KANSAS NEWBORN SCREENING ADVISORY COUNCIL MEETING MINUTES

ONLINE VIA ZOOM MEETING

MAY 9, 2024

Members Present

Dr Carolina Beltran, Dr. Kourtney Bettinger, Dr. Britton Zuccarelli, Dr Mike Lewis, Dr. Jennifer Gannon, Meghan Strenk, Karey Padding, Julie Wellner, Shobana Kubendran, Michelle Leeker, Dr. Jack Staddon

Members Absent

Emily Barr, Grace Brouillette, Dr. Laurie Gwyn, Dr. Thomas Loew, Dr. Selina Gierer, Karen Brahman, Gail Webster

KDHE Staff Present

Michelle Black, Drew Duncan, Michelle Mills, Marilee Lowrey, Connie Neuhofel, Shane Morris, Elizabeth Schardine, Zac Leeker, Abegail Bauer, Faith Smith, Gaven Stuhlsatz, Paige Leonard, Sally Brownlee, Timothy Kim, Tyler Brinlee, William Moore, Karen Perez, Lauren Large, Mercedes Robinson,

Others Present

Charlotte Buchanan, Sara Hinton (Commonsense Childbirth), Dr. Bryce Heese, Dawn Mercer (Revity), Emily Stelle (RD at CMH), Emily Teague (KU Wichita Genetic Counselor), Erica Zarse (Midwest Pediatric Specialist, Ped Endo), Randi Gadea, Kayla Jameson (CHCSEK FRS), Dr. Max Fedlt (CMH Endo)

Action items from today's meeting.

1. Dr. Staddon asked how many low risk HgB there were in the same time frame.
2. Discussion of how reporting forms are completed. Dr. Lewis is willing to review the reporting forms and how further discussion.
3. Meeting Frequency to 6 months and review – Gail Webster from 8/31/2023.

Approval of Last Month's Meeting Minutes –

Moved to approve by Dr. Lewis
Seconded by Michelle Leeker

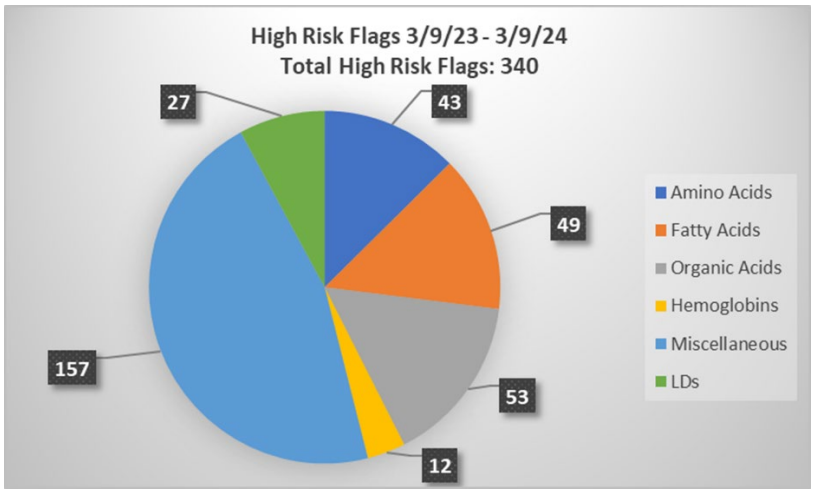
Action Items from last month-

Updates on CF – Proposed by Dr. Beltran
Will present data on condition specific information later
Meeting Frequency to 6 months and review – Gail Webster
Provided mid-year update for condition numbers and ongoing activities
Was able to provide a deeper dive in conditions, outcomes, and breakdown of all abnormal flags – to be presented later.

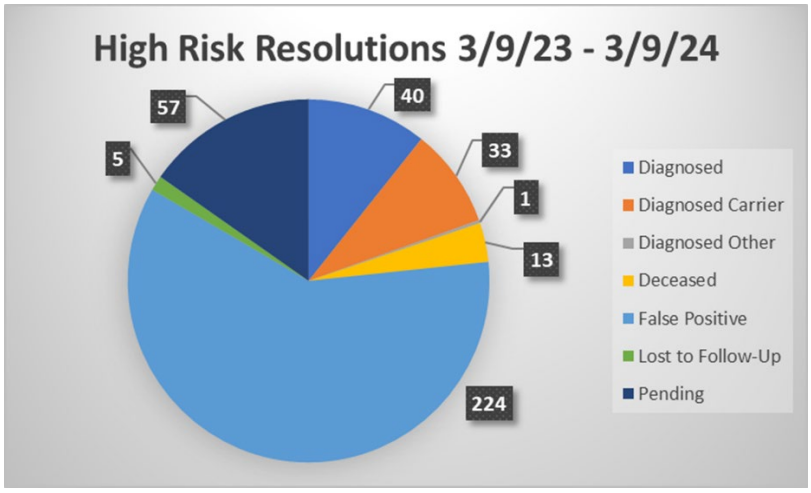
- **General Program Updates-**
    - Data System Update
        - Vendor Selected
        - Awaiting contract from DoA
        - Initial Pre-planning phases
    - Legislative Updates
        - No new legislation
        - Budget proviso to be revisited in 2025
- **Conferences**
    - APHL Conference
        - Upcoming 10/20-2024
    - ALD Alliance Meeting
        - 3/20 Lab Staff attended.
    - Heartland Regional Genetics
        - 4/23-4/25 of 2024. Staff attended.
- Dr. Lewis asked about states that are doing great in NBS.
    - Zac responded with there are many variables including how states fund the NBS. Kansas is only one of 4 states that still do not charge families for NBS. It is also based on how states count their screens.
        - Dr. Staddon stated
          Michigan ~10 million population; Kansas ~3 million. This could be part of funding difference
    - Dr. Gannon asked about how states are counting second tier. Zac responded with some states screen for Zellweger's but not all babies are screened for Zellweger's but it is being counted.
        - Dr. Gannon clarified what is a second tier test and what is a second tier screen.
    - Michelle Mills mentioned.
        - APHL formed a counting subcommittee as a guide for counting. They hav e made suggestions but nothing has been stated definitively.
        - Tennessee counts primary and secondary conditions. MO counts primary and secondary. KS only counts primary conditions.
- **Staffing**-
    - NBS Long Term Follow Up Program Manger
        - Christina Ferguson, hired on January 22, 2024
        - The position is also providing support to grant writing.
        - Currently connecting with other states as we build out what specifically LTFU means in Kansas and specifying what datapoints we aim to collect in order to make fruitful changes to our program.

- **New AC Member-**
    - Pediatric Hematologist/Oncologist
        - Dr. Jack Staddon, KU School of Medicine - Wichita

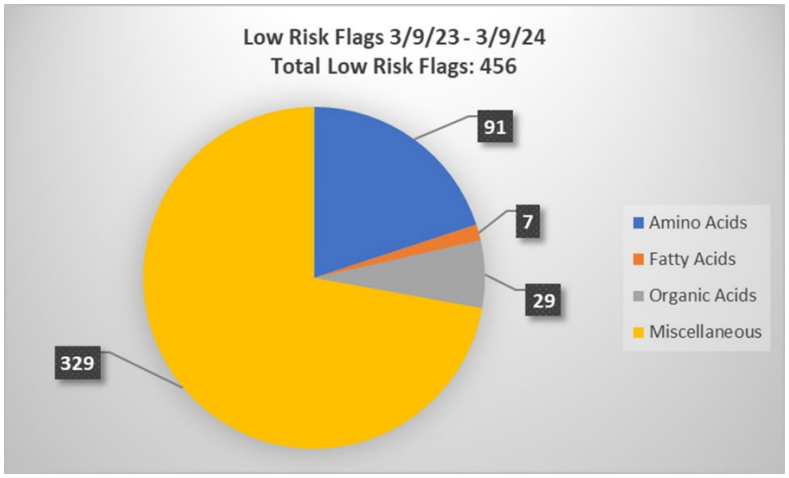
- **HRSA NBS Propel Grant Updates**
    - Accomplishments – Year 1
        - X-linked Adrenoleukodystrophy (X-ALD) screening went live in February 2024, aligning KDHE NBS with 35 of the 37 Recommended Uniform Screening Panel (RUSP) conditions.
        - The Mucopolysaccharidosis Type II (MPSII) pilot began in March 2024, with an anticipated go live date of the fourth quarter of calendar year 2024, further aligning KDHE NBS with the RUSP.
        - Started development of LTFU program
        - Specialty Clinics Contract
        - Contract with Sickle Cell Association of the Midwest
    - Upcoming Year 2
        - Continued Development and Implementation of LTFU
        - Evaluation and Lab onboarding of new conditions
        - Purchase of 2 new plate readers
    - Michelle Leeker asked Do we have a timeline of being able to add new diseases from the RUSP?
        - Zac stated that he believes the lab presentation will address this question later in the meeting.
        - Michelle Mills stated We are not under a legislative RUSP alignment, but we are always trying to bring on new conditions in a timely manner.
- **Special Health Care Needs Program**
    - Recently restructured and have been placed into Screening and Surveillance section along with Newborn Screening, Birth Defects, CCHD/Hearing, and Outreach/Education
    - Current Program Projects/Focus
        - Review and evaluation of forms and letters
        - Designated effort towards hemophilia direct assistance payments with targeted outreach
        - Development of regionally specific outreach and resources folder
    - Karen Perez introduced herself as the new SHCN program manager.
- **Condition Discussion**
    - High Risk Flags for 3/9/23 to 3/9/2024



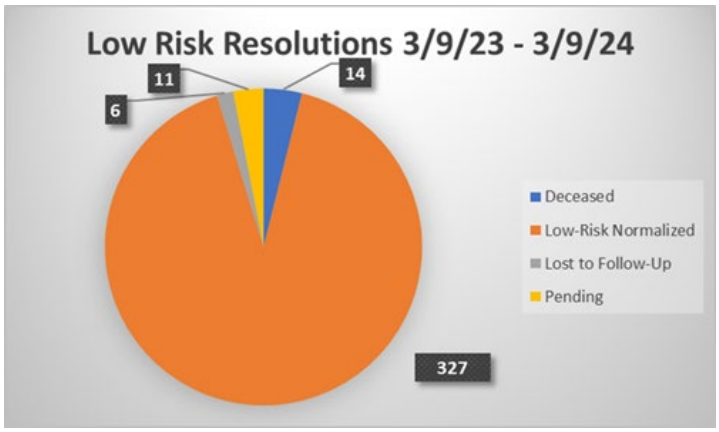
- High Risk Resolutions 3/9/2024



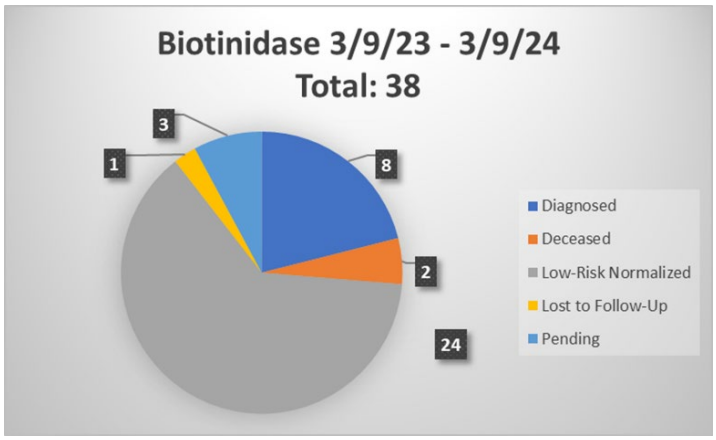
- Dr. Staddon asked how many low risk HgB there were in the same time frame.
  - Low Risk Flags 3/9/2023 to 3/9/2024



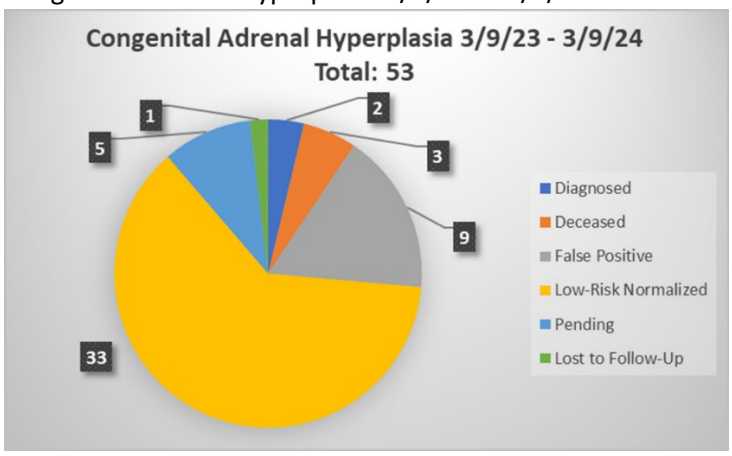
  - Low Risk Resolutions 3/9/23 to 3/9/24



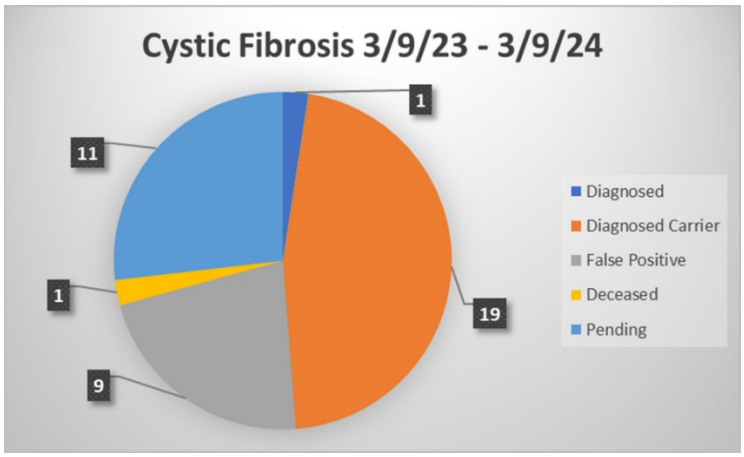
- Biotinidase 3/9/24 to 3/9/24

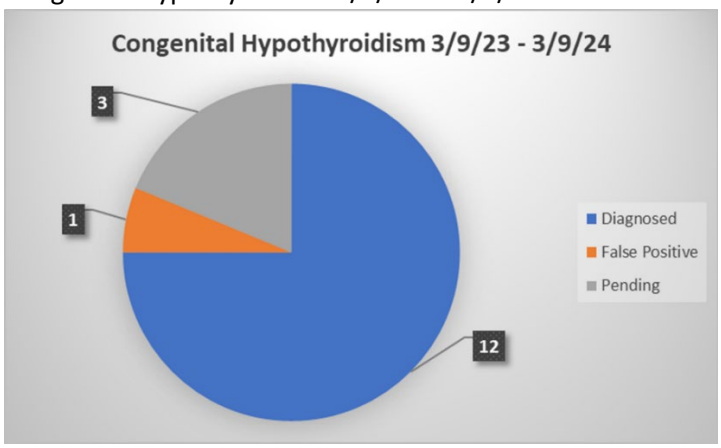


- Congenital Adrenal Hyperplasia 3/9/23 to 3/9/24

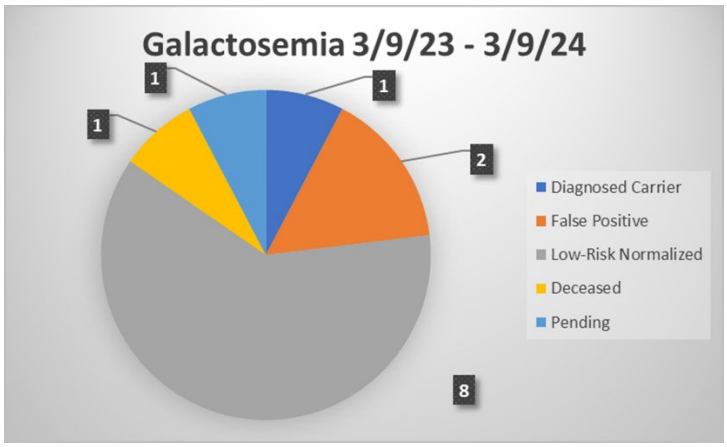


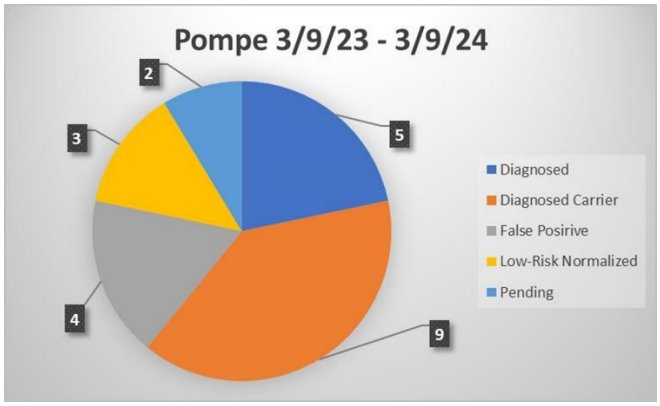
- Cystic Fibrosis 3/9/23 to 3/9/24

- 

  - Discussion of how reporting forms are completed. Dr. Lewis is willing to review the reporting forms and how further discussion. Action Item
  - Julie Wellner mentioned that The Wisconsin report says what variants are reported so you should know if carrier or not.
  - Randi Gadea mentioned Regarding the forms - from our perspective, the forms offer the options of "diagnosis excluded" or "diagnosis confirmed". There is not an option on either for "carrier" so if that is something that could be added to the form for clarity I would be open to it!
  - Zac mentioned that we have a medical director now that is able to help the program with making medical decisions.
  - Zac mentioned if any other providers are wanting to look at reporting forms we are willing to review.
- Congenital Hypothyroidism 3/9/23 to 3/9/24
- 

- Galactosemia 3/9/23 to 3/9/24

- 

- Pompe 3/9/23 to 3/9/24
- 

- **HgB Trait Discussion (a follow up)**
  - Misconceptions surrounding trait and 'no negative health impacts'
  - Had conversations with Dr. Shurney from the Sickle Cell Foundation of Minnesota, Dr. Staddon from KU – Wichita, CMH Pediatric Hematology/Oncology, and Kevin Wake from the Uriel E. Owens Sickle Cell Disease Association of the Midwest to review letters and protocols
  - Updates include: Revised parent and provider letters with updated timeline and intervention recommendations and increased parent and family information surrounding SC disease and Trait
- **MPS II Pilot Phase**
  - Total of 7000 specimens have been ran since beginning pilot phase
  - Have yet to have a single high-risk flag during pilot

- **Presentation by Sara Hinton about Chagas Disease**
  - **Not sure what we need to put for her presentation**
  - **Sara commented after her presentation.**
    - Ididn't go over neonatal presentation of Chugs disease so I'll put a brief o verview here:

      Neonates with congenital Chagas disease often have no clinical indication of infection, but 10%-

40% present with low birth weight, prematurity, low Apgar scores, hypoto nicity, fever, hepatosplenomegaly, or anemia. More severe presentations c an include myocarditis, meningoencephalitis, and pneumonitis. The more severe manifestations, while rare, have a high risk of mortality.

- **KHEL Updates**
  - Newborn Screening Laboratory Updates
    - Update on New Conditions
      - X-linked Adrenoleukodystrophy
        - Go Live was February 1, 2024
      - Mucopolysaccharidosis II
        - Pilot in progress
        - Estimated Go Live August 2024
      - Krabbe
        - Capable of piloting and Go Live in 2025
      - GAMT
        - Capable of Method Development and Go Live in 2025
  - Recommended Uniform Screening Panel
    - The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) met January 29-30, 2024.
      - Krabbe was recommended for RUSP inclusion status on January 30, 2024, with a 10 to 3 vote.
      - Duchenne's Muscular Dystrophy was voted to move forward to full evidence review on August 9, 2023.
        - Literature review expected to be completed prior to May 2024.
        - Public Health System Impact Assessment expected to be completed prior to May 2024.
      - cCMV was returned with request for more data and encouraged to resubmit nomination package.
      - Nomination packages currently under consideration:
        - Metachromatic Leukodystrophy (MLD)
        - Biliary Atresia (BA)
        - Duchenne's Muscular Dystrophy (DMD)
      - Currently meeting May 9-10, 2024.
        - Final Evidence Review of DMD will be presented.
  - **Collaborative Laboratory Integrated Reports (CLIR)**
    - **About CLIR**

      Collaborative Laboratory Integrated Reports (**CLIR**) is an interactive Web Tool created jointly by staff of the Biochemical Genetics Laboratory, Department of L Medicine and Pathology, and of the Department of Information Technology, **Mayo Clinic**. Key contributors and collaborators are located at **Oslo University H Norway**, and at the **California Department of Public Health**.
  - CLIR Data 2021-2023 completed
    - Tyler Brinlee, KS NBS Scientist
    - Timothy Kim, KS NBS Scientist
  - CLIR Data - working on 2024

- CLIR reference ranges are derived by analysis of hundreds of thousands of data points from different laboratories
- Some states say it is too complicated to contribute and won't use CLIR

- **Quality Indicators**
  - **Total Summary of Turnaround Time for Newborn Screening**
    National Goal: 7 Days from Birth to Collection (6 Days from Collection to Report)
    Acceptible Critera: 3 Days from Collection to Receipt + 3 Days from Receipt to Report = 6 Days Collection to Report

    2023

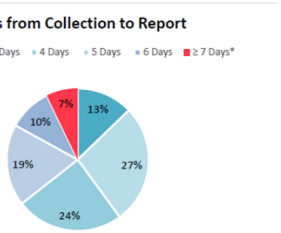
Total Samples Received	39599
Total Samples Reported	39776

    Days from Collection to Report

≤ 2 Days	5230
3 Days	11100
4 Days	10003
5 Days	7588
6 Days	4034
≥ 7 Days*	2924

    Days from Collection to Receipt

≤ 3 Days	33894
≥ 4 Days	6985

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    Acceptible Critera: 3 Days from Collection to Receipt + 3 Days from Receipt to Report = 6 Days Collection to Report

    Days from Receipt to Report

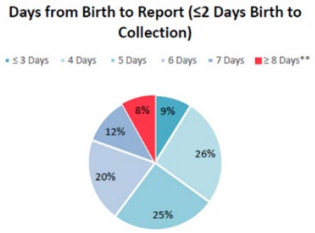
≤ 2 Days	32767
3 Days	6129
4 Days	1392
≥ 5 Days	591

    Day from Birth to Collection

≤ 2 Days	35048
≥ 3 Days	5831

    Days from Birth to Report (≤2 Days Birth to Collection)

≤ 3 Days	3119
4 Days	9131
5 Days	8859
6 Days	7089
7 Days	4004
≥ 8 Days**	2846

- **PVV and FPR for 2023**

  Positive Predictive Value (PPV): The positive predictive value of a test is the probability that the patient has a condition when restricted to those patients who test positive (referrals for confirmation). **PPV for NBS Combined = 32.69%**

BIOT = 100%	**AAD = 8.82%**	**SMA = 100.0%**
CAH = 18.18%	**OAD = 2.44%**	**X-ALD = 75.0 %**
CF = 16.66%	**FAD = 28.00%**	**MPS I = 100.0%**
CH = 100.0%	**HGB = 100.0%**	**Pompe = 66.67%**
GALT = 100.00%	**SCID = 1.69%**	**CF Carrier = 62.96%**
GALT Carrier = 100.00%	**DiGeorge = 1.69%**	**SCID Other = 3.33%**

  False Positive Rate (FPR): The false positive rate of a newborn screening program is expressed as the proportion of positive tests in patients proven by follow up evaluation not to have one of the conditions targeted by the newborn screening program. **FPR for NBS Combined = 0.33%.**

BIOT = 0.00%	**AAD = 0.07%**	**SMA = 0.00%**
CAH = 0.02%	**OAD =0.09%**	**X-ALD = 0.002%**
CF = 0.02%	**FAD = 0.04%**	**MPS I = 0.00%**
CH = 0.00%	**HGB = 0.00%**	**Pompe = 0.01%**
GALT = 0.00%	**SCID = 0.13%**	

- Dr. Gannon asked in the chat.
  - Michelle, I'm wondering if babies are getting exposed to something prior to the screen that is causing the C3 false positive?
  - Michelle Mills stated it was likely.

- Sara asked what kind of exposure and Dr. Gannon stated she does not know what kind of exposure.

- **Detection Rate and Sensitivity**

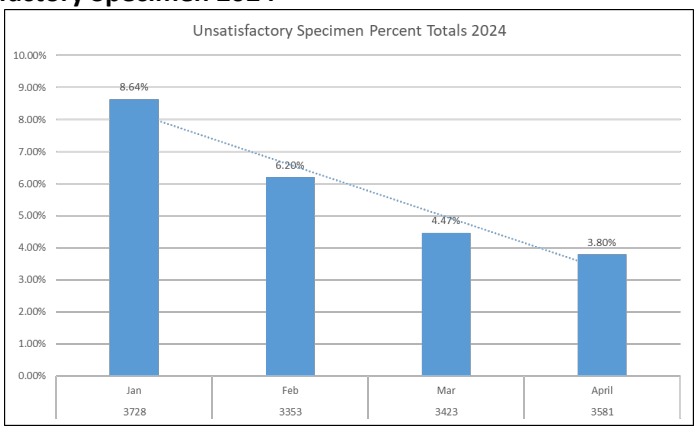
  - **Detection Rate (DR)**: The detection rate of a newborn screening program is expressed as the number of neonates that on average needs to be tested to detect one affected patient. This is often referred to as the "Prevalence". To get a more truthful detection rate, a state NBS program should use combined data from four to five years of screening. **DR for NBS Combined = 1:512 (with 60 open cases).**

BIOT = 1:10,757	**AAD = 1:14,342**	**SMA = 1:21,514**
CAH = 1:21,514	**OAD = 1:43,028**	**X-ALD = 1:43,028 male (1:21,514 Female)**
CF = 1:21,514	**FAD = 1:6,147**	**MPS I Carrier = 1:21,514**
CH = 1:3,585	**HGB = 1:10,757**	**Pompe = 1:3,586**
GALT = 1:21,514	**SCID = 1:43,028**	**CF Carrier = 1:2,531**
GALT Carrier = 1:21,514	**DiGeorge = 1:43,028**	**SCID Other = 1:21,514**

  - **Sensitivity**: Sensitivity is the ability of a test to identify individuals who have a given disease or disorder. The more sensitive a test is the fewer "false-negative" results it produces. False-negative results fail to expose disease states that may be present and can lead to developmental disabilities and/or death in the infant, and subsequently distrust in the testing method. Although the goal has historically been to achieve 100% sensitivity, even at the expense of specificity if necessary, it may not be applicable for some of the metabolic disorders that are not reliably detectable in the immediate newborn period. Consequently, without mandatory repeat screening on the entire newborn population, there will always be the possibility of missing some rare delayed onset and/or milder genetic variations of the conditions that we screen for. The SMA screening test has an expected 5% false negative rate.

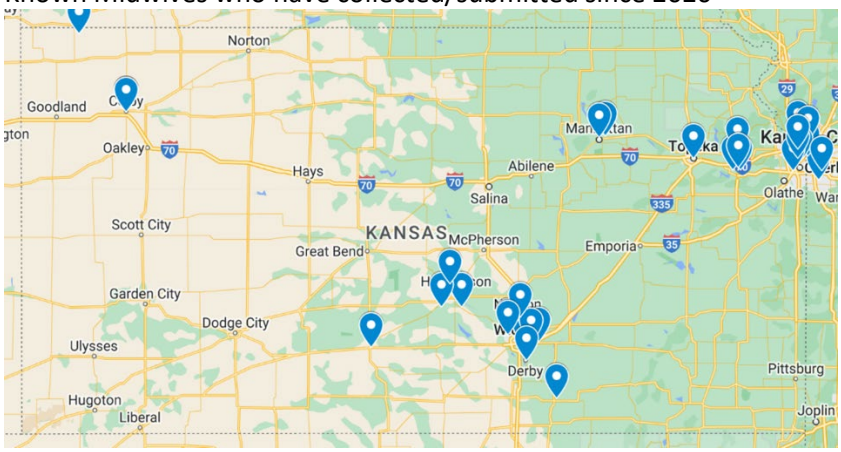
  - **Sensitivity = 100%**

- **Unsatisfactory Specimen 2024**

  - 

- **New Laboratory**
  - Official opening date will be Summer 2025
  - Lab will be moving the majority of the 1st Quarter 2025
  - Julie Wellner asked if follow up was moving to the new lab also and the answer is no but the new lab is only a block away from where follow up located.

- **Follow up Updates**
  - Specialist Consultation Contracts
    - Have finalized contracts, all parties signed, and routed to 5th floor contracts team
  - Contracts in place for CMH Genetics and KUMC Genetics
  - Comprehensive Letter Review Continued.

- Dr. Staddon suggested
  My suggestion would be to send the medical translation letter to specialis ts in our group who speak Spanish. Sometimes even medical interpreters are inadequate.
- Dr. Zuccarilli stated
  I'm a certified bilingual provider (Spanish) and am happy to review any ne urological condition letters
- Working on a full medical Spanish translation for each conditions parent letter
- Plan to add condition specific info sheet to parent letters in the near future
- Will be adding QR codes to letters when new website is finalized to direct families to additional condition information and supports.
- Exploring Midwives Contracts.
  - Issue: Have noticed a barrier in recollection for rural families who received an unsatisfactory specimen from hospital collections.
  - Concept: Contract with midwives who are willing to drive to families nearby to recollect the specimen.
  - Notable desrts. North Central, Southwest and Southeast.
  - Known Midwives who have collected/submitted since 2020
  - 

- Recollection discussion
  - Dr. Staddon asked
    Pediatrician or family practice can't redraw in their office or rural a ccess hospital?
  - Drew stated that
    Several don't have the collection cards, or familiarity with the colle ction process to ensure a valid collection.
  - Dr. Lewis asked what happens when another state collects on their card and sends it to KS.
    - Michelle Mills stated that if KHEL receives a card from a bordering state they call the collecting facility to inform them and then over nights the specimen to the correct state lab.

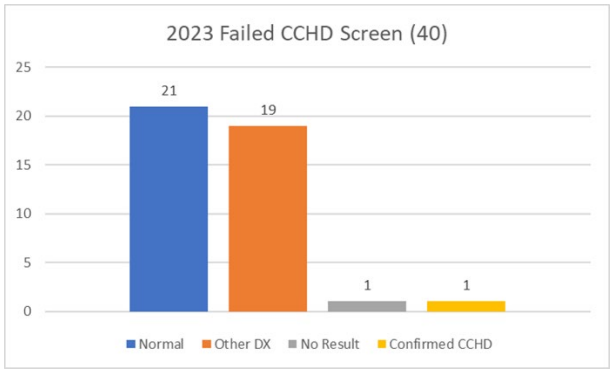
- Michelle Black stated that we are aware of situations where a KS card was sent to NE and NE completed the testing in their state.

- **Hearing and CCHD updates**

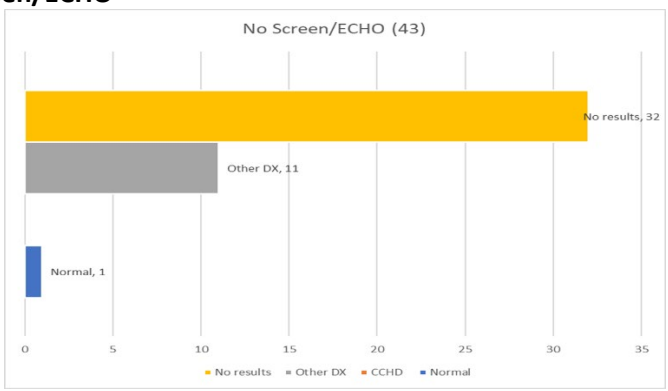
State of Kansas Newborn Screening

Births	35764
Deaths	96
No Consent	79
No Consent Medical	2
Discharged Before Screen	54
Transf/NICU/No Results	2791
No results reported	486
Pass	32129
Failed Screen	40
ECHO/No Screened	43

- **2023 Failed CCHD Screen**



- **No Screen/ECHO**



- **2022 and 2023 data**

2023 Annual Preliminary Data					CDC National Average		
	2022		*2023		2021 NA	2020 NA	2019 NA
Total Birth	36,249		35,848				
Total Screened	35,877	99%	35476	99%	98%	98%	98%
Total OHB Births	1012		1016				
Total OHB screened	835	83%	867	85%			
Total screened < 1 month	35,123	98%	34773	98%	95.5%	97.2%	97.7%
Total who need Diagnostic Evaluation	592		508				
Total who received a Diagnostic Eval	446		329				
Total Diagnosed < 3 months	289	65%	224	68%	42.7%	60.6%	79%
Total with no diagnosis	147		179				
Awaiting DX	10		39				
Moved, Adopted, declined, expired	57		31				
LFU Unresponsive	53		1				
LFU Unable to contact	25		1				
Unknown	2		107				
Diagnosis LFU	80/592	14.95%	107/509	21.00%	32.4%	29.8%	27.50%
Total hearing loss	77		79				
Total Eligible for EI Services	70		69				
Total enrolled in EI	61	87%	48	70%	60.2%	61.4%	61.7%
Total enrolled in EI before 6 months	43	70.49%	40	83%	42.2%	72.4%	73%
Awaiting IFSP	1		5				

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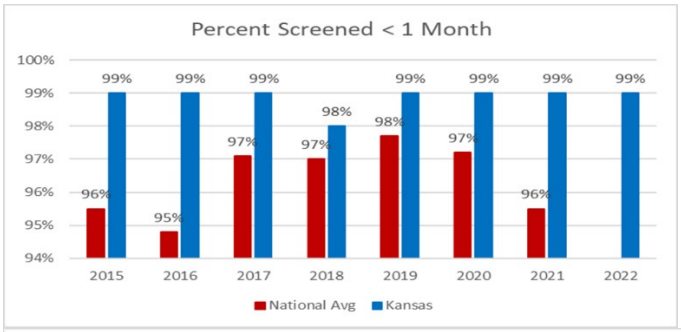
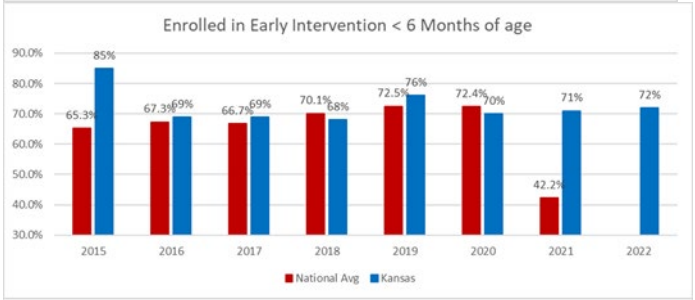
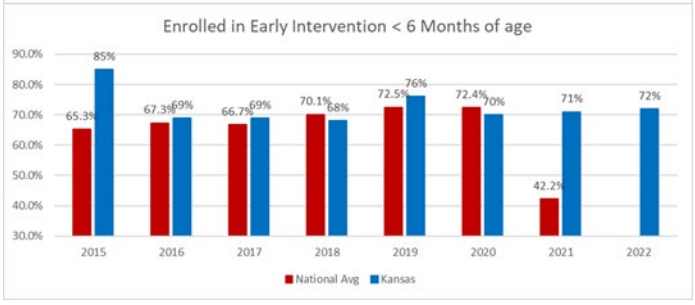
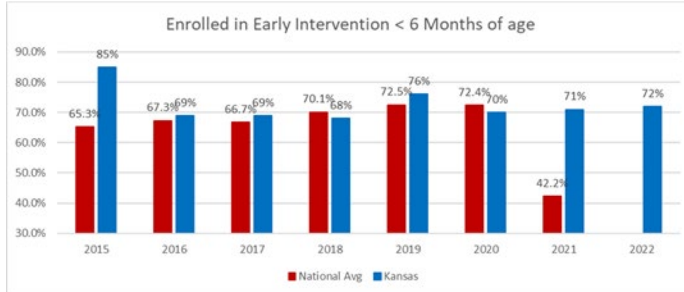
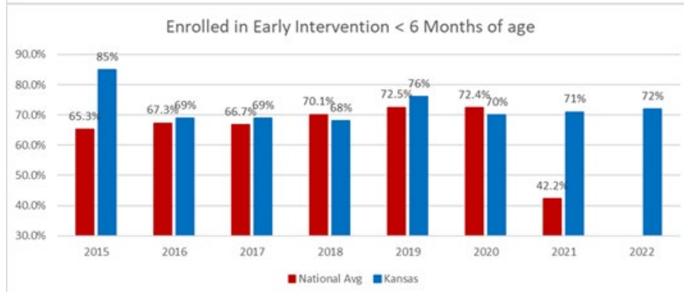
- **Longitudinal look at data**
  - 

  - 

  - 

- 
Enrolled in Early Intervention < 6 Months of age
National Avg
Kansas
- 
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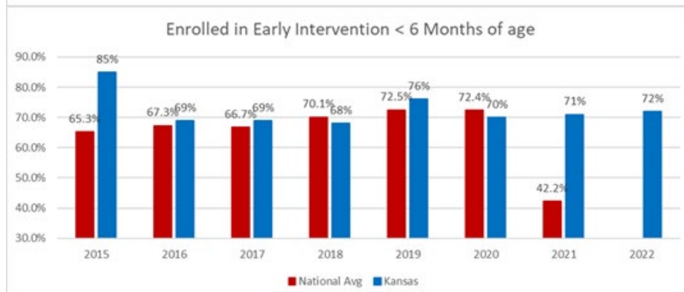
- 
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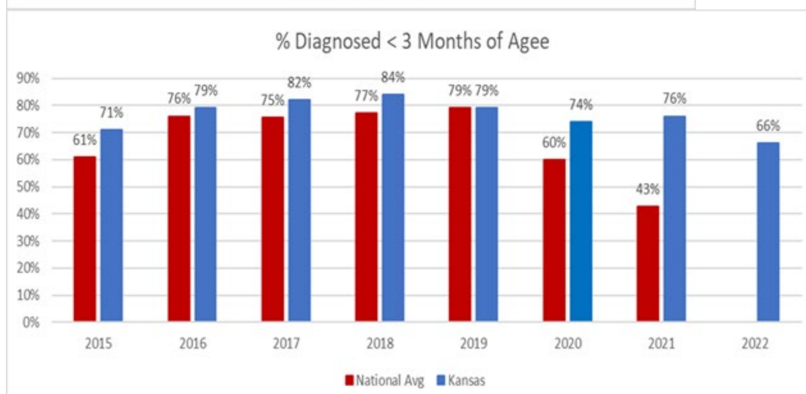
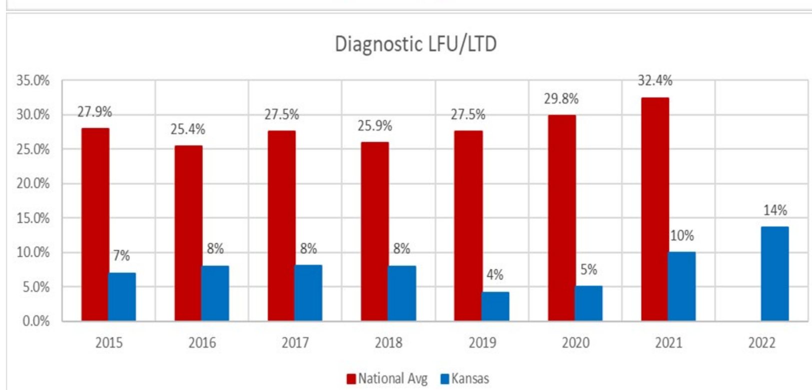
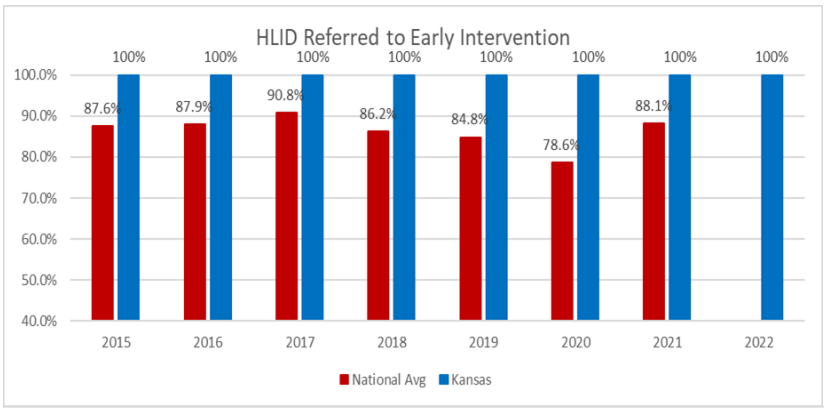
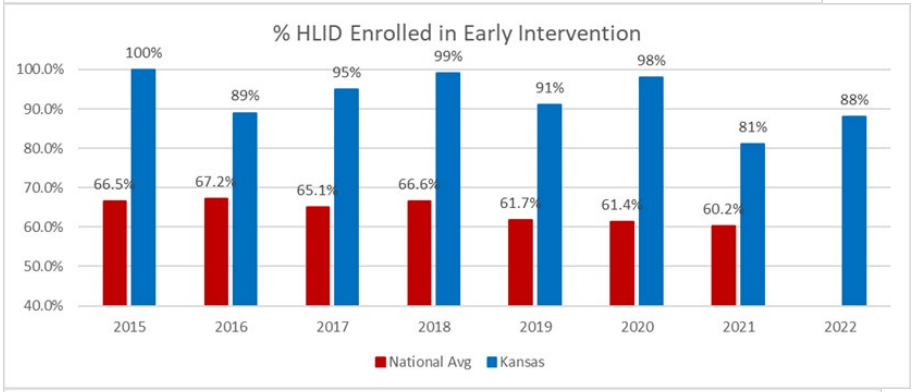
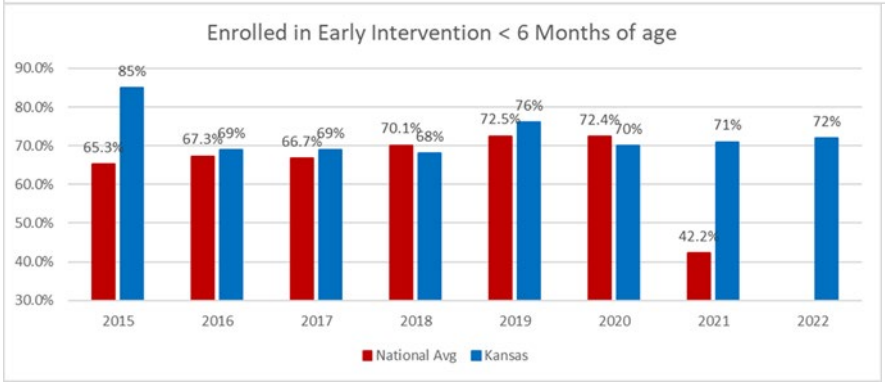
National Avg
Kansas
- 
% Diagnosed < 3 Months of Agee
National Avg
Kansas
- 
Diagnostic LFU/LTD
National Avg
Kansas

- 

- 

- 

- **Education and Outreach**
  - Increased visits to hospitals, clinics and labs in 2023 and 2024.
  - Continuing to increase presence at community health fairs and other cross-cutting events.
    - Baby Showers
    - Housing Conference
    - GPHC
    - NEK Head Start Conf
  - **Guide to Kansas Family Support**
    - <u>Hospital and Birthing Centers</u>
      - More than 10,000 pamphlets have been disseminated to various hospitals and birthing centers.
      - Sent to every hospital/birthing center in Kansas.

- Will send to every county health center by end of year.
- Education to introduce brochure during the prenatal period.

- <u>Additional Resource Locations</u>
  - Over 40,000 pamphlets have been distributed to non-birthing locations.
  - Average of 25 to 100 pamphlets per location, some of which include:
  - Public Libraries
  - Workforce Centers
  - Childcare Centers
  - Non-profits
  - Health Depts.
  - DCF
  - Goodwill
  - Housing Authority locations/clinics
- Newsletter Numbers and Updates
  - Newborn Screening Newsletter combined with several other public health programs.
  - Previous number of subscribers = 281
  - Total Subscribers now over 420
  - GovDelivery replaced MailChimp
- Screening and Surveillance Vanity Website
  - In the QA portion of the project with expected launch timeline of 2-3 weeks.
  - Included family survey on new website to capture first-hand experiences from families on their newborn screening experience.
  - KMCHC Perinatal/Infant Workgroup Project
    - Plan to leverage new website to host a tab for “new parents”; will provide critical information after birth to mothers and families. Post cards to families with a “well screen”.
- Annual Awards
  - Overhauled Annual Awards
  - New Graphics
  - New Pictures
  - Split awardees into three different categories
    - Midwives
    - Birthing Hospitals
    - Screening Facilities
  - Awardees can be recognized for Gold or Silver
    - Gold = 3 or more areas of recognition
    - Silver = 2 areas of recognition

    - Six different areas of recognition
        - Complete Demographics
        - CCHD Screening
        - Hearing Screening
        - Collection Age
        - Transit Time
        - Unsatisfactory Rate
    - Prizes Changed
        - No banner
        - Gold and Silver each get a media package.

- **Comments/Discussion**
- **Chair and Vice Chair Nominations**
    - Dr. Gannon nominated Dr. Carolina Beltran for Council Chair.
    - Dr. Lewis seconded the motion for Dr. Beltran
    - Dr. Bettinger asked for a vote to confirm Dr. Beltran
        - Members voted to approve the motion.
    - Dr. Beltran nominated Dr. Michael Lewis for a co-chair.
    - Julie Wellner seconded the motion for Dr. Lewis
        - Members voted to approve.
- Dr. Beltran thanked the council for trusting her to assist the council. She also thanked Dr. Gannon for her support in leading the council and her knowledge if moving the council forward.
- **Next meeting is mid-November**
    - Will send out a Doddle poll