NEWBORN SCREENING FAMILY OUTCOMES

Listening Sessions

2024

A Summary of the Family Listening Sessions held by the Mountain States Regional Genetics Network (MSRGN)

INTRODUCTION TO THE PROJECT

Introduction

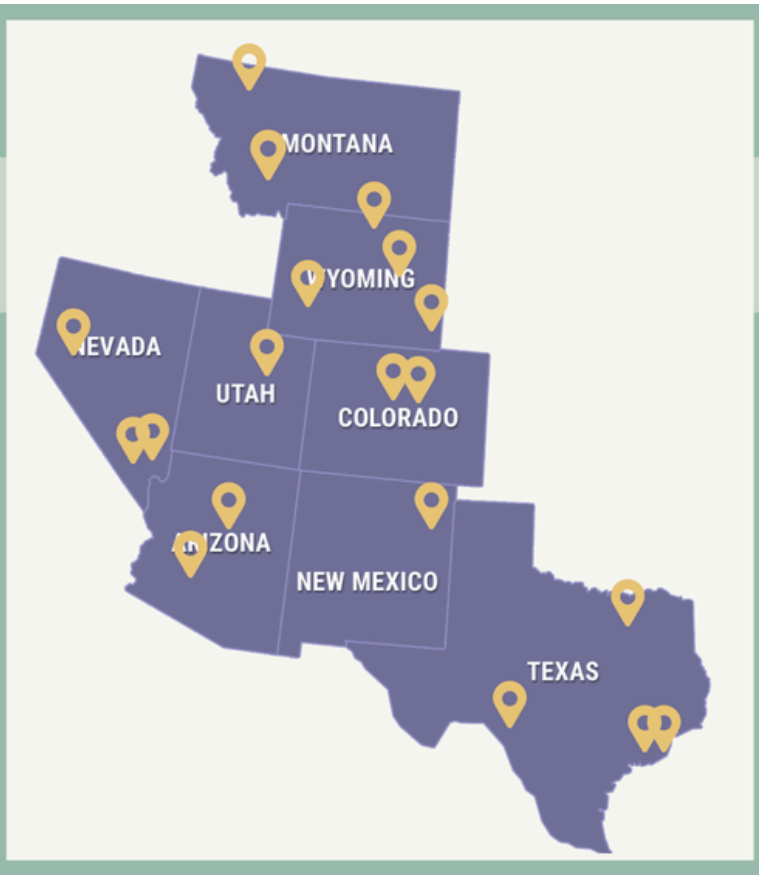
FAMILY OUTCOMES OF NEWBORN SCREENING

In August of 2023, HRSA reached out to Mountain States Regional Genetics Network (MSRGN) for assistance with a family outcome newborn screening project. Supplemental funding was provided to MSRGN and each Regional Genetics Network (7) to support engagement of family leaders, especially those from underserved populations. The goal was to provide meaningful input on measures of newborn screening family engagement and family outcomes for use in an instrument (to be created by RTI) to assess outcomes of families with children identified through NBS. This project was to be done in partnership with RTI, the NBS Excel awardee for the family outcomes instrument. After two national coordinating calls with RTI, HRSA and all the RGN representatives (October 2023 and December 2023), MSRGN had 3 additional calls with RTI staff while the plans for the MSRGN regional activities for this project were finalized.

MSRGN management team approved the proposal of two family listening sessions around the topic of family outcomes from newborn screening. The family recruitment goal for the listening sessions was at least two families per state in the MSRGN region (8 states: AZ, CO, MT, NV, NM, TX, UT, WY) who had experienced receiving a positive newborn screen result for a blood spot condition for their child or loved one. A flyer about the sessions was developed and approved by HRSA for circulation and distribution beginning in early January 2024. The two listening sessions were held March 11 and March 15, 2024 with additional one-on-one sessions held throughout March and April 2024.

WHERE ARE THE FAMILIES FROM?

Nineteen (19) families were recruited and attended one of the listening sessions or met one-on-one with the MSRGN Project Manager to provide input on family outcomes. At least one family from each of the 8 mountain states was engaged in the listening sessions or one on one calls.



The MSRGN Region

- Where Families are from
  - *2 Arizona*
  - *2 Colorado*
  - *2 Montana*
  - *3 Nevada*
  - *1 New Mexico*
  - *4 Texas*
  - *1 Utah*
  - *4 Wyoming*

Zip Codes from Medically Underserved Areas (MUA): 13/19 or 68.4%

Wyoming Representation

In the MSRGN region, Wyoming represents a state with no practicing genetic providers in the state. Genetic services for families with a positive newborn screen are contracted through the department of health with providers in other states (currently Colorado). Families often times travel to neighboring states for genetic services as well.
Due to this genetic unreservedness of the entire state of Wyoming, having 4 families represent the Wyoming experience around a positive newborn screening was a unique and important perspective captured in these listening sessions.

False Positives, Borderline Results, and Missed Diagnoses

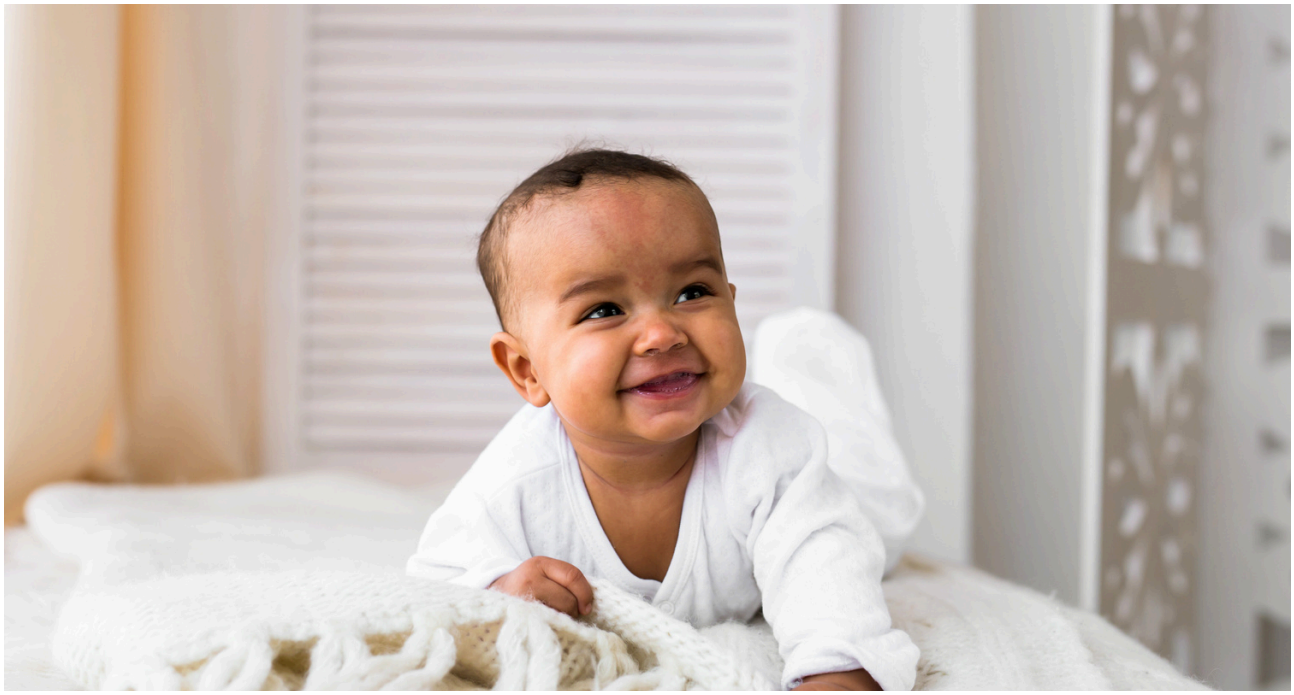
The recruiting flyer for this event identified that MSRGN was looking for families from our 8-state region who had received a positive newborn screen (by blood spot screening) for their child or loved one. Throughout the recruiting period, a few families expressed interest who had experiences with NBS results which involved missed diagnoses, borderline results and false positives. It was determined by the MSRGN management team to include these individual's perspectives as their experiences showed the broad spectrum of family outcomes which confront the NBS system.

NEWBORN SCREENING CONDITIONS REPRESENTED

Families who had received a positive newborn screen by bloodspot screening (excluding hearing loss and congenital heart screening) from the Mountain States regions were encouraged to participate. Ten different conditions were represented with the greatest representation from families with a phenylketonuria (PKU) diagnosis (7 families) and adrenoleukodystrophy (ALD and XALD) diagnosis (4 families). All conditions represented are listed in the graphic below.

NBS Conditions

- 7 Phenylketonuria (PKU)
- 4 X-linked Adrenoleukodystrophy (XALD & ALD)
- 2 Homocystinuria (HCU)
  - 1 caught by NBS
  - 1 missed by NBS
- Severe Combined Immunodeficiency (SCID)
- Maple syrup urine disease
- Sickle cell disease
- Galactosemia
- Congenital Adrenal Hyperplasia
- False Positive very long chain acyl-coa dehydrogenase deficiency



WHAT DID YOU CONSIDER MOST IMPORTANT FOR YOUR FAMILY FOLLOWING A NBS DIAGNOSIS?

Question choices were collected by asking this open-ended question:"What do you feel are the top 3 family outcomes of newborn screening for your family?" on the interest form filled out by families prior to the listening session. Families were then asked during the listening session on Mentimeter to give each option a score from 0 to 5.

On a 5 point scale the "Child Health Status" option yielded the highest combined score of a 4.9

Child Health Status
4.9

Monitoring & Management of NBS Condition
4.7

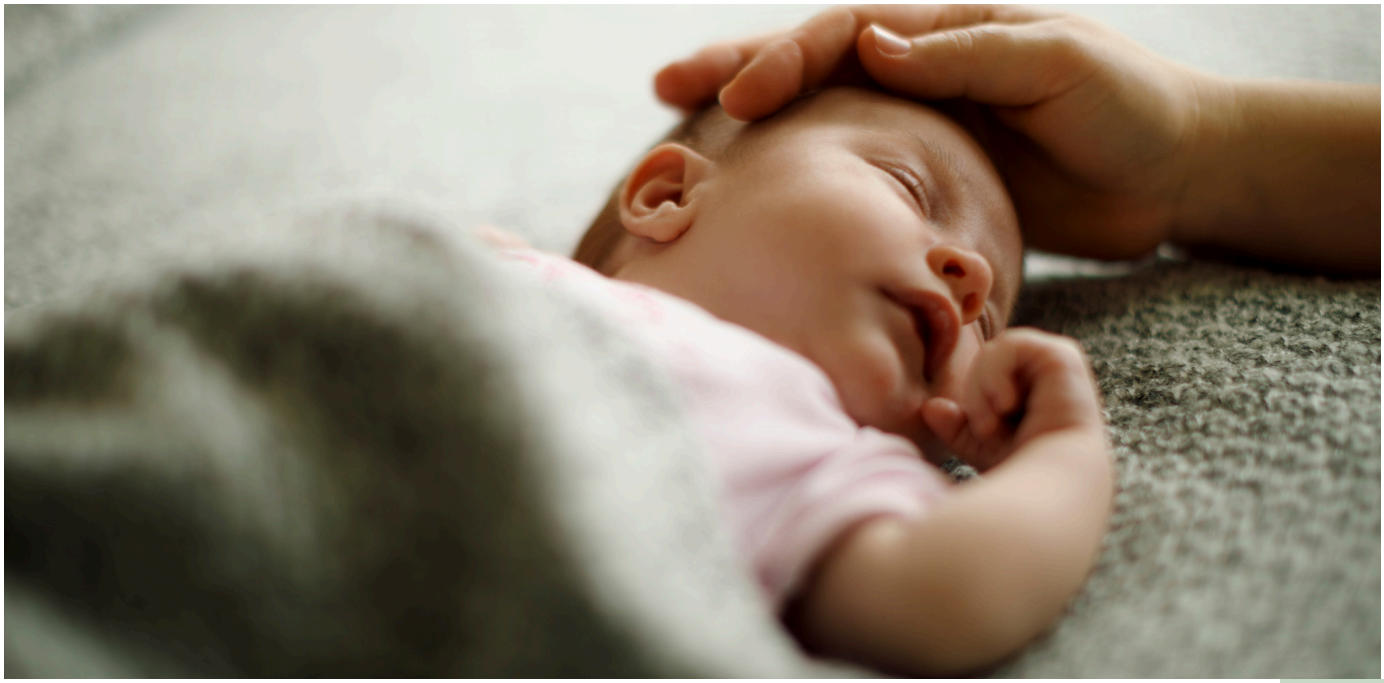
Education & Learning about NBS Condition
4.6

Child is Thriving
4.5

WHAT DID YOU CONSIDER MOST IMPORTANT FOR YOUR FAMILY FOLLOWING A NBS DIAGNOSIS?

As reported by families participating in the listening sessions, in their own words...
(via Mentimeter)

1. Early education.
2. Support and education.
3. We are able to be proactive to help him versus reactive which will lead to a better, healthier, life.
4. Resources and reassurance. Guidance with insurance and getting medical formula and food.
5. Making sure my child had the care he deserved and that we as a family advocated for our son. Education was extremely important to us. Making sure he lived a normal healthy life.
6. Education.
7. Most important for our family was finding out how to deal with the day to day of PKU, the changes in levels and having a better understanding that my child could live a healthy lifestyle still.
8. Having a treatment plan put into place immediately even if it could have been a fluke.
9. Informed education and timelines. We had multiple appts with genetics, nutrition and medical experts, very helpful.
10. To make sure we knew how to properly treat her diagnosis to keep her healthy and happy.
11. My son being alive.
12. Immediate referral.
13. Information and access and availability of a specialist for the diagnosis.
14. Ability to monitor and treat before life-threatening symptoms appear.
15. Positive treatment and that first phone call having someone with more knowledge about what your child has and how you will move forward.
16. Knowledge of the disease and the ability to proactively monitor before symptoms arise.
17. So many answers that changed our lives for the health of our child. We have an amazing group of people in our corner.
18. The most import part for our family after NBS is being connected to health resources to support the best outcomes for our child, being empowered with the knowledge of a diagnosis for better awareness.
19. Access to treatment.



HOW DO YOU DEFINE THRIVING FOR YOUR CHILD?

As reported by families participating in the listening sessions, in their own words...
(via Mentimeter)

1. Is able to lead a healthy (physically, mentally), inclusive life, able to do the things she wants to do in life.
2. Being able to be like a normal kid, going to school, birthday parties etc. also being able to ween of medications.
3. Child is happy and is living a fulfilling life, they are able to do whatever they want to do.
4. Child feels happy, safe, and loved. Less medical more fun!
5. Healthy, being successful with his treatment and doing the best he can in his condition.
6. My child is living a full life full of love and support for his condition.
7. Healthy emotionally , physically and mentally.
8. Thriving for my child looks like, being able to go to regular school with normal children, playing , dancing , running reading, not going to the ER or being admitted to the hospital for a year.
9. Participating in activities and school, personalty, make friends and memories.
10. Living a healthy life, with minimal major healthcare interferences. Without good health, life is very difficult.
11. Being able to do what other kids his age do, reasonably and is happy.
12. Most importantly happy and healthy. Able to get the life saving treatment that they need without having to jump through hoops.
13. Levels are consistently good, my child FEELS healthy. In my case, making sure my child can do normal things with treatment that other children can.
14. Happy and healthy! Having good levels.
15. Having good levels and able to maintain them. Meeting all milestone goals. Happy and loved!
16. The child having medical needs met through their treatment.
17. Feeling hope for the future, not feeling a victim to his disease.
18. Living at their full potential.

HOW DO YOU MEASURE IF YOUR CHILD IS THRIVING ON A DAILY BASIS?

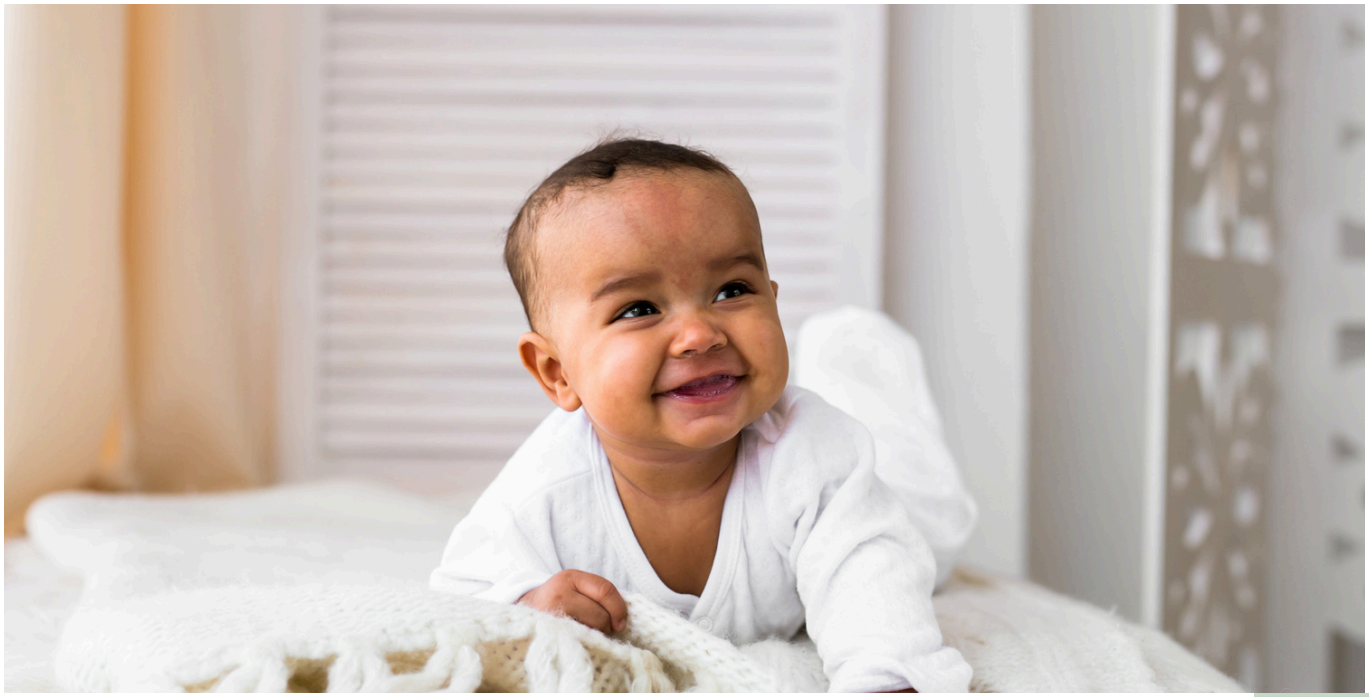
As reported by families participating in the listening sessions, in their own words...
(via Mentimeter)

1. IThat she is LIVING life, not just focused on treatment. Doing the things she wants to do.
2. How he's physically feeling and looking.
3. Their life is not that different from the neighbor kids who don't have their condition.
4. Daily health check (physical, emotional).
5. Emotionally stable and being happy in his choices.
6. He is able to live a "normal " life minus some extra doctor appointments.
7. How his attitude is for the day!
8. Her being able to go to school, not having SCD paid that requires medication or ER visits, drinking water daily without me telling her, having the strength to carry all her junk to her room.
9. Appropriate behavior/reactions which would suggest normal or high levels.
10. We're used to daily meds, dietary restrictions, etc. but the goal each day is to minimize missed activities and increase joy. When our kid is down due to health reasons, it's very apparent.
11. Being able to go to school like others because his medicine can be controlled and he can wake up and learn every day!
12. Smiling, laughing, feeling loved and important, levels are within range, feeling satisfied.
13. Cortisol and Aldosterone levels are within range. Not feeling moody (common side effect or hydrocortisone). And not missing out on things because of his condition.
14. They can keep up with their peers. Don't feel different. Are able to be successful with their diet, meds and formula.
15. My child goes to bed with full belly, a roof over their head, a loving and supporting family.
16. By their mental behavior.
17. Working towards and achieving his goals.
18. Healthy and happy- access to and taking their medical foods and medication.

WHAT ARE SOME FAMILY OUTCOMES FROM YOUR NBS JOURNEY THAT COULD BE IMPROVED FOR OTHER FAMILIES?

As reported by families participating in the listening sessions, in their own words...
(via Mentimeter)

1. Early education at time of positive result, not giving false hope, timeline of events to ease parents mind.
2. Physician training for delivery of initial diagnosis - very emotional and confusing time for families, followed by comprehensive and compassionate family education and support.
3. Resources when given the diagnosis rather than what the state provides. Most diseases have groups that provide the information families want.
4. How information is given (not via voicemail or nurse).
5. Don't give false hope, answer their questions and make them feel heard. Then help pave the way to clear education and resources.
6. The providers not giving false hope about it being a false positive.Giving information on the suspected condition. What possible concerns to look out for in your child before the other test comes back.
7. Better education for families before the test is even performed. More support along the journey.
8. If my childs diagnosis was put in more laymen's terms and not dr'ed up it would be easier to understand and not panic about. When your told something is wrong with your child you go panic mode.
9. Starting on treatment immediately, Not waiting on a 2nd set of test to come in.
10. Better testing to minimize miss diagnosis. Simplistic explanations of disease course and treatment. Honest answers.
11. Make sure the person calling is informed on the diagnosis, information, resources.
12. Better follow up testing.
13. Have information packets ready to be sent to the family with the diagnosis with accurate information - maybe with a list of specialists for that condition as well.
14. Coaching for pediatricians (doctor makes the call, has been briefed on the condition and possible risks - i.e. siblings), wasn't told not to google - wish they would have said that.
15. Instead of being told to not research it, be informed the the information is out dated and treatment is successful and people are thriving every day with this rare disease.
16. Pediatricians have better resources to provide families. I had to find everything myself.
17. More education for the doctors for when the screenings come back . Make sure they don't give false hope.
18. If the geneticist let me know that NBS would be how my child wou instead of giving me this dismal fate without confirmation and telling me at 5 months to consider abortion.
19. Communication of results, Access to treatment, Family support.



RANK IN ORDER OF IMPORTANCE TO YOUR FAMILY...

Domains provided by RTI from previous research on this topic of family outcomes from NBS. Families were asked to rank order these using the Mentimeter platform during the listening sessions.

Ranked from 1st order of importance to 5th by families

1. **Access to information about your child's condition**
2. **Ability to meet your child's needs**
3. **Access to high-quality services**
4. **Ability to meet financial needs of child's condition**
5. **Access to social support for your family**

THEMES WHICH EMERGED FROM THE DISCUSSION

After family introductions and the interactive questions on Mentimeter, families were asked to share verbally or in the Zoom chat in more detail about some of the top family outcomes identified during the Mentimeter feedback and rankings. The items listed below were unanticipated themes that emerged through the family's answers and sharing of their journeys. We list them here as we believe these areas are very important to also capture and attempt to quantify in a family outcomes instrument.

Awareness of NBS Prior to Screening

Prior to receiving a call that their child had a positive NBS result, many families shared that they were not aware of the NBS, what it tested for, or that their baby was tested. A couple of families that were aware of NBS reported knowing either due to working in the profession or family history.

Communication of NBS Result

Many families reported that when being informed of a possible positive NBS result communication was confusing, uninformative, and often from unknown providers. In some cases families were not told the specific condition indicated on the result or what symptoms to watch for, while many other families reported being told it was likely a false positive and their child would just need to be screened again.

Emotions Associated with Receiving Result

Upon receiving a positive NBS result families reported feelings of grief and uncertainty. Families reported that these emotions still come up years later when discussing their NBS experience and have ultimately lead to feelings of PTSD surrounding the experience.

Confirmatory Testing and Treatment

After receiving a positive NBS result families needed confirmatory testing and treatment. Families reported being faced with significant insurance and financial barriers. Many families also needed to travel out of state to seek care, and some continue to travel significant distances for ongoing monitoring and management of NBS condition multiple times a year.

COMMUNICATION OF NBS RESULTS... FROM A FAMILY PERSPECTIVE

We have some PTSD from the way it was communicated with us because of the lack of information that was provided. I think that contributes to how scary each MRI is. If we had had accurate information up front, I think it might make all the monitoring a bit easier to handle.

We were told "keep your baby as healthy as possible and stay away from people" and then we had to wait for appointments or results it was painful.

We received a voicemail from my pediatrician on a Friday night from an unknown number. So I didn't answer, and she just was like, “Hey I have some information. Call me back tomorrow I'll leave some notes with the nurse, it's likely a fluke.” And so I called back the next day, and then the nurse just read the notes, whatever the doctor wrote, and well so you start googling it. And you don't want to go down that rabbit hole as I'm sure all y'all know.

When we got our phone call, it was really just okay. We have this. We're trying to find more information for me, for you like, sit tight. It's like I'm holding my new baby who has potentially, you know, and we're being left to Google. And all we're getting is your child will die from this. And how do you deal with that? That is the biggest barrier of like the grief that you go through, and no one prepares you for having anything like that. And so when we can't provide accurate information up front, we're left to that, and that is more traumatizing than just being given a set of accurate information up front. In fact there it's very often that I find myself almost with PTSD symptoms of that uncertainty. That period of Googling where we weren't able to really understand what was going on.

COMMUNICATION OF NBS RESULTS...MORE THOUGHTS

So the only thing I was gonna say was not taking it seriously as a parent. I didn't take the borderline results seriously, because she told me not to, so I didn't worry about it. I went on with my day, you know, monitored him as I could for what I knew, but the doctor's not taking it seriously really makes you as a patient, not take it as seriously as you should. And so this experience has really given me a voice too. If I feel like something is wrong- the doctors are not always right, and they're not always going to care as much as you do about your child. And so I think this really gives, I am sure, all of us a voice to advocate for our kids. even if the doctors disagree.

When we got our positive it was the nurse practitioner and she didn't give us much information. By that time my son was 4 days old, and he was throwing up, it was not like a spit up. It was he was just throwing up, and there was some dry blood in there. But you know, even with that, like lethargic, and and I mean he already showed symptoms. But even with that she told us it could be a fluke. Just watch for symptoms. but didn't really mention what were the symptoms. So I Googled everything right after that. And I am glad I did Google this stuff because I knew what to look for, so I didn't wait for the second set of newborn screening. I just went in one day. I think it was day 6, 7. I went in cause at that point he was seizing, and he wouldn't wake up. So it was. I was like, I am not waiting for.
So I wish, yeah, I do wish like she had a little more information and stuff like that.

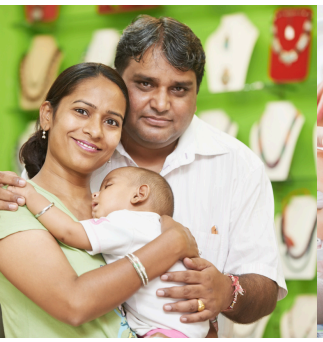
~

I'm like he's throwing up a lot. And then she'd kind of just brushed it off....yeah, it wasn't great.

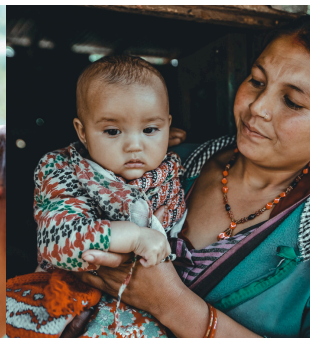
A nurse called us, told us the (name of the condition) and scheduled an appointment as if what she just said didn't turn our world upside down. They put us with the first available provider, a PA, who came in with sheets printed off from google. Having a loose understanding that our kid could die from this thing and the providers didn't even know what it was scary. I think they took it seriously, maybe even overkill, but initially really didn't understand it - which put a lot of unnecessary fear in our house.

THE FAMILIES SAY...

These quotes from families were taken from the transcript of the listening sessions. Any identifying information provided by the family has been removed.







I just want to say it has been so great hearing everyone's stories. I wish they would connect folks with positive finding to someone that has gone through it to help them navigate or give support to the family

I just wanna tell everybody~
Life's gonna present a bucket full of problems.
Being alive is a miracle and regardless how you come out and start into this world, given a chance at life.
Lucky we live in an age where we have some chance of resolving some problems that our young one's face, and that's a positive thing to look forward to.
I'm just really grateful for the newborn screenings to indicate what's going on. So we can address it, you know. And I just really appreciate that.

THE FAMILIES SAY SOME MORE...

The positive newborn screen for us was super scary all on its own. But we found it particularly frustrating because we felt like we were in a system trying to navigate and figure out what to do and what was next. And the system was fighting against us instead of with us. And so that was a really hard time.

But all healthcare systems are on, I guess, a system to be able to communicate better. But since some of our doctors are here and some are, you know, 1,000 miles away, and they leave it on us, the patient, to be able to share the information between the 2 places. So you know now, computers don't even have a disk thing. So we're having to keep my mom with her old computer so that we can upload a disk to be able to get a disk to another state.
Because, you know it's not reliable to ship it, because now, you mail something and you don't know if it's gonna get there or damaged, and you only have one copy. So we just wish information could be better, cause every time we do anything we get stressed out.

I like the honesty of a lot of the medical professionals.
but I like the honesty of the physicians, the nurses, the clinicians saying, “I don't know. But let us find out. Let us bring some people in here, and we'll work on it.”... We would go into a room with our doctor, and we would have 4 or 5 other people come in
anywhere from geneticists to residents to add to their advice, or to, you know, learn so that they would know for the next person. So I think that open honesty of
“you know what- we don't know. But let's find out. Let's see what we know what we can do, what resources we can pull together.” So that was helpful for me because I was lost. I didn't know anything about the disease.

THE FAMILIES' FINAL THOUGHTS...

You feel like you are in a constant state of fight or flight, if its not a doctors appointment, or blood draw, its insurance denials for your child's lifesaving treatment. Better family support throughout life stages, and making sure that all the pieces are in place. Not leaving families to address all of this while also learning a new rare disorder and how to manage that.

I think I get more advice and input from social groups than our team. Such as treatment options and routes of treatment you can take. We don't really get that guidance from our team, and I lean on the community a lot.

I think it's just important that teams kind of offer up options to people so like you know what is available to you, or what different routes you can take. You know, as far as we do frequent blood work. Being able to do that at home is really nice, because for the first 2 and a half months of my child's life we were taking her to and from the hospital 3 times a week to be poked and then within that, just because there were so many different moving parts, there were a couple of times where her blood work got lost somewhere, and so we didn't get a result. So we'd have to go in and poke her a fourth or a fifth time for the week. So I mean just being able to kind of pick and choose what works for your family. And having those options, I think, is a big thing as far as treatment goes and things like that.

ADVICE FROM NBS FAMILIES TO FAMILIES BEHIND THEM ON THE PATH

At the conclusion of the listening sessions, there were a few questions that we ran out of time to ask the families. We chose one of these questions (in the box below) to reach out via email to each participant individually and ask them to respond. Nearly every family shared their feedback and advice for others. We felt this powerful advice from those who have navigated this journey may be invaluable to those families a bit further behind them on the path, and so we include some of that advice here in hopes it will reach those who need it.

Question

Imagine speaking with a family who has just received a positive NBS for the same condition that your child received. What is the most important piece of wisdom, advice or awareness you would share with them?

ADVICE FROM NBS FAMILIES TO FAMILIES

1

Knowing early that your child has ALD is scary but also the greatest gift. To be able to monitor and treat if needed provides so much hope that your child and you will be ok. There is so much support out there and such a close ALD community of families, doctors, and researchers.

I would advise to keep the child isolated from others if they are diagnosed with SCID. And get in touch with a specialist on steps you can take to keep your child healthy until further testing. Also call their insurance provider to see about expediting the referrals. Lastly, not to allow this information to shadow the beautiful newborn stage and enjoy the milestones.

As you embark on this journey, remember that a network of patients, caregivers, and advocates is here to support you. The progress in medical treatments for SCID is a beacon of hope, ensuring that your child can thrive while awaiting further breakthroughs.
If you have any questions, need guidance, or simply wish to talk, please do not hesitate to reach out. Our community is your community, and together, we are stronger.

While we are all dealing with something different and might not know 100% what they are dealing with, we are all on the same journey.

My response would be - Your child's life was just saved by newborn screening. Your child has a diagnosis that comes with a treatment, so learn the new things. But more than anything remember to love them first, be their parents first. Soak up their smell and their size, their personality, and all their features. I was so caught up in the diagnosis that I didn't make time to bond and enjoy those really early moments. It is easier said than done when your world is flipped upside down, but find support and connection with those in the same rare disease space as you. They are invaluable.

Breathe. It will be ok. It feels so overwhelming right now with the information overload, but I promise that your baby will be ok. There are hard things that you will face, and it will feel like a roller-coaster, but don't look back on this first year of life and only remember the hard. Don't forget to remember the good. The first words and rolling over and steps. The first time they smile or laugh. Those are the important things! Also, you ALWAYS know best for your child and don't be afraid to stand firm on your hard no's! You have a world wide support system built in now and have 100s of people cheering for you and willing to shed tears with you, or give you ideas when you are stuck. You've got this! And that baby is a blessing and will be so supported!

ADVICE FROM NBS FAMILIES TO FAMILIES

2

I would share that this diagnosis does not define your child. It's going to feel like every wish and hope for your child was just lost, the plan you envision banishes but it will be okay! Take in your new baby snuggles and don't worry about the things you can't control.

You may be grieving the loss of the life you imagined for your child right now. That's understandable and totally normal. Remember that grief is a process, not a state of being. Allow yourself the tears and know you will get to the other side of this. It's hard, but try to enjoy every moment with your baby - take pictures, sing songs, smile, laugh, enjoy the cuddles and the middle of the night wake ups. Your baby needs you right now, and you are the best person to be there for them. This will be hard but you will be ok, and so will your baby. Knowing this diagnosis early is a blessing even if it doesn't feel like it right now. There are doctors who will help you understand your child's diagnosis, there are support groups where you can feel part of a community, and you will become a parent expert. Sometimes that will give you confidence while other times it will make you very frustrated. Try to find a mentor quickly - a parent who has been through exactly what you are going through, with your same diagnosis - to help you navigate these first weeks, months, years. It will all be okay. Just take one step at a time.

I would tell them that it is going to be ok!!! To seek out other families to not only learn more about PKU/food ideas/etc, but to see that their children are doing great. Also, don't be afraid to ask questions or to ask for help, you will need it.

I know ALD seems really scary right now, but you will figure out your rhythm. You will feel like you can breathe again. You will be able to do all the things you want to do with your child and not feel hopeless. Your child can and will thrive. They will learn to be resilient - and so will you! Join ALD connect/ALD alliance, go to the zoom support meetings, learn from and lean on your ALD community. Be diligent, keep on top of your blood work, MRIs, and appointments. You live in the best time ever to get this diagnosis. We know more now than ever before and we have new treatments in clinical trials, and being designed every day. Welcome to the best club you never wanted to join!

One thing I was scared of was the horror stories about children who are improperly managed and what my child's life would look like.

I would tell someone that their child is going to live a full, happy, healthy life regardless of the diagnosis. They will play sports and they will look like a 'normal' child. The medicine is the hardest thing to get into the groove of but once you do, you'll be able to do it in your sleep in a few months. They talk about stress dosing a lot, but in reality we don't have to stress dose as often as they think.

I would also tell them that even though the doctors say that they will not have to have extra salt in their everyday life, take it from me and the other parents that have CAH children, they will. They will need salt when it is hot outside or when they have overexerted themselves. This is something I had to learn as I continued to read.

ADVICE FROM NBS FAMILIES TO FAMILIES

The best piece of wisdom would be the that grief and joy will, in fact, coexist, also you'll grow into the best version of yourself.

The advice I would give is take one day at a time because thinking about the future will be overwhelming and you'll miss the joy in the present time.

The last is find your community, that's where your real support system will be happening.

I would say to the family to always be an advocate for their child. They know them best and will be become an expert in HCU. I would also say it is overwhelming now but it will become there new “normal”. It is a hard but manageable disorder and many of the children can live a mostly normal childhood.

3

Breathe. This next chapter of your life is going to be a challenge, but you love your child and everything ahead of you, even the worst of the worst days, will be worth it. Lean on your people, accept help before you need it, ask questions early and often, connect with people who have been through what you're about to go through (keep in mind your experience might differ from theirs for better or worse, but you're not alone), ... be ready to be vulnerable and communicate honestly because holding in all of the stress, anxiety, questions, fear, guilt, etc. can be really damaging to your mental health and your relationships, keep detailed records of everything (appointments, symptoms, food, medications, suggested treatment options, pictures/videos of health episodes, reactions, etc.). You might not even yet know how important certain details might be later. Find something for you to bring yourself peace when you're really having a hard time. When you're at your best, or at least not at your worst, you have more to give to your child so learning to at least sometimes prioritize your wellbeing is important. If you have other kids make certain to really communicate what is going on and set aside time for just them, read books that bring clarity to difficult situations and can maybe help you understand of iterate the battles that you're fighting (Atlas of the Heart by Brene Brown and Solving for Why by Mark Shrime, MD), keep track of financial records, expenses, etc. and see if you can qualify for any tax breaks, financial assistance, etc. because having a kid with healthcare issues is not only time consuming, it's very expensive.
Having a child with a health condition is HARD. You will grieve for the life that you hoped for in your head, and that's okay. You will have people who do not and cannot understand your situation, and that's okay. You'll have many more who do and show you kindness in ways that you'll remember for a lifetime. Learning to decipher between the two early on will save your sanity.
Everything will be okay, even when it's not okay... you've got this! When kids are in need, parents become superheros and find strength that they didn't even know they had... everything you need to fight for your kid is already inside of you. Get yourself battle ready and be their warrior.

CONCLUSIONS

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In conclusion, the 19 families that were involved in this project had invaluable insights and experiences both positive and negative, that can help to inform further work by RTI on a Family Outcomes instrument. These insights can also be leveraged to make improvements in the overall quality of the newborn screening programs and how those programs can become more family centered in their approach to providing a public health service. Our hope is that the words we heard and which are documented here from families in the Mountain States Region will go beyond just our region to further highlight the need for the family perspective, input and feedback in all aspects of newborn screening. These families have experienced the NBS system in the most intimate of experiences for their beloved children and they have valuable lessons to teach us all about how to improve their own family outcomes and the outcomes for those who will follow.

Final Thoughts

All families, in the end, were very appreciative of newborn screening

The families expressed being thankful, blessed, glad the system existed and for the results they received for their child because parents were able to seek specialty care early on. Many parents also expressed that without NBS they were told their child would die or experience irrevocable damage.

Families need support to not feel isolated, alone and afraid

Families reported having to find their own support and that they were not connected to that support from either the NBS system or the specialty provider. Facebook groups, national organizations and finding other families near them were regarded as extremely helpful and necessary.

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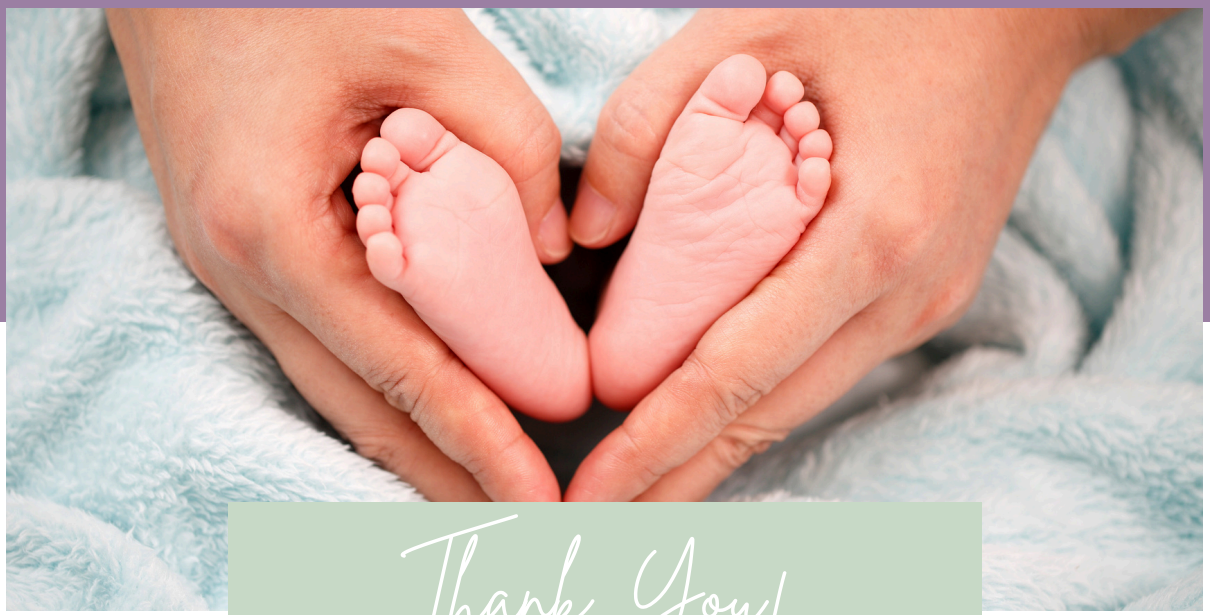

Thank You!