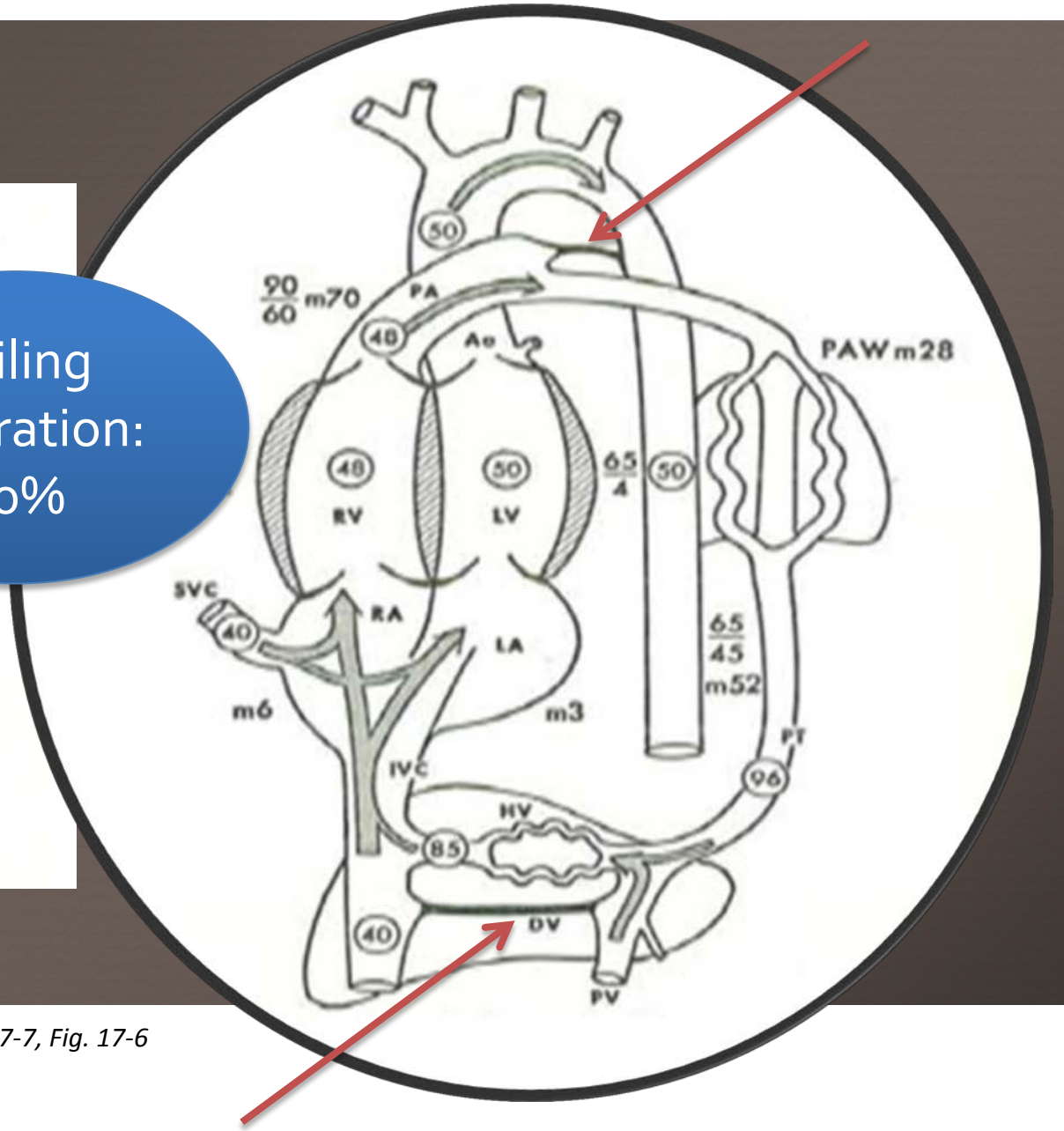
Modified from *Rudolph, 1974, Fig. 17-7*

Children's National Heart Institute
Part of the Children's National Health System

TAPVD



Failing Saturation: 50%



Modified from *Rudolph, 1974, Fig. 17-7, Fig. 17-6*

Children's National Heart Institute
Part of the Children's National Health System

ORIGINAL ARTICLE

Prenatal screening for major congenital heart disease: assessing performance by combining national cardiac audit with maternity data

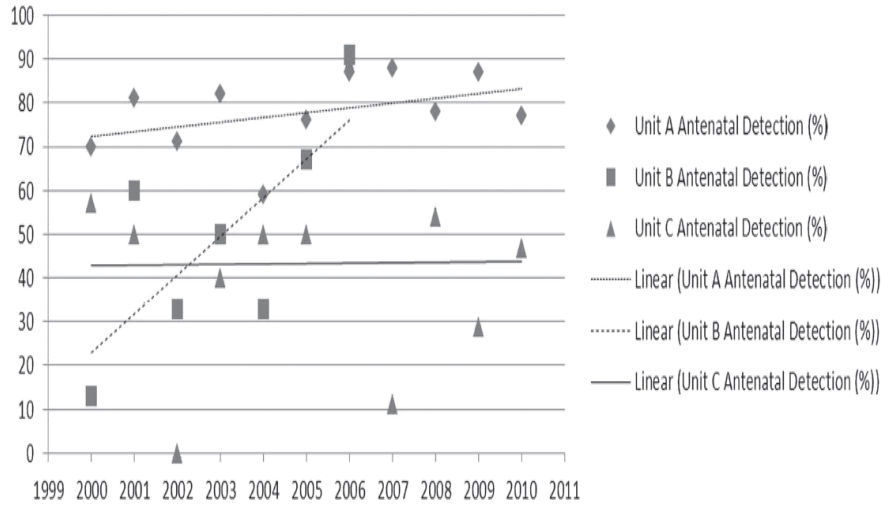


Figure 1 Linear trends for prenatal detection of major congenital heart disease in three screening hospitals over 11 years showing a steady and high detection rate in Hospital A, improvement in Hospital B (data only between 2000–2006), and a steady but poorer detection rate in hospital C. ♦: Hospital A; ■: Hospital B; ▲: Hospital C, and linear trends for Hospital A: ...·····; Hospital B: ····· and Hospital C ———

Conclusions:

- **Wide inter-hospital variation** in prenatal and antenatal detection for TGA and CoA
- **Manual linkage is impractical** on a national basis
- Need for **national validated database** or registry to raise the standard of care
- Hospitals need **granularity** to understand deficiencies and institute actions to improve performance

Gardiner HM, *et al*. *Heart* 2013;**0**:1–8. doi:10.1136/heartjnl-2013-304640

Children's National
Heart Institute
Part of the Children's National Health System

The Toolkit & Heart Smart Videos

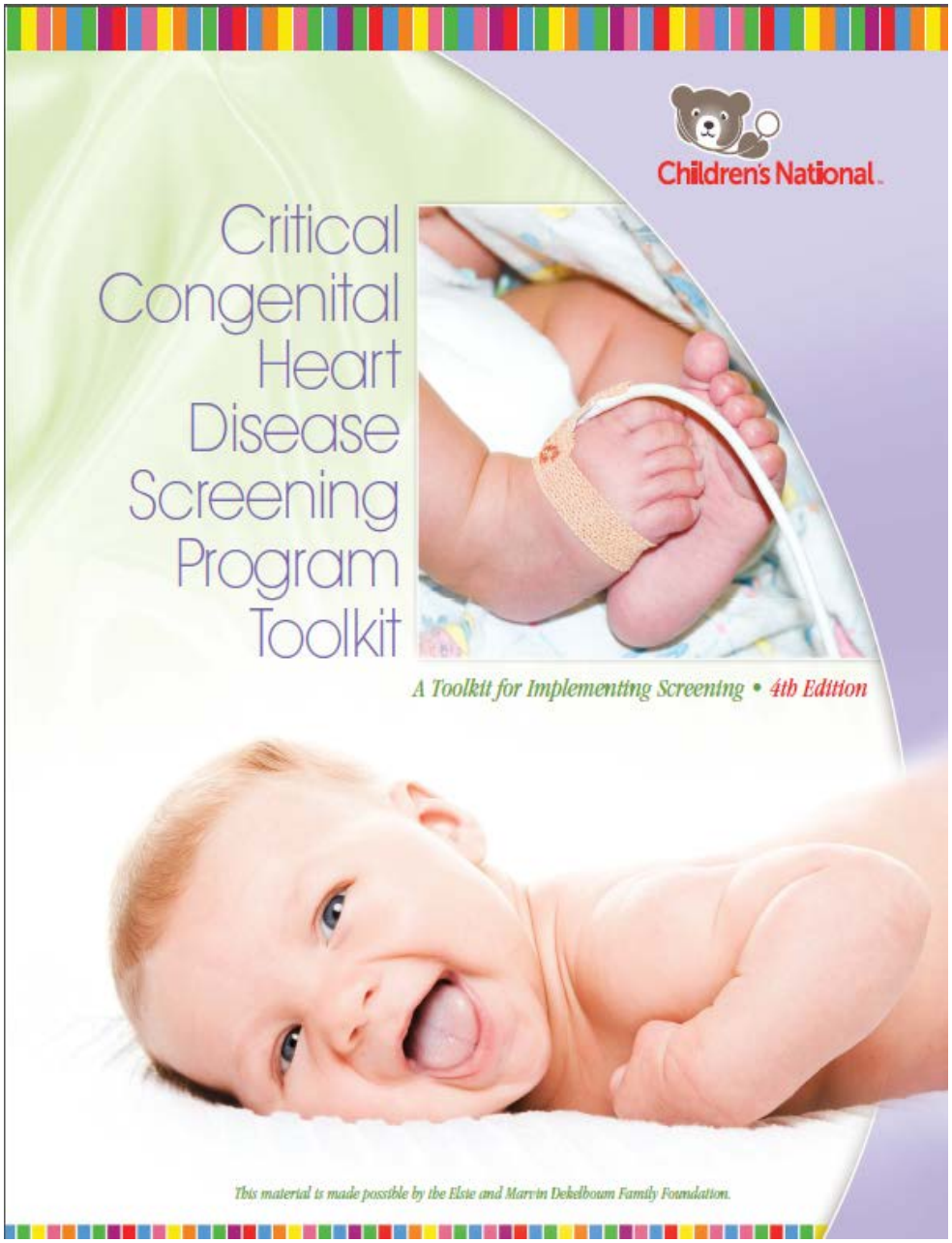
Toolkit Includes:

- Implementation Recommendations
- Screening Protocol
- Education for Families
- Competencies for Providers
- Advocacy Resources and Stories

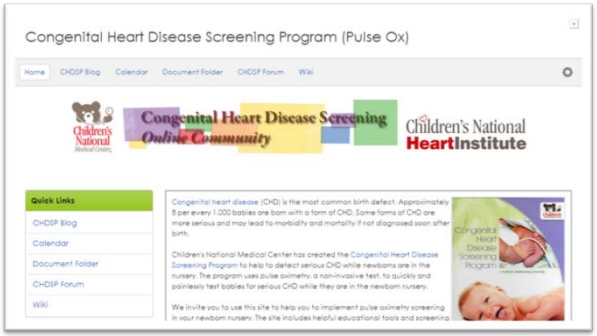
Educational Videos translated into these five languages :

مرحبا
привет
¡hola
nǐ hǎo
bonjour

Children's National
Heart Institute
Part of the Children's National Health System



Online Resources



Children's National Pulse Ox Program
www.childrensnational.org/pulseox/

Center for Disease Control

www.cdc.gov/ncbddd/pediatricgenetics/cchdscreening.Html

Baby's First Test

http://www.babysfirsttest.com//

Parent Advocacy Groups

http://1in100.org/ ; www.tchin.org

Newborn Coalition

http://newborncoalition.com/

ACMG CCHD ACTion Sheet

NewSTEPs

https://www.newsteps.org/

CDC
CENTERS FOR DISEASE CONTROL AND PREVENTION

One in One Hundred

newborn coalition

Children's National
Heart Institute
Part of the Children's National Health System

Thank You

Questions?

Children's National

Heart Institute

Part of the Children's National Health System

Contact Information

www.childrensnational.org/pulseox
pulseox@childrensnational.org

Gerard R. Martin, MD, FAAP, FACC, FAHA
gmartin@childrensnational.org

Lowell Frank, MD
lfrank@childrensnational.org

Lisa A. Hom, RN, Esq.
lhom@childrensnational.org
202-476-5063

Children's National
Heart Institute
Part of the Children's National Health System