KANSAS NEWBORN SCREENING ADVISORY COUNCIL MEETING MINUTES

ONLINE VIA ZOOM MEETING

NOVEMBER 14, 2024

Members Present

Dr Carolina Beltran, Dr. Britton Zuccarelli, Dr Mike Lewis, Dr. Jennifer Gannon, Julie Wellner, Michelle Leeker, Karen Brahman

Members Absent

Emily Barr, Grace Brouillette, Dr. Laurie Gwyn, Dr. Thomas Loew, Dr. Selina Gierer, Gail Webster, Dr. Jack Staddon, Meghan Strenk, Karey Padding, Dr. Kourtney Bettinger

KDHE Staff Present

Michelle Black, Drew Duncan, Michelle Mills, Connie Neuhofel, Shane Morris, Zac Leeker, Faith Smith, William Moore, Lauren Large, Abegail Bauer, Mercedes Robinson, Jacqueline Fung, Tyler Brinlee, Timothy Kim, Gaven Stuhlsatz,

Others Present

Charlotte Buchanan, Stelle (RD at CMH), Emily Teague (KU Wichita Genetic Counselor), Brooklyn Tolbert (KU Wesley genetics nurse), Victoria Prater-Rochier, KU Wesley genetics.

Action items from today's meeting.

- Email out the bylaws of the council for the review before the next meeting.
- Talk about making a subcommittee for bylaw reviews.
- How should we address gaps in participation.
- Consider a matrix or framework to determine order of adding new conditions

Approval of Last Month's Meeting Minutes –

Dr. Lewis motioned to approved and Michelle Leeker seconded.

Action Items from last month-

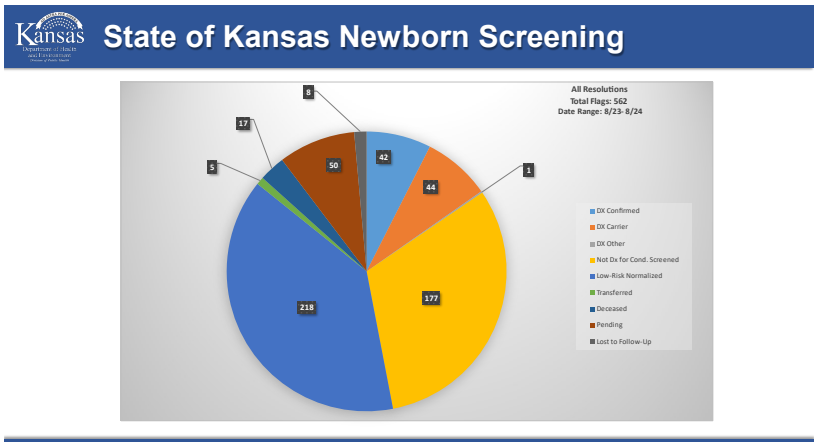
- Dr. Staddon question about # of low risk HgB cases. Have since compiled detailed breakdown of all case counts.
    - Going forward we plan to break down the cases into high risk and low risk.
- Dr. Lewis to review CF Dx Reporting Forms with Program
    - Ongoing and continuing to meet with Dr. Lewis to improve the letters.

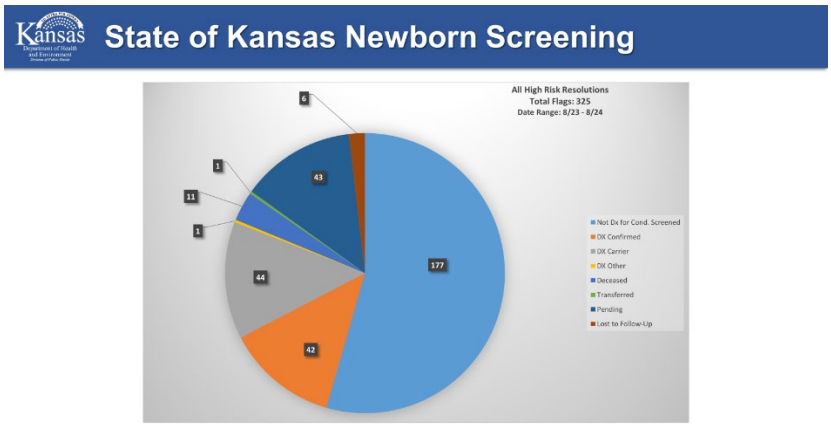
- **General Program Updates-**
    - Data System Update
        - Information gathering activity complete

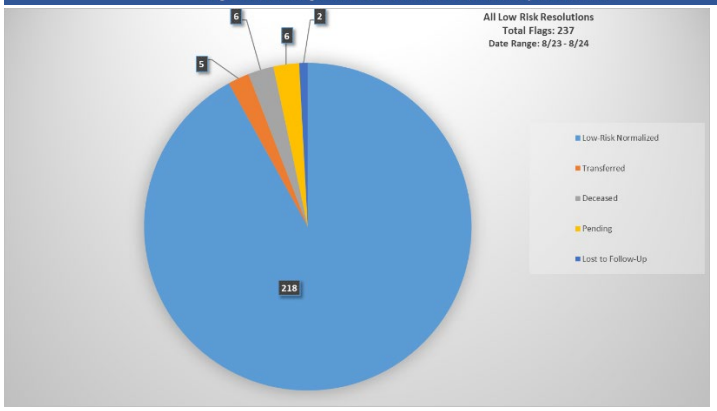
        - In pre-development stages
        - Initial project meetings have begun
    - Legislative Updates
        - Budget Proviso extended to 2026
        - Discussions with Lab for Legislative requests for 2025 session
    - Conferences
        - APHL Conference (Omaha, NE)
        - 10/20-24
- **Follow-Up Updates**
    - NBS Genetic Counselor - Jacqueline Fung, CGC, Hired 10/14/24
    - HRSA NBS Propel Grant Updates -Lab
        - Accomplishments – Year 1
            - Went live with X-ALD on Feb 1, 2024
            - MPS II pilot was started March 1, 2024
            - Assay development for quantitative BIO and GALT assays started
        - Upcoming/Current Activities – Year 2
            - Went live with MPS II on August 1, 2024
            - Purchased additional new instrument for installation in the New Lab for instrument redundancy and increased testing capacity.
            - Purchased additional instrumentation to begin assay validations for Krabbe post Lab Move.
        - HRSA NBS Propel Grant Updates -Follow up
            - Accomplishments – Year 1
                - Initiated contracts with Specialty Clinics
                - Established partnership with Uriel E. Owens Sickle Cell Association of Midwest
                - Launched Long-term Follow-up (LTFU):
                - Conducted outreach and surveys with families of children diagnosed with hemoglobin (Hgb) disorders over the past 9 years to assess their needs.
                - Started preparations to expand outreach to a new condition.
            - Upcoming – Year 2
                - Implementation of Genetic Counseling supports
                - Expand surveys to additional families and conditions.
                - Define data collection requirements for long-term follow-up.
                - Identify and provide resources for newly diagnosed families.
        - APHL Conference -Omaha, NE (10/20-10/24)

- Some staff provided "key takeaways"
- FHIR system – In development and allows NBS programs to pull data from numerous different EHRs when patient information is missing, or wrong provider might be listed.
- Staff retention and recruitment strategies were presented. Data around turnover rates for the two youngest generations in the work force. Establishing clear career pipelines through local universities. "The Public Health Millennial" provided great resources on this.
- COOP/Disaster Response Plan presentation from NC. What happens when roads are gone and specimens can't be transported?
- Face to face collaboration with other state's NBS programs.
- KS is excelling with collaboration between NBS and other MCH programs

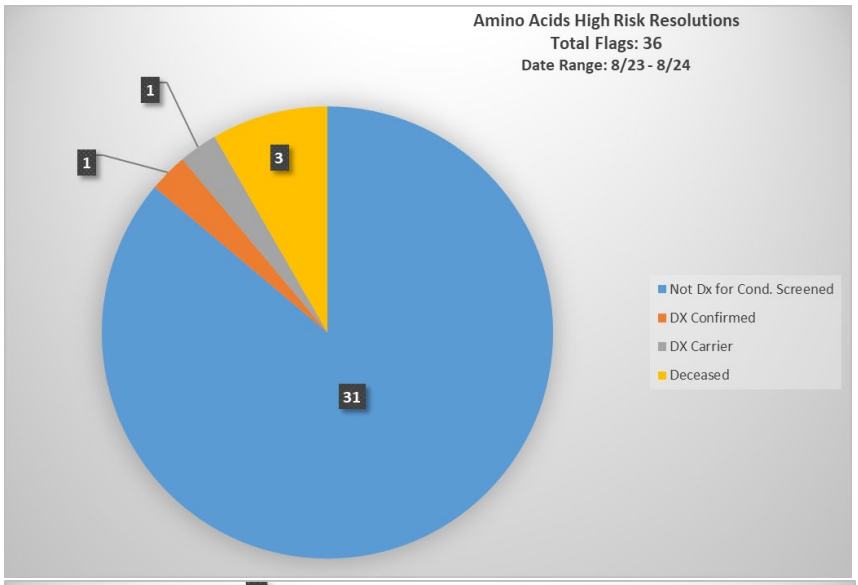
- ~~Guest Presenter~~
  - ~~Dr. Matthew Ellinwood, National MPS Society~~
- GC introduction
  - Jackie introduced herself to the council.
  - Zac explained that Jackie would be focusing on HGB and HGB trait first and then moving to other conditions.
  - Also addressed the propel grant and how the educational materials and handouts are developed.
    - Christina stated that they would connect with the specialist to develop the documents and utilize the CDC information.
- Discussion about making a subcommittee to review the A/C bylaws.
  - Victoria Prater-Rochier volunteered, and Dr. Beltran also agreed to participate.
  - Dr. Gannon asked for clarification on specific goals and anticipated time commitment?
  - Julie asked that we email the bylaws for review before the next meeting.
- Data Presentation

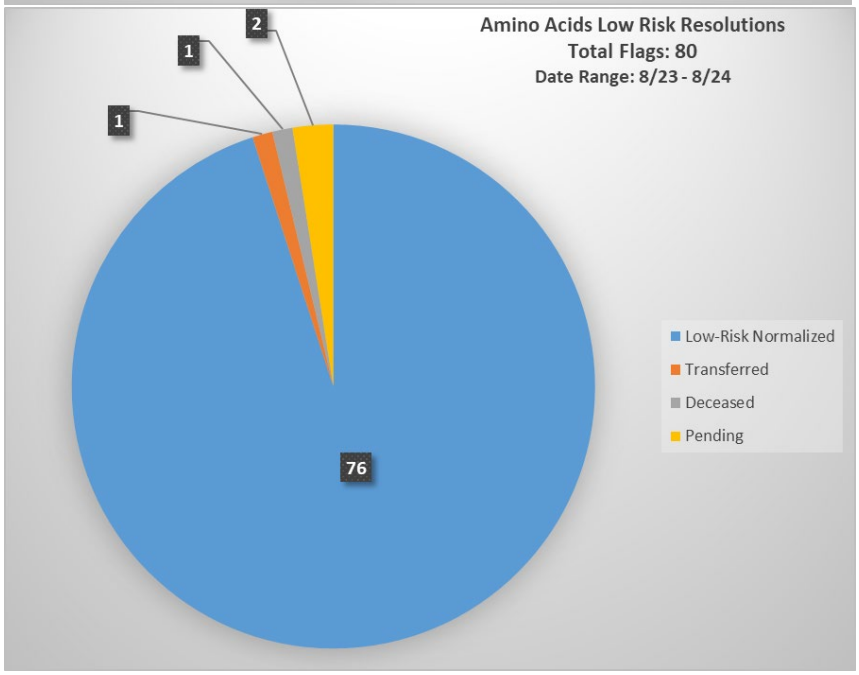




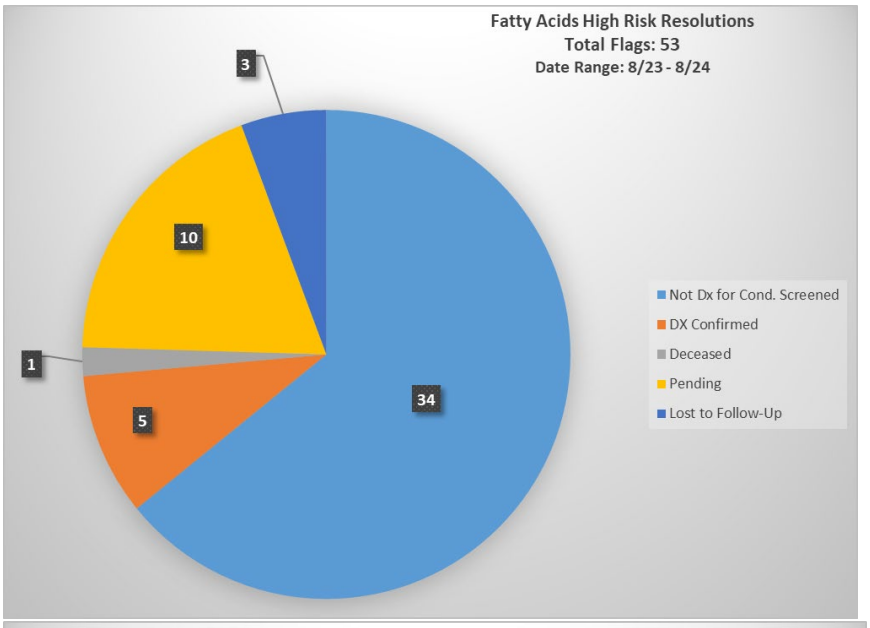


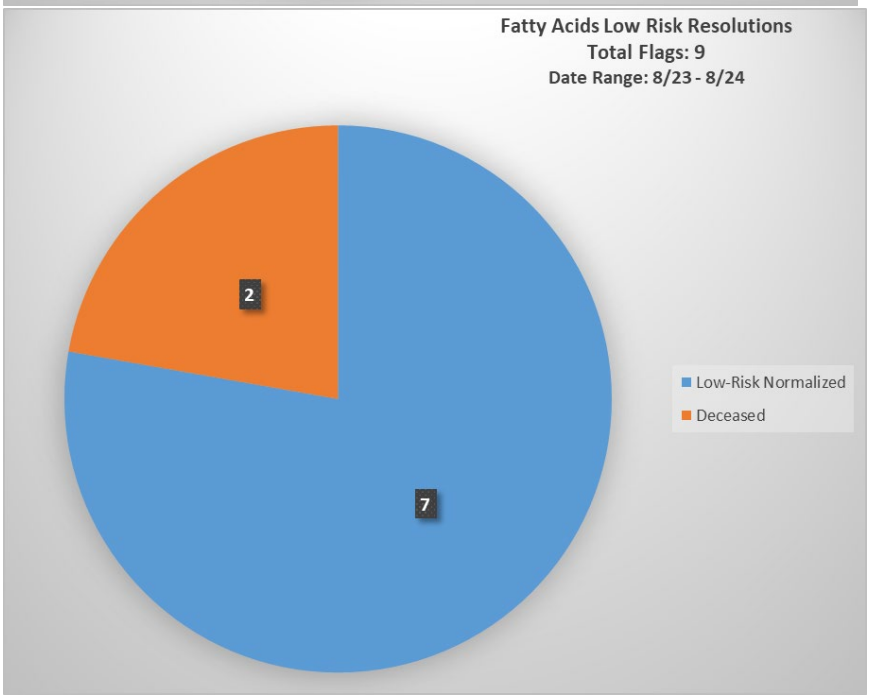
- Conditions: ASA, CIT, PKU, MCY, MSUD, TYR 1

- 

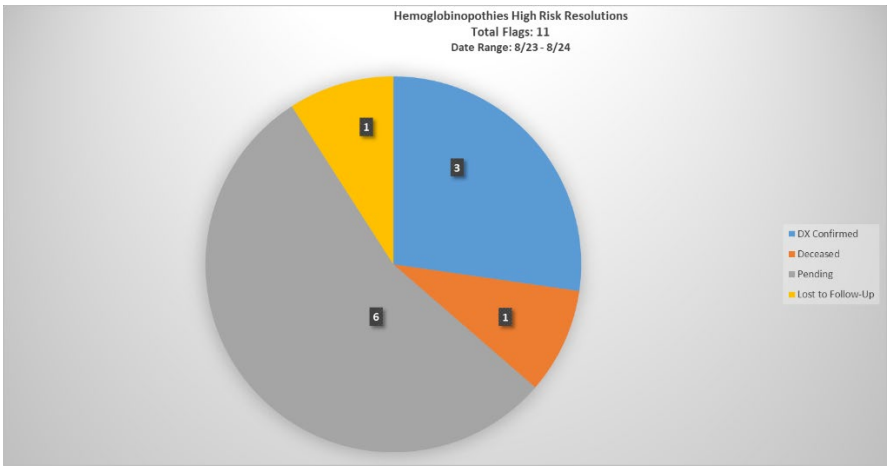
- 

- Condition: CUD, LCHAD, MCAD, TFP, VLCAD

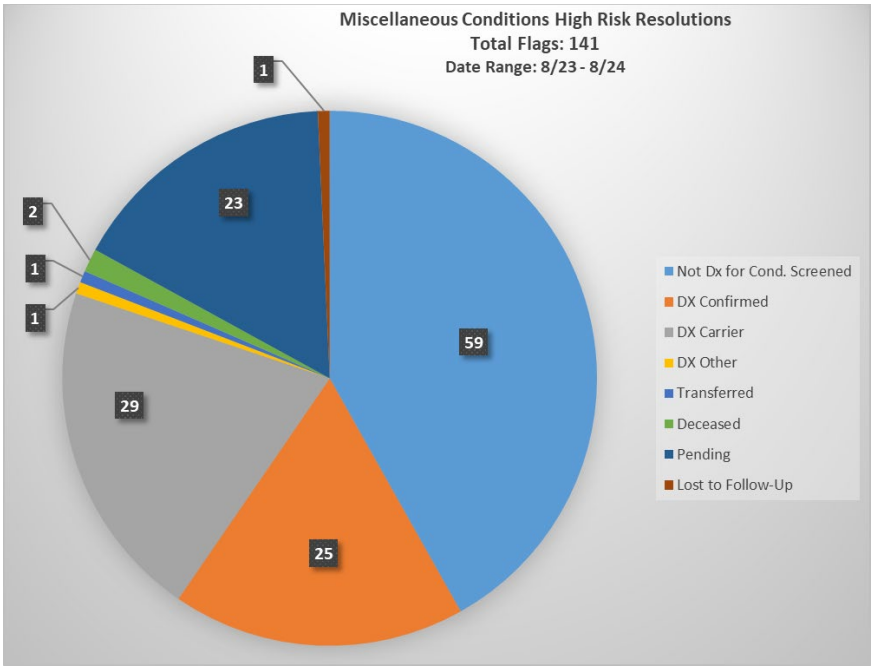


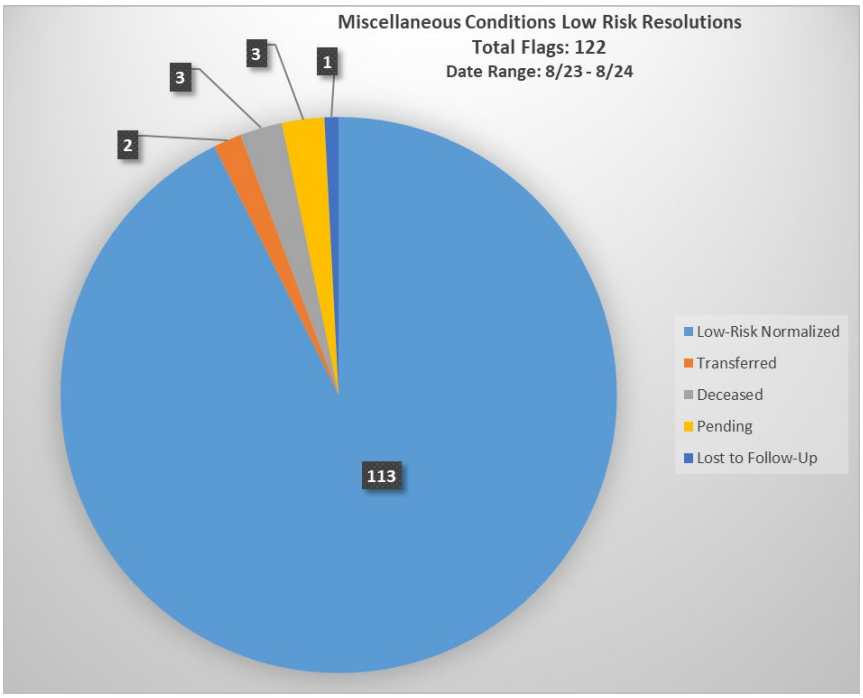


- Condition: Beta-Thalassemia, Hb S/C, Hb SS

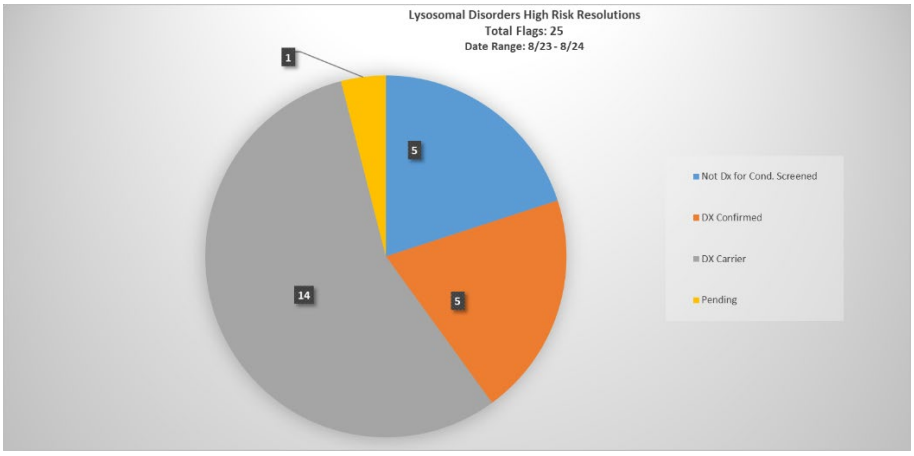


- Condition: Biot, CAH, CF, CH, GALT, SCID, SMA, X-ALD





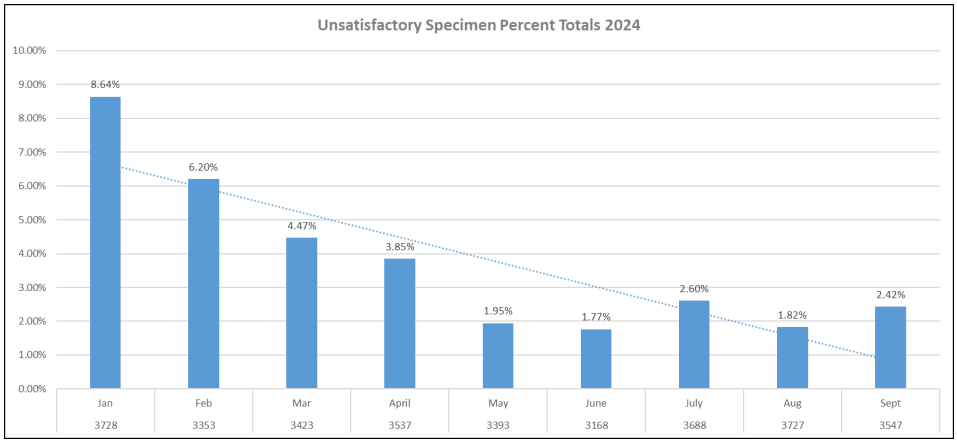
- Conditions: Pompe, MPS I, MPS II



- KHEL Updates
  - Updates on New Conditions
    - X-linked Adrenoleukodystrophy - Go Live February 1, 2024
    - Cutoff is 0.14 µmol/L for low risk/repeat and 0.20 µmol/L for high risk/refer
    - 1 Dx male (C26:0 LPC = 0.78975), 1 False Positive male (C26:0 LPC = 0.321), 4 pending
    - Since February 1, 2024, high risk/refer are 5 male, 1 female
    - Mucopolysaccharidosis II - Go Live was August 1, 2024
    - Cutoff is 0.20 µmol/L for low risk/repeat and 0.30 µmol/L for high risk/refer
    - No high risk/refer screened since August 1, 2024

- Krabbe
- ETA Go Live 4th Quarter 2025
- GAMT
- Method Development, ETA Go Live end of 2025/early 2026

- Recommended Uniform Screening Panel
  - The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) is meeting today, November 14, 2024.
  - Krabbe was approved for RUSP inclusion status on July 1, 2024. It is specifically stated as infantile Krabbe disease (Low galactocerebrosidase, or GALC, and psychosine ≥ 10 nM).
  - Duchenne's Muscular Dystrophy – unanimous vote to postpone decision on May 10, 2024.
  - The Committee is seeking additional information. The decision will be revisited no later than May 10, 2025.
  - cCMV has been returned with request for more data and encouraged to resubmit nomination package.
  - Nomination packages currently under consideration:
  - Metachromatic Leukodystrophy (MLD)
  - Biliary Atresia (BA)
- Unsatisfactory Specimens 2024



  - New Laboratory
    - Official opening date will be Spring 2025
    - Lab will be moving the majority of the 1st quarter of 2025

- Conversation was had about creating framework on how to determine what order to bring on new conditions and to what relevance the RUSP plays in the decision.
- Drew brought up the new work with Maternial Child health early initiatives.
- Guest Presentation: Dr. Joanne Kurtzberg, Krabbe Disease
- New Condition Nomination

- Michelle Leeker made a motion to start piloting Krabbe and GAMT while bringing on the condition to the Kansas panel after a successful pilot.
    - Dr. Lewis seconded the motion.
    - We do not have a quorum on the call today and the vote will have to be done by email.
- Karen asked if the cap needed to be revisited if we continue to add conditions to the Kansas Newborn Screening program.
    - Drew stated that budget is always a concern, and we continue to monitor the budget and advocate for increased funding.
- Dr. Gannon suggested that maybe a white paper would be the most appropriate first step for bringing on a new condition.

- Comments, Discussion, or new business
- Next meeting date
    - Tentatively April.May
        - In person in our brand-new Lab
        - Will send out a doodle pool for specific dates.