FOUNDATION FIGHTING BLINDNESS

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Survey of Affected Individuals

Usher Syndrome Type 1B

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USH1B Survey of Affected Individuals and Caregivers

Objectives: Gather information from the community of affected individuals to share with researchers.

Recruitment: My Retina Tracker Registry e-blast and informal patient networks

Mode of Administration: Self-administered through Survey Monkey (English only)

Dates of data collection: August 5, 2021 – September 6, 2021

Number of Respondents:

- Total = 81
- Adult caregivers = 57
- Affected adults = 14
- Other = 10 (most parents of affect adults)

Number of affected individuals: 97 (16 families have >1 affected individuals)

Diagnostic Journey

What age (in years) was the affected individual first diagnosed clinically?

Respondents (N)	73
Mean	8.1
Median	6
25^{th}, 75^{th} percentile	2, 12

What age (in years) was the affected individual first diagnosed genetically?

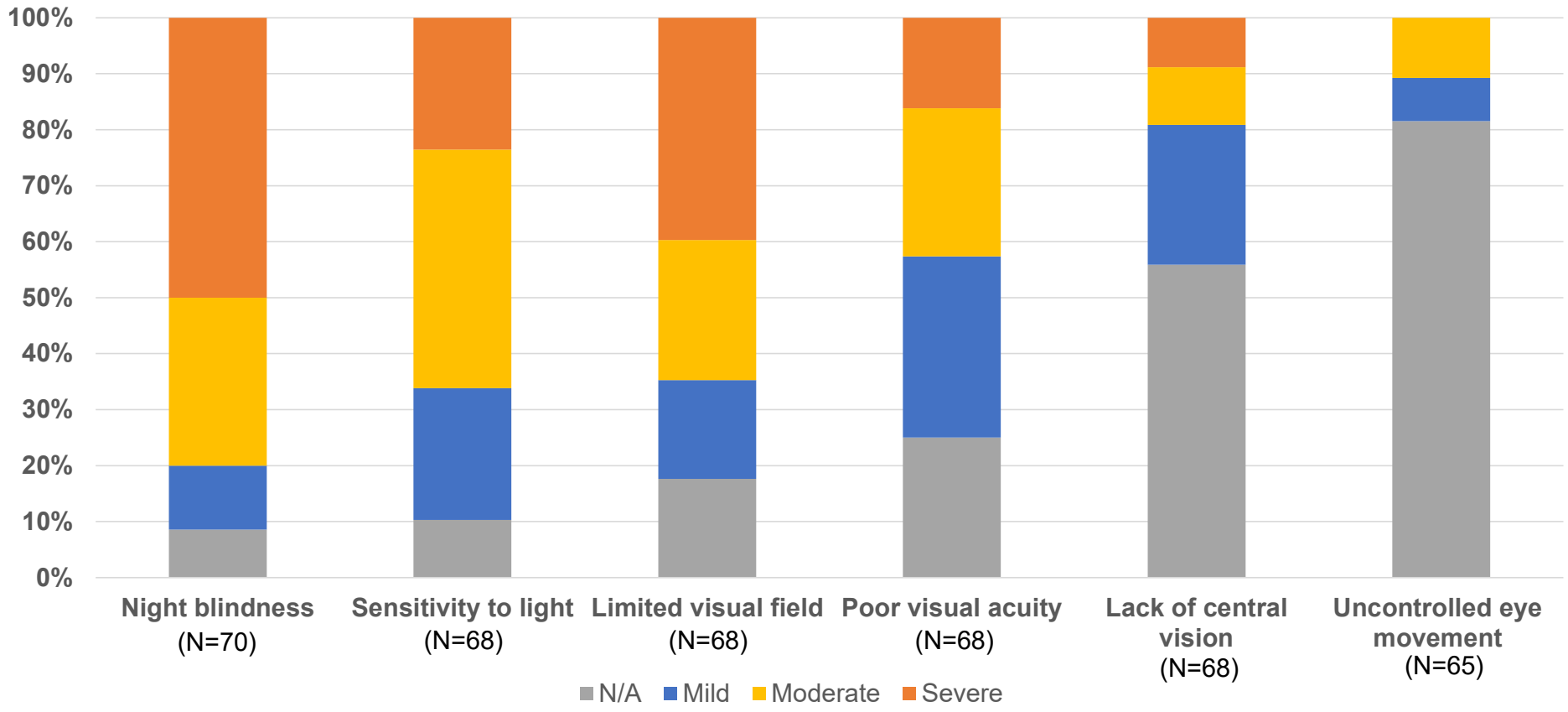
Respondents (N)	73
Mean	10.3
Median	6
25^{th}, 75^{th} percentile	2, 14

Current Best Corrected Visual Acuity in the Worse Eye

Best Corrected Visual Acuity	Number	(%)
20/20 to 20/30	27	41%
20/40 to 20/60	15	23%
20/80 to 20/160	6	9%
20/200 to 20/400	6	9%
20/500 to 20/1000	2	3%
Counting fingers / Light perception / No light perception	10	15%

For context, the average (median) current age of individuals reporting BCVA is 19.5 (14) years.

Self-Rated Severity of Visual Symptoms



Other Related Conditions

Are there other symptoms or physical health conditions (eye-related or otherwise) that are related to the individual's diagnosed genetic variant?

Coded Response	# Responses (out of 55 responses)
Deafness	28
Poor balance	22
Vestibular dysfunction unspecified	11
Autism	4
Cataracts	2
Cystoid macular edema	2
Vestibular migraine	2
Other headache	2
Low muscle strength / tone	2
Vestibular areflexia	2
Others (with 1 each)	10

Almost all respondents reported deafness, poor balance or other symptoms of vestibular dysfunction.

Worries

Thinking about your / the affected individual's condition, what worries you most?

Coded Response	# Responses (out of 78 responses)
Total loss of vision	24
Progressive vision loss	18
Unknown future	13
Participating and thriving	9
Mental health	8
Loss of independence / reliance on caregiver	7
Accidents / injuries / safety	6
Inability to work	3
Not being able to communicate	2
Others (with 1 each)	5

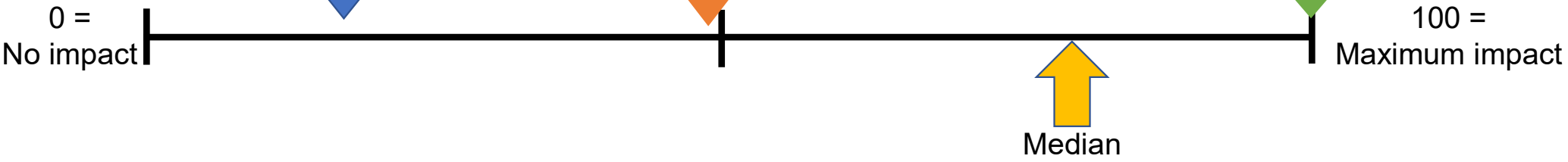
Over half of the respondents (42/78) reported that further loss of vision was their biggest worry.

Impact on Family

"We have to adapt and overcome…. Ush1b has changed the way I think about accessibility in our home, vacations, schooling, extra curricular, social emotional health. It has impacted everyday activities but at the same time we adjust and the way things are done become the typical."

"It had a massive impact in the years following diagnosis and meant that we introduced Braille and touch typing. It allowed us to plan. It was also a weight on our shoulders about how and when to tell our son."

"For his siblings, it's mostly the attention that they feel their brothers get and always having to help. Growing up to quickly because of it. For us parents, it's changed our lives drastically, most notably having to learn to be an advocate and teaching them how to advocate for themselves. Also, grief from all the uncertainty that comes and goes."



63 respondents rated the impact of Ush1B on their family.

Participation in Clinical Trials

Based on what you know today, what's your comfort level about participating in a trial with FDA oversight to treat your Ush1b? (0 = not at all comfortable; 100 = totally comfortable)

Respondents (N)	62
Mean	58.4
Median	60
25th, 75th percentile	40, 80

What are the top 1 to 3 questions you have about being in a trial?

Coded response	# Responses
Safety / expected side effects	38
Will vision get worse?	18
Expected benefits	14
Able to participate in future trials?	7
How long trial involvement?	7
Affect future access to treatment?	4
Trial appropriate for me or my child?	4
How is treatment administered?	4

Motivations

What are your top 1 to 3 motivations (or desired outcomes) for participating in a clinical trial?

Coded response	# Responses (out of 58)
Stop progression	26
Improve lost vision	17
Finding a cure	13
Getting life back / independence	9
Advance treatments	8
Provide hope	5
Help others	5
Understand sign language better	1
See family	1

Anything Else for the Research Community to Know?

“I would like them to keep in mind that because children are getting diagnosed at such a young age now that it is our hope a cure will be found before it’s too late. ”

“Just always hopeful that something can be done to slow down the progression.”

“The diagnoses and the progression of the disease is both devastating and emotional draining. As a parent you want the best opportunities for your child to be happy and successful in life. With the diagnoses you know what’s coming but not how fast or how severe. Technology has helped us overcome the hearing loss part of Ush1B, while the vision part is still so individual and unknown. It makes it impossible to plan for the future without fear or worries. As our daughter is planning a carrier in Animation where she will be dependent on her vision and as parents we tell her to go for her dreams. But we don’t know if it’s the right advice. It’s devastating enough as a parent, we couldn’t imagine being the one having the disease. This is a panic situation where time is of essence as she loses more vision each day. It’s urgent and we have waited so long for research….”

“We need to save the next generations of children with ush 1b. We as parents need the be kept informed of any clinical trails from the very first stages. We would travel the world to join a clinical trial if it would benefit my son.”

“We have loved attending the combined family -research conferences. I know these are hard for researchers. But they’re immensely valuable for families and a great benefit for us especially. Please. We would love more of these…”

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Thank you!

Appendix (1)

As we plan to share the results of this (anonymous) community survey with the academic research and physician community, is there anything else you feel is important for them to know? Please share any last thoughts, comments, or questions!
Help find a cure
also deaf and afraid to attempt to find methods to hear. soon to be blind and deaf
Restore vision is my hoping!
Thank you for all the work you do. Although the patient pool may be small, the need for a treatment is huge.
Just always hopeful that something can be done to slow down the progression.
Share how study was impartial, fair and not motivated by Big Pharma getting rich

Appendix (2)

As we plan to share the results of this (anonymous) community survey with the academic research and physician community, is there anything else you feel is important for them to know? Please share any last thoughts, comments, or questions!
1. With one-degree field of vision and blind spots scattered throughout my visual field, it is still very difficult to navigate websites and software apps, even with accessibility features. My eyes can't line up, making it difficult to pinpoint. Because of this, I also walked on a street, instead of a sidewalk, a couple times and hit my head and body frequently. 2. I have always struggled communicating with both the hearing and Deaf folks. I lack social language becauseI was never able to put words with facial expressions and body gestures. 3. Canes don't work. We DB folks need eye guides/interpreters. 4. I cannot follow movements, including closed captions. Summary: I wish I learned how to be DeafBlind, not Deaf. To survive. To have decent lives. To both work and play.
The diagnoses and the progression of the disease is both devastating and emotional draining. As a parent you want the best opportunities for your child to be happy and successful in life. With the diagnoses you know what's coming but not how fast or how severe. Technology has helped us overcome the hearing loss part of Ush1B, while the vision part is still so individual and unknown. It makes it impossible to plan for the future without fear or worries. As our daughter is planning a carrier in Animation where she will be dependent on her vision and as parents we tell her to go for her dreams. But we don't know if it's the right advice. It's devastating enough as a parent, we couldn't imagine being the one having the disease. This is a panic situation where time is of essence as she loses more vision each day. It's urgent and we have waited so long for research….

Appendix (3)

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Along with clinical trials, I'm very interested to know if there are any longitudinal studies for 1B currently in progress? I haven't been able to find anything. Additionally, I'm curious about research into atypical Ushers. From my (limited) understanding, I believe MYO7A is associated with an atypical presentation, and it would be great if there was more research into this and also translation of existing research in the area.
The emotional and mental impact of this diagnosis has been devastating to my family. While my son is still young, we have noticed his reactions to sunlight and darkness which shows that his vision is deteriorating even at this young age, and that breaks my heart. Ushers 1B typically progresses at a faster rate than other Ushers sub-types, as you know, and part of me wishes I could stop time so that my son can maintain his vision and see the world fully. Despite this fear and sadness, I have hope in our wonderful medical and research communities. There are many talented and caring individuals who are working on developing a cure for this type of blindness so that children like my son do not have to face the possibility of a dark, or limited, future. Please know that your work is SO important to families like mine. Our son's livelihood is literally in your hands. Thank you for all that you do.

Appendix (4)

As we plan to share the results of this (anonymous) community survey with the academic research and physician community, is there anything else you feel is important for them to know? Please share any last thoughts, comments, or questions!
How and why Usher can appear in families when no one in the past ever experienced symptoms or the syndrome from either parent's history. Please keep in mind that for families living this everyday this is not "research" its life. Please don't forget the human element with this syndrome and that its experienced uniquely by everyone.
I would like them to keep in mind that because children are getting diagnosed at such a young age now that it is our hope a cure will be found before it’s too late.
Sight is beautiful and we need to finds a cure , how can us Ushers help with this process?
XXXXX has gotten more tired with a toddler. Her eyes get tired and naps when toddler naps
Our daughter is just beginning her high school career aged 11yrs, and thinks that she has all options available to her in her future career and life. We desperately want this to be the case, and yet we know that by the time she leaves high school she is likely to have experienced further and significant vision loss, meaning that she is dependent on aids and support to lead the independent life she dreams of. Her vision is changing now - she does not have 5-10 years left to wait for treatments. We are desperate to support the research and clinical community to achieve outcomes for our daughter and her peers. Thank you for every drop of effort you put into this work
Raising awareness about the negative impacts of consanguineous marriages is critical - especially in developing countries. All of the cases of Usher syndrome and RP that I am aware of (at least among my family in India) are among people whose parents were first cousins

Appendix (5)

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14 years ago Dr. Elliot Berson told me to have hope. He said treatments would be about 10 years away. It's discouraging that it's been now 14 years and still no treatments.
No. We're still new to it all.
Not that I can think of as far as I know but other than if this trial safe to participate. I hope this clinical trial is going to go really well and smoothly!
We need more information about the condition in general. It seems to vary so much in individuals. We had a very bleak view of what it meant to have the disease but it has so far been very life enhancing for our son. He has had so many opportunities in life as a result of his disability and lives life to the fullest.
Please let me know if I can volunteer any hours for your organization. I 100% support your endeavors in treating Ush1b blindness.
We have loved attending the combined family -research conferences. I know these are hard for researchers. But they're immensely valuable for families and a great benefit for us especially. Please. We would love more of these…

Appendix (6)

As we plan to share the results of this (anonymous) community survey with the academic research and physician community, is there anything else you feel is important for them to know? Please share any last thoughts, comments, or questions!
qualitative outcomes (impact on daily life) to be defined by patients/patient reps; results/learnings/conclusions of the trial should be used to get approval for USH1b treatment - guarantee to pursue search for treatment/cure USH1b (and not only use the results for the research other disease areas for the sake of economic interests; in case of outsourcing of licences, guarantee that USH1b trial will not be punched out of the pipeline due to other (more beneficial?) interests of big pharma;
What can we do to support this for us extremely important research? Hoping for a cure!!!
We need to save the next generations of children with ush 1b. We as parents need the be kept informed of any clinical trails from the very first stages. We would travel the world to join a clinical trial if it would benefit my son.
Please have more research and resources available for those of us living with this condition. Perhaps there can be more innovation in products and therapeutics that can make living with this a little less difficult and provide comfort for an extremely challenging state of being
We feel that the disease should be addressed with a more global (less segmented) approach. The medical staff is each focused on its area of expertise (eye, ear, balance) with no overall coordination while the combined effect of deafness, RP and vestibular issue creates a unique disability with unique impacts on day-to-day life for which we feel a lack of support. Besides, the combined effect of the eye the the vestibular conditions is strongly underestimated.
As a family we have been very disappointed with the general attitude from the medical community about helping us to accept the diagnosis. We have been directed towards third sector organisations for support which have been brilliant but we feel that there should be something offered by the NHS to support families after such an important genetic diagnosis is given. Being told repeatedly 'not to worry' about the future without any support has not been helpful.

Appendix (7)

As we plan to share the results of this (anonymous) community survey with the academic research and physician community, is there anything else you feel is important for them to know? Please share any last thoughts, comments, or questions!
Combined hearing and vision loss has devastating effects on individuals because of how it affects their quality of life and independence.
There are more people than you would think impacted worldwide. Just look at the Facebook groups with people begging for help. For us in particular, we are concerned for our child's future.
During of trials. We've hearing of clinical trials for years, how much time do we have for authorization of any of the clinical trials.
I hope that when a cure is found, I believe that it will become a treatment ,that concerns all patients, be a more general one and not an individual one, and that everyone should worry if this treatment concerns their own exon.
We are worried about our daughter vision loss due to usher syndrome
Urgency

Appendix (8)

As we plan to share the results of this (anonymous) community survey with the academic research and physician community, is there anything else you feel is important for them to know? Please share any last thoughts, comments, or questions!
As a community member that feels confident in education and resource side of the diagnosis, our family struggles to have a true understanding of the clinical side. I think it is imperative we bridge this gap. There is a disconnect between providing the diagnosis, maintaining eye health care, and having resources to support daily life. The diagnosis of Usher syndrome can be exhausting because families are consistently juggling many service providers and tend to become the experts. Families are yearning for information that can be comprehended at their level of understanding. While we are eager for a cure, there is a lack of responsiveness from families because they truly just don't have an ounce of energy left to digest information that seems like a foreign language. There is also a need to educate children during appropriate years about the clinical aspect of their diagnosis. We focus a lot on self advocacy; however, this component is missing. At the end of the day the reality is, if these trials are not available by ages when they parents will be at the forefront of the decision, the choice is theirs. So educating our youth is a need. In my professional opinion if the clinical and educational community could collaborate we would see an increase in participation and data collection to support the efforts of both sides. Taking the time to hear from the individuals/children as well would also be "eye opening". Best of luck and much sincere appreciation for your efforts.
The urgency of this research to families with children with Usher 1B as a way of preserving the children's vision an subsequent life experience/opportunities