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DEPARTMENT OF HEALTH

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South Dakota Newborn Screening Advisory Committee Meeting

April 12, 2023

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Welcome & Introductions

Beth Dokken, Director
Division of Family & Community Health

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Review and Adoption of Newborn Screening Purpose and Mission Statements

Bernadette Boes, RN
Newborn Screening Program Coordinator

Purpose of the South Dakota Newborn Screening Advisory Committee and Role of the Committee Member

- The South Dakota Newborn Screening Advisory Committee provides expertise, guidance and recommendations to the South Dakota DOH Newborn Screening Program regarding programmatic decisions.
- To assist the Department of Health in reviewing policies and procedures of the Newborn Screening Program
- The group convenes on an annual basis to receive updates on the status of the newborn screen program and to discuss the addition of new disorders to the South Dakota panel of disorders.
- The advisory committee consists of newborn screening stakeholders and partners, including pediatric specialists, Medicaid, nurse leaders, lab personal and family interested in promoting newborn screening.

Purpose Statement

The purpose of newborn screening is to detect potentially fatal or disabling conditions in newborns as early as possible, before the infant displays any signs or symptoms of a disease or condition.

Mission Statement

The Newborn Screening Program seeks to decrease the morbidity and mortality of infants born in South Dakota through timely testing, diagnosis, and high quality newborn screening follow-up.

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Newborn Screening 2022 Findings and Program Updates

Bernadette Boes, RN
Newborn Screening Coordinator

Newborn Screening 2022
By the numbers

Bloodspot – Diagnosed / Carriers

Congenital Hypothyroidism / CH	6
Congenital adrenal hyperplasia / CAH	2
Cystic Fibrous / CF	3
carriers - 11	
Severe Combined Immunodeficiency / SCID	1
other immunodeficiency identified 1	
Medium-chain acyl-CoA dehydrogenase deficiency / MCADD	3
carriers – 1	
Very-long-chain acyl-CoA dehydrogenase deficiency / VLCAD	0
carriers - 6	
POMPE	0
carriers 2	
hemoglobinopathies / Sickle Cell / HGB/SS	1
carriers - 76	

Newborn Screening Formal Nomination Process, Form, and Development of Nomination Subcommittee

Bernadette Boes, RN
Newborn Screening Coordinator

Proposed Formal Condition Nomination and Review

How is a Condition Reviewed by the Newborn Screening Advisory Committee?

Establish an improved process by which conditions are reviewed for addition to South Dakota's newborn screening panel.

Establish newborn screening nomination committee

Nomination Review Process

1. Nomination Presented to the Newly established Newborn Screening Nomination Committee

2. Committee meets to discuss if Condition qualifies for Newborn Screening Nomination Status by reviewing the following nomination criteria:

- **Criterion 1:** There is support from an appropriate screening facility and the nominated condition is considered feasible to add (i.e., newborn screening lab for blood spot screens and hospitals for point of care screens).

- **Criterion 2:** Clinical specialist(s) are available, ready to accept referrals, and willing to manage patients found through screening.

- **Criterion 3:** The nominated condition can be found between 24 and 48 hours of life through screening but cannot be identified clinically in that time frame.

- **Criterion 4:** There is a screening test available now or expected within 12 months that can be done quickly and is successful in finding affected newborns.

- **Criterion 5:** There is safe and effective treatment and/or intervention available which provides significant improvement in the quality of life when administered early.

- **Criterion 6:** There is an infantile onset form of this condition.

Nomination Process

Did the Nomination Committee agree or disagree if the condition met criteria?

- If yes – Prepare Presentation for Newborn Screening Advisory Committee and provide potential process for implementation and full analysis of disorder. The Newborn Screening Advisory Committee then votes on the disorder.
    - If the committee votes yes then DOH would move though their processes to include the condition.
    - If the committee votes no then the nominator would be notified of the outcome of the vote.
- If No- the nominator would be notified of the decision made by the nominating committee.

Advisory Nomination Sub-Committee Membership

Ethics advisor
Disorder specialist
Genetic consultant
Pediatrician
Medicaid representative
Genetic counselor
Short term follow up staff
Social worker
CYSHCN nurse practitioner

How to Nominate A Condition

- If you would like to nominate a condition to be considered for addition to the newborn screening panel, please contact the NSAC nomination committee at bernadette.boes@state.sd.us.
- The Newborn Screening coordinator will follow-up with you to discuss the condition you wish to nominate and describe the nomination and advisory committee process.
- Following the discussion with the Newborn Screening coordinator, should you want to formally nominate a condition, you must complete the *Condition Nomination Form* and the *Conflict of Interest Disclosure Form - Newborn Screening Condition Nomination.*

Next Steps

- A disorder has already been submitted for review by the Nominating Committee.
- Over the summer we will work to establish the nominating committee with a goal of meeting in September.
- If you are interested in being part of the nominating committee please reach out to Bernadette.

Discussion on Membership Expansion

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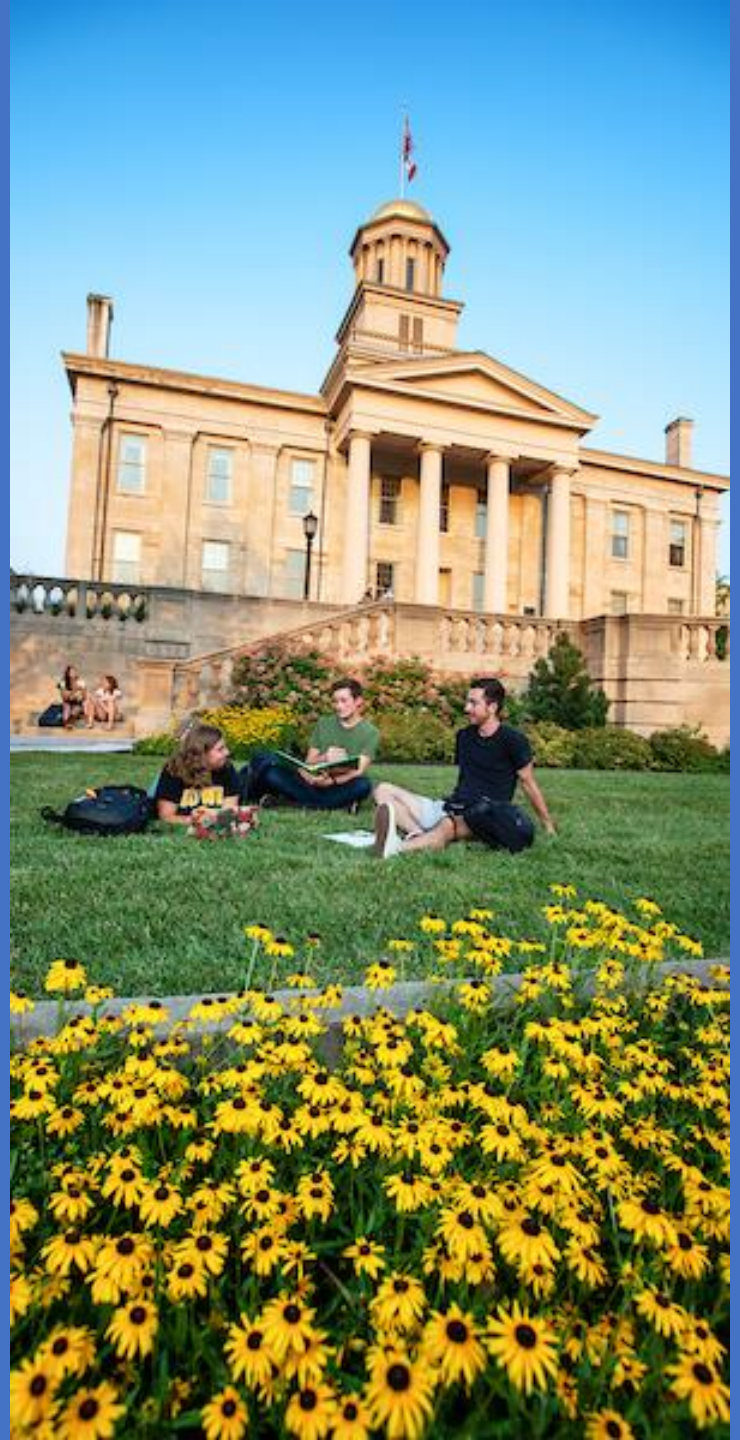
NewSTEPS – Newborn Screening Technical Assistance and Evaluation Program

Program with APHL – Association of Public Health Laboratories (HRSA Funded)

University of Iowa State Hygienic Lab

Pompe Update

9/1/2022 – 3/31/2023



Lysosomal Storage Disorders

- Group of diseases caused by a buildup of macromolecules in the lysosomes of a cell*(Platt et al., 2018)*
- Generally caused by enzyme dysfunction*(Platt et al., 2018)*
- Typically treated with enzyme replacement therapy
- 3 LSDs currently on the RUSP
- Pompe added 2015
- Testing for Pompe for South Dakota newborns began September 1st, 2022

Pompe Disease

- Caused by deficiency in acid alpha glucosidase enzyme(GAA)*(Taverna, S. et al., 2020)*
- Leads to a buildup of glycosaminoglycans(GAGS) in the cell *(Taverna, S. et al., 2020)*
- Test for GAA enzyme activity using Flow Injection Tandem Mass Spectrometry

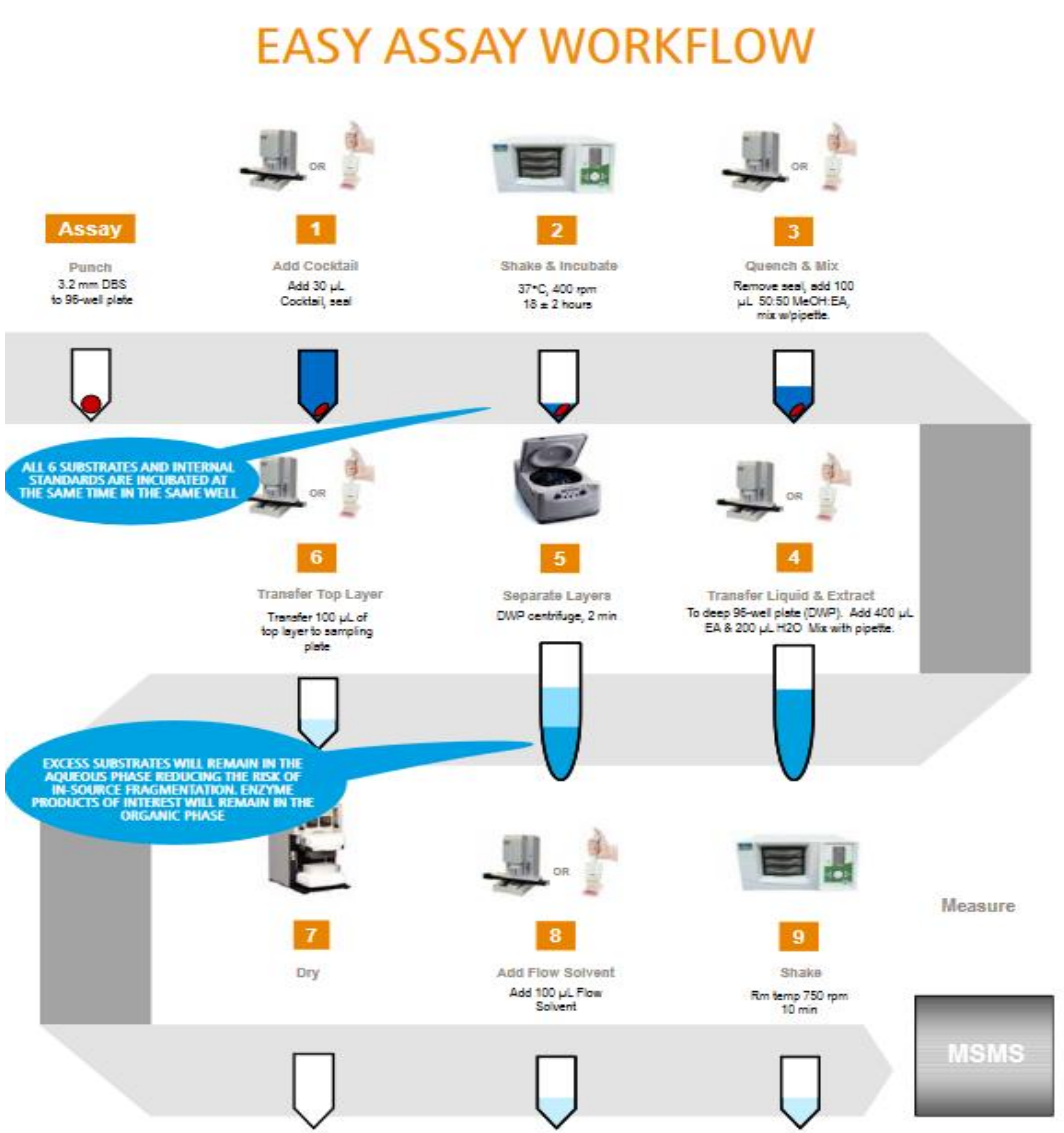
Pompe Disease(cont.)

- 2nd Tier testing performed by Mayo Clinic Labs – uses the ratio of (creatine/creatinine)/GAA
- It is currently in the process of being added to the Iowa panel – Estimated 5/1/2023

Methodology

- Flow injection Tandem Mass Spectrometry
- 3-hour incubation
- Approx. 3.5-hour total processing time
- 3-hour plate processing on instrument

Procedure



From Perkin Elmer NeoLSD Kit Brocure

Pompe testing

- Testing initiated on 9/1/2022
- 7333 total specimens tested as of 4/5/2023
- 2 Presumptive Positive specimens identified
- Second tier analysis categorized both as pseudo deficiency or carrier status.
- No true positives identified so far.

Questions?

References

1.Platt, F, et al. Lysosomal storage diseases. *Nature Review Disease Primers*. 4 (2018)

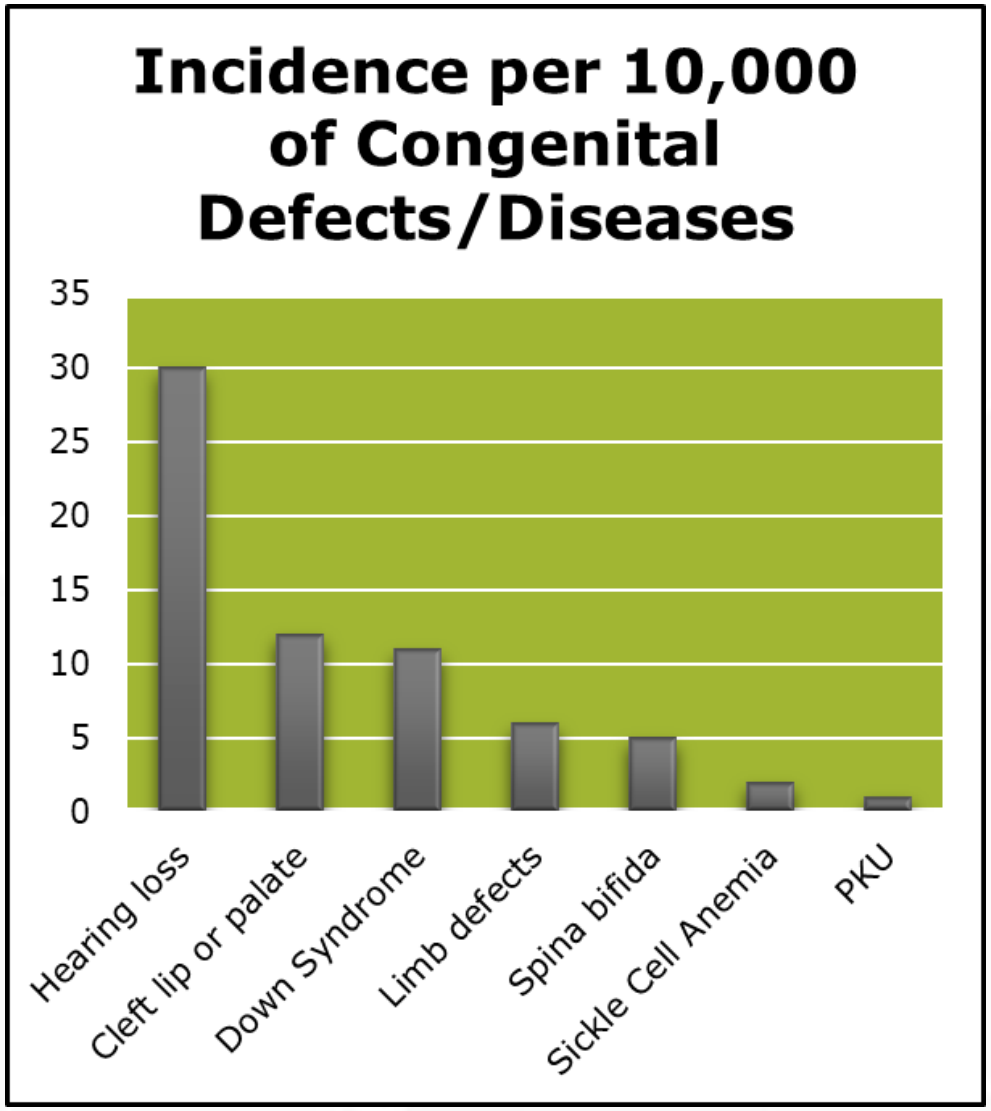
2.Taverna, S. et al. Pompe disease: pathogenesis, molecular genetics and diagnosis. *Aging(Albany, NY)* 2020 August 15; 12(15) 15856-15874

SD EHDI Collaborative

SD EHDI (Early Hearing Detection and Intervention) Collaborative is a partnership between the University of South Dakota and the South Dakota Department of Health that was formed in 2015 to allow the initiation of HRSA grant quality improvement activities within the South Dakota Newborn Hearing Screening Program.

https://doh.sd.gov/family/newborn/Screening/hearing/
https://ehdi136.com
https://mchb.hrsa.gov/programs-impact/early-hearing-detection-intervention-ehdi

Hearing Loss is the Most Common Birth Anomaly



- Hearing loss is the most common birth anomaly
  - About 2 or 3 out of every 1000 children in the United States are born deaf or hard-of-hearing (NIDCD, 2011)
    - ~ 12,000 per year
  - Another 4,000 to 6,000 infants and children (0-3 years) acquire late onset hearing loss
    - That means they would have passed newborn screening

Untreated Hearing Loss

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Children with untreated hearing loss are at risk for:

- Delayed receptive and expressive language
- Negative effects on social, cognitive, and psychosocial development
- Isolation & withdraw
- Learning difficulties
- Repeated classes
- Decreased test scores (even those with minimal hearing loss)
- Distracted or disruptive behaviors

Early Identification Leads to Better Outcomes

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Children who began intervention before 6-months of age typically have better speech, language, and academic outcomes than their same-aged peers who may have started intervention after 6-months of age (Yoshinaga-Itano et al., 1998).

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How do we identify hearing loss early?

What is early?

EHDI: EARLY HEARING DETECTION AND INTERVENTION

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Screening

- No later than 1 month of age

Diagnosis

- No later than 3 months of age

Early Intervention

- No later than 6 months of age

1:3:6

http://www.jcih.org/

EHDI OVERVIEW

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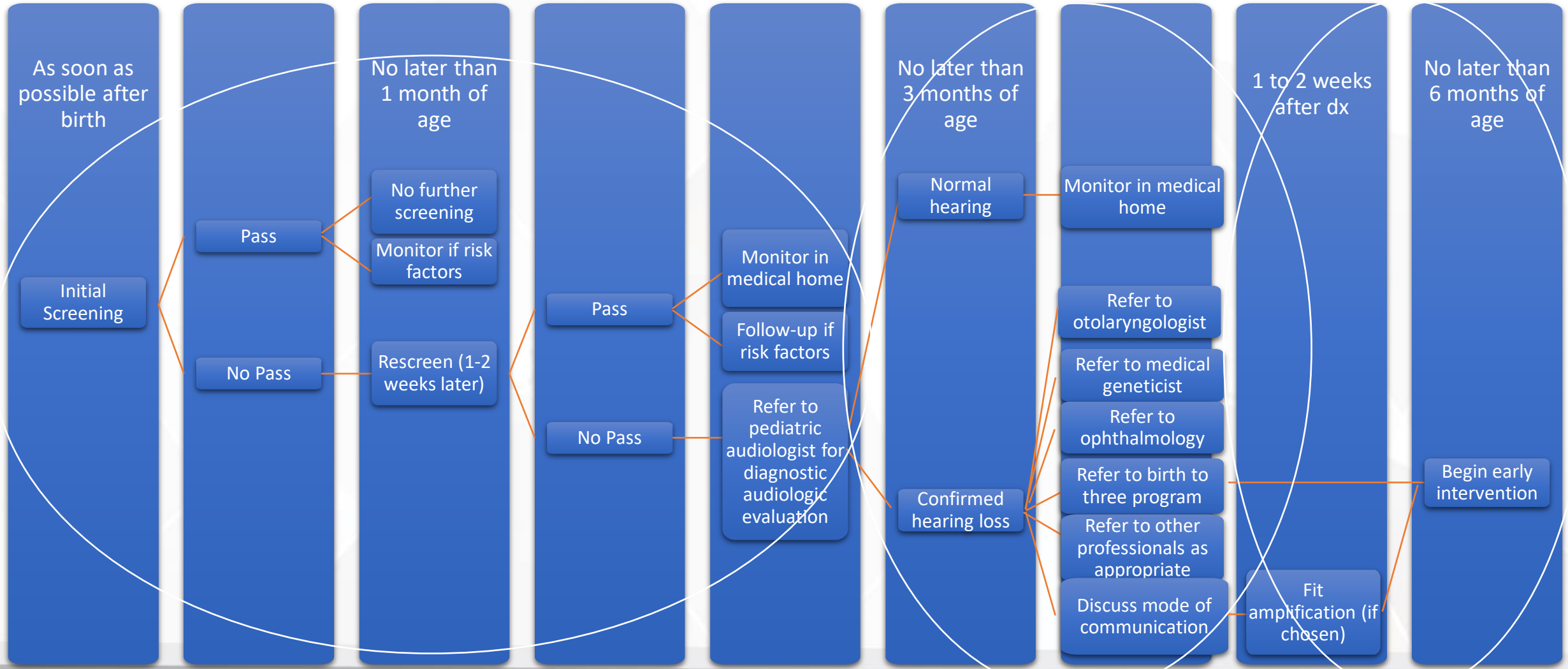

Does UNHS Improve the Age of Identification and Intervention?

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Before UNHS

- Median age of identification was 12-13 months

After UNHS

- Median age of identification was 3 months

EHDI 10 years later (Holte et al., 2012)

Grant Funding

U.S. Department of Health and Human Services Human Resources and Services Administration (HRSA)

Funding Title: Early Hearing Detection and Intervention Program

Funding Opportunity Number: HRSA-20-047

Period of Performance: April 1, 2020, through March 31, 2024

https://www.hrsa.gov/grants/find-funding/HRSA-20-047
https://www.grants.gov/web/grants/view-opportunity.html?oppId=318268

Program Funding Purpose

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- "The purpose of this program is to support comprehensive and coordinated state and territory EHDI systems of care so families with newborns, infants, and young children up to 3 years of age who are deaf or hard-of-hearing (DHH) receive appropriate and timely services that include hearing screening, diagnosis, and early intervention (EI).
- Support EHDI systems of care within states and territories to:
- Lead efforts to engage all EHDI system stakeholders at the state/territory level to improve developmental outcomes of children who are DHH;
- Provide a coordinated infrastructure to:
  - **Ensure that newborns are screened by 1 month of age, diagnosed by 3 months of age, and enrolled in EI by 6 months of age (1-3-6 recommendations); and**
  - **Reduce loss to follow-up/loss to documentation.**
- Identify ways to expand state/territory capacity to support hearing screening in young children up to 3 years of age;
- Strengthen capacity to provide family support and engage families with children who are DHH and adults who are DHH throughout the EHDI system;
- Engage, educate, and train health professionals and service providers in the EHDI system about the 1-3-6 recommendations; the need for hearing screening up to age 3, the benefits of a family-centered medical home and the importance of communicating accurate, comprehensive, up-to-date, evidence-based information to families to facilitate the decision-making process; and
- Facilitate improved coordination of care and services for children who are DHH and their families through the development of mechanisms for formal communication, training, referrals, and/or data sharing between the state/territory EHDI Program and the Individuals with Disabilities Education Act (IDEA) Program for Infants and Toddlers with Disabilities (Part C) Program."

https://www.infanthearing.org/stategrants/grants_hrsa_2019/MCHB%20HRSA-20-047%20state%20grants.pdf

Program Objectives

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"The recipient is funded to achieve, collect, and report on the following:

- By March 2024: Using the state/territory's data from the 2017 CDC (Centers for Disease Control and Prevention) EHDI Hearing Screening and Follow-up Survey (HSFS) as baseline data:
  - Increase by 1 percent from baseline per year, or achieve at least a 95 percent screening rate, whichever is less, the number of infants that completed a newborn hearing screen no later than 1 month of age.
  - Increase by 10 percent from baseline, or achieve a minimum rate of 85 percent, the number of infants that completed a diagnostic audiological evaluation no later than 3 months of age.
  - Increase by 15 percent from baseline, or achieve a minimum rate of 80 percent, the number of infants identified to be DHH that are enrolled in EI services no later than 6 months of age.
- Using data collected from year 1 as baseline data:
  - Increase by 20 percent from baseline the number of families enrolled in family-to-family support services by no later than 6 months of age.
  - Increase by 10 percent the number of families enrolled in DHH adult-to-family support services by no later than 9 months of age.
  - Increase by 10 percent the number of health professionals and service providers trained on key aspects of the EHDI Program."

https://www.infanthearing.org/stategrants/grants_hrsa_2019/MCHB%20HRSA-20-047%20state%20grants.pdf

Program Description

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"Successful recipients will be expected to address the following activities:

- A. Lead efforts to engage and coordinate all stakeholders in the state/territory EHDI system to meet the goals of this program.
- B. Engage, educate, and train health professionals and service providers in the EHDI system.
- C. Strengthen capacity to provide family support and engage families with children who are DHH as well adults who are DHH throughout the EHDI system.
- D. Facilitate improved coordination of care and services for families and children who are DHH through the development of mechanisms for formal communication, training, referrals and/or data sharing between the state/territory EHDI Program and early childhood programs including the IDEA Part C program."

https://www.infanthearing.org/stategrants/grants_hrsa_2019/MCHB%20HRSA-20-047%20state%20grants.pdf

Workplan

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Addresses program goals with a number of activities and tasks, highlights include;

- Maintance, reestablishment, and expansion of the state's teleaudiology infrastructure.
- Coordinated screening efforts for children up to three years of age.
- Assessing access to hearing screening for high LTF populations.
- Develop, maintain, and promote a website.
- Continue, restructure, and convene once annually at minimum, an advisory committee. (A minimum of 25 percent of the committee must be comprised of parents of children who are DHH and adults who are DHH.)
- Conduct outreach and education statewide to health professionals and service providers regarding the 1-3-6 recommendations and the need for hearing screenings up to age three.
- Support family engagement and family support activities via programs and activities that provide direct family-to-family support services and DHH adult-to-family support services.

Newborn Hearing Screen Rate by One Month of Age in South Dakota 2012-2021

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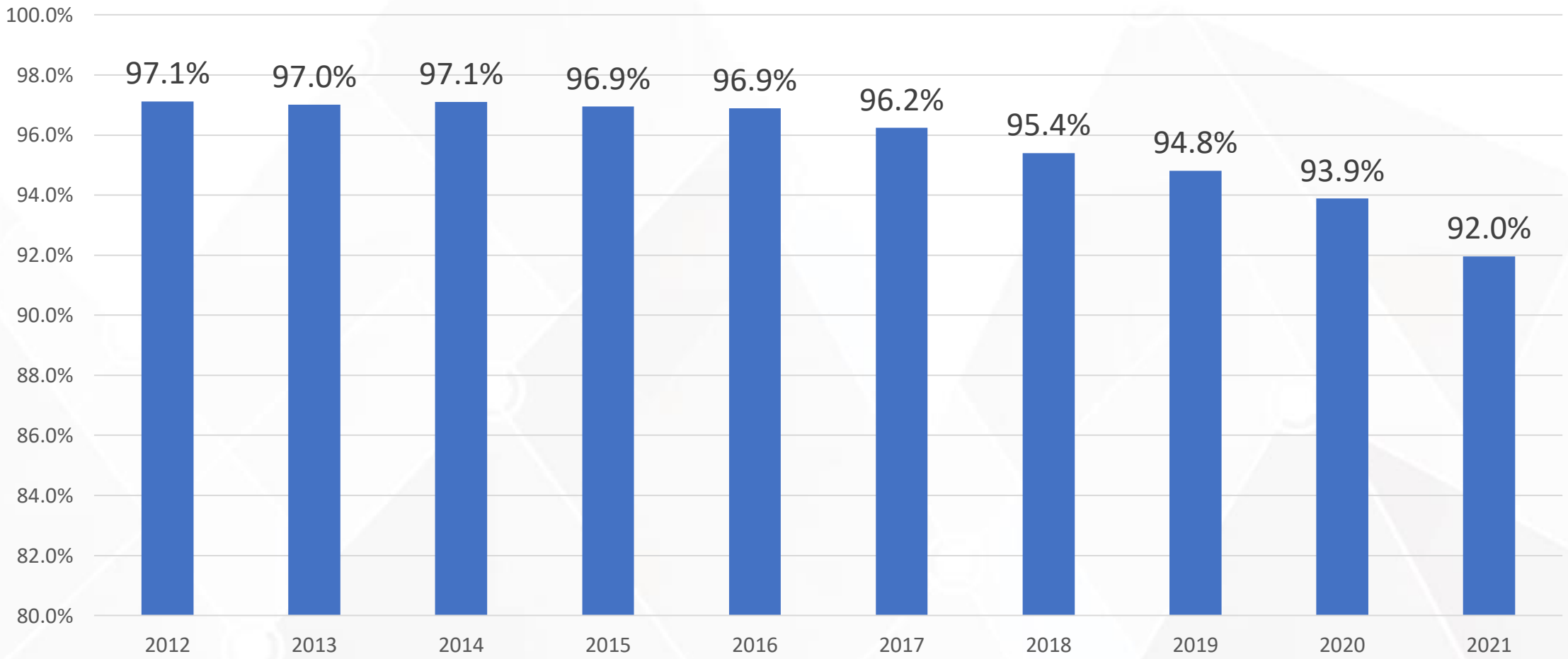

Infant Hearing Screen Rate in South Dakota 2012-2021

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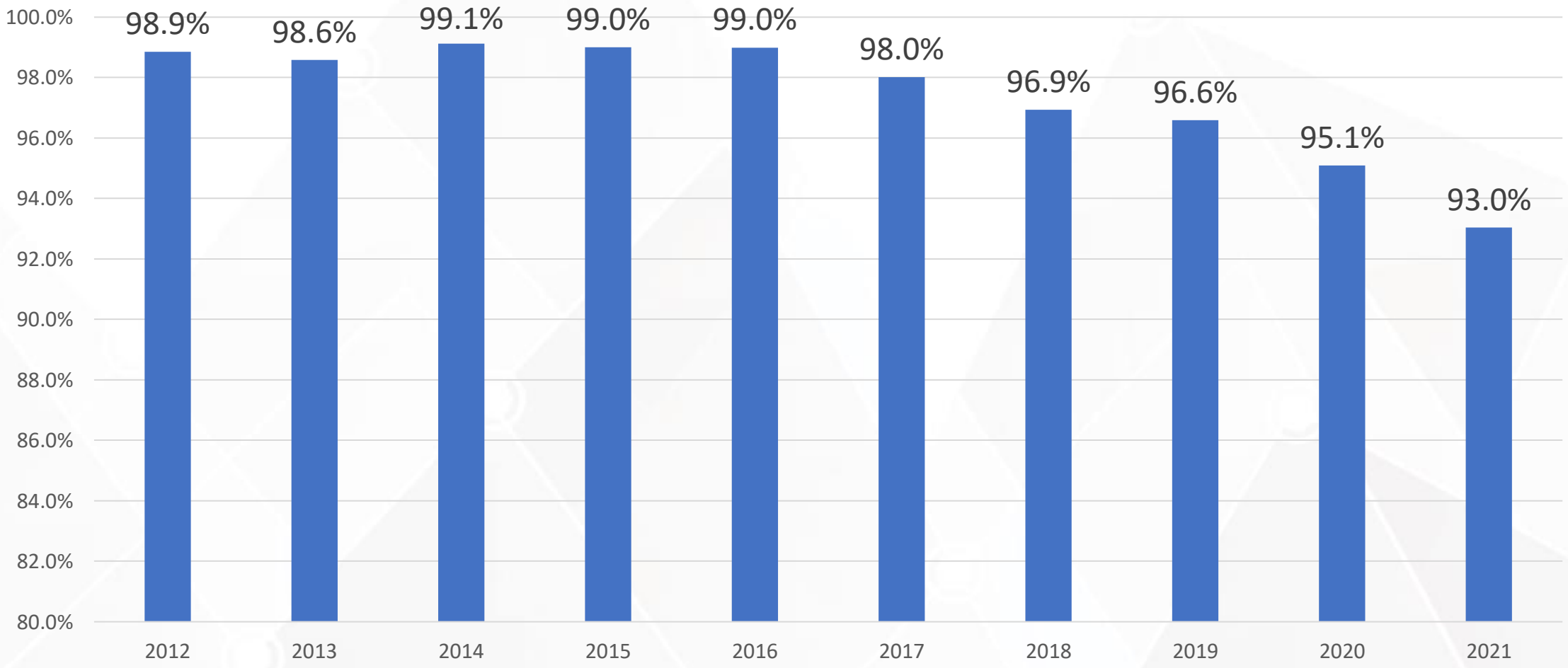

Lost to Follow up Rate for Infants Needing Hearing Diagnostic Testing in South Dakota 2012-2021

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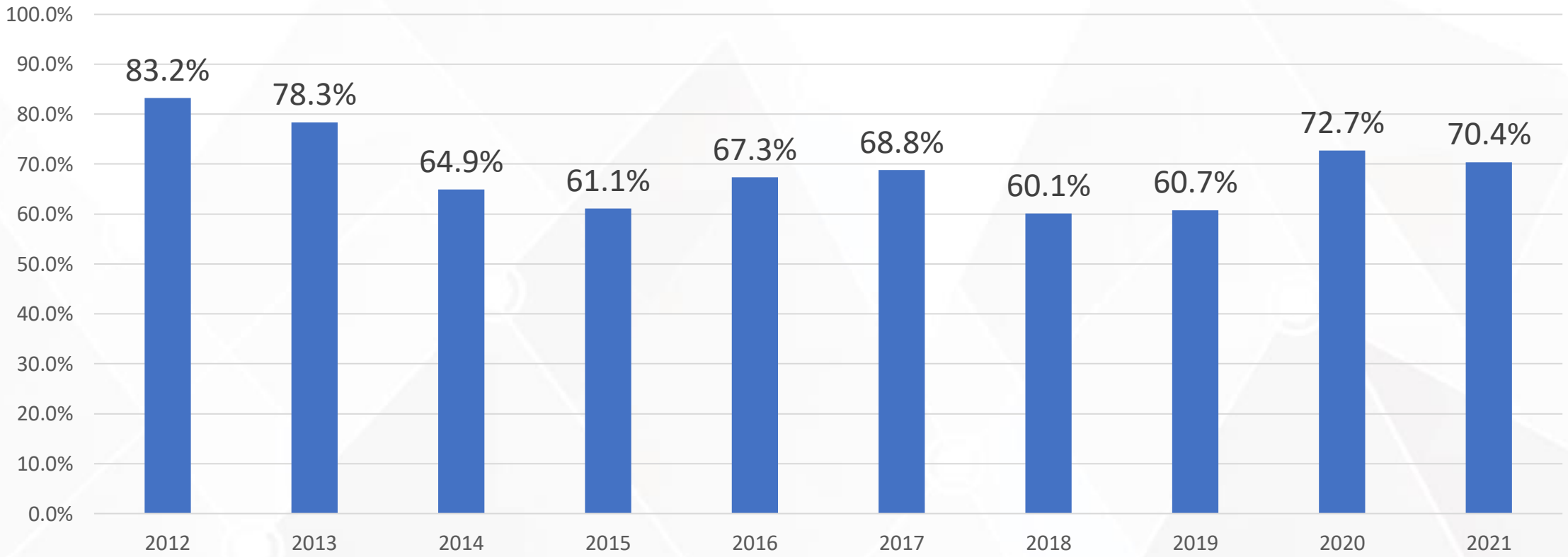

Successes

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- Infant screening rate near the national rate
- Three teleaudiology spoke sites
- Working relationship with out of hospital birth professional licensure board since inception
- Establishment of South Dakota Hands & Voices Chapter
- Establishment of web-based application and resource site
- 2022 Annual EHDI meeting Website of the Year Award
- Student and parent presentation posters in 2022 and 2023 at Annual EHDI Meeting

Tele-Audiology

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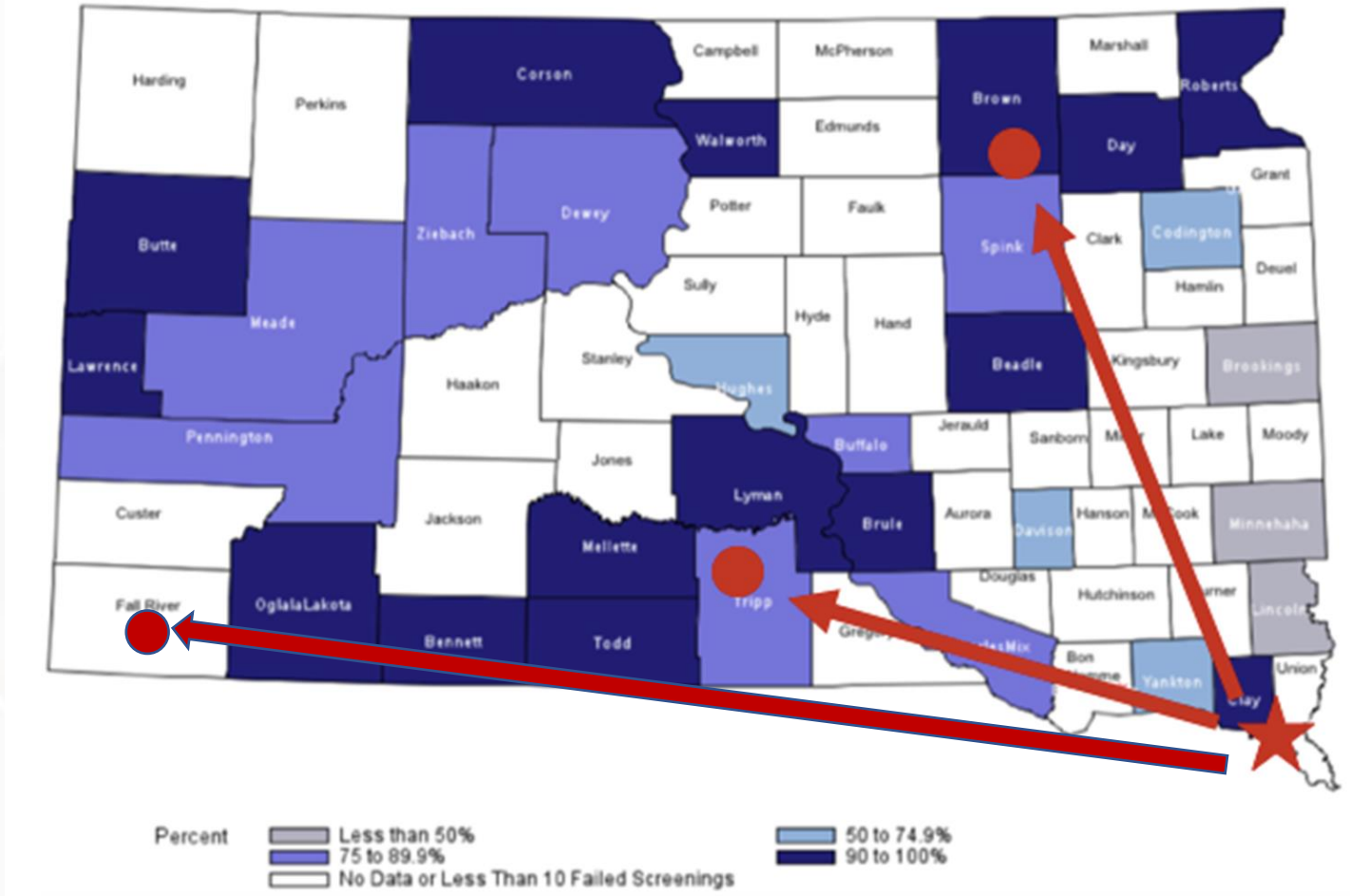
- Partnerships with Winner Regional Hospital, Sanford in Aberdeen, Fall River Health Services in Hot Springs
- Continuing to stress the importance of follow-up care protocol for after a diagnosis of hearing loss
- Onsite training for tele-audiology staff



Newborn Hearing Screening and EHDI

- **Successes and progress over the past year**

- August 2022 Follow Up Program Started

- September 2022 Outreach to Certified Nurse Midwives and Certified Nurse Midwives, Provided Education, Brochures and Need Assessment

- October 2022 – DOH purchased 10 Portable Hearing Screeners

- December 2022 through March 2023 – Distributed 6 screeners to high volume midwives.

- March 2023 – In partnership with SDSD, Identified High Loss to Follow Up in IHS locations – with IHS involvement and SDSD acting as liaison they will provide education, direction, screener responsibility and placement of remaining screeners at these locations

Newborn Hearing Screening Follow Up

What we started and where we want to continue to build

1 Month - Monthly Missed or Not Passed Hearing Screen Reports Sent to Birthing Hospitals, Free Standing Birth Centers, Midwives NBS asked for updates and follow up

2 Month – NBS staff Follow up with Primary Care Provider(PCP) via Fax and parent with a letter with most recent results. Both parties provided with developmental education, recommendations and necessary follow up

3 Month – Check if not passed 2nd screens or diagnostic referrals to Audiology have been complete and recorded

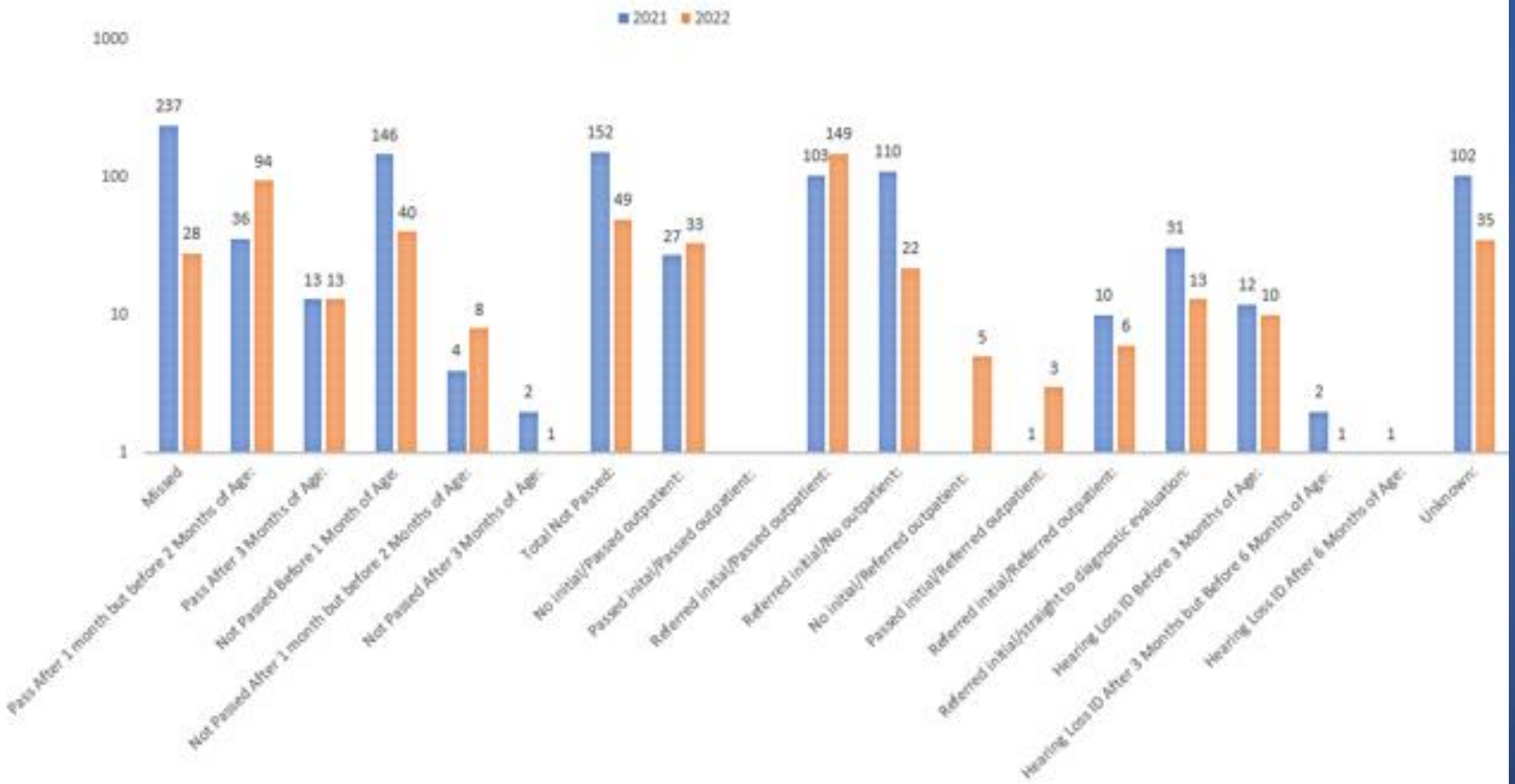
4 Month – Diagnosed Children with Hearing Loss – Check with Audiologist for referral to Birth to Three and SDSD. Check for Birth to Three consents. Remind Audiologist and Notify PCP regarding need or sent referral and consents

5 Months – Follow Up with outstanding missed, not passed, need for diagnostics with PCP

6 Months – Check on Birth to Three Referrals

8 Months – Check Birth to Three Enrollment

NON FOLLOW UP VS. FOLLOW UP COMPARISON



Year	2021	2022
Total Occurrent Births According to Vital Records:	5140	5008
Missed	237	28
Pass Before 1 Month of Age:	4496	4701
Pass After 1 month but before 2 Months of Age:	36	94
Pass After 3 Months of Age:	13	13
Total Passed:	4545	4808
Not Passed Before 1 Month of Age:	146	40
Not Passed After 1 month but before 2 Months of Age:	4	8
Not Passed After 3 Months of Age:	2	1
Total Not Passed:	152	49
Passed initial/No outpatient:	4415	4627
No initial/Passed outpatient:	27	33
Passed inital/Passed outpatient:	0	0
Referred initial/Passed outpatient:	103	149
Referred initial/No outpatient:	110	22
No initial/Referred outpatient:	0	5
Passed initial/Referred outpatient:	1	3
Referred initial/Referred outpatient:	10	6
Referred initial/straight to diagnostic evaluation:	31	13
Hearing Loss ID Before 3 Months of Age:	12	10
Hearing Loss ID After 3 Months but Before 6 Months of Age:	2	1
Hearing Loss ID After 6 Months of Age:	1	0
Unknown:	102	35

NEXT STEPS for Hearing Program Planning – Newborn Hearing Screening with EHDI Follow Up

- USD EHDI grant management – Outside agency outreach
- DOH – Coordination between partnering agencies
- Short term follow up – USD graduate students Fall 2023
- SDSD Diagnostic follow up, Birth to Three refer data, resources, outreach
- Net Smart / My Insight / Vital Records – Case Management Module Addition – Over 2 years out – Modifications to EVRRS until Module complete
- Increase Screeners into more Community Health Offices for Rural Population outreach and failed follow up at Birthing facilities and high-risk infants
- EHDI – Increase tele audiology sites for diagnostic follow up

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Partnership Outreach – Development of Newborn Hearing Interagency Group

- Help develop needed protocols, procedure and policies and data sharing interagency agreements with all parties involved.
- DOH, Vital Stats, USD EHDI, SDSD, DOE B to 3
- Meet quarterly for feedback and process improvements

Funding Sources

MCH Block Grant – Coordinator Salary

EHDI grant – limited funds and has been decreasing every cycle, 25% to Parent Groups

CDC EHDI – not guarantee, not every state gets grant Limited number of awardees- data needs only

Increase NBS fee flat rate fee - to cover Newborn Hearing

Other Legislative Funds ongoing or one time – to expand screener and tele audiology footprint

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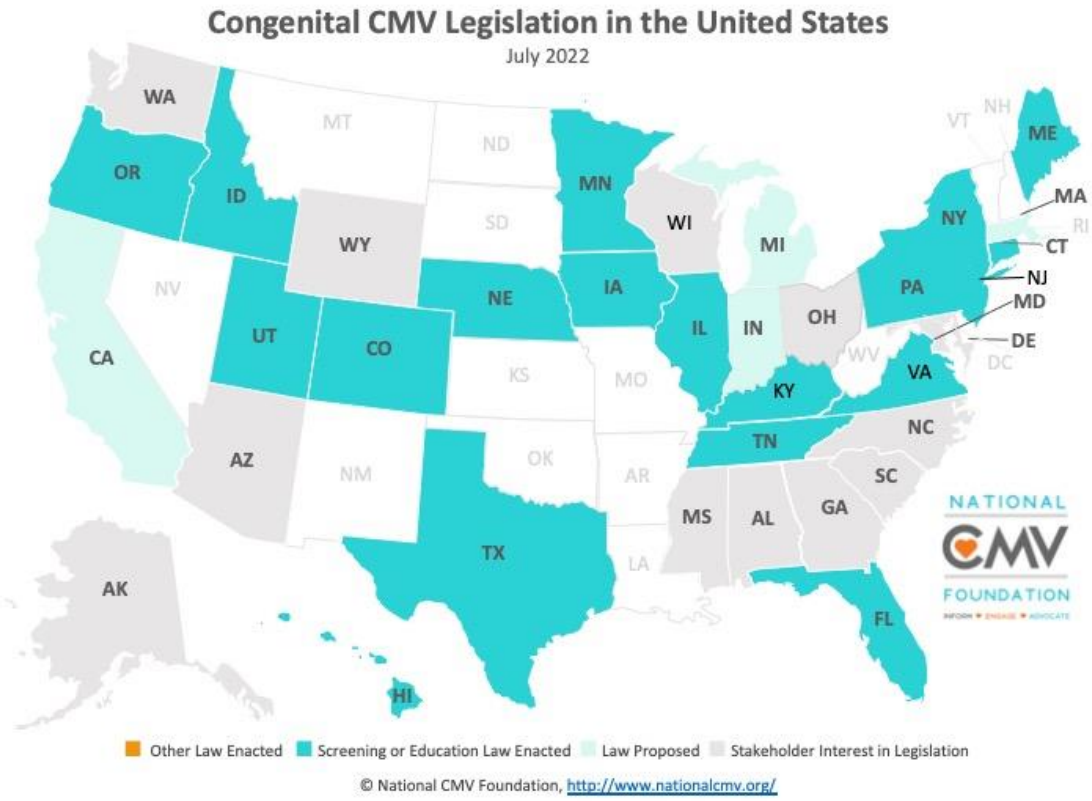
Public Comments

Current Status of cCMV Screening

- Without screening, less than 5% of children born with cCMV are diagnosed.[4]
- Less than 10% of infants with symptomatic cCMV are diagnosed.[5]
- Hearing-targeted cCMV screening will only identify 7% of all children born with cCMV.[3]
- cCMV is more common than the 29 combined metabolic and endocrine disorders on the RUSP.[6]
- CMV testing needs to be done within the first 3 weeks of life if using urine or saliva.[6]
- CMV dried blood spot testing has improved greatly in recent years (75% sensitivity).[2]
- "Treatment" can improve outcomes and can include:
  - Monitoring of hearing
  - Antiviral treatment (if appropriate)
  - Early intervention (automatic qualifier in many states)
- Research has shown that parent attitudes toward cCMV screening are positive.[1]



1.Diener, M. et al.
2. Dollard, SC. et al.
3. Fowler, K. et al.
4. Grosse, S. et al.
5. Sorichetti, B. et al.
6. www.nationalcmv.org

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- National CMV Foundation: www.nationalcmv.org

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Committee Discussion

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Adjourn

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