



**X-Linked Retinitis Pigmentosa**

**Externally Led Patient-Focused Drug Development**

**Voice of the Patient Report**

Meeting Date: 7 June 2022

**Meeting hosted by:** Foundation Fighting Blindness

**Submitted to:** The U.S. Food and Drug Administration (FDA)

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## **X-Linked Retinitis Pigmentosa Voice of the Patient Report**

### *Driving Research, Saving Vision*

The urgent mission of the Foundation Fighting Blindness (“the Foundation”) is to drive the research that will provide preventions, treatments and cures for people affected by retinitis pigmentosa, age-related macular degeneration, Usher syndrome and the entire spectrum of retinal degenerative diseases. The Foundation is a beacon for those affected by these blinding diseases. This *Voice of the Patient* report was prepared by the Foundation as a summary of the input shared by people and families living with X-linked Retinitis Pigmentosa (XLRP) during an Externally-Led Patient Focused Drug Development Meeting (EL-PFDD). This meeting was hosted virtually on June 7, 2022.

**Authors and Collaborators:** This report was prepared and submitted on behalf of the Foundation by Todd Durham, PhD., Sr.

Vice President, Clinical & Outcomes Research, Joan Fisher, Sr. Research Specialist, Janet Cheetham, PharmD, consultant, and Rebecca Ratchford, Senior Administrative Assistant, Foundation Fighting Blindness and by Chrystal Palaty, medical writer.

Consulting Partners include James Valentine, Esq. and Larry Bauer, RN, MA, from Hyman, Phelps & McNamara, P.C.

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Janet Cheetham, PharmD has consulting relationships with companies developing therapeutics and technologies to advance health.

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James Valentine, Esq. and Larry Bauer, RN, MA are employed by Hyman, Phelps & McNamara, P.C., a law firm that represents patient advocacy organizations and companies that are developing therapeutics and technologies to advance health.

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**Point of Contact:** For questions related to this report please contact Todd Durham, PhD., Sr. Vice President, Clinical & Outcomes Research, Foundation Fighting Blindness, [Info@FightingBlindness.org](mailto:Info@FightingBlindness.org)

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phoning in or contributing their insights online. We dedicate this report to all of you.

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Finally, thank you also to AGTC and Biogen for so generously providing financial support for this meeting.

*Disclaimer:*

*Foundation Fighting Blindness recommends all individuals with XLRP consult their physician before starting or changing their treatments.*

## **Executive Summary and Key Meeting Insights**

The Foundation Fighting Blindness (“the Foundation”) hosted the X-linked retinitis pigmentosa (XLRP) Externally-Led Patient Focused Drug Development (EL-PFDD) meeting on June 7, 2022. This meeting was held to provide patient and caregiver perspective of the symptoms and burdens associated with XLRP in daily life, as well as the massive unmet treatment needs experienced by those who live with XLRP every day. The information gathered at the meeting is presented in this *Voice of the Patient* report, a high-level summary of the perspectives generously shared by the patients and caregivers of individuals living with XLRP, who participated in the June 7, 2022, EL-PFDD meeting. This report also includes selected comments that were submitted through a public online portal via the FightingBlindness.org website and the results of submissions by My Retina Tracker® Registry members via MyRetinaTracker.org of “A Survey to Explore the Impacts of X-Linked Retinitis Pigmentosa on Daily Living and Goals for Treatment”.

The information in the *Voice of the Patient* report may be used to guide therapeutic development and inform the FDA’s benefit-risk evaluations when assessing therapies to address XLRP. The hope is that this information will catalyze better treatments and ultimately a cure for those affected by XLRP.

The Foundation has provided this report to the FDA, government agencies, regulatory authorities, medical products developers, academics, and clinicians, and it is publicly available for the many stakeholders in the XLRP community, including the Foundation’s partners and advocacy organizations. Note that the input received from the June 7, 2022, EL-PFDD meeting reflects a wide range of XLRP experiences, however not all symptoms and impacts may be captured in this report.

## Key meeting themes and insights:

- 1. X-linked retinitis pigmentosa (XLRP) is characterized by progressive vision loss.** Symptoms range from difficulties adapting to changes in lighting, diminishing night vision, a loss of peripheral vision, and eventually progress to a loss of central vision. Individuals living with XLRP live in fear of deteriorating vision loss and eventual blindness.
- 2. XLRP is not just about vision loss but involves a gradual deterioration of independence.** Dealing with XLRP is a constant and daily battle. As vision deteriorates, all activities of daily living are impacted. Individuals living with XLRP first lose their ability to drive at night, and then during the day. Participating in social engagements and events becomes more challenging, they lose the ability to read, and employment and careers are severely impacted. Roles within families change and self-esteem plummets. They experience injuries and many experience stigma and discrimination.
- 3. Currently, there are no approved treatments for XLRP.** Many individuals have tried the few available therapies including vitamin A. Several tried investigational therapies in clinical trials, some of which involved high risk, invasive interventions.
- 4. As vision deteriorates, people with XLRP are forced to continually adapt and find new strategies and technologies.** These include combinations of portable lighting devices, magnifying devices, UV sunglasses, audio reading and other modifications. Many described having to eventually use both visual and audio modifications.
- 5. Approaches to managing XLRP work “somewhat”, however most do not treat the target symptoms, slow progressive vision loss, nor replace or restore sight.** There are currently no approved medications for XLRP and access to experimental treatments is limited, often due to restrictions in clinical trial eligibility. As a result, some individuals affected with XLRP are not using any treatment.
- 6. Short of a cure, those living with XLRP need a therapy to improve night vision, visual acuity, and peripheral vision.** Many meeting participants asked

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for a therapy that would prevent their vision from deteriorating further, with the hope of maintaining their independence, returning to school or work or walking without assistance. They asked for improvements to central vision, broader clinical trial inclusion criteria, specifically to include women, and more research for XLRP.



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## Introduction and Meeting Overview

### Clinical overview of X-Linked Retinitis Pigmentosa<sup>1</sup>

X-linked retinitis pigmentosa (XLRP) is a progressive degenerative genetic disease. XLRP is one of the most common and the most severe forms of retinitis pigmentosa. Vision loss starts in the first few decades of life and progresses to the point of legal and/or complete blindness, usually when an individual reaches their 40s or 50s. An estimated 15,000 people in the United States are currently afflicted with XLRP.

XLRP is caused by variations in the *RPGR* gene and less commonly, in the *RP2* gene, leading to the loss of photoreceptor cells in the retina. Rods, the cells responsible for vision in the dark, degenerate first. Night vision problems are typically one of the first XLRP symptoms noted, then peripheral vision, side vision, day vision, and then central vision are progressively affected.

*“The typical course for RP is losing first your night vision, followed by peripheral vision, and then eventually your central vision.”* - Jonathan Tamaiev

*“XLRP robs you of your vision slowly over time until you're blind.”* - Greg Dubecky

*RPGR* gene variations manifest in two XLRP phenotypes. The most common phenotype is rod-cone dystrophy, characterized by loss of peripheral vision and night vision. The second, less common type, is cone/cone-rod dystrophy, which affects the central retina, affecting central vision, color vision, and contrast sensitivity.

*RPGR* and *RP2* genes are located on the X chromosome and can be passed down by either parent. Typically, XLRP affects males more severely than females, and women were not recognized as having symptoms but were believed to be carriers only. However, 15% of men living with XLRP do not demonstrate a classic inheritance pattern. In about 8.5% of affected families, a dominant inherited pattern is observed, which means that multiple generations of both males and

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<sup>1</sup> Clinical overview extracted from presentation by Dr. Thiran Jayasundera, Professor of Ophthalmic Genetics, Associate Chair, Strategic Implementation at the Kellogg Eye Center, University of Michigan at the XLRP on the June 7, 2022.

females with gene variations can experience symptoms. Generally, families experience phenotypic conservation, which means that affected individuals can expect to experience the same type and degree of vision loss as other family members with XLRP.

An XLRP clinical diagnosis requires a detailed family history, including symptom onset, duration and progression. An examination includes a best corrected visual acuity (BCVA) test, assessment of intraocular pressure and cataracts, and a full retinal examination, using imaging optical coherence tomography, widefield fundus photo and autofluorescence. Functional testing is conducted using an electroretinogram (ERG), and perimetry or visual field testing. Ultimately, genetic testing confirms the causative gene variation, provides a more accurate prognosis, and enables screening of other family members. Identifying the underlying causative gene variation will help to elucidate the basic biological defect and may eventually direct personalized therapeutic interventions.

At the time of this report, there are not yet any effective treatments to stop or reverse the deterioration of photoreceptor cells, but a number of new treatments are in development. Gene therapy or gene modification approaches are being tested in patients with early-stage disease, while stem cell therapies are being tested in patients at the later XLRP stages. Novel approaches including optogenetics or visual prosthesis that can substitute for the lost photoreceptors are being pioneered for late-stage disease.

### **Meeting summary**

The XLRP Externally-Led Patient Focused Drug Development (EL-PFDD) meeting was held virtually on June 7, 2022. The meeting represented an important opportunity for people with XLRP to educate FDA staff and other key stakeholders about patient and family perspectives on the challenges and unmet treatment needs of those living with X-Linked Retinitis Pigmentosa (XLRP). The hope was that this meeting would encourage future research and successful new product development for people living with XLRP who urgently need safe and efficacious treatment options. The XLRP EL-PFDD meeting was co-moderated by Todd Durham, PhD, Senior Vice President of Clinical and Outcomes Research from the Foundation Fighting Blindness (the “Foundation”), and by James Valentine, JD, MHS, from Hyman, Phelps and McNamara.

**Dr. Durham** opened the meeting by introducing the Foundation’s mission and providing a brief introduction to XLRP. **Dr. Ekaterini Tsilou MD**, a Medical Officer in the Office of Tissues and Advanced Therapies in the Center for Biologics Evaluation and Research (CBER) at the FDA, provided some opening comments from the FDA. She briefly explained the FDA’s role in reviewing drugs for a wide variety of ophthalmic diseases, including XLRP, and explained the bigger role in the FDA in public health and oversight. She thanked all the patients for their participation and commented she looks forward to incorporating what is learned into the FDA’s thinking about and understanding of XLRP. **Dr. Thiran Jayasundera**, Professor of Ophthalmic Genetics, Associate Chair, Strategic Implementation at the Kellogg Eye Center, University of Michigan, presented a clinical overview of XLRP to create a scientific foundation for the rest of the meeting. Key points from his presentation are summarized in the clinical overview on the previous page.

Dr. Durham introduced the moderator, Dr. James Valentine, JD, MHS who provided an overview of the meeting structure and encouraged individuals living with XLRP to contribute to the dialogue via online polling, calling in by phone, and contributing written comments using the online portal.

Online polling was used to determine the demographics of the meeting attendees who were representing those living with XLRP and are presented in **Appendix 1**. Most attendees are located the Eastern US, with strong representation from the US Mountain and Central time zones and the US Pacific time zone, as well as representatives from Asia and Canada. Approximately 2/3 of attendees were male, a third were female, and 5% were non-binary. All age ranges were represented with a little over one third of the attendees were in the 51-70 age category, followed by another third in the 31-50 age category. A majority of those represented in the polls had a genetic diagnosis with a variant or mutation in the *RPGR* gene, fewer in the *RP2* gene and some that are unsure about their genetic diagnosis. These demographics differ slightly from the individuals who participated in the “Survey to Explore the Impacts of X-Linked Retinitis Pigmentosa on Daily Living and Goals for Treatment”, summarized in **Appendix 2**. Survey participants were exclusively US-based, affected males with a *RPGR* gene variation.

The live meeting was attended by 77 individuals, including 30 XLRP affected individuals and 10 caregivers, family members or friends. A total of six attendees were from the FDA, in addition to six scientists, 17 individuals from industry, three healthcare workers, four from non-profits including the Foundation and one consultant.

The XLRP EL-PFDD meeting was structured around two key topics. Session 1 was *Living with XLRP: Symptoms and Daily Impacts*. Session 2 was *Current and Future Approaches to Treatment for XLRP*. The meeting agenda is in **Appendix 3**, and the questions provided for meeting discussion are in **Appendix 4**.

The morning session continued with pre-recorded patients selected to represent a range of individuals at different points of their XLRP progression. They shared stories of their diagnosis in late childhood or their early teens and how they are impacted by progressive vision loss. Many described how several other family members were also living with XLRP. They shared their worries about their futures and the futures of their family members and especially their children who are affected.

James Valentine moderated a discussion between several people who served on a live Zoom panel as well as people who dialed in by phone. Additional relevant comments entered through an online submission form were read by Dr. Durham. The names of panelists and callers are listed in **Appendix 5**.

The afternoon session opened with a pre-recorded panel of patients who described different medical therapies and other approaches they have used to address their vision loss. Many described their participation in clinical trials, and the results that they had experienced to date. Several described trying more alternative approaches, pointing out that when they feel good, their vision is better. Finally, patients concluded by describing their wishes and hopes for treatments and the trade-offs that they are willing to accept. Again, meeting attendees participated in online polling, called in and submitted written comments which were added to the moderated discussion by James Valentine and Dr. Durham. At the end of the meeting, Larry Bauer, RN, MA provided a reflective summary of the key meeting messages and Dr. Durham concluded the meeting by thanking all the participants and attendees.

The online polling results from Topic 1 and Topic 2 are included in **Appendices 6** and **7**, respectively. Survey results are presented in **Appendix 2** for comparison. To include as many voices as possible, the online comment submission portal was open for four weeks after the meeting. Selected comments are included in the body of this report, and all submitted comments are included in an accompanying PDF, which is available at <https://www.fightingblindness.org/xlrp-pfdd>.

This *Voice of the Patient* report is provided to all XLRP stakeholders including the US FDA, other government agencies, regulatory authorities, medical products developers, academics, clinicians, and any other interested individuals. The final report, the meeting transcript, and a video of the meeting can be found at <https://www.fightingblindness.org/xlrp-pfdd>. According to YouTube statistics, the meeting has been streamed over 660 times as of September 20, 2022.

While XLRP is a specific subset of Retinitis Pigmentosa (RP), these terms were used interchangeably during the meeting and in this report to refer to XLRP.

## Session 1: Living with XLRP: Symptoms and Daily Impacts

**XLRP-related health concerns:** All individuals living with XLRP experience difficulties in adapting to changes in lighting. Most also experience decreased night vision and loss of peripheral vision. These are also the top three most troublesome XLRP-related health concerns.

Meeting attendees used online polling to first select all of the XLRP-related manifestations that they or their loved ones ever had and which were the three most troublesome. Many, like Robert, wanted to pick all of the symptoms as being most troublesome.

*“Of all of those six [symptoms listed in the polls], I have experienced all six of those over my 40 years of knowing I have XLRP. ... It's hard to say which one is the most impactful because it is a spectrum and it is fluctuating throughout the day.” – Robert (caller)*

**XLRP is characterized by changing symptoms and ongoing progressive vision loss requiring constant accommodation, adjustment and adaptation.** This was a key point emphasized during this session.

*“Individual symptoms are not the most challenging part of having XLRP. The most difficult aspect of XLRP is the ever-changing nature of the symptoms and the ever-increasing disability. I recognized long ago that my coping mechanisms are only temporarily useful at best.” - William Hartman*

*“It was progressive for me...and as the changes came about so did the losses...loss of driving at night, loss of not being able to drive at all, loss of not being able to go where I want when I want, loss of not being able to see my wife's face, my children's faces, and my grandchildren's faces, loss of reading, loss of not being able to see wonderful sunsets but only gray...so many losses.” – Tom, comment submitted online*

**Individuals living with XLRP emphasized how progressive visual impairment differs from blindness.**

*“The slow journey into sight loss presents a series of physical and emotional challenges. Learning how to effectively use a white cane and travel with*



*confidence is a daunting task on its own. And summoning the courage to begin using a cane in public for the first time is much more difficult than it may seem. Beyond the practicality of the white cane as a tool, it also represents a notable shift in identity. And I think a lot of people who grow into sight loss, struggle with this transition.” - Darryl Adams*

The poll results are presented in **Appendix 6, Q1 & Q2** and described in the sections below.

### **Difficulty adapting to changes in lighting**

This XLRP-related health concern was experienced by 100% of the individuals living with XLRP represented by the poll results and was selected as one of the top three most troublesome XLRP-related health concerns by 61% of respondents. Difficulty adapting to changes in lighting impacts all aspects of life. Many described how the increased light of summer makes life easier for them.

*“The transition from bright to dark, it affects all of the things that I deal with, leaving my house, going into businesses, going into restaurants/stores, whatever the case might be.” – Robert (caller)*

*“There are lots of times where the sun is coming down in the earlier evening or later afternoon and it's so bright that if my windshield isn't totally clean, I will get a whiteout experience. I just can't see anything. ...I'm finding it's this light/dark game that I'm playing. Sunglasses on, oops, sunglasses off, regular glasses on, quick because we're going through a tunnel or the light level is changing because of trees. So, it's kind of crazy to have both glasses ready there, whipping one out after the other, but that's the life I've started to live.” - Lisa Lloyd*

### **Decreased night vision**

Decreased night vision was selected as the most troublesome XLRP-related health concern by 76% of respondents and is experienced by total of 94% of the individuals living with XLRP represented by the poll results. Often decreased night vision was the first symptom of XLRP.

*“I first remember noticing there was something wrong with my vision. I remember playing tag or hide and seek with other children. And I couldn't*

*understand how they could see at night. I would just stand there and they would just disappear into the night to go hide.” - Joaquin Cornejo*

*“I would say my loss of night vision, trouble distinguishing colors and inability to process images [are the] most significant symptoms. Losing my night vision has made it very hard for me to function in dimly lit situations, having trouble distinguishing colors and processing images makes it very hard for me to see pictures, watch TV or recognize faces.”- Jeffrey, comment submitted online*

*“Every major decision in my life has been dictated by the ticking time bomb that is my vision. There are no breaks, there's no time for rest. Disease is insidious. Each day, I see slightly less than the day before. I gave up driving at night one year after obtaining my license. The list of ways RP has impacted my life is endless, not driving at night was just the beginning.” - Jonathan Tamaiev*

### **Loss of peripheral vision**

Loss of peripheral vision is experienced by 84% of the individuals represented by the poll results, and selected by 58% as one of their top most troublesome XLRP-related health concerns.

*“The most significant impact of XLRP on my life is the loss of peripheral vision. The most significant effect of that is not being able to drive. This is a big loss of freedom.” – Aaron, comment submitted online*

*“My son was diagnosed at age five. He is now 19. And although he tries to remain positive, the peripheral and night vision loss has certainly impacted both him and our family over the years.”- Kim, comment submitted online*

Joaquin described how he adapted to the loss of his peripheral vision. *“I would have to scan constantly and turn my whole head left or right just to see. But even then, that was not good enough.” - Joaquin Cornejo*

### **Discomfort from bright lighting**

Discomfort from bright lighting, also known as photophobia is experienced by 69% of the individuals represented by the poll results, and was selected by 24% as one of their top three most troublesome XLRP-related health concerns.

*“The sun started to get too bright for me, even with prescription sunglasses.” - Joaquin Cornejo*

Lisa’s extreme photophobia caused her to pass out during eye exams and performances. *“I’m living a life where I’m not sure where bright lights are all of a sudden going to intervene”.* - Lisa Lloyd

### **Anxiety or depression**

Anxiety or depression is experienced by two-thirds, or 66% of the individuals represented by the poll results and was selected by 24% as one of their top three most troublesome XLRP-related health concerns. The reasons for vision-loss related anxiety and depression were varied and ranged from the deterioration of self-esteem and self-confidence, safety concerns, and living with the knowledge that they would gradually go blind by middle age.

*“By the time I got my diagnosis (at age 14), I had low self-esteem, seriously considering trying to kill myself because I couldn’t participate in life like my brothers and sisters and my friends, and classmates. Can’t play hide and go seek, can’t see in the dark, I was horrible at sports. So, getting the diagnosis was a relief because it was my eyes and not me.” – Robert (caller)*

*“I face anxiety, depression, anger, and frustration constantly. This interferes with every social interaction and relationships with my family. I continually struggle to suppress these feelings in order to be a normally functioning human being. Those feelings are not me, and it’s a constant fight to be the positive upbeat person that I truly am inside.” – Paul 3, comment submitted online*

Greg described how his wife’s vision loss caused anxiety, *“Whenever she was in an unfamiliar environment. Stairs, traffic, obstacles are drivers of anxiety then and are especially now.”* - Greg Dubecky

*“The depression was related in being legally blind because it’s a slow progression and then eventually, you hit the wall where you’re just not able to function anymore. So, I hit that wall and then I wasn’t working. So the loss of the job, loss of [being] the breadwinner, so it all came down on me and the depression hit really hard.” – Allan (caller)*

Michael described his increased anxiety. *“The first time that I really noticed it -- where I would feel my hands clenching and becoming fearful -- were in more social situations, walking into a room where you have no idea what the lighting may or may not be. And of course, now you're going to be in a scenario where there may be food, knocking things over, spilling things.”* - Michael Stone

**Many individuals with XLRP live with the knowledge that they will be blind by middle age.**

*“I learned about [XLRP] when I was in grade school. And they told me by the time I was 35 or 40 I could go totally blind.”* - Dave (caller)

*“I knew walking into the ophthalmologist office, that the chance of me being diagnosed with RP was high, but when the words hit me and I was informed that I need to seriously reconsider my future and brace for blindness by the age of 40, I was devastated for a while. I was mad, sad, and upset with the world.”* - Jonathan Tamaiev

**Loss of central vision**

Loss of central vision is experienced by 59% of the individuals represented by the poll results, and was selected by 39% as one of their top most troublesome XLRP-related health concerns. The loss of central vision is a more advanced symptom of the disease for some individuals living with XLRP.

*“Now that I've lost my central vision, I consider myself blind. Want to know how it looks? It's a lot like watching an old black and white television tuned to a channel with a weak signal. Everything is flickering, static, gray and indistinct. I see something, but I almost never know what I'm looking at.”* - Paul Martz

*“One of the most significant symptoms for me is the loss of my central vision...no detail, fuzziness...like being in a white fog or a blizzard.”* – Tom, comment submitted online

*“My lack of central vision is this inability to actually read labels in the grocery store or find the signs that say where I need to go.”* – Robert (caller)

*"I don't even bother to rely upon my remaining eyesight much. Most times I close my eyes, so I can concentrate better. My tiny bit of eyesight is more of a hindrance than a help." - Robert Pawlick*

### **Other XLRP-related health concerns**

While no one selected experiencing "other" XLRP-related health concerns as a poll response, additional health concerns were mentioned throughout the meeting and 6% indicated that their "other" XLRP-related health concerns were among their top three most troubling. Individuals living with XLRP described their overwhelming fatigue, cataracts, glaucoma, sleep disturbances and physical injuries.

- **Overwhelming fatigue**

Many meeting attendees described how their vision deterioration contributed to overwhelming fatigue, not only for their eyes, but in the entire body.

*"I'm a teacher, so a good amount of my sight, I'm looking at text. Whether it's on paper, on a screen. ...I'm in the middle of my job, halfway through and my eyes are tired regularly." - Andrea Bullock*

April's son is living with XLRP and his colleagues don't understand why he need to take frequent rest breaks. *"His eyes get super tired and then it affects his whole physicality." - April Lifriu*

*"It's more than just your eyes getting tired. It's what happens to your...physicality and your soul. ... The fatigue was a surprise to me. ...I can't over emphasize that enough, what a shocker that is and that how each one of these little challenges that we're all noting forces us into a darker place, just because of that level of exhaustion. I find that even life's most simple tasks, ... I got to give myself a time out and sit down and break." - Michael Stone.*

- **Cataracts and glaucoma**

*"I ended up getting cataracts which is common. And then when I had my cataract surgery, I ended up with glaucoma which was one of the side effects that can happen. I had a lot of complications, and I actually am due to have cataract surgery in two weeks." – Susan (caller)*

- **Sleep disturbances.**

*“For many years, right around dusk, I would start to wake up, right when it's time to start getting ready for bed. ...Now all of a sudden, your sleep patterns are completely off and anybody knows if you can't sleep, you're not good for anything.” - Michael Stone*

*“If you can't sleep, it makes everything harder during the day. ... If I don't sleep well, my vision's bad the next day.” - Lisa Lloyd*

- **Physical injuries**

*“While vision loss and night blindness are the major symptoms of RP, the trauma produced by dealing with these symptoms is the most challenging symptom of all. All of the bumps and bruises are worse, from spills and accidents leave marks on the body and spirit.” – Adam, comment submitted online*

*“My parents noticed that I would often trip or bump into things when moving into dimly lit areas, such as a movie theater, or even just around the yard or house at night.” - William Hartman*

*“We live in a world right now where people are very distracted. They're not paying attention. Whether or not I'm with a service dog or with a cane, they don't recognize what that is. So, what's happening is that I've been in almost this state of fight or flight because it's a constant safety issue. Cars are not stopping at stop signs, where you expect them to be. People just bumping into and can be quite violent, actually.” - Michael Stone*

*“My dad was just hit [by a car] last month. Most people who are blind, at some point, do get hit by a car in my experience because people are very distracted. This is a huge problem. A lot of the cars are becoming close to silent, which makes it even harder for our population.” - Lisa Lloyd*

**Impacts:** Driving, participating in social life and reading are the top three activities of daily life affected by XLRP

Poll respondents used online polling to select their top three activities of daily life that are most important to them or their loved one struggles with due to XLRP. Poll results are presented in **Appendix 6, Q3** and illustrated with patient comments below.



## Driving

Driving was the top choice, selected by 84% of the individuals represented by the poll results, as a specific activity of daily life that is important, but they are not able to do or struggle with due to XLRP. Many individuals living with XLRP described how they first lost their ability to drive at night and then the day, as their vision diminished. The inability to drive is closely tied to the loss of independence and the erosion of self-esteem.

*“My night driving diminished to a point where I was very dependent upon the painted lines on the road in order to stay in my lane. It was quite difficult when meeting cars on a two lane road at night. My driver's license was taken away after I had an accident at age 22 when the state found out I was legally blind. I had to rely on friends, family, and public transportation thereafter.”* - Comment submitted by the Foundation Fighting Blindness on behalf of an individual affected with XLRP

*“Losing independence was mostly from not being able to drive, [as a result of] the night blindness and the limited visual field. That's such a big adjustment. Losing one's license and not being able to drive and having to depend on other people and services ... to get around, that's just huge.”* - Rod (caller)

*“None of this journey has been easy due to my inability to drive. At night, I rely on public transportation to commute for almost two hours each way across east Brooklyn.”* As a medical resident, Jonathan was required to do long, overnight shifts and selected his residency based on the availability of public transportation. *“I did not feel comfortable living in a city where I could not safely rely on public transportation.”* - Jonathan Tamaiev

William described how vision loss continued to surprise him. *“Surprise, I'm taking away your ability to drive. ... No more driving to work, no more driving to get groceries, no more driving for kid drop offs or pickups at school. No more driving the kids to practice or games.”* - William Hartman

For April's sister, *“Driving was her biggest impact. I think she went through a decade of depression trying to accept that and now she's in that accepting stage.”* - April Lifriu

### Participating in social engagements or social events

Participating in social engagements or social events was selected by 50% of the individuals represented by the poll results as a specific activity of daily life that is important, but they are not able to do or struggle with due to XLRP. Many described feeling socially isolated, and listed reasons including not being able to drive, the inability to negotiate crowds, or places including public washrooms, and by a lack of self-confidence.

The loss of peripheral and night vision loss has impacted Kim's son socially. *"Things we take for granted on a daily basis, inability for your child to participate in his fifth grade dance because the lights were dimmed and he became frightened, embarrassed and frozen in a corner because he could no longer see."* – Kim, comment submitted online

*"I have lost the ability to recognize people's faces. I can very rarely make eye contact, and this has all been happening to me over the past two, two years or so. It's all very new and very fresh to me."* - Mark Curley

This was another unpleasant surprise presented to William by vision loss. *"Surprise, starting today under all lighting conditions, I'm going to make you look very clumsy in a public crowd of people. Try not to bump into them so much. I suggest you look around yourself three to four times every time before you dare take a single step. Do you feel the confidence wound bleeding, getting bigger?"* He added, *"The pain will be ever present and will spread as you realize people increasingly isolate you and treat you differently."* - William Hartman.

*"My biggest fear is having to use a public restroom. They can be very dark and the different layouts can cause me big issues. I feel like a freak using a bright flashlight to navigate them. I would rather stay home."* – Mike 1, comment submitted online

Patricia's son, *"Always made friends so easily, but his college friends now live all over the country. ... Making new friends is much more challenging. He is unable to participate in the activities he could do as a teen."* – Patricia, comment submitted online



## Reading

Reading was the third most impacted activity, selected by almost a third, or 31% of the individuals living represented by the poll results as a specific activity of daily life that is important, but they are not able to do or struggle with due to XLRP.

*“The biggest way this impacts my life, I can't read. ...I have a seven-year-old grandson and I'd love to read aloud to him, but without central vision, I can't read print, not even large print books. ... Basically without central vision, I have no good options for reading to my grandson.”* - Paul Martz

*“My visual acuity is still very good, but my central vision has shrunk so much that I will soon no longer be able to read. This will truly be life altering, and cut me off from so much of the world.”* – Laura, comment submitted online

Tim's inability to read limits his participation. *“I wasn't even able to fill out the poll questions. I mean, I can't tell, I couldn't see them or even find out where I was supposed to go to fill out the poll questions.”* - Tim (caller)

## Attending school or having a job

Attending school or having a job was selected by 28% of the individuals living with XLRP represented by the poll results as an important activity of daily life impacted by XLRP.

### **Many described challenges getting their needs recognized and met in educational situations.**

For Joaquin, *“Even when I was in grade school, other kids made fun of me for bumping into stuff. But they didn't know my situation. Neither did I. I really didn't understand it. Even the teachers didn't know, they just sat me in front of the classroom and that was as best they could do for me. It was still challenging.”* Things became even more difficult at high school. *“In high school they used a white board. Even sitting in the front, I really couldn't see what was on the board or even watching movies in school when they were turning off the lights. ... Graduating high school was really challenging due to my vision and my RP.”* - Joaquin Cornejo

*“The schools don't give sometimes the right support. We had to sue our school district because [our daughter] couldn't get the support she needed. Time and time again, the schools don't understand what that young student who's losing their vision needs. They're clueless because it's 1% of the population that's blind.” - Lisa Lloyd*

**For many, vision deterioration impacted their employment, created challenges in finding new jobs, and resulted in workplace injuries.**

Vision deterioration interfered with William’s work. *“You just want to be able to do your job like everyone else, but you find it difficult to concentrate. You're bereft of your independence, having such little confidence, such little pride, and self-esteem left, you're consumed by your thoughts. - William Hartman*

Changes in lighting impacted Michael at work. *“I will tell you that work has been amongst the toughest because that was something that you would think that you would be able to control because it would be something constant, but even there, it wasn't.” - Michael Stone*

Allan, a physical therapist, is having a hard time finding employment. *“I've never had problems getting work before so I was applying for jobs and went on five interviews but what happened was I hit the wall and became legally blind. And so, I was bumping into things. I was being very careful during the interviews and then, I wasn't hired. I went from working to not working.” – Allan (caller)*

*“I tried working at a body shop, but I had to stop working there because I kept bumping into stuff, hurting my shins, cutting myself. So, I just stopped working there entirely.” - Joaquin Cornejo*

**Several experienced stigma and workplace discrimination.**

*“I don't look forward to the task of trying to put [my colleagues] at ease with my visual impairment or the various difficulties in social interaction. This usually entails me making an announcement about my disability many*

*times in asking various people for assistance all the while, putting on a strong face and assuring them that I'm fit for the job.” - William Hartman*

Paige’s fiancé’s medical profession refused to acknowledge or accommodate his vision loss. *“He has this incredible skill but wasn't able to meet these other criteria that would've had nothing to do with the path he was going. ... He was not able to complete residency because of his vision. Therefore, he is not board certified and that has limited his job opportunities.” - Paige (caller)*

**Many described how XLRP influenced their career choice or how they found specific opportunities for visually impaired people.**

After graduation from medical school, where he had a position working in a retinitis pigmentosa research lab, Jonathan said, *“I proceeded with a career in psychiatry, a field of medicine where I could have a lasting clinical role without my vision.” - Jonathan Tamaiev*

*“Believing I needed to be more competitively employable because of my failing eyesight, I got grants and student loans and earned my MBA from Thunderbird, rated the number one international management school in the world. ... “I'm in training to become a vendor through Tennessee Business Enterprises program, TBE. This is possible due to the Randolph-Sheppard Act, 1936, which grants blind vendors the exclusive right to operate vending machines on federal, state and other properties. - Robert Pawlick*

*“I applied for vocational rehab and disability ... I received a training through a foundation for Blind Children/ Adult Services transition program here in Phoenix and then, mobility and computer skills. And so, I transitioned to IT working with Blind Institute of Technology in Salesforce training program. And so, it made everything better, got back to work.” – Allan (caller)*

**Some were forced to retire or take disability.**

*“Being diagnosed with RP, I was encouraged to pick a career compatible with going blind. I chose computer programming. I used ZoomText screen reader and a CCTV to read papers & books, as well as fill out paperwork from age 18 to age 43, at which time making things bigger wasn't working*

*anymore, and I took disability.”* - Comment submitted by the Foundation Fighting Blindness on behalf of an individual affected with XLRP

*“I’m 52...I retired from work at the end of last year because of my vision.”* - Matthew Bacho

*“Taking disability meant leaving the workforce, taking a cut in pay and ending up reversing the roles of me and my wife. She had to go to work to help us get by. ...Learning to give things up and accept, it is a big part of having RP.”* – Rod, comment submitted online

### **Participating in sports or other recreational activities**

Participating in sports or other recreational activities was selected by 28% of the individuals represented by the poll results as an activity of daily life impacted by XLRP. Many described how playing ball was often unsafe.

*“I used to be incredibly active and tennis, driving, skateboarding, you name it. I used to do a lot of it and thoroughly enjoyed it. And these are things that just disappear as retinitis pigmentosa does his job on you.”* – Mark Curley

The loss of night vision affects Polly’s son. *“It has impacted him playing with friends outside at dusk or later in the backyard, navigating a movie theater on his own and feeling safe in his room at night.”* – Polly, comment submitted online

For Greg’s son, *“Marty was never able to play sports he wanted to as a younger child. Any sport with a ball was impossible and often unsafe.”* - Greg Dubecky

*“RP’s biggest impact on my son’s life so far has been in physical education class in his elementary school. Whenever they play ball sports, he struggles significantly and his classmates are too young to understand his difficulty seeing balls thrown at him.”* - SW, comment submitted online

### **Some described successful adaptations.**

*“I bought an indoor bike because I couldn’t bike anymore.”* – Susan (caller)

*I've led an active life and I played soccer as a child. And as my eyesight began to get worse, I gravitated towards sports that focused more on individual achievement, such as skiing, running and cycling.” - Darryl Adams*

### **Travel or vacationing**

Travel or vacationing was selected by 25% of the individuals living with XLRP represented by the poll results as an activity of daily life impacted by XLRP.

*“Travel is difficult. It can be done but the logistics are so much more difficult.” – Patricia, comment submitted online*

*“My worst days come when I am in different environments that may require me to travel. I have to rely on Uber or Lyft to travel that can be very unreliable (leading to frustrating situations). When in unfamiliar environments, I may bump into things, get lost, need help from others, and I often get myself into embarrassing situations.” – Jeffrey, comment submitted online*

Lute, Richard’s service dog, described how, *“Denver Airport is one of our biggest challenges. When we go by ourselves on a trip, we navigate together through a lot of people to find our departure gate. Dad holds onto my handle and leash as we slip in and out through the crowds with him asking, ‘Is this gate 32?’ When we fly back to Denver, I find the escalator from the gate area down to the train. He’ll say, ‘Hop up,’ and I move with him onto the escalator and down we go. We follow the crowd onto the next train and off it goes to baggage claim. Everybody is in a rush to get off, go up the escalator, get their bags and go home. We just go with the flow.” - Lute, Richard Faubion’s seeing eye dog*

### **Self care or chores**

Self care or chores was selected by 19% of the individuals living with XLRP represented by the poll results as an important activity of daily life that is impacted by XLRP.

*“As everyone with this disease does, I struggle daily with normal tasks and my world keeps getting smaller and darker.” – Denise, one comment submitted online*

*“Well, right now there's not a whole lot I can do. I mean, I'm lucky if to be able to fix some of my own meals. And I mean, I have to have someone come in and clean my house and I'm not able to watch TV.” - Tim (caller)*

Difficulties in adjusting to lighting can create safety concerns. *“It could be something as innocuous as putting toothpaste on your toothbrush or trying to cook your food in a kitchen where there's a safety concern to it.” - Michael Stone*

*“In my personal life, some of my most difficult days are just spent trying to triage paper mail. It's recently become very difficult for me to read regular mail and it triggers a fear in me of completely losing all vision one day.” - William Hartman*

### **Walking or ambulating**

Walking or ambulating was selected by 16% of the individuals living with XLRP represented by the poll results as an activity of daily life impacted by XLRP. During the meeting, many described using a service dog or a white cane when walking.

*“I used to be fine during the day walking around by myself, but now I use a walking cane just so I don't hurt myself.” - Joaquin Cornejo*

*“I also can't navigate a new place by myself if the lighting is poor. I do not have training with a white cane, so I am currently holding onto a person I know while walking around. – Ava, comment submitted online*

*“I can't walk to the car and get something out of the car on my own because it's very dangerous. When you're visually impaired, if you take the wrong step, you could fall, you could break a bone. And I've literally gotten to the point now where I rely heavily on having a shoulder or an elbow to hang onto when I'm in public.” – Mark Curley*

### **Other**

Other activities of daily life were selected by 9% of individuals living with XLRP represented by the poll results, as being impacted by XLRP. Many individuals living with XLRP have gradually lost their independence and are now dependent on family members. Loss of independence is also the top worry and is addressed by the next poll question. Many of the meeting attendees experienced vision-loss

related stigma and a lack of understanding. Some even described being made fun of by others at school and at work.

*Vision loss caused Greg's son to feel embarrassment on vacation. "We entered a dimly-lit restaurant at the end of a great day at the beach. The restaurant lighting was not something our son previously had difficulty with, but even at his young age, he lost enough low vision by this time in his young life that he couldn't navigate well enough to avoid a pole in the middle of the room, running face-first into that pole. Sadly, this was to the amusement of his friends and unknown patrons in the restaurant, both thinking he was simply awkward and not that he had challenges they didn't. Embarrassment followed but fear dictated the remainder of the vacation and carried over into regular life." - Greg Dubecky*

*"What we all suffer is ignorance, discrimination, and exhaustion from having to constantly re-learn how to navigate a society that is not designed to support us. ... I am living an alternative experience. I have a disability because the world tells me I am disabled. I believe I am just living a different way than folks with working retinas." – Adam, comment submitted online*

### **Spending time outdoors**

Spending time outdoors was selected by 3% of the individuals living with XLRP represented by the poll results as a specific activity of daily life impacted by XLRP.

*Joaquin is now unable to spend as much time outside with his children. "I was really outdoors with them, and I can't do that no more. I can't go outside and ride a bike. I can't play basketball, softball. I just can't do outdoor walks with the family. It's just really hard." - Joaquin Cornejo*

*"After 20 years as a master gardener, I am now giving up gardening because one, sunlight is too bright and two, I can no longer distinguish between plants and weeds." – Laura, comment submitted online*

**Top three worries:** loss of independence, not knowing how XLRP will progress, and impacts on relationships with family and friendships/not being able to care for family.

Poll respondents used online polling to select their top three worries about XLRP in the future. Poll results are presented in **Appendix 6, Q4** and illustrated with patient comments below.

### **Loss of independence/who will care for them in the future**

Loss of independence was the top worry overall, selected as a top three worry by 90% of poll respondents and this worry generated many comments throughout the meeting and online. This included a closely related worry about who will care for them in the future, as many living with XLRP depend heavily on their partner and children.

*“The constant fear of when I will lose another crucial means of independence lingers over me. What will happen when I can no longer drive at all? What will happen when I can no longer see my kids' faces? What will happen when I can longer see my computer screen, I cope by proactively taking steps to secure and maintain my independence, but I live with a constant fear that one day I won't be able to stay in front of it.” - Jonathan Tamaiev*

*“It's very humiliating in certain situations that I can't do things on my own. ... I'm still trying to learn this thing, how to just kind of swallow my pride and rely on people to help get me around.” – Mark Curley*

*“I have become more reliant on my wife for certain activities such as organizing clothing, medication and financial management.” – Paul 2, comment submitted online*

*“Although my husband is my helper, I feel very isolated at home, and worry about how I would cope if something happened to him.” – Laura, comment submitted online*

Tim's eyesight is getting progressively worse, *“And it's just a tremendous worry because when I go totally blind, I mean I don't know what I'm going to do. I have to depend on my daughter to take me anywhere. Well before,*



*my wife used to, but she passed away 14 months ago. It had to be her for pretty much the last three years, and it was just very difficult.* - Tim Hill

*"I'm pretty fortunate in the fact that I have a wife that has sight. So my concern is if someday when that... if that could change. ...Life, you never know who's going to be around one day to the next."* - Rod (caller)

### **The stress of not knowing how XLRP will progress**

The stress of not knowing how XLRP will progress was selected as a top worry by 71% of those living with XLRP represented by the poll results. Many of the concerns were focused on loved ones – especially children – living with XLRP. Others expressed fears for going completely blind in their lifetime.

*"My internal dialogue goes, as it has for the past 28 years, wondering, waiting, worrying about my wife and son's progressive vision loss because of XLRP. For half my life now, this insidious disease and how it affects loved ones, friends, and others is the first thing I think about in the morning, what I think about regularly throughout the day, and the last thing I think about before sleep. Witnessing one loved one struggle with progressive vision loss is not something I wish on anyone. Witnessing three is nearly unbearable."* - Greg Dubecky

*"It is a feeling of not knowing what the future holds that I feel is the most difficult aspect of the disease. Living with XLRP is a lifelong series of adjustments, and continuous learning and adaptations, but never mastery."* - Darryl Adams

*"On my worst days, I'm consumed with worry. Will a treatment be approved in time to help my son? Will we be able to find a clinical trial accepting new pediatric patients? I'm terrified he will lose his vision before a treatment is available and then it'll be too late."* – Polly, comment submitted online

*"I'm losing eyesight at a tremendous level, and it's really, really worrying me. ... My greatest fear is that I'm 66 years old, and I'm afraid that the cure is not going to be found in my lifetime and that I will go totally blind."* - Tim Hill

*“The hardest thing for me has been the anxiety of anticipating vision loss in the future. While that’s given me a great deal of anxiety it has also set my life in a direction that I would not change for the world.” – Bryson, comment submitted online*

### **The impacts of relationships with family and friends / Not being able to care for your family**

The impacts of relationships with family and friends and not being able to care for your family were selected as top worries by 39% and 35% of those living with XLRP, respectively. Many comments were submitted by individuals who felt that they were not fulfilling their role in supporting their families or caring for children as they wanted to.

*“My family don't know the full extent of my vision. I haven't told them. I'm just trying to be strong for them and not let them worry about me. ...My daughter tries to show me stuff, she's really excited, and I just can't see what she's showing me. Or if me and my wife have date night, she asks me ... how does she look and does she look pretty, and I really can't tell what she looks like. I just tell her she looks beautiful. ... XLRP, it's just killing me inside.” - Joaquin Cornejo*

Andrea has two daughters, *“My biggest fear is that I will hinder them from being able to be successful in their development, and with their desires, and growing into healthy young ladies.” – Andrea Bullock*

*“My dad went blind when I was three, so I started taking care of him when I was four. My mom left and that's pretty common too. The spouse will often leave when the other spouse goes blind. I don't think that my husband will leave me, but it definitely is a bit of a concern.” – Lisa Lloyd*

*“When my wife and I married it was expected that we would share the responsibilities and day-to-day life; that has changed for my wife as she has had to take on more and more of the everyday life ‘things’. It can be very tiring for her. We don't always see eye to eye on things due to the fact that I cannot see things as they are.” – Tom, comment submitted online*

*"As a grandmother, I cannot read to my grandchildren, admire what they want to show me, or even babysit without assistance." – Laura, comment submitted online*

*"I'm a caregiver for my dad who's 81 and blind with X-linked RP, I'm a caregiver for my daughter who's totally blind and 15 years old with LCA, and I'm losing my vision, right? My concern is, how am I going to continue to care for everybody?" - Lisa Lloyd*

### **Having to change jobs or stop working**

Having to change jobs or stop working was selected as a top worry by 26% of those living with XLRP represented by the poll results.

*"About the only vision I have left is a little ability to see my computer screen to continue to do my job, and that's getting more and more difficult. ... I am concerned about how long am I going to be able to do the job that I do, sitting in front of a computer screen all day long. I have skills, I have intelligence, I have the ability to work, and yet the challenges for folks with low vision and blindness in this country right now, even around COVID, is challenging, very, very challenging." – Robert (caller)*

*"My fiancé gets fatigued quickly. He works on a computer for 10 to 11 hours during the week. ... The stress of work and trying to always prove himself really greatly affects his mental state and anxiety. And he is very worried about not being able to see his computer one day." – Paige, comment submitted online*

### **Many worried about how their children will support themselves in the future.**

*"His visual world is closing in on him daily as he prepares now for a career, which in itself is difficult for someone with a visual impairment. He also needs to consider a career that he can adapt to. This weighs very heavily on [my son]." - Greg Dubecky*

*"He's 22 and he doesn't know what to do with his life. There's a sense of feeling inadequate because so much in life is depending on your vision and your eyes. He's not a computer person, he's not a tech person, so he's not*

*going to fall in that category. He's a hard worker. ... This disease can take a toll like on your ego and your ability and your confidence.” - April Lifriu*

*“He is currently employed, but as his ability to read becomes more challenging what will become of his job? He has a graduate degree and a Masters degree but I know that blind individuals are among the most underemployed. As parents we obviously worry about his long-term future.”*  
– Patricia, comment submitted online

## Family planning

Family planning was selected as a top worry by 19% of those by the poll results. Many were worried about passing XLRP to their future children, and several described adoptions and using IVF to conceive.

*“We found ourselves not only wondering what this disease could mean for us but also our future family, leaving us with the very unenviable decision as to whether we should have children of our own. ... What's more is we'd be putting our children in the same position we're in, deciding if growing a family was the right thing to do.” - Greg Dubecky*

*“Realizing that scientific progress regarding genetic treatments and cures was still decades away, we took matters into our own hands. We adopted all of our children. ... Adoption can't truly prevent the disease from being within the genetic makeup of our children, but adopting did effectively eliminate any chance of RP being passed from me to our children.” - Robert Pawlick*

*“I now face the decision of using IVF for all future pregnancies.” - Jonathan Tamaiev*

## Other worries

Other worries were selected by 10% of the of those living with XLRP represented by polls. Other worries included injuries on the job and in the world, vulnerability, social isolation, alcohol abuse, safety in emergency situations.

April worries about her son's safety at work. *“Every day I fear that he's going to get hurt “because it's a dangerous environment, but he jumps in and he's willing to work.” - April Lifriu*

*“As we lose our independence, we do become more vulnerable. And I don't mean that just in a physical way, but in many other ways. For example, it's just been brought to my attention that my dad's being financially taken advantage of and someone's using his credit cards again and again and again. And when you hand over that power, you have to be careful. Sometimes even in the hands of other family members, you become very vulnerable in lots of different ways.” - Lisa Lloyd*

*“The other thing that worries me is my dad's drinking. The isolation, ... can easily lead to drinking and depression so I think those go hand in hand. And then, that's not something we've talked about too much [in the meeting] but it is pretty common in the population.” – Lisa Lloyd*

*“Graphical information is inaccessible to me. ...Many health websites presented COVID data as graphical charts that I couldn't see. It was bad for me mentally. I wanted to know, was the situation in my county getting better or worse? It made me feel like we've got this global pandemic going on, but we're not going to tell the blind people how bad it's getting.” - Paul Martz*

*“I live here in Colorado and in January, the town I live in was devastated by fires and many of my friends lost their homes. But if that was my situation and it was dangerously close to it being me and living in an area where one doesn't expect that to happen, that would've been it. I wouldn't have been able to navigate that. ...I would've been dependent on somebody actually coming and rescuing us.” - Michael Stone*

## Session 2: Current & Future Approaches to Treatment for XLRP

*"No matter how I adjust and adapt and train, whether it's cane training or OT at a blind rehab center, or prescription drugs for anxiety, or training to use computers and smartphones, the bottom line is that I'm still blind." –*

Paul 3, comment submitted online

### Limited medications and medical treatments are available to treat the symptoms associated with XLRP.

Poll respondents used online polling to select all medications or medical treatments that they or their loved one have used, currently or previously, to treat symptoms associated with XLRP. The poll had few options to choose from, and respondents only selected an average of 2.6 responses. Poll results are presented in **Appendix 7, Q1** and illustrated with patient comments below. Individuals living with XLRP also described some of the specific downsides and trade-offs that they were willing to tolerate in exchange for the opportunity to improve their vision.

#### Vitamin A

Vitamin A was the top selected medication option, selected by 75% of the individuals represented by the poll responses. Some of the downsides mentioned included lack of efficacy and side effects, including the potential for liver damage.

*"I did try the vitamin A palmitate. I don't really know how well it worked, but I didn't really like the idea of having to have my liver tested every three months. Once I started got to my childbearing years, I stopped taking that." – Susan (caller)*

*"The very first trial I was in, through the National Eye Institute at NIH, was vitamin A. And I'm assuming that my expectations were not very high, but I don't remember any improvement at all." - Matthew Bacho*

*"I've tried various things throughout the years. I still to this day take vitamin A with palmitate, and I also take DHA, several other vitamins." - Mark Curley*

## Investigational therapy in a clinical trial

Investigational therapy in a clinical trial was selected by half, or 50% of the individuals represented by the poll responses. Many panelists and discussion starters described their participation in gene therapy and stem cell trials, and most were enthusiastic about the opportunity to advance the science, even participating as controls. Several shared their hope that their treated eye had less vision and light loss than in the untreated eye.

Mark was optimistic about the initial results of his gene therapy trial. *“They treated just my left eye only, and I'm coming up on my three-year checkup at the end of this month. And the goal of the trial in the treatment is to halt any further progression of the retinitis pigmentosa. ...I just have a lot of hope that it is working and that it will not progress any further.”* - Mark Curley

Matthew's gene therapy trial was his fourth trial. *“Not only am I interested in improving, if I can, my vision, but I'm also interested in advancing the science. My experience with the gene therapy has been interesting. My background is science, and so I understand it to a great extent and I was really excited to be a part of that.”* - Matthew Bacho

Richard didn't experience positive results from his stem cell trial, but he felt participating was important. As dictated by his guide dog Lute, *“He knows there are risks with a new treatment, but without a risk, there could not be any vision gained. He got some edema from the stem cell trial, but only had 20/400 vision then and the risk was worth the investment of two years flying back and forth to the trial site.”* - Lute, Richard Faubion's seeing eye dog

Investigational trial downsides: only one eye can be treated at once, arduous testing requirements, the need for reduced expectations, non-optimal dosing, limited eligibility.

*“The flip side of this is, my right eye is still untreated, and it is progressing. ... I hope and I pray that we can get both eyes treated very shortly.”* - Mark Curley.



*“Tests indicate that the sensitivity in the treated eye to light has improved, but it does not translate into my day-to-day activities. Now that I've lowered my expectations and I've got a chance to look at the physiology, the infrared photography, photographs of my retina, it appeared to me that perhaps it wasn't getting thinner at the same rate as the untreated eye. And I haven't received any information about that, but right now, I think my goal is, the realistic goal is to slow down progression, at least.” - Matthew Bacho*

*“I had gene therapy in one eye in 2017. ... The treated eye also took a one-off hit from the invasive nature of the surgery itself. And however, I only got a low dose, and I am confident that once optimal doses have been found, gene therapy will be effective, especially for younger patients. At 46 at the time, I was probably too old to benefit from gene therapy.” – Mike 2, comment submitted online.*

*“I even tried to qualify for a clinical trial, and they told me my eyesight was too bad.” – Tim Hill*

### **Other medications or supplements**

Other medications or supplements not listed in the response options were selected by half, or 50% of the individuals represented by the poll responses. These included vitamin and mineral supplements, DHEA, lutein and zeaxanthin and sleep medications.

*“Ever since my diagnosis 34 years ago, I've been taking a daily regimen of vitamins and minerals believed to help slow RP's impact on remaining eyesight. Though considered a homeopathic treatment, this combination includes FFB and doctor recommended vitamin A palmitate. My eyesight has lasted 20 years longer than my great uncle's. I believe these supplements are solely responsible for slowing my RP's deterioration.” - Robert Pawlick*

*“I take higher levels of DHA lutein and zeaxanthin daily, general liquid vitamins and eat mostly organic food, drinking a lot of filtered water.”–Lisa, comment submitted online*

*“VisiClear, ... It's 10 different kinds of vision kinds of vitamins. ... I take two capsules in the morning before eating or anything. I've been doing it now*



*for four and a half months and now my peripheral vision, things are getting brighter. ... And I just noticed that things are brightening up, and I'm very encouraged.” – Dave (caller)*

### **Medications to manage anxiety or depression**

Medications to manage anxiety or depression were selected by 30% of the individuals represented by the poll responses. Some described how anxiety medications unexpectedly helped their vision.

*“RP took to on me physically and mentally. I gained 90 pounds and I'm now taking medications to treat anxiety, depression, diabetes, high blood pressure and high cholesterol.” - Robert Pawlick*

Susan described some alternative approaches to deal with her anxiety. *“Whatever it is that I'm doing that's maybe making my anxiety better, my vision is just so much more clear. ... I was taking this Nordic brand fish oil, and I also started doing this tapping thing. It touches the pressure points, and I have to say I feel I don't have as bad a vision days. I do feel like my vision is clearer which I know sounds really crazy, but it's working. And I do feel that our vision can be tied to how we feel. And I think when we are stressed, it compounds not being able to see well.” – Susan (caller)*

### **Cannabidiol or CBD**

Cannabidiol or CBD was selected by 20% of the individuals living with XLRP represented by the poll responses, but there were no comments made during the meeting or submitted online.

### **I have not used any treatments recently**

This option was selected by 20% of the individuals living with XLRP represented by the poll responses. Many, like Tim, were told that there are no treatments available for XLRP.

*“Every time I go to the ophthalmologist or I even went to the Wilmer Eye Institute at Johns Hopkins, and they just tell me that there's nothing they can do for me.” - Tim Hill*

## Argus II

Argus II retinal implant was selected by 10% of the individuals living with XLRP represented by the poll responses.

*Lisa Lloyd's father received his implant in 2007. "He was able to get back some usable vision after not having any for 25 plus years. We were really excited about that. The vision that he was able to get back allowed him to mobilize much easier and especially in terms of crossing the street and staying in the white lines, and knowing that he was in the crosswalk. ...It was just a great relief to know that he could see more high contrast. ...Just a little bit of usable vision after having none for so long was a dream come true for us." - Lisa Lloyd*

Argus II had several downsides. Invasive surgery was required for installation and the company that produced the retinal implant went out of business.

*"What that means for him is that he has the retinal implant still behind his left eye. It is not going to be removed. ...Unfortunately, now he doesn't have the what he needs device-wise for it to function. He just has it in the back of his eye, but the computer system that works with it and the glasses, they're not all functioning together as a unit anymore. He has lost that ability to navigate a little bit easier." – Lisa Lloyd*

**Most individuals living with XLRP have tried many different tools, technologies, approaches, and strategies – almost always in combination - to help manage their XLRP.**

*"Technology is great, and helps with independence, but certainly doesn't compare to having vision." – Rod, comment submitted online*

Poll respondents used online polling to select all the approaches, besides medications or treatments that they or their loved one have used, currently or previously, to help manage symptoms associated with XLRP. Poll respondents selected an average of 4.6 responses each. Poll results are presented in **Appendix 7, Q2** and illustrated with patient comments below. Most meeting attendees described all their different strategies, which emphasized the point that was made earlier, that individuals living with XLRP are required to make constant accommodations, adjustments and adaptations to deal with their ongoing progressive vision loss and changing symptoms.

### Portable lighting devices

Portable lighting devices was the top choice of approaches to manage vision loss, selected by 68% of individuals represented by the poll results. Some described using bright lights in their homes.

*"I use simple things like lights and a cane, a magnifying glass." - Matthew Bacho*

*"The lights in our home are LED. ...We modified the lighting when we moved in and put in a sola-tube in the kitchen where I cook." – Lisa, comment submitted online*

### Magnifying device for computer

Magnifying device for computer was selected by 58% of individuals living with XLRP represented by the poll results. Many also described using high contrast themes in order to see better.

*"My interest in assistive technology really began about 20 years ago when I began experiencing incompatibilities with my work environment, due to my progressive sight loss. I began learning about various technologies, such as screen readers and screen magnification that were available to help me continue doing my job." - Darryl Adams*

For Jon, who has end-stage RP, *"You grab onto whatever you can to help get access to the information you need and make things accessible. And for me, a lot of magnification, screen readers and things of that sort, so that's where I've principally benefited. Every day at work to be able to blow things up." - Jon Erickson*

*"I must use assistive technologies on my computer, especially larger fonts and high contrast themes in order to do my work." - William Hartman*

*"I noticed that some of the panel members use "inverted" or "white-on-black" high contrast themes on their computers. This relatively simple setting has extended my ability to use computer displays instead of just text-to-speech. If I use typical, black-on-white contrast, it's almost as intense as looking into the sun." – Joseh, comment submitted online*

## Device/software to facilitate audio reading

Device or software to facilitate audio reading from a computer or other device was selected by 47% of individuals living with XLRP represented by the poll results, and meeting attendees described many of the tools they used. For many, print material remains a challenge, especially mail.

Paul used screen reading software to read his presentation during the EL-PFDD meeting. *“A computer voice reads each line and I hear it in my earbud, and then I repeat back what I hear out loud.”* He continued, *“I depend on my wife to read most print material and when she's not around, I use text to speech apps like Seeing AI. It's a great app. You just point your smartphone and it reads the text. It reads printed letters and addresses on envelopes. It's invaluable, but there's still situations where I struggle.”* - Paul Martz

*“My computer experience goes back 40 years and my career has always been computer dependent, RP took its toll on my ability to use my management software tools. I adapted by learning screen reading programs, such as JAWS, Job Access With Speech, ZoomText and Apple's VoiceOver. My first and only smartphone remains the Apple iPhone, it's VoiceOver software and mobility apps help me navigate shop and find things I misplaced.”* - Robert Pawlick

*“Dad has a lot of devices which help him in his work and to communicate with people. He uses JAWS on his computer; it reads what's on his computer screen to him. Dad wears an iPhone on his belt that also talks to him all the time. It tells him who's calling, if he has a new text message or email and when his appointments and meetings are. ...He also uses a GPS app to tell him where he is.”* – Lute, Richard Faubion's seeing eye dog

*“I've been blessed. I have had a career through using different technologies, tape recorders when I was in college and such, but computers with the JAWS and the clear reader with the scans of the print, and the talking cellphone which has been a big help.”* - Dave (caller)

### Use of UV sunglasses

Use of UV sunglasses was selected by 47% of individuals living with XLRP represented by the poll results.

*"I wear prescription sunglasses, a visor and protective shoes, so I don't stub my toes. I keep my regular glasses handy in case the light levels dip down and I need them quickly, especially while driving. I know my day driving days are numbered."* - Lisa, comment submitted online

*"If you're curious about my glasses, no, they don't help me see, I'm blind, but they do protect my eyes."* – Paul Martz

### School/work modifications

School or workplace modifications were selected by 42% of individuals living with XLRP represented by the poll results. This also includes home modifications.

Daryl personally facilitated many assistive technology solutions.

*"Throughout my career, I've worked on numerous assistive technology solutions, including an electronic reader, a spatial awareness wearable and enhanced indoor wayfinding to assist people who are blind or visually impaired."* - Darryl Adams, Director of Accessibility at Intel

*"We bought a one level home with no steps intentionally."* – Lisa, comment submitted online

### Low vision specialist

Low vision specialist was also selected by 42% of individuals living with XLRP represented by the poll results. One meeting attendee mentioned how expensive this option could be.

*"We see a retina specialist yearly to track progression XLRP."* – Christine, comment submitted online

*"I just wanted to mention it's \$225 per hour for the assistive technology support in our area currently."* - Lisa Lloyd

### Cataract surgery

Cataract surgery was selected by 37% of individuals living with XLRP represented by the poll results.

*"I am getting the cataract surgery in two weeks, which I'm very anxious about, but I'm optimistic that maybe that'll even make it even more clear."*  
– Susan (caller)

Matthew's cataract surgery downsides included surgical risk, anxiety and the fact that he was awake during the procedure. *"The gene therapy I underwent was just fine because they put me completely under. But I know with cataract surgery, they don't do that, and that was quite uncomfortable. You're always afraid that you'll move, right? And they're asking you questions. So, that was nerve-wracking when I did that back in the '90s."* - Matthew Bacho

### Lenses to magnify central vision

Lenses to magnify central vision were also selected by 37% of individuals living with XLRP represented by the poll results.

*"I am grateful to have the little bit of central vision I have today. With corrective lenses I have 20/50 acuity in both eyes and I'm able to make a living as a visual artist. When people inevitably ask, 'How do you paint and draw without sight?' I am quick to remind them that blindness is a spectrum, and my vision is sort of like 'looking through a straw'."* – Paul 1, comment submitted online

*"I adjusted first by wearing prism magnifying glasses to read with directed light."* – Paul 2, comment submitted online

### Behavioral or psychotherapy

Behavioral or psychotherapy was selected by 21% of individuals represented by the poll results, but there were few comments. Robert described the many therapeutic approaches and their benefits.

*"As a result of seeing a psychiatrist and taking the prescribed medicines, I'm exercising, eating healthier and maintaining a better attitude these days. As therapy, I've been writing and publishing novels and short stories online,*

*woodworking, specifically churning bowls and plates has been a productive therapy for me as well. Helping to rebuild my self-esteem, I remain my family's handyman. I can repair or replace everything in our home.” - Robert Pawlick*

### **Closed-circuit television**

Closed-circuit television was selected by 21% of individuals living with XLRP represented by the poll results. This was mentioned often by meeting attendees and in the online comments.

*“When my central vision was intact, I could read large print. I could use magnification on my computer to blow up the size of the text. I had a CCTV, which is a camera and screen system that turns normal print into large, high contrast text. But as my central vision got worse, those options stopped working for me.”- Paul Martz*

### **We are currently not doing anything**

We are currently not doing anything was selected by 16% of individuals living with XLRP represented by the poll results. This is hardly surprising, as many individuals living with XLRP who attended the meeting were told that there were few other options available for them.

*“My doctor just pretty much telling me, "Hey, tough luck." There was nothing he could do to help me.”- Joaquin Cornejo*

*Every time I go to the ophthalmologist ... they just tell me that there's nothing they can do for me.” - Tim Hill*

### **Retinal prosthesis (artificial retina)**

Retinal prosthesis, which is artificial retina was selected by 11% of individuals living with XLRP represented by the poll results. Elsewhere in the report, Lisa Lloyd described her father’s experience with a retinal prosthesis.

### **Other**

Other approaches were selected by 11% of individuals living with XLRP represented by the poll results. During the meeting and in the submitted



comments, other approaches including the use of canes, exercise and sleep, acupuncture, yoga, adaptive strategies, training or rehabilitation, sighted guides.

- **Canes**

Lute described how Richard, *“Used a white cane for 15 years to find his way around, going to the eye doctor at least every other year for checkups.”* - Lute, Richard Faubion’s seeing eye dog

*“I’d never leave the house without a white cane.”* - Comment submitted by the Foundation Fighting Blindness on behalf of an individual affected with XLRP

*“Once I knew how to use a mobility cane and assistive technology with confidence, I knew that I could carve my own path in life and still be successful in my career.”* – Adam, comment submitted online

- **Exercise and sleep**

*“I get daily exercise, at least seven hours of sleep. Otherwise, I can’t see my best the next day.”* – Lisa, comment submitted online

*“I still have been very active my whole life, and I go walking every day a mile around the neighborhood.”* - Dave (caller)

- **Acupuncture & Yoga**

*“I feel like acupuncture is helpful. It helped brighten up my vision, and I actually started doing it again recently.”* - Mark Curley

Kathy experienced, *“Improved night vision and acuity through acupuncture. I am torn between continuing this therapy which I started 10 years ago, and giving it up all together. My progression seems like it has picked up in the last year or so that I haven’t done acupuncture. Is this a coincidence? I just don’t know.”* – Kathy, comment submitted online

Matt enrolled in clinical trials for acupuncture and yoga. *“My expectations for those were not also very high, but I was interested in those therapies, and I found them very intriguing and indeed yoga did improve my balance, so I was kind of happy about that. But no, I did not see any noticeable improvement in my vision.”* - Matthew Bacho



- **Adaptive strategies, training and rehabilitation**

*“Continual progression of blindness requires lots of ongoing adjustment to cope and perform.” – Rod, comment submitted online*

*“It was liberating for me when I attended adjustment to blindness training and learned essentially how to be a blind person in the world. Once I knew how to use a mobility cane and assistive technology with confidence, I knew that I could carve my own path in life and still be successful in my career. It was invaluable and life-affirming.” – Adam, comment submitted online*

*“I believe education and experience were the best tools for overcoming fear, so immediately after my diagnosis, I learned everything possible about RP. Tennessee's vocational rehabilitation, VOC Rehab office, provided with financial assistance in white cane training so that I could attend college and earn my bachelor's degree in management. ...Sure, I haven't directly benefited from scientific advances, gene therapies, clinical trials or studies. I do believe I've successfully mitigated many RPs effects, reducing it to nothing more than a constant aggravation. Through education and training, RP has not disabled me.” - Robert Pawlick*

*“One treatment I wanted to share with you is about 22 years with the Bureau of Blind Services Rehabilitation. Instructor was my job, teaching people how to live independently within their own home with visual impairments, because I was one myself.” - Dave (caller)*

- **Sighted Guides**

*“As my sight loss progressed, I was faced with a decision, I would need to stop participating in the activities that I loved or I would need to find alternative ways to participate. And for me, the answer was finding sighted guides to help me keep going.” - Darryl Adams*

- **Augmented reality smart glasses**

*“For a massive boost to night vision, I am using augmented reality smart glasses. Other patients should test out various VR, AR glasses.” – Mike 2, comment submitted online.*

## Service dog

Service dog was selected by was selected by 5% of individuals living with XLRP represented by the poll results.

*Lute is Richard's black Labrador retriever guide dog. "We go for long walks almost every day, and it's my job to keep him safe; watching for cracks, bumps, hanging limbs, avoiding distractions and oncoming cars if we're on a road and not a sidewalk. ... When it's time to go outside, he reaches out to find me so I can walk beside him with my harness and leash on. ... Even if Dad does get some of his sight back, I know we'll always be together because we're bonded for life." - Lute, Richard Faubion's seeing eye dog*

*"I did go get a seeing eye dog which I thought is invaluable. I can walk for miles on my own if I want." – Susan (caller)*

**Drawbacks of current approaches:** Almost half of individuals living with XLRP reported that their current approaches to managing XLRP are not very effective and almost half reported not having any therapeutic options to manage XLRP. Poll respondents used online polling to select the top three biggest drawbacks of current approaches to managing XLRP. The top two responses were split equally between "Not very effective at treating target symptoms" and "Not applicable as I am not using any". Poll results are presented in **Appendix 7, Q4** and illustrated with patient comments below. The treatments, devices and accommodations that exist are not very helpful and are not stopping the progression.

### Not very effective at treating target symptoms

Almost half, or 47% of individuals represented in the polls indicated that their treatment approaches were not very effective at treating target symptoms or no longer worked as their vision continued to deteriorate.

"Not very effective at treating target symptoms" was reflected in the response to the poll question, "How well does your current regimen aid with symptoms or control your or your loved one's disease overall?" Only 6% of poll respondents selected "to a great extent". While 59% of poll respondents indicated that their current regimen works "somewhat", 18% responded that their current regimen aids "somewhat", 6% selected "not at all". A further 12% responded "Not

applicable because they are not using anything". Poll results are presented in **Appendix 7, Q3**.

*"Well, I've used a lot of different things, like screen readers and I have a white cane, and sunglasses and different things. ... I've tried all these different devices to do things like help me watch television, and none of them work." - Tim Hill*

*"How well do these treatments address effects... I answered "somewhat" because use of a cane, inverting colors on computer, ZoomText software, iPhone/Siri, CCTV, and Uber/Lyft services are not always a guarantee every day. Software updates often change the layout. Siri can misspeak or miss text or notifications. Uber/Lyft are not always reliable or helpful. All of these have limitations. They do not replace fully one having eyesight." – Paige, comment submitted online*

*"Assistive technology helps me tremendously. I still do have many limitations, and I still ask for help quite frequently. But, with assistive technology, I function very well and I am able to complete nearly all of my daily tasks." – Jeffrey, comment submitted online*

### **Not applicable as I am not using any**

Almost half, or 47% of individuals represented in the polls indicated that this question was not applicable as they were not using any treatments, consistent with responses to earlier poll questions.

### **Limited availability or accessibility**

Almost a quarter, or 26% of poll respondents indicated that there was limited availability or accessibility to treatments for XLRP. Some of the reasons include location far from a clinical trial centre, being excluded from trials because of sex, comorbidities, bureaucracy, or having insufficient vision.

*"I don't have as much access as some of the others to do a clinical trial. ... I'd have somebody drive me for six hours or more." - Tim Hill*

*"I'm a female with the RPGR version of RP and although I was told I was a carrier, I started losing night vision in my teens and have been legally blind since my late 40s. My niece whose dad has RP is following my pattern, yet*

*we are not considered eligible for the gene therapy trials due to our sex, which is maddening.*" – Laura, comment submitted online

*"I have not been able to participate in any trials or research for RP because I also have dry macular degeneration. Will there ever be a chance for me to take advantage of any new developments?"*- Denise, comment submitted online

*"I've been applying for clinical studies because I see real hope in them. However, the spots are very limited and the inclusion criteria are very specific. Imagine that you knew that that there exists a treatment that can stop or even slow the progression of blindness. And it's sitting in a freezer in a lab somewhere, and bureaucracy and red tape is keeping you away from it until potentially it's too late."* - Jonathan Tamaiev

*"[For gene therapy clinical trials] you do have to have enough vision to save, to make that meaningful. For a lot of us at this end stage RP, we're hanging our hats on stem cells to really be able to regenerate those cells, those rods and cones."* - Jon Erickson

*"I recently tried to sign up for a clinical study, but I was told that my eyesight was too bad. That they wouldn't take me."* - Tim Hill

### **Only treats some, but not all symptoms**

A further 21% of respondents selected only treats some, but not all the symptoms as a downside.

### **Route of administration**

Although only 11% of poll respondents selected route of administration as a downside, there were several quotes about surgeries or injections.

Lisa's father had the Argus II implanted. *"Just go into the operation, it was a little scary taking out the eyeball and getting it in back there and stapling in there and all of that. And he did get a slight infection afterwards, but it cleared up after a few days, thank goodness."* – Lisa Lloyd

Route of administration was problematic for Matthew during a gene therapy trial. *"In my case, [the administration] was a sub-retinal [injection] and they asked me ahead of time if they could treat the fovea, which was*

*really probably only part of the retina that I was using. I agreed. ... Unfortunately, it led to a problem with acuity. ...I see that they're working on intravitreal administration. I'm wondering if that perhaps, that would avoid such problems.” - Matthew Bacho*

### **High cost or co-pay, not covered by insurance**

This option was selected by 5% of those responding to the polls, but there were few quotes to support this.

### **Other drawbacks**

This option was selected by 5% of poll respondents. Many of the other drawbacks, such as technology no longer working as vision deteriorates or because companies have gone out of business, were discussed previously. Other drawbacks described included technology doesn't always work in real world situations, is continually being updated and the challenge of switching between visual and audio formats especially as vision deteriorates.

- **Technology doesn't always work in real-world situations, is continually being updated.**

Text to speech apps aren't proficient at reading directions on food packages. *“It reads everything it sees. It runs lines from different blocks of text together. What am I supposed to do with that? Why aren't the apps smart enough to sort out the blocks of text? It's even more of a challenge when a text is on a plastic bag or something round, like a bottle or can.” - Paul Martz*

*“Then there's the dreaded software update. It's guaranteed to change the app layout, like somebody rearranged the furniture.” - Paul Martz*

*“The technology is great, but it's also constantly changing so it's a challenge just to keep up with all of that.” – Rod (caller)*

- **Having to negotiate both visual and audio formats, especially as vision deteriorates**

*“We're somewhat in this visual world and we, at some point, have to transition to more audio and that's just a fuzzy line. And so, I think a lot of us find ourselves straddling, having to rely on vision to accomplish work,*

*daily life, etc., and needing to pivot somewhat with different skills and technologies to facilitate that.” - Jon Erickson*

*“I learned all the zoom text and all the different technologies to make things bigger, ... that worked well for about 18 years. But then, in my early forties, it got to a point where making things bigger didn't work. So going from using technology to make things bigger to doing things audibly is ... as big as adjustment as not driving.” – Rod (caller)*

*“I did try to learn to work audibly, instead of visually, it was just such a big change, and added to my stress. Ended up staying on disability instead of returning to work.” - Comment submitted by the Foundation Fighting Blindness on behalf of an individual affected with XLRP*

*“I consider myself fortunate to live in the digitized era and have worked hard transitioning from a visual to auditory learner.” – Paul 2, comment submitted online*

### **Requires too much effort/time commitment and side effects**

Although these drawbacks were not selected by any of the poll respondents, meeting attendees made comments about the time required for clinical trial testing and side effects.

*Matt described all the tests required for clinical trials. “Those some tests can be sometimes a little frustrating, but I've gotten used to them and the technology's gotten better. It seems like back in the 1990s, I wasn't sure that the [researchers] were getting a lot of data, but I think the technology has really improved. And I think that makes it worthwhile and helps me as a participant know that they're getting some good data” . - Matthew Bacho*

*"My fiancé has expressed concern for trying a new treatment that would possibly make his current vision worse. Side effects would be a major factor in his decision to try a new treatment." – Paige, comment submitted online*

### **Top three aspects for a possible new drug today: Improvement to night vision, visual acuity, and peripheral vision.**

Poll respondents used online polling to select the top three aspects of their condition that they would rank as most important for a possible new drug today.



Poll results are presented in **Appendix 7, Q5** and illustrated with patient comments. This poll did not necessarily capture all the possible options as *many* meeting participants mentioned that they wanted a drug that stopped or slowed progression, and this is described under Other. Some asked for improvement to all aspects of vision to preserve a sense independence.

*“The option for ‘all of the above’ was not available, but I would say all of the above most certainly. But to be a little bit more specific, what I've had the toughest time dealing with, especially over the past two or three years is, I'm losing my sense of independence. ...So if I could truly pick one of those options, it would be that, and of course along with gaining independence is overall improvement and a cure. So that's kind of what that leads to.”* -

Mark Curley

### **Help with night vision**

A total of 68% poll respondents selected help with night vision as one of their top three aspects of XLRP that they would rank as most important for a possible new drug today. The impacts and challenges of low night vision, especially driving, socializing and safety, were discussed earlier in this report.

### **Improving visual acuity**

Improving visual acuity was also selected by 68% of poll respondents as one of their top three aspects of XLRP that they would rank as most important for a possible new drug today.

Tim explained that to maintain his independence he needs, *“Just more acuity. Just be able to clear up my vision to where I can at least see just a little bit better. I've lived with the loss of peripheral vision for so long that I'm used to that, but I think more acuity is what I really need... to do things where I don't have to have help with everything.”* - Tim Hill

*“If I could see enough for good orientation and mobility, then when Lute and I are walking around town, maybe we could see the large store signs to know where we are. Now I use a GPS app that tells me where I am, but it would be nice to be able to confirm that with a little vision.”* Richard also wants to be able to *“watch more television and actually see what's going on*

*rather than just seeing shapes and hearing lots of voices in music.” - Richard Faubion*

### **Help with peripheral vision**

Help with peripheral vision was selected by 47% of poll respondents as one of their top three aspects of XLRP that they would rank as most important for a possible new drug today.

*"My sons, 16 and 20, both are affected with XLRP. Their limited peripheral and night vision affects all the other choices in this poll. And we are hopeful that both will qualify and participate in clinical trials in the next few years."*

– Nancy, comment submitted online

### **Attending school or having a job**

This was selected as a top three choice of 32% of poll respondents as one of their top three aspects of XLRP that they would rank as most important for a possible new drug today. This is not surprising: so many meeting participants described how their XLRP impacted their employment and careers.

Matt selected this option. *“Based on my experience in trying to get a job and starting a career when I was younger, just getting your foot in the door and getting into school.”- Matthew Bacho*

### **Walking/ambulating without assistance**

Walking and ambulating without assistance were selected as a top three choice by 32% of poll respondents.

*“Mobility in general, being able to get around, being independent, not having to rely on others.” – Matthew Bacho*

### **Participating in sports and other recreational activities**

This option was selected as a top three choice of 16% of poll respondents.

### **Participating in social engagements and events**

Participating in social engagements and events was selected as a top three choice of 16% of poll respondents as one of their top three aspects of XLRP that they would rank as most important for a possible new drug today.

*“If I could gain any sight back, I would really like to see faces and people again.” – Jeffrey, comment submitted online*



*"I've learned over the years about the importance of nonverbal communication. So social interactions, being able to make eye contact, seeing a smile or a wave and not having to explain why you didn't wave back, I mean, I think those are things, from my personal experience, which would be a great thing to get back or to improve." – Matthew Bacho*

### **Using a computer**

Using a computer was selected as a top three choice by 5% of poll respondents.

### **Travel or Vacationing**

Travel or vacationing was not selected as a top three choice by any of the poll respondents.

*I want to travel. I want to explore. I want to continue to see." - Jonathan Tamaiev*

### **Other - preserving remaining vision, helping central vision, treating second eye, cure/restoration of vision.**

Although none of the poll respondents picked 'other' as one of their top three choices several other options mentioned during the meeting including preserving remaining vision, improvement of central vision, having second eye treated, vision restoration

- **Preserving remaining vision - stopping the progression of vision loss**

*"If there were only a way to stop the progression, I would at least have some semblance of control over my future." - William Hartman*

*"A treatment with the potential to stop the disease progression will effectively ease the way for those who are in the earlier stages of decline and it will allow them to plan for a life, where they can rely on their eyesight, even if it's not perfect. ... Finding ways to stop my sight loss progression would mean the world to me." - Darryl Adams*

- **Improvement in central vision**

*"I just wish I had better central vision at this point... So that was my choice as well is just to get better central vision and be able to better see you guys*

*on the screen and read.” When asked how much improvement he wanted, Jon replied “20/20. How about that?” - Jon Erickson*

*“If I had lost only my peripheral vision, I'd spend the rest of my life as someone with a vision impairment, and that would stink, but it would still be better than losing my central vision and leaving me blind, which is where I am now. So if there's one treatment the RP community needs, it's restoration of central vision.” - Paul Martz*

- **Treatment of second eye for those who participated in clinical trials**

*“These phase one, two patients took on a lot of risk and spent huge amounts of time and endured big disruptions to their lives, to advance these treatments to the next stage and should be offered the opportunity to have their other eye treated as soon as possible.” Jeff hoped that patients who participated in the phase I and II trial cohorts, could participate in a crossover cohort or could have their untreated eye treated as part of a compassionate use program.*

- **Vision restoration**

*"This is an easy answer for me. In order of my preferred priority, one, vision restored, two, vision enhanced, three vision stabilized, and in all cases without a big risk to health and adverse side effects." - Comment submitted by the Foundation Fighting Blindness on behalf of an individual affected with XLRP*

*“I told myself at a young age, I would never allow myself to hope for a cure. ... I'm in a place in my life now where I have the ability to critically evaluate the research out there. At this point, I went back on my word to myself. I am hopeful. I do not need to lose my vision. This is becoming more and more clear each day. However, each day that the treatment is delayed. I am one step closer to irreversible blindness.” - Jonathan Tamaiev*

- **Greater inclusion in clinical trials, especially for females**

*“I'd like to express our family's frustrations with the eligibility criteria for the various X-linked RPGR clinical studies. ...Clinical trials already have enough safety and efficacy data to broaden their inclusion criteria to include persons older than 65 y/o, women, any one eye that is free of calcium*

*deposits or nevus, and MRS scores below 2.0 dB.” – Michael, comment submitted online*

*“VERY FRUSTRATING that us XLRP females (later stages) are not allowed to participate in the gene therapy trials. This kind of discrimination would not be allowed in other fields!” – Lisa, comment submitted online*

*“I am a 65-year-old female with significant symptoms and I am also very frustrated with the lack of access of clinical trials for me!!!” – Beth, comment submitted online*

- **More research specifically for women and for XLRP specifically**

*“I’d be interested in hearing about research relating to women affected with XLRP” – Kathy, comment submitted online*

*“It seems like everything is geared towards more macular degeneration, rather than people with just a little bit of central vision, which is what I have. I’ve never had more than nine degrees peripheral, and I’ve never had any night vision, but it’s just my eyesight - it just keeps getting progressively worse.” - Tim Hill*

*“I’ve participated in natural history and visual field measurement studies, and I was recently a control participant in a gene therapy clinical trial. I strongly believe that consistent scientific advancement in this field is essential to making progress in the pursuit of site restoration. I’m excited to have the chance to participate in the science that promises to restore eyesight and improve the lives of so many.” - Darryl Adams*

## Incorporating Patient Input into a Benefit-Risk Assessment Framework

*When deciding whether or not to use a treatment, I would consider cost, the risk of adverse events, frequency of treatments, duration of treatment, feasibility of treatment.” – Jeffrey, comment submitted online*

The FDA uses a Benefit-Risk Assessment Framework which includes decision factors such as the analysis of condition, current treatment options, benefit, risk, and risk management. The Framework provides an important context for drug regulatory decision-making and includes valuable information for weighing the specific benefits and risks of a particular medical product under review.

**Table 1** speaks to the challenges of living with XLRP. It serves as the proposed introductory framework for the Analysis of Condition and Current Treatment Option to be adapted and incorporated in the FDA’s Benefit-Risk Assessment. This may enable a more comprehensive understanding of this unique condition for key reviewers in the FDA Centers and Divisions who would be evaluating new treatments for XLRP. The data resulting from this meeting may help inform the development of XLRP -specific clinically meaningful endpoints for current and future clinical trials, as well as encourage additional researchers and industry to investigate options for treatments.

The information presented captures the perspectives of patients living with XLRP presented at the June 7, 2022, meeting. It includes information from the polling results, as well as comments submitted before, during, and after the meeting through the online portal. This table was formatted to be understandable by screen reader software.

Note that the information in this sample framework is likely to evolve over time.

**TABLE 1 XLRP Benefit-Risk Table – *See the Voice of the Patient Report for a more detailed narrative.***

<b>ANALYSIS OF CONDITION/ IMPACTS ON ACTIVITIES OF DAILY LIVING</b>	
<b>EVIDENCE AND UNCERTAINTIES:</b>	<p><b>X-linked retinitis pigmentosa (XLRP) is characterized by progressive vision loss.</b></p> <p>Symptoms range from difficulties adapting to changes in lighting, diminishing night vision, a loss of peripheral vision, and eventually progress to a loss of central vision. Individuals living with XLRP live in fear of deteriorating vision loss and eventual blindness.</p>
<b>CONCLUSIONS AND REASONS:</b>	<p><b>XLRP is not just about vision loss but involves a gradual deterioration of independence.</b></p> <p>All activities of daily living are impacted as vision deteriorates. Individuals living with XLRP lose their ability to drive. Participating in social engagements and events becomes more challenging, they lose the ability to read, and employment and careers are severely impacted. Roles within families change and self-esteem plummets. They experience injuries from falls, bumping into objects, or worse. Many experience stigma and discrimination.</p>

## CURRENT TREATMENT OPTIONS/ PROSPECTS FOR FUTURE TREATMENTS

### EVIDENCE AND UNCERTAINTIES:

**There are no approved treatments for XLRP.**

Many individuals have tried the few available therapies as well as investigational therapies in clinical trials, some of which involved high risk, invasive interventions.

**As vision deteriorates, people with XLRP are forced to continually adapt and find new strategies and technologies.**

These include combinations of portable lighting devices, magnifying devices, UV sunglasses, audio reading and other modifications. Many described having to eventually use both visual and audio modifications as their vision deteriorated.

**Approaches to managing XLRP work “somewhat”, however most do not treat the target symptoms, slow progressive vision loss, nor replace or restore sight.**

There are currently no approved medications for XLRP and access to experimental treatments is limited, often due to restrictions in clinical trial eligibility. As a result, some individuals affected with XLRP are not using any treatment.

### CONCLUSIONS AND REASONS:

**Short of a cure, those living with XLRP need a therapy to improve night vision, visual acuity, and peripheral vision. Individuals with XLRP need a therapy to prevent their vision from deteriorating further.**

They also asked for more XLRP research and a broader inclusion criterion for clinical trial especially for women.

## Conclusion

At the start of the XLRP EL-PFDD meeting, Dr. Todd Durham, PhD, said, “*Those listening and learning today, I ask that you thoughtfully consider these stories and imagine what life is like with XLRP and what a treatment might do to improve their lives.*”

This meeting provided an opportunity for the FDA, government agencies, regulatory authorities, pharmaceutical and biotechnology companies, scientists, and health care professionals to hear directly from the patients with XLRP.

Meeting attendees heard about the myriad of symptoms that individuals living with XLRP experienced, the frightening progression of vision loss, and the gradual deterioration of independence and self-esteem. They heard about changes to family roles and relationships, employment and how individuals living with XLRP have to give up the recreation and sports that they love, and their worries for their future care and the continuing manifestations of disease progression. Many expressed their worries for the futures of their children and loved ones living with XLRP.

Individuals living with XLRP described the few therapeutic options available and the many tools and techniques that they used to adapt to their diminishing vision. Even when used in combination, these solutions only worked “somewhat” to alleviate symptoms and many are not using any approaches. In the absence of vision restoration, individuals living with XLRP would like vision loss to be slowed or stopped and improved night vision, visual acuity, and central vision. Most wish for a restoration of independence

The Foundation Fighting Blindness is grateful for this opportunity to share our voices through this *Voice of the Patient Report* and we sincerely thank all individuals and family members with XLRP who shared their insights and experiences at the EL-PFDD meeting, submitted online comments, and participated in the survey. We hope and expect this meeting will encourage future research and successful new product development for people living with XLRP who urgently need safe and efficacious treatment options.

*“My eyes have been opened as to how inaccessible and challenging so much in this world is to those with vision impairments.” – Paige, comment submitted online*

*“While it's important to embrace new tools and techniques to manage living with sight loss progression, having actual treatments available would fundamentally change the lives of so many.” - Darryl Adams*

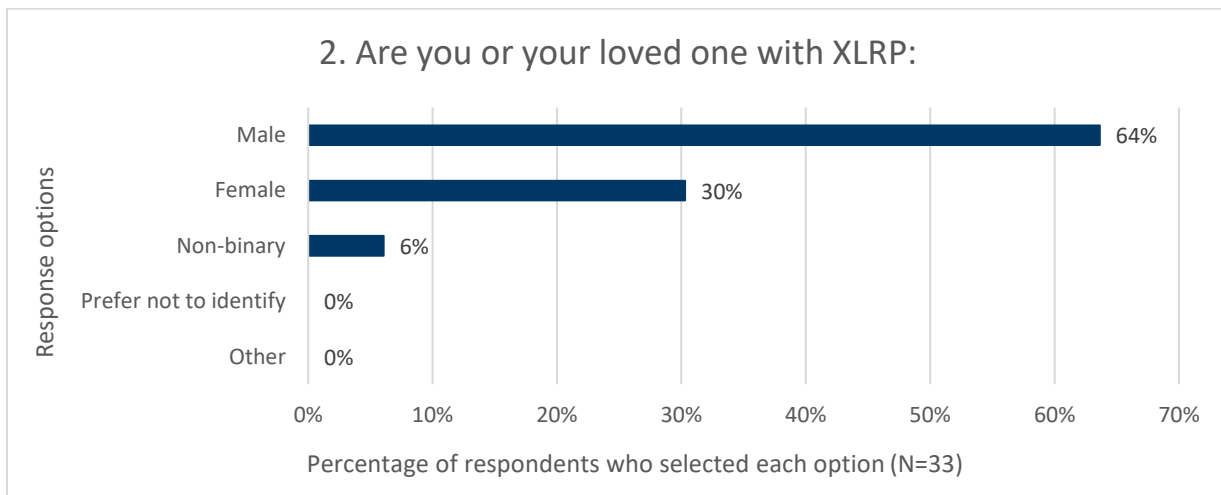
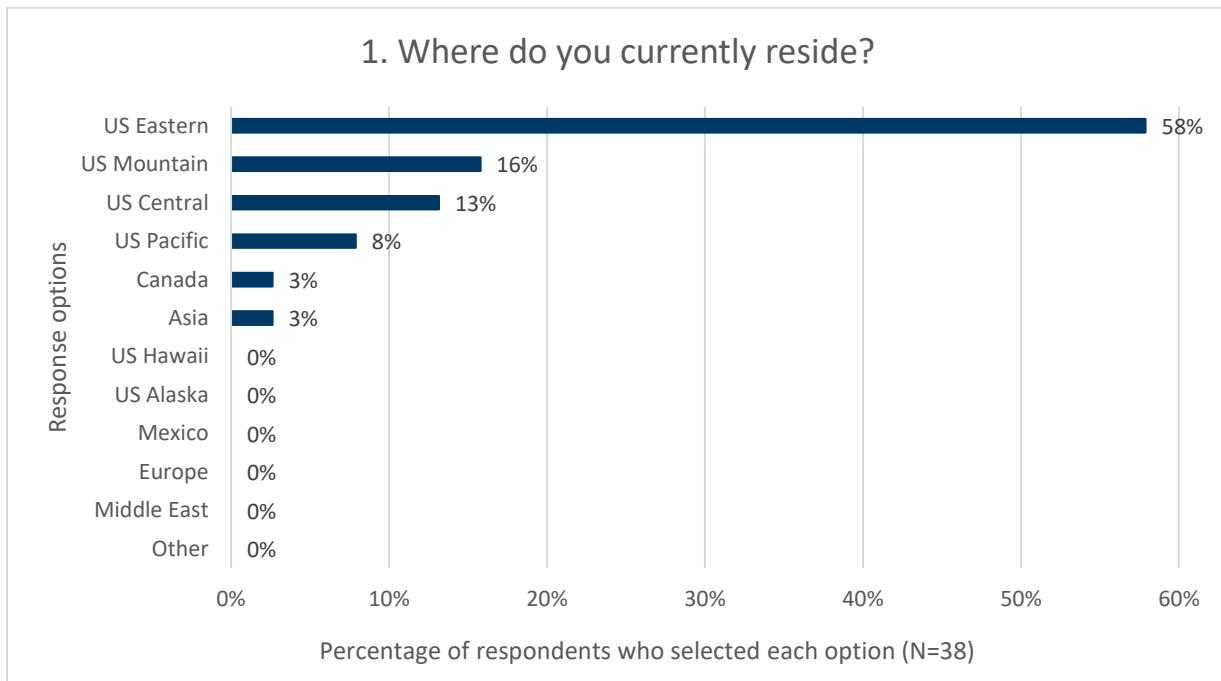
*“XLRP has taken and continues to take so much from me. I will continue to fight, fight back and find ways to cope in the hope that one day very soon, there will be a treatment to halt the progression that would finally allow me to catch my breath from the relentless race to deal with the ever-increasing disabilities inflicted by XLRP. That would allow me to experience a brief moment of peace from the years of XLRP distress.” - William Hartman*

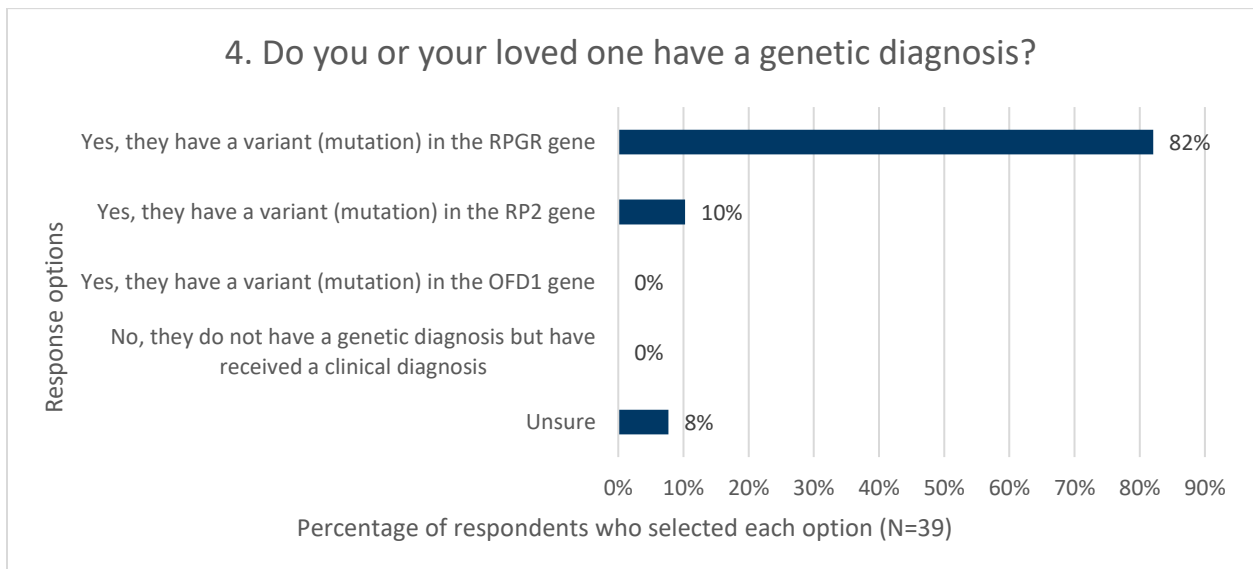
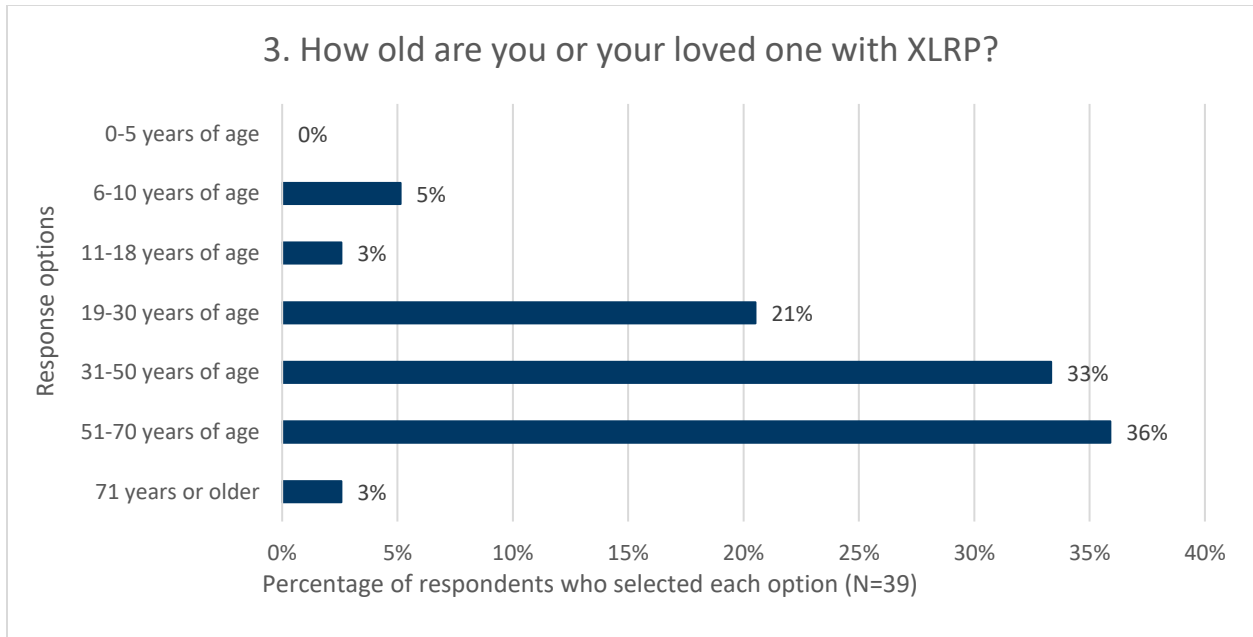


## Appendix 1: Demographics

The graphs below include all attendees who chose to participate in online voting. The number of affected individuals and caregivers who responded to each polling question is shown below the X axis (N=x).

While the response rates for these polling questions is not considered scientific data, it provides a snapshot of those who participated in the XLRP EL-PFDD meeting and is intended to complement the live and pre-recorded comments throughout the meeting.





## **Appendix 2: Survey to Explore the Impacts of X-Linked Retinitis Pigmentosa on Daily Living and Goals for Treatment - Results**

Prior to the EL-PFDD, the Foundation conducted a survey of males with RPGR-associated XLRP who were members of the My Retina Tracker Registry. The full report is included at <https://www.fightingblindness.org/xlrp-pfdd>.

The key results from the survey are as follows:

- There were 41 respondents to the survey.
- The mean age was 51 years and the mean age of first visual symptoms was 14 years.
- The most common first visual symptom, reported by 84% of respondents, was difficulty seeing in dim or dark light.
- Nearly all respondents (90% or more) reported that the following activities of daily living were difficult because of their XLRP: navigating spaces in dim light; navigating spaces that are unfamiliar; finding misplaced items; participating in sports or other physical activities; and shopping in stores or pharmacies.
- The most common current visual symptoms, reported by 90% or more respondents, are difficulty seeing in dim light or the dark and difficulty telling one color from another.
- When asked about the risks from receiving a future potential treatment that would stop the progression of their XLRP, more than 60% reported that they would be “very concerned” about the risk of permanent loss of central vision.

### Appendix 3: Meeting Agenda EL-PFDD for XLRP

**June 7, 2022**      *All times are Eastern Daylight Time*

10:00 – 10:05AM    Opening remarks  
*Todd Durham, PhD*, Senior Vice President, Clinical & Outcomes Research, Foundation Fighting Blindness

10:05 – 10:15 AM    FDA Remarks - The Role of Patients in Drug Development  
*Ekaterini Tsilou, MD*, Medical Officer in the Office of Tissues and Advanced Therapies in the Center for Biologics Evaluation and Research (CBER) at the FDA

10:15 – 10:30AM    Clinical Overview of XLRP  
*Thiran Jayasundera, MD, MS*, Professor of Ophthalmic Genetics, Associate Chair, Strategic Implementation at the Kellogg Eye Center, University of Michigan

10:30 – 10:35AM    Overview of Discussion Format  
*James Valentine, JD, MHS*, Hyman, Phelps & McNamara, meeting moderator

10:35 – 10:40AM    Audience Demographic Polling Questions

#### **Session 1: Living with XLRP - Symptoms and Daily Impact**

10:40 – 11:05AM    Patient & caregiver perspectives on symptoms and daily impacts - 5 pre-recorded panelists

11:05 – 12:30PM    Audience discussion and remote polling on Topic 1

12:30 – 1:00PM      Break

#### **Session 2: Current & Future Approaches to Treatment for XLRP**

1:00 – 1:25PM      Panel 2: Patient & caregiver perspectives on current and future treatments- 5 pre-recorded panelists

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1:25 – 2:45PM Audience Discussion and remote polling on Topic 2

2:45 – 2:55PM Meeting Summary  
*Larry Bauer, RN, MA, HPA, Hyman, Phelps & McNamara*

2:55 – 3:00PM Wrap Up and Thank You  
*Todd Durham, PhD, Foundation Fighting Blindness*

## Appendix 4: Meeting Discussion Questions

### Session 1: Living with XLRP: Symptoms and Daily Impacts

1. Of all the symptoms and health effects of XLRP, which 1-3 symptoms have the most significant impact on you or your loved one's life?
2. How does XLRP affect you or your loved one on best and on worst days?
3. How has your or your loved one's symptoms changed over time? How has the ability to cope with the symptoms changed over time?
4. Are there specific activities that are important to you or your loved one that you cannot do at all or as fully as you would like because of XLRP?
5. What do you fear the most as you or your loved one gets older? What worries you most about you or your loved one's condition?

### Session 2: Current & Future Approaches to Treatment for XLRP

1. What are you currently doing to manage your or your loved one's XLRP symptoms?
2. How well do these treatments address the most significant symptoms and health effects of XLRP?
3. What are the most significant downsides to your or your loved one's current treatments and how do they affect daily life?
4. Short of a complete cure, what specific things would you look for in an ideal treatment for XLRP? What factors would be important in deciding whether to use a new treatment?

## Appendix 5: Panel Participants, Discussion Starters and Callers

### Session 1: Living with XLRP: Symptoms and Daily Impacts

#### Patient/caregiver testimonials

- **Jonathan Tamaiev**, 27 years old and diagnosed with XLRP at the age of 13
- **William Hartman**, 50 years old and diagnosed with XLRP at the age of six. Genetically tested and confirmed to have an *RPGR* gene variation
- **Joaquin Cornejo**, 41 years old, experienced XLRP-related challenges since the age of five
- **Greg Dubecky**, caregiver with a wife, son and brother-in-law are all living with XLRP

#### Zoom discussion starters

- **Michael Stone**, living with XLRP
- **Lisa Lloyd**, diagnosed with XLRP at age 22, caregiver for her 15-year-old daughter living with Leber congenital amaurosis (LCA) and her 81-year-old father living with XLRP. She is part of an XLRP family
- **Andrea Bullock**, living with XLRP
- **April Lifriu**, living with XLRP and caregiver of two children living with XLRP. She is part of an XLRP family and has a sister living with XLRP

#### Callers

- **Robert**, age 55 years old and diagnosed at the age of 14. He is part of an XLRP family and has two sisters and a son living with XLRP
- **Tim**, 66 years old and diagnosed with XLRP at the age of six, with the *RPGR-ORF15* gene variation
- **Rod**, 61 years old and diagnosed with XLRP at the age of 17
- **Paige**, fiancée of Jeff who is living with XLRP
- **Allan**, living with XLRP and part of an XLRP family

### Session 2: Current & Future Approaches to Treatment for XLRP

#### Patient/caregiver testimonials

- **Richard Faubion**, living with XLRP and owner of Lute, a black Labrador retriever guide dog.

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- **Paul Martz**, 59 years old and diagnosed with RP at the age of 13
- **Robert Pawlick**, living with XLRP for 34 years and from an XLRP family, with his maternal uncles affected. His testimonial was read by his wife, Lisa Pawlick
- **Darryl Adams**, diagnosed with XLRP at the age of 14 years old

### **Zoom discussion starters**

- **Jon Erickson**, living with XLRP.
- **Mark Curley**, aged 48 years old and living with XLRP over 35 years
- **Matthew Bacho**, 52 years old and originally diagnosed at the age of 13, and confirmed as XLRP eight years ago
- **Tim Hill**, 66 years old and diagnosed with XLRP at the age of six, with the *RPGR-ORF15* gene variation

### **Callers**

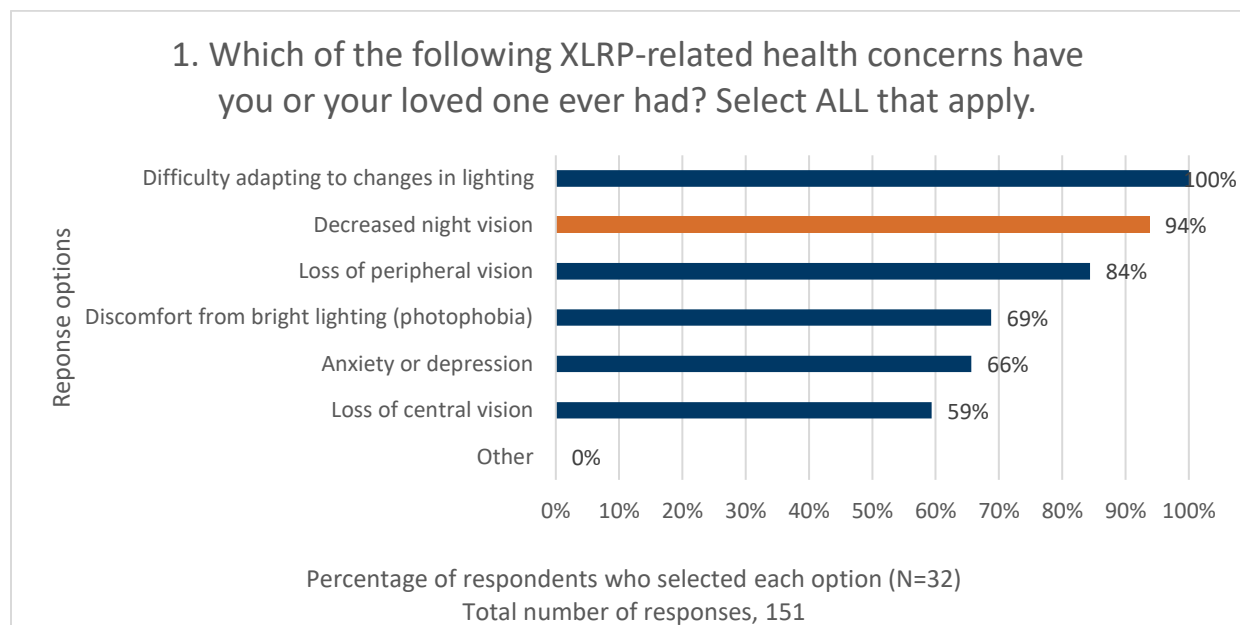
- **Dave**, 70 years old, living with XLRP since grade school
- **Lisa**, diagnosed with XLRP at age 22, caregiver for her 15-year-old daughter living with Leber congenital amaurosis (LCA) and her 81-year-old father living with XLRP. She is part of an XLRP family.
- **Susan**, 60 years old and living with XLRP



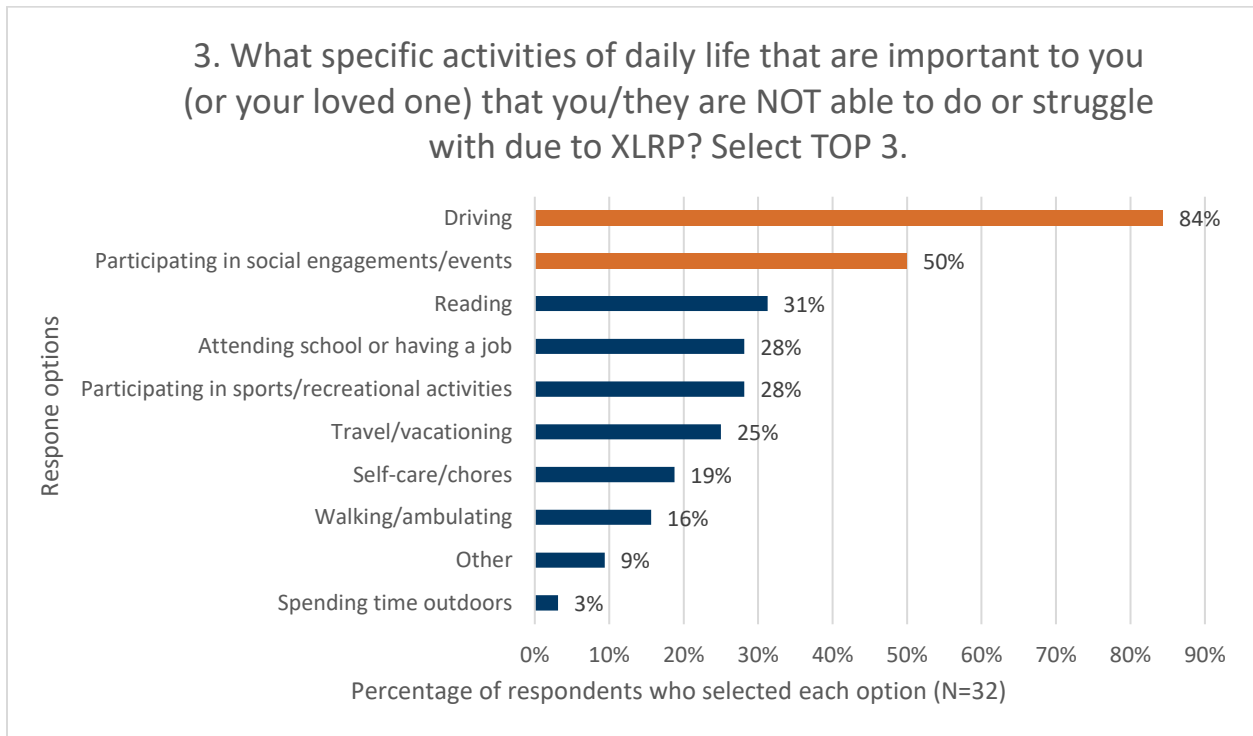
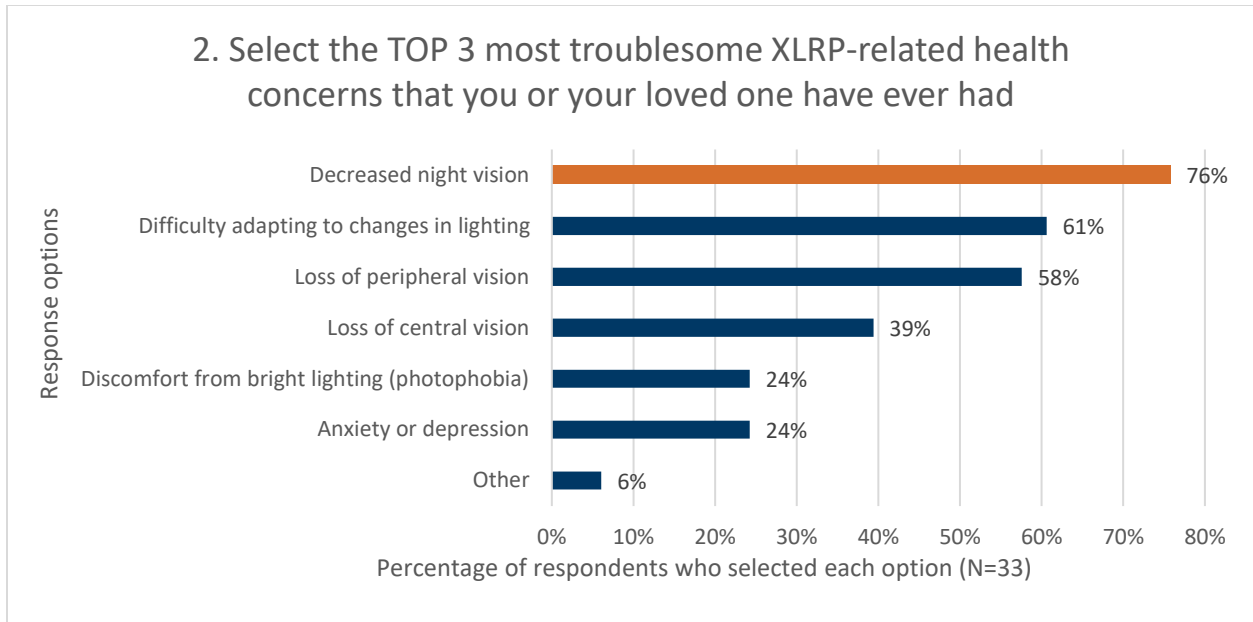
### Appendix 6: Session 1 Polling Results

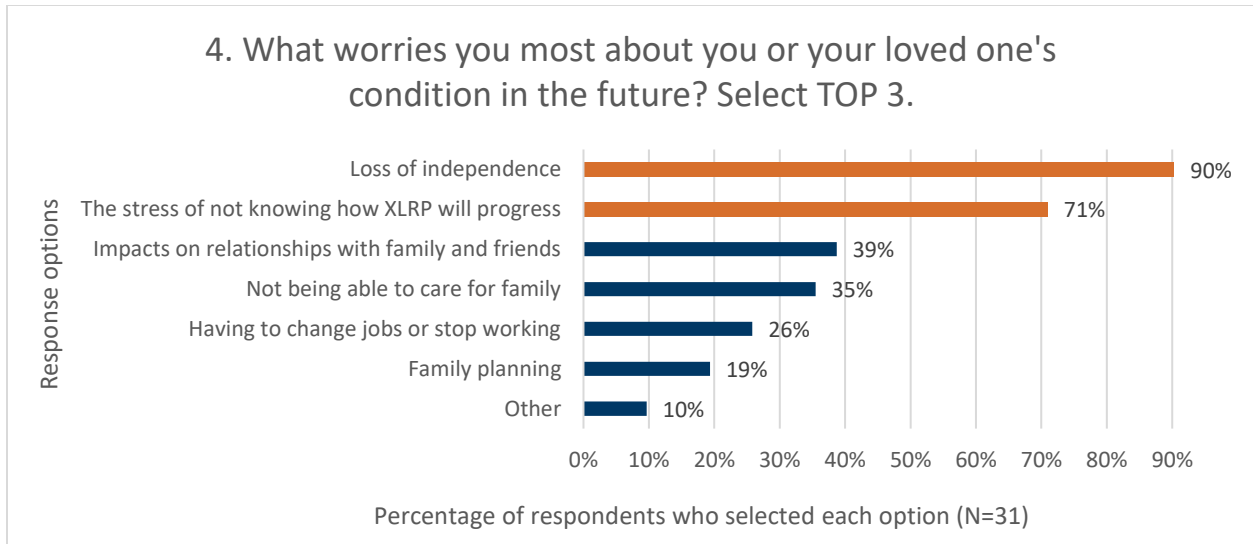
The graphs include those attendees who chose to participate in online voting. The number of patients who responded to each polling question is shown below the X axis. For most questions, poll respondents could select more than one response. The total of poll responses is also shown below the X axis.

While the response rate data for these polling questions is not considered scientific data, it provides a snapshot of who participated in the XLRP EL-PFDD meeting and is intended to complement the live and pre-recorded comments throughout the meeting.



Respondents each selected an average of 4.7 responses to this poll question.

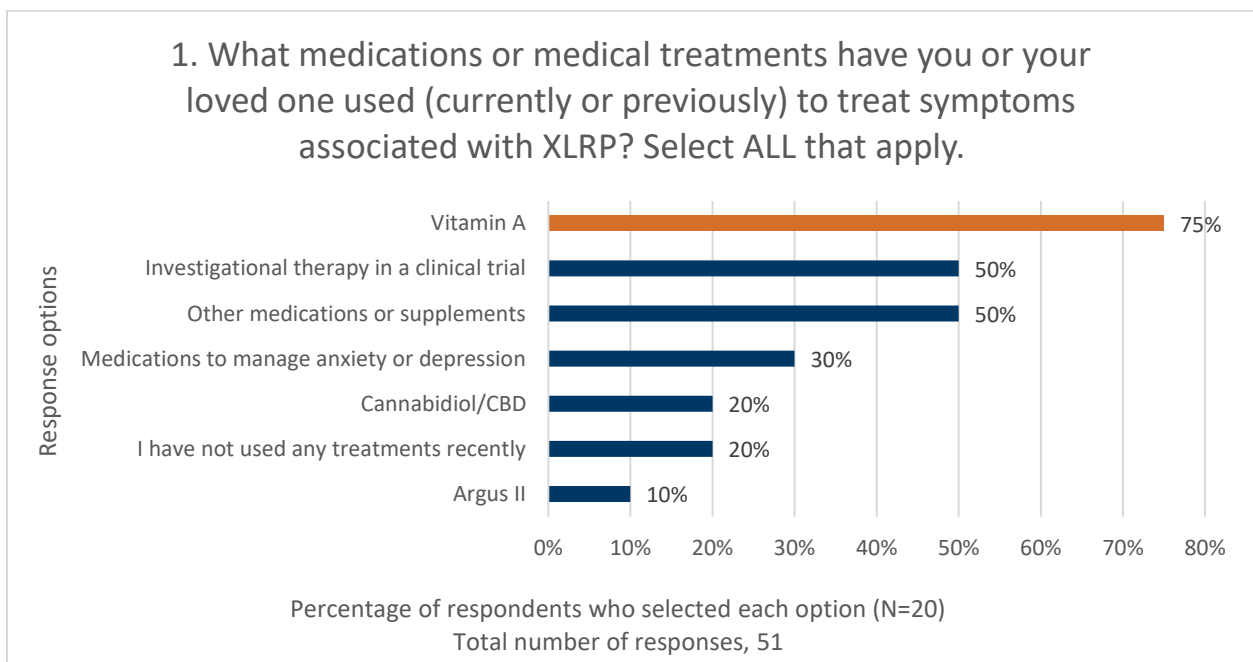




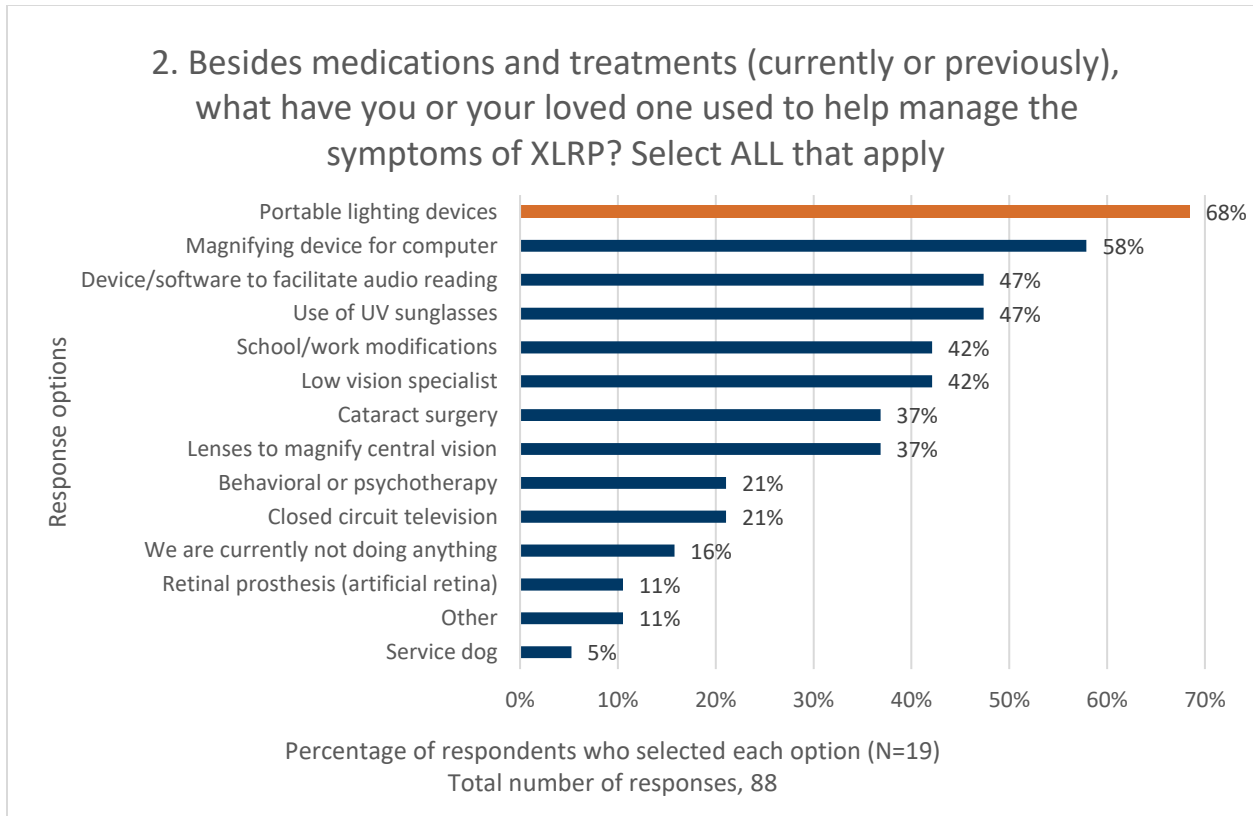
### Appendix 7: Session 2 Polling Results

The graphs include those attendees who chose to participate in online voting. The number of patients who responded to each polling question is shown below the X axis. For most questions, poll respondents could select more than one response. The total of poll responses is also shown below the X axis.

While the response rate data for these polling questions is not considered scientific data, it provides a snapshot of who participated in the XLRP EL-PFDD meeting and is intended to complement the live and pre-recorded comments throughout the meeting.



Respondents each selected an average of 2.6 responses to this poll question.



Respondents each selected an average of 4.6 responses to this poll question.

