# KANSAS NEWBORN SCREENING ADVISORY COUNCIL MEETING

## ONLINE VIA ZOOM MEETING

# MINUTES

## JANUARY 20, 2022

**Members Present**

Emily Barr, Dr. Jennifer Gannon, Dr. Carolina Beltran, Dr. Grace Brouillette, Julie Wellner, Dr. Kourtney Bettinger, Dr. Merlin Butler, Dr. Laurie Gwyn, Michelle Leeker, Dr. Brittan Zuccarelli, Shobana Kubendran, Dr. Mike Lewis, Karey Padding

**Members Absent**

Dr. Selina Gierer, Dr. Thomas Loew

**KDHE Staff Present**

Michelle Black, Drew Duncan, Kinsey Anderson, Michelle Mills, Marilee Lowrey, Connie Neuhofel, Shane Morris, Liz Schardine, Phillip Adam, Patrick Hopkins

**Others Present**

Randi Gadea, Dr. Bryce Heese, Hari Patel, Charlotte Buchanan, Gail Webster, Jake Ginter, Kayzy Bigler, Dennis Dobson, Michelle Knoll, Illana Chilton, Meghan Strenk, Kristin Finn, Karen Braman

**ACITON ITEMS**:

1) Have HgB materials reviewed by families and council members.

**Meeting Recording:**

*Link*: https://us02web.zoom.us/rec/share/xZkK6I7Ca0TflcCstyahR9BPgZtlIj8jnzIELz5odbS4OlA_MHJxnvsWxJDc9zev.PFN7ScnxfgvsZpi2

*Access Passcode*: KSNBSAC2022!

**Minutes**

1. Housekeeping
    a. Attendees answered icebreaker question in the chat: what was the best thing that happened to you in 2021?
    b. Approval of previous minutes (with no edits): Dr. Gwyn moved to approved and Dr. Bettinger seconded.
2. Follow Up Updates
    a. Legislation/Regulations
        i. Legislative session is starting up again
        ii. Regulations revisions have begun
        iii. Karey Padding is willing to testify again for the program
    b. Data system review
        i. Third party review of our combined program data systems.
    c. Interagency Agreement
        i. Formal agreement of collaboration between KHEL and KDHE
        ii. Programs worked closely to outline appendices
    d. Staffing changes:
        i. BFH Reorganization (BFH reorg 2.0)
        ii. F/U staffing- functioning without an admin and a data manager since May of 2021. Added a support specialist.
            1. Posted for a data manager and a full-time admin.
        iii. Lab staffing- They have one open position. Working with the staff they have as staff are being pulled to work COVID.
3. 2021 Continuous Quality Improvement Highlights.
    a. Abnormal Notification system improvements (Ilana Chilton is willing to assist in this effort)
    b. NBS Office hours
    c. Upgraded instrumentation for CAH CH and CF
    d. Shared program one drive
    e. Paperless case management workflow.
4. 2021 KS NBS Presentations and honors
    a. 2 Presentations
    b. 2 Posters
    c. Recognition
        i. Best Poster award winner Michelle Mills tied with MO, CA and Baebies, Inc.
        ii. Everyday Life Saver Award Nomination- Kinsey Anderson
    d. Invited to present in a panel discussion in the 3rd session
    e. *"Using CQI to Improve Newborn Screening Timeliness"*
    f. Invited to present an overview of our implementation of DMF screening for Pompe and MPS-I
5. Data Discussion
    a. Collection Age
        i. Steady decrease in collection time through 2019 and early 2020
        ii. Follow up experienced staffing transition in September 2020

1. Reduced education activities between September 2020 and February 2021

iii. 2021 overall trending down with education activities:
1. Increased frequency of webinars
2. Monthly Newsletters Reminders

iv. Temporary increase during fall 2021 COVID-19 surge

b. Transit Time
i. 2019 68.6 Average hours 2.9 days
ii. 2020 62.4 Average hours 2.6 days
iii. 2021 60.9 Average hours 2.5 days
iv. Activities for Improvement
1. Targeted facility education & Timeliness Tips poster
2. APHL Sunday Courier Project began August 2020
v. Impactful Events
1. COVID-19 shut downs and staffing shortages
2. Seasonal shipping delays with winter weather

c. Time from Receipt to Report
i. Decrease in early 2019 likely from laboratory protocol change – same-day confirmations for flagged specimens
ii. Winter weather causes shipping delays – samples received after laboratory cutoff times
1. Adds up to 24 hours of additional time to report due to specimens being run the day after receipt
iii. Slight increase in 2021 due to addition of Lysosomal Storage Disorders
1. September to November impacted by delays in LSD 2nd tier screening results

d. Time from Birth to Report
i. Decrease in early 2019 likely from laboratory protocol change – same-day confirmations for flagged specimens
ii. Winter weather causes shipping delays – samples received after laboratory cutoff times
1. Adds up to 24 hours of additional time to report due to specimens being run the day after receipt
iii. Slight increase in 2021
1. due to addition of Lysosomal Storage Disorders
2. September to November impacted by delays in LSD 2nd tier screening results

e. Percent of Initial Specimens Reported in Given Timeframes
i. Overall, percent of initial specimens reported in given timeframes is relatively stable
ii. Changes between 2020 & 2021 are likely related to:
1. The increased collection age in early 2021
2. Transit delays in early 2021

f.

| | | Initial Samples Collected Between ≤ 48 Hours | | | | | Initial Samples Collected > 48 Hours | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | *Total Initial Samples* | *Total ≤ 48h* | *% of Initial* | *Reported in 8+ days* | *% of Initial* | *% of ≤ 48h* | *Total > 48h* | *% of Initial* | *Reported in 8+ days* | *% of Initial* | *% of > 48h* |
| **2019** | 37,270 | 34,876 | 93.6% | 5101 | 13.7% | 14.6% | 2,394 | 6.4% | 1328 | 3.6% | 55.5% |
| **2020** | 36,473 | 34,398 | 94.3% | 3533 | 9.7% | 10.3% | 2,075 | 5.7% | 1222 | 3.4% | 58.9% |
| **2021** | 33,243 | 30,713 | 92.4% | 3890 | 11.7% | 12.7% | 2,530 | 7.6% | 1811 | 5.4% | 71.6% |

g.

| | All Samples Collected ≤ 48 Hours | All Samples Collected > 48 Hours |
|---|---|---|
| **2019** | 40.2 Hours (1.7 Days) | 42.0 Hours (1.8 Days) |
| **2020** | 38.2 Hours (1.6 Days) | 39.2 Hours (1.6 Days) |
| **2021** | 43.5 Hours (1.8 Days) | 45.8 Hours (1.9 Days) |

h. Follow Up Timeliness
    i. Total number of initial calls made 449
        1. Goal 2 hours; 2021 Average 2.3
    ii. Percent of calls made in Given Timeframes

1.

| Time Frame | Percent |
|---|---|
| 1 hour | 65.7% |
| 2 hours | 22.5% |
| 2+ hours | 10.7% |

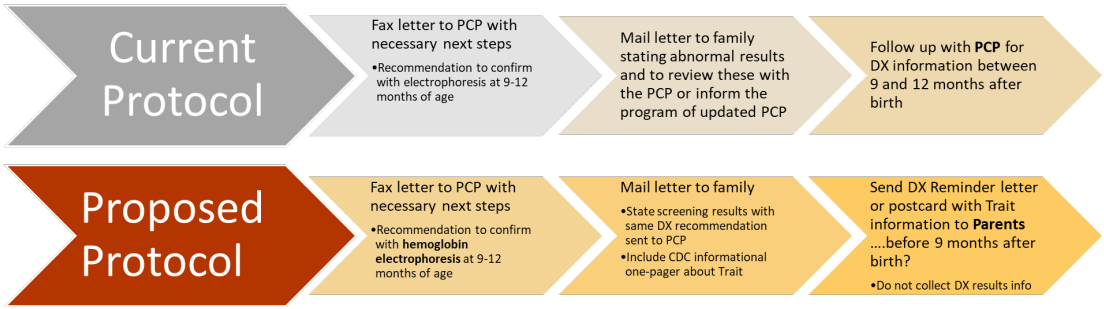
i. Unsatisfactory Specimens –
    i. 2020 Trendline was trending down and in 2021 we saw a trendline that continued to climb as facilities face staffing issues.
    ii. Goal is 1.3% and our average is 5%

6. Follow up Contracts
    a. Medical Director
        i. Provide oversight and support for the follow up program
        ii. Need for generalist and public health perspective
            1. Pediatrician, Family Practice, Preventative Medicine Credential
            2. Public Health Experience
    b. Specialist Contracts
        i. Intention is for several contracts
            1. Metabolic/Genetics, Hematology, Endocrinology, Immunology
            2. Need clear idea of estimated number of calls expected for each (avg. # of high risk/refer results)
        ii. Purpose - PCP consultation after follow-up recommendation for referral
            1. NOT a state referral center model
    c. Considering: Follow-Up Support Contract
        i. Discussion highlights about potential follow-up support contract(clinical):
            1. Concern that a clinical follow up support contract would overlap or infringe on the specialist role of consulting with PCP and/or families
            2. Concern about specialist calling families directly without having been called by the PCP
            3. Kinsey clarified purpose is simply to ensure that family received screening result information from PCP and to direct family to additional,

high quality information, if family feels they didn't get enough or good quality information.
4. Concern that if we reach out to family without knowing what step of the diagnostic process they're in, it could create more concern and questions for the family
5. Dr. Beltran - If PCP is the one giving all information to the family, then PCP is the one that requires support. Biggest thing is to identify if PCP was able to follow the recommendations and identify the support that they need
6. Concerns with specialist capacity to be obligated to call families for every high risk/refer level result – hence idea for separate follow up contract to fulfill that function
7. Idea that PCPs can inform families who will be calling them to mitigate concern about cold calls to families from people they don't know

ii. Additional comments available in the chat log following the minutes
iii. Further discussion was recommended and Dr. Bettinger, Dr. Lewis, Kirsten Finn, Gail Webster are interested in

7. Follow up Contracts Estimated Timeline
    a. Medical Director – Announcement in February 2022
    b. Specialist and Follow-Up Support Contracts
        i. March 2022 – Number of contracts determined, and position descriptions finalized
        ii. April 2022 – Begin recruitment
        iii. May 2022 – Begin contract negotiation and concurrence process
        iv. July 1, 2022 (SFY23) – Contract Start Date
8. HgB Trait Protocol Review
    a. What we've learned:
        i. HgB Trait results make up most of the program's Lost To Follow Up (LTU)
        ii. Many primary care providers either don't communicate these results to families at all or communicate that Trait is benign
        iii. In 2020 we had *at least* 353 infants with HgB Trait screening results, which represents:
            1. 1% of all infants screened in 2020
            2. 11% of all infants that we followed up on in 2020
    b. The program does not do anything meaningful with diagnostic information for HgB Trait and our energies may be better distributed to adding new conditions to our screening panel
    c. Why is a change needed?
        i. Better communication of HgB Trait results and implications to families
        ii. Redistribution of follow up team's capacity
    d. Key changes
        i. Screening results, disorder information, and reminder for DX testing sent directly to families
        ii. No more collection of diagnostic testing results




e.

f. There were discussions in regard to be way to inform families without alarming them.
    i. Making sure the letter is clear. Plain language. What is the parent supposed to do.
    ii. Discussion on this topic was recorded in the chat.

g. Meghan brought up that large populations that would flag for HgB do not read English so it could be beneficial to have in different language.

**h. Action items:**
    **i. Have HgB materials reviewed by families and council members.**
    **ii. Have HbB materials available in more than one language – avail. in most common languages for populations most likely to be affected**

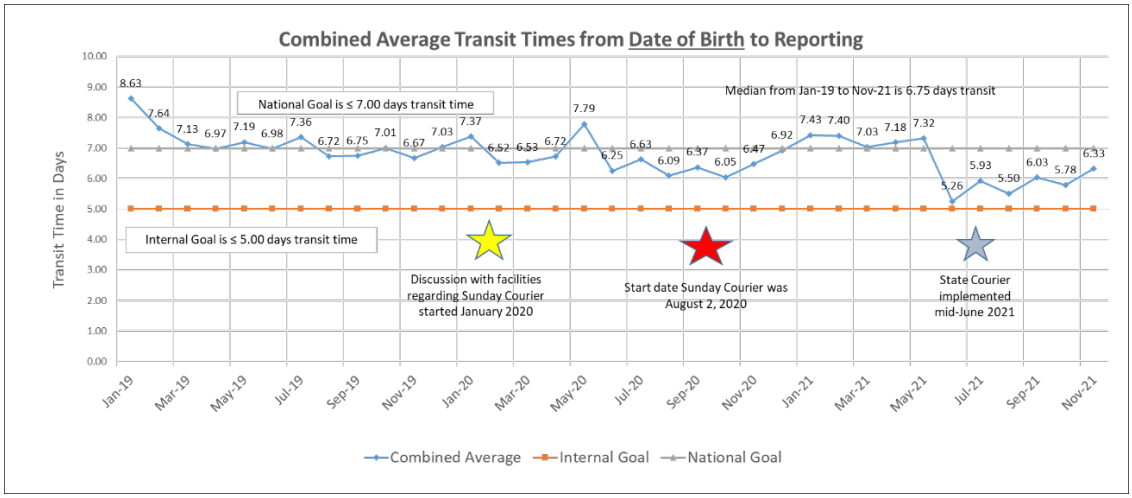
9. Special Health Care Needs (SHCN) program
    a. Program Purpose; To promote the functional skills of persons, who have or are at risk for, a disability or chronic disease.
    b. Target Population-
        i. State Statute -KSA 6-5a01
            1. A child with special health care needs" means a person under 21 years of age who has an organic disease, defect or condition which may hinder the achievement of normal physical growth and development
        ii. Maternal and Child Health Bureau
            1. "Children and youth with special health care needs (CYSHCN) are those who have, or are at risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally"
    c. SHCN Core Functions – Services provided. Provides specialized medical services to: Ages 0-21 with eligible medical conditions.
        i. *Diagnostic – evaluation and related testing (not for second opinions)
        ii. Direct Assistance Programs (DAP'S) – Client chosen funding assistance
        iii. Care Coordination – development of an Action Plan and assistance with community resources
        iv. Special Bequest – services otherwise not covered to improve quality of life (ex: mobility items, assistive technology, non-clinical/indirect services, etc).
        v. *Family System Navigation Trainings – Free one day training for families who have a child or loved one with any disability or SHCN
        vi. *Supporting You (SHCN) - A peer to peer mentoring program for families who have loved ones with special health care needs/disability

vii. * Open to all families who have a loved one with a special health care need

d. SHCN Eligible Conditions Spina Bifida, Cleft Lip/Cleft Palate, Acquired or congenital health disease, Burns requiring surgical intervention, Orthopedic Conditions*, Limited gastrointestinal or genitourinary conditions requiring surgery, Hearing Loss, Vision disorders (limited) Craniofacial anomalies (select), seizures- outpatient care and prescriptions only, Juvenile Rheumatoid Arthritis, Genetic and Metabolic Conditions.

e. SHCN Satellite Offices -SFY 2022 Map shown in Power Point

10. KHEL updates
    a. Newborn Screening – staff turnover
    b. New instrumentation for Congenital Adrenal Hyperplasia, Congenital Hypothyroidism and Cystic Fibrosis is live, as of December 13, 2021
    c. Proposing age related cutoffs for Congenital Hypothyroidism
11. Covid 19 and Newborn Screening
    a. NBS Staff are now assisting with COVID daily
    b. COVID staff who had previously been assisting in NBS have been pulled back to COVID
12. Sunday Courier Grant
    a.



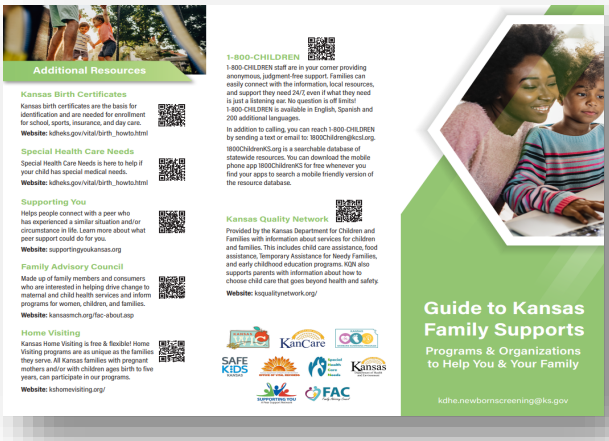

13. X-ALD
    a. Have instrumentation and will continue to move forward with timeline.
14. Education and Outreach Updates
    a. Facility Recognition Award
        i. 2020 Annual Awards announced on December 2nd
        ii. 3 Best of Best Facilities; Newman Regional Health, Hutchinson Regional Medical Center and Sabetha Community Hospital Inc.
        iii. 2021 – discussion begun on additions to current version
            1. i.e. success stories; tips and tricks for success section
    b. KDHE Website
        i. Soft launched new website in December
        ii. Several issues identified, being documented and tracked for resolution
        iii. If visiting website, please let us know of missing information, non-functional links, other issues: email shane.morris@ks.gov
    c. Online Training Webinars (3rd and 4th Quarter of 2021)

i.

| | Newsletter | Calendar | Email | Webinar Name |
|---|---|---|---|---|
| Total Registrations | 8 | 14 | 3 | Blood Spot Collection Technique |
| Total Registrations | 4 | 3 | 5 | NBS Basics and Metabolic Review |
| Total Visitors (clicks) | 2314 | 24 | 57 | Blood Spot Collection Technique |
| Total Visitors (clicks) | 235 | 12 | 35 | NBS Basics and Metabolic Review |

1. Totals:
    a. Blood Spot Collection = 25
    b. NBS Basics = 12

d. Combo Brochure Progress

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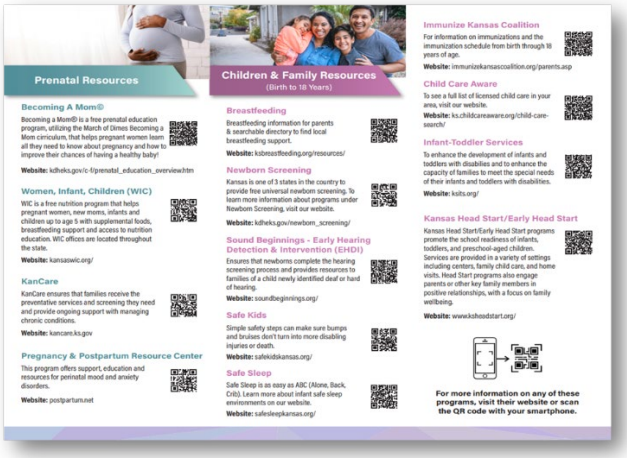
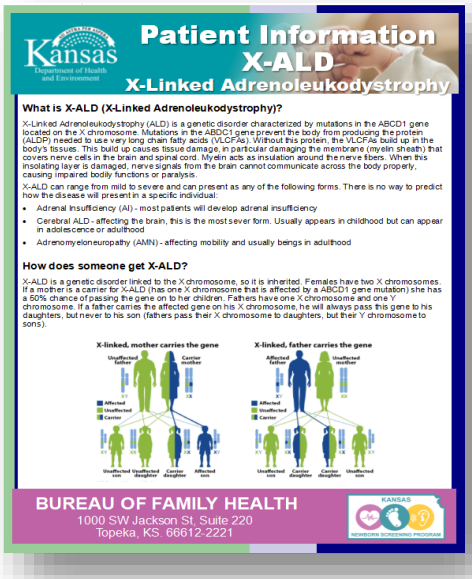



ii.

e. X-ALD Info Briefs



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# Patient Information
# X-ALD
## X-Linked Adrenoleukodystrophy

**What is X-ALD (X-Linked Adrenoleukodystrophy)?**

X-Linked Adrenoleukodystrophy (ALD) is a genetic disorder characterized by mutations in the ABCD1 gene located on the X chromosome. Mutations in the ABDC1 gene prevent the body from producing the protein (ALDP) needed to use very long chain fatty acids (VLCFAs). Without this protein, the VLCFAs build up in the body's tissues. This build up causes tissue damage, in particular damaging the membrane (myelin sheath) that covers nerve cells in the brain and spinal cord. Myelin acts as insulation around the nerve fibers. When this insulating layer is damaged, nerve signals from the brain cannot communicate across the body properly, causing impaired bodily functions or paralysis.

X-ALD can range from mild to severe and can present as any of the following forms. There is no way to predict how the disease will present in a specific individual:

- Adrenal Insufficiency (AI) - most patients will develop adrenal insufficiency
- Cerebral ALD - affecting the brain, this is the most sever form. Usually appears in childhood but can appear in adolescence or adulthood
- Adrenomyeloneuropathy (AMN) - affecting mobility and usually beings in adulthood

**How does someone get X-ALD?**

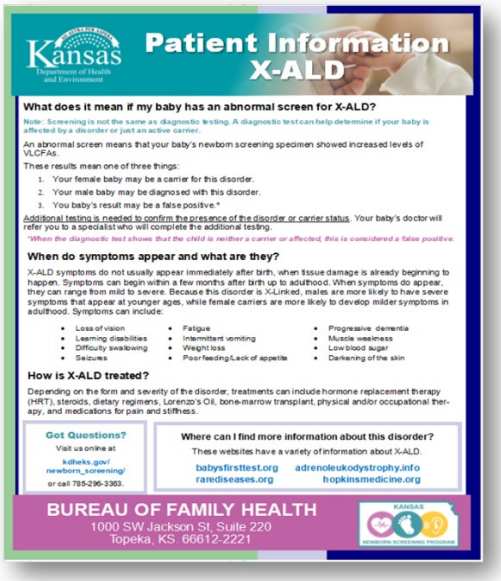
X-ALD is a genetic disorder linked to the X chromosome, so it is inherited. Females have two X chromosomes. If a mother is a carrier for X-ALD (has one X chromosome that is affected by a ABCD1 gene mutation) she has a 50% chance of passing the gene on to her children. Fathers have one X chromosome and one Y chromosome. If a father carries the affected gene on his X chromosome, he will always pass this gene to his daughters, but never to his son (fathers pass their X chromosome to daughters, but their Y chromosome to sons).




**BUREAU OF FAMILY HEALTH**
1000 SW Jackson St, Suite 220
Topeka, KS. 66612-2221

KANSAS NEWBORN SCREENING PROGRAM

i.



Kansas Department of Health and Environment

# Patient Information
# X-ALD

**What does it mean if my baby has an abnormal screen for X-ALD?**

Note: Screening is not the same as diagnostic testing. A diagnostic test can help determine if your baby is affected by a disorder or just an active carrier.

An abnormal screen means that your baby's newborn screening specimen showed increased levels of VLCFAs.

These results mean one of three things:

1. Your female baby may be a carrier for this disorder.
2. Your male baby may be diagnosed with this disorder.
3. You baby's result may be a false positive.*

Additional testing is needed to confirm the presence of the disorder or carrier status. Your baby's doctor will refer you to a specialist who will complete the additional testing.

*When the diagnostic test shows that the child is neither a carrier or affected, this is considered a false positive.

**When do symptoms appear and what are they?**

X-ALD symptoms do not usually appear immediately after birth, when tissue damage is already beginning to happen. Symptoms can begin within a few months after birth up to adulthood. When symptoms do appear, they can range from mild to severe. Because this disorder is X-Linked, males are more likely to have severe symptoms that appear at younger ages, while female carriers are more likely to develop milder symptoms in adulthood. Symptoms can include:

- Loss of vision
- Learning disabilities
- Difficulty swallowing
- Seizures
- Fatigue
- Intermittent vomiting
- Weight loss
- Poor feeding/Lack of appetite
- Progressive dementia
- Muscle weakness
- Low blood sugar
- Darkening of the skin

**How is X-ALD treated?**

Depending on the form and severity of the disorder, treatments can include hormone replacement therapy (HRT), steroids, dietary regimens, Lorenzo's Oil, bone-marrow transplant, physical and/or occupational therapy, and medications for pain and stiffness.

**Got Questions?**
Visit us online at
kdheks.gov/newborn_screening/
or call 785-296-3363.

**Where can I find more information about this disorder?**
These websites have a variety of information about X-ALD.

babysfirsttest.org
adrenoleukodystrophy.info
rarediseases.org
hopkinsmedicine.org

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KANSAS NEWBORN SCREENING PROGRAM

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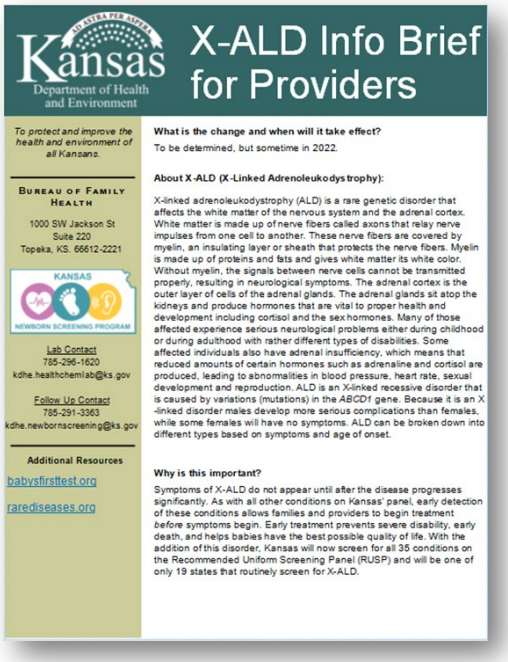

Kansas Department of Health and Environment

# X-ALD Info Brief for Providers

*To protect and improve the health and environment of all Kansans.*

**Bureau of Family Health**

1000 SW Jackson St
Suite 220
Topeka, KS. 66612-2221

KANSAS NEWBORN SCREENING PROGRAM

Lab Contact
785-296-1620
kdhe.healthchemlab@ks.gov

Follow Up Contact
785-291-3363
kdhe.newbornscreening@ks.gov

**Additional Resources**

babysfirsttest.org

rarediseases.org

**What is the change and when will it take effect?**

To be determined, but sometime in 2022.

**About X-ALD (X-Linked Adrenoleukodystrophy):**

X-linked adrenoleukodystrophy (ALD) is a rare genetic disorder that affects the white matter of the nervous system and the adrenal cortex. White matter is made up of nerve fibers called axons that relay nerve impulses from one cell to another. These nerve fibers are covered by myelin, an insulating layer or sheath that protects the nerve fibers. Myelin is made up of proteins and fats and gives white matter its white color. Without myelin, the signals between nerve cells cannot be transmitted properly, resulting in neurological symptoms. The adrenal cortex is the outer layer of cells of the adrenal glands. The adrenal glands sit atop the kidneys and produce hormones that are vital to proper health and development including cortisol and the sex hormones. Many of those affected experience serious neurological problems either during childhood or during adulthood with rather different types of disabilities. Some affected individuals also have adrenal insufficiency, which means that reduced amounts of certain hormones such as adrenaline and cortisol are produced, leading to abnormalities in blood pressure, heart rate, sexual development and reproduction. ALD is an X-linked recessive disorder that is caused by variations (mutations) in the *ABCD1* gene. Because it is an X-linked disorder males develop more serious complications than females, while some females will have no symptoms. ALD can be broken down into different types based on symptoms and age of onset.

**Why is this important?**

Symptoms of X-ALD do not appear until after the disease progresses significantly. As with all other conditions on Kansas' panel, early detection of these conditions allows families and providers to begin treatment *before* symptoms begin. Early treatment prevents severe disability, early death, and helps babies have the best possible quality of life. With the addition of this disorder, Kansas will now screen for all 35 conditions on the Recommended Uniform Screening Panel (RUSP) and will be one of only 19 states that routinely screen for X-ALD.

iii.

f. Continuing Outreach
    i. Collaboration & Outreach Efforts
    ii. Lending support to our SHCN partners with development of "post-card"
    iii. 2021 outreach plan includes making contact with 3 facilities/birthing providers each week
    iv. Development of CCHD resource for families is in progress

15. Subcommittee Updates
    a. X-ALD Subcommittee Updates
        i. Updates to projected timeline – projected go live 3rd quarter 2022
        ii. New instrumentation installed
        iii. Lab working on validation process and hiring and training staff
        iv. Follow up letters and provider resources in their second draft
        v. Data system preparations for both lab and follow up in process
16. Comments and Discussions
    a. Shobana commented that she really appreciated having slides ahead of time for review.
17. Contracts for medical specialist
    a. Committee continued discussions on what are options are for having a specialist being able to assist the follow up program.
    b. We discussed benefits and concerns of the burden it would provide to the system.
    c. Benefit would be that this model would defray the cost that specialist are using dealing with NBS.
    d. Clarified when parents are getting letters and how they are followed up.
    e. Discussed when we close as lost to follow up and subsets of lost to follow up.
18. Dr. Bettinger motioned to adjourn and Dr. Gywn seconded the motion.

# Kansas Newborn Screening Advisory Council Chat Logs - Jan 2022

## Contracts Discussion

Kirsten Finn:

> I know within the ALD community, as an example, in other states many families have not had adequate information and it was quite stressful to them in many instances.

Michelle J. Mills-KS NBS Laboratory Manager:

> This is good feedback, Kirsten. We want to assist families.

Gail Webster Blessed Event Birth Services, CPM:

> As a midwife, I see my clients being uncomfortable and concerned about being contacted by providers they are not familiar with or have not initiated the contact.

Shobana Kubendran:

> Can follow up support contract be rolled into the contract with specialist? The "follow up nurse" will work with the specialist recommendation for follow up

Gail Webster Blessed Event Birth Services, CPM:

> If I have an abnormal screening that requires follow up, I think it would be advantageous that I make initial contact and give my client the name of providers who would be contacting them for additional follow up.

Michael Lewis:

> I think Shobana might be onto the right answer. The volume of times families are told 'this is usually a false positive' for CF testing with positive CFTR mutations are identified is high so they aren't feeling the need to come for a sweat chloride. As we know, the DNA isn't changing no matter how many times the newborn screening is done.

Randi Gadea:

> What if the families had a number they could call if they had additional questions? This would let them seek out the amount of information that they want.

Karey Padding, LMSW - Parent Advocate - ED of The Treehouse:

> As a parent, if something is time sensitive and I need to act, I want to have clear and concise information and instruction.

Carolina Beltran: Just wanted to comment- Jen, we do same - we contact PCP before calling patients parents

# HGB Trait Protocol Discussion

Julie Wellner RN KU Wichita Genetics: Who is interpreting the results?

Julie Wellner RN KU Wichita Genetics: The follow up labs?

Julie Wellner RN KU Wichita Genetics: SO the specialist are consulted for these cases?

Kirsten Finn:

I think as long as it is confirmed the conversation takes place before a letter is received.

Kourtney Bettinger, pediatrician, KU-KC:

I like empowering families with the one-page CDC info sheet (assuming it's written in a format understandable to families)

Michelle Leeker:

As a parent, I like the new protocol, would recommend reminder letter 5-6 months.

Julie Wellner RN KU Wichita Genetics:

You are linked to vital statistics? Do they capture language

Julie Wellner RN KU Wichita Genetics:

You may be able to look at that before your send a letter

Shane Morris, KDHE, Educator:

There is a section on the birth certificate that asks for primary language spoken in home. It is sometimes blank because it's not a required field.