North Dakota Newborn Screening Conference

Welcome

Internet Passcode: 12351235

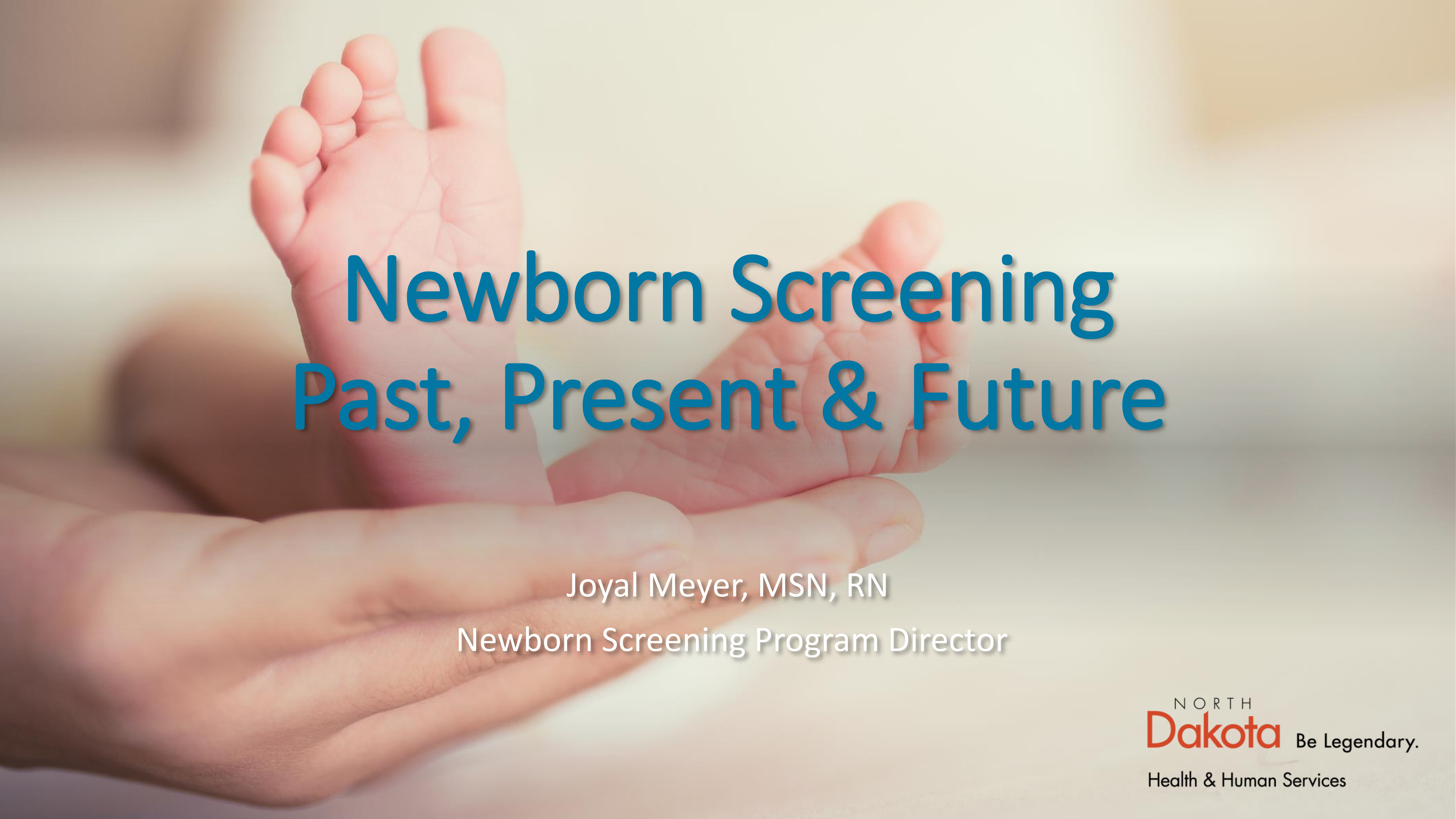
Scan To Take Pre Test

https://ndhealth.co1.qualtrics.com/jfe/form/SV_20lIVn29P6GRgHA

JOYAL MEYER RN, MSN

DISCLOSURES

- Nothing to disclose



Newborn Screening
Past, Present & Future

Joyal Meyer, MSN, RN

Newborn Screening Program Director

NORTH
Dakota Be Legendary.
Health & Human Services

Objectives

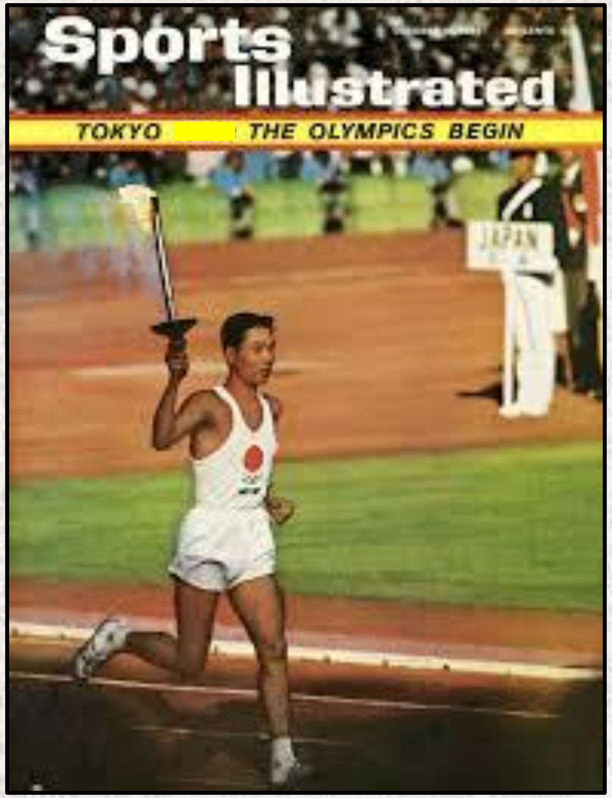
1

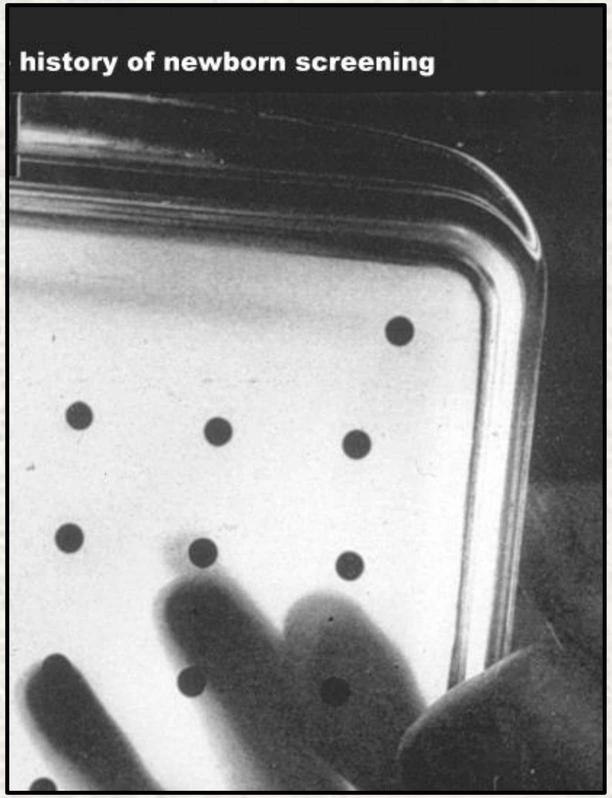
1. Identify the different parts of newborn screening.

2

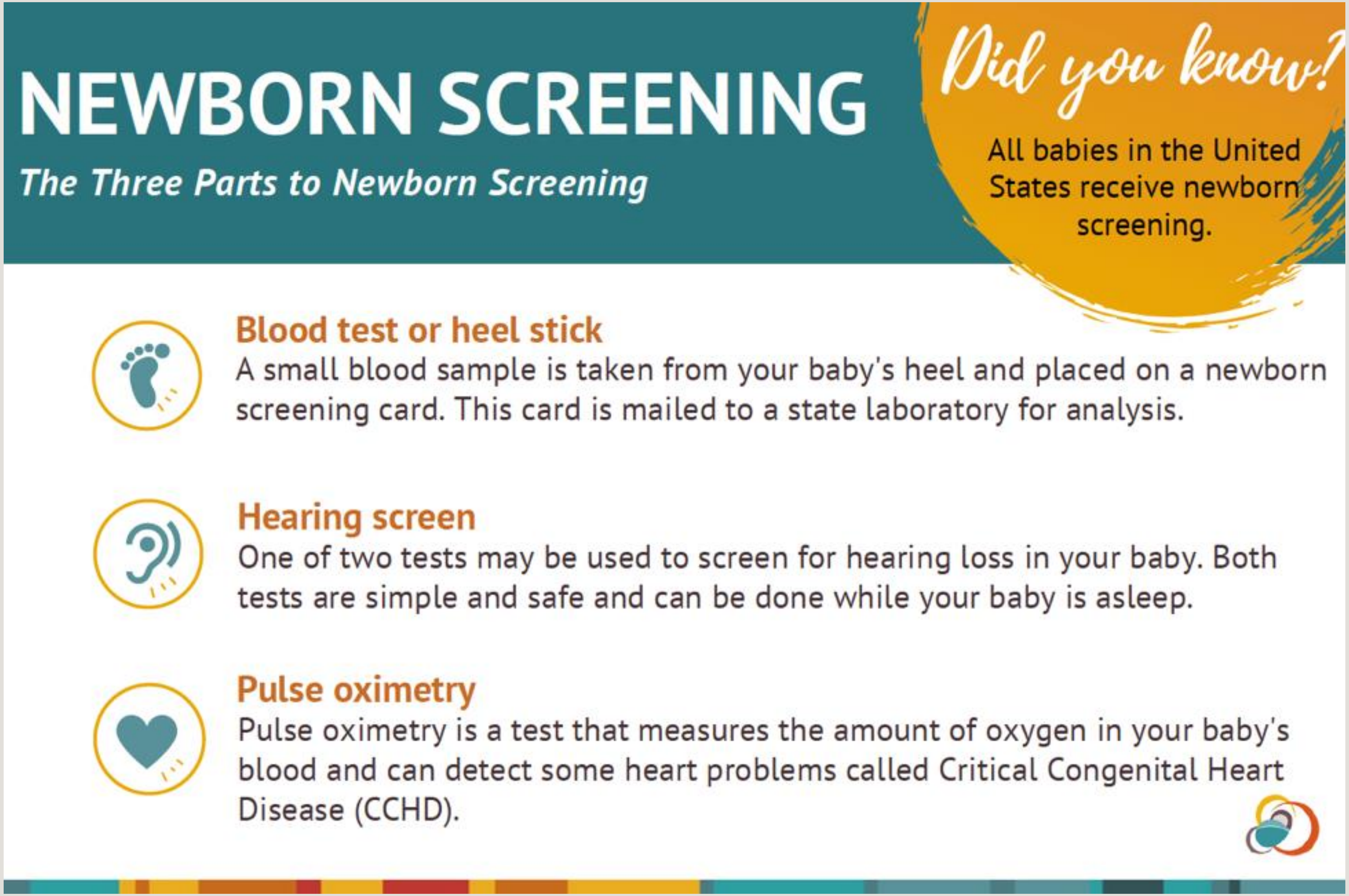
2. Recognize which disorders are considered time critical.

What Year Were These Events?





1964



NEWBORN SCREENING

The Three Parts to Newborn Screening

Did you know?

All babies in the United States receive newborn screening.

Blood test or heel stick

A small blood sample is taken from your baby's heel and placed on a newborn screening card. This card is mailed to a state laboratory for analysis.

Hearing screen

One of two tests may be used to screen for hearing loss in your baby. Both tests are simple and safe and can be done while your baby is asleep.

Pulse oximetry

Pulse oximetry is a test that measures the amount of oxygen in your baby's blood and can detect some heart problems called Critical Congenital Heart Disease (CCHD).

Slide Courtesy of Baby's First Test

What Screenings Are Mandated in ND?

Blood Spot Screening ✓

Hearing Screening

Pulse Oximetry Screening ✓

Timeline for NBS Disorders in ND

1964	Phenylketonuria (PKU)
1977	Congenital Hypothyroidism (CH)
1991	Galactosemia
1992	Congenital Adrenal Hyperplasia & Maple Syrup Urine Disease (MSUD)
2003	MCAD, Biotinidase, Sickle Cell, Hemoglobinopathies
2004	Disorders Detected by Tandem Mass Spectrometry Officially on NBS Panel
2006	Cystic Fibrosis (CF)
2016	Severe Combined Immunodeficiency (SCID)
2021	Spinal Muscular Atrophy (SMA)

Recommended Uniform Screening Panel (RUSP)

Advisory Committee on Heritable Disorders in Newborns & Children (ACHDNC)

Primary Conditions

Secondary Conditions

National Push for Timeliness in NBS

A JOURNAL SENTINEL WATCHDOG REPORT

Deadly Delays

The nation's newborn screening programs depend on speed and science to save babies from rare diseases. But thousands of hospitals fall short, deadly delays are ignored and failures are hidden from public view — while babies and their families suffer.

Kristyna Wentz-Graff/Journal Sentinel

Delays at hospitals across the country undermine newborn screening programs, putting babies at risk of disability and death

By Ellen Gabler of the Journal Sentinel staff

Milwaukee Journal Sentinel released on November 16, 2013

Time Critical Disorders

Organic Acid Conditions

- Propionic Acidemia
- Methylmalonic Acidemia (methylmalonyl-CoA mutase)
- Isovaleric Acidemia
- 3-Hydroxy-3- Methyglutaric Aciduria
- Holocarboxylase Synthase Deficiency
- Beta-Ketothiolase Deficiency
- Glutaric Acidemia, Type I

Fatty Acid Oxidation Disorders

- Medium-chain acyl- CoA Dehydrogenase Deficiency
- Very long-chain acyl- CoA Dehydrogenase Deficiency
- Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency
- Trifunctional Protein Deficiency
- Glutaric Acidemia, Type II
- Carnitine Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyltransferase Type II Deficiency

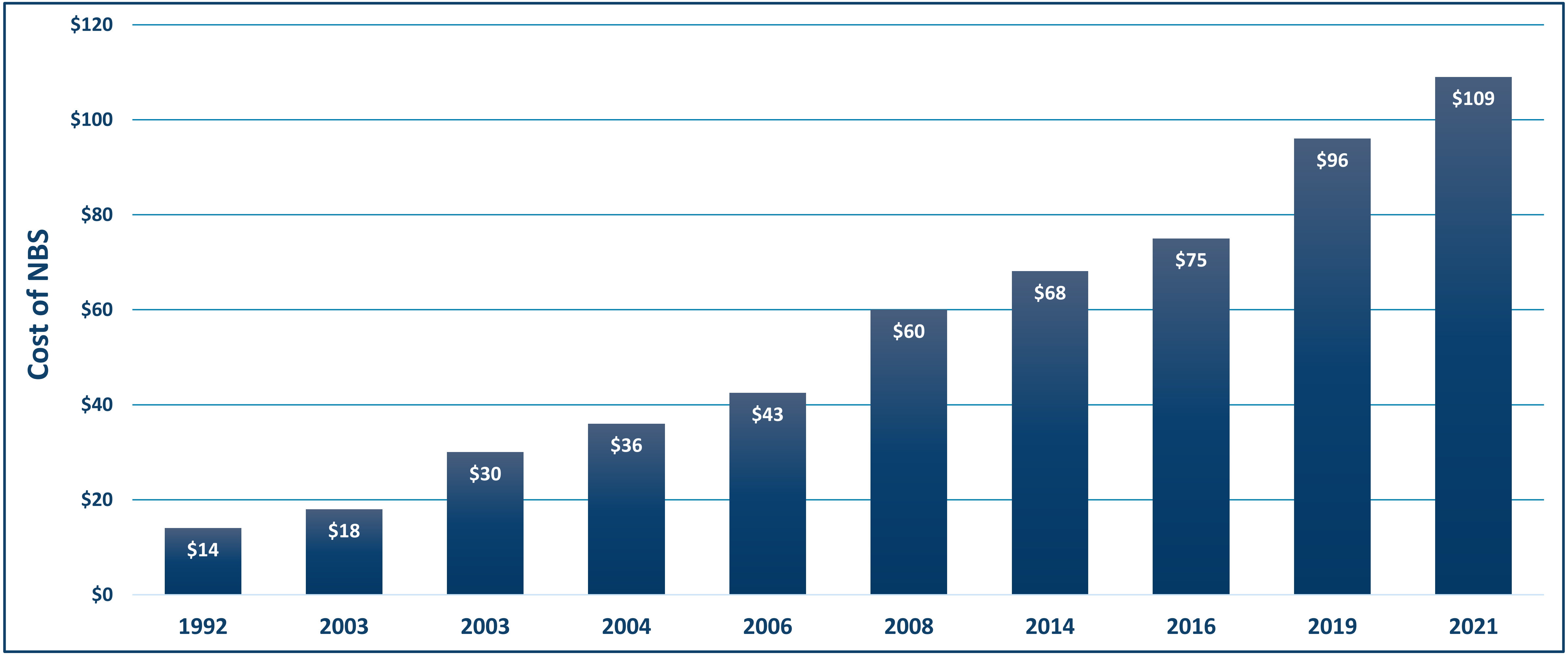
Amino Acid Disorders

- Argininosuccinic Aciduria
- Citrullinemia, Type I
- Maple Syrup Urine Disease

Other Disorders

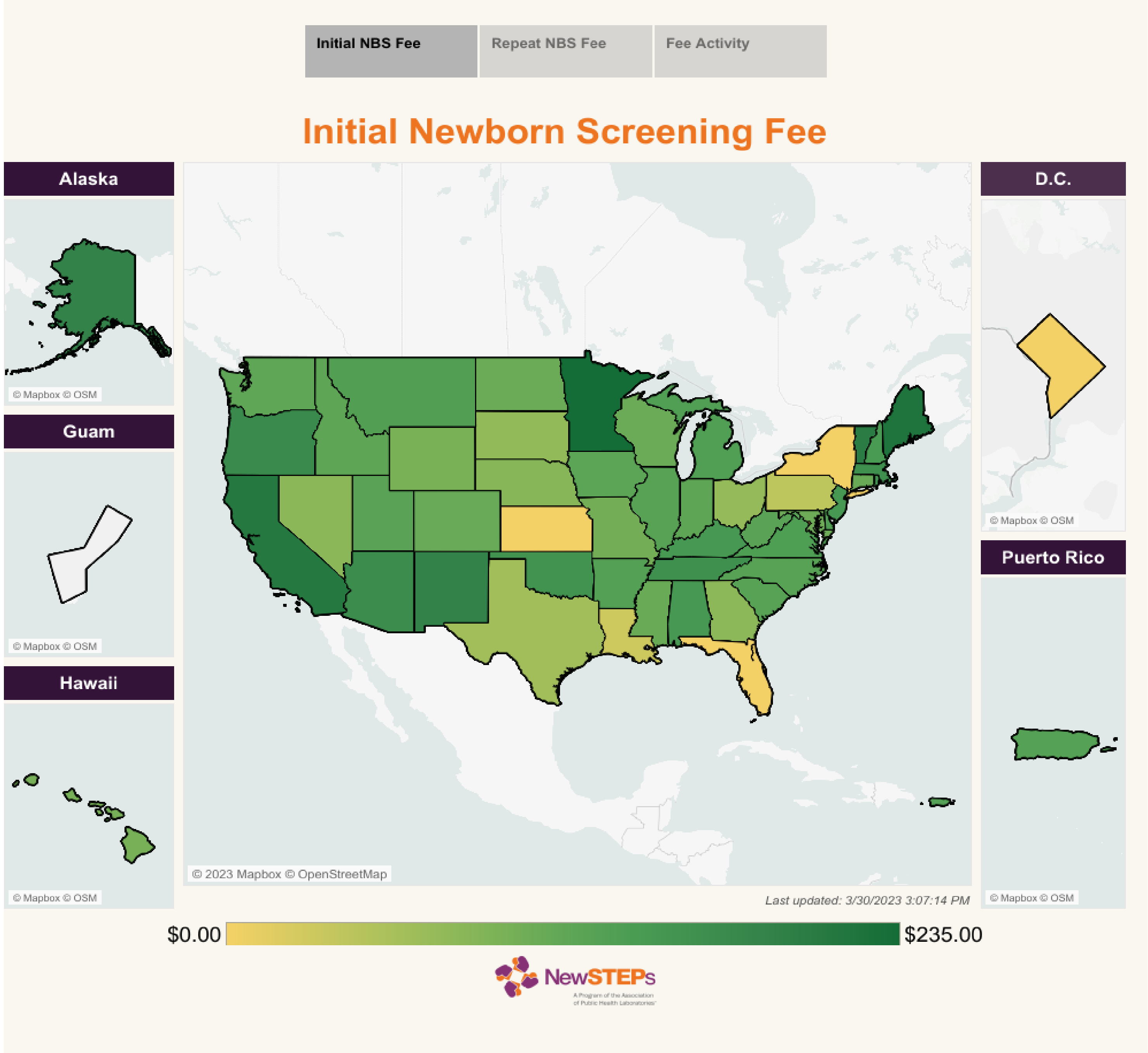
- Classic Galactosemia
- Congenital Adrenal Hyperplasia

Newborn Screening Fee History in ND



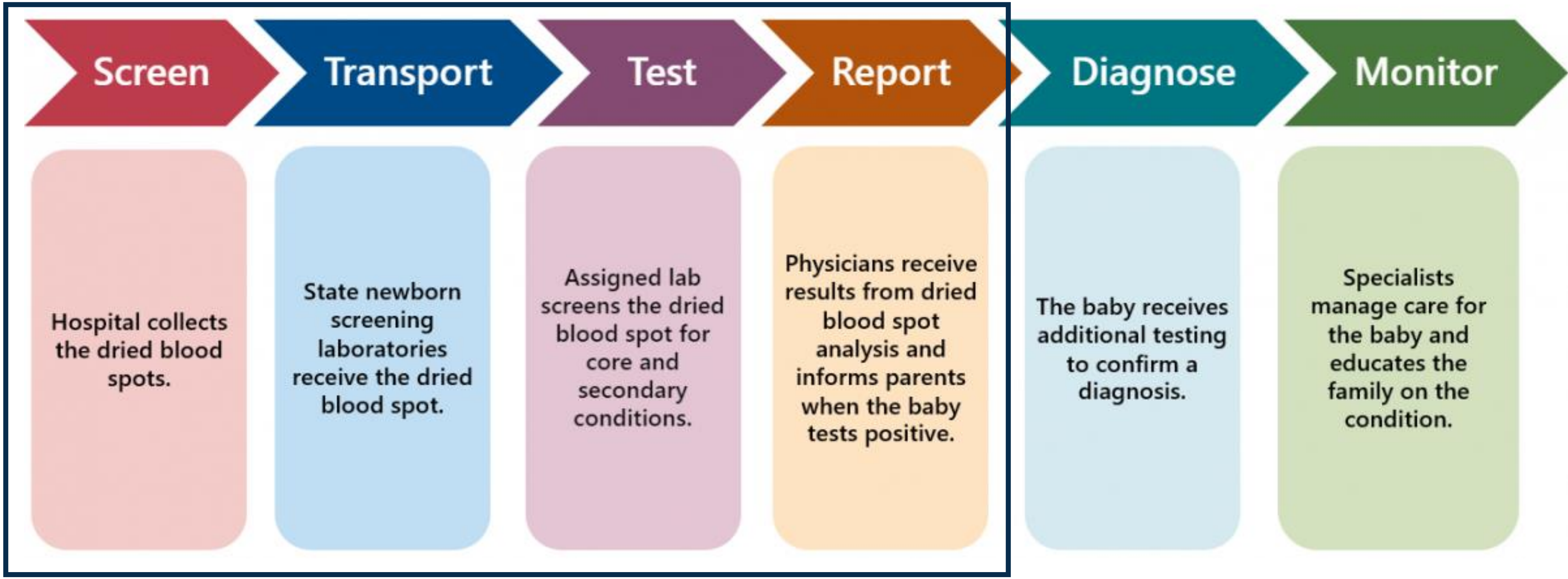
How Do We Compare?

- States in Yellow Have No NBS Fee
  - Kansas
  - Florida
  - New York
- States in Dark Green Have the Highest NBS Fee
  - Minnesota
  - Delaware
  - California



North Dakota

Newborn Screening Process



Screen

Hospital collects the dried blood spots.

Transport

State newborn screening laboratories receive the dried blood spot.

Test

Assigned lab screens the dried blood spot for core and secondary conditions.

Report

Physicians receive results from dried blood spot analysis and informs parents when the baby tests positive.

Diagnose

The baby receives additional testing to confirm a diagnosis.

Monitor

Specialists manage care for the baby and educates the family on the condition.

Image Courtesy of CDC Contingency Plan Version II

Timely Specimen Collection Run Chart

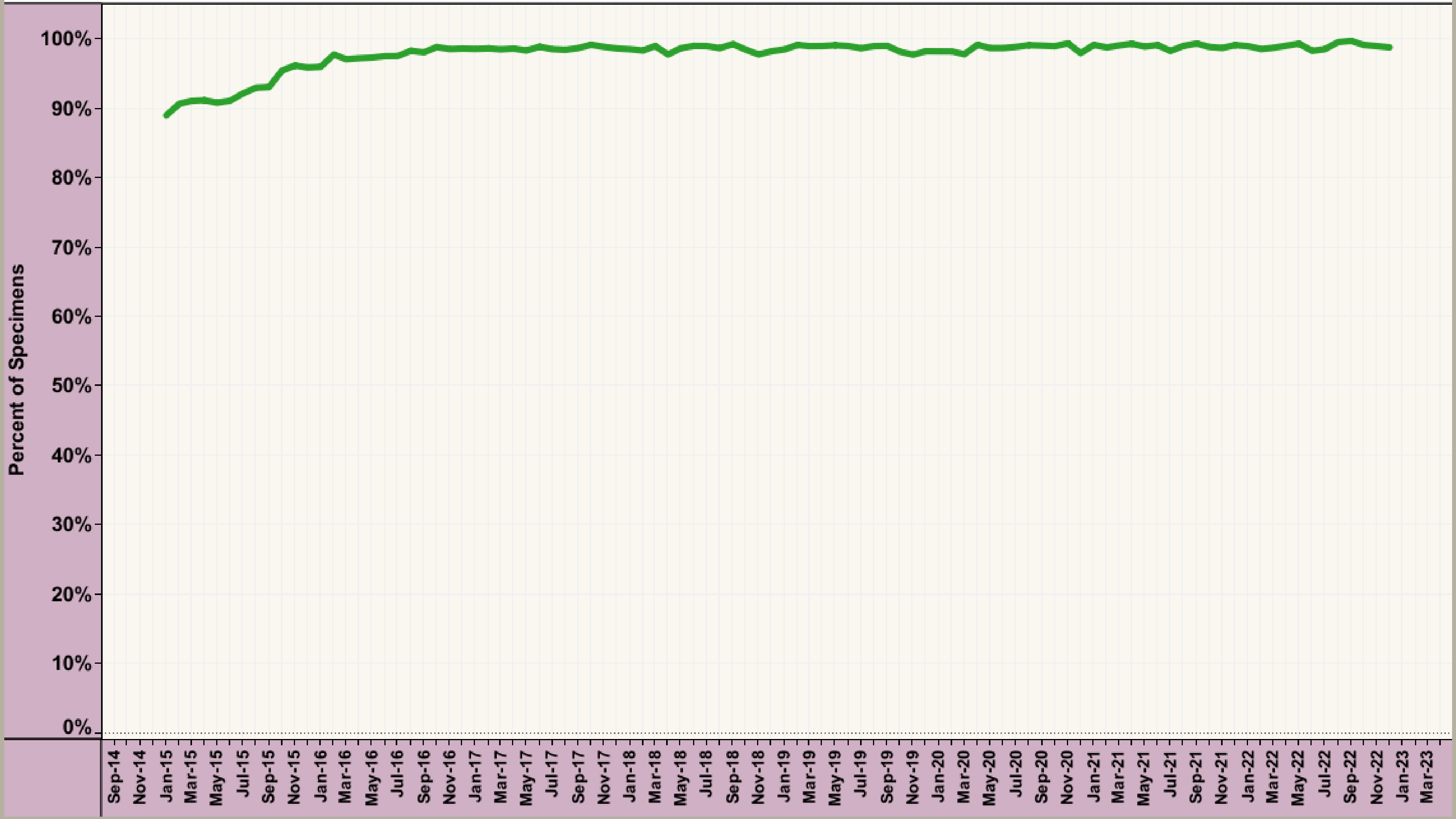
Percent of first dried blood spot specimens collected in ***<=48 Hours*** *from birth*

Select Time Interval
<=48 Hours

Select Median
No Median

Select NBS Program ID
186

Change Date Range
Jan 2015 to Dec 2022



Birth to Reporting Time Critical Results Run Chart

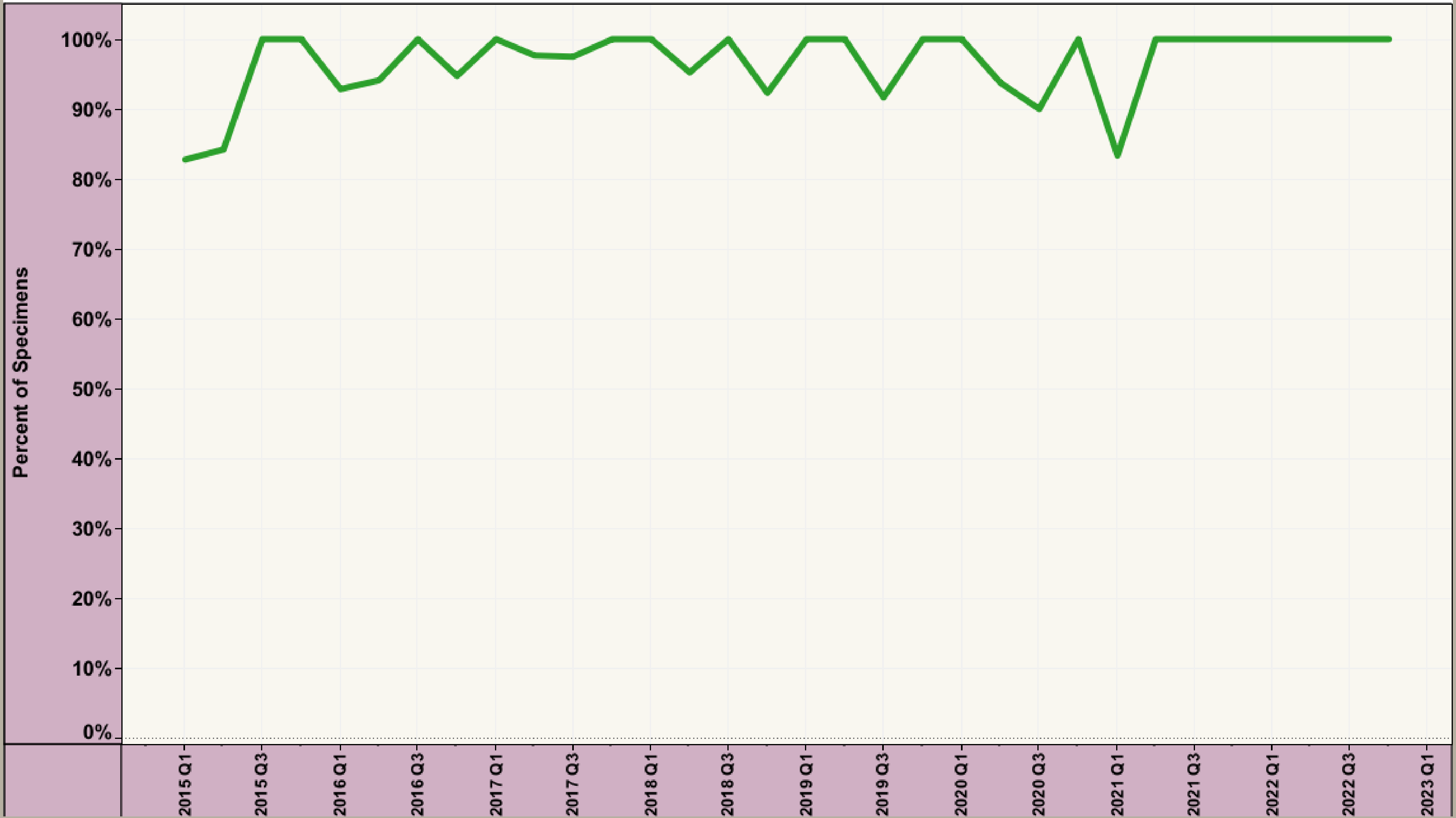
Percent of specimens with a time critical result reported on ***<=Day 5*** *after birth*

Select Time Interval
<=Day 5

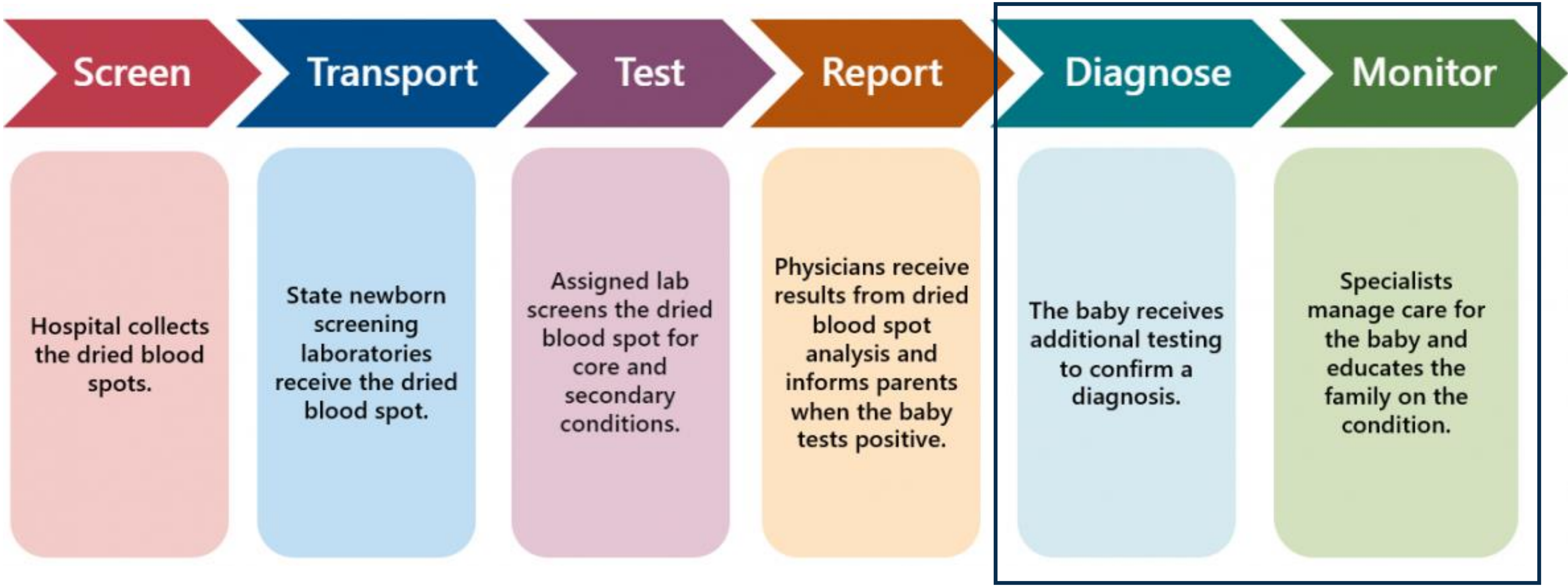
Select Median
No Median

Select NBS Program ID
186

Change Date Range
2015 Q1 to 2022 Q4



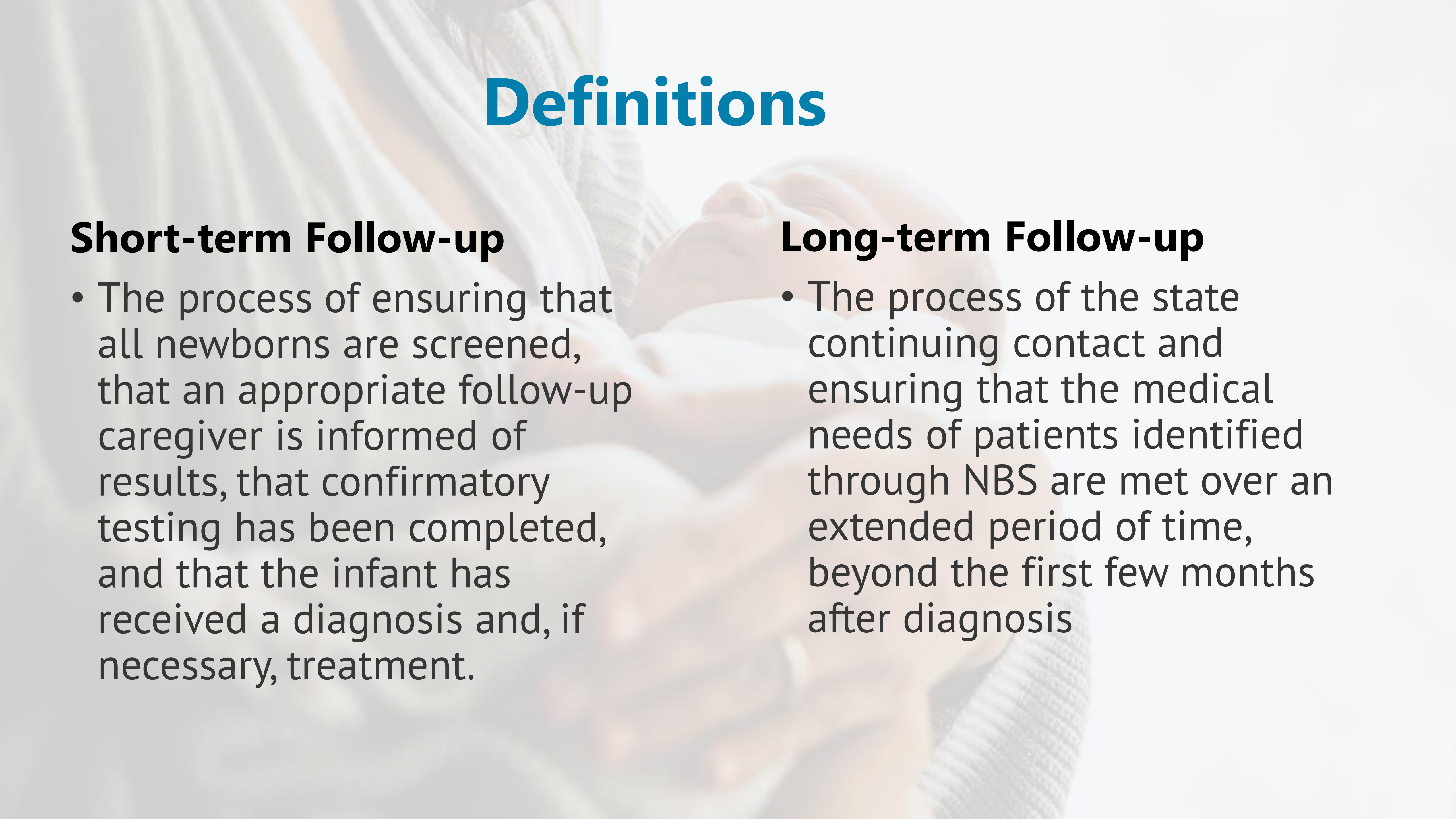
Newborn Screening Process



Screen	Transport	Test	Report	Diagnose	Monitor
Hospital collects the dried blood spots.	State newborn screening laboratories receive the dried blood spot.	Assigned lab screens the dried blood spot for core and secondary conditions.	Physicians receive results from dried blood spot analysis and informs parents when the baby tests positive.	The baby receives additional testing to confirm a diagnosis.	Specialists manage care for the baby and educates the family on the condition.

Image Courtesy of CDC Contingency Plan Version II

Definitions



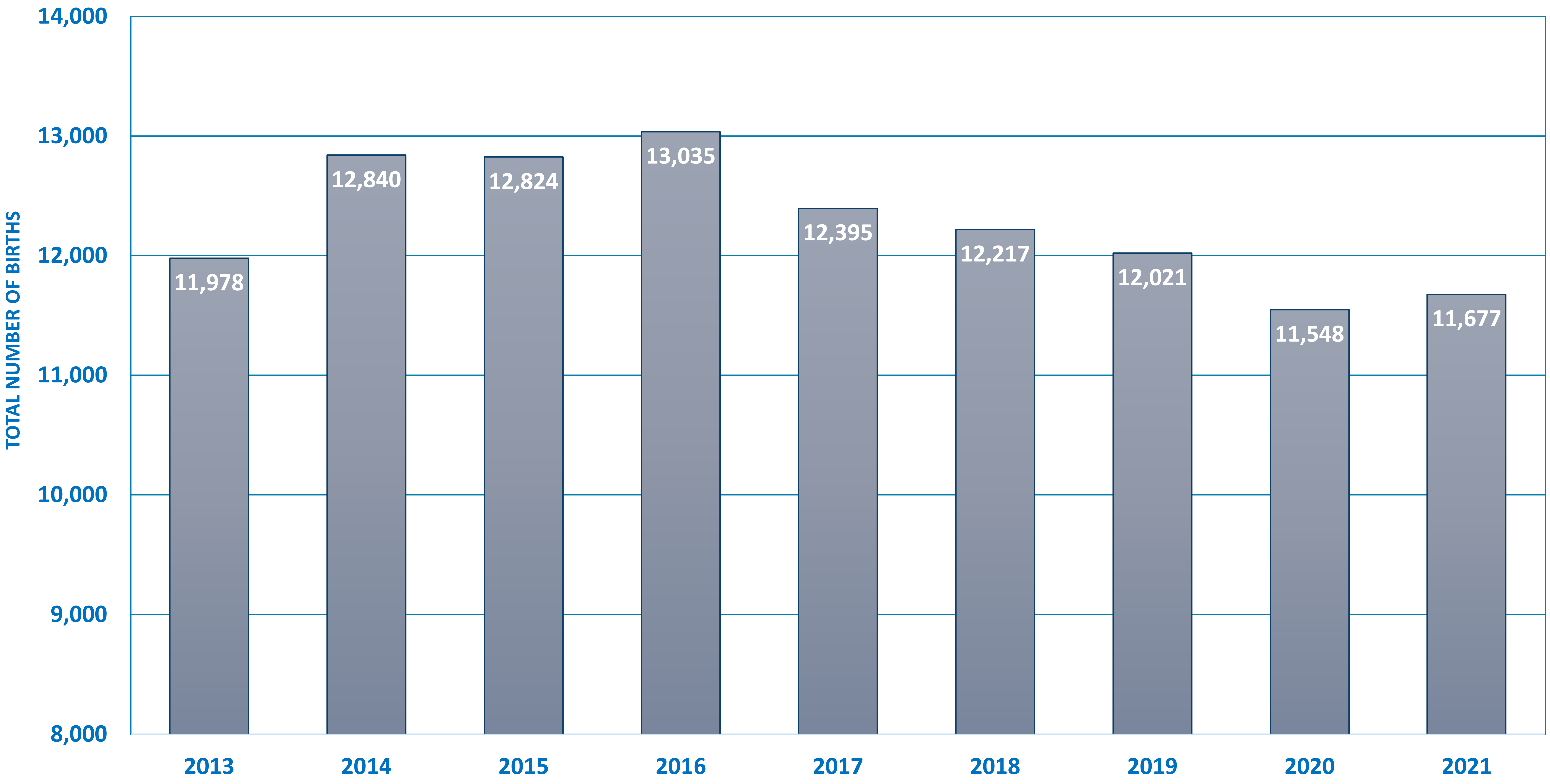
Short-term Follow-up

- The process of ensuring that all newborns are screened, that an appropriate follow-up caregiver is informed of results, that confirmatory testing has been completed, and that the infant has received a diagnosis and, if necessary, treatment.

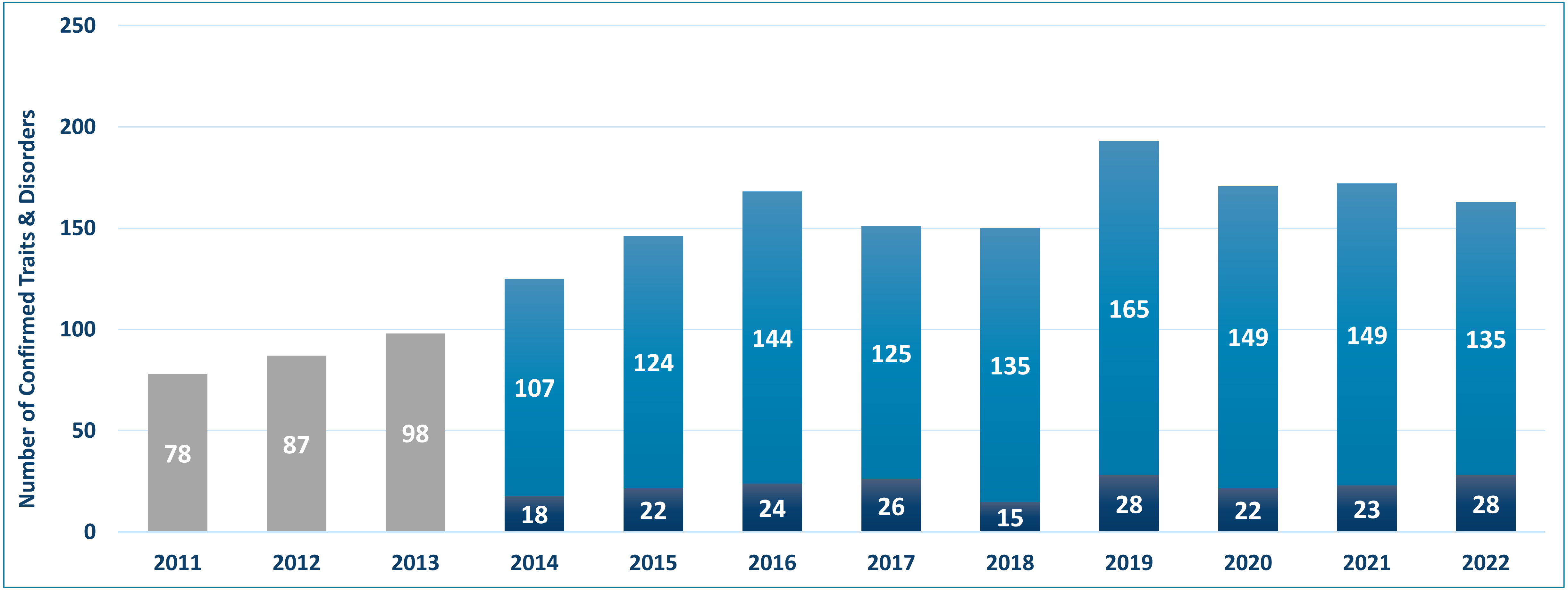
Long-term Follow-up

- The process of the state continuing contact and ensuring that the medical needs of patients identified through NBS are met over an extended period of time, beyond the first few months after diagnosis

North Dakota Occurrent Birth Rate

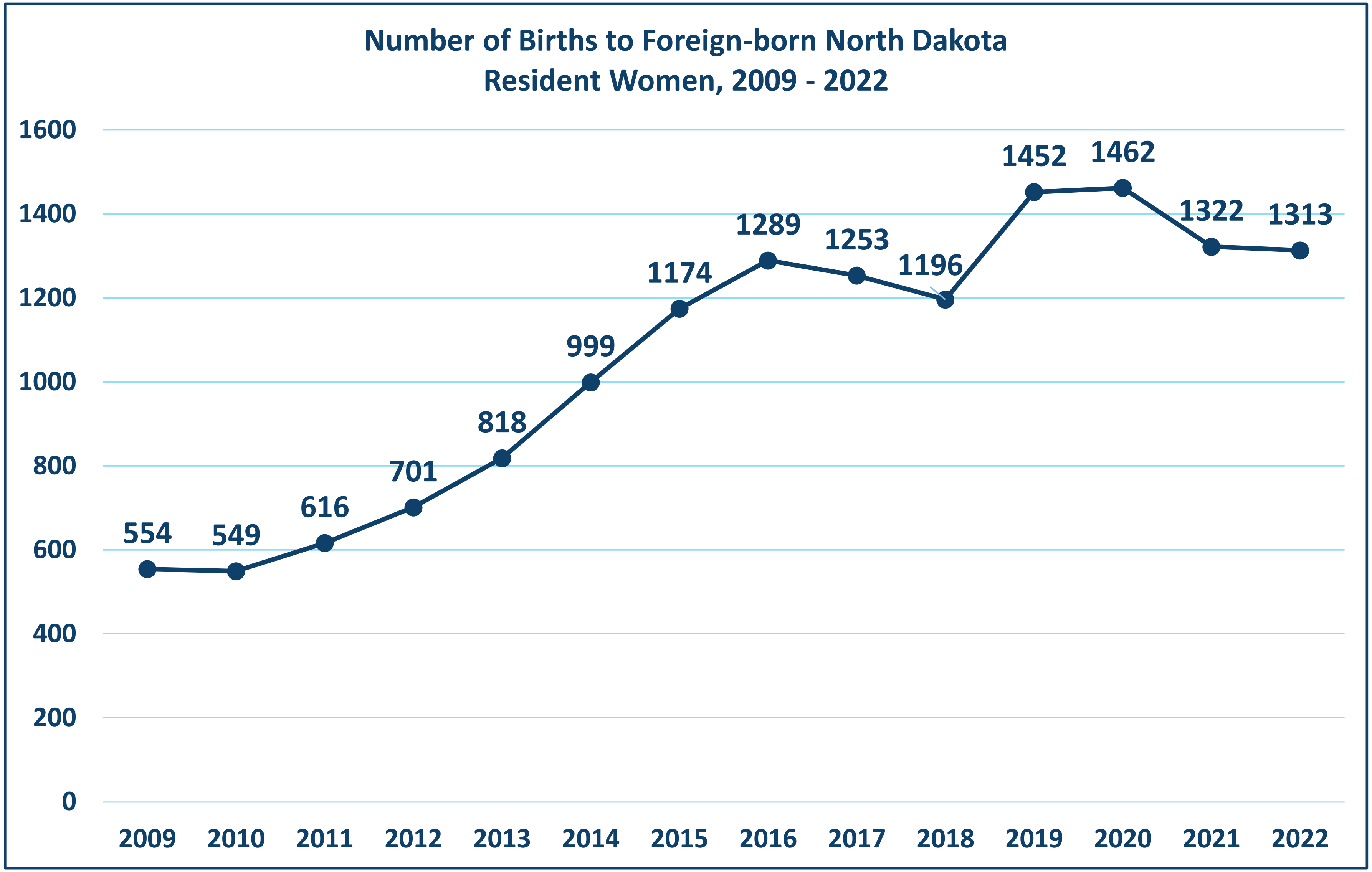


Confirmed Newborn Screening Cases



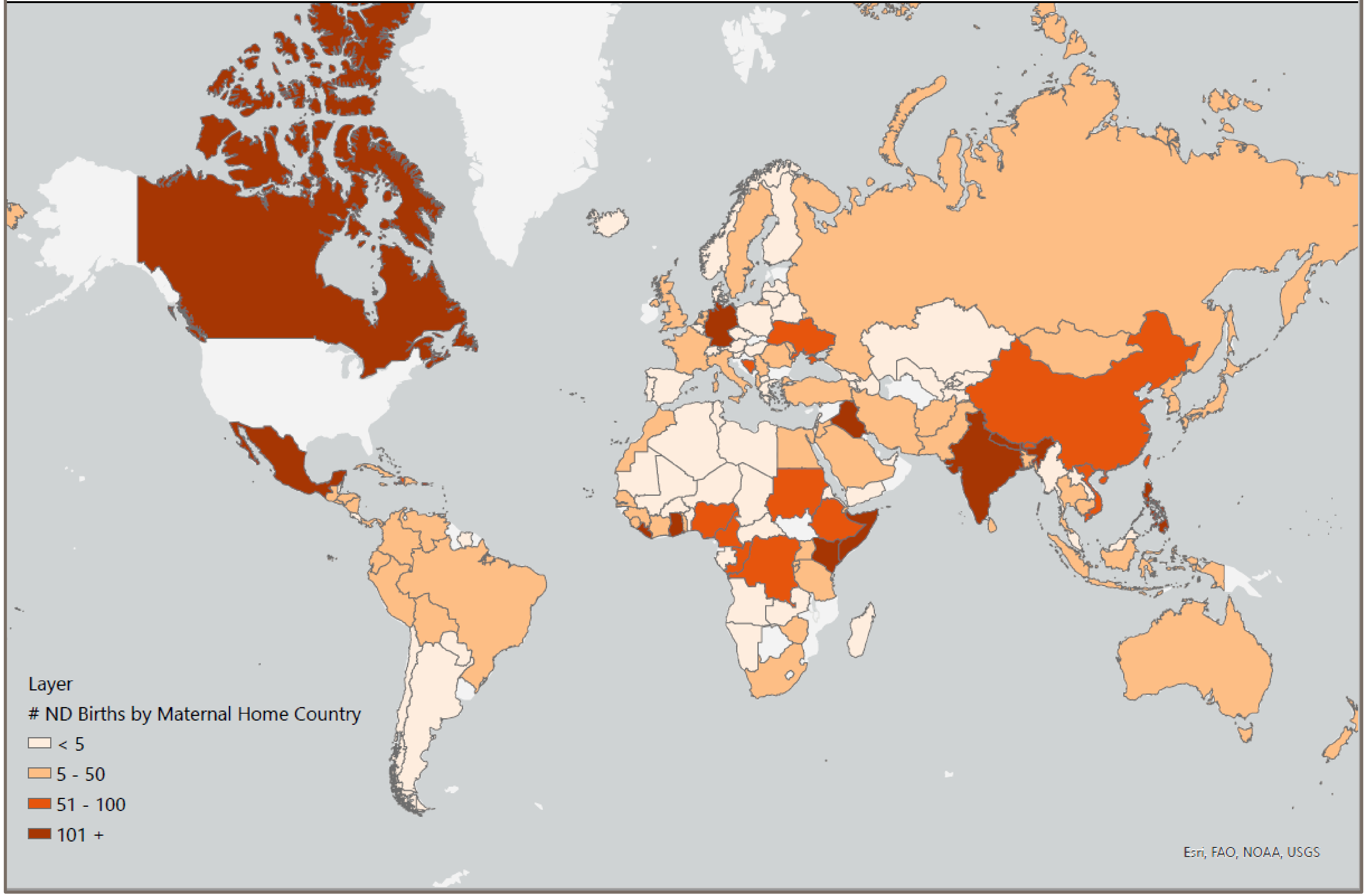
*Data Source: Newborn Screening Case Log

Increasing Diversity in North Dakota



*Data source: ND Vital Records

Number of North Dakota Resident Births by Mother's Home Country, 2019 - 2023*



Esri, FAO, NOAA, USGS

NBS Long-term Follow-up

Newborn Screening Welcome Card

CONGRATULATIONS
ON THE BIRTH OF YOUR BABY!

May your journey
be filled with support,
love, and warm cuddles.

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Health & Human Services

North Dakota
Newborn Screening
Program

ENGLISH: In order to best serve you, interpreter services are available upon request.
SPANISH (LA): A fin de servirle mejor, los servicios de interpretación están disponibles a solicitud.
FRENCH: Afin de mieux vous servir, des services d'interprétation sont disponibles sur demande.
RUSSIAN: По запросу доступны услуги переводчика в целях обеспечения наилучшего обслуживания.
NEPALI: तपाईंलाई सर्वोत्तम सेवा दिनको लागि, आग्रह अनुरूप दोभासे सेवाहरू पनि उपलब्ध छन्।
SOMALI: Si sida ugu fiican laguugu adeego, adeegyada turjubaanka ayaa la heli karaa markii la codsado.
SWAHILI: Ili kuwahudumia bora zaidi, huduma za ukalimani zinapatikana kwa ombi.
ARABIC: لخدمتك بطريقة مُثلى، تتاح خدمات الترجمة الشفوية عند الطلب.
BOSNIAN: Kako bismo vam pružili najbolje usluge, usluge prevođenja će vam biti dostupne na zahtjev.
KURDISH: بو ئەوەی بتوانین بە باشترین شێوە خزمەتت بکەین، خزمەتگوزاری وەرگێڕان، ئەگەر داوا بکرێت، لە بەردەستە.
FULANI: Ngam huuwa neeki ma no woodata, kuude nantinooɓe ɗo waagi to ɗaɓɓitiɗa.
HAITIAN CREOLE: Pou sèvi-ou pi byen, sèvis entèprèt yo disponib sou demann.
CHINESE: 为了给您最好的服务，口译服务可按需提供。

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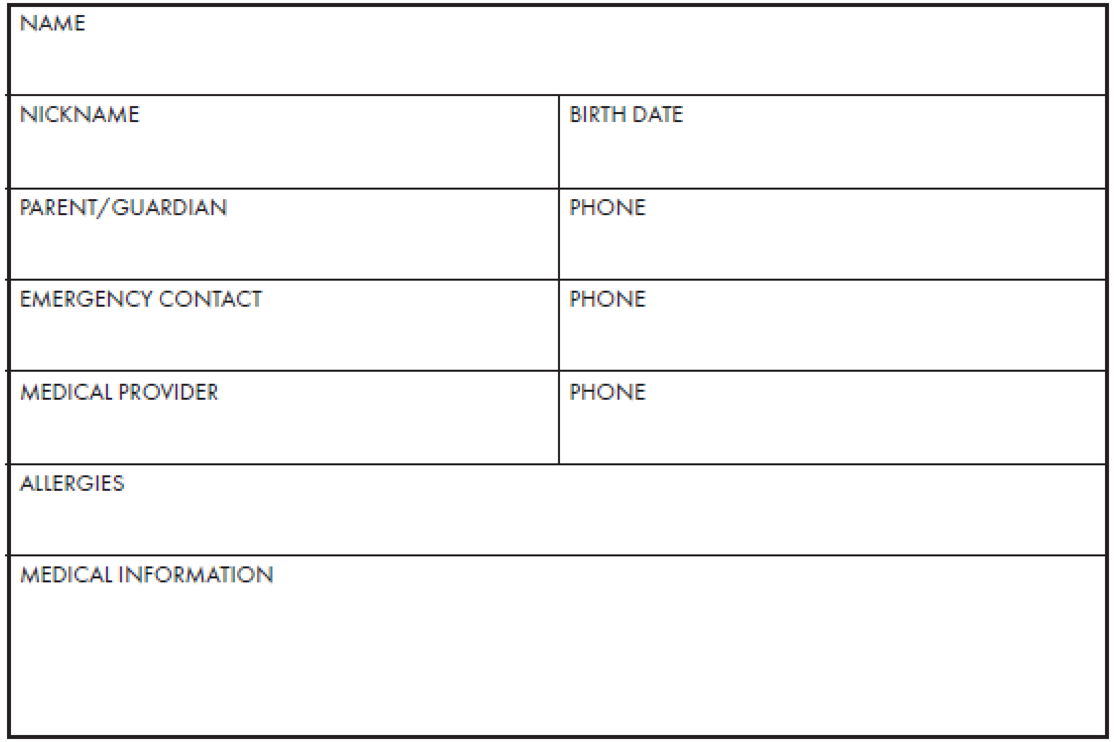
Newborn Screening
Program

Emergency Car Seat Sticker

CHILD EMERGENCY CONTACT

MEDICAL ID CARD ENCLOSED

NORTH Dakota | Health
Be Legendary.™

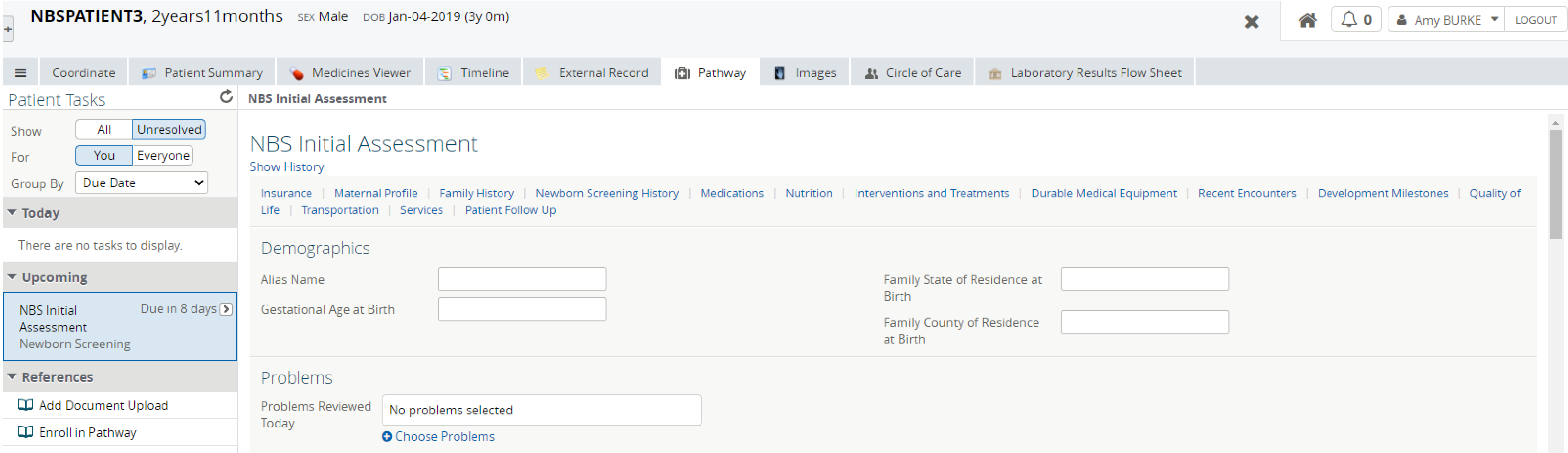


NAME	
NICKNAME	BIRTH DATE
PARENT/GUARDIAN	PHONE
EMERGENCY CONTACT	PHONE
MEDICAL PROVIDER	PHONE
ALLERGIES	
MEDICAL INFORMATION	

North Dakota Long-term Follow-up

- Initial contact with family within 2 weeks of diagnosis
- Quarterly contacts for the first year of life
- Annual contacts through age 6

NBS Care Coordination Pathway | Assessment



NBSPATIENT3, 2years11months SEX Male DOB Jan-04-2019 (3y 0m)

Patient Tasks

Show: All | Unresolved

For: You | Everyone

Group By: Due Date

Today

There are no tasks to display.

Upcoming

NBS Initial Assessment — Due in 8 days
Newborn Screening

References

Add Document Upload

Enroll in Pathway

NBS Initial Assessment

NBS Initial Assessment

Show History

Insurance | Maternal Profile | Family History | Newborn Screening History | Medications | Nutrition | Interventions and Treatments | Durable Medical Equipment | Recent Encounters | Development Milestones | Quality of Life | Transportation | Services | Patient Follow Up

Demographics

Alias Name:

Gestational Age at Birth:

Family State of Residence at Birth:

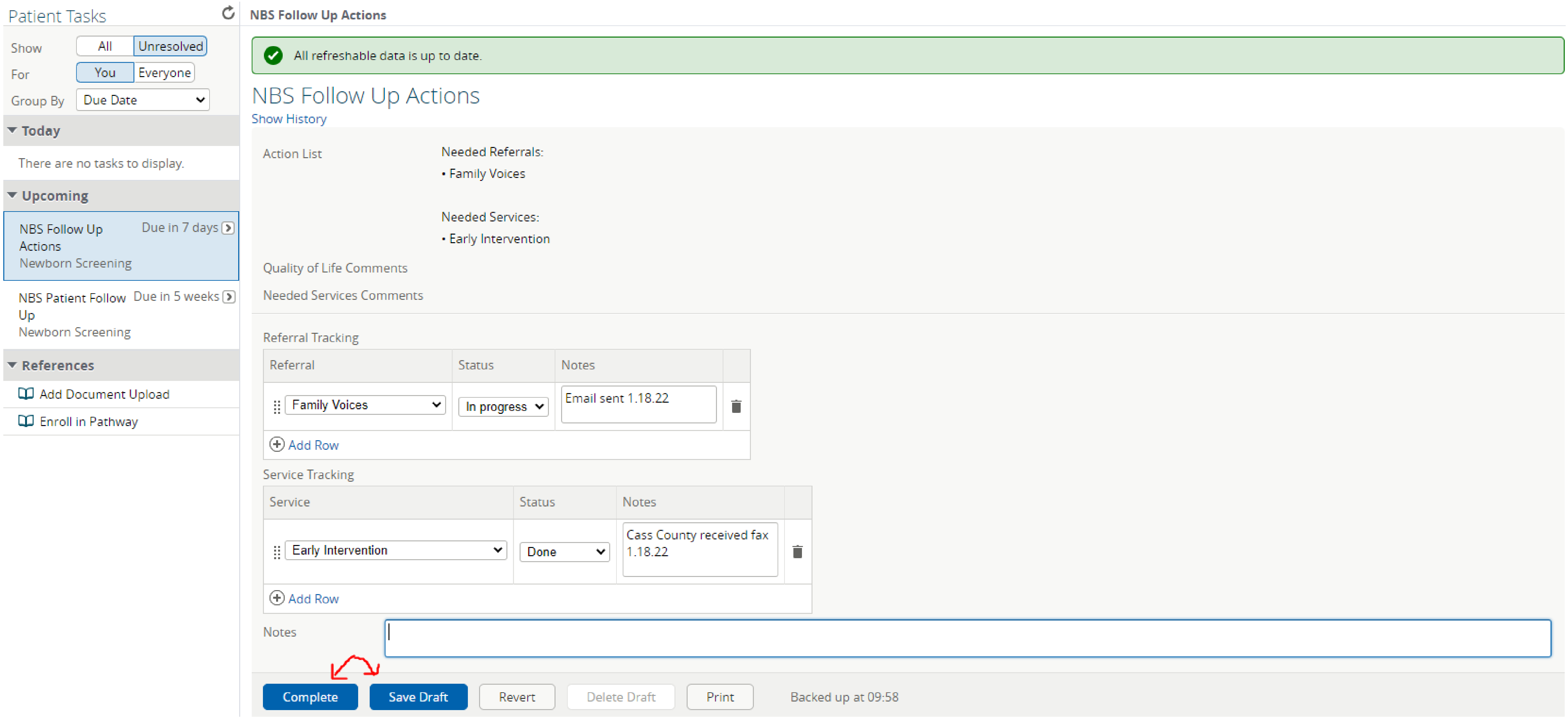
Family County of Residence at Birth:

Problems

Problems Reviewed Today: No problems selected

Choose Problems

NBS Care Coordination Pathway | Referrals



There are no tasks to display.

NBS Follow Up Actions Due in 7 days
Newborn Screening

NBS Patient Follow Up Due in 5 weeks
Newborn Screening

Add Document Upload
Enroll in Pathway

All refreshable data is up to date.

Action List

Needed Referrals:
- Family Voices

Needed Services:
- Early Intervention

Quality of Life Comments

Needed Services Comments

Referral Tracking

Referral	Status	Notes
Family Voices	In progress	Email sent 1.18.22

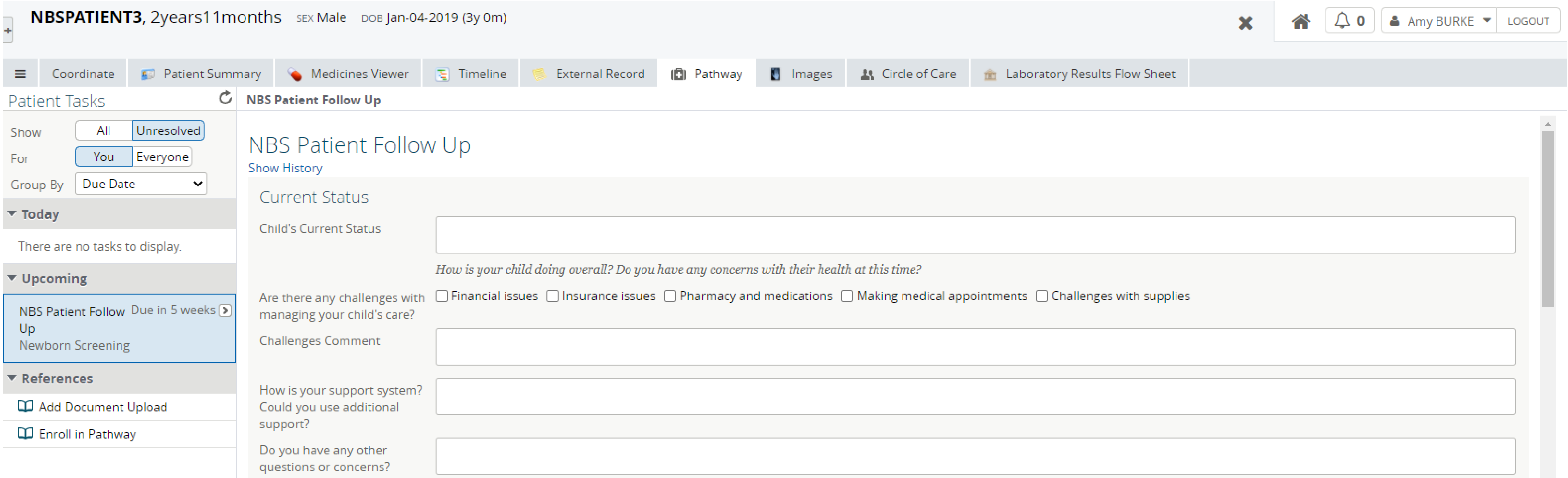
Service Tracking

Service	Status	Notes
Early Intervention	Done	Cass County received fax 1.18.22

Notes

Backed up at 09:58

NBS Care Coordination Pathway | Follow-up



NBSPATIENT3, 2years11months SEX Male DOB Jan-04-2019 (3y 0m)

Patient Tasks

Show: All | Unresolved

For: You | Everyone

Group By: Due Date

Today

There are no tasks to display.

Upcoming

NBS Patient Follow Up — Due in 5 weeks
Newborn Screening

References

NBS Patient Follow Up

NBS Patient Follow Up

Current Status

Child's Current Status

How is your child doing overall? Do you have any concerns with their health at this time?

Are there any challenges with managing your child's care? ☐ Financial issues ☐ Insurance issues ☐ Pharmacy and medications ☐ Making medical appointments ☐ Challenges with supplies

Challenges Comment

How is your support system? Could you use additional support?

Do you have any other questions or concerns?

3 Years of Long-term Follow-up

- Need for outreach clinics – rural nature and long distances, weather impacts – transportation

- Not all families need LTFU, but most have found it beneficial – because their child is doing well

- Many families struggle with navigating insurance, billing, prior authorizations

- There is a great need for additional supportive services for these families

MYTH	FACT
“All my other children are healthy, so this baby won’t have a disorder.”	A disorder can happen in families with no history of disease.
“If my baby is sick, I will know it.”	Most babies with a disorder appear healthy at birth.
“The government is storing the blood spots for research purposes.”	No research will be done on blood spots without parental consent.
“If my baby had a heart condition, it would have been found prenatally.”	Not all cardiac conditions are identified through a prenatal ultrasound.
“I can ring a bell or make loud noises to determine my baby has a hearing loss.”	Many babies with significant hearing loss will respond to sounds.

WHAT IS ON THE HORIZON FOR NBS IN ND?

- New Disorder Expansion
- Expand Services and Outreach to Families
- Contingency Planning for Emergency Preparedness & Response
- Working with National to Improve Services Offered to Families with Children who have Cystic Fibrosis
- Public Outreach Campaign

Newborn Screening's Junior Ambassadors

Thank you!

Joyal Meyer

jbmeyer@nd.gov

NORTH Dakota Be Legendary.

Health & Human Services

AMY GAVIGLIO CGC, MS

DISCLOSURES

- Received speaker fees from Worldwide Clinical Trials & Orchard Therapeutics
- These companies will not be discussed today.

YOU DO WHAT? THE ROLE OF GENETIC COUNSELORS ACROSS THE LIFESPAN

Amy Gaviglio MS CGC

Connetics Consulting

May 3, 2023

DISCLOSURES

Speaking honorariums received from Orchard Therapeutics and Worldwide Clinical Trials.

OUTLINE

- Essential Public Health Services & Public Health Genetic Counseling
- Newborn Screening Journey & the Role of Genetic Counselors
- Future Considerations

STORIES HELP US: WHAT IS NBS TO FAMILIES?

MEET OUR AMBASSADORS

FAMILY LEADERS IN
NEWBORN SCREENING

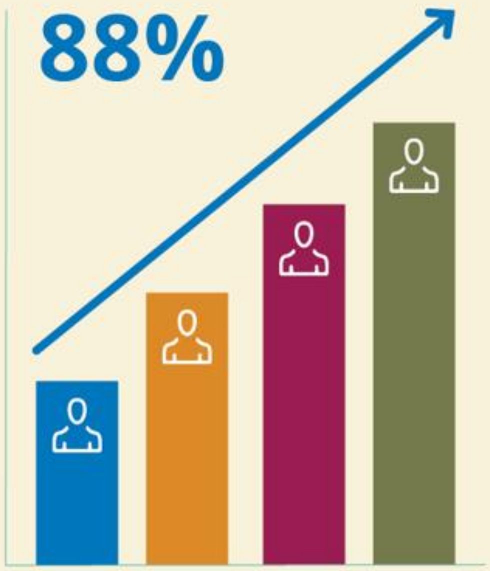
VOLUME 1 2021-2022

https://www.youtube.com/watch?v=nniHSCPLO9k

WHERE ARE GENETIC COUNSELORS?

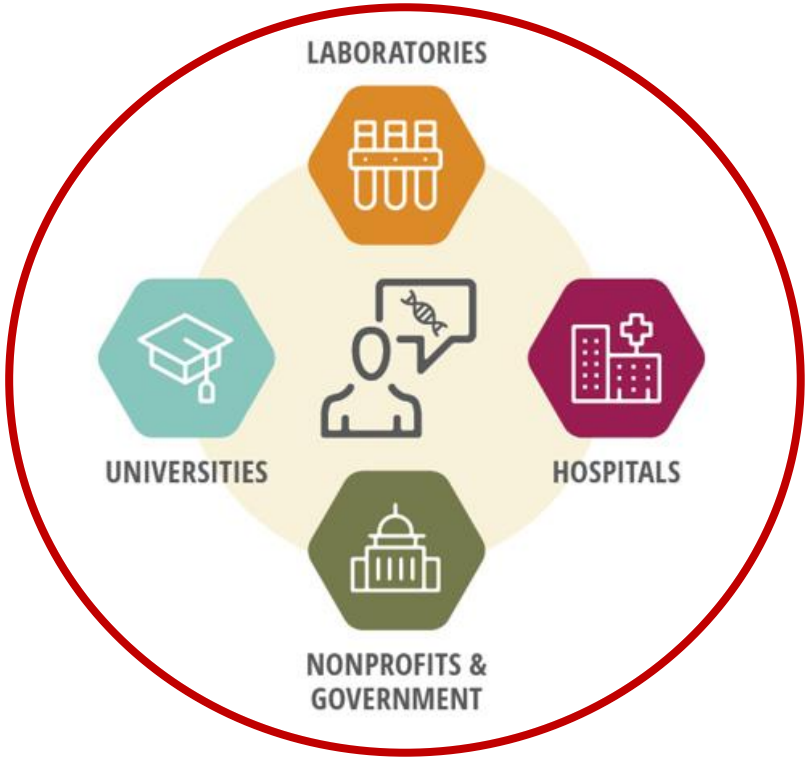
The number of Certified Genetic Counselors (CGCs) continues to rise – there has been an **88% increase since 2011**. By 2030, there are likely to be over 10,000 CGCs.

There are **more than 6,000 CGCs today.**



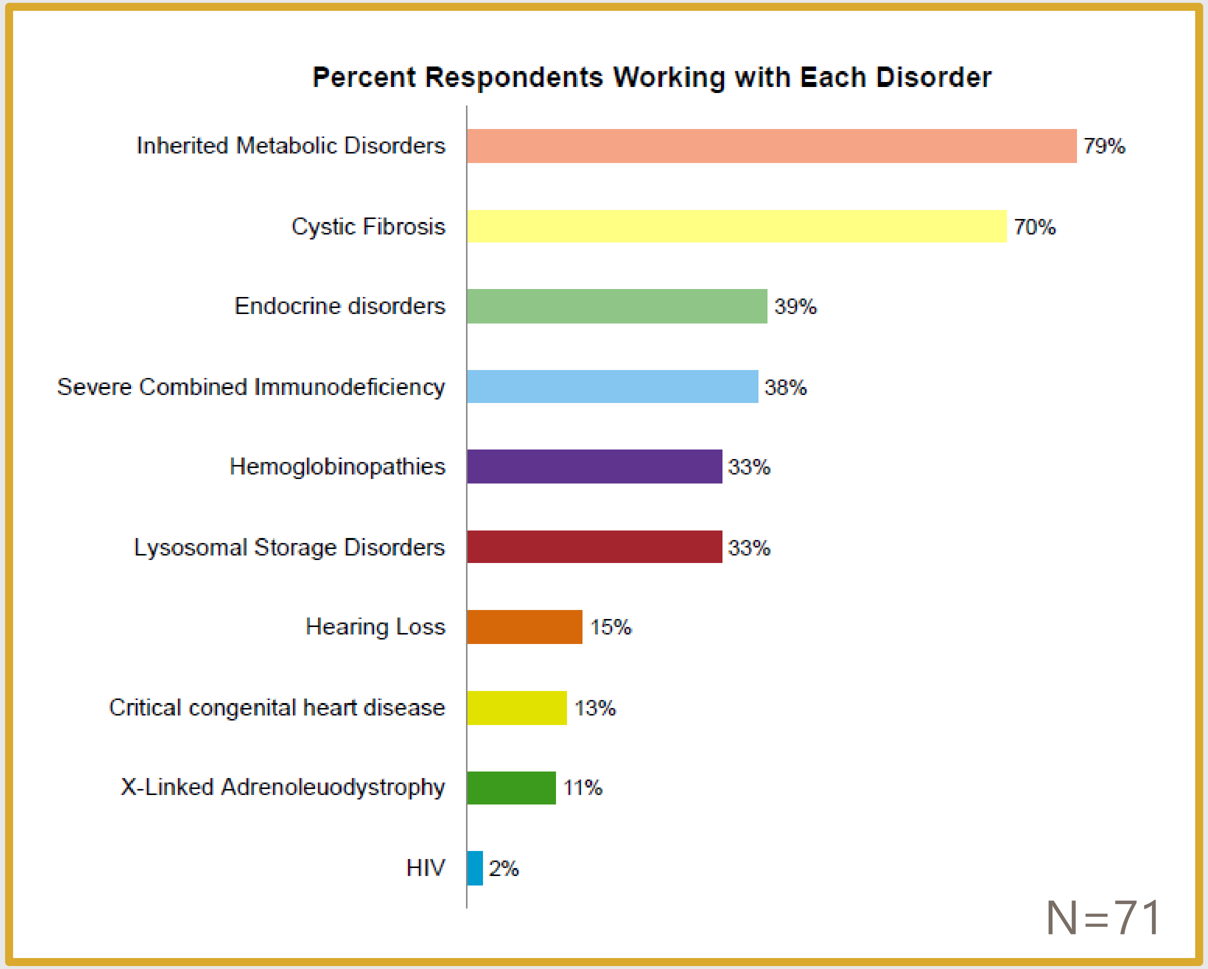
SPECIALTY AREAS

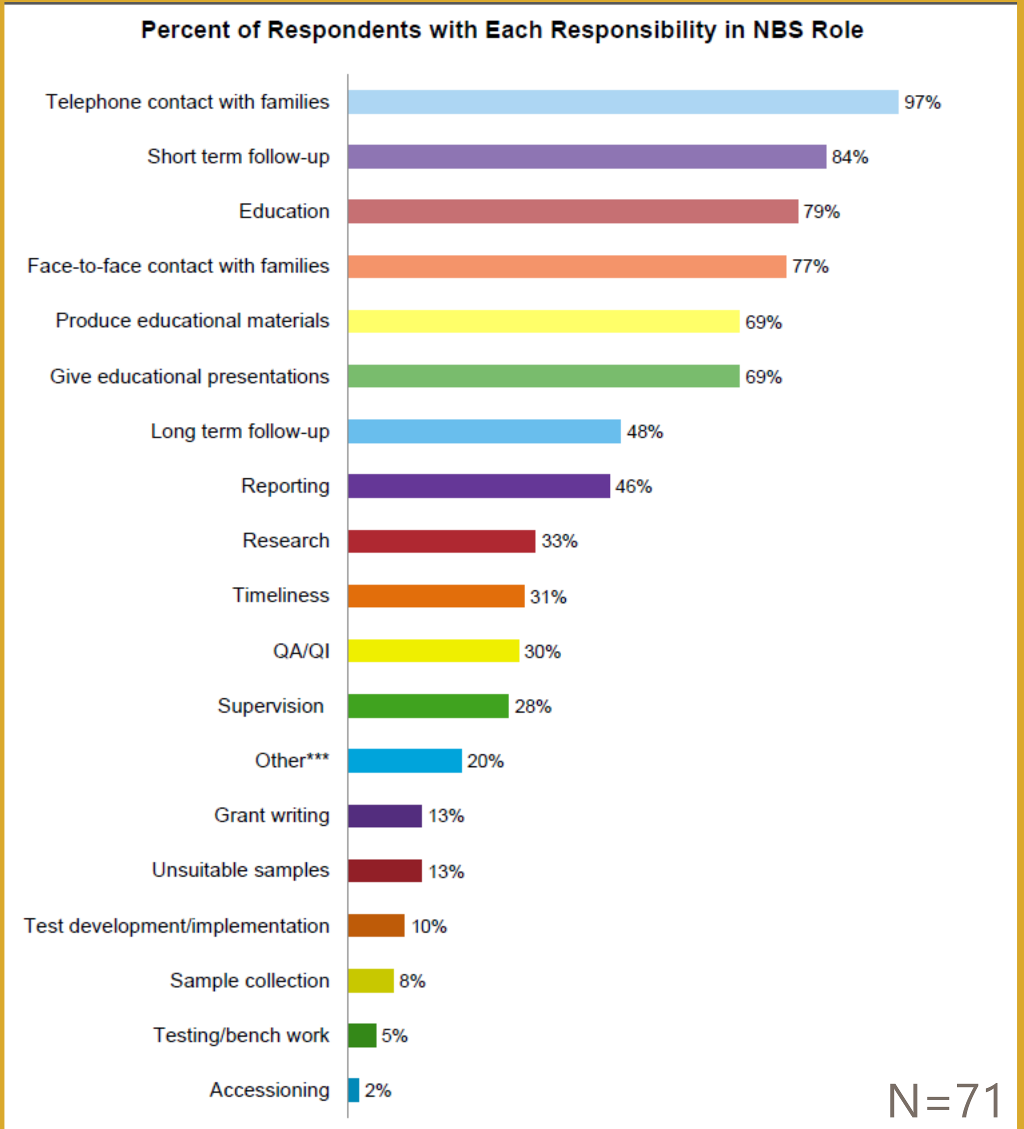
GENETIC COUNSELORS WORK IN A VARIETY OF SETTINGS



2022 NSGC Professional Status Survey

GENETIC COUNSELORS IN OR WORKING WITH NBS PROGRAMS

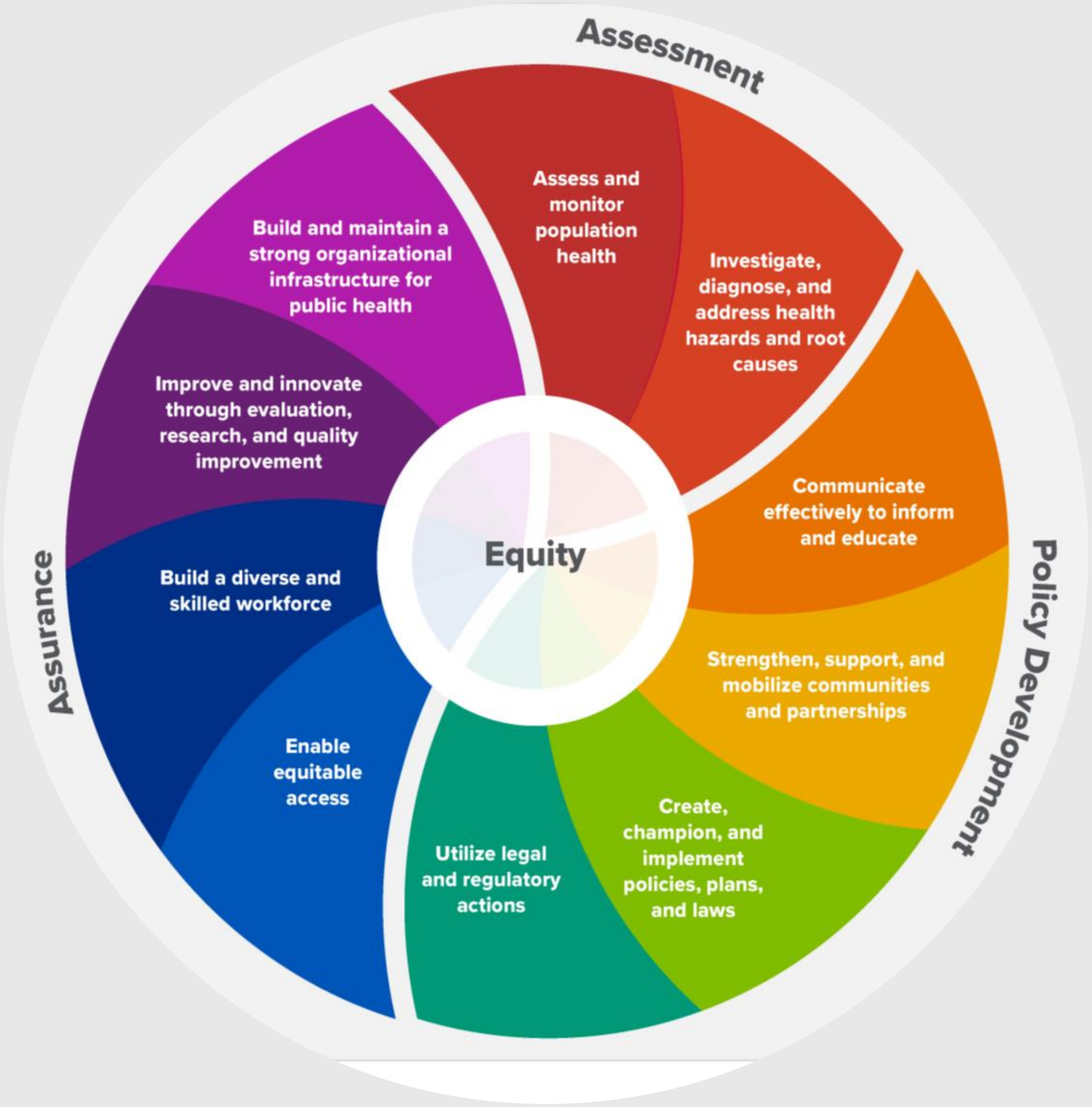

Percent Respondents Working with Each Disorder


Percent of Respondents with Each Responsibility in NBS Role

***Outreach, project development and management, data management, IT website development, IT LIMS development, coordinating sample storage,

Bradley & Vogel. 2016. An Exploration of the Roles and Responsibilities of Genetic Counselors Working With Newborn Screening Programs. APHL NBS Poster

10 ESSENTIAL PUBLIC HEALTH SERVICES



Genetic Counseling Practice-Based Competency Domains:

1. **Genetics Expertise and Analysis**
2. **Interpersonal, Psychosocial, and Counseling Skills**
3. **Education**
4. **Professional Development and Practice**

https://www.cdc.gov/publichealthgateway/publichealthservices/essentialhealthservices.html

Institutions

- Hospitals;
- Community health centers;
- Higher education programs

Examples

- FQHCs
- CEPH
- ACGC

Organizations

- Non-profit advocacy organizations;
- Non-profit member organizations;
- Genetic testing companies;
- Grant programs

Examples

- NORD
- NSGC
- NCC

Public Health Agencies

- Local health departments;
- State health departments;
- Federal agencies

Examples

- State Public Health Genomics Programs
- HRSA
- CDC

Researchers

- Newborn screening researchers;
- Carrier testing researchers;
- Clinical researchers;
- Basic researchers

Individuals

- Individuals with genetic conditions;
- Families with a history of genetic conditions;
- Caregivers
- General Public

Genetics Providers

- Geneticist
- Genetic counselors
- Genetic nurses
- Metabolic dietitians

Non-Genetics Healthcare Providers

- Primary care providers;
- Specialty providers (oncologists, neurologists, cardiologists, etc.)
- Other healthcare professionals (healthcare interpreters, occupational therapists, etc.)

Public Health Professionals

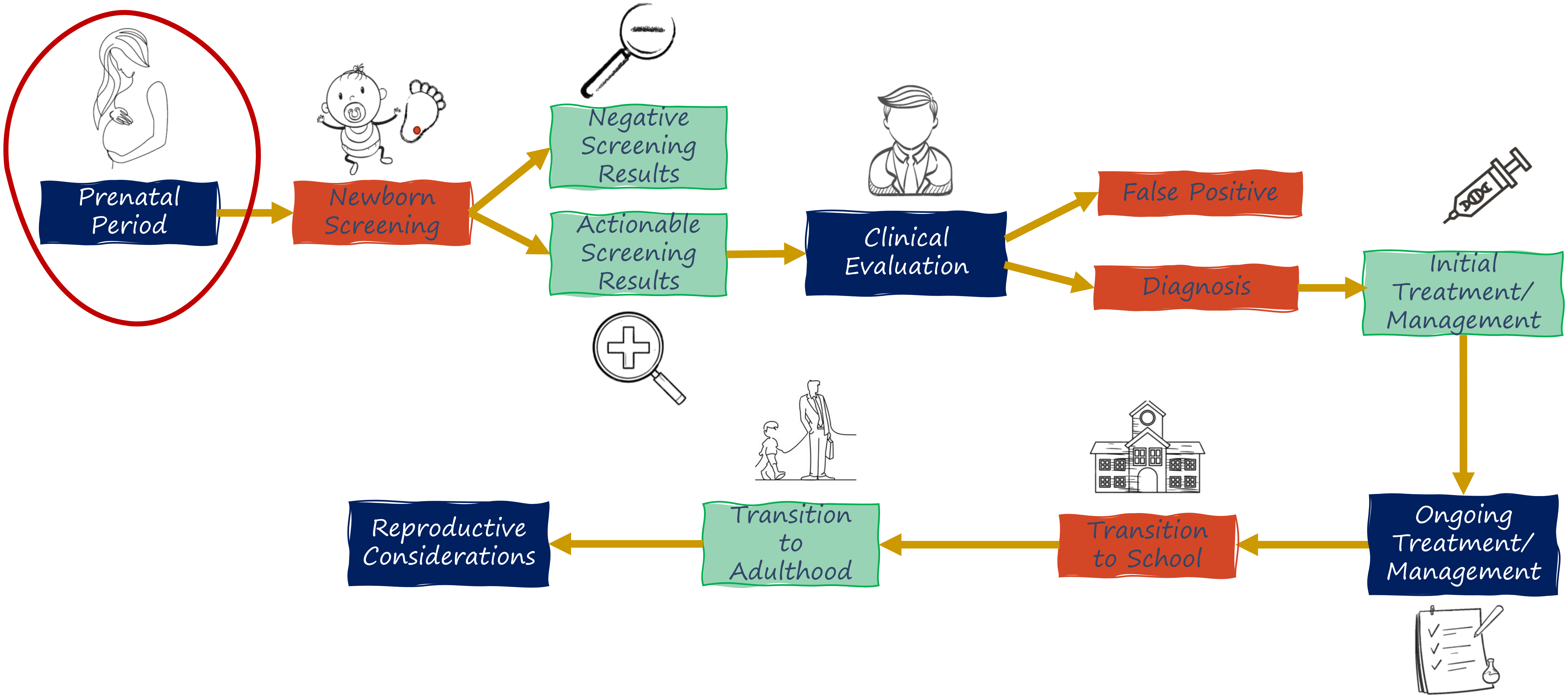
- Local and state health department officials;
- Laboratorians;
- Health education specialists;
- Federal public health professionals

NEWBORN SCREENING JOURNEY & GENETIC COUNSELING

GENETIC COUNSELORS FILL MANY ROLES WITHIN AND RELATED TO NBS

- State Genetic Services programs/coordinators
- State Newborn Screening Advisory Board Members
- NORD/Genetic Alliance/non-profits that support families
- Patient and professional education
- Laboratory liaisons
- NBS data management
- Screening result notification and case management
- QI/QC
- Clinical service provision – short and long term
- Clinical research/trials
- Industry/Pharma

THE NBS AND RARE DISEASE JOURNEY



Prenatal Period

Newborn Screening

Negative Screening Results

Actionable Screening Results

Clinical Evaluation

False Positive

Diagnosis

Initial Treatment/ Management

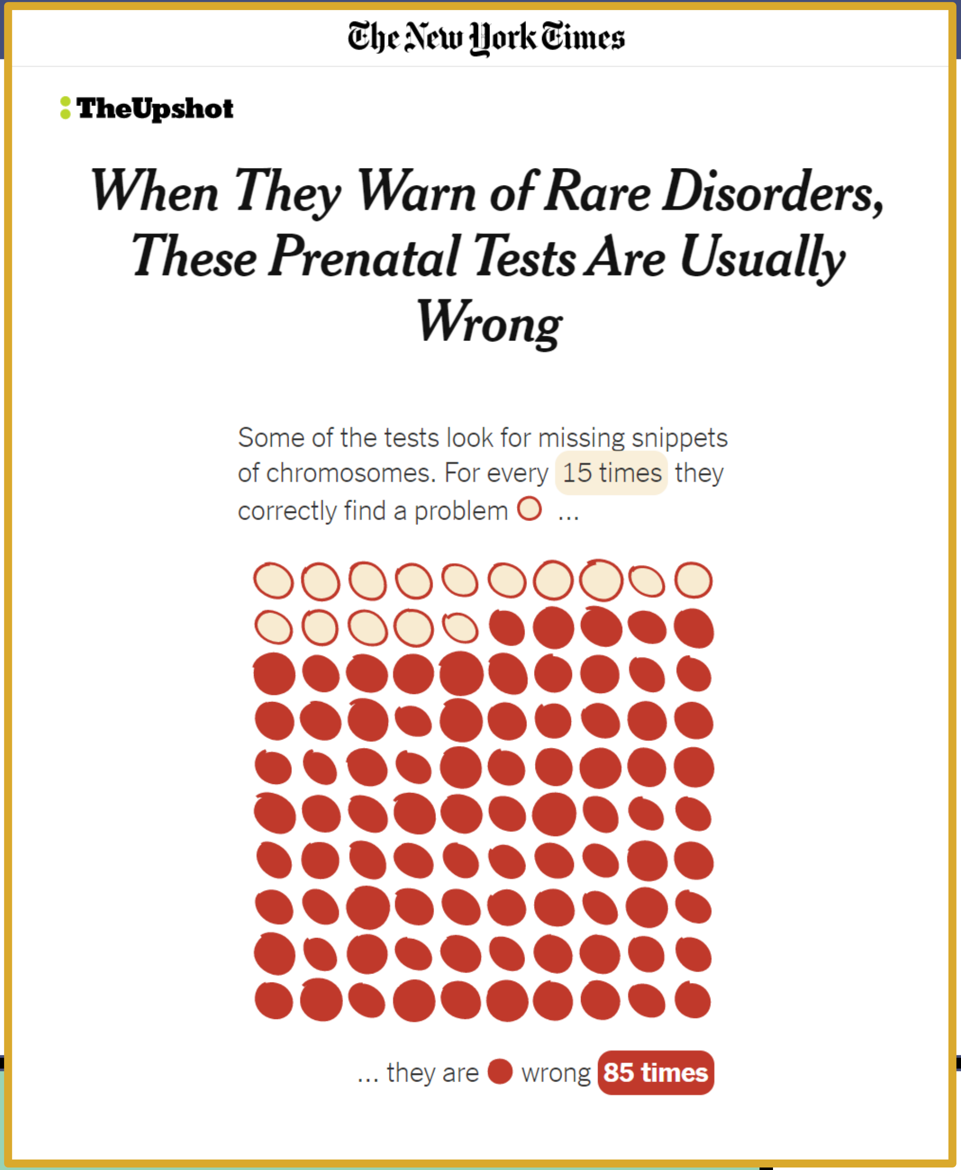
Ongoing Treatment/ Management

Transition to School

Transition to Adulthood

Reproductive Considerations

SCREENING VS DIAGNOSIS



The New York Times

TheUpshot

When They Warn of Rare Disorders, These Prenatal Tests Are Usually Wrong

Some of the tests look for missing snippets of chromosomes. For every 15 times they correctly find a problem ○ …

… they are ● wrong **85 times**

Forbes

EDITORS' PICK

What The NYTimes Got Wrong On Prenatal Screening

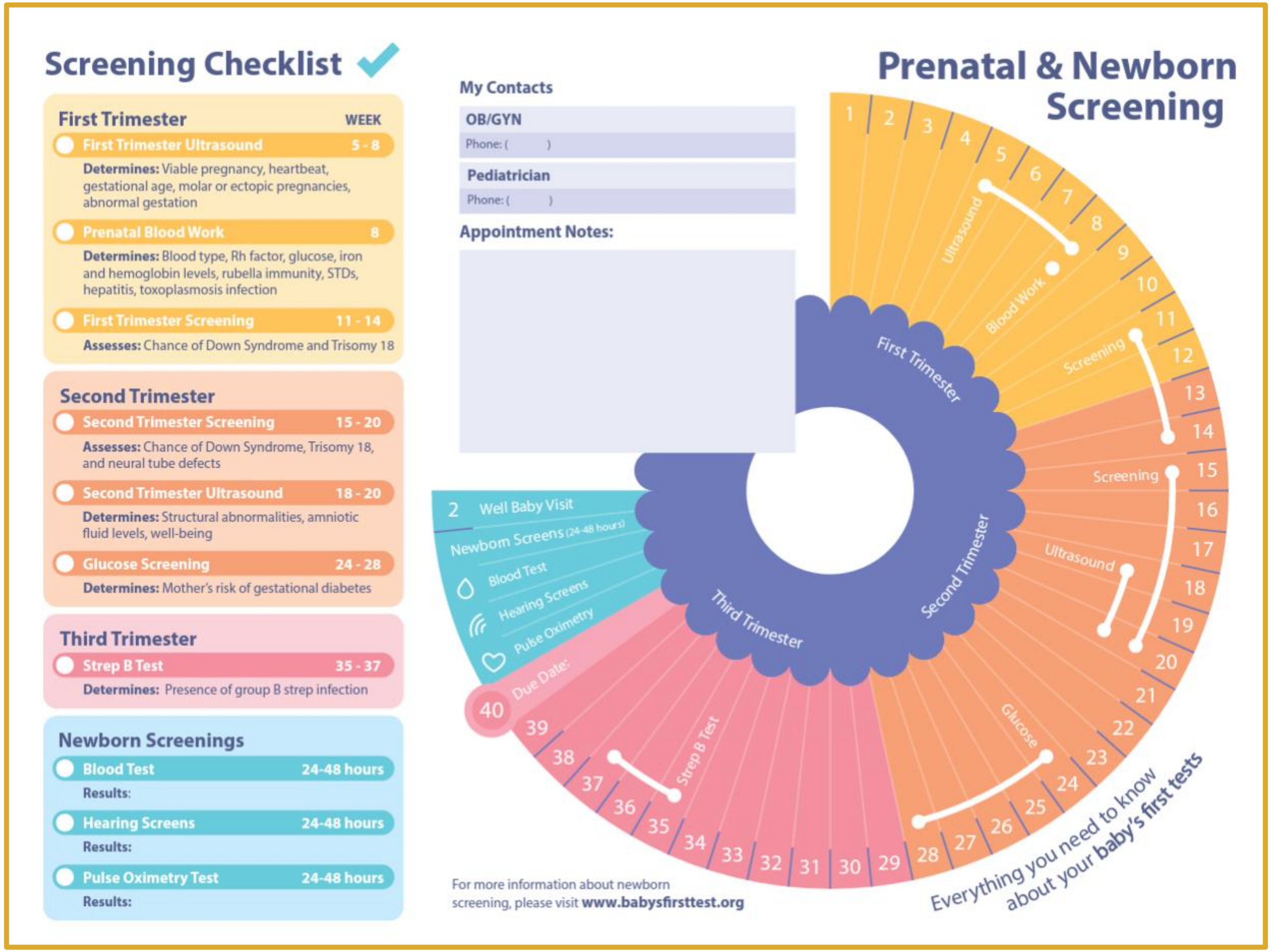
Ellen Matloff Contributor ⓘ

I cover genetic counseling, testing and digital health.

SCREENING VS DIAGNOSTIC TESTS

	Screening Test	**Diagnostic Test**
Population (offered the test)	• Those **without symptoms** of disease where early detection is important	• Those **with symptoms** • Those undergoing further work-up **after a positive screening test**
Results	• Result is an **estimate of level of risk** • Determines whether a diagnostic test is warranted	• Result usually provides a more **definitive diagnosis**
Test Metrics	• Cutoffs set towards high **sensitivity** • Acceptance of **false positive** results	• Cutoffs set towards high **specificity** • Higher **precision and accuracy**
Invasiveness	• Usually **non-invasive**	• May be **invasive**

SPECTRUM OF PRENATAL TO NEONATAL SCREENING



Screening Checklist ✓

First Trimester WEEK

○ **First Trimester Ultrasound** 5 - 8

Determines: Viable pregnancy, heartbeat, gestational age, molar or ectopic pregnancies, abnormal gestation

○ **Prenatal Blood Work** 8

Determines: Blood type, Rh factor, glucose, iron and hemoglobin levels, rubella immunity, STDs, hepatitis, toxoplasmosis infection

○ **First Trimester Screening** 11 - 14

Assesses: Chance of Down Syndrome and Trisomy 18

Second Trimester

○ **Second Trimester Screening** 15 - 20

Assesses: Chance of Down Syndrome, Trisomy 18, and neural tube defects

○ **Second Trimester Ultrasound** 18 - 20

Determines: Structural abnormalities, amniotic fluid levels, well-being

○ **Glucose Screening** 24 - 28

Determines: Mother's risk of gestational diabetes

Third Trimester

○ **Strep B Test** 35 - 37

Determines: Presence of group B strep infection

Newborn Screenings

○ **Blood Test** 24-48 hours

Results:

○ **Hearing Screens** 24-48 hours

Results:

○ **Pulse Oximetry Test** 24-48 hours

Results:

My Contacts

OB/GYN

Phone: (   )

Pediatrician

Phone: (   )

Appointment Notes:

Prenatal & Newborn Screening

For more information about newborn screening, please visit **www.babysfirsttest.org**

https://babysfirsttest.org/newborn-screening/resources/prenatal-and-newborn-screening-brochure

INTERSECTION OF PRENATAL & NEONATAL SCREENING

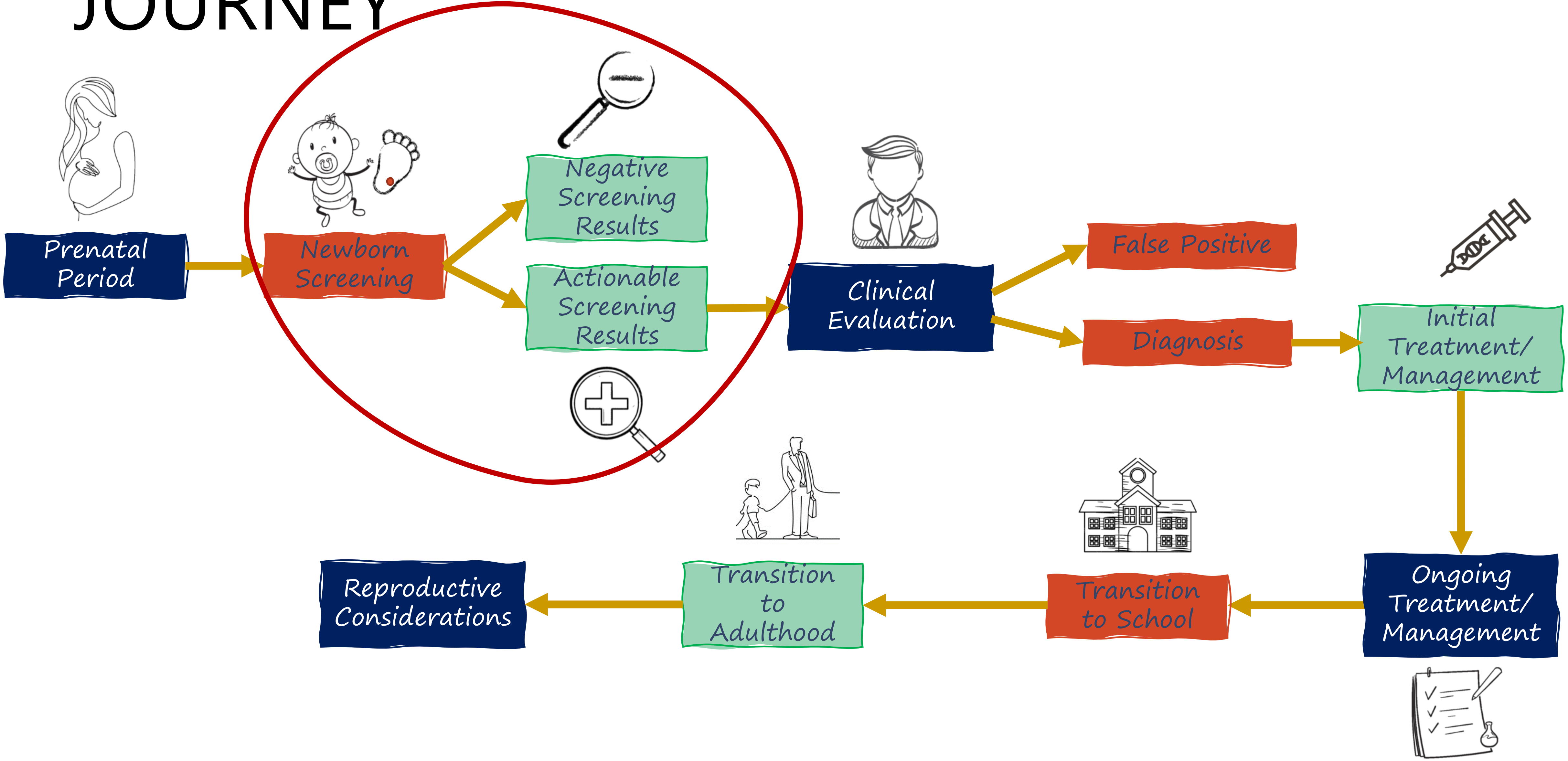
- **Sickle Cell Disease**
  - A SickleDex may be used, which has risk for FPs and FNs
- **Cystic Fibrosis**
  - Variant panel utilized prenatally may differ from what is utilized within the NBS program
  - False sense of security may be provided prenatally
- **Spinal Muscular Atrophy**
  - Most NBS programs will not detect carriers

WHAT PARENTS WANT TO KNOW IN PRENATAL PERIOD: COMMUNICATION ROLE FOR GENETIC COUNSELORS

- All newborn babies get tested for some rare disorders before they leave the hospital.
- Babies with these diseases may look at healthy at birth.
- Serious problems can be prevented if we find out about the disease right away.
- To do the test, a few drops of blood will be taken from your baby's heel.
- Your baby's doctor and birth facility will get a copy of the test results. Ask about the results at your first visit.
- Some babies need to be retested. If your baby needs to be retested you will be notified.
- Talk to your baby's healthcare provider if you have questions.

Rose NC, Dolan SM. Newborn screening and the obstetrician. Obstet Gynecol. 2012 Oct;120(4):908-17

THE NBS AND RARE DISEASE JOURNEY



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Transition to Adulthood

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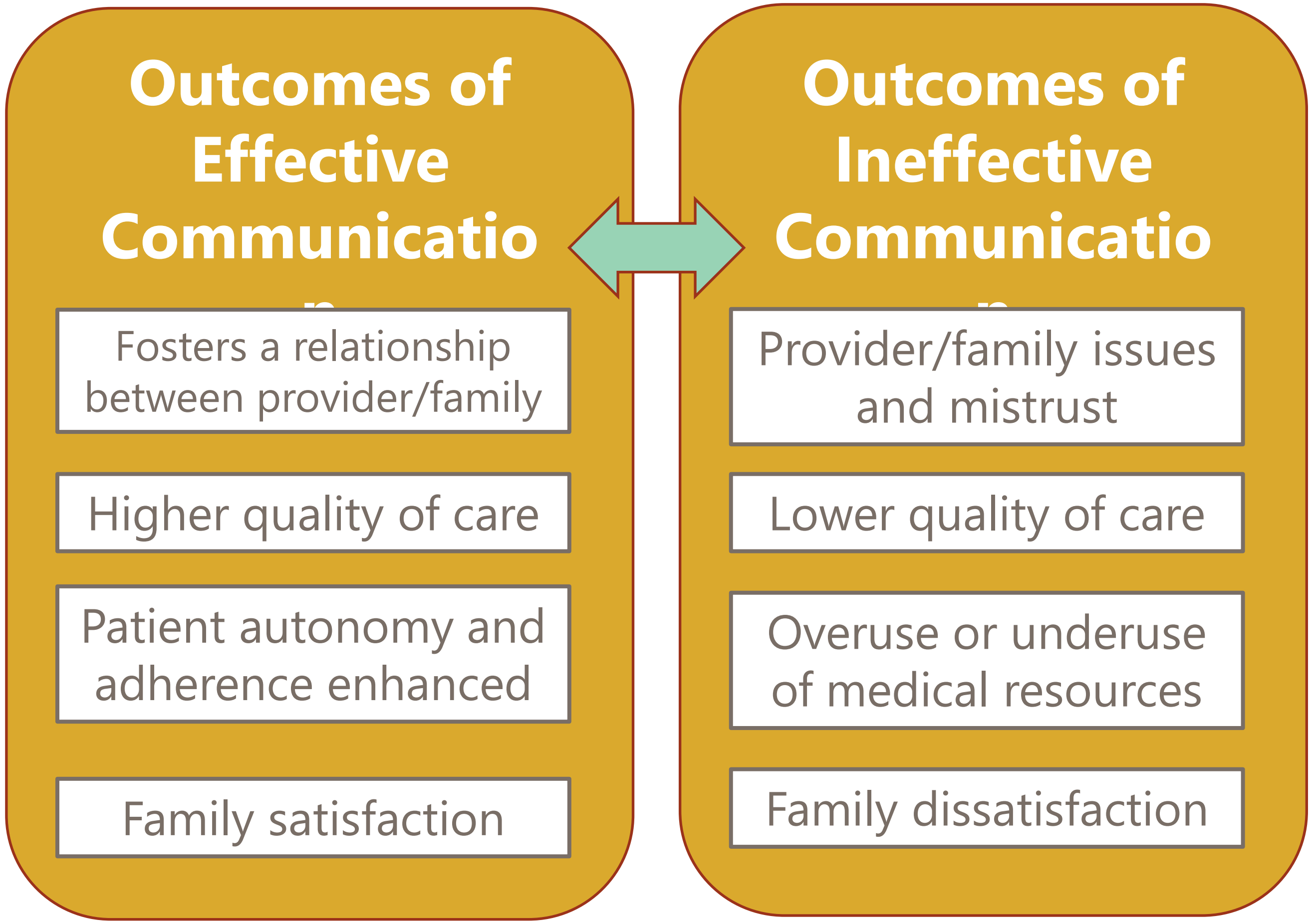
IMPORTANCE OF EFFECTIVE COMMUNICATION

#1 ISSUE IN HEALTHCARE COMMUNICATION IS THE ILLUSION IT HAS TAKEN PLACE

"Communicating NBS results is a delicate task and should be carefully and appropriately crafted to prepare for a range of outcomes, which could vary considerably given the wide range of clinical profiles of positive cases."

Fusco, Francesco et al. "Delivering Positive Newborn Screening Results: Cost Analysis of Existing Practice versus Innovative, Co-Designed Strategies from the ReSPoND Study." International journal of neonatal screening vol. 8,1 19. 14 Mar. 2022.

IMPORTANCE OF EFFECTIVE COMMUNICATION



Outcomes of Effective Communication

- Fosters a relationship between provider/family
- Higher quality of care
- Patient autonomy and adherence enhanced
- Family satisfaction

Outcomes of Ineffective Communication

- Provider/family issues and mistrust
- Lower quality of care
- Overuse or underuse of medical resources
- Family dissatisfaction

These individuals are our conduits to our main partners… the families

- Our screening results are only as good as the follow-up done
- Fact sheets/resources are only as good as their dissemination

Adapted from Rocque, R. 2015. PLOS ONE 10(10): e0139577

Family Quotes

Conway, M., Vuong, T. T., Hart, K., Rohrwasser, A., & Eilbeck, K. (2022). Pain points in parents' interactions with newborn screening systems: a qualitative study. *BMC pediatrics*, *22*(1), 167.

Bani, M., et al. (2023). Parents' experience of the communication process of positivity at newborn screening for metabolic diseases: A qualitative study. *Child: Care, Health and Development*, 1– 11.

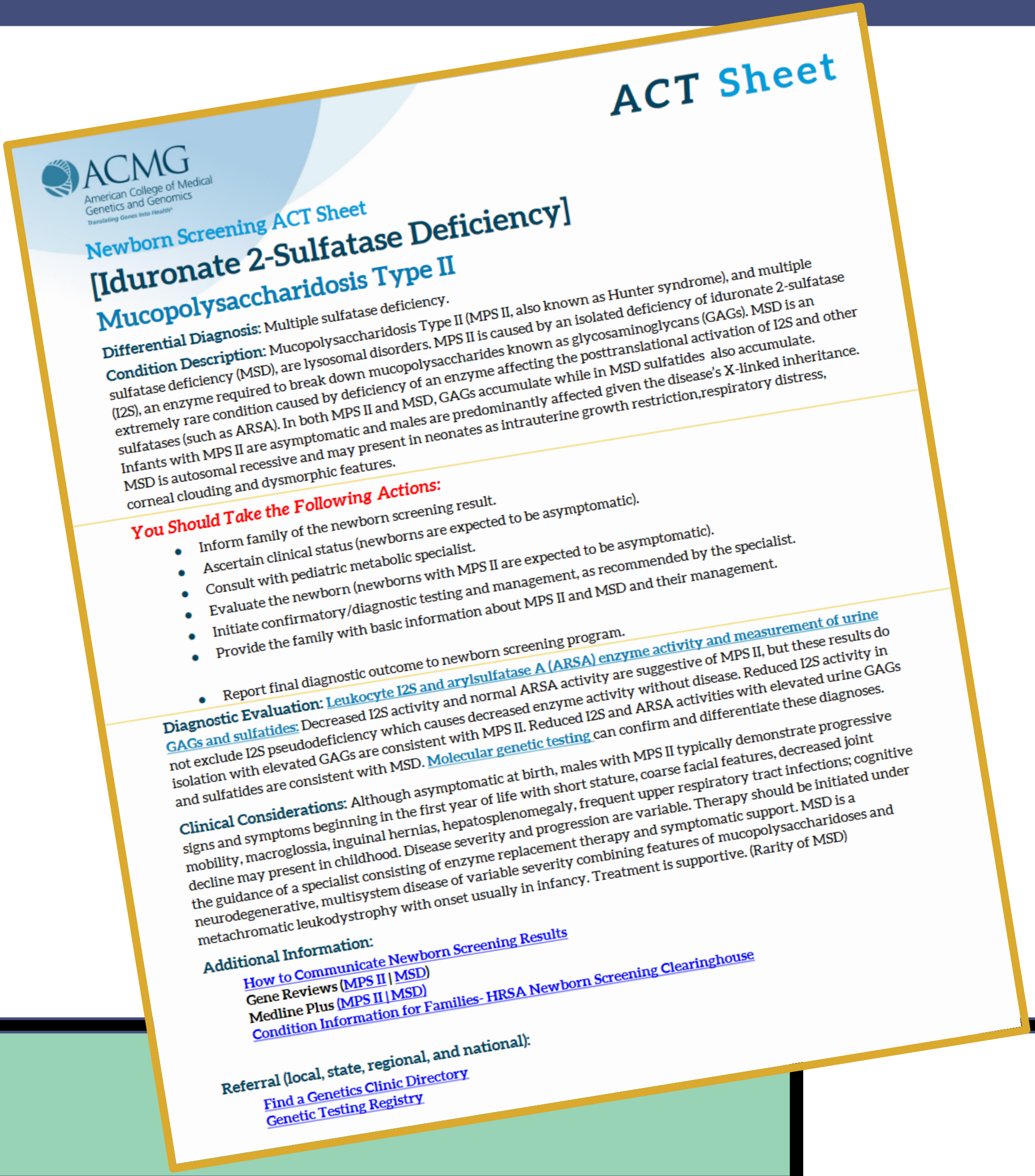
“We were left to ourselves and so we went on Google, which I know is wrong, and on the Internet I obviously found unpleasant things, therefore I had a moment of discomfort.”

“… the individual who communicated the results was the receptionist from the pediatrician’s office … [She] said that there had been a positive for PKU. My wife asked what that was and the receptionist replied that she didn’t know, but it meant that our baby could be retarded. So that was obviously traumatic.”

“It was very difficult. One of the things I did, being we live in the middle of nowhere and there was such little knowledge from everyone, is I called almost daily for a week until I got someone who would actually talk to me. I called constantly to find someone to talk to because no one else knew anything.”

“The communication was fully verbal. As a parent, you feel anxious, and because of that, I missed some pieces of what was being said, I also wish I could record them. If I had that information written on paper, I could return to it afterwards with a clearer mind.”

RESULT COMMUNICATION SHEETS



ACT Sheet

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health

Newborn Screening ACT Sheet

[Iduronate 2-Sulfatase Deficiency]

Mucopolysaccharidosis Type II

Differential Diagnosis: Multiple sulfatase deficiency.

Condition Description: Mucopolysaccharidosis Type II (MPS II, also known as Hunter syndrome), and multiple sulfatase deficiency (MSD), are lysosomal disorders. MPS II is caused by an isolated deficiency of iduronate 2-sulfatase (I2S), an enzyme required to break down mucopolysaccharides known as glycosaminoglycans (GAGs). MSD is an extremely rare condition caused by deficiency of an enzyme affecting the posttranslational activation of I2S and other sulfatases (such as ARSA). In both MPS II and MSD, GAGs accumulate while in MSD sulfatides also accumulate. Infants with MPS II are asymptomatic and males are predominantly affected given the disease's X-linked inheritance. MSD is autosomal recessive and may present in neonates as intrauterine growth restriction,respiratory distress, corneal clouding and dysmorphic features.

You Should Take the Following Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (newborns are expected to be asymptomatic).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (newborns with MPS II are expected to be asymptomatic).
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide the family with basic information about MPS II and MSD and their management.
- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Leukocyte I2S and arylsulfatase A (ARSA) enzyme activity and measurement of urine GAGs and sulfatides: Decreased I2S activity and normal ARSA activity are suggestive of MPS II, but these results do not exclude I2S pseudodeficiency which causes decreased enzyme activity without disease. Reduced I2S activity in isolation with elevated GAGs are consistent with MPS II. Reduced I2S and ARSA activities with elevated urine GAGs and sulfatides are consistent with MSD. Molecular genetic testing can confirm and differentiate these diagnoses.

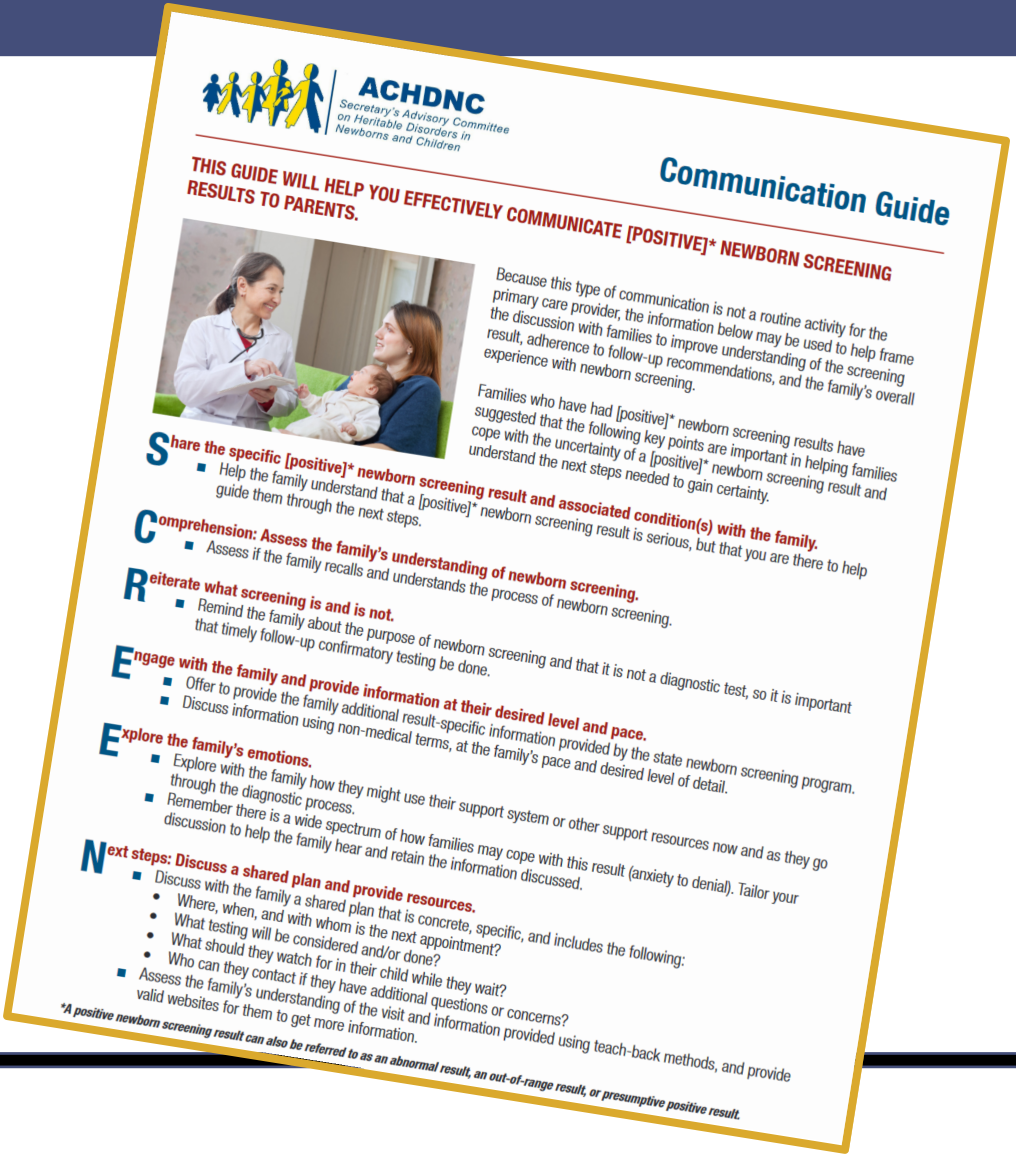
Clinical Considerations: Although asymptomatic at birth, males with MPS II typically demonstrate progressive signs and symptoms beginning in the first year of life with short stature, coarse facial features, decreased joint mobility, macroglossia, inguinal hernias, hepatosplenomegaly, frequent upper respiratory tract infections; cognitive decline may present in childhood. Disease severity and progression are variable. Therapy should be initiated under the guidance of a specialist consisting of enzyme replacement therapy and symptomatic support. MSD is a neurodegenerative, multisystem disease of variable severity combining features of mucopolysaccharidoses and metachromatic leukodystrophy with onset usually in infancy. Treatment is supportive. (Rarity of MSD)

Additional Information:

How to Communicate Newborn Screening Results
Gene Reviews (MPS II | MSD)
Medline Plus (MPS II | MSD)
Condition Information for Families- HRSA Newborn Screening Clearinghouse

Referral (local, state, regional, and national):

Find a Genetics Clinic Directory
Genetic Testing Registry



ACHDNC
Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

Communication Guide

THIS GUIDE WILL HELP YOU EFFECTIVELY COMMUNICATE [POSITIVE]* NEWBORN SCREENING RESULTS TO PARENTS.

Because this type of communication is not a routine activity for the primary care provider, the information below may be used to help frame the discussion with families to improve understanding of the screening result, adherence to follow-up recommendations, and the family's overall experience with newborn screening.

Families who have had [positive]* newborn screening results have suggested that the following key points are important in helping families cope with the uncertainty of a [positive]* newborn screening result and understand the next steps needed to gain certainty.

Share the specific [positive]* newborn screening result and associated condition(s) with the family.
- Help the family understand that a [positive]* newborn screening result is serious, but that you are there to help guide them through the next steps.

Comprehension: Assess the family's understanding of newborn screening.
- Assess if the family recalls and understands the process of newborn screening.

Reiterate what screening is and is not.
- Remind the family about the purpose of newborn screening and that it is not a diagnostic test, so it is important that timely follow-up confirmatory testing be done.

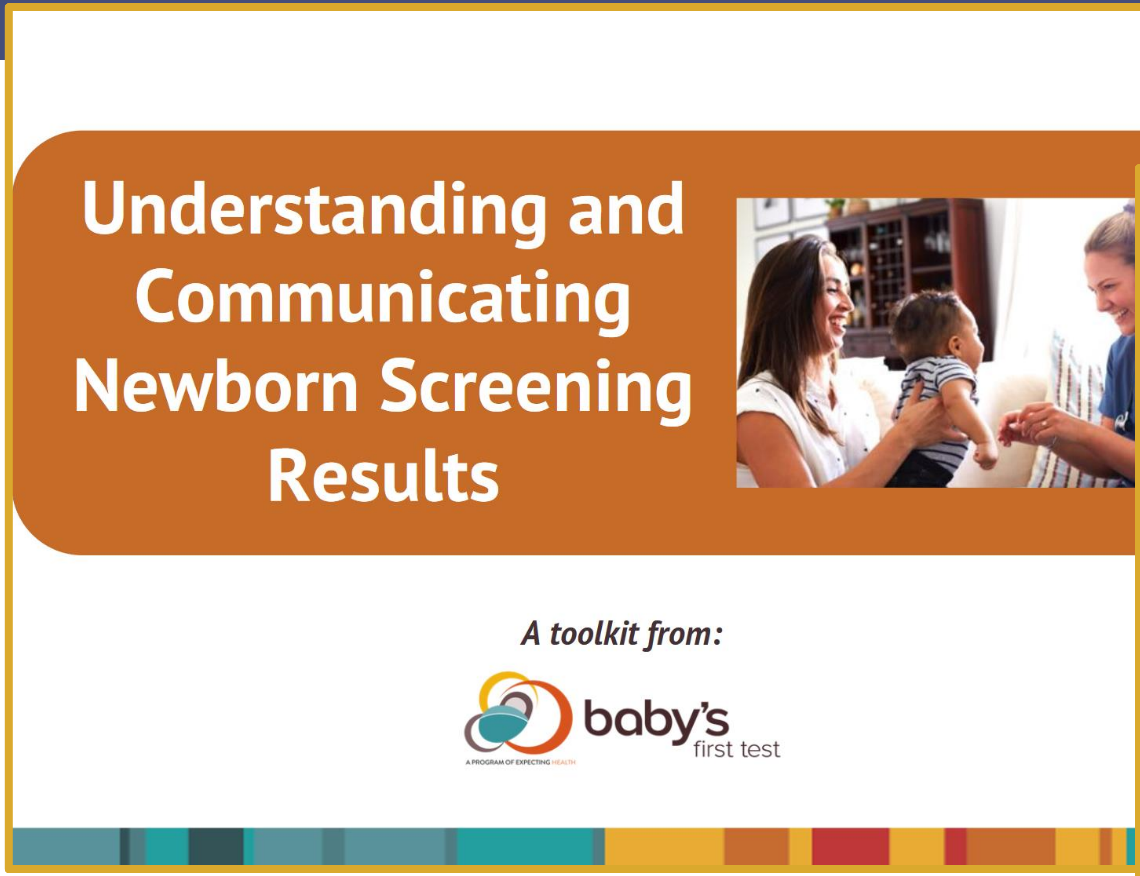
Engage with the family and provide information at their desired level and pace.
- Offer to provide the family additional result-specific information provided by the state newborn screening program.
- Discuss information using non-medical terms, at the family's pace and desired level of detail.

Explore the family's emotions.
- Explore with the family how they might use their support system or other support resources now and as they go through the diagnostic process.
- Remember there is a wide spectrum of how families may cope with this result (anxiety to denial). Tailor your discussion to help the family hear and retain the information discussed.

Next steps: Discuss a shared plan and provide resources.
- Discuss with the family a shared plan that is concrete, specific, and includes the following:
  - Where, when, and with whom is the next appointment?
  - What testing will be considered and/or done?
  - What should they watch for in their child while they wait?
  - Who can they contact if they have additional questions or concerns?
- Assess the family's understanding of the visit and information provided using teach-back methods, and provide valid websites for them to get more information.

*A positive newborn screening result can also be referred to as an abnormal result, an out-of-range result, or presumptive positive result.

BABY'S FIRST TEST RESULT COMMUNICATION TOOLKIT

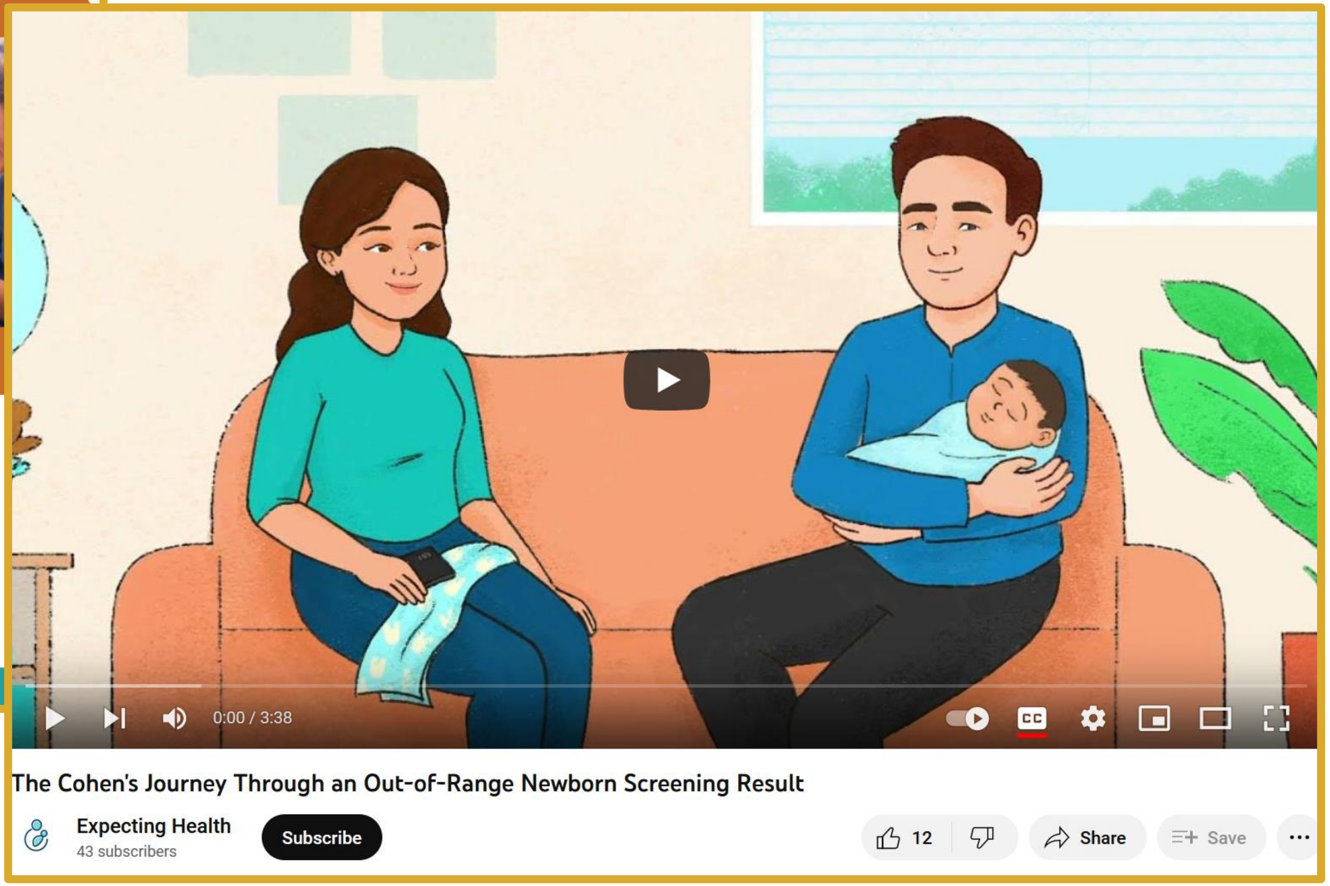


Understanding and Communicating Newborn Screening Results

A toolkit from:

baby's first test

A PROGRAM OF EXPECTING HEALTH



The Cohen's Journey Through an Out-of-Range Newborn Screening Result

Expecting Health

43 subscribers

https://www.babysfirsttest.org/sites/default/files/Results_Toolkit.pdf

https://www.youtube.com/watch?v=cmWvANOlSNk

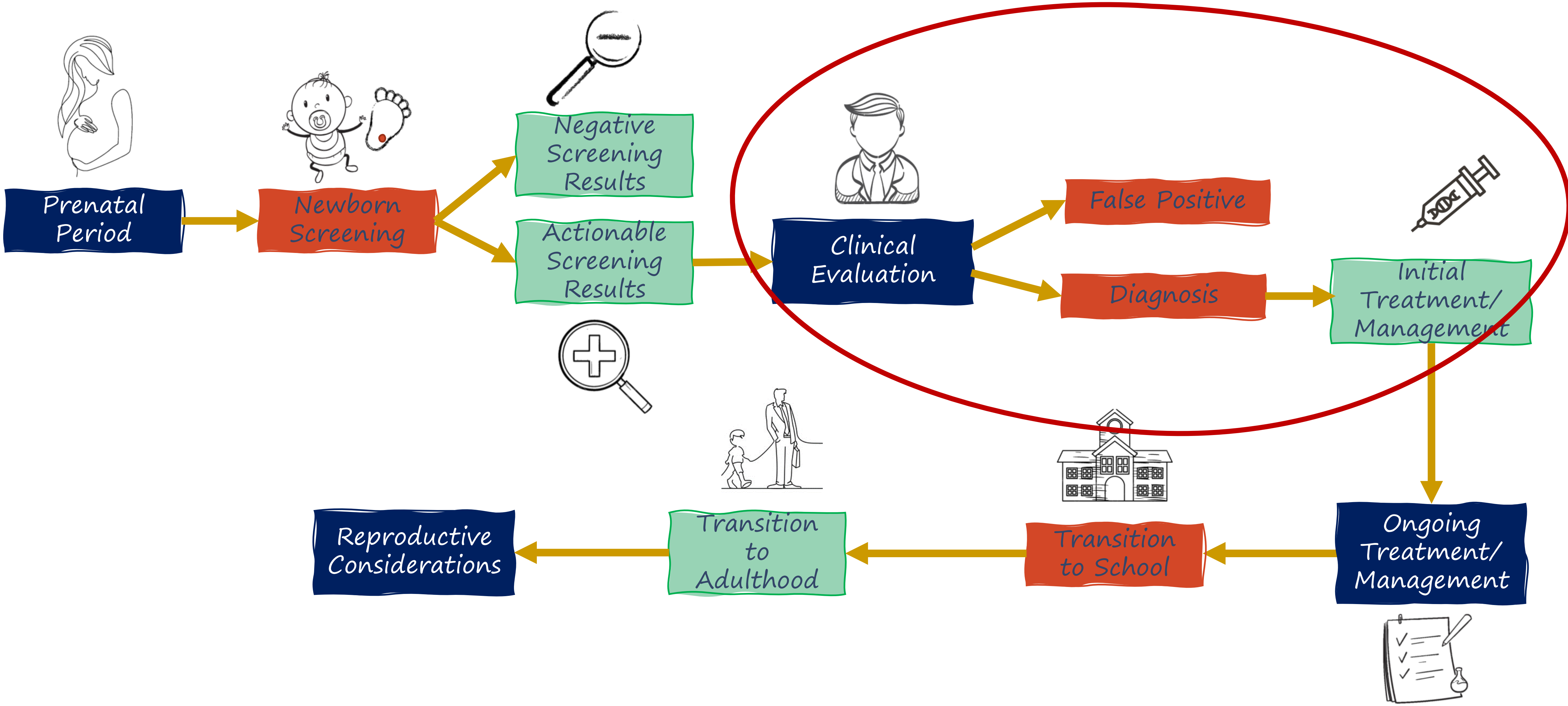
ADDITIONAL COMMUNICATION CONSIDERATIONS

- **For the love of all things… don't tell parents, "Don't Google it"**
  - Provide reliable and updated resources instead

- **Provide Evidence-Based Hope**

- **There is a reason we are doing NBS**
  - NBS-identified families react differently than clinically-identified families – may need more support in accepting disease and treatments

THE NBS AND RARE DISEASE JOURNEY



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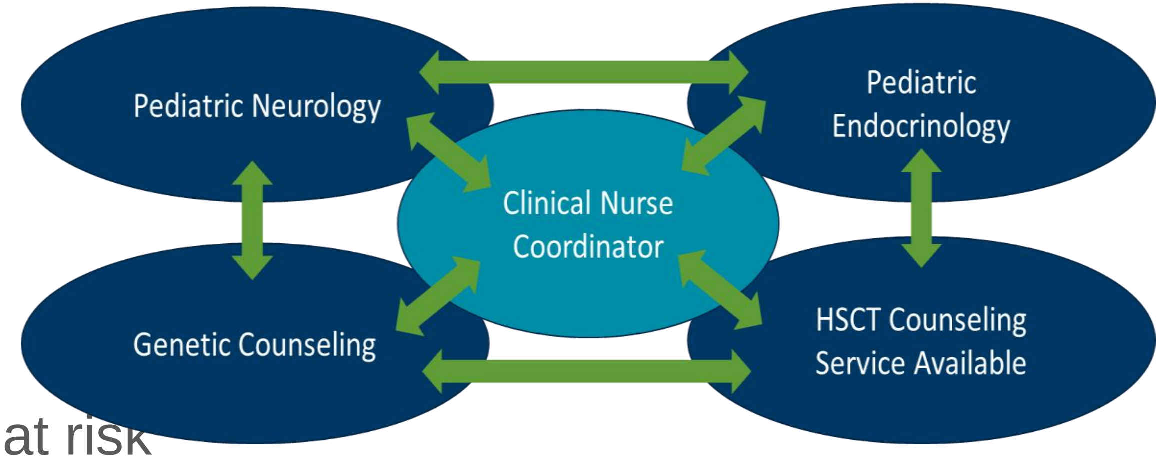
GENETIC COUNSELING AS PART OF INITIAL VISIT: X-ALD EXAMPLE

- **Genetic Counselors are part of the X-ALD team.**
- **They help to:**
  - Identify the cause of disease
  - Interpret genetic testing results
  - Counsel families
  - Determine of other family members are at risk
  - Coordinate family testing
  - Provide support and resources

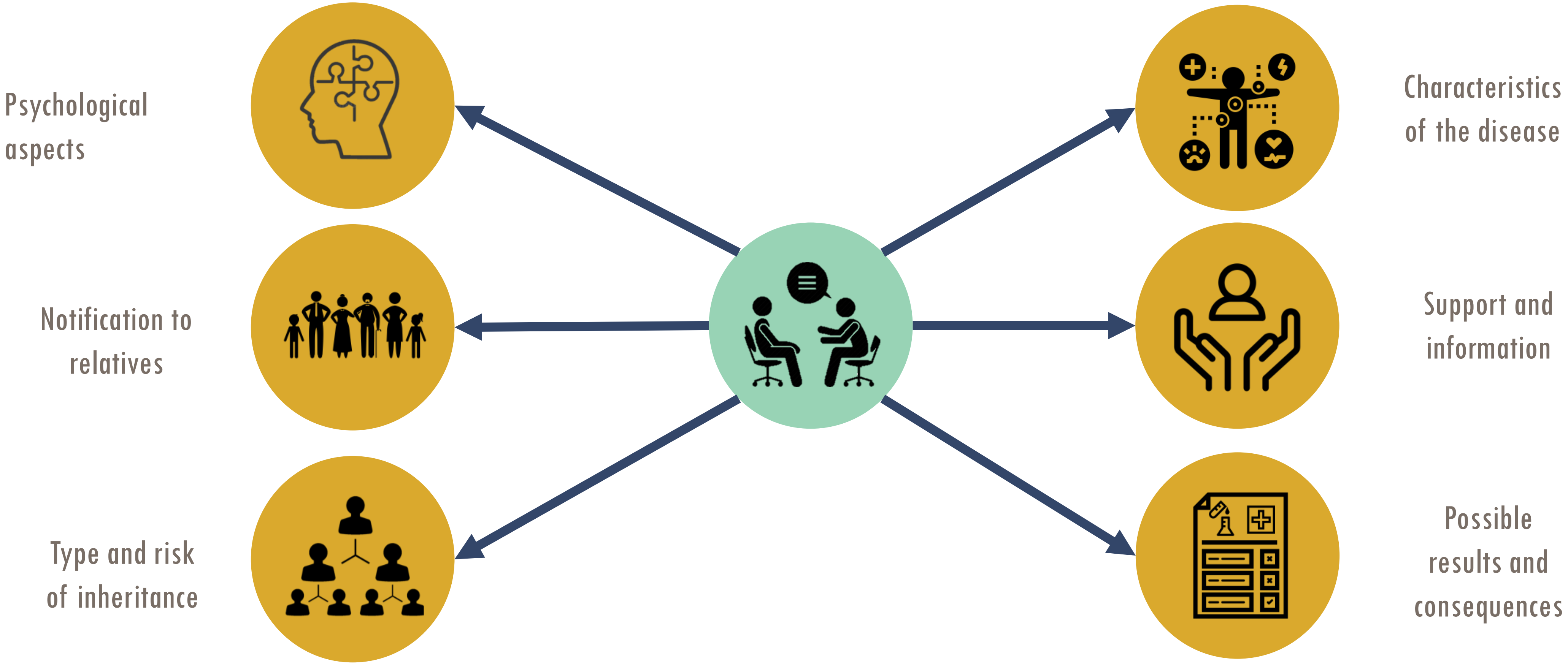


GENETIC COUNSELING FOR CARRIER/TRAIT FINDINGS

- **Newborn Screening Regularly Detects Carriers and Trait Status for:**
    - Cystic Fibrosis
    - Sickle Cell/Hemoglobinopathies
- **Genetic Counseling is Recommended for these findings**
    - CF Foundation policy; exploring access issues via telehealth
    - Certificate programs available for Sickle Cell Trait counseling (not restricted to genetic counselors)

ANATOMY OF A GENETIC COUNSELING SESSION

Family & Patient Goal Setting with Shared Decision Making



Psychological aspects

Notification to relatives

Type and risk of inheritance

Characteristics of the disease

Support and information

Possible results and consequences

Traumatic events, like getting a genetic diagnosis and caring for someone with a genetic condition, can have lasting effects on health and well-being.

Before Diagnosis

- Diagnostic Delay
- Frequent Medical Consults, Tests, Treatments
- Feelings of Fear, Frustration, and Anxiety
- Avoidance and/or hypervigilance

At Time of Diagnosis

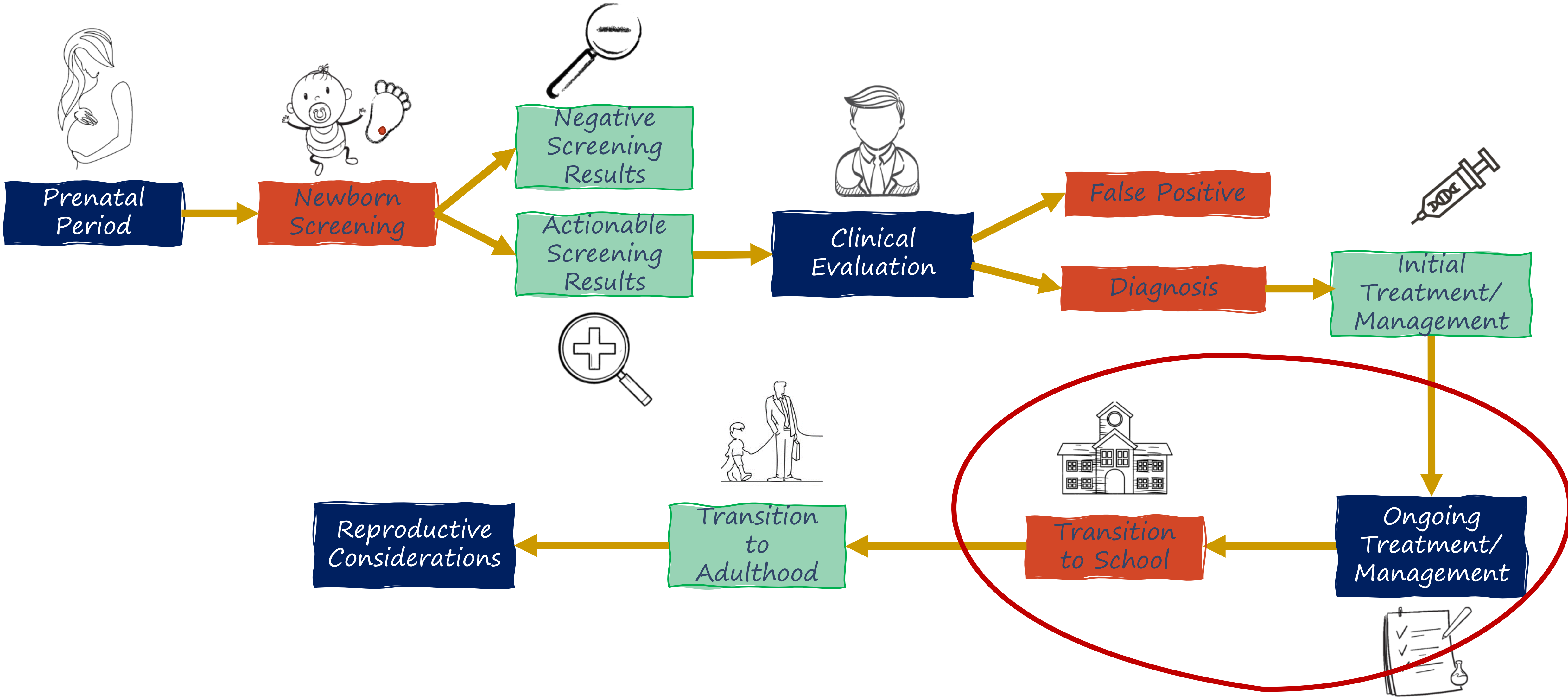
- Emotional Impact of Diagnosis
- Feelings of Shock and Grief
- Anger and Resentment at Diagnostic Odyssey

After Diagnosis

- Concerns over Lack of Awareness
- Burden of Being the Expert
- Feelings of objectification
- Potential for re-traumatization

Chivroli, J., *et al.* 10.13140/RG.2.2.27558.47684. (2019).

THE NBS AND RARE DISEASE JOURNEY



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DISCUSSIONS OF TREATMENT OPTIONS/MANAGEMENT PLAN

HSCT

Medical Foods/ Formula

Vitamins/ Supplements

Gene Therapy

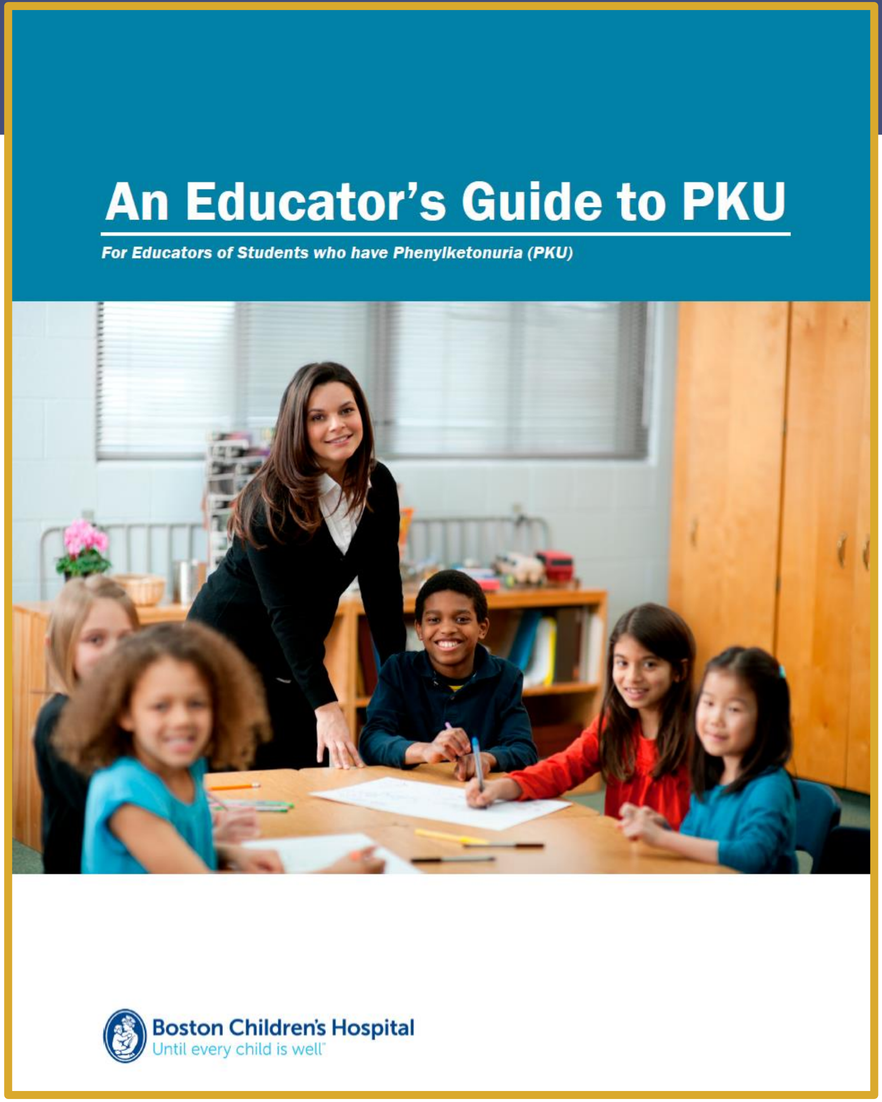
Enzyme Replacement Therapy/ Infusions

Many Others

- Families identified through Newborn Screening may be faced with several treatment options; including whether or not to participate in a clinical trial
- Treatment and management may also require significant time and effort
  - May require travel away from home for months

TRANSITIONING TO SCHOOL AGE

- **Transition guidance may include:**
  - How to talk to classmates and teachers
  - Consideration of nutrition needs
  - Consideration of other ancillary needs (more breaks, hydration, sick days, etc)
  - Individualized Education Plans

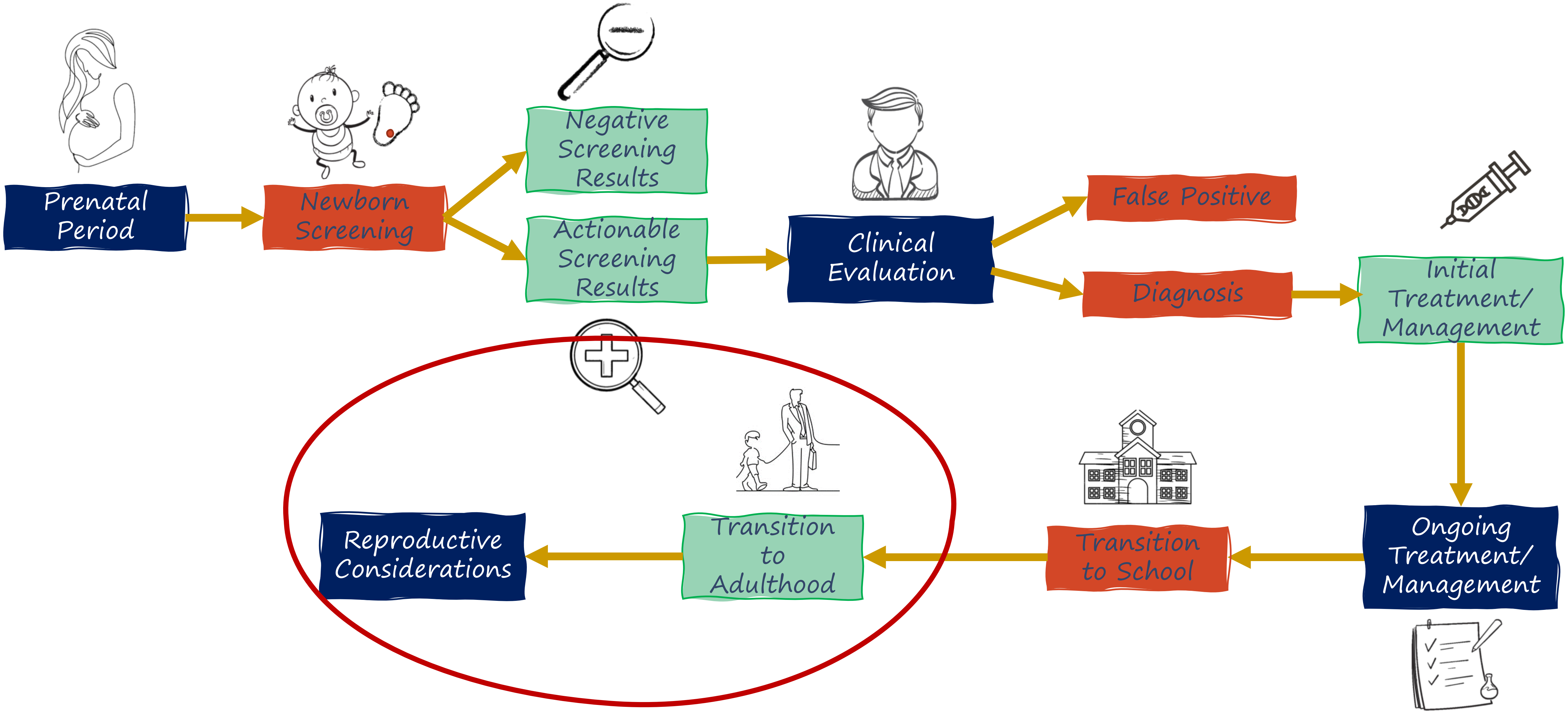


An Educator's Guide to PKU

For Educators of Students who have Phenylketonuria (PKU)

Boston Children's Hospital
Until every child is well

THE NBS AND RARE DISEASE JOURNEY



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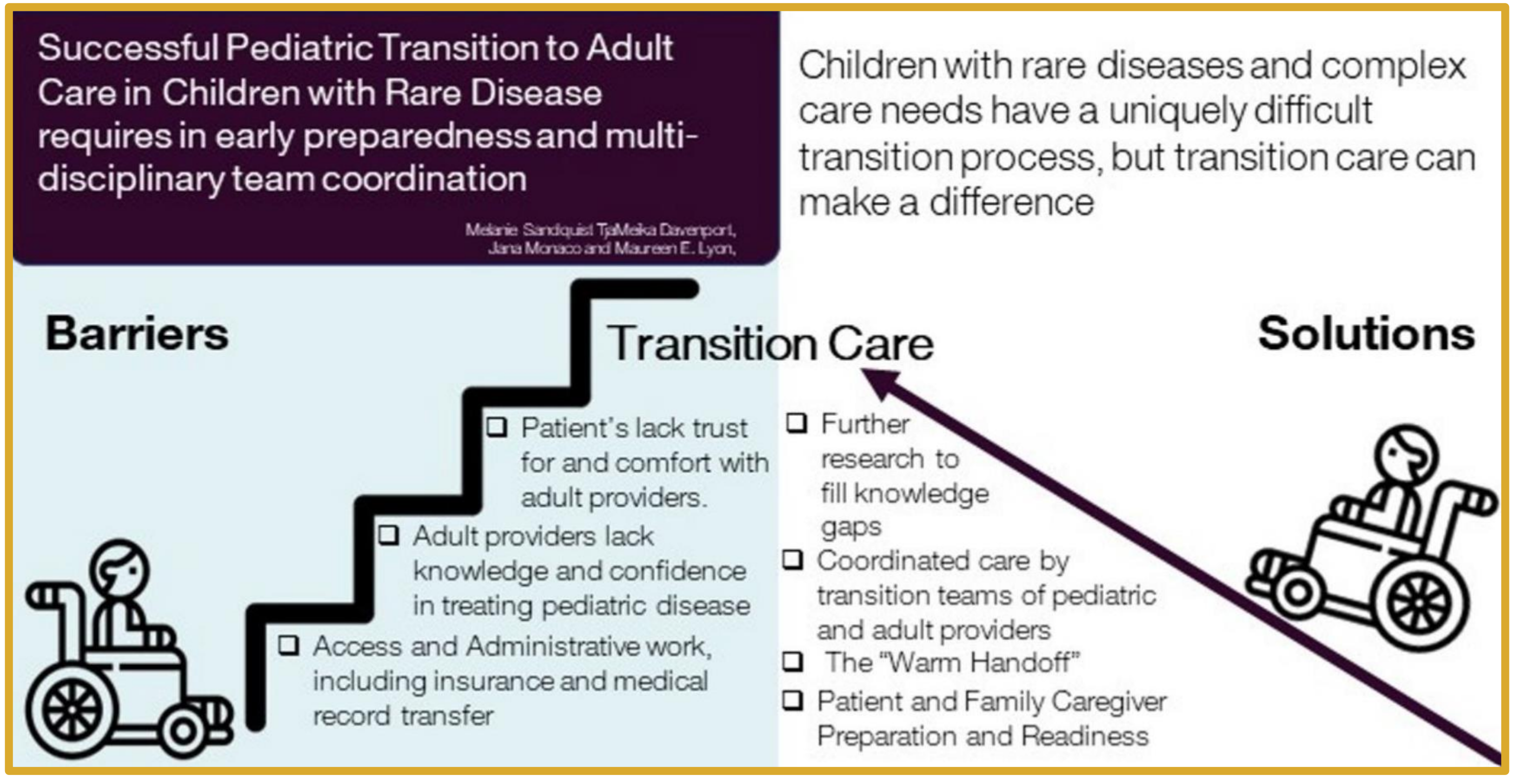
TRANSITIONING TO ADULTHOOD



Successful Pediatric Transition to Adult Care in Children with Rare Disease requires in early preparedness and multi-disciplinary team coordination

Melanie Sandquist TjaMeika Davenport,
Jana Monaco and Maureen E. Lyon,

Children with rare diseases and complex care needs have a uniquely difficult transition process, but transition care can make a difference

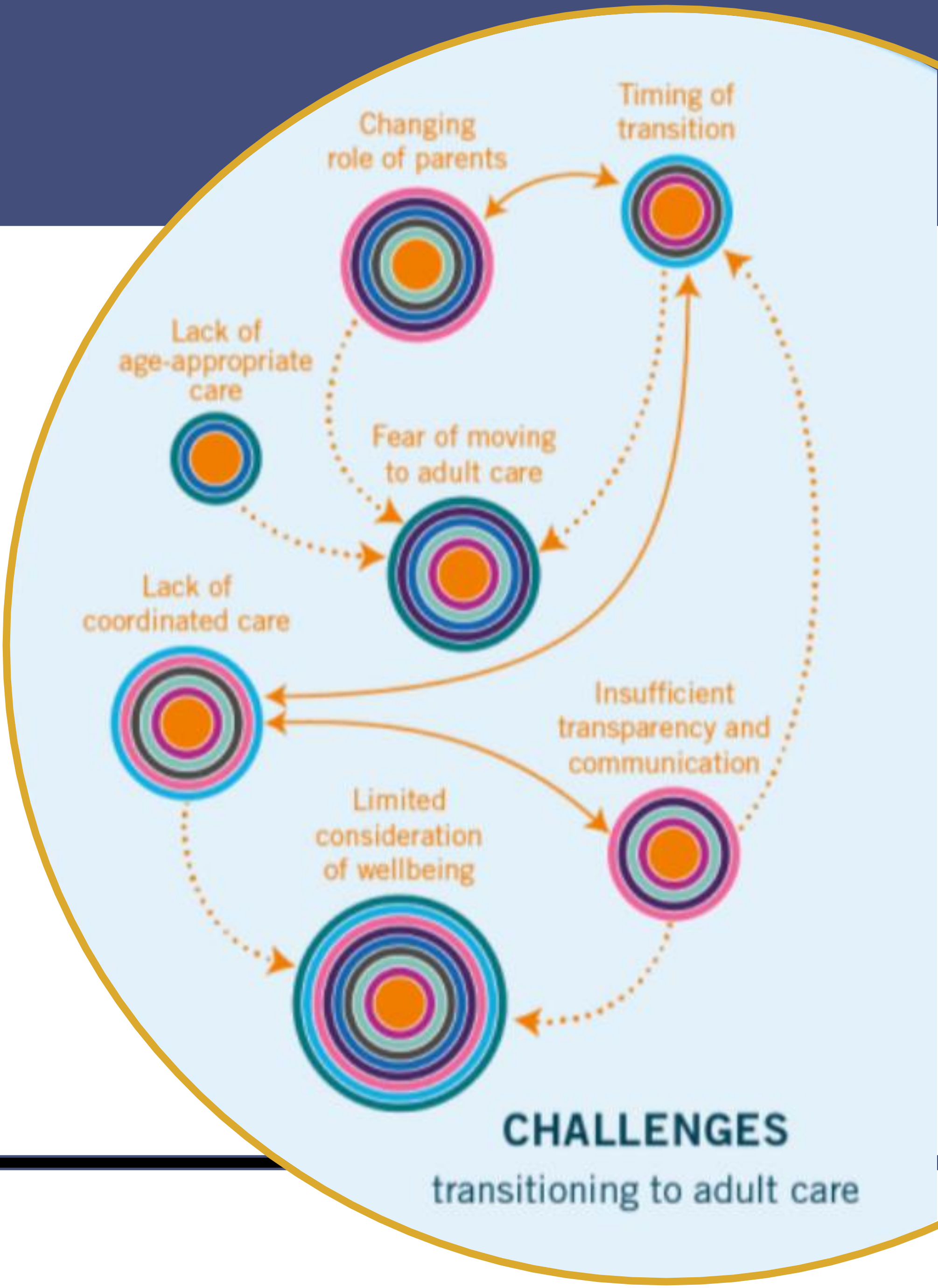
Barriers

Transition Care

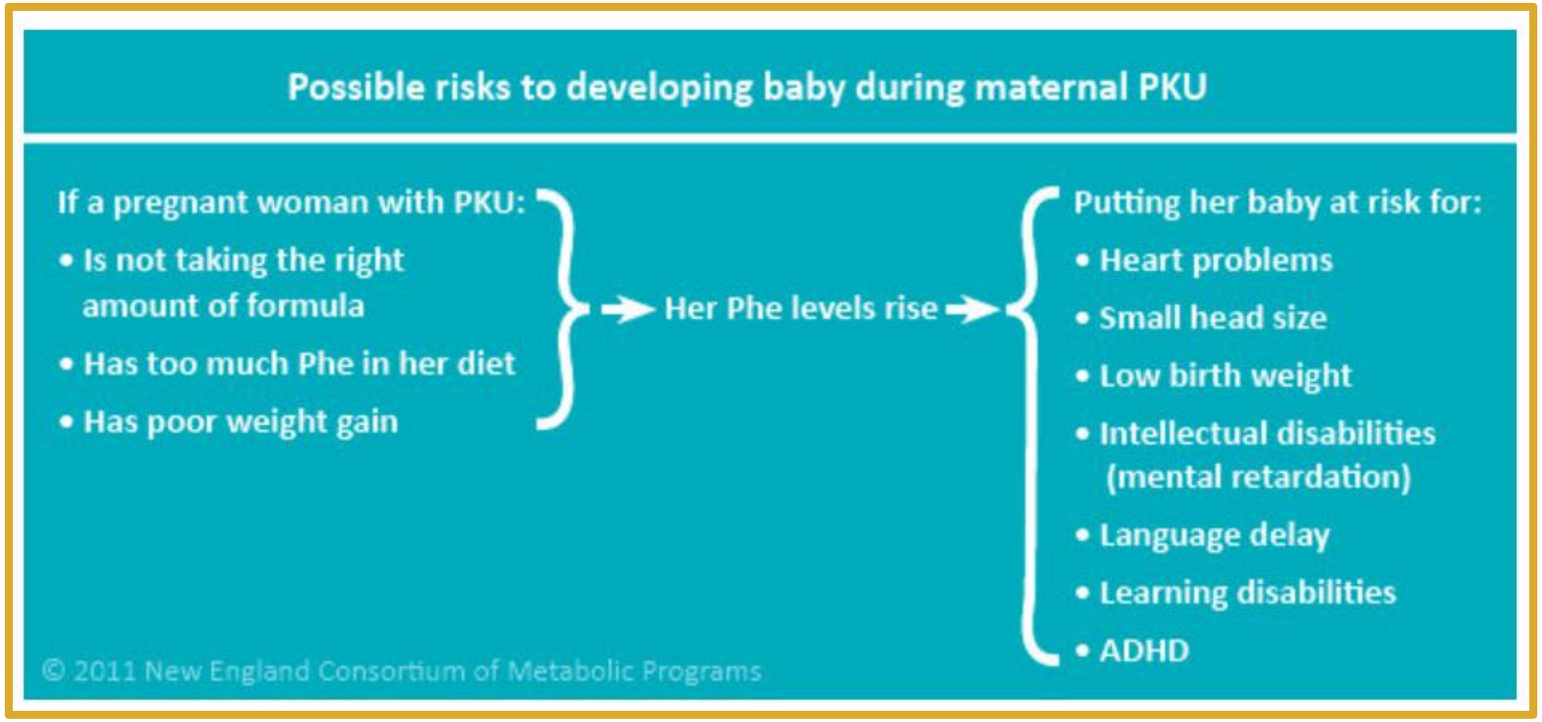
Solutions

- ☐ Patient's lack trust for and comfort with adult providers.
- ☐ Adult providers lack knowledge and confidence in treating pediatric disease
- ☐ Access and Administrative work, including insurance and medical record transfer

- ☐ Further research to fill knowledge gaps
- ☐ Coordinated care by transition teams of pediatric and adult providers
- ☐ The "Warm Handoff"
- ☐ Patient and Family Caregiver Preparation and Readiness



CARE OF PREGNANT INDIVIDUALS WITH NBS DISEASES



Possible risks to developing baby during maternal PKU

If a pregnant woman with PKU:
- Is not taking the right amount of formula
- Has too much Phe in her diet
- Has poor weight gain

→ Her Phe levels rise →

Putting her baby at risk for:
- Heart problems
- Small head size
- Low birth weight
- Intellectual disabilities (mental retardation)
- Language delay
- Learning disabilities
- ADHD

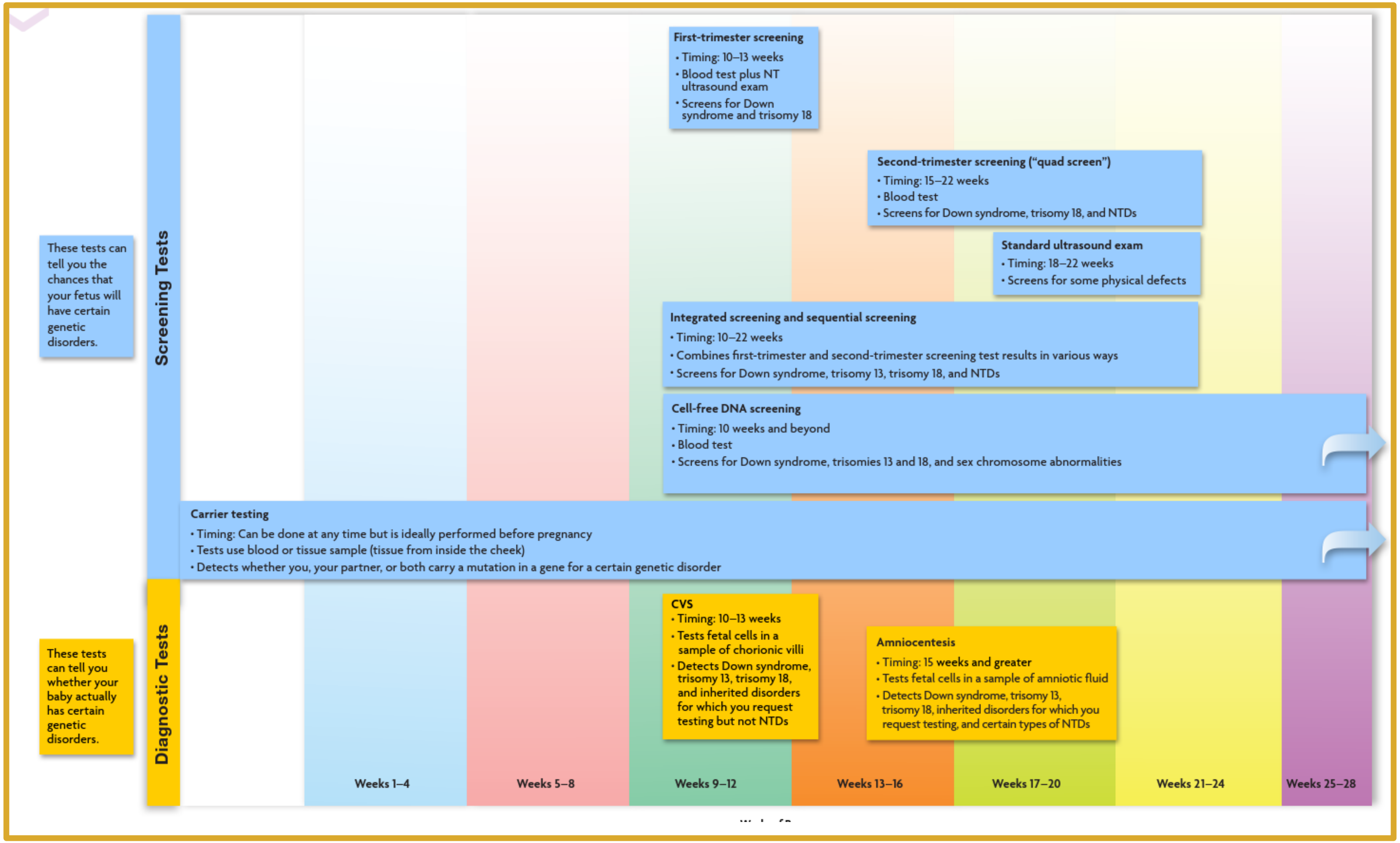
© 2011 New England Consortium of Metabolic Programs

- **Numerous NBS diseases have pregnancy implications:**
  - Urea cycle diseases
  - Maternal PKU
  - Homocystinuria
  - MSUD
  - Galactosemia
  - Fatty acid oxidation disorders
  - Organic acidemias
  - Lysosomal storage diseases

https://www.newenglandconsortium.org/about-maternal-pku

Wilcox (2018). Impact of pregnancy on inborn errors of metabolism. *Reviews in Endocrine and Metaoblic Disorders.* **19**, 13–33

REPRODUCTIVE DECISION MAKING



Screening Tests

These tests can tell you the chances that your fetus will have certain genetic disorders.

First-trimester screening
- Timing: 10–13 weeks
- Blood test plus NT ultrasound exam
- Screens for Down syndrome and trisomy 18

Second-trimester screening ("quad screen")
- Timing: 15–22 weeks
- Blood test
- Screens for Down syndrome, trisomy 18, and NTDs

Standard ultrasound exam
- Timing: 18–22 weeks
- Screens for some physical defects

Integrated screening and sequential screening
- Timing: 10–22 weeks
- Combines first-trimester and second-trimester screening test results in various ways
- Screens for Down syndrome, trisomy 13, trisomy 18, and NTDs

Cell-free DNA screening
- Timing: 10 weeks and beyond
- Blood test
- Screens for Down syndrome, trisomies 13 and 18, and sex chromosome abnormalities

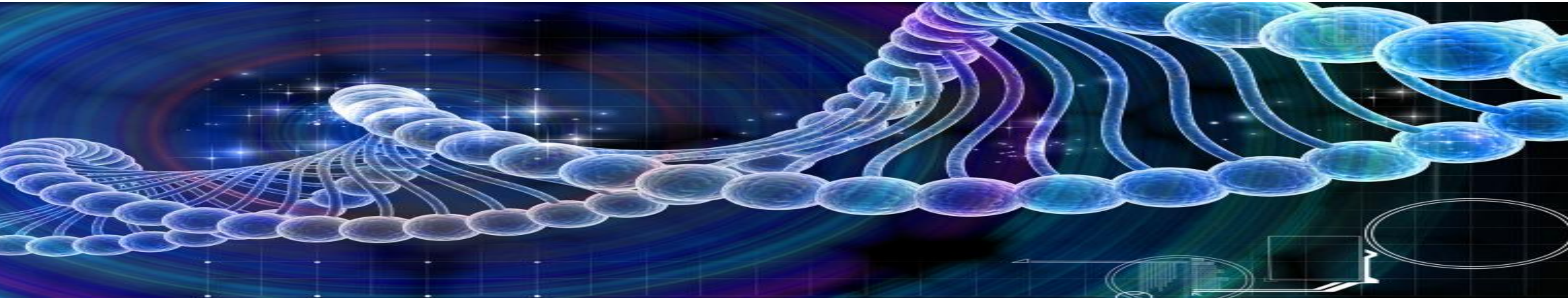
Carrier testing
- Timing: Can be done at any time but is ideally performed before pregnancy
- Tests use blood or tissue sample (tissue from inside the cheek)
- Detects whether you, your partner, or both carry a mutation in a gene for a certain genetic disorder

Diagnostic Tests

These tests can tell you whether your baby actually has certain genetic disorders.

CVS
- Timing: 10–13 weeks
- Tests fetal cells in a sample of chorionic villi
- Detects Down syndrome, trisomy 13, trisomy 18, and inherited disorders for which you request testing but not NTDs

Amniocentesis
- Timing: 15 weeks and greater
- Tests fetal cells in a sample of amniotic fluid
- Detects Down syndrome, trisomy 13, trisomy 18, inherited disorders for which you request testing, and certain types of NTDs



FUTURE CONSIDERATIONS

A NOTE ON TERMINOLOGY

Newborn Screening

- Population-based Public Health program
- State-mandated/Opt-out
- Relies on doctrines like *parens patriae*
- Diseases must meet certain criteria (eg, effective treatment is available)

Sequencing of Newborns/Children

- Typically done in clinic/hospital
- Consented
- Relies on medical guidelines for reportable findings
- May detect both treatable and untreatable diseases

OVERVIEW OF WGS IN HEALTHY NEWBORNS: CURRENT PROJECTS*

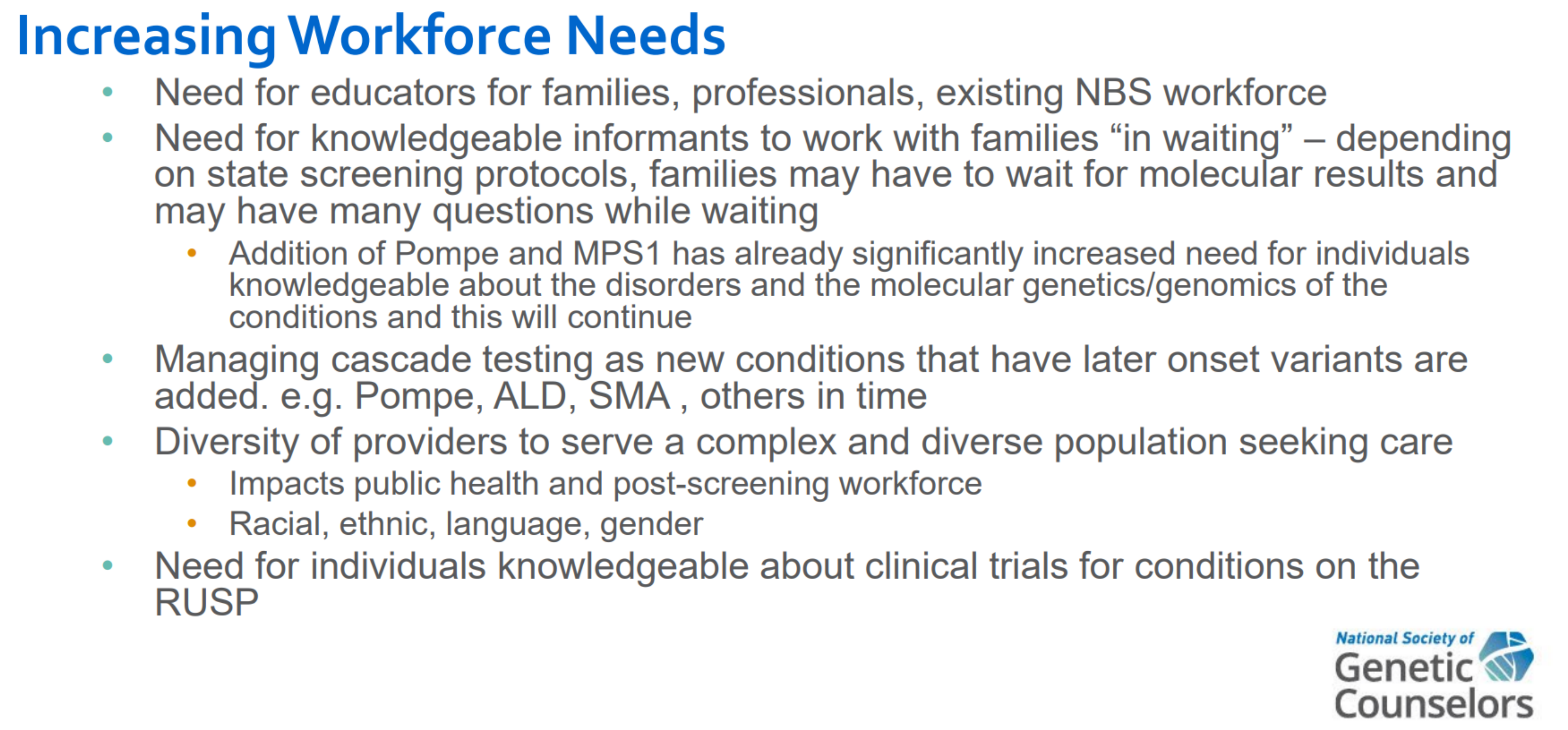
* Table likely not exhaustive

PROJECT	LOCATION
BabySeq2	US
Baby Beyond	Australia
BeginNGS	US/Greece
EarlyCheck	US
Genomics England	UK
GUARDIAN	US
Screen4Care	Europe

Commonalities:

- Using Whole Genome backbone to essentially conduct a large gene panel
  - Filtering to specific genes (childhood-onset/treatable dz)
  - Filtering to pathogenic/likely pathogenic variants
- Questions around:
  - Treatability vs. actionability vs. manageability

WORKFORCE CAPACITY CONSIDERATIONS



Increasing Workforce Needs

- Need for educators for families, professionals, existing NBS workforce
- Need for knowledgeable informants to work with families “in waiting” – depending on state screening protocols, families may have to wait for molecular results and may have many questions while waiting
  - Addition of Pompe and MPS1 has already significantly increased need for individuals knowledgeable about the disorders and the molecular genetics/genomics of the conditions and this will continue
- Managing cascade testing as new conditions that have later onset variants are added. e.g. Pompe, ALD, SMA , others in time
- Diversity of providers to serve a complex and diverse population seeking care
  - Impacts public health and post-screening workforce
  - Racial, ethnic, language, gender
- Need for individuals knowledgeable about clinical trials for conditions on the RUSP

FUTURE NEEDS

"After looking at all your test results and consulting many experts, it's my medical opinion that you have something I can't pronounce."

- Ever increasing need to educate providers
- Increasing prenatal NBS education
- Working with families "in waiting"
- Coordination and planning with primary care
- Explanation of molecular results coming from NBS programs
- Support for families
- Ongoing QI and data coordination

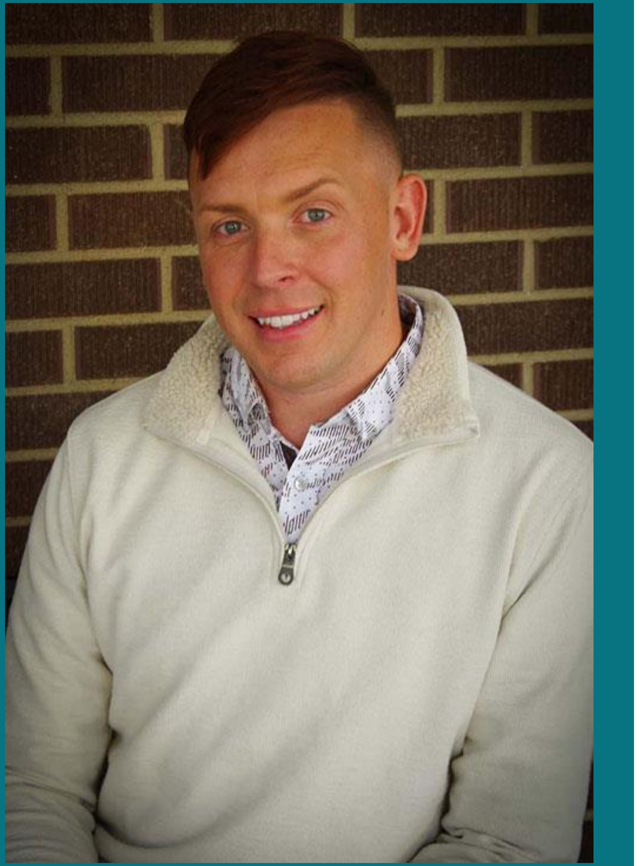
THANK YOU!

Amy Gaviglio, MS, CGC

amy.gaviglio@outlook.com

TAMI DECOTEAU
PHD

JAREB KINNEY
MA, CMHC, LPCC



DISCLOSURES

- Nothing to disclose

The Impact of Trauma of Child Development

Newborn Screening Conference

Presenters

Jareb Kinney, MA. Licensed Professional Clinical Counselor

Dr. Tami DeCoteau, Ph.D. Licensed Psychologist

Presentation Overview

- Defining Trauma
- Adverse Childhood Experiences
- What are Adverse Childhood Experiences
- Intergenerational Trauma effects on children
- Summarizing the Impact of Different Traumas on Child Development and Trauma Informed Care

What is Trauma and Toxic Stress

- Trauma is the result of exposure to distressing or life-threatening events that have lasting negative effects on an individual's functioning and well-being.
- Traumatic experiences include but are not limited to physical, sexual, and emotional abuse, childhood neglect, living with a family member with mental health or substance use disorders, sudden separation from a loved one, poverty, racism, discrimination, oppression, violence in the community, war, or terrorism.
- Trauma has particularly damaging long-term effects on children's developing brains and is commonly referred to as adverse childhood experiences (ACEs).

What is Trauma and Toxic Stress Cont.

Toxic stress is prolonged or repeated activation of the stress response system in children without the buffering presence of a supportive, nurturing adult. This type of stress can occur when a child experiences strong, frequent, or prolonged adversity, such as abuse, neglect, exposure to violence or substance abuse, or living in poverty. Toxic stress can affect a child's brain development, lead to long-term health problems, and negatively impact their social, emotional, and cognitive functioning.

Adverse Childhood Experiences

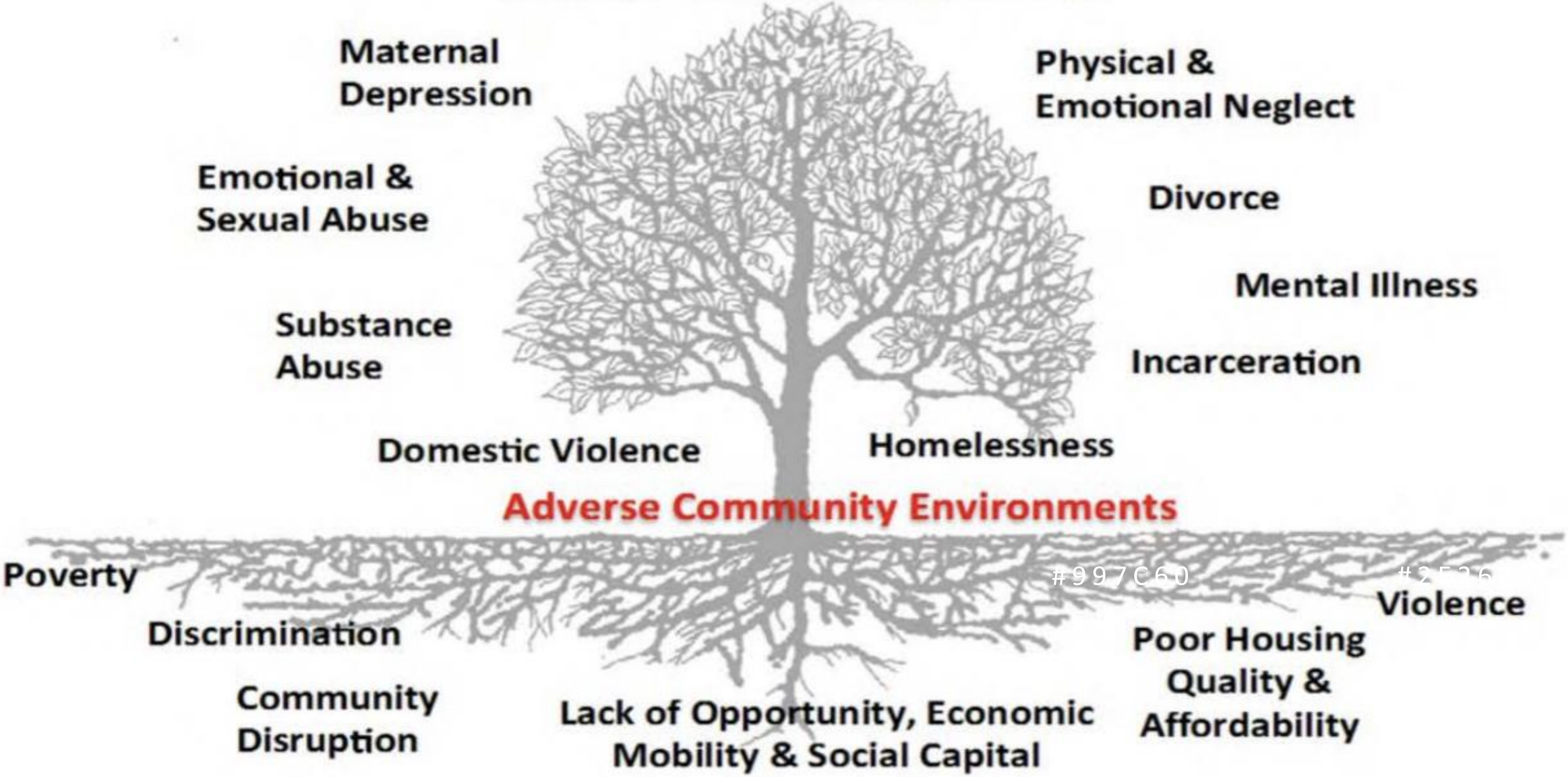
- ACES is the largest and first research study that established a direct association between childhood trauma and adult ill health. The study found that the greater the number of advers childhood experiences, the greater the possibility of disabilities and dysfunction in adult life.

- ACEs Questionnaire (Felitti et al., 1998) is a 10-item measure used to measure childhood trauma. The questionnaire assesses 10 types of childhood trauma measured in the ACE Study. Five are personal: physical abuse, verbal abuse, sexual abuse, physical neglect, and emotional neglect.

What are Adverse Childhood Experiences

- Adverse childhood experiences (ACES) abuse, neglect, or household challenges children may face.
- ACES are linked to health problems in adulthood, including heart disease, cancer, and poor health.
- The impact of ACES is cumulative and can lead to multiple risk factors for leading causes of death in adults.
- ACES can lead to coping behaviors such as smoking, alcohol, or drug abuse, which can contribute to long-term health problems.
- Children exposed to high levels of ACES may develop anxiety, anger, and depression, leading to chronic use of coping behaviors.
- ACES are associated with long-term health problems due to the positive effects of health-risk behaviors, such as smoking.

The Pair of ACEs



Ellis, W., Dietz, W. (2017) A New Framework for Addressing Adverse Childhood and Community Experiences: The Building Community Resilience (BCR) Model. Academic Pediatrics. 17 (2017) pp. S86-S93. DOI information: 10.1016/j.acap.2016.12.011

2 times

as likely to smoke

2.5 times

more likely to have sexually-transmitted infections

4 times

more likely to have chronic obstructive pulmonary disease

7 times

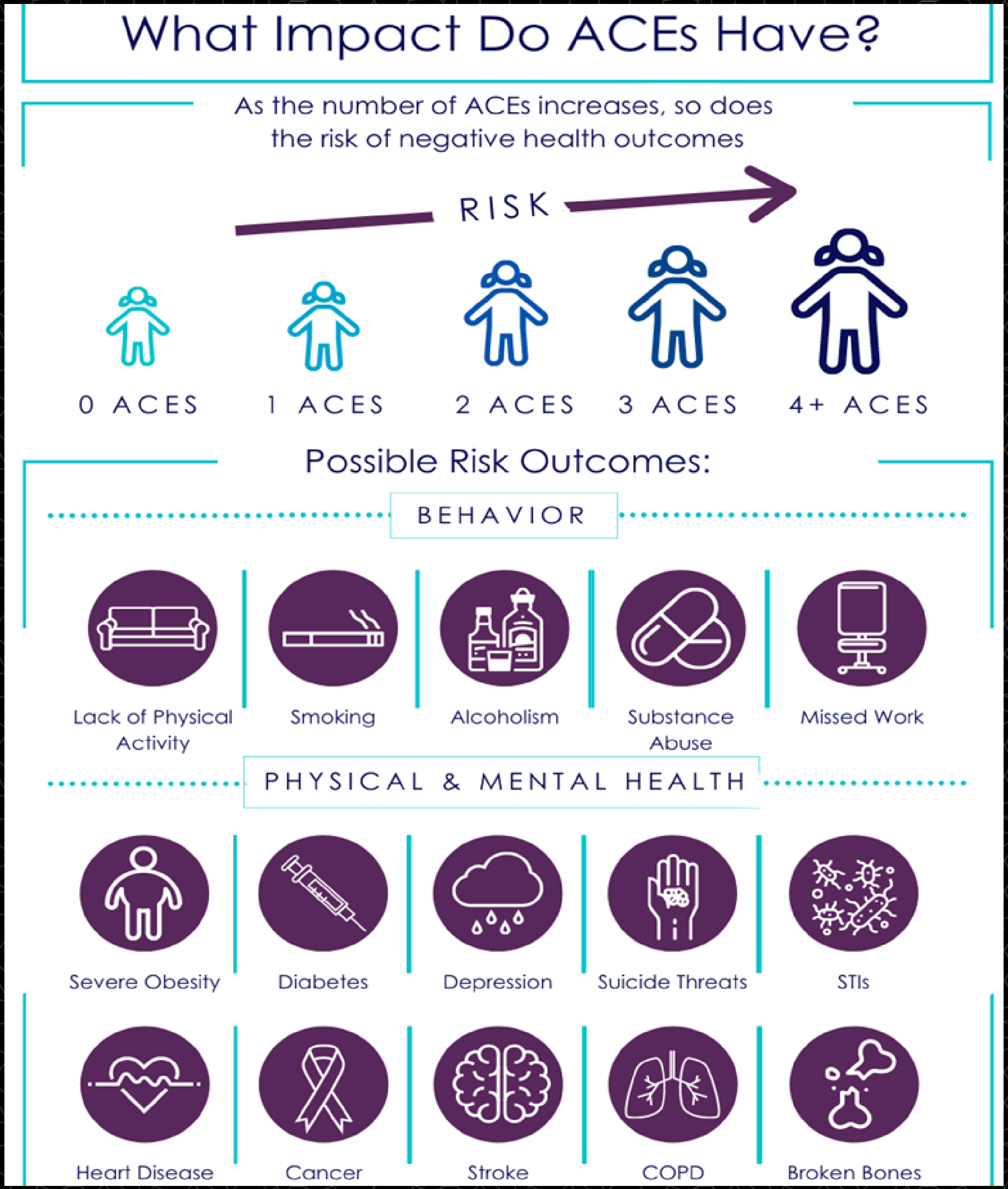
more likely to consider themselves an alcoholic

10 times

as likely to have injected street drugs

12 times

as likely to have attempted suicide



What Impact Do ACEs Have?

As the number of ACEs increases, so does the risk of negative health outcomes

Possible Risk Outcomes:

BEHAVIOR

- Lack of Physical Activity
- Smoking
- Alcoholism
- Substance Abuse
- Missed Work

PHYSICAL & MENTAL HEALTH

- Severe Obesity
- Diabetes
- Depression
- Suicide Threats
- STIs
- Heart Disease
- Cancer
- Stroke
- COPD
- Broken Bones

Adverse Childhood Experiences and Birth Complications

- One study by Mersky and Lee (2019) showed that 84.4% of low-income women receiving home visiting services had at least one ACE, and 68.2% reported multiple ACEs.
- Higher ACE scores were associated with a greater likelihood of pregnancy loss, preterm birth, and low birthweight, with each additional ACE associated with a 12% increase in pregnancy loss, a 7% increase in preterm birth, and an 8% increase in low birthweight.
- Greater exposure to ACEs increases the risk of pregnancy loss, preterm birth, and low birthweight.
- The associations between ACEs and preterm birth and low birthweight appear to have threshold effects, meaning some poor birth outcomes may only occur with a critical level of adversity.
- Further research is needed to investigate non-additive and interactive effects of ACEs, as well as the timing, frequency, severity, and duration of adverse experiences.
- Biological changes such as compromised neuroendocrine and immune functions, as well as psychosocial pathways such as elevated levels of stress and anxiety, tobacco and substance use, and exposure to adverse adult experiences may be factors (Mersky & Lee, 2019).

The Intergenerational Transmission of Adversity

- Intergenerational trauma is a cycle in which trauma from past generations is passed down to current generations, affecting the physical health and brain development of children who are surrounded by it.
- Adverse Childhood Experiences (ACEs) can lead to intergenerational trauma, and when traumas and adverse experiences remain unaddressed, individuals may repeat patterns learned in their own childhoods.
- Intergenerational trauma is not just a pattern of behavior that is learned and repeated but a brain development issue.
- nationally-recognized approaches focus on helping young children and their caregivers. It works to intervene in the cycle of intergenerational trauma by focusing on the needs of the child while also addressing how past trauma impacts the relationships between parents or caregivers and their children. (Center for Child Counseling)

The Intergenerational Transmission of Adversity Cont.

- Adverse Childhood Experiences (ACEs) have negative, lasting effects on physical and mental health of exposed children during childhood and later in life.
- ACEs affect future generations through intergenerational transmission of risk from mother to child, but the exact mechanism is unclear.
- A recent study examined the association between maternal ACEs, neonatal brain development, and infant emotionality at six months of age.
- Maternal ACEs were associated with smaller amygdala volumes in newborns, and both high maternal ACEs and smaller left amygdala volume were associated with higher infant negative emotionality at six months.
- The observed changes in brain structure are likely attributable to changes in the prenatal environment rather than exposures after delivery.
- The mechanisms underlying the association between maternal ACEs and infant brain structure remain unclear, but alterations in gestational biology, including endocrine and immune systems, have been implicated.
- ACEs are highly prevalent, and prevention initiatives in children are critical. Understanding the transmission of risk between mother and child may lead to targeted interventions that mitigate risk in children. (Nonacs et al. 2022)

Adverse Childhood Experiences and Birth Complications

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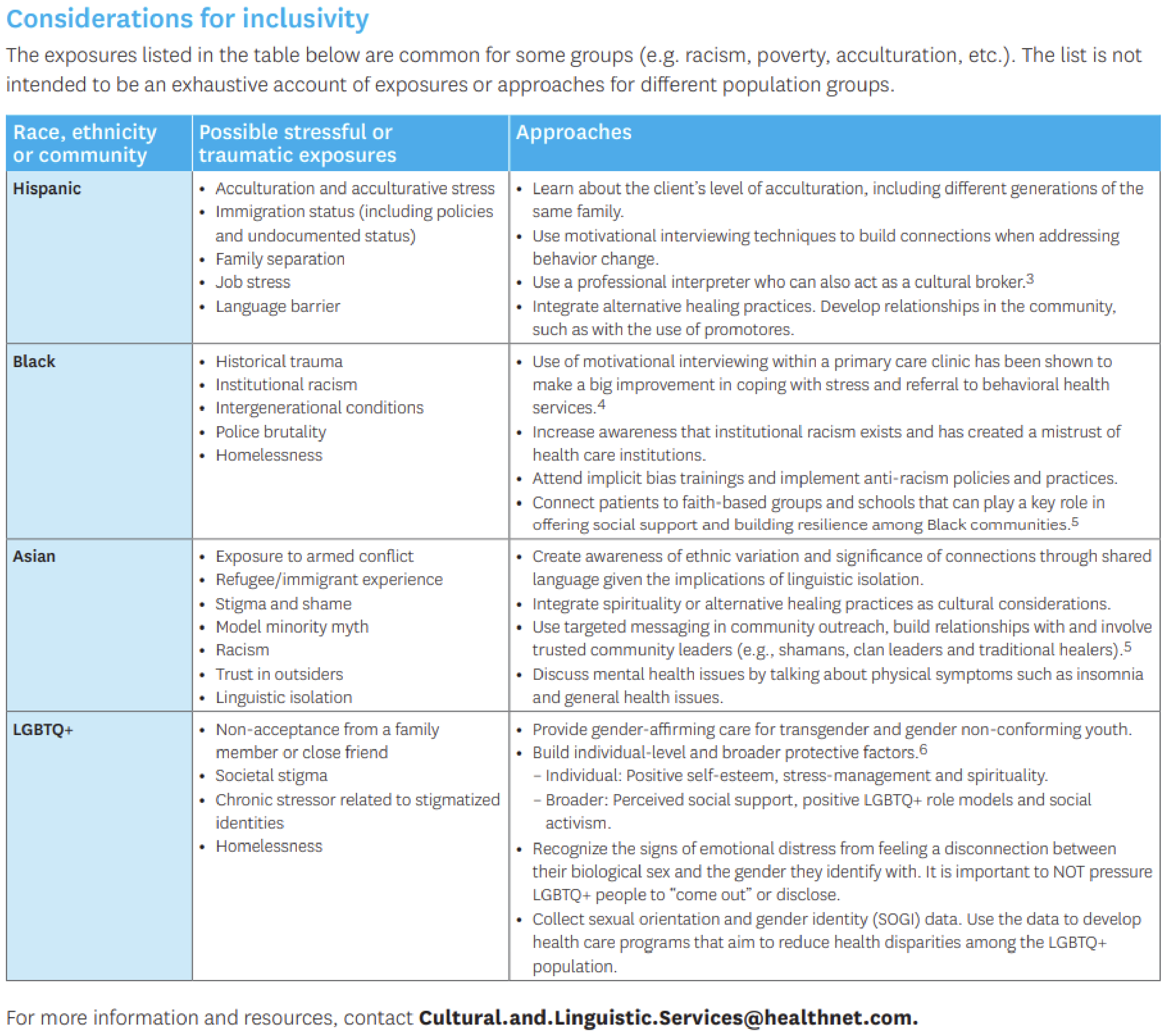
Cultural Considerations



Considerations for inclusivity

The exposures listed in the table below are common for some groups (e.g. racism, poverty, acculturation, etc.). The list is not intended to be an exhaustive account of exposures or approaches for different population groups.

Race, ethnicity or community	Possible stressful or traumatic exposures	Approaches
Hispanic	• Acculturation and acculturative stress • Immigration status (including policies and undocumented status) • Family separation • Job stress • Language barrier	• Learn about the client's level of acculturation, including different generations of the same family. • Use motivational interviewing techniques to build connections when addressing behavior change. • Use a professional interpreter who can also act as a cultural broker.[3] • Integrate alternative healing practices. Develop relationships in the community, such as with the use of promotores.
Black	• Historical trauma • Institutional racism • Intergenerational conditions • Police brutality • Homelessness	• Use of motivational interviewing within a primary care clinic has been shown to make a big improvement in coping with stress and referral to behavioral health services.[4] • Increase awareness that institutional racism exists and has created a mistrust of health care institutions. • Attend implicit bias trainings and implement anti-racism policies and practices. • Connect patients to faith-based groups and schools that can play a key role in offering social support and building resilience among Black communities.[5]
Asian	• Exposure to armed conflict • Refugee/immigrant experience • Stigma and shame • Model minority myth • Racism • Trust in outsiders • Linguistic isolation	• Create awareness of ethnic variation and significance of connections through shared language given the implications of linguistic isolation. • Integrate spirituality or alternative healing practices as cultural considerations. • Use targeted messaging in community outreach, build relationships with and involve trusted community leaders (e.g., shamans, clan leaders and traditional healers).[5] • Discuss mental health issues by talking about physical symptoms such as insomnia and general health issues.
LGBTQ+	• Non-acceptance from a family member or close friend • Societal stigma • Chronic stressor related to stigmatized identities • Homelessness	• Provide gender-affirming care for transgender and gender non-conforming youth. • Build individual-level and broader protective factors.[6] – Individual: Positive self-esteem, stress-management and spirituality. – Broader: Perceived social support, positive LGBTQ+ role models and social activism. • Recognize the signs of emotional distress from feeling a disconnection between their biological sex and the gender they identify with. It is important to NOT pressure LGBTQ+ people to "come out" or disclose. • Collect sexual orientation and gender identity (SOGI) data. Use the data to develop health care programs that aim to reduce health disparities among the LGBTQ+ population.

For more information and resources, contact **Cultural.and.Linguistic.Services@healthnet.com.**

Trauma-Informed Care

Trauma-informed care supports the health needs of patients who have experienced Adverse Childhood Experiences (ACEs) and toxic stress. It involves understanding trauma prevalence and effects, training staff, integrating trauma knowledge into practices, and avoiding re-traumatization by providing non-judgmental support. Key principles include safety, trust, patient-centered care, and cultural sensitivity. ACE screening can induce emotional reactions, including shame, anger, and empowerment. Providers can avoid re-traumatization by maintaining emotional safety, assessing patient strengths, providing supportive responses, empowering patients with stress-regulation education, referring to trauma-specific therapy, and practicing compassionate resilience for their own well-being.

References

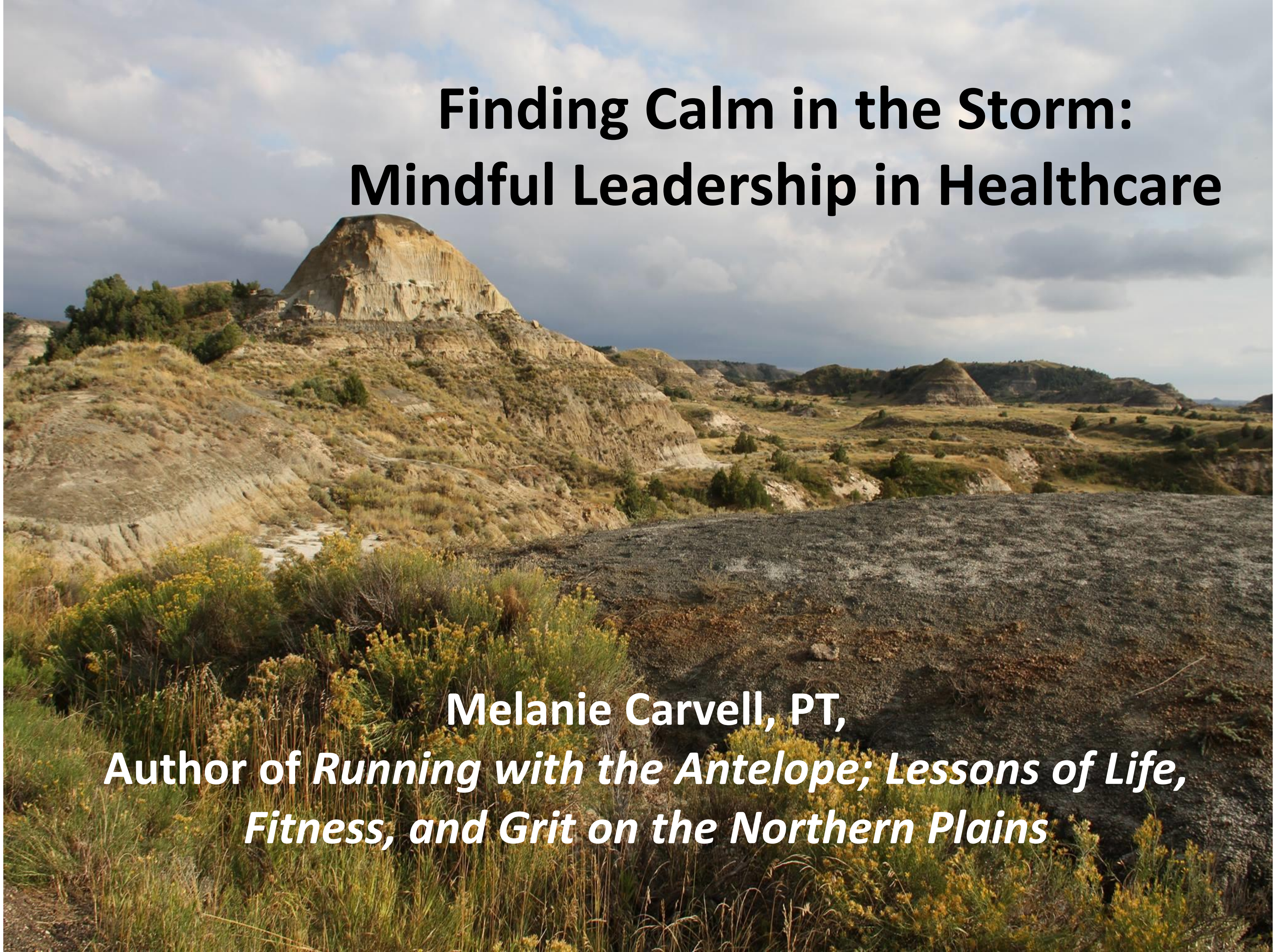
- https://www.traumainformedcare.chcs.org/what-is-trauma/
- https://www.centerforchildcounseling.org/aces-and-intergenerational-trauma/
- Mersky JP, Lee CP. Adverse childhood experiences and poor birth outcomes in a diverse, low-income sample. BMC Pregnancy Childbirth. 2019 Oct 28;19(1):387. doi: 10.1186/s12884-019-2560-8. PMID: 31660899; PMCID: PMC6819344.
- https://womensmentalhealth.org/
- https://www.acesaware.org/


SPIDER
MANDAN
DRAKES
adidas

MELANIE CARVELL PT

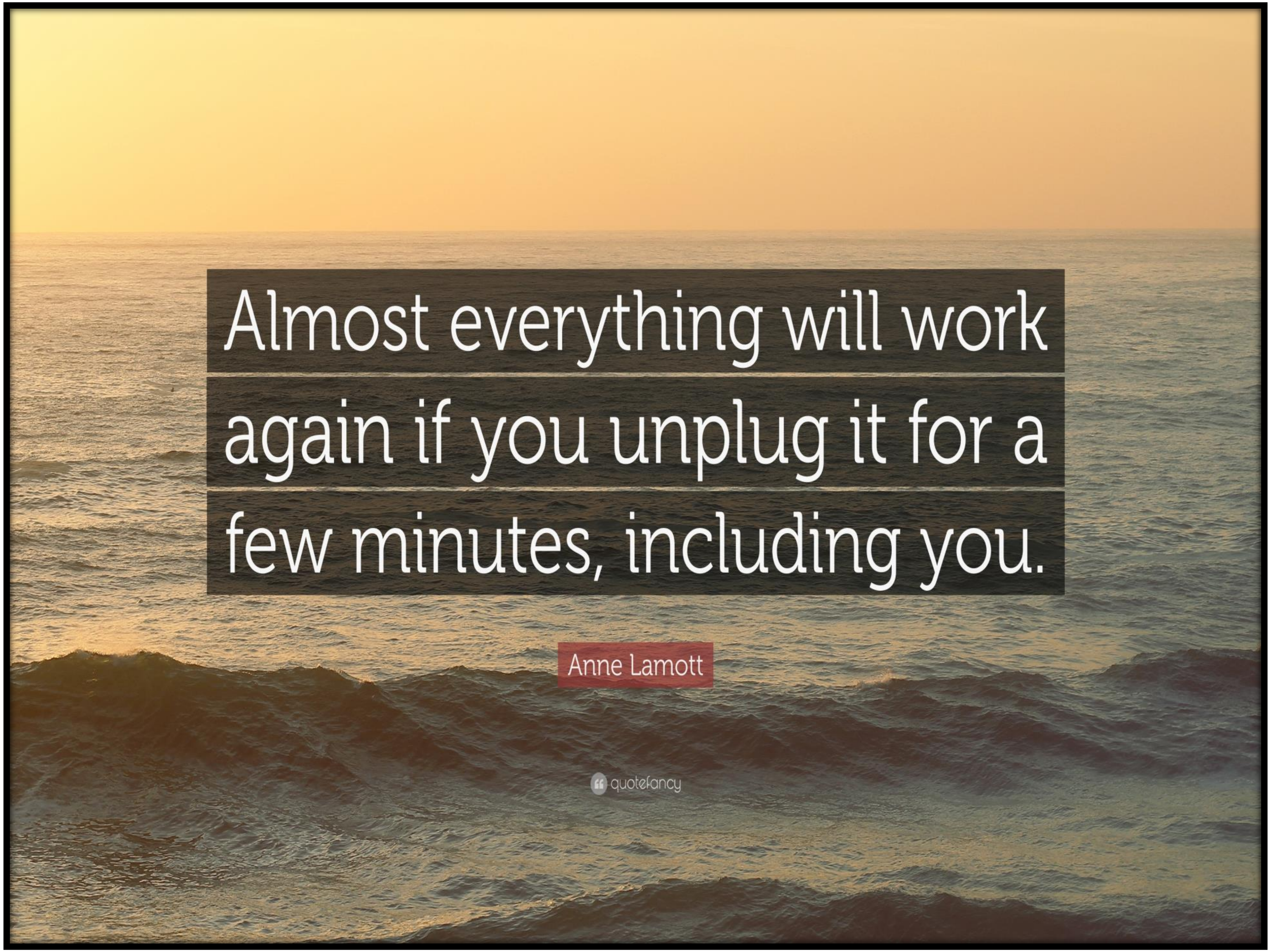
DISCLOSURES

- Nothing to disclose



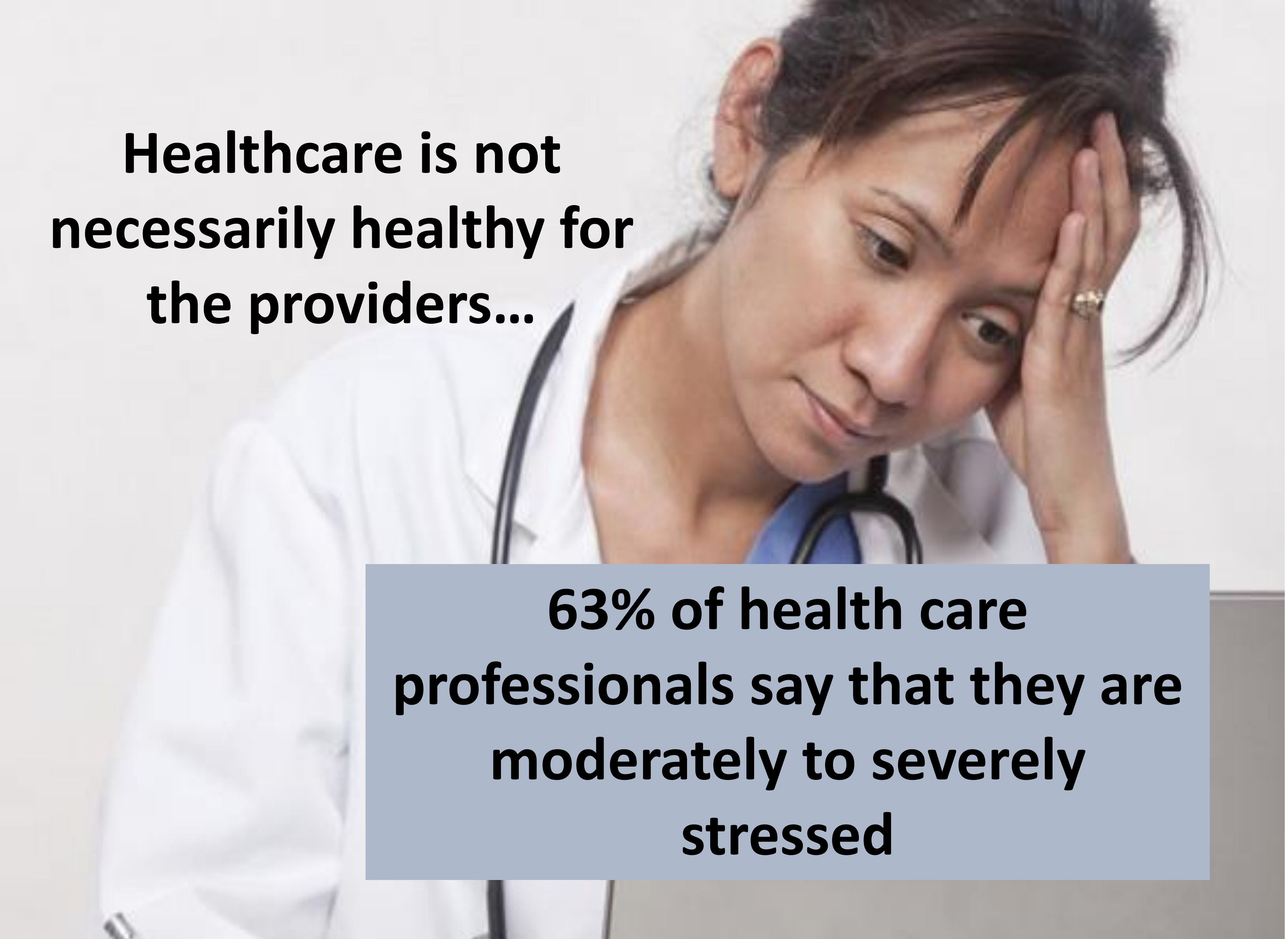
Finding Calm in the Storm:
Mindful Leadership in Healthcare

Melanie Carvell, PT,
Author of *Running with the Antelope; Lessons of Life, Fitness, and Grit on the Northern Plains*



Almost everything will work again if you unplug it for a few minutes, including you.

Anne Lamott



Healthcare is not necessarily healthy for the providers…

63% of health care professionals say that they are moderately to severely stressed

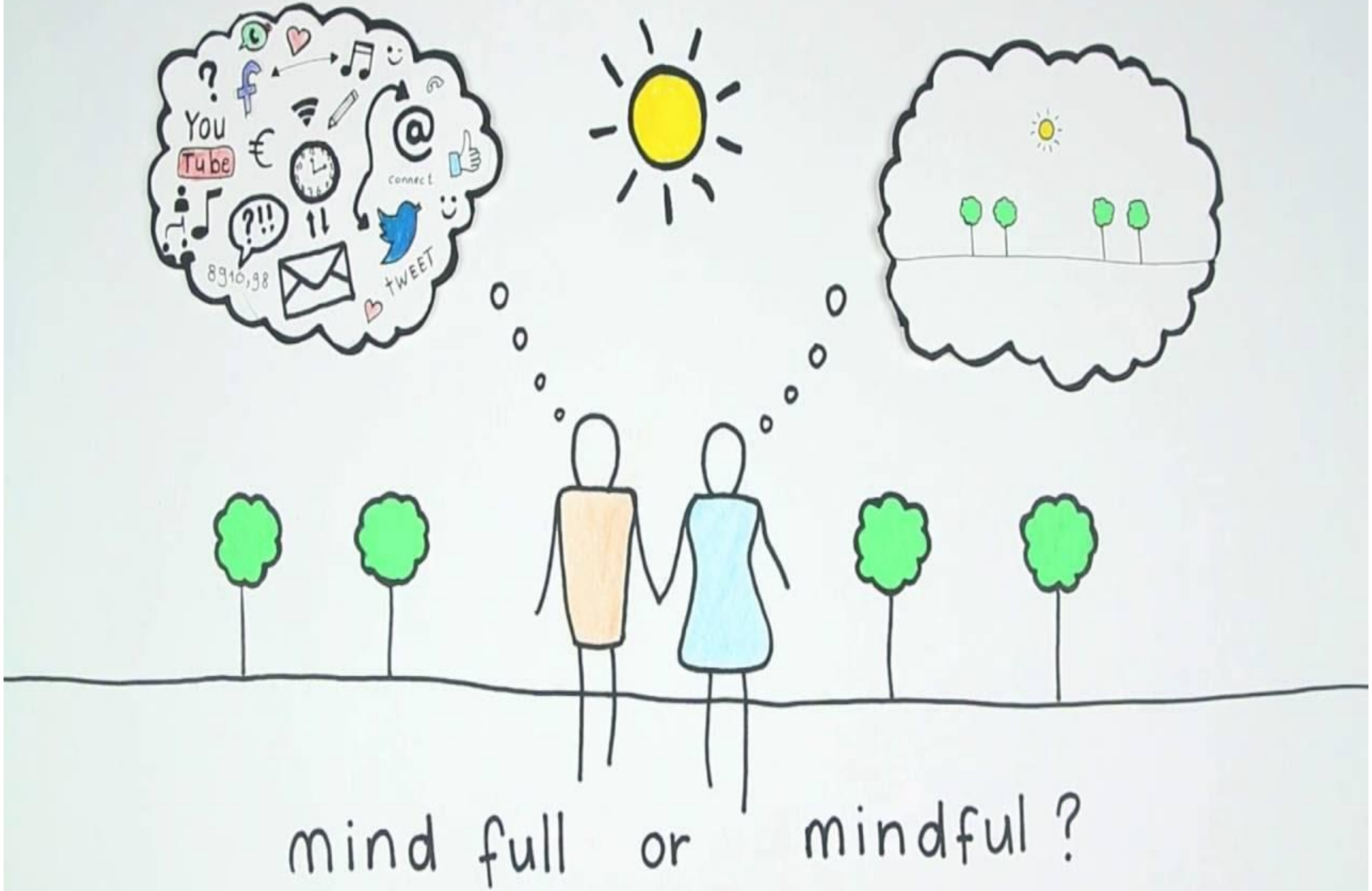
There is a reason why in-flight safety demonstrations always instruct us to secure our own masks first before assisting others

Mindfulness is about being PRESENT…simply being aware of what is happening right now, without the need to judge or control.


now
tomorrow
yesterday

Characteristics of mindful leaders:

- They are less likely to be reactive, jump to conclusions, or to take things too personally
- They can appreciate both sides of a situation
- They understand all things come and go
- They are much less likely to "multi-task"
- They are more likely to eat and drink mindfully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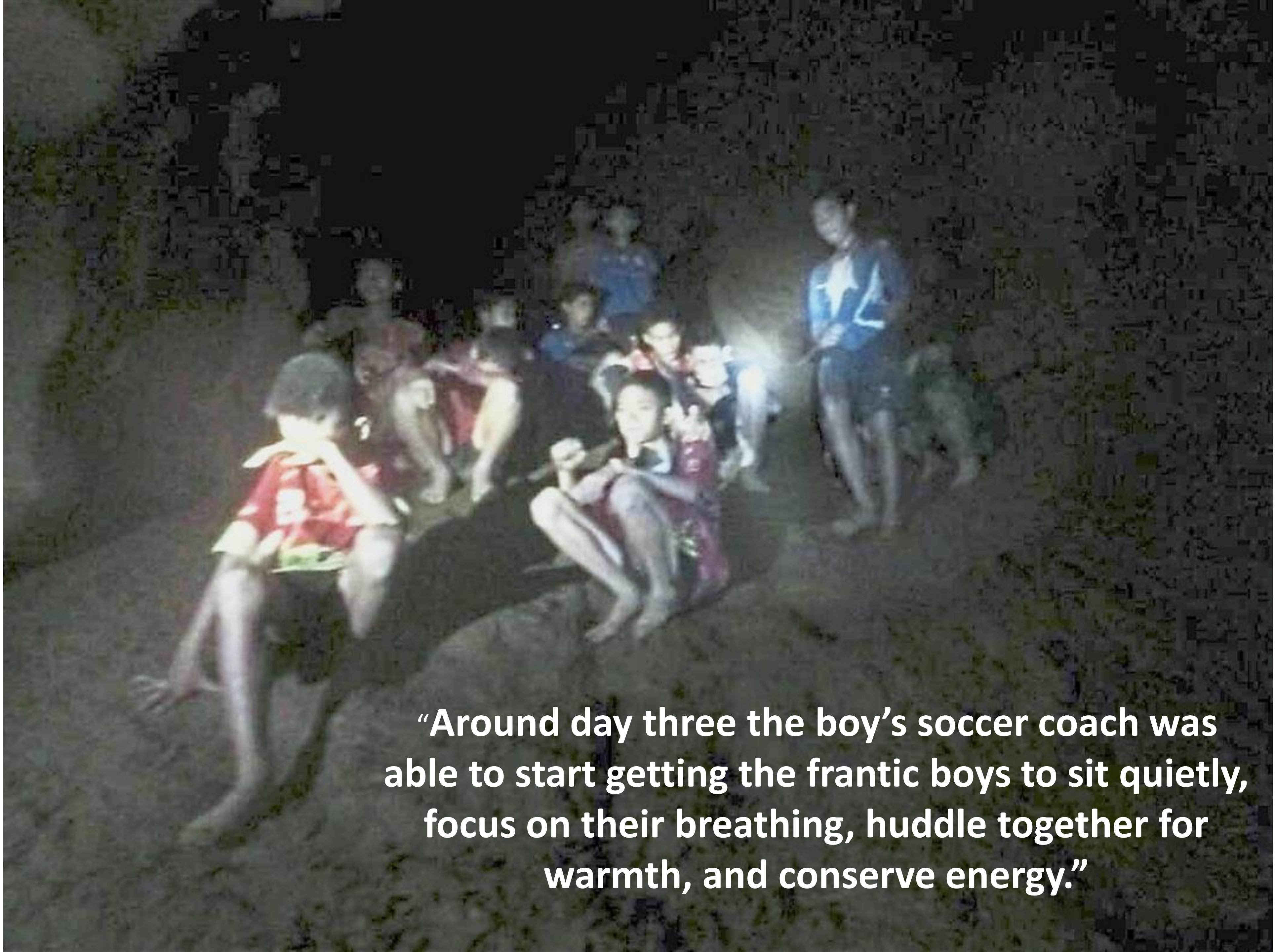
TWEET
mind full or mindful ?

How do you leave others feeling after you have crossed their path?

“How The Beatles’ Meditation Technique Could Cure Depression” THE INDEPENDENT	“Can Meditation Top Medication?” The Boston Globe	“Why Meditation Helps You Focus: Mindfulness Improves Brain Wiring In Just A Month” Daily Mail	“Meditation Boosts Part Of Brain Where ADD, Addictions Reside” ars technica	“Why Meditation And Orgasm Feel The Same To The Brain” THE HUFFINGTON POST
“Meditation Boosts Genes That Promote Good Health” NewScientist	“It’s Not Just For Your Brain: Meditating Can Actually Change Your DNA” FAST COMPANY	“We Need To Take Meditation More Seriously As Medicine” TIME	“Re-Wiring Your Brain For Happiness: How Meditation Can Physically Change The Brain” abc NEWS	“Meditation ‘Makes People More Intelligent By Growing The Brain’” The Telegraph
“Short-Circuit Stress: Mindful Meditation May Be Key To Better Sleep” TODAY	“Meditation Gives Brain A Charge” The Washington Post	“Mindfulness As Good As Antidepressant Drugs, Study Says” CNN	“Even Beginners Can Curb Pain With Meditation” npr	“Meditation Can Improve Your Memory, Focus, And Productivity At Work” lifehacker
“Meditation Gives Brain Power A Boost: Study” FOX NEWS	“Meditation Makes People More Rational Decision-Makers” USA TODAY	“Study: Meditation Improves Memory, Attention” the Atlantic	“Want A Sharp And Youthful Mind? Meditate” TORONTO STAR	“Meditation Is Proven To Be The Serene Way To Get Smarter” Daily Mail



“Around day three the boy’s soccer coach was able to start getting the frantic boys to sit quietly, focus on their breathing, huddle together for warmth, and conserve energy.”

Meditation trains us to “be” with whatever is happening rather than getting upset and over-reactive

Meditation myths:

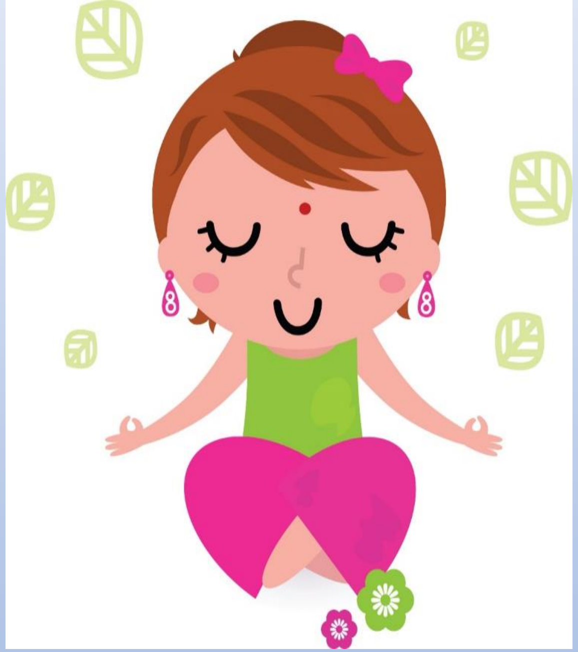
- It’s complicated
- You need to do it an hour a day
- You need to chant in a foreign language
- You need to empty your mind completely
- You need to sit in the lotus position
- You need to have a dedicated time and complete quiet
- It is a weird religious activity
- It will make you lose your edge
- Strange things will happen

Meditation is a powerful mindfulness practice…

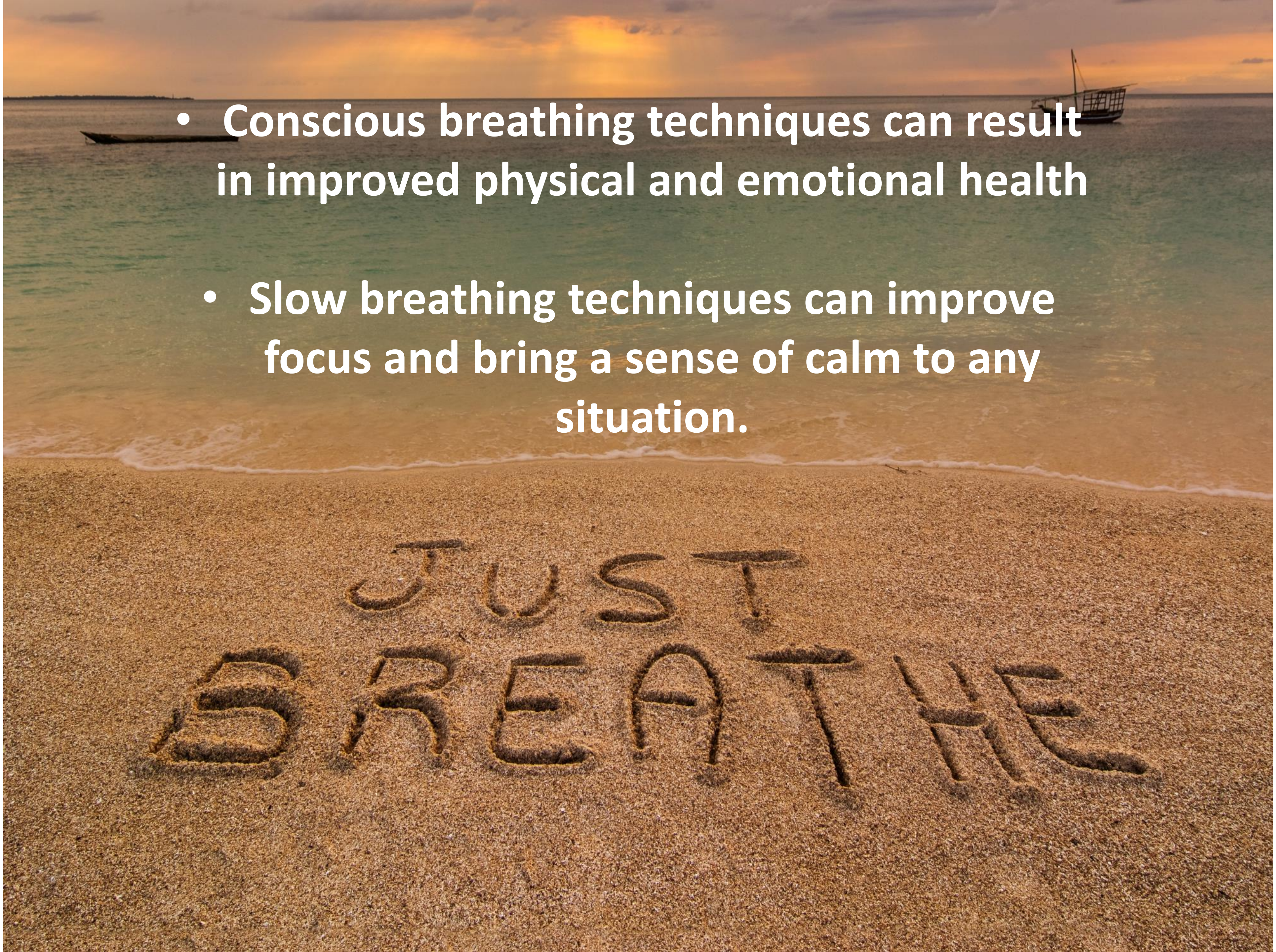
- Sleep
- Cardiovascular health
- Wound healing
- Immunity
- Focus
- Performance
- Memory
- Self Control

- Acute and Chronic Pain
- High blood pressure
- Inflammation
- Insomnia
- Stress
- Anxiety and depression
- PTSD
- Addictive behaviors

Fight or Flight



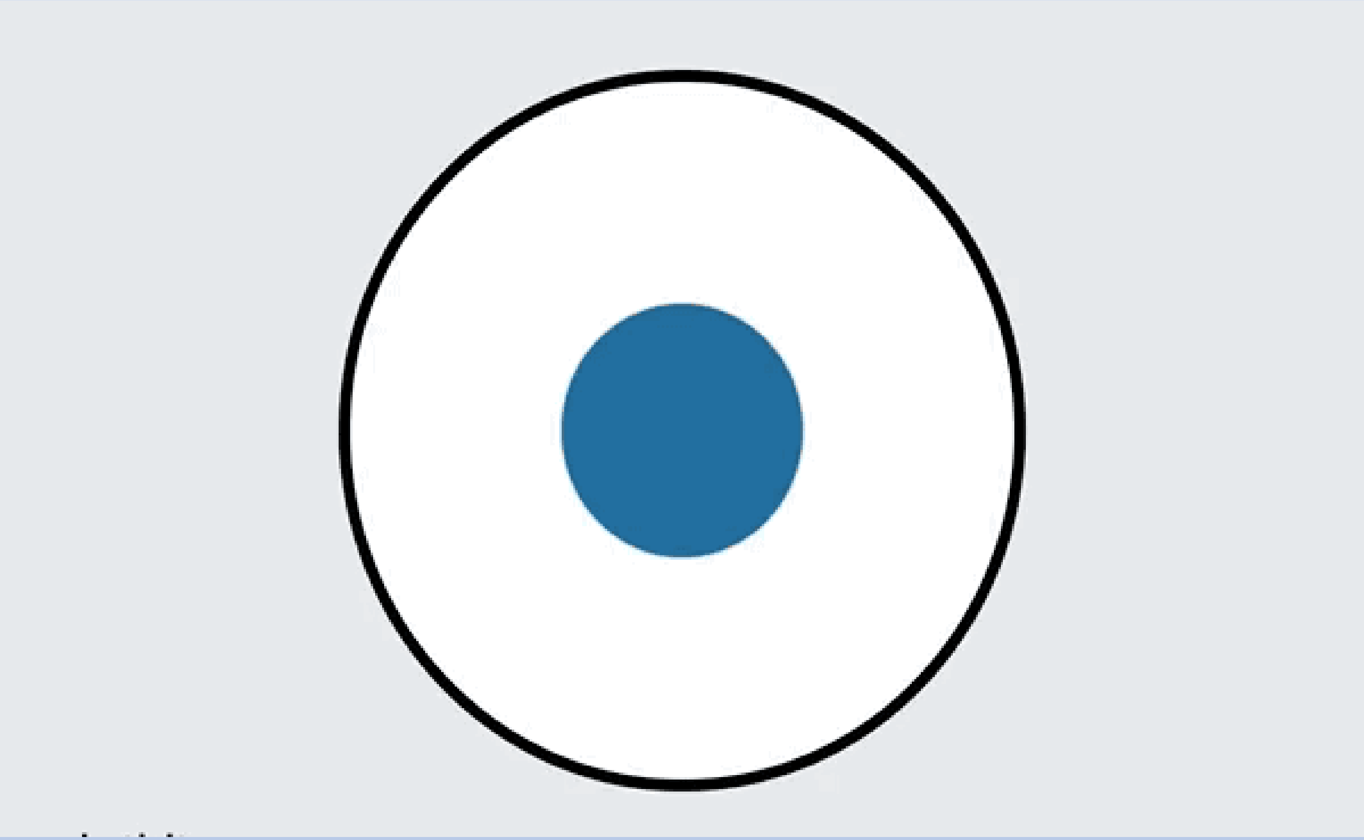
Rest and Restore



- Conscious breathing techniques can result in improved physical and emotional health

- Slow breathing techniques can improve focus and bring a sense of calm to any situation.

A Quick Shot of Calm…
Tactical or “Box” Breathing



Stressful event → **React with anger, frustration, and worry**

Stressful event → **Breathe, pause and think; respond calmly and consciously**

STOP,
BREATHE
& THINK APP
stopbreathethink.org

unplug
meditation

getting the
whole world
meditating
one person at
a time

www.insighttimer.com

10% HAPPIER
WITH DAN HARRIS

HEADSPACE®

Calm

- If you can sit quietly after difficult news…
- If in financial downturns you remain perfectly calm...
- If you can happily eat whatever is on your plate…
- If you can love everyone around you unconditionally…
- If you can always find contentment just where you are…

You are probably…

Thank you!

For more information:

melaniecarvell.com

melanieacarvell@gmail.com

RUNNING
with the
ANTELOPE

Life, Fitness, and Grit on the Northern Plains

Melanie Carvell

Foreword by Clay S. Jenkinson

BRADLEY MARINO MD
MMP, MSCE, MBA

DISCLOSURES

- See slide in presentation

Neurodevelopmental and Care Coordination Needs of Children with Congenital Heart Disease

Bradley S. Marino, MD, MPP, MSCE, MBA

Ronald and Helen Ross Distinguished Chair in Pediatric Cardiology
Executive Co-Director, Pediatric and Adult Congenital Heart Center
Cleveland Clinic Children’s

Conflict of Interest Disclosures for

Bradley S. Marino, MD, MPP, MSCE, MBA

Grant/Research Support	NIH (U24, RO1) Additional Ventures Single Ventricle Research Fund
Consultant	Nothing to disclose
Speakers Bureau	Nothing to disclose
Stock Shareholder	Nothing to disclose
Other (identify)	1. Creator of the Pediatric Cardiac Quality of Life Inventory 2. National leadership roles with the CNOC, CHPHC, AHA

STS Congenital Heart Surgery Data Summary

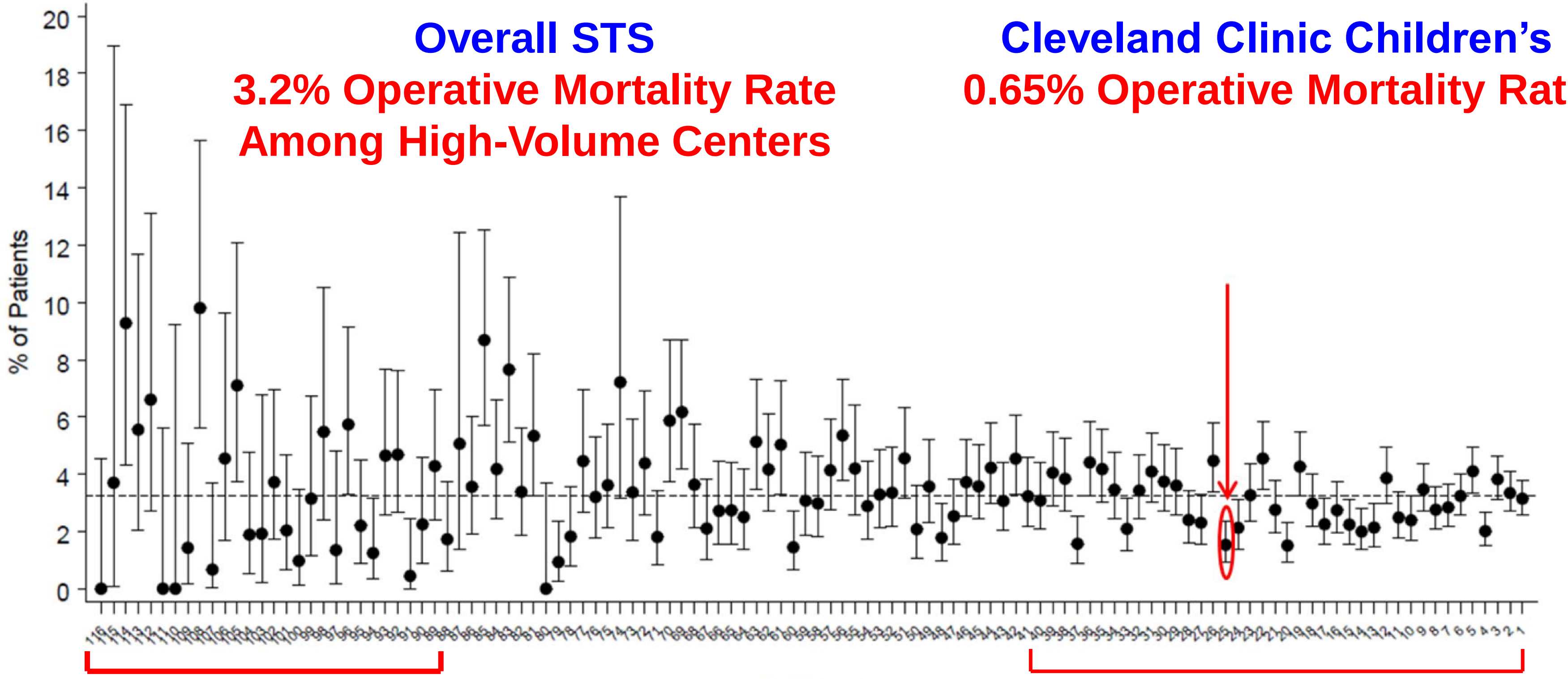
Duke Clinical Research Institute

STS Congenital Heart Surgery Operative Mortality
STS Period Jul 2015 - Jun 2019

Figure 1:

All Patients

Operative Mortality + 95% CI



Overall STS
3.2% Operative Mortality Rate Among High-Volume Centers

Cleveland Clinic Children's
0.65% Operative Mortality Rate

Low Volume Centers

High Volume Centers

Dotted line on graph represents overall value for the STS: 3.23%

Important Morbidities after Repair vs Palliation:

- Neurodevelopmental/Neurocognitive
- Psychosocial and Psychiatric
- Late surgery or catheter re-interventions
- Arrhythmias
- Chronic heart failure/transplantation
- Pulmonary hypertension
- Endocarditis
- Other important end organ dysfunction:
  - Chronic renal and liver insufficiency
  - Coagulopathy leading to thromboembolic complication

Important Morbidities after Repair vs Palliation:

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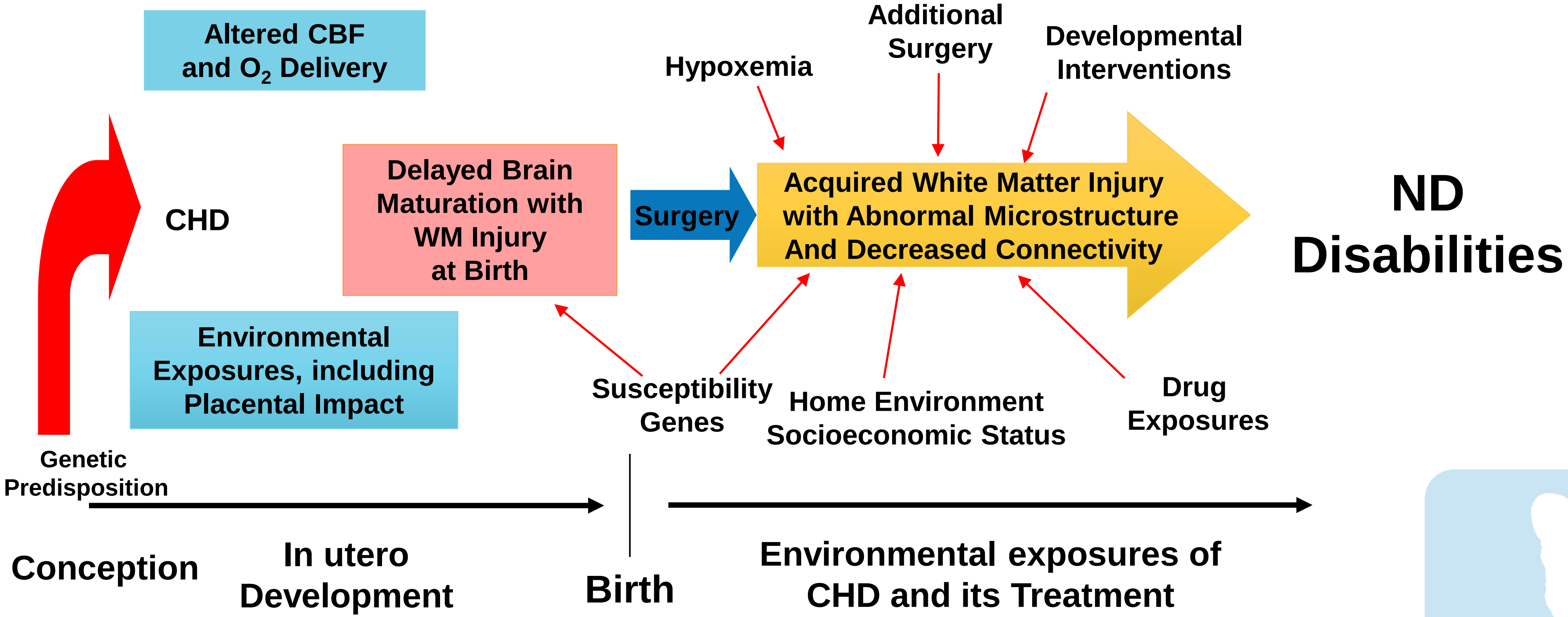
Model for Neurobehavioral Dysfunction in CHD Surgical Survivors



Altered CBF and O_2 Delivery

CHD

Environmental Exposures, including Placental Impact

Genetic Predisposition

Delayed Brain Maturation with WM Injury at Birth

Surgery

Hypoxemia

Additional Surgery

Developmental Interventions

Acquired White Matter Injury with Abnormal Microstructure And Decreased Connectivity

ND Disabilities

Susceptibility Genes

Home Environment Socioeconomic Status

Drug Exposures

Conception

In utero Development

Birth

Environmental exposures of CHD and its Treatment

Adapted from J. William Gaynor

ND Phenotype in CHD Survivors

- Mild cognitive impairment - normal to slightly lower IQ and Academic Achievement
- Impaired pragmatic language
- Decreased visual construction and perception
- Poor executive functioning
- Inattention and increased impulsivity (ADHD)
- Diminished fine and gross motor skills

Marino et al, Circulation, 2012

ND Phenotype in CHD Survivors

- Individual deficits or delays may be mild, but often occur across multiple domains
- “High prevalence – low severity” picture doesn’t meet classic criteria of a “learning disability”
- Many of these children have difficulties in school, but may not qualify for special services

Psychosocial Phenotype in CHD Survivors

- Impaired social interaction and deficits in social cognition

  Bellinger et al, *Cardiology Young*, 2008

- Impaired core communication skills – incidence of autism spectrum disorders

  Swillen et al, *Am J Med Gen*, 2015

- Increased incidence of Psychiatric disorders

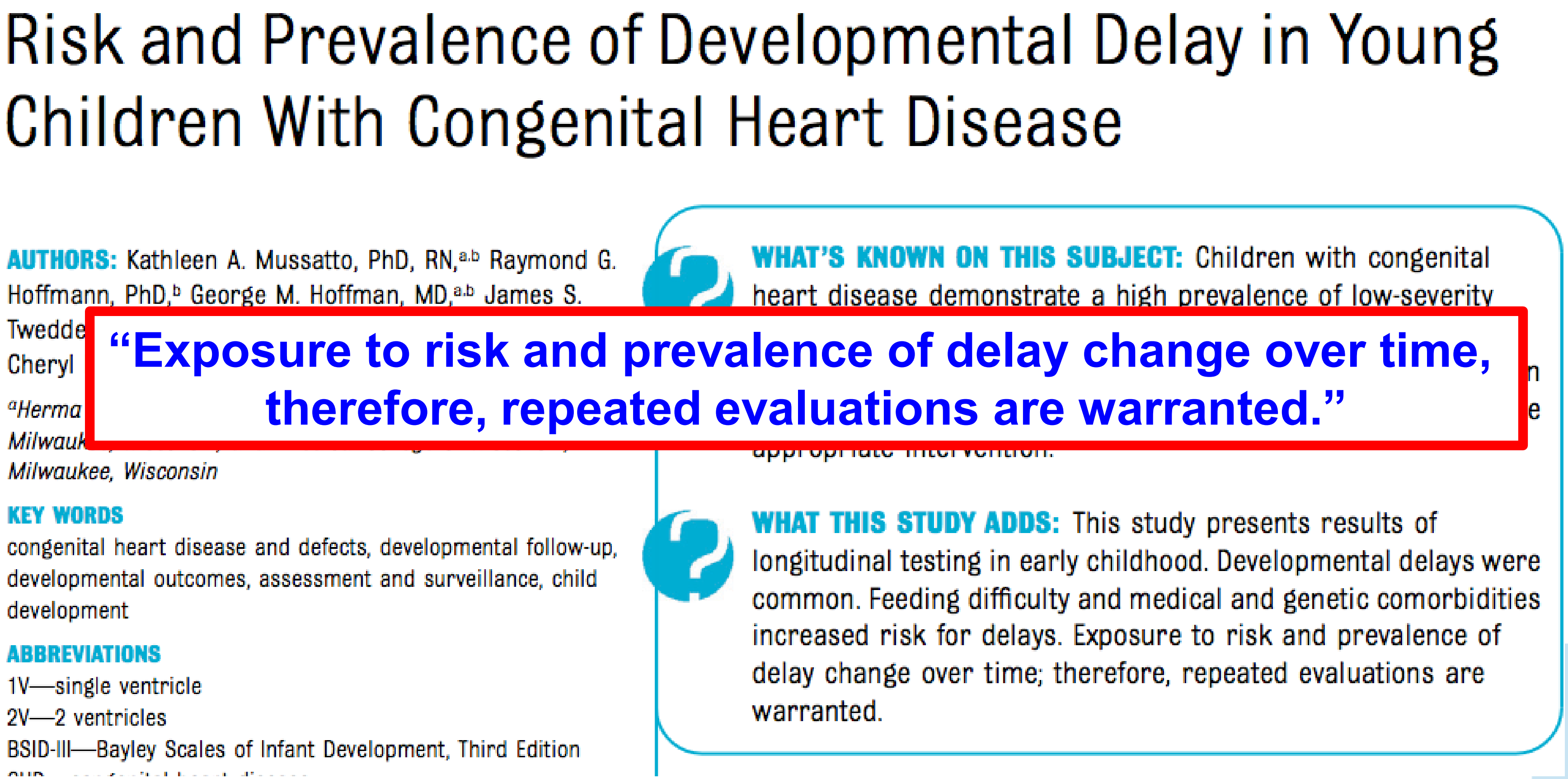
  Olsen et al, *Circulation*, 2011

- Issues with behavioral/emotional functioning

  Marino et al, *Circulation*, 2012

  - Anxiety
  - Depression
  - Post-traumatic stress

Developmental Delay Changes Over Time



Risk and Prevalence of Developmental Delay in Young Children With Congenital Heart Disease

AUTHORS: Kathleen A. Mussatto, PhD, RN,[a,b] Raymond G. Hoffmann, PhD,[b] George M. Hoffman, MD,[a,b] James S. Twedde

Cheryl

[a]Herma

Milwauk

Milwaukee, Wisconsin

KEY WORDS

congenital heart disease and defects, developmental follow-up, developmental outcomes, assessment and surveillance, child development

ABBREVIATIONS

1V—single ventricle

2V—2 ventricles

BSID-III—Bayley Scales of Infant Development, Third Edition

WHAT'S KNOWN ON THIS SUBJECT: Children with congenital heart disease demonstrate a high prevalence of low-severity

appropriate intervention.

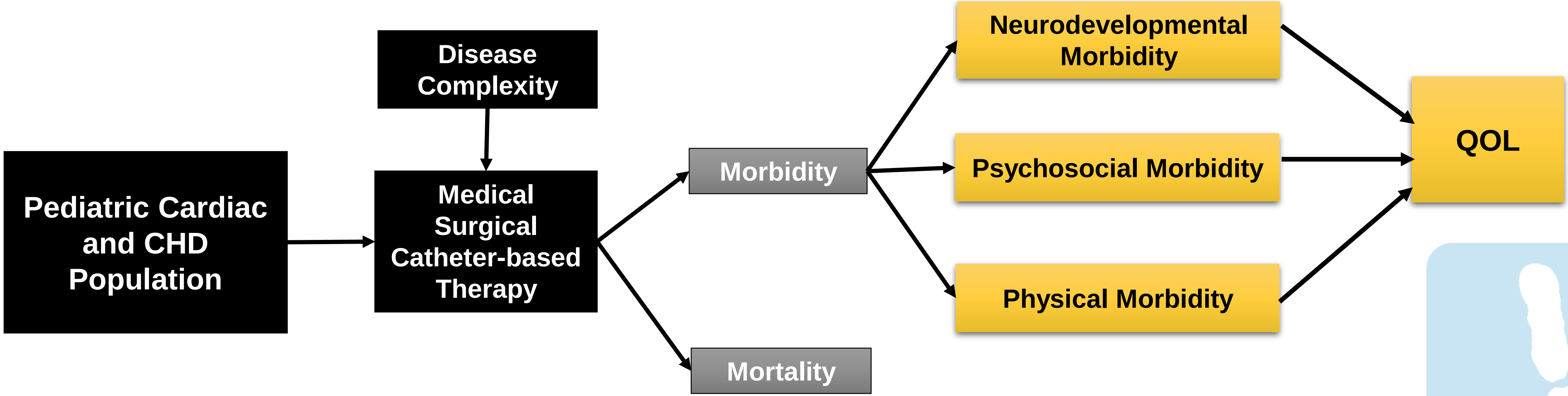
WHAT THIS STUDY ADDS: This study presents results of longitudinal testing in early childhood. Developmental delays were common. Feeding difficulty and medical and genetic comorbidities increased risk for delays. Exposure to risk and prevalence of delay change over time; therefore, repeated evaluations are warranted.

"Exposure to risk and prevalence of delay change over time, therefore, repeated evaluations are warranted."

Mussatto KW, *Pediatrics* 2014

How Should We Maximize Long-term Outcomes in Children with CHD?

PROM: Health-related Quality of Life is a new Vital Sign



Disease Complexity

Pediatric Cardiac and CHD Population

Medical Surgical Catheter-based Therapy

Morbidity

Mortality

Neurodevelopmental Morbidity

Psychosocial Morbidity

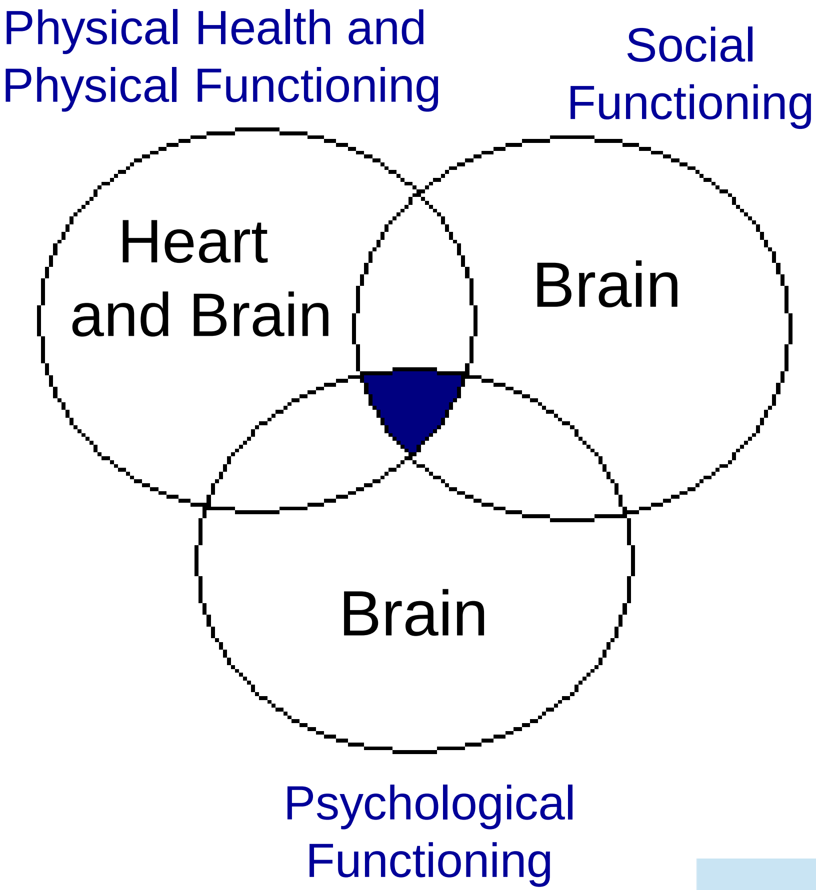
Physical Morbidity

QOL

Definition of Quality of Life (QOL)

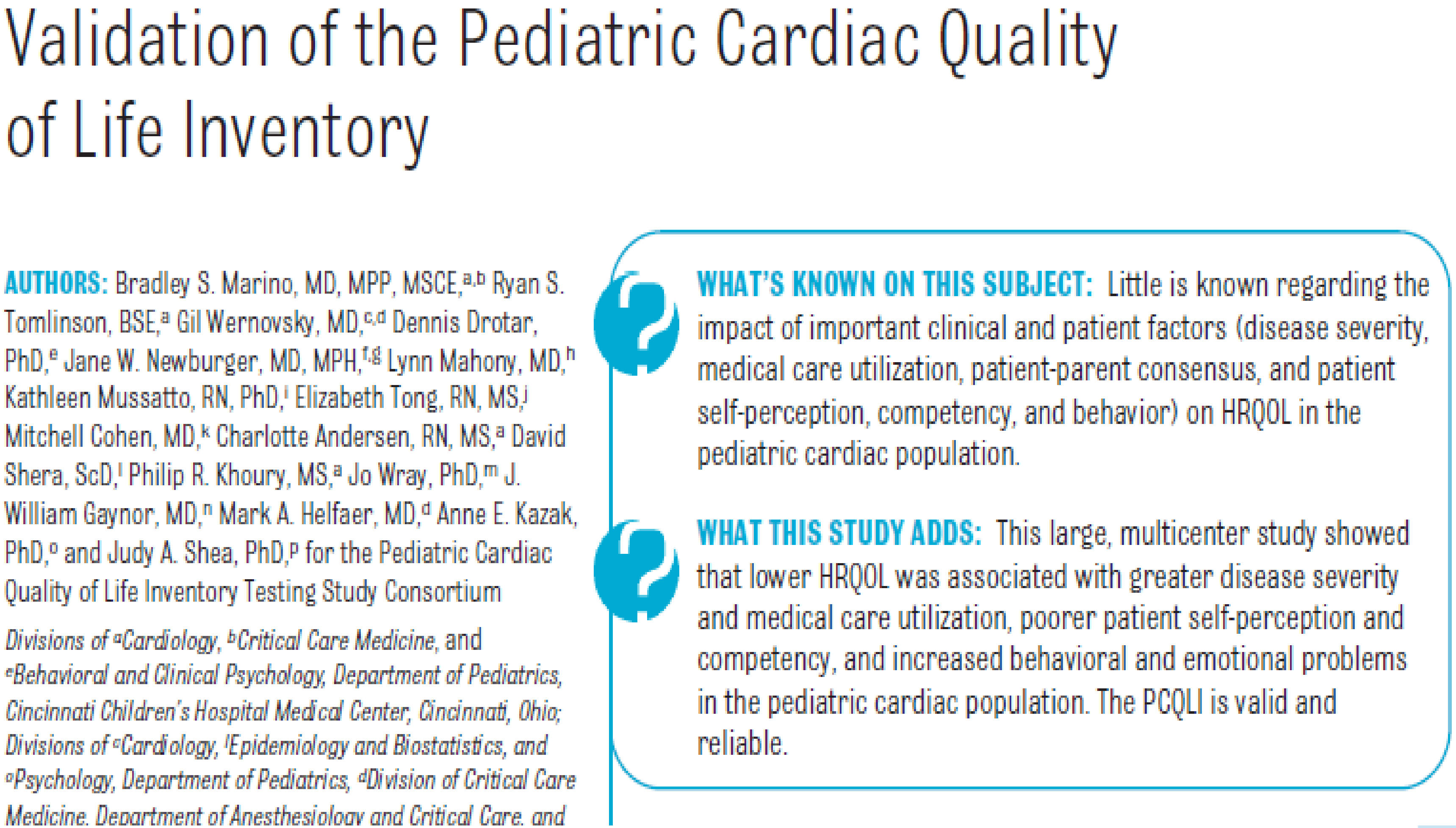
- **Physical Health and Physical Functioning**
- **Psychological Functioning**
- **Social Functioning**

- QOL describes a child’s ability to function in situational contexts (family, school, peer) and derive personal satisfaction from doing so



Ware 1984, Aaronson 1988

Quantitation of HRQOL with a Cardiac-Specific QOL Measure



Validation of the Pediatric Cardiac Quality of Life Inventory

AUTHORS: Bradley S. Marino, MD, MPP, MSCE,[a,b] Ryan S. Tomlinson, BSE,[a] Gil Wernovsky, MD,[c,d] Dennis Drotar, PhD,[e] Jane W. Newburger, MD, MPH,[f,g] Lynn Mahony, MD,[h] Kathleen Mussatto, RN, PhD,[i] Elizabeth Tong, RN, MS,[j] Mitchell Cohen, MD,[k] Charlotte Andersen, RN, MS,[a] David Shera, ScD,[l] Philip R. Khoury, MS,[a] Jo Wray, PhD,[m] J. William Gaynor, MD,[n] Mark A. Helfaer, MD,[d] Anne E. Kazak, PhD,[o] and Judy A. Shea, PhD,[p] for the Pediatric Cardiac Quality of Life Inventory Testing Study Consortium

Divisions of [a]Cardiology, [b]Critical Care Medicine, and *[e]Behavioral and Clinical Psychology, Department of Pediatrics, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio; Divisions of [c]Cardiology, [l]Epidemiology and Biostatistics, and [o]Psychology, Department of Pediatrics, [d]Division of Critical Care Medicine, Department of Anesthesiology and Critical Care, and*

WHAT'S KNOWN ON THIS SUBJECT: Little is known regarding the impact of important clinical and patient factors (disease severity, medical care utilization, patient-parent consensus, and patient self-perception, competency, and behavior) on HRQOL in the pediatric cardiac population.

WHAT THIS STUDY ADDS: This large, multicenter study showed that lower HRQOL was associated with greater disease severity and medical care utilization, poorer patient self-perception and competency, and increased behavioral and emotional problems in the pediatric cardiac population. The PCQLI is valid and reliable.

Marino et al, *Pediatrics* 2010

Pediatric Cardiac Quality of Life Inventory Research Consortium [19 Centers US and UK]

QOL in CHD survivors is lower than heart-healthy children

Mellion et al, J Pediatrics, 2014

QOL in CHD survivors worsens with increasing disease complexity

Marino et al, Quality of Life Research 2008
Marino et al, Pediatrics 2010
Wray et al, Cardiology Young 2012

Increasing medical care utilization (number of surgeries, cardiac catheterizations, hospitalizations, and physician visits annually) is associated with lower QOL in CHD survivors

Marino et al, Pediatrics 2010
Wray et al, Cardiology Young 2012

There is wide variation of QOL score within cardiac sub-groups

Marino et al, Pediatrics 2010
Wray et al, Cardiology Young 2012

“Nurturing” Neurodevelopmental and Psychosocial Resilience to Improve HRQOL

- Neurobehavioral and psychotherapy in the child and adolescent to minimize impact of social cognition issues, psychiatric issues, autism spectrum and affective disorders, and ADHD

- Psychosocial support to have the child improve their self-perception

- Therapy in the patient and parent to:
  - Prevent and treat PTSD symptomatology prior to invasive procedures, in the ICU, and during follow-up
  - Prevent and treat Anxiety and depression

- Programs to reduce parental stress relative to raising a child with chronic disease

Marino et al, *Pediatrics,* 2010
Marino et al, *Circulation,* 2012
Gerstle et al, J *Pediatrics,* 2016

Ernst, Marino et al, *Ped Cardiology,* 2018
Marino et al, *J Pediatr*, 2016
Qadir et al, *Pediatrics*, 2020 (Abstract – Submitted Manuscript)

AHA Scientific Statement

Neurodevelopmental Outcomes in Children With Congenital Heart Disease: Evaluation and Management

A Scientific Statement From the American Heart Association

This statement has been approved by the American Academy of Pediatrics.

Bradley S. Marino, MD, MPP, MSCE, FAHA, Co-Chair; Paul H. Lipkin, MD;
Jane W. Newburger, MD, MPH, FAHA; Georgina Peacock, MD, MPH; Marsha Gerdes, PhD;
J. William Gaynor, MD; Kathleen A. Mussatto, PhD, RN; Karen Uzark, PhD, CNP, FAHA;
Caren S. Goldberg, MD, MS; Walter H. Johnson, Jr, MD; Jennifer Li, MD;
Sabrina E. Smith, MD, PhD; David C. Bellinger, PhD; William T. Mahle, MD, FAHA, Co-Chair; on behalf of the American Heart Association Congenital Heart Defects Committee of the Council on Cardiovascular Disease in the Young, Council on Cardiovascular Nursing, and Stroke Council

Background—The goal of this statement was to review the available literature on surveillance, screening, evaluation, and management strategies and put forward a scientific statement that would comprehensively review the literature and create recommendations to optimize neurodevelopmental outcome in the pediatric congenital heart disease (CHD) population.

Methods and Results—A writing group appointed by the American Heart Association and American Academy of Pediatrics reviewed the available literature addressing developmental disorder and disability and developmental delay in the CHD population, with specific attention given to surveillance, screening, evaluation, and management strategies. MEDLINE and Google Scholar database searches from 1966 to 2011 were performed for English-language articles cross-referencing CHD with pertinent search terms. The reference lists of identified articles were also searched. The American College of Cardiology/American Heart Association classification of recommendations and levels of evidence for practice guidelines were used. A management algorithm was devised that stratified children with CHD on the basis of established risk factors. For those deemed to be at high risk for developmental disorder or disabilities or for developmental delay, formal, periodic developmental and medical evaluations are recommended. A CHD algorithm for surveillance, screening, evaluation, reevaluation, and management of developmental disorder or disability has been constructed to serve as a supplement to the 2006 American Academy of Pediatrics statement on developmental surveillance and screening. The proposed algorithm is designed to be carried out within the context of the medical home. This scientific statement is meant for medical providers within the medical home who care for patients with CHD.

Conclusions—Children with CHD are at increased risk of developmental disorder or disabilities or developmental delay. Periodic developmental surveillance, screening, evaluation, and reevaluation throughout childhood may enhance identification of significant deficits, allowing for appropriate therapies and education to enhance later academic, behavioral, psychosocial, and adaptive functioning. (*Circulation*. 2012;126:1143-1172.)

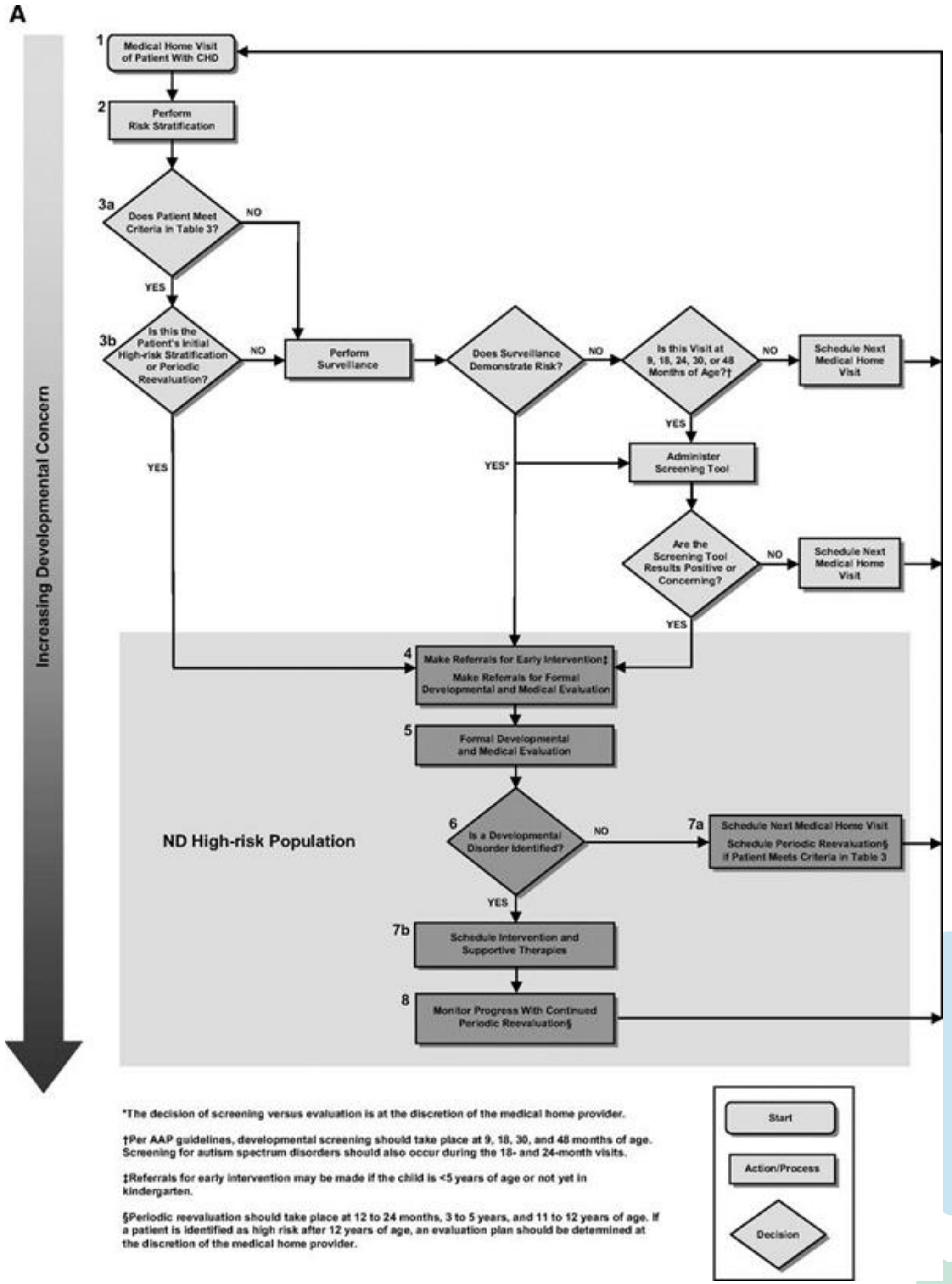
Marino et al, *Circulation,* 2012

Goals of ND Assessment in Pediatric Heart Disease and CHD Survivors

- To diagnose developmental disability and developmental delay through **Surveillance, Screening, Evaluation, and Management**
- To put interventions in place to prevent or treat the ND and Psychosocial phenotype noted in the pediatric cardiac population to maximize long-term outcome
  - To maximize health-related QOL
  - To maximize educational attainment
  - To reduce the incidence of anxiety, depression, and stress in family members and family dysfunction
  - To maximize adult transition and ACHD outcome

2012 AHA/AAP ND algorithm for children with CHD:

1. **Surveillance**
2. **Screening**
3. **Evaluation**
4. **Management**



*The decision of screening versus evaluation is at the discretion of the medical home provider.

†Per AAP guidelines, developmental screening should take place at 9, 18, 30, and 48 months of age. Screening for autism spectrum disorders should also occur during the 18- and 24-month visits.

‡Referrals for early intervention may be made if the child is <5 years of age or not yet in kindergarten.

§Periodic reevaluation should take place at 12 to 24 months, 3 to 5 years, and 11 to 12 years of age. If a patient is identified as high risk after 12 years of age, an evaluation plan should be determined at the discretion of the medical home provider.

AHA/AAP Risk Stratification: High-Risk Patients

- Neonates or infants requiring open heart surgery (cyanotic and acyanotic types)
- Children with other cyanotic heart lesions not requiring open heart surgery in the neonatal or infant period
- Children with any combination of CHD and other co-morbidities
- Other conditions determined at the discretion of the medical home providers

AHA/AAP Risk Stratification: High-Risk Patients

- CHD and ANY of the following co-morbidities
  - Prematurity (< 37 weeks)
  - Developmental delay recognized in infancy
  - Suspected genetic abnormality and/or syndrome
  - History of mechanical support (ECMO and/or VAD)
  - Heart transplantation
  - CPR at any point
  - Prolonged hospitalization (>2 week LOS in the hospital)
  - Perioperative seizures related to CHD surgery
  - Significant abnormalities on neuroimaging and/or microcephaly

AHA/AAP ND Recommendations For Patients Stratified as High-Risk

- Referral to formal developmental and medical evaluation
- Referral to early intervention services or early childhood special education services prior to confirmation of a specific developmental diagnosis
- Periodic re-evaluations for DDs and developmental delays at 12-24 mo, 3-5 yrs, and 11-12 yrs of age
- Referral of young adults for higher education and/or vocational counseling

CNOC

Cardiac Neurodevelopmental Outcome Collaborative

CNOC Vision

- Multi-center, multi-national, multi-disciplinary group of healthcare professionals committed to working together and partnering with families to optimize neurodevelopmental outcomes for individuals with pediatric and congenital heart disease through ***clinical, quality, and research initiatives, intending to maximize QOL across the lifespan.***

3 Countries
46 Participating Institutions
1000 Members

CNOC
Cardiac Neurodevelopmental
Outcome Collaborative

Cardiac Neurodevelopmental Outcome Collaborative (CNOC) Evaluation Protocols

Cardiology in the Young

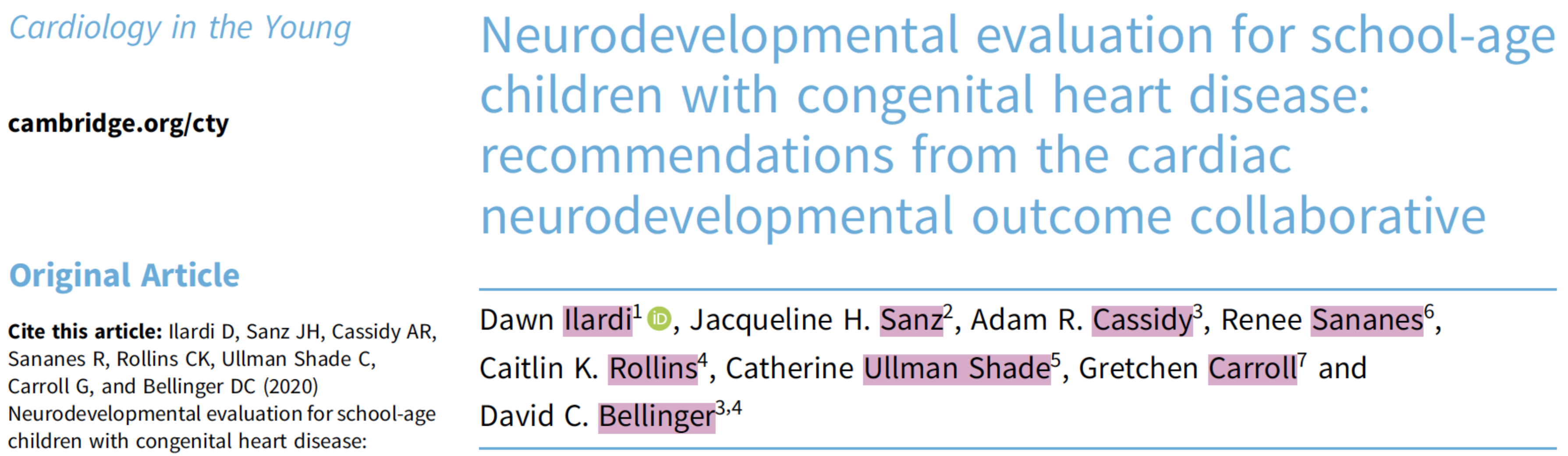
cambridge.org/cty

Original Article

Cite this article: Ware J, Butcher JL, Latal B, Sadhwani A, Rollins CK, Brosig Soto CL, Butler SC, Eiler-Sims PB, Ullman Shade CV, and Wernovsky G (2020) Neurodevelopmental evaluation strategies for children with congenital heart disease aged birth through 5 years: recommendations from the cardiac

Neurodevelopmental evaluation strategies for children with congenital heart disease aged birth through 5 years: recommendations from the cardiac neurodevelopmental outcome collaborative

Janice Ware[1], Jennifer L. Butcher[2], Beatrice Latal[3,4], Anjali Sadhwani[5], Caitlin K. Rollins[6], Cheryl L. Brosig Soto[7], Samantha C. Butler[5], Patricia B. Eiler-Sims[8], Catherine V. Ullman Shade[9] and Gil Wernovsky[10,11,12]



Cardiology in the Young

cambridge.org/cty

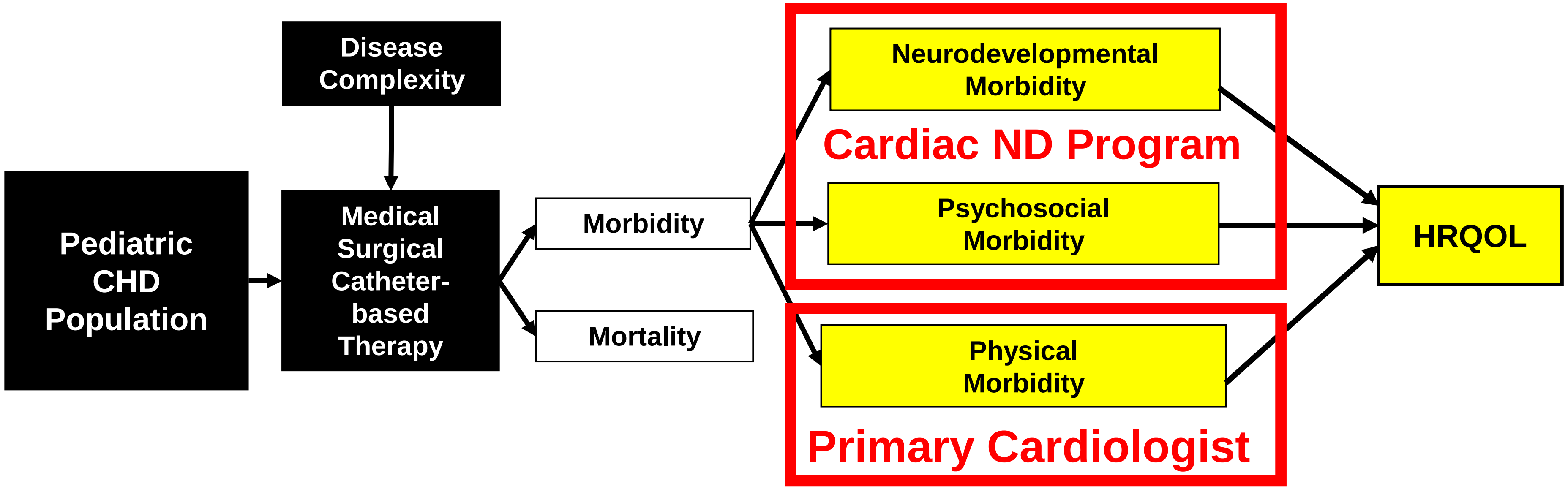
Original Article

Cite this article: Ilardi D, Sanz JH, Cassidy AR, Sananes R, Rollins CK, Ullman Shade C, Carroll G, and Bellinger DC (2020) Neurodevelopmental evaluation for school-age children with congenital heart disease:

Neurodevelopmental evaluation for school-age children with congenital heart disease: recommendations from the cardiac neurodevelopmental outcome collaborative

Dawn Ilardi[1], Jacqueline H. Sanz[2], Adam R. Cassidy[3], Renee Sananes[6], Caitlin K. Rollins[4], Catherine Ullman Shade[5], Gretchen Carroll[7] and David C. Bellinger[3,4]

How Should We Care for Children with Heart Disease and their Families Long-term?



Disease Complexity

Pediatric CHD Population

Medical Surgical Catheter-based Therapy

Morbidity

Mortality

Neurodevelopmental Morbidity

Cardiac ND Program

Psychosocial Morbidity

Physical Morbidity

Primary Cardiologist

HRQOL

The Cardiac ND Program Addresses a Critical Gap in How We Care for Children with Heart Disease and their Families

Monitoring and Intervening on Development

NICU-Cardiac Neurodevelopmental Program

Supporting development throughout childhood and adolescence

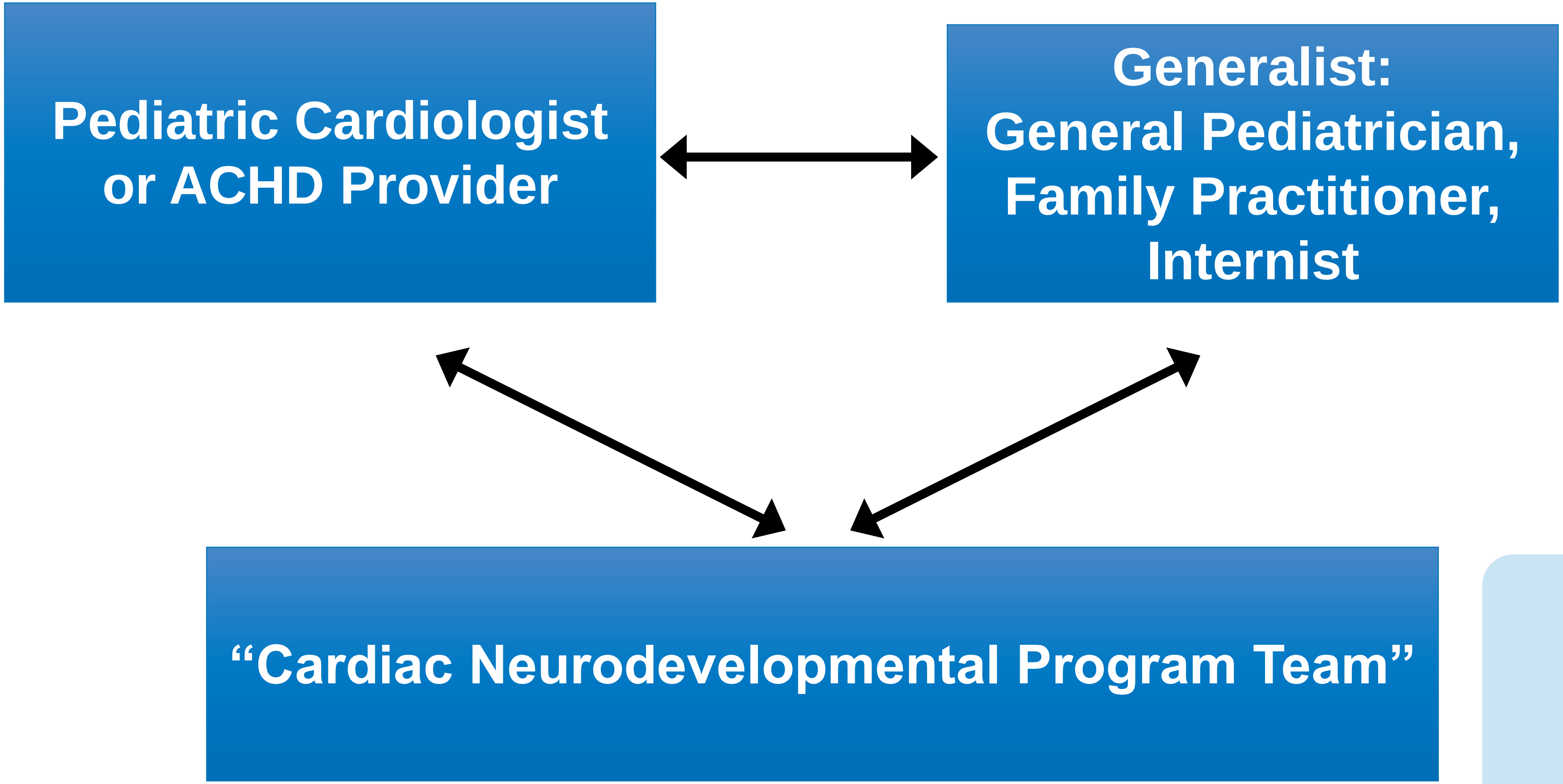
Ann & Robert H. Lurie Children's Hospital of Chicago

Cardiac ND Follow-Up Clinic at

Ann & Robert H. Lurie Children's Hospital of Chicago

- Offered to high risk NICU/Cardiac patients
- Regular developmental evaluations infancy through adolescence
- Psychology and Neuropsychology evaluation and management
- Physical, occupational, speech therapy
- Dieticians, social workers, nurses and APNs
- Care from Developmental Pediatricians and Special Educators

Care Model within the Medical Home



Pediatric Cardiologist or ACHD Provider

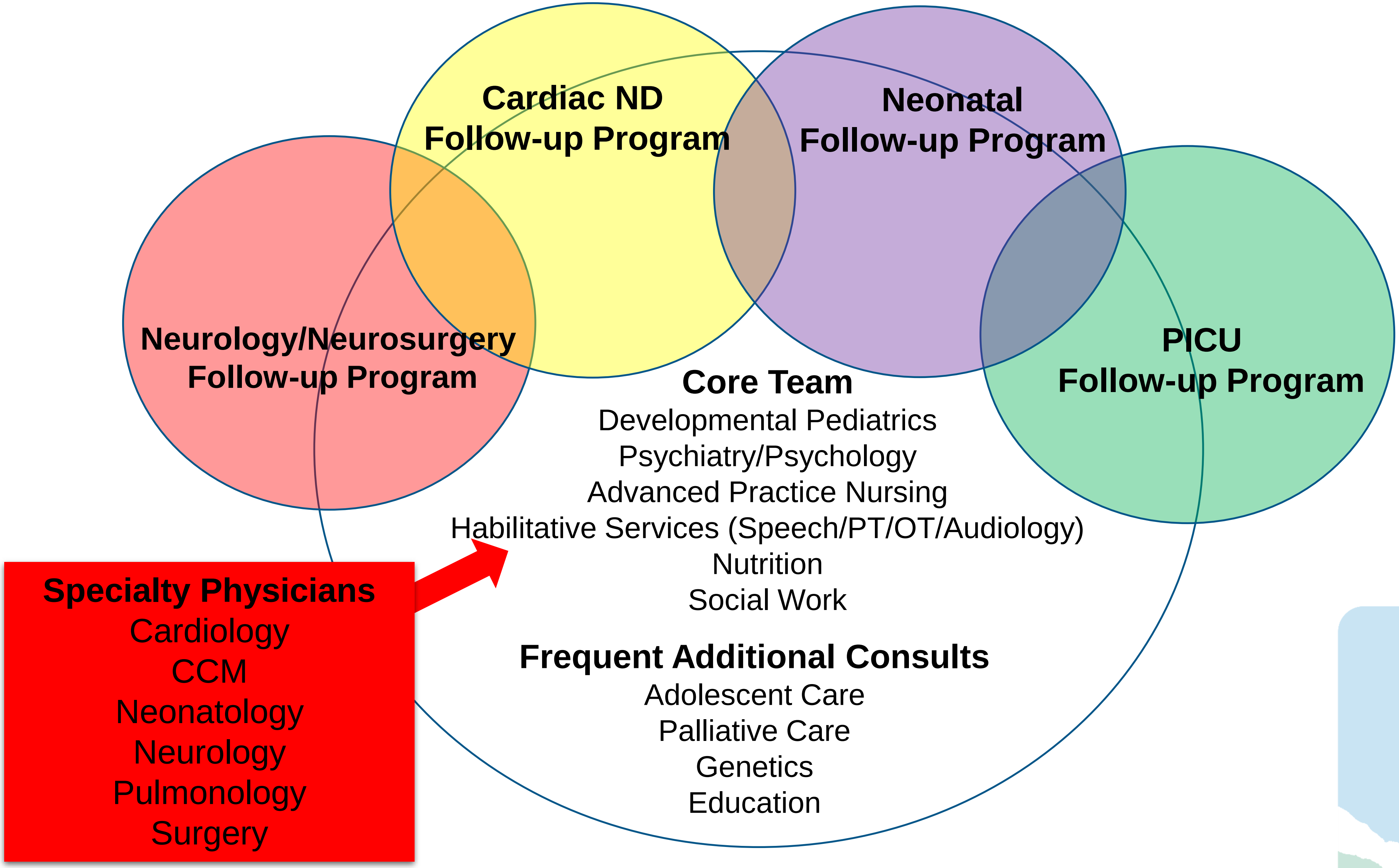
Generalist:
General Pediatrician,
Family Practitioner,
Internist

“Cardiac Neurodevelopmental Program Team”

Care Model Within the Medical Home

- Separate high-risk NICU and cardiac populations and have two separate clinics with no interaction
- Separate NICU and cardiac populations, but non-MD core staff work in both for efficiency
- Single developmental follow-up program for high-risk NICU and cardiac populations
- Singular developmental follow-up medical home for high-risk children that includes many populations

Development of a Singular Developmental Medical Home for High-Risk Children



Cardiac ND
Follow-up Program

Neonatal
Follow-up Program

Neurology/Neurosurgery
Follow-up Program

PICU
Follow-up Program

Core Team
Developmental Pediatrics
Psychiatry/Psychology
Advanced Practice Nursing
Habilitative Services (Speech/PT/OT/Audiology)
Nutrition
Social Work

Frequent Additional Consults
Adolescent Care
Palliative Care
Genetics
Education

Specialty Physicians
Cardiology
CCM
Neonatology
Neurology
Pulmonology
Surgery

The “CNP’ Team

- “Developmental Intensivist”
- Developmental Pediatrician
- Physical Therapist/Occupational Therapist
- Educator
- Access to:
  - Psychologist/Neuropsychologist
  - Geneticist
  - Neurologist
  - Dietician
  - Speech Language Pathologist

The “CNP’ Team

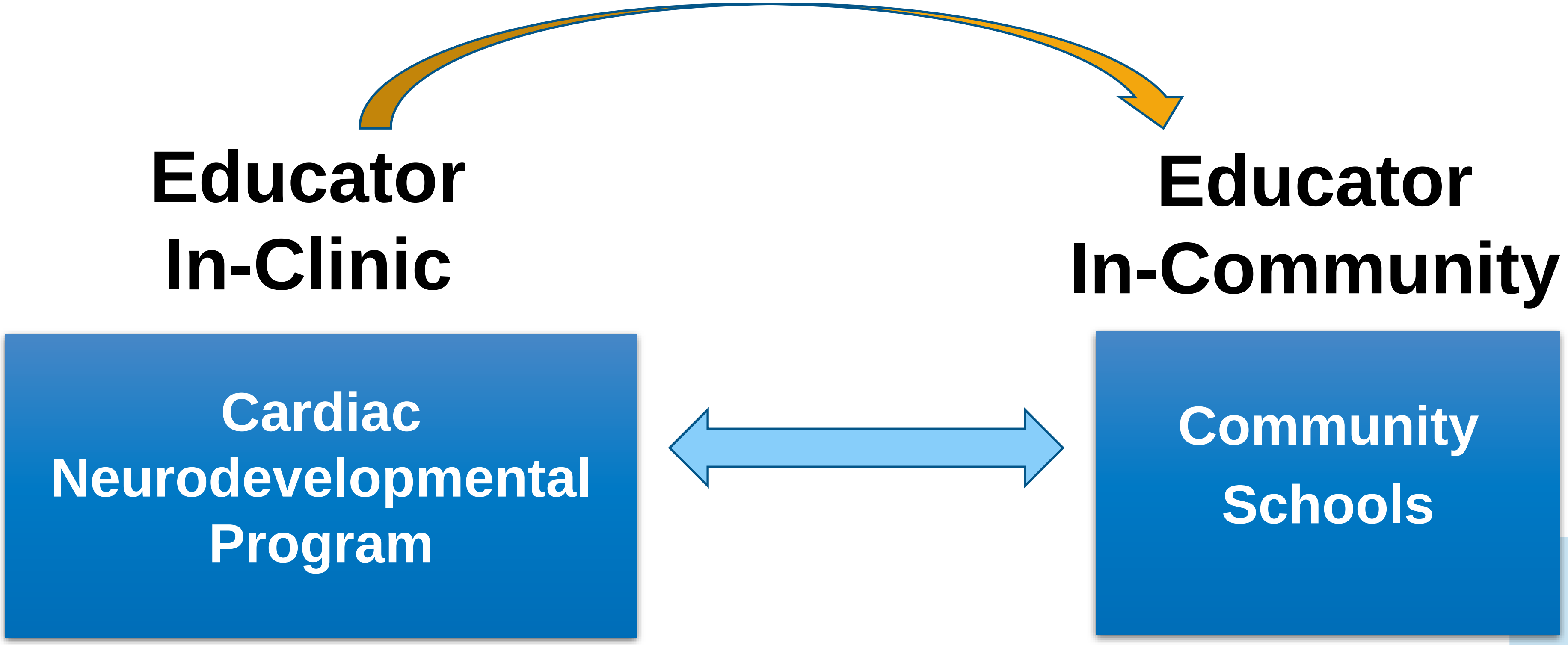
- “Developmental Intensivist”
- Developmental Pediatrician
- Physical Therapist/Occupational Therapist
- **Educator**
- Access to:
  - Psychologist/Neuropsychologist
  - Geneticist
  - Neurologist
  - Dietician
  - Speech Language Pathologist

Learning Challenges Are Common in CHD

- Students with complex CHD have generally lower academic achievement than typical peers
  - **1/3-1/2** of students with complex CHD receive special education programming
  - **1 in 6** are placed in substantially-separate classrooms
  - **1 in 5** repeat a grade at least once
  - **1 in 4** receive occupational therapy, physical therapy, speech therapy, and/or psychotherapy

Bellinger et al., 2011; Bellinger, Watson, et al., 2015; Jaworski, White, DeMaso, Newburger, Bellinger, & Cassidy, 2017; Shillingford et al., 2008

School Intervention Program Follow-Through



Educator
In-Clinic

Educator
In-Community

Cardiac Neurodevelopmental Program

Community Schools

Neurodevelopmental Evaluation without Communication to the Schools

Getting Patients into Clinics –
The “Big Lie” and the “Grand Re-education”

Cardiovascular Care AND Neurodevelopmental and Psychosocial Support Across the Lifespan

Fetal Life

Neonate/Infant

Child/Adolescent

Adolescent-
Young Adult

Fetal Care Center

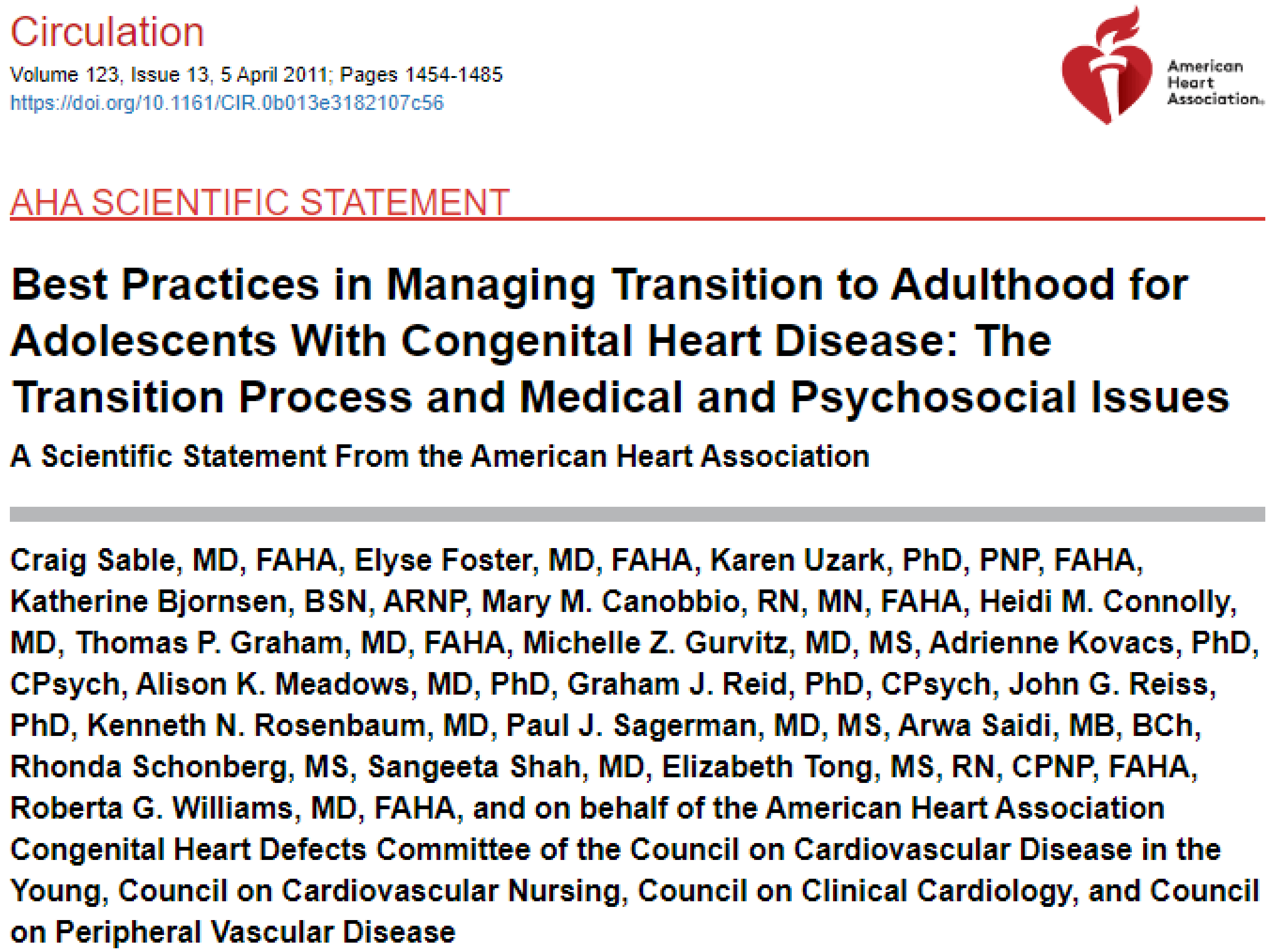
Perioperative CICU

Developmental Support Program

Cardiovascular Bridge Programs

Comprehensive Cardiovascular Care
AND
Neurodevelopmental and Psychosocial Follow-up

AHA Best Practices for ACHD Transition



Circulation

Volume 123, Issue 13, 5 April 2011; Pages 1454-1485

https://doi.org/10.1161/CIR.0b013e3182107c56

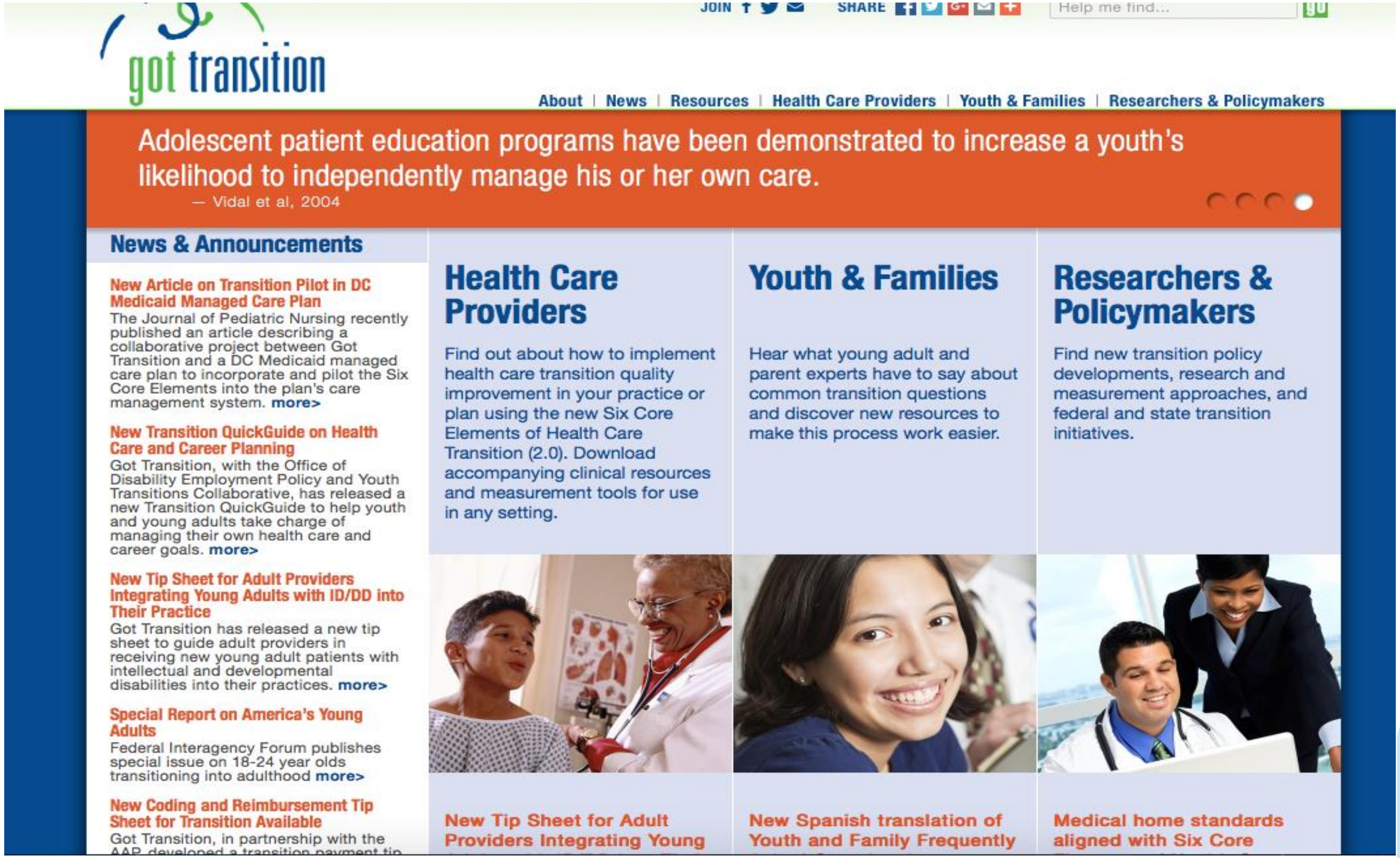
AHA SCIENTIFIC STATEMENT

Best Practices in Managing Transition to Adulthood for Adolescents With Congenital Heart Disease: The Transition Process and Medical and Psychosocial Issues

A Scientific Statement From the American Heart Association

Craig Sable, MD, FAHA, Elyse Foster, MD, FAHA, Karen Uzark, PhD, PNP, FAHA, Katherine Bjornsen, BSN, ARNP, Mary M. Canobbio, RN, MN, FAHA, Heidi M. Connolly, MD, Thomas P. Graham, MD, FAHA, Michelle Z. Gurvitz, MD, MS, Adrienne Kovacs, PhD, CPsych, Alison K. Meadows, MD, PhD, Graham J. Reid, PhD, CPsych, John G. Reiss, PhD, Kenneth N. Rosenbaum, MD, Paul J. Sagerman, MD, MS, Arwa Saidi, MB, BCh, Rhonda Schonberg, MS, Sangeeta Shah, MD, Elizabeth Tong, MS, RN, CPNP, FAHA, Roberta G. Williams, MD, FAHA, and on behalf of the American Heart Association Congenital Heart Defects Committee of the Council on Cardiovascular Disease in the Young, Council on Cardiovascular Nursing, Council on Clinical Cardiology, and Council on Peripheral Vascular Disease

Transition



Adolescent patient education programs have been demonstrated to increase a youth's likelihood to independently manage his or her own care.

— Vidal et al, 2004

News & Announcements

New Article on Transition Pilot in DC Medicaid Managed Care Plan
The Journal of Pediatric Nursing recently published an article describing a collaborative project between Got Transition and a DC Medicaid managed care plan to incorporate and pilot the Six Core Elements into the plan's care management system. more>

New Transition QuickGuide on Health Care and Career Planning
Got Transition, with the Office of Disability Employment Policy and Youth Transitions Collaborative, has released a new Transition QuickGuide to help youth and young adults take charge of managing their own health care and career goals. more>

New Tip Sheet for Adult Providers Integrating Young Adults with ID/DD into Their Practice
Got Transition has released a new tip sheet to guide adult providers in receiving new young adult patients with intellectual and developmental disabilities into their practices. more>

Special Report on America's Young Adults
Federal Interagency Forum publishes special issue on 18-24 year olds transitioning into adulthood more>

New Coding and Reimbursement Tip Sheet for Transition Available
Got Transition, in partnership with the AAP developed a transition payment tip

Health Care Providers

Find out about how to implement health care transition quality improvement in your practice or plan using the new Six Core Elements of Health Care Transition (2.0). Download accompanying clinical resources and measurement tools for use in any setting.

New Tip Sheet for Adult Providers Integrating Young

Youth & Families

Hear what young adult and parent experts have to say about common transition questions and discover new resources to make this process work easier.

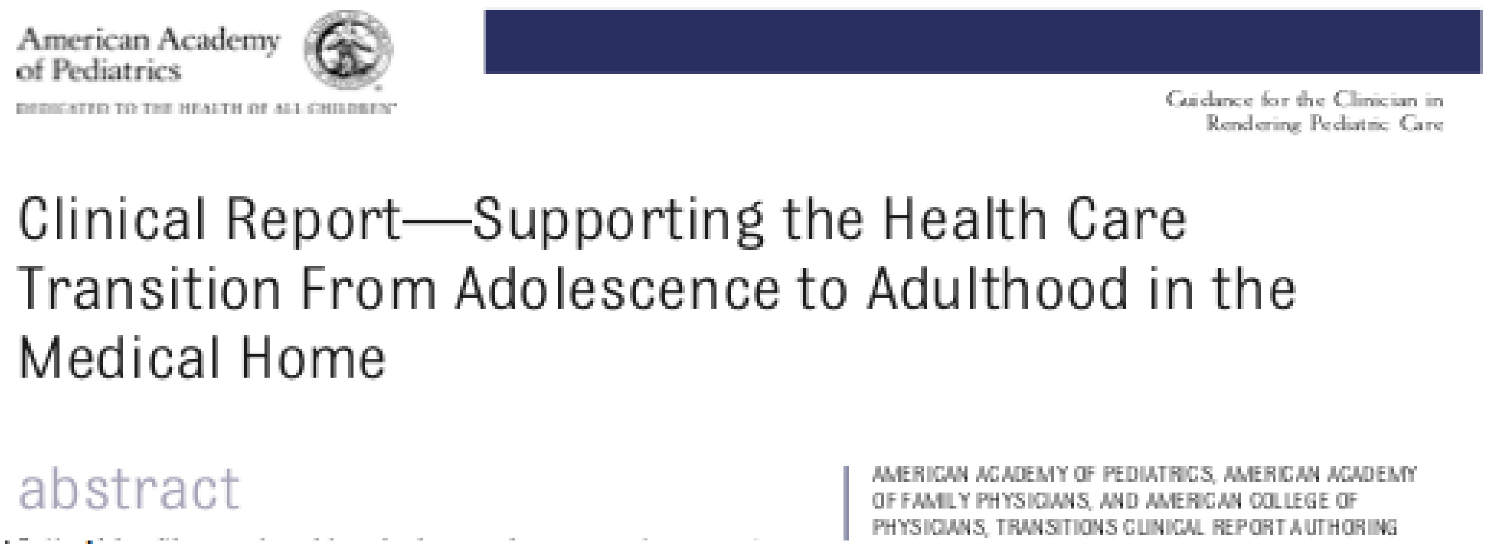
New Spanish translation of Youth and Family Frequently

Researchers & Policymakers

Find new transition policy developments, research and measurement approaches, and federal and state transition initiatives.

Medical home standards aligned with Six Core

www.gottransition.org



American Academy of Pediatrics

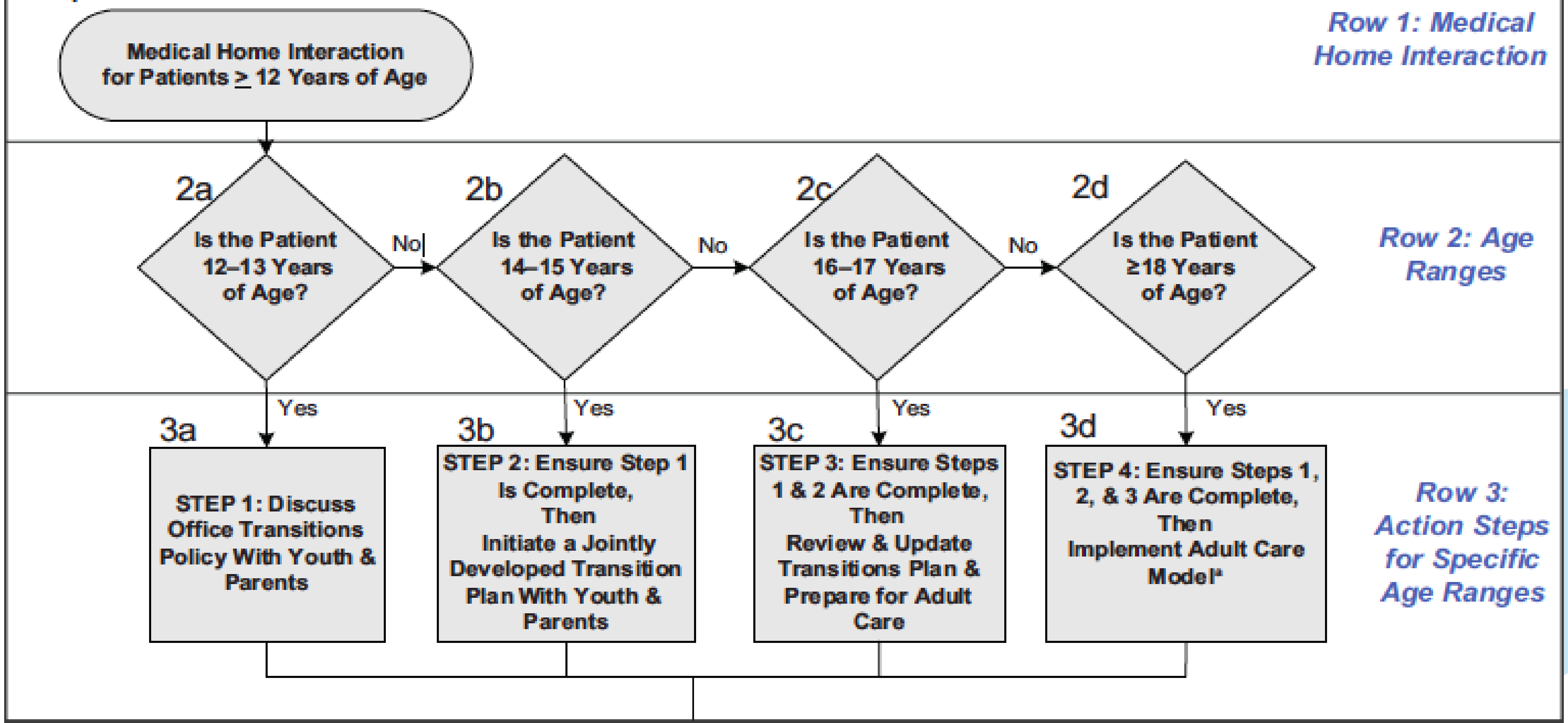
DEDICATED TO THE HEALTH OF ALL CHILDREN™

Guidance for the Clinician in
Rendering Pediatric Care

Clinical Report—Supporting the Health Care Transition From Adolescence to Adulthood in the Medical Home

abstract

AMERICAN ACADEMY OF PEDIATRICS, AMERICAN ACADEMY OF FAMILY PHYSICIANS, AND AMERICAN COLLEGE OF PHYSICIANS, TRANSITIONS CLINICAL REPORT AUTHORING



Medical Home Interaction for Patients ≥ 12 Years of Age

2a Is the Patient 12–13 Years of Age?

2b Is the Patient 14–15 Years of Age?

2c Is the Patient 16–17 Years of Age?

2d Is the Patient ≥18 Years of Age?

3a STEP 1: Discuss Office Transitions Policy With Youth & Parents

3b STEP 2: Ensure Step 1 Is Complete, Then Initiate a Jointly Developed Transition Plan With Youth & Parents

3c STEP 3: Ensure Steps 1 & 2 Are Complete, Then Review & Update Transitions Plan & Prepare for Adult Care

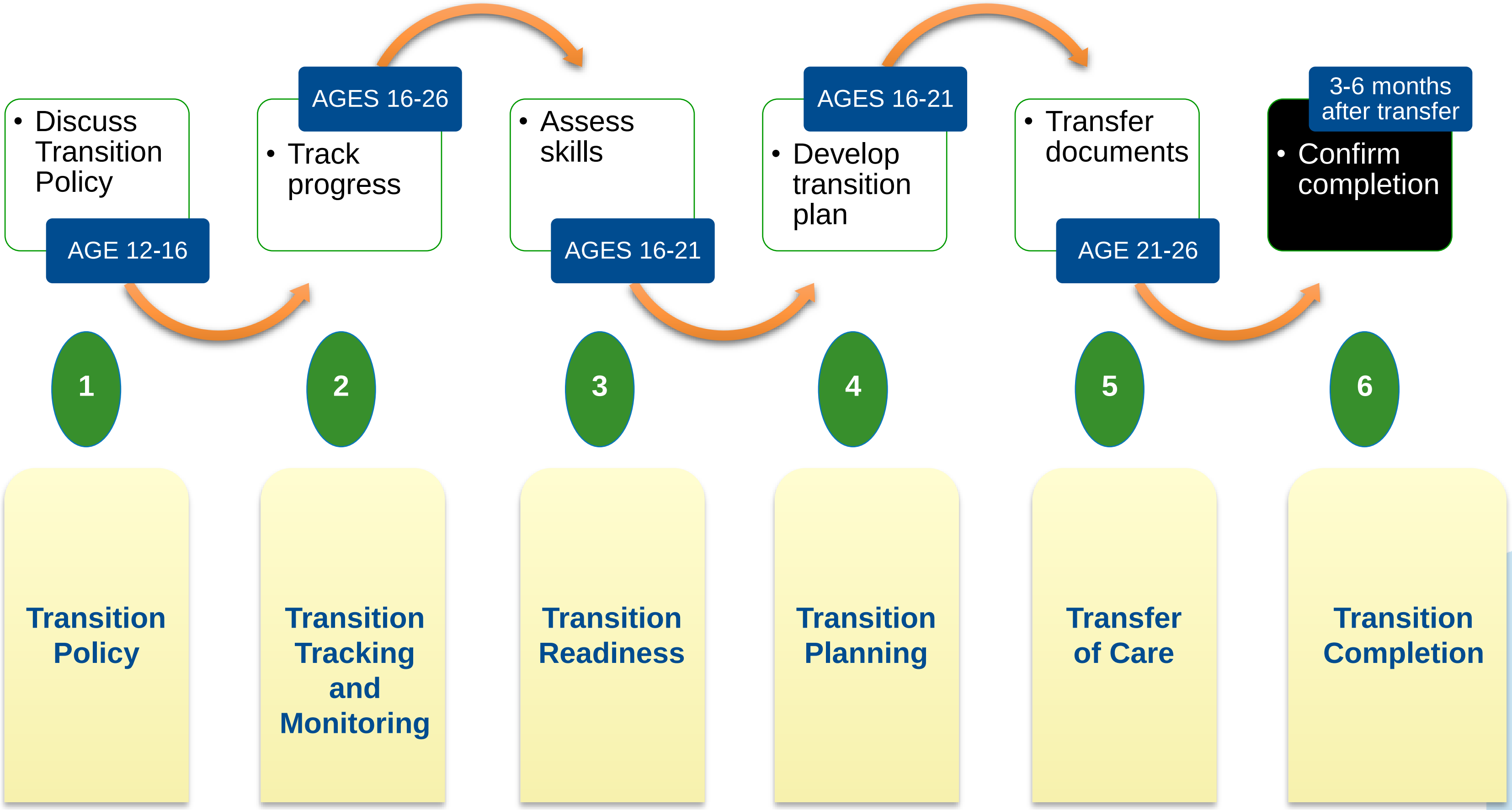
3d STEP 4: Ensure Steps 1, 2, & 3 Are Complete, Then Implement Adult Care Model[a]

Row 1: Medical Home Interaction

Row 2: Age Ranges

Row 3: Action Steps for Specific Age Ranges

Six Core Elements of Transition



- Discuss Transition Policy

AGE 12-16

1

Transition Policy

AGES 16-26

- Track progress

2

Transition Tracking and Monitoring

- Assess skills

AGES 16-21

3

Transition Readiness

AGES 16-21

- Develop transition plan

4

Transition Planning

- Transfer documents

AGE 21-26

5

Transfer of Care

3-6 months after transfer

- Confirm completion

6

Transition Completion

Summary

- CHD is the most common birth defect
- Survival after neonatal heart surgery is expected
- Neurobehavioral dysfunction in CHD surgical survivors results from white matter injury
- Maximize long-term HRQOL though:
  - Adhering to AHA/AAP and CNOC evaluation and management guidelines
  - Monitor and intervene on development through cardiac neurodevelopmental programs
  - Appropriately “transition” teens with CHD to ACHD medical care and mental health support programs

Cleveland Clinic Children’s

JOAN CONNELL MD
FAAP, MPH, MS, RPh

DISCLOSURES

- Nothing to disclose

Comprehensive Care Coordination within ND

Where are We Now, Where Could We Be?

Joan Connell, MD MPH
Medical Director, NDDHHS Newborn Screening Program

Objectives

- Review Definition/Concept of Comprehensive Care Coordination
- Discuss models of care coordination currently used in North Dakota
- Determine what modifications are necessary to make comprehensive care coordination available to children with special health care needs in North Dakota

1. Defining Care Coordination

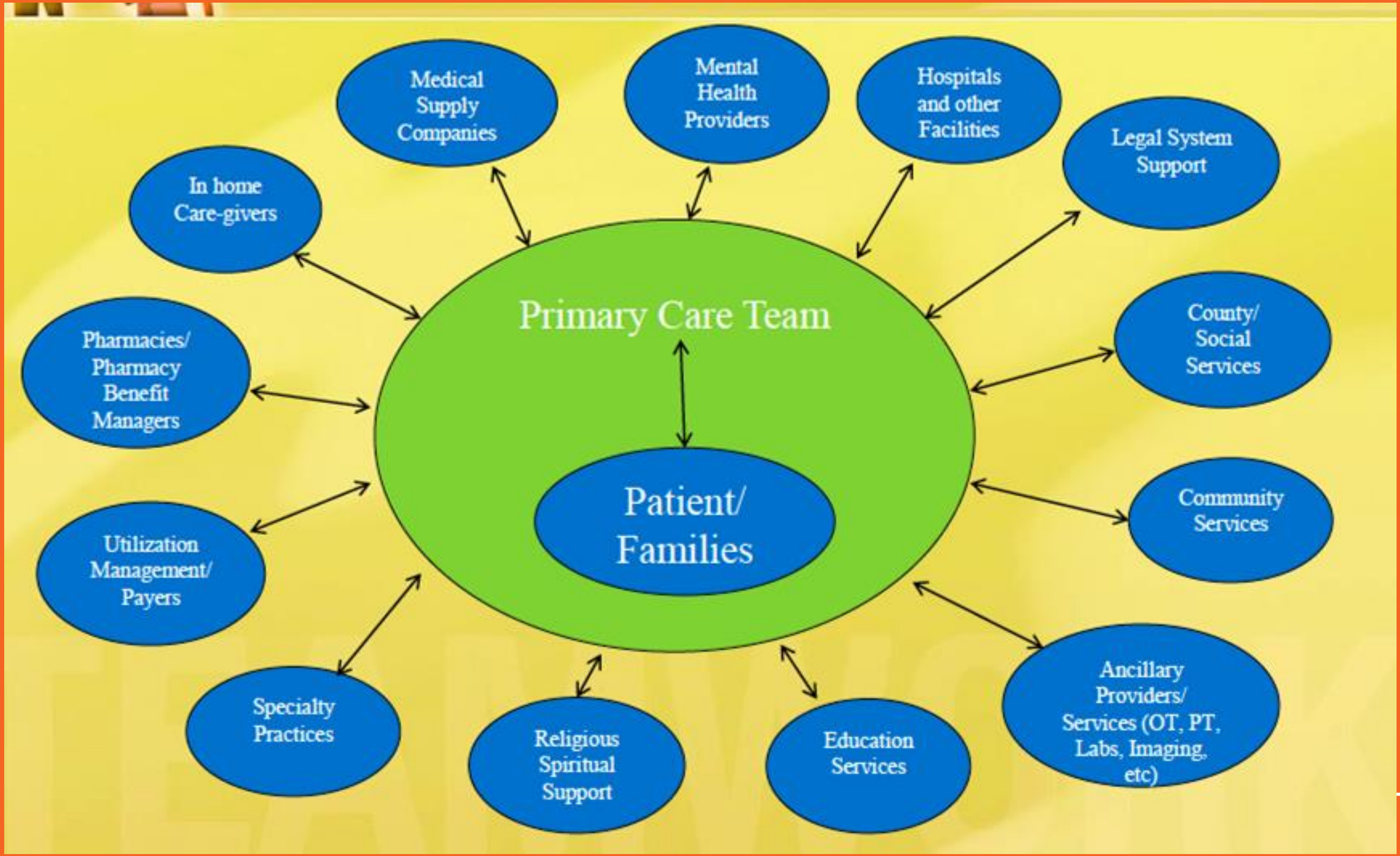
- "The deliberate organization of patient care activities between **two or more participants** involved in a patient's care to facilitate the appropriate delivery of health services." McDonald, 2007

Another perspective…

“Care coordination is a function that helps ensure that the **patient’s needs and preferences** for health services and **information sharing** across people, functions, and sites are met over time. Coordination **maximizes the value of services** delivered to patients by facilitating beneficial, efficient, safe, and **high-quality patient experiences** and **improved healthcare outcomes.”**

National Quality Forum 2006

Another Perspective…



Primary Care Team

Patient/ Families

Medical Supply Companies

Mental Health Providers

Hospitals and other Facilities

Legal System Support

County/ Social Services

Community Services

Ancillary Providers/ Services (OT, PT, Labs, Imaging, etc)

Education Services

Religious Spiritual Support

Specialty Practices

Utilization Management/ Payers

Pharmacies/ Pharmacy Benefit Managers

In home Care-givers

5 Key Elements of Care Coordination

Numerous participants are typically involved in care coordination

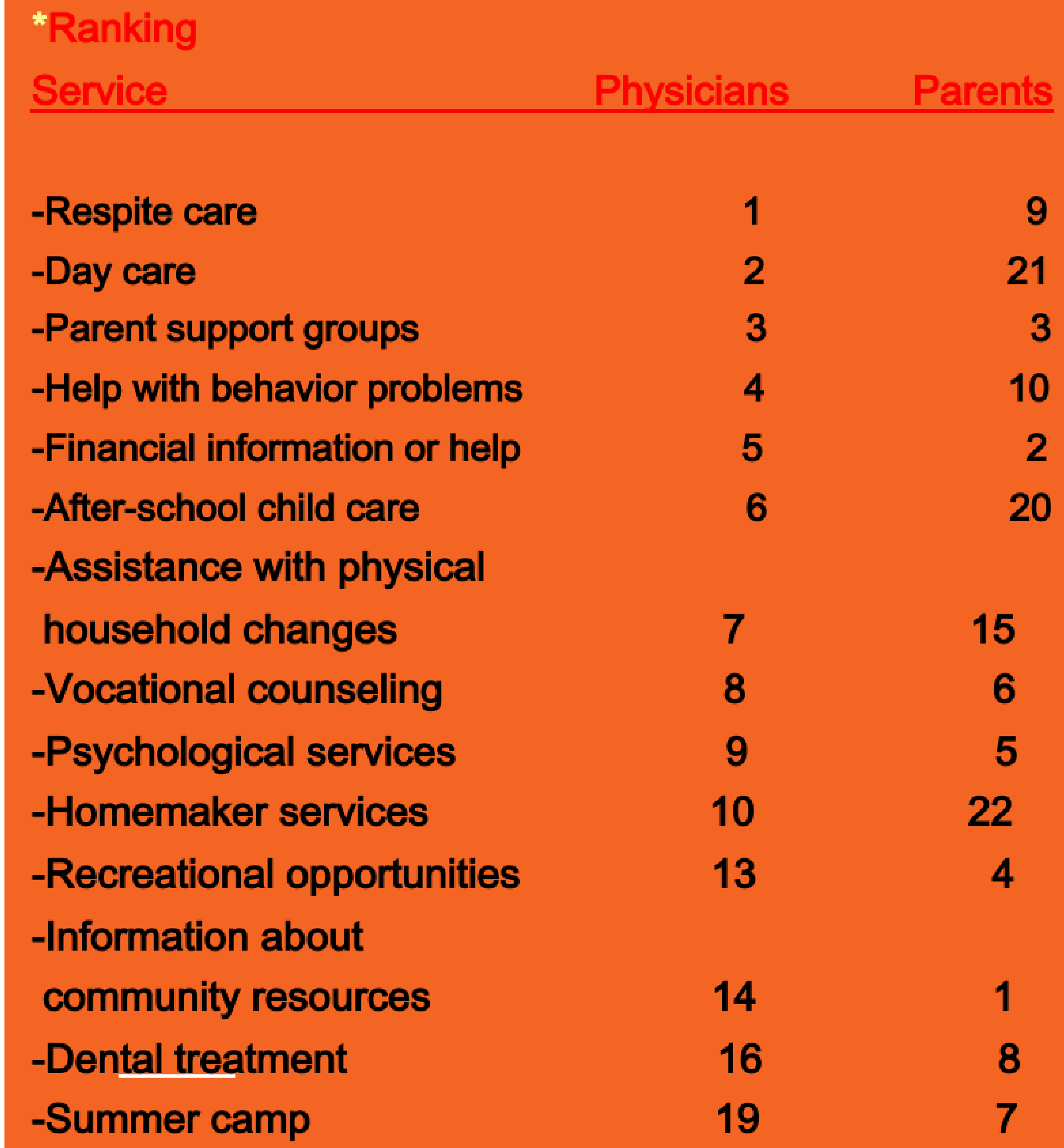
Coordination is necessary when participants are dependent upon each other to carry out disparate activities in a patient’s care

In order to carry out these activities in a coordinated way, each participant needs adequate knowledge about their own and others’ roles, and available resources

In order to manage all required patient care activities, participants rely on exchange of information and

Integration of care activities has the goal of facilitating appropriate delivery of health care services

Which Services Do Parents *Really* Want Help Coordinating?



*Ranking

Service	Physicians	Parents
-Respite care	1	9
-Day care	2	21
-Parent support groups	3	3
-Help with behavior problems	4	10
-Financial information or help	5	2
-After-school child care	6	20
-Assistance with physical household changes	7	15
-Vocational counseling	8	6
-Psychological services	9	5
-Homemaker services	10	22
-Recreational opportunities	13	4
-Information about community resources	14	1
-Dental treatment	16	8
-Summer camp	19	7

Examples of Programs Utilizing Case Management/Care Coordination in North Dakota

Who is Eligible?

BCBS ND
Case Management

Patients with serious illness…

Case management referrals can be initiated by:

- The member/patient
- The member's authorized representative
- A health care provider

Assessments

BCBS ND
Case Management

Case management often includes voluntary assessments including:

- Comprehensive health screening
- Screening for depression and anxiety
- Assessment of Member’s Health Engagement
- Medication reconciliation

Interventions
BCBS ND Case Management

- Goal setting with members to achieve optimal health outcomes
- Motivational interviewing to assess barriers to change
- Assessment of member engagement into their health
- Providing education regarding health risks and needs assessment
- Collaboration/referral to Patient Centered Medical Home/primary care provider
- Transition of care planning for complex cases understand their benefits
- Coordination of local, regional and nationwide health care services
- Ongoing case management for especially complex and chronic cases
- Referrals to disease management professionals for rare and complex disease management
- Assisting members in making informed health care decisions
- Connecting the members to the right resources within BCBSND to help them understand their benefits

Funding Source: Blue Cross Blue Shield Insured

Who is Eligible?

ND Medicaid Targeted Case Management

https://www.hhs.nd.gov/sites/www/files/documents/targeted-case-management-smi-sed.pdf

- Serious Mental Illness/Emotional Disturbance
  - Mental/substance use/developmental disability with WHODAS>=25 in need of emergency or long term services
- Child Welfare
  - Victim Abuse/Neglect
  - Placed by an agency from another state
  - Receiving services dictated by a court order or voluntarily
- High Risk Pregnant Women

Assessments

ND Medicaid Targeted Case Management

Comprehensive assessment and periodic reassessment of individual needs to determine the need for medical, educational, social or other services. These assessment activities include

1) taking client history;
2) identifying the individual's needs and completing related documentation; and
3) gathering information from other sources such as family members, medical providers, social workers, and educators (if necessary), to form a complete assessment of the eligible individual.

Interventions
ND Medicaid Targeted Case Management

- Development (and periodic revision) of a specific care plan

- Referral and related activities
- Monitoring and follow-up activities
- Review Requirements - for care plan and (re)assessment

- Collateral Contacts

Funding Source: Medicaid State and Federal $$

Who is Eligible?

ND Medicaid 1915i State Plan Amendment

https://www.hhs.nd.gov/1915i

North Dakota's 1915(i) will serve individuals with behavioral health conditions meeting the following eligibility criteria:

1. The individual is age 0+; and
2. The individual is currently enrolled in ND Medicaid or Medicaid Expansion; and
3. The individual's household income is at or below 150% of the Federal Poverty Level https://www.hhs.nd.gov/sites/www/files/documents/1915i/Federal%20Poverty%20Level%20Table.pdf; and
4. The individual's WHODAS score is 25 or above; and
5. The individual resides in and will receive services in a setting meeting the federal home and community-based setting requirements, and
6. The individual has one or more of the qualifying Diagnoses. Click here for list of diagnoses.

Assessments

ND Medicaid 1915i State Plan Amendment

Dependent upon whom is providing the care coordination/services https://www.hhs.nd.gov/sites/www/files/documents/1915i/Providers%20by%20Service%20and%20Region.pdf

Interventions

ND Medicaid 1915i State Plan Amendment

- Care Coordination (0+)
- Training and Supports for unpaid caregivers (0+)
- Community Transitional services (0+)
- Benefits planning (0+)
- Non-medical transportation (0+)
- Respite (0-21)
- Prevocational training (17.5)
- Supported education (5+)
- Supported employment (14+)
- Housing support services (17.5)
- Family peer support (0-18)
- Peer support (17.5)

Funding Source: Medicaid State and Federal $$

Who is Eligible?

Family Voices of North Dakota

http://fvnd.org/

- Families with children (up to age 21y) who are struggling to navigate care.

Assessments

Family Voices of North Dakota

- Informal assessments that are based on what families are expressing as needs

Interventions
Family Voices of North Dakota

- Connect families to community supports- including housing, local food pantries, housing assistance…
- Assist families with health care, educational, human services systems navigation
- Provide emotional support
- Follow up to make sure services/supports in place

Funding Source: Federal/State Grants Private Donations

Services that Care Coordinators Help Coordinate

BCBS FVND NDMA TCM 1915i

-Respite care x
PS x

-Day care
PS x

-Parent support groups x PS
x

-Help with behavior problems x PS
x

-Financial information or help PS
x

-After-school child care PS
x

-Assistance with physical PS
x

PS- Peer Support Specialist

Services that Care Coordinators Help Coordinate

Service BCBS NDMA TCM
1915i FVND

-Vocational counseling x x
x

-Psychological services x x x

-Homemaker services PS
x

-Recreational opportunities PS x

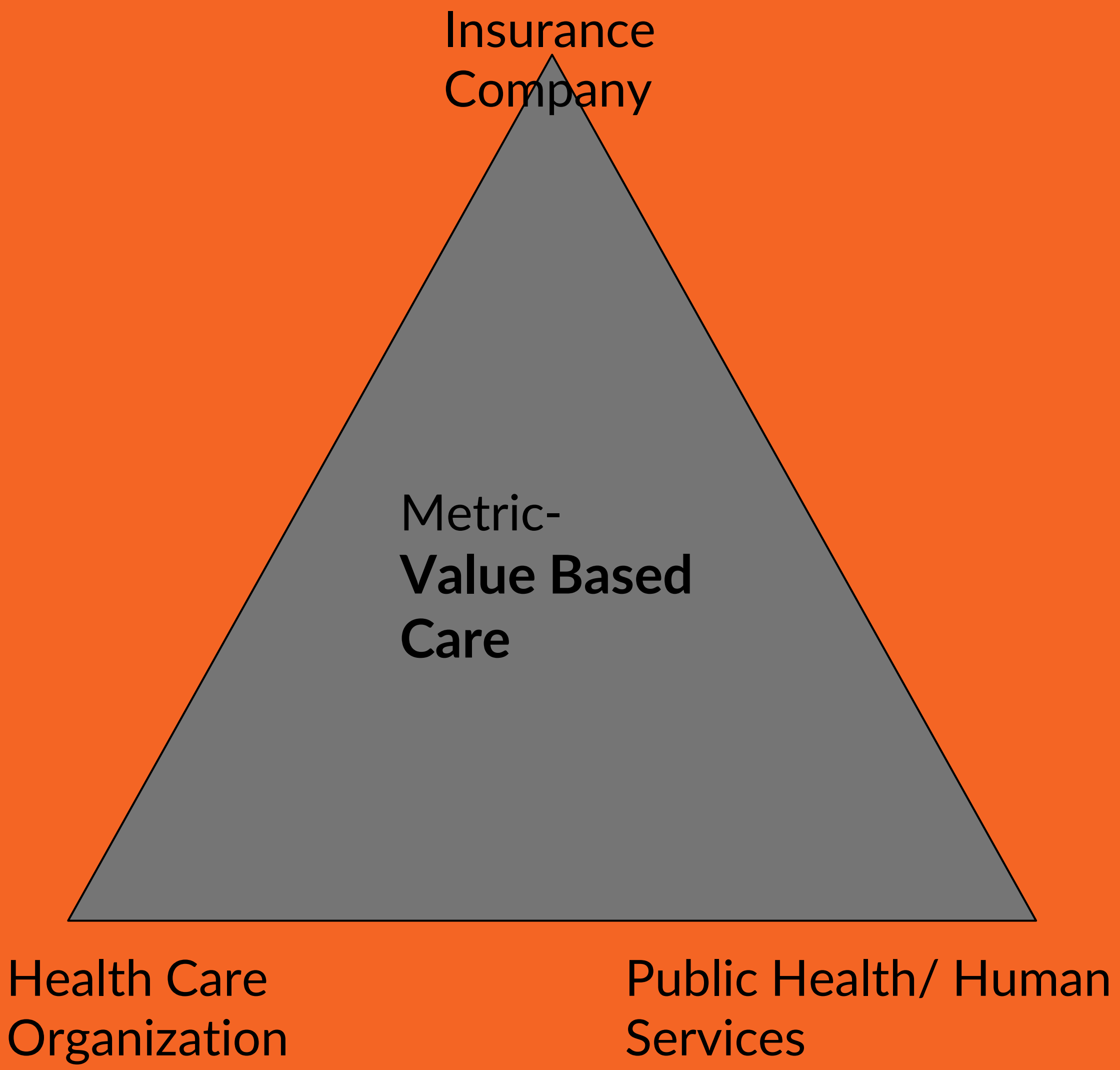
-Information about

community resources x PS
x

-Dental treatment x
PS x

PS- Peer Support Specialist

How Could We Optimize This??

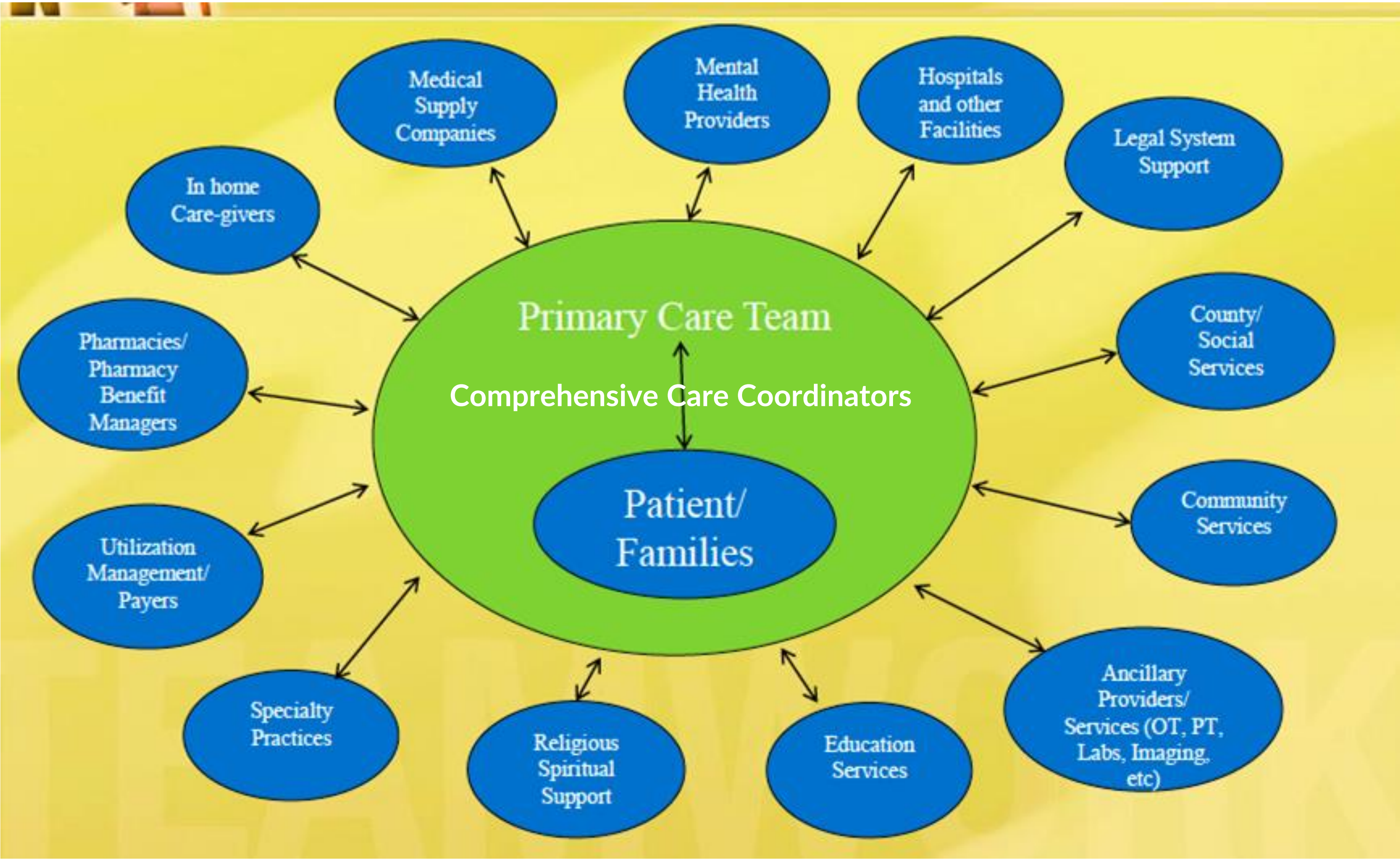


Insurance Company

Metric-
Value Based Care

Health Care Organization

Public Health/ Human Services



Value Based Care= High Quality Care= Equitable Care

Next Steps

Promote Use of Care Coordinators and Care Coordinator Systems (211)

Ask… no, PUSH your Congressional Representatives to enact legislation to transition reimbursement model from fee-for-service to value based care

Approach your patient/child comprehensively to better understand/provide/recommend what they truly need

?? Questions ??

BETH VANNOY, JD

DISCLOSURES

- Nothing to disclose

Living with MCADD

Featuring Alec Hudson Vannoy

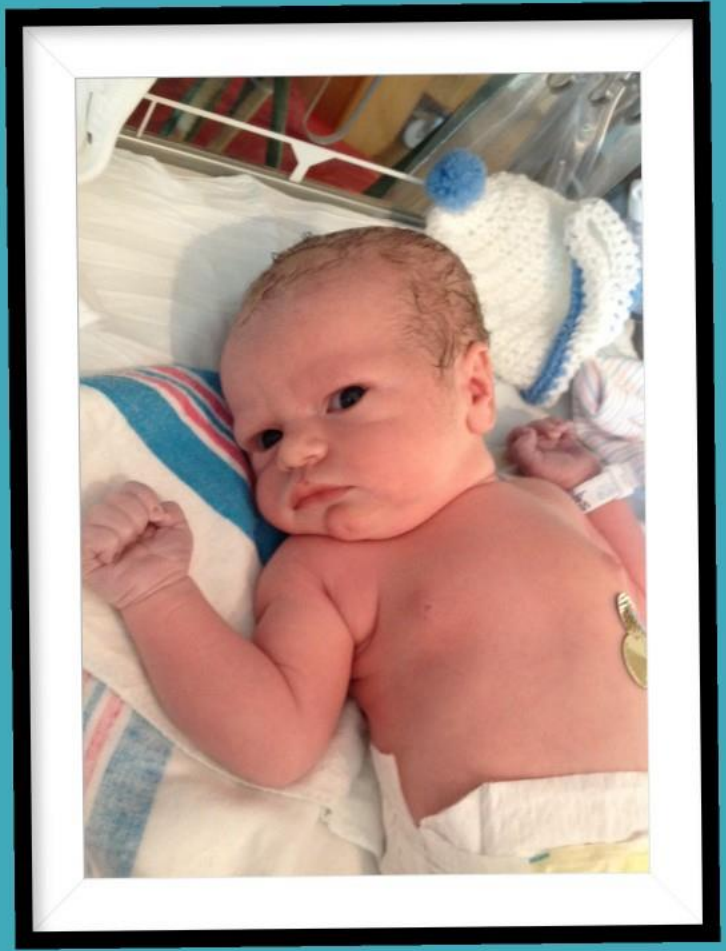
presented by Beth H. Vannoy

Our Rare Disease Diagnosis through Newborn Screening:

Living with MCADD

Alec Hudson Vannoy

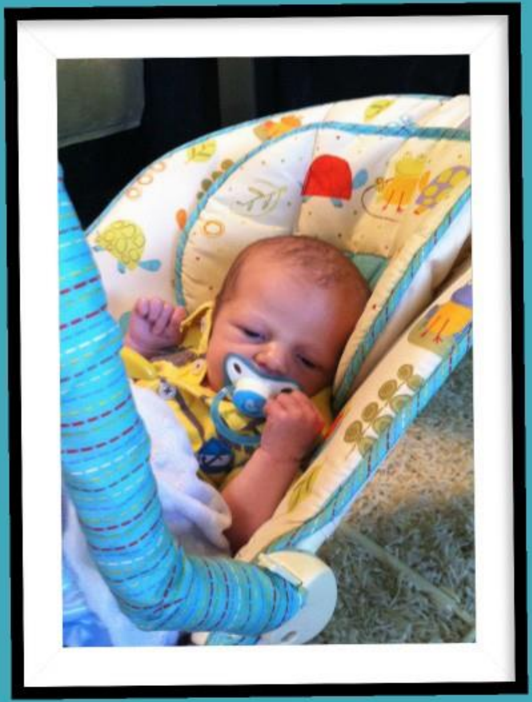
June 20, 2012

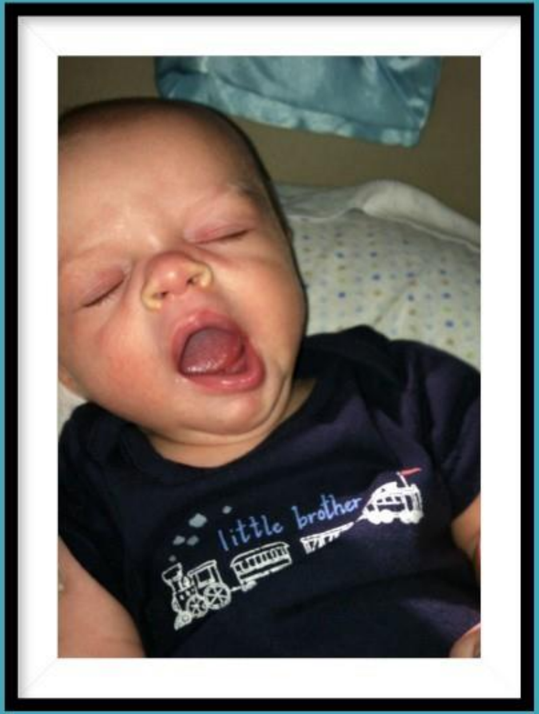


Heel Prick and Headed Home!

Thursday,
June 21, 2012

First few days were filled with naps....





Isn't that just what babies do?

Saturday
June 23, 2012

Newborn Screening Results

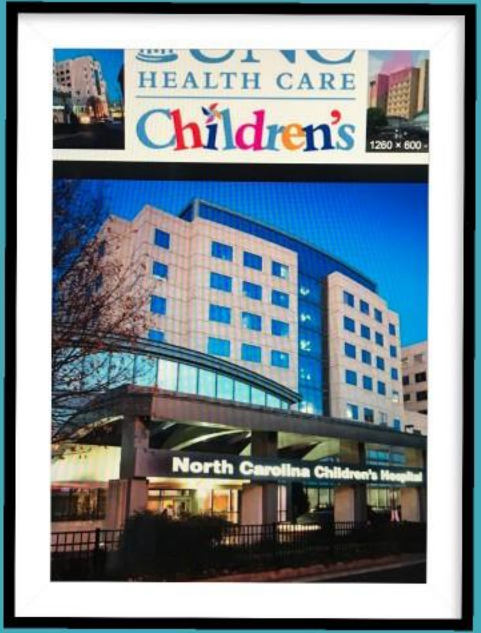
- Results received through Pediatrician on Tuesday, June 26, 2012.
- Appointment with Metabolic Specialist at UNC Children's Hospital on Wednesday, June 27, 2012.

What is MCAD Disorder?

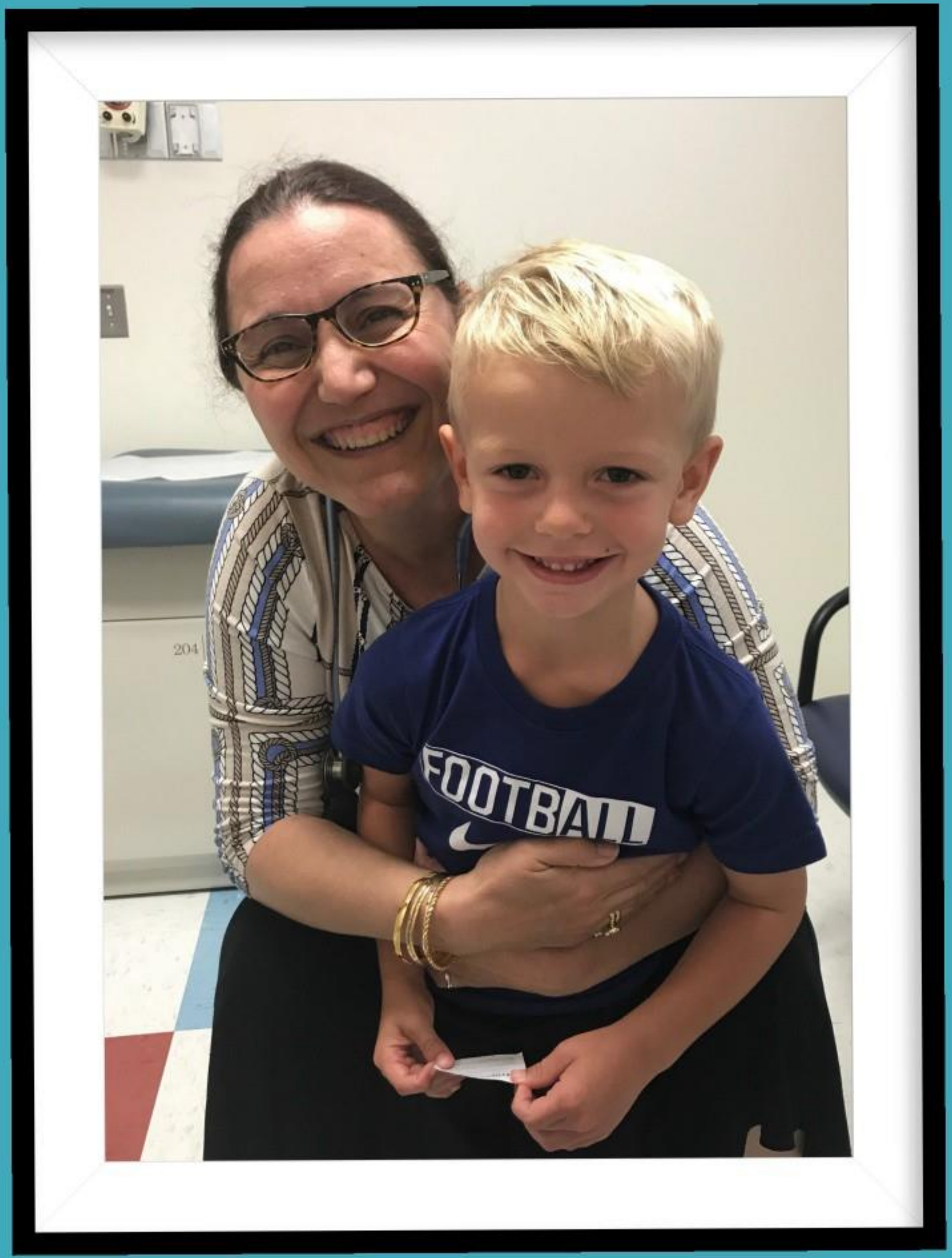
Medium-Chain Acyl-CoA Dehydrogenase Disorder

Our First Appointment

June 27, 2012



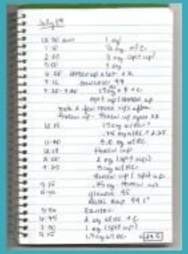
- Met our team - Metabolic Specialist & Nutritionist
- Additional Testing to confirm NBS Results - Urinalysis & Bloodwork
- Test results received about 2.5 weeks later confirmed Alec's MCADD diagnosis.
  - Alec is homozygous for K329E mutation.



Dr. Muge Calikoglu
UNC Children's Hospital

Alec's first 12 months of life

- Fed Alec every 3 hours;



- Heel pricks;

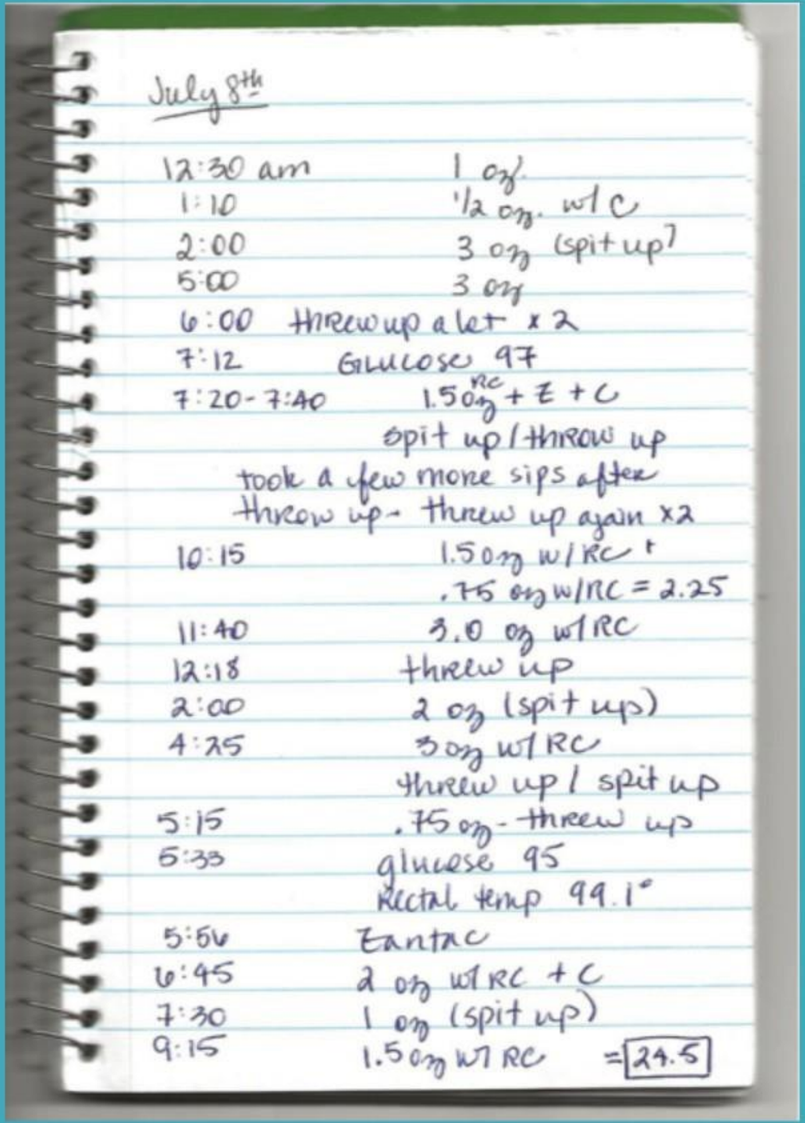
- Entrusting Alec to a babysitter so that I could return to work; and

WHEN I AM AFRAID,
I WILL TRUST IN YOU.
PSALMS 56:3

- Learning to juggle.

Alec's first 12 months of life

Fed Alec every 3 hours:



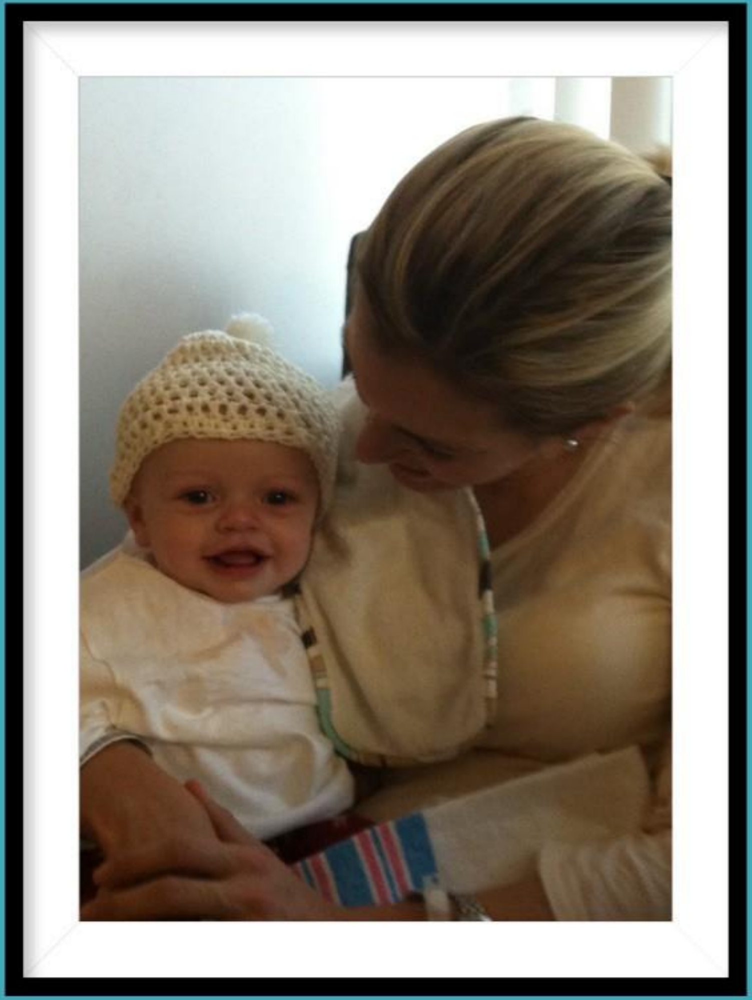
July 8th

12:30 am	1 oz.
1:10	½ oz. w/ C
2:00	3 oz (spit up)
5:00	3 oz
6:00	threw up a lot x 2
7:12	GLUCOSE 97
7:20 - 7:40	1.5 $\overset{RC}{oz}$ + Z + C
	spit up / throw up
	took a few more sips after throw up - threw up again x2
10:15	1.5 oz w/ RC +
	.75 oz w/ RC = 2.25
11:40	3.0 oz w/ RC
12:18	threw up
2:00	2 oz (spit ups)
4:25	3 oz w/ RC
	threw up / spit up
5:15	.75 oz - threw up
5:33	glucose 95
	Rectal temp 99.1°
5:56	Zantac
6:45	2 oz w/ RC + C
7:30	1 oz (spit up)
9:15	1.5 oz w/ RC = $\boxed{24.5}$

Hospital Visits

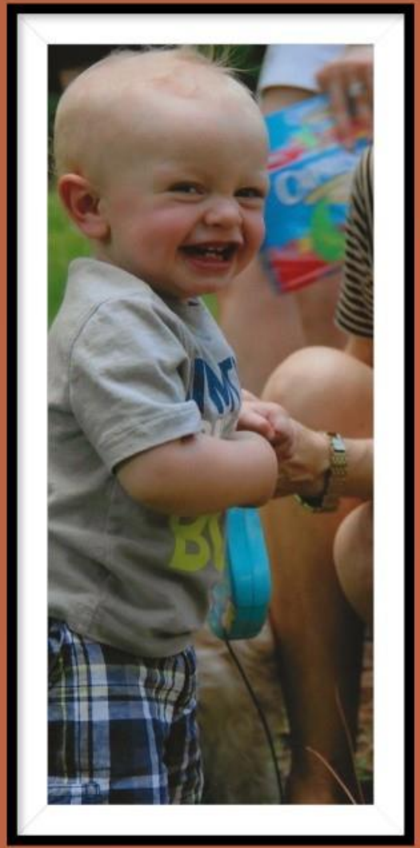
First Overnight Hospital Visit

November 25-26, 2012



Diagnosis: Bronchiolitis

MCADD Complications: Diarrhea and Vomiting



Alec turns One!!

First Year Milestones

- ✓ We can sleep 5 to 6 hours!
- ✓ No more bottles.... We begin eating "real" food!
- ✓ We start preschool!

With Preschool Comes....

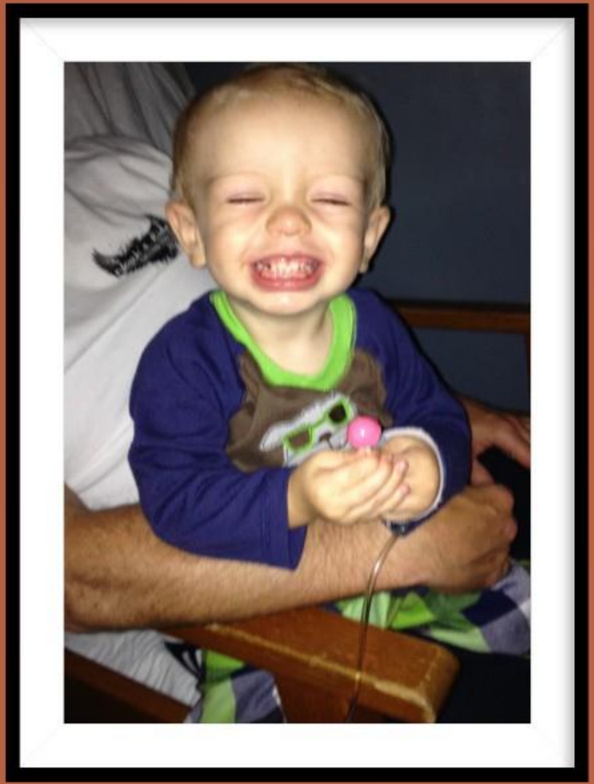
Stomach Bug

Hand, Foot & Mouth

With Preschool Comes....

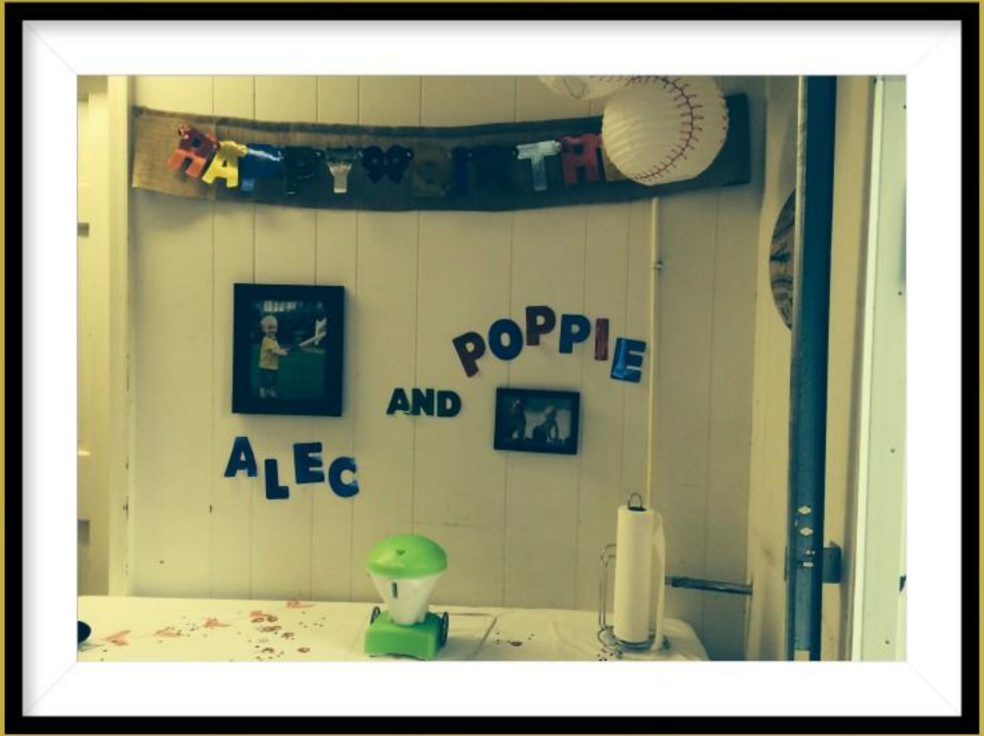
Sickness

Stomach Bug



Hand, Foot & Mouth

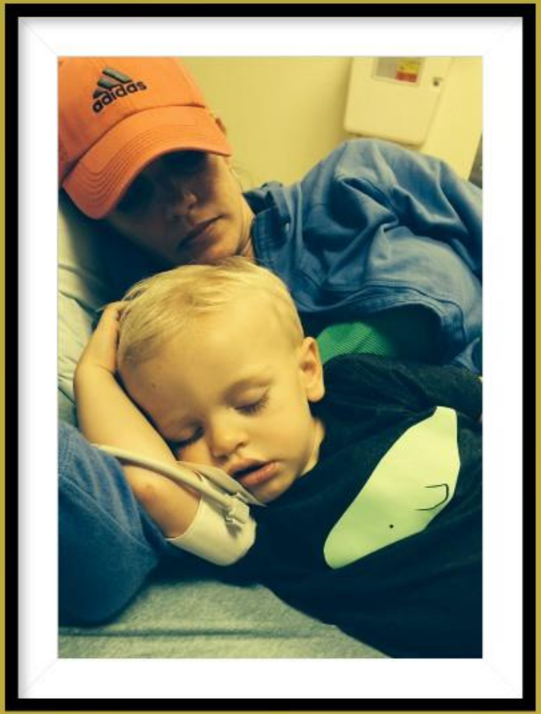
Alec turns 2!

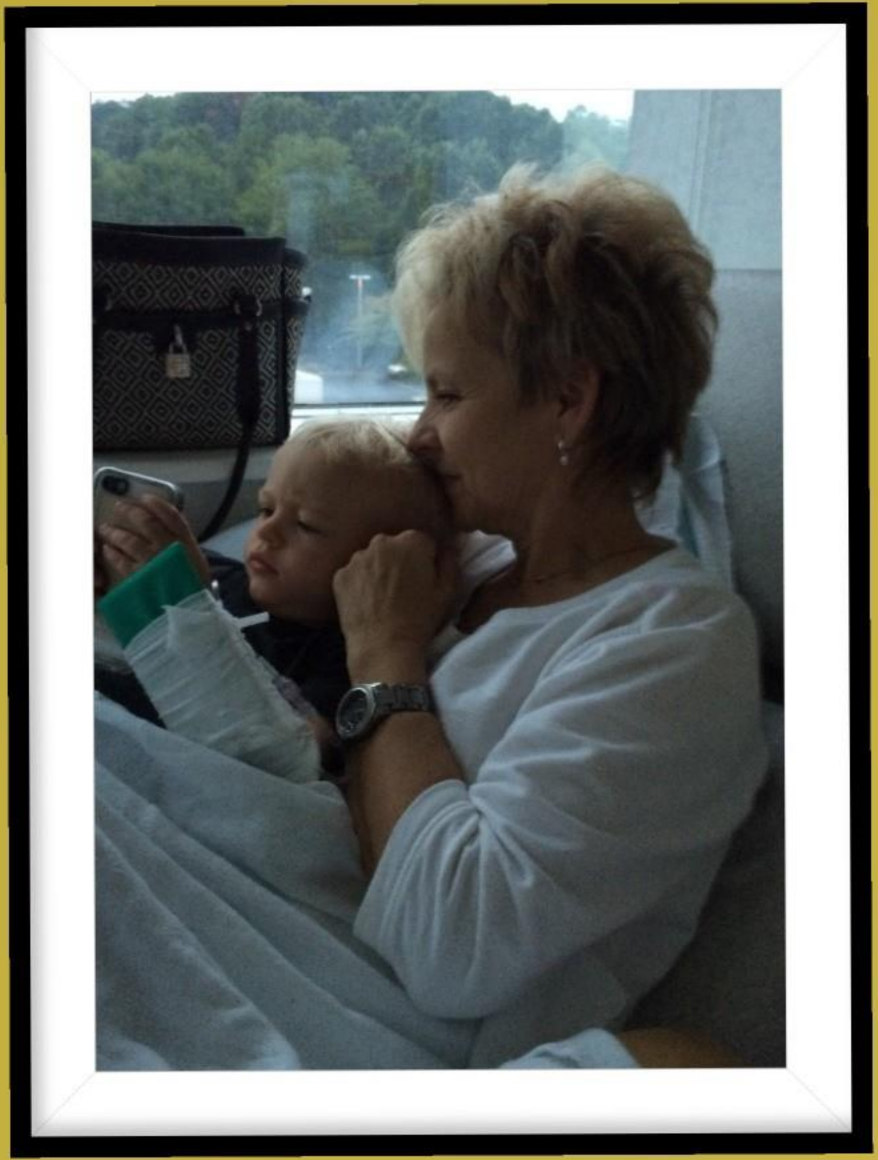


Food Allergies

Reaction to Pine Nuts

August 1, 2014



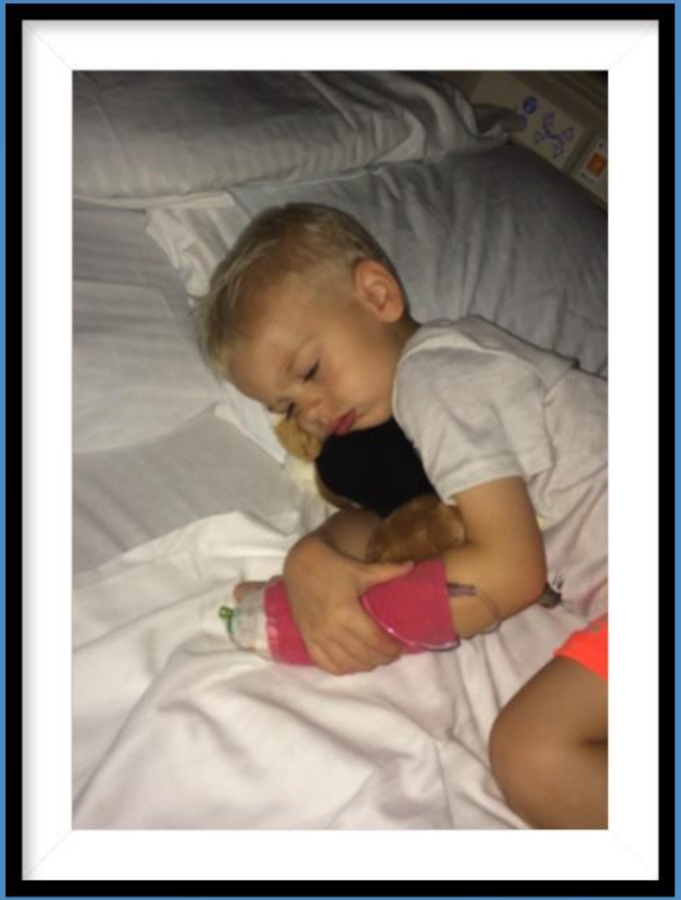


Alec turns 3!

Tonsillectomy and Adenoidectomy

July 13, 2015





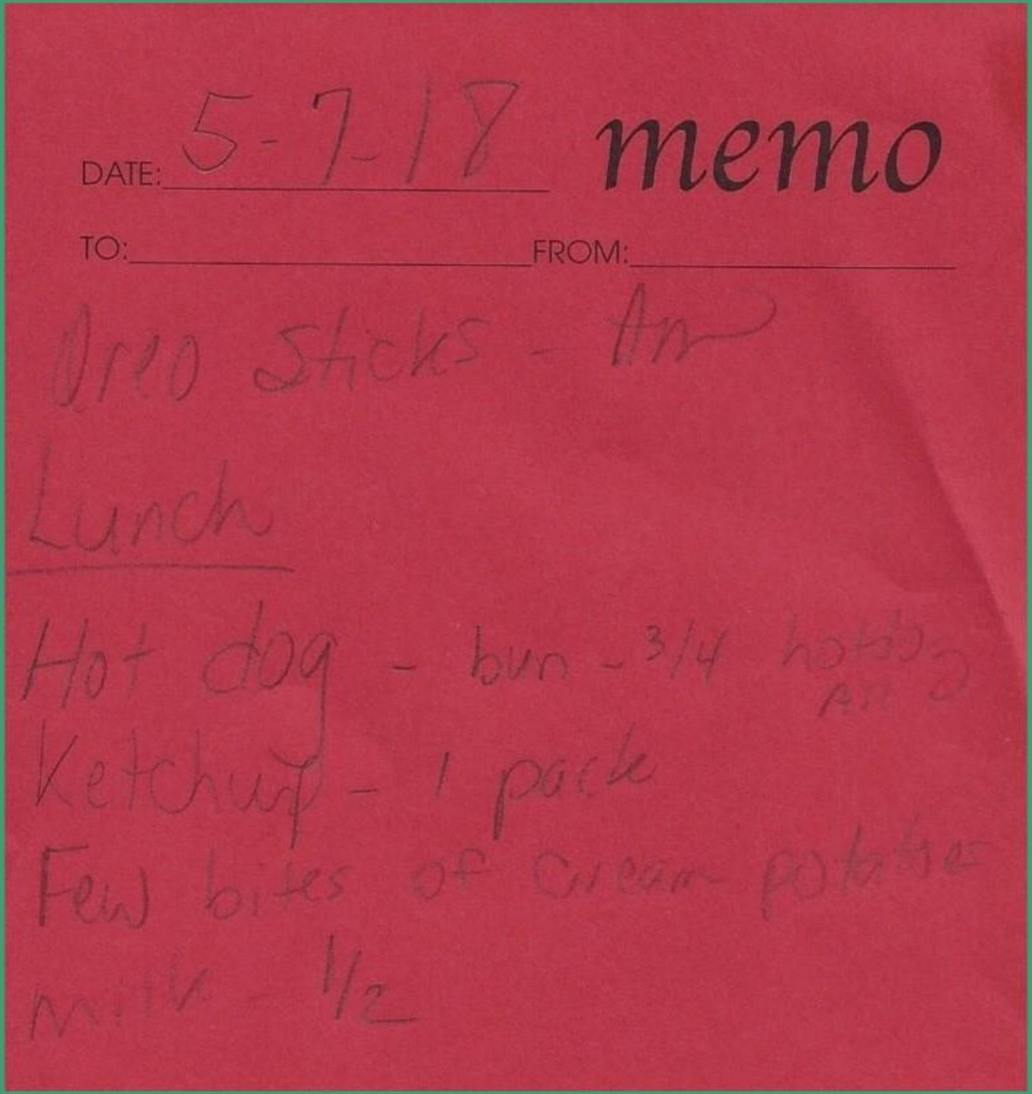
On the road to recovery….

Alec turns 4!

Alec turns 5!

Kindergarten, here we come!

Alec's Lunchroom Report



DATE: 5-7-18 *memo*

TO: FROM:

Oreo Sticks - An

Lunch

Hot dog - bun - 3/4 hotdog An

Ketchup - 1 pack

Few bites of cream potatoes

milk - 1/2

Allergy Testing

Designed by Freepik

"MCADD Normal"

Medicine Daily

SPORTS!!

Family Trips & Vacations

- Travel Times
- Hotel Accomodations
- Meal Times
- Snacks

Field Trips

Zootastic

Raleigh, NC

Article published by the National Institute of Health:

Parental Experiences of Raising a Child With Medium Chain Acyl-CoA Dehydrogenase Deficiency

Published in 2017.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5419063

Charlotte H.

Newborn Screening

minutes matter

MCADD AWARENESS

Emery G.

Summary

Charlotte H.

April 1, 2021 - April 5, 2021

Alabama


CHARLOTTE
ANN HALL
A MOMENT IN OUR ARMS
FOREVER IN OUR HEARTS
APR. 1.
2021
APR. 5.
2021

Emery G.

July 15, 2021 -
July 16, 2021
Maryland

1
2
3
4
5
6
7
8
9
10
11
12


One
month old


Two
months old


Three
months old


Four
months old


Five
months old
E


Six
months old


Seven
months old
love


Eight
months old


Nine
months old


Ten
months old


Eleven
months old

One Years Old.

Our sweet Emery, today you are one years old. How has it been one year already?!

A year ago today you came into this world. We were so in love with you and couldn't wait to bring you home from the hospital and show you off to family and friends. Little did we know your life here on earth would be so short. This past year we've had to learn how to navigate a new way of life; constantly wondering about your milestones and what you would look like, and all the "should-have-beens". It hasn't been an easy journey, but we are doing our best to honor and remember you every chance we get.

We love and miss you so much baby girl and will always be your Mommy and Daddy.

Newborn Screening

Background

Timeliness

Government Regulation

Background

- Every baby born in the United States will be screened unless a parent decides to opt out for religious reasons.
- The blood test is generally performed when a baby is 24 to 48 hours old. This timing is important because certain conditions may go undetected if the blood sample is drawn before 24 hours of age.

Government Regulation

- The Advisory Committee on Heritable Disorders in Newborns and Children
- Recommended Uniform Screening Panel
- Newborn Screening in Your State

Timeliness

HRSA: Federal Advisory Committee Newborn Screening Timeliness Goals

Summary

1. All States test for the same disorders through their NBS Program.

2. All States require NBS results to be returned within 2 days.

3. MCADD is a life threatening condition that requires earily identification and treatment.

4. All babies with MCADD will show symptoms within the first 24 hours of life.

Summary

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False

Beth H. Vannoy

www.MinutesMatter-MCADD.org

Beth@MinutesMatter-MCADD.org

(336) 469-4833

Living with MCADD

Featuring Alec Hudson Vannoy

presented by Beth H. Vannoy

2012

2013

2014

2015

2016

2017

2018-2023

Scan To Take Post Test

https://ndhealth.co1.qualtrics.com/jfe/form/SV_2mi557RbNEpjwJE

Scan To Take Evaluation

https://ndhealth.co1.qualtrics.com/jfe/form/SV_br9VreOovRCkxg2

CONFERENCE EVALUATION

- QR Code Placeholder