



VOICE OF THE PATIENT REPORT

Galactosemia

Externally-Led Patient-Focused
Drug Development Meeting

Meeting Date: September 1, 2022

Report Date: May 1, 2023

MEETING CO-HOSTED BY:

The Galactosemia Foundation
and the National Organization
for Rare Disorders, Inc. (NORD)



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Galactosemia Voice of the Patient Report

The Galactosemia Foundation (GF) and the National Organization for Rare Disorders (NORD®), Inc. collaboratively prepared this Voice of the Patient report as a summary of the input shared by people and families living with Galactosemia during an Externally-Led Patient Focused Drug Development Meeting (EL-PFDD). This report reflects the host organizations’ account of the perspectives of patients and caregivers who participated in the public EL-PFDD meeting, held virtually on September 1, 2022.

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- Center for Drug Evaluation and Research (CDER)
- Center for Biologics Evaluation and Research (CBER)
- U.S. Food and Drug Administration (FDA)

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The primary goal of patient focused drug development is to ensure that patients' experience, perspectives, needs, and priorities are meaningfully incorporated into drug development and evaluation. The information we learned today from patients can help investigators and sponsors to design better clinical trials by focusing on demonstrating that the drug has an effect on an outcome measure that patients report are clinically meaningful to them."



- SHEILA FARRELL, MD, REVIEWER, DIVISION OF RARE DISEASE AND MEDICAL GENETICS, FDA

Executive Summary and Key Insights about Living with Galactosemia

Galactosemia is a rare, severe, and highly variable condition caused by an inborn error of metabolism, characterized by the inability to metabolize the sugar galactose. There are several forms of the disease, which are caused by different defects in the galactose metabolic pathways; not all of the different forms of galactosemia are included in newborn screening tests. All infants with classic galactosemia born in the United States are now detected by newborn screening, which enables the rapid dietary restriction of galactose. However, galactose is produced endogenously, so despite restricting galactose in the diet, most individuals living with galactosemia have a constellation of long-term complications. These can include cognitive difficulties, speech and language difficulties, neurological problems, socio-emotional problems, low bone mineral density, primary ovarian insufficiency among girls and women and a pre-pubertal growth delay. The long-term outcomes for patients living with galactosemia are variable.

Together, GF and NORD hosted the Galactosemia EL-PFDD meeting on September 1, 2022. This meeting was held to provide patient and caregiver perspectives on the symptoms and burdens associated with galactosemia in daily life, as well as the massive unmet treatment needs experienced by families who live with galactosemia every day.

The objective of the meeting was to increase the understanding of how patients, families and caregivers experience and manage galactosemia, the factors that are considered when the treatments are chosen, and side effects and complications of current available treatments. This may, in turn, help research and the FDA understand patient preferences when developing new therapies and evaluating the benefit-risk for new treatment options.

"We hope that by sharing our unique stories, we can both educate and advocate for future galactosemic patients and their families. - Natalie, Caregiver of a 25 and a 22-year-old patient"

The voices of people living with galactosemia, their families and their caregivers were heard through courageous patient and caregiver testimonies, live polling of the broader audience, a chat discussion during the meeting, and a post meeting survey. The meeting was held virtually to enable as many community members to participate as possible and to allow many different voices to be heard. The EL-PFDD meeting attracted registrations from 612 patients, caregivers, researchers, and other advocates, and the live event was attended by 350 participants.

Testimony and facilitated discussion resulted in a number of key insights about living with galactosemia:

- 1. Individuals living with galactosemia experience many symptoms and have variable outcomes.** Neurological complications include learning disabilities, cognitive delays, apraxia, speech impairments, and memory problems. Pre-ovarian insufficiency in female patients, anxiety and depression, tremors and seizures as well as many other health effects impact life on a daily basis.
- 2. The impacts and challenges of galactosemia persist for a lifetime.** Communication challenges impact relationships and the ability to socialize. Individuals living with galactosemia experience many challenges with education, employment and sports and they have to work much harder than others at school, to obtain a job and to maintain employment. Some are unable to live independently. The social and financial impacts on families are enormous.
- 3. Patients and families worry about disease progression and the emergence of new symptoms including seizures.** Individuals living with galactosemia worry that their disease will worsen and that they will be unable to live independently in the future.
- 4. The leading galactosemia treatment strategy is to eliminate galactose from the diet, which is not sufficient to prevent long-term challenges.** Endogenous galactose production means that a galactose-free diet does not prevent the disease.
- 5. Galactosemia has tremendous unmet treatment needs.** There are no FDA-approved treatments to address the underlying cause of galactosemia. Throughout life, neurological damage continues to accumulate, including the development of seizures and decreased muscle.
- 6. Families try many types of treatments and therapies to help their children learn, to thrive socially, and to have their best chance at a full life.** Treatments are mostly targeted towards symptom management. Many take vitamin D and calcium for bone health, along with probiotics and other medications. Many engage with therapies including speech, feeding, occupational and motor therapies, however despite their persistent hard work, individuals with galactosemia may still fall behind. Some treatments come with side effects, therapy can be incredibly costly, and many individuals and their families feel exhausted and overwhelmed.
- 7. Families are desperate for more effective treatments to prevent or minimize the long-term complications of galactosemia.** Research and development in this area is desperately needed. Many would like to participate in clinical trials to ensure better outcomes for themselves and for others.
- 8. Newborn screening is critical yet even with early identification of infants with galactosemia, available treatments may not be effective.**



Cassidie Archuleta began her advocacy work in 2009 when her first and only child, Alenna, was diagnosed with classic galactosemia.

Galactosemia Externally-Led PFDD Rationale and Design

The patient perspective is critical in helping the US Food and Drug Administration (FDA) understand the context in which regulatory decisions are made for new drugs. Externally-Led Patient-Focused Drug Development (EL-PFDD) meetings provide opportunities for patients, their families, and caregivers to share critical information about the impact of their disease on their daily lives and their experiences with currently available treatments. Patients' experiences provide valuable insight for the FDA and other key stakeholders including researchers, medical product developers and healthcare providers.

The goals of this meeting were as follows:

- Provide researchers, drug developers, and the FDA with a robust understanding of patients' and caregivers' experiences with galactosemia, including how individuals view their quality of life, which aspects of the disease are most challenging for them, and how they currently address their disease symptoms.
- Identify key insight for research and clinical trial design from individual affected by galactosemia and their caregivers, so that the outcome of potential therapeutics can be measured in ways that are both clinically sound and therapeutically impactful.

GF and NORD collaboratively prepared this Voice of the Patient report, a high-level summary of the perspectives generously shared by patients living with galactosemia and caregivers who participated in the September 1, 2022, EL-PFDD meeting. The GF and NORD have provided this Voice of the Patient report to the US FDA, government agencies, regulatory authorities, medical product developers, academics, and clinicians, and it is publicly available for the many stakeholders in the galactosemia community.

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 galactosemia. The hope is that this information will catalyze better treatments and ultimately a cure for those affected by this disease.

Note that the input received reflects a wide range of galactosemia experiences, however not all symptoms and impacts may be captured in this report.



A video recording of the entire EL-PFDD meeting for galactosemia can be viewed at: rarediseases.org/externally-led-patient-focused-drug-development-meeting-for-galactosemia/

Galactosemia EL-PFDD Meeting Summary

The Galactosemia EL-PFDD meeting was co-moderated by **Debbie Drell**, Senior Director of Membership, NORD, and **Scott Saylor**, Board Member, Galactosemia Foundation.

Peter Saltonstall, the President and CEO of NORD, and **Nicole Casale**, President of Galactosemia Foundation officially opened the meeting and thanked all meeting participants for their attendance. **Judith Fridovich-Keil, PhD**, professor at the Department of Human Genetics at Emory University School of Medicine provided a clinical overview of galactosemia as a foundation for the meeting.

Dr. Sheila Farrell, a pediatrician and a reviewer in the Division of Rare Diseases in Medical Genetics at the US FDA, provided welcome remarks on behalf of the FDA. She described the role of the FDA in protecting public health, provided an overview of the drug development process, and explained that while the FDA does not develop drugs, they are integrated through the drug development process. She identified the different points during the drug development process where patient perspectives are important and necessary. Dr. Farrell thanked all the patients and their families for taking the time to participate in this meeting.

Debbie Drell and Scott Saylor provided an overview of the meeting structure and introduced the discussion format. They invited patients, families and caregivers of individuals living with galactosemia to contribute to the discussion through online polling, by speaking through Zoom, by sharing and asking questions using the online chat, and by emailing additional comments.

The Galactosemia EL-PFDD meeting was structured around two key topics. The morning session was structured around *Topic 1: Living with Galactosemia—Burdens and Symptoms*, which addressed the economic, emotional, and professional toll on patients, caregivers and family members. The afternoon session addressed *Topic 2: Perspective on Current and Future Treatments*, dealt with the unmet treatment needs and hopes for future galactosemia therapies. The meeting agenda is in **Appendix 1**.

The morning session continued with a panel of patients and caregivers who were selected to represent a range of experiences of patients and families living with galactosemia. Meeting attendees participated in online polling and Debbie Drell and Scott Saylor moderated a discussion between a live Zoom panel and meeting participants. Galactosemia community members shared their perspectives and connected over a live chat.

The afternoon session opened with a panel of patients and caregivers who spoke about their galactosemia treatment experiences, and their hopes for future treatments. Debbie Drell and Scott Saylor again invited meeting attendees to participate in online polling, to be part of the moderated discussion over zoom, to share experiences and support one another over the live chat. At the end of the meeting, **Brittany Cudzilo**, Vice President of the Galactosemia Foundation, provided a reflective summary of the key messages heard throughout the meeting. Debbie Drell and Scott Saylor concluded the meeting with a heartfelt thank you to all the participants and attendees. To include as many voices as possible, additional comments were accepted by email for 30 days following the meeting.

The biographies of all meeting speakers and a list of those who chose to speak during the meeting are included in **Appendix 2**. **Appendix 3** includes the meeting demographic, polling and discussion questions and demographic polling results are in **Appendix 4**. Online meeting poll results are included in **Appendix 5**. Although an additional survey was conducted after the meeting, the results were similar to the online meeting poll results, so are not included in this report. Additional patient comments that were submitted after the meeting are included in a separate PDF with selected excerpts included in the body of the VOP report.

Clinical Overview of Galactosemia¹



What is galactosemia and what causes it?

Galactosemia is a rare disease caused by an inborn error of metabolism, characterized by an inability to metabolize the sugar galactose. There are several forms of the disease. Galactose is consumed by humans in milk and dairy products, and importantly, small quantities of galactose are produced endogenously by the body.

Galactosemia is caused by deficiencies in the enzymes of galactose metabolism, which make up the Leloir pathway. Different forms of the disease are caused by different defects in the Leloir pathway. Type one galactosemia, or classic galactosemia (CG) is caused by a deficiency of the galactose-1-phosphate uridylyltransferase gene, or GALT, which is responsible for breaking down galactose. Galactosemia is passed down genetically through two autosomal recessive variant genes. In rare cases, a patient may have a new variant, not inherited from a parent. Patients with classic galactosemia, typically have a profound loss of GALT activity. Patients with clinical variant galactosemia, have a milder form of the disease, with residual (1-10%) GALT enzyme activity. Patients with Duarte galactosemia or biochemical variants have a specific combination of GALT alleles, with about 25% residual GALT activity intact, and recent studies show that these individuals are asymptomatic.

In addition to GALT, the other enzymes involved in the Leloir pathway include galactokinase (GALK), UDPgal 4'-epimerase (GALE) and mutarotase enzymes, responsible for Types 2, 3, and 4 galactosemia, respectively.

The prevalence of galactosemia in the United States is about one in 50,000 births, which equates to about 80 newborn diagnoses each year. There are currently about 3000 people in the United States living with galactosemia.



How is galactosemia diagnosed?

All infants with classic galactosemia born in the United States are now detected by newborn screening, which enables the rapid dietary restriction of galactose; unfortunately, not all of the different forms of galactosemia are included in newborn screening tests. In the US, newborn screening for classic galactosemia began in the 1960s, but it wasn't until 2004 that all states added galactosemia testing to their panel. Screening approaches vary from one state to another and can include a subset or combination of different analyses including GALT activity, total galactose or Tgal (galactose plus gal-1P activity combined), and GALT mutation testing. A positive screening test result requires a more rigorous follow up to confirm the diagnosis.



What are the symptoms of galactosemia?

Individuals living with galactosemia are born healthy but experience an acute metabolic crisis when they are exposed to galactose from breast milk or formula. Following their initial galactose exposure, symptoms begin with jaundice, vomiting, diarrhea, failure to thrive and then rapidly progress to cataract formation, liver and spleen damage, sepsis and even death. Once a diagnosis is made and infants are switched to an appropriate formula, metabolic symptoms begin to resolve. Dietary restrictions save an infant's life in the short-term but do not prevent lifelong complications from the disorder, as galactose is produced endogenously and damage continues to accumulate.



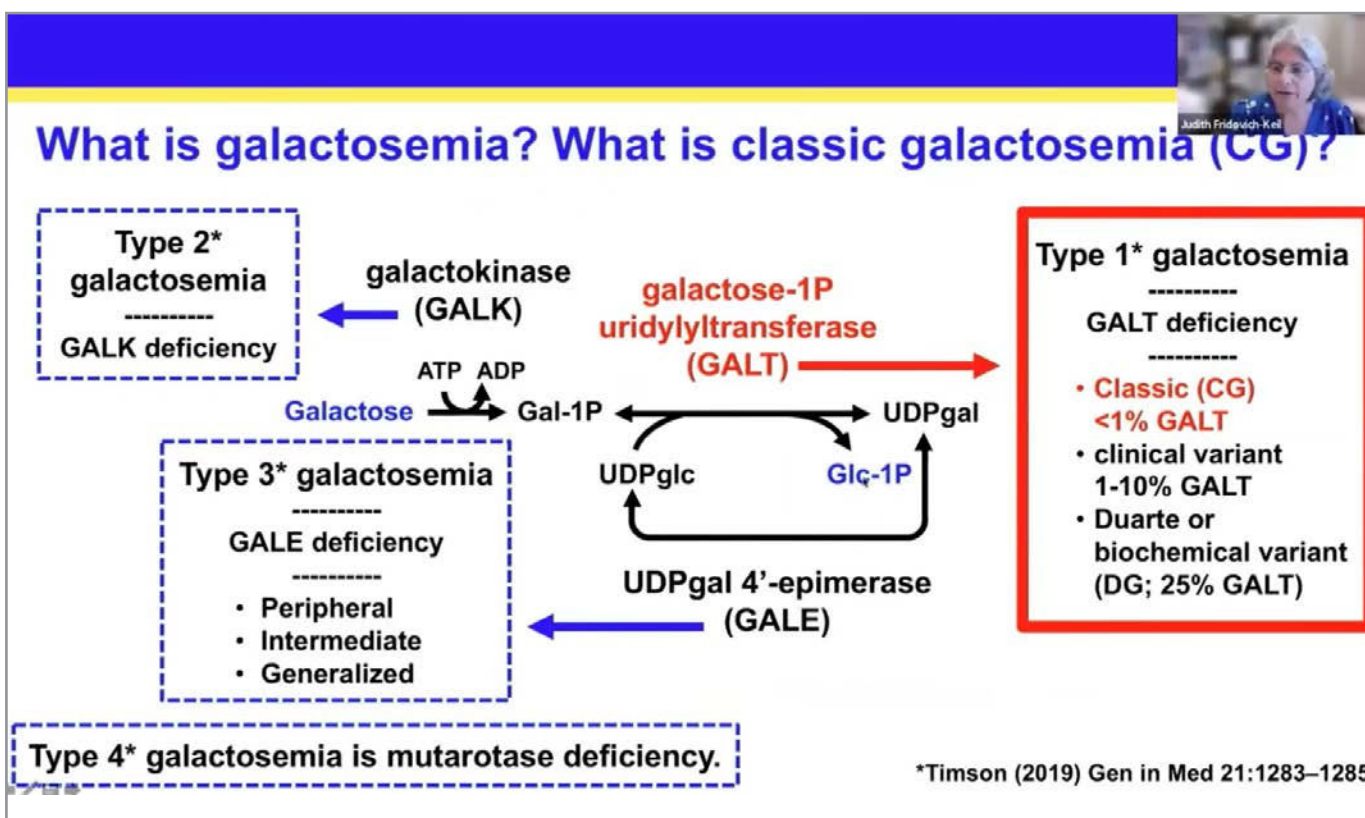
How is galactosemia currently treated and managed?

The primary treatment option is the immediate and continued dietary restriction of galactose. Most individuals living with galactosemia receive extensive therapies including speech and occupational therapies, as well as special educational services.



What are the outcomes for individuals living with galactosemia?

The challenges of galactosemia are lifelong, lasting into adulthood. Despite the restriction of galactose in the diet, most individuals living with galactosemia have a constellation of long-term complications, including cognitive difficulties, speech and language difficulties, neurological problems, socio-emotional problems, low bone mineral density, primary ovarian insufficiency among girls and women, a pre-pubertal growth delay, and challenges with adaptive behavior. The long-term outcomes for patients living with galactosemia are also variable from one patient to the next.



Judith Fridovich-Keil, PhD, Professor of Human Genetics at Emory University School of Medicine, presenting an overview of Galactosemia

¹ Most of this content was extracted from the September 1, 2022, presentation by Judith Fridovich-Keil, PhD, Professor, Department of Human Genetics, Emory University School of Medicine.

Demographic Snapshot of Meeting Participants

The Galactosemia EL-PFDD meeting was attended via a livestream webcast, with 350 live participants. An online chat allowed patients and community members to add their responses during the presentations, to ask each other questions and most importantly, to reach out to support each other.

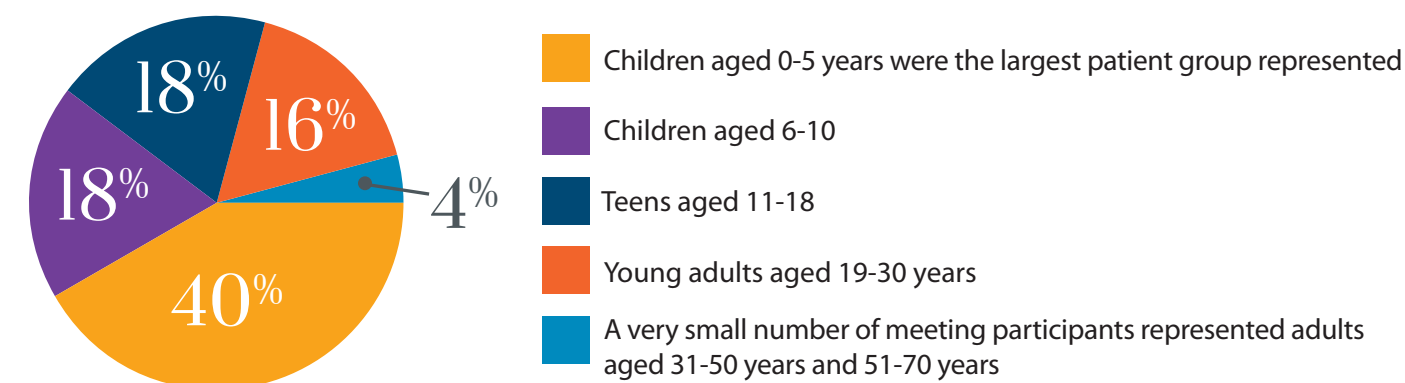
"I love all these people signing in, in support of our Galactosemia community, like the close friends, aunts, uncles, nannies, and more. Thank you to all of you for your support!!"

- Cassidie, Caregiver of a 13-year-old patient (via chat)

A demographic snapshot of meeting attendees was created from online polling and the chat.

- Polling data demonstrated that majority of meeting attendees were a caregiver of someone with classic galactosemia (92%), and the rest were individuals living with galactosemia, either classic galactosemia (4%) or clinical variant galactosemia (4%).
- Chat participants identified themselves as patients, parents, grandparents, aunts and nannies, aunts and uncles, cousins, good friends of patients, as well as patient advocates.
- Chat participants represented many locations across the US: Maryland, Arkansas, Ohio, Nebraska, New Jersey, New York, South Carolina, North Carolina, Tennessee, Pennsylvania, Iowa, Georgia, Florida, California, Virginia, Michigan, Kentucky, Oklahoma, Wisconsin, Texas, Missouri, Illinois, Minnesota. Others attended from Canada, Sweden, Singapore and Zimbabwe. This is consistent with the results of online polling.
- Over half of patients represented at the meeting were male (58%), and the rest were female (42%). No one identified as 'other' or 'preferred not to identify'.
- Most of the poll respondents represented individuals with classic galactosemia diagnosed by genetic testing (88%), followed by those who had diagnosis from newborn screening but did not have a diagnosis from genetic testing (10%), and the rest were unsure of the genetic diagnosis.

Representation by Age



There were no individuals aged 71 years or older represented. The oldest meeting attendee living with galactosemia was Jerry, who at 62, is one of the oldest living galactosemia patients.

Demographic questions are listed in **Appendix 3**, and online demographic polling results are shown in **Appendix 4**. Meeting attendance was dynamic, with more participants joining throughout the day, so demographics may have shifted throughout the meeting. Note that not all of the meeting participants chose to participate in online polling.



“
It is now clear to researchers that this disease is progressive, and it will likely get worse as a patient ages. Personally, I have already seen a slight regression in Jacob’s ability to walk, balance and the time it takes for him to respond to a question.”

- NATALIE, CAREGIVER OF A 25 AND A 22-YEAR-OLD PATIENT

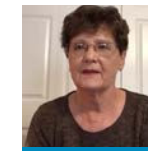
Voice of the Patient - Topic 1: Living with Galactosemia—Burdens and Symptoms

This section highlights some of the key galactosemia themes that were emphasized throughout the meeting, as well as the online meeting poll results. Galactosemia symptoms, impacts and burdens on daily life, and worries are illustrated with quotes from patients and community members.

Galactosemia is diagnosed very early in life and is always traumatic. This theme was mentioned throughout the meeting but not captured in the polls. Parents described how their newborns became gravely ill, and many infants nearly died before a galactosemia diagnosis was made. Mothers were horrified to find out that it was their breastmilk that was causing their child’s symptoms. Several parents spoke of children who died within the first weeks or months of life.



“My daughter Alenna was diagnosed with classic galactosemia on her fifth day of life. Before diagnosis, she had elevated bilirubin levels, projectile vomiting, and had diarrhea that gave her such an intense diaper rash, her skin would peel at the slightest touch. Once diagnosed and put on dairy free formula, her acute symptoms faded.” - Cassidie, Caregiver of a 13-year-old patient

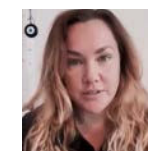


“Jerry was in a coma at two days, three days, diagnosed right before he [would have] passed away. ...I had lost two other children from this disease. My second and my third children were both girls, who appeared to be healthy at first, the girls passed away undiagnosed, at one and a half months of age each.”
- Elaine, Caregiver of a 62-year-old patient

“He was in and out of hospitals for five weeks until finally we got the correct diagnosis. At that point he was severely jaundiced, nearly comatose, and blind in both eyes due to severe cataracts and a retinal hemorrhage. An MRI showed serious damage to his brain.”
- Brian, Caregiver of a 29-year-old patient

Many caregivers reported that their loved ones experienced progressive and degenerative symptoms. This important point was raised several times throughout the meeting. Several patients only first started to experience seizures years after diagnosis. Patients and caregivers expressed fears and worries about a decline in their abilities as they aged.

“Since his body is constantly producing galactose, the same toxin that made him so sick, classic galactosemia is considered progressive. We insisted we’d travel anywhere in the country for the best treatment available, but unfortunately, our medical team explained to us that galactosemia is an ultra rare disease and research is scarce.”
- Amber, Caregiver of an eight-year-old CG patient



“As Penelope grew, I learned more about her struggles. One week she could talk, then the next day she’s babbling and reverting to toddler, baby like behavior.”
- Gillian, Caregiver of a five-year-old patient

SYMPTOMS AND IMPACTS

Learning disability, cognitive delay, speech impairment, and impaired memory

Learning disabilities, cognitive delays and speech impairment are frequently experienced galactosemia symptoms and were also selected as the most troublesome symptoms in the polls, in **Appendix 5 Q1 & Q2**. Impaired memory is a closely related symptom.

Children with galactosemia are often bright and intelligent, yet typically struggle to meet developmental milestones, have different learning styles than their peers, and some are held back in school. Some experience speech apraxia, and have a hard time verbalizing. Many of those living with galactosemia have challenges adjusting to change.

“When I was younger, I had a lot of difficulties with school, and just speech delays, learning disabilities. And I had to go out of school, and be held back, and then have one-on-one tutoring that helped me a lot. And even after that, I’ve still been having some kind of problems with learning disabilities.” - Jamie, Patient, 26 years old

“Along with learning delays, he deals with speech delays, fine motor issues, and behavioral problems. He is quickly falling behind his peers. When Adrian is asked a question, he often takes a while to form his thoughts and many times, he is passed by because people think he doesn’t have anything to say. ... Behavior is also difficult for Adrian, particularly when things do not go according to his plan.”

- Amy, Caregiver of a six-year-old patient

“He was just recently diagnosed with a subcategory of central auditory processing, which is like an organizational output deficit. He has processing issues, auditory visual processing, handwriting is well below age level, gross motor is affected. Cognitively, the speech and language disorders have impacted him across his academics. He struggles with math, he struggles with all the aspects of language, vocabulary, things like that, syntax, everything.” - Kelly Foley, Parent



Throughout my school days, I was always a low-average student. Math was always my most difficult subject and still is today. I struggle with making change, leaving tips at restaurants, percents and telling time. Seeing signs in stores for a percentage off sale is hard for me to understand. Overall, it has been hard in many life circumstances because I struggle with any math that I need to process at the moment. It was not until I met other people living with galactosemia that I realized we all had comprehension similarities, including math.”



**- MAUREEN,
PATIENT,
50 YEARS OLD**

Patients and caregivers described how learning disabilities, cognitive delays and speech impairments have affected them and their loved ones, particularly at school.

“I had problems in school doing English and math. Certain subjects, it affected us a little bit more than others. Having English, making things to make sense. I think it’s easier for us talk than it is for us to write stuff. It is all jumbled. So it takes 10 times to write out a paper to make it make sense. And then math ... it really affects us a lot.” - Brett, Patient, 35 years old

“School was hard for Jerry. He was behind for his age in everything, from reading to comprehension. He was taken out of the classroom for special education. He was bullied for what he didn’t and couldn’t answer in regular class. ... He had a hard time verbalizing what was bothering him, and would become angry and frustrated when the stresses and anxiety became too much for him.” - Elaine, Caregiver of a 62-year-old patient

Some like Erin, were told that they would not be able to learn:

“Some of my teachers, even throughout my schooling, have told my parents - and they’ve told me many times - that I’m just never going to learn whatever the subject they’re teaching. ... It’s hard to tell them I just need a little bit extra help.” - Erin, Patient, 27 years old

Patients discussed their challenges with school and how hard they had to work to keep up. Some struggled to speak up, had a hard time focusing and making decisions on their own. Some described challenges with time management and remembering important information. Parents shared the challenges and delays in toilet-training their children.

Liver failure, jaundice, or sepsis as well as failure to thrive and lethargy (infants)

Liver failure, jaundice or sepsis are frequently experienced galactosemia-related health concerns and were also selected as the second most troublesome symptoms in the online poll. Liver failure and jaundice are often the first traumatic signs of metabolic crisis, appearing shortly after birth. Failure to thrive and lethargy are symptoms that also appear shortly after birth.

“Jerry was in a coma with yellow skin and high bilirubin count, being fed through an IV. But, like his sisters, he wasn’t getting any better.” - Elaine, Caregiver of a 62-year-old patient

“Unfortunately, an hour after getting home from that doctor’s visit, I was holding her limp and almost lifeless in my arms. Penelope was hospitalized with metabolic crisis, liver failure, and hypoglycemia. A little crash cart rested outside her room, in case of cardiac or respiratory arrest.” - Gillian, Caregiver of a five-year-old patient

“Typically, newborns have a bilirubin level of around five. Newborns with jaundice may have 10 to 15. Jacob’s bilirubin level was 44, which is incredibly dangerous and causes what is called kernicterus. This is when bilirubin crosses the blood brain barrier and damages the brain and spinal cord. His situation was critical, and he was immediately taken to the nearest NICU.” - Natalie, Caregiver of a 25 and a 22-year-old patient

Parents described the first traumatic weeks of life including multiple blood transfusions, hematology consults and conversations with transplant teams.

Tremors and/or seizures

At the EL-PFDD meeting, tremors and seizures were selected as the third most troublesome galactosemia-related symptom. Parents described many different types of seizures yet for some, seizures only started later in life, bringing the question of disease progression to the forefront.

“At two years old, I noticed absence seizures. . . . Unmedicated, she has seizures every hour in clusters. I was told she’s now at risk for status epilepticus, a seizure that never stops, and that she could die from. I carry a rescue med everywhere I go. I also carry the weight of her dying in her sleep. Seizures complicate everything. A simple ear infection and stress from her sensory processing disorder can trigger more seizures.” - Gillian, Caregiver of a five-year-old patient



“Garrett was in fifth grade when he experienced his first seizure event. We were told that every child could have a seizure and that if he started having any more, that we would need to have those looked into. Later that same year, he had a clustering event which led him to taking seizure medication to assist in controlling them.” - Heather, Caregiver of three CG patients, 24, 21 and 16 years old

“When I was 20, I developed seizures and that was difficult for three years until I was put on medications.” - Jamie, Patient, 26 years old

“Around age 55, Jerry started having visual seizures, which are now under control with medication. In hindsight, he may have been having small seizures all along. We noticed short moments of blanking out or disengagement, but he seemed to snap out of it when we’d call his name.” - Elaine, Caregiver of a 62-year-old patient

Primary ovarian insufficiency

In live polling, primary ovarian insufficiency (POI) was identified by approximately 50% of females living with galactosemia. Patients and caregivers described what POI looks like for themselves and their loved ones. During the meeting and in this report, a number of similar terms including primary ovarian insufficiency, pre-ovarian insufficiency, and premature ovarian failure (POF) were used interchangeably.

“After going through puberty, I began to have unusual symptoms, such as hot flashes, mood swings and night sweats. After about a year of experiencing these symptoms and having blood work, I was diagnosed at the age of 18 as having premature ovarian insufficiency, which is a direct result of galactosemia.” - Maureen, Patient, 50 years old

“Most girls with galactosemia are infertile caused by POF, premature ovarian failure. POF can cause osteopenia and osteoporosis. Treatment for this is hormone replacement therapy, birth control, indefinitely. It’s heartbreaking. They also have tend to be delayed in puberty, some only reaching puberty with medical intervention. Most people think of puberty as a dreadful time. We rejoice in it. Recently, Alenna has shown signs that she’s naturally maturing. You would think that this would be a great relief to me, but the truth is, about 80% of girls that naturally mature will then go into POF after only a few years.” – Cassidie, Caregiver of an 13-year-old patient

One of the biggest impacts of POI is infertility, and many patients are severely impacted by not being able to have biological children.

“I was told long ago that having galactosemia was like looking through a glass window and watching others being a part of eating foods I could not have. I could see them through the window and through the glass, but could not break it or join them. This theme is true for infertility. I can see moms and children through the glass, but cannot experience what it is truly like to be a mom. All my life I longed to be on the other side of that window.” - Maureen, Patient, 50 years old

“When I heard that I likely would not be able to have children, I felt like a small piece of me died. I played it off like I didn’t care. But in reality, I was wanting to have children and I felt it was so unfair that I likely wouldn’t have the chance.” – Shelby, Patient, 31 years old

Patients discussed the heartbreak of POI-related infertility and many caregivers shared their sadness that their daughters with galactosemia may not have children of their own. Some shared stories of their successful conceptions and adoptions.

Anxiety and depression

Anxiety and depression were frequently identified as some of the most troublesome symptoms. Some have generalized anxiety while others described more specific anxieties.

“One of my daughters really struggles with anxiety and sleeping alone.” - Tara, Caregiver of two CG patients, 10 and 8 years old

Amy’s son developed anxiety about blood draws required for his clinical trial. “He is fortunate that once he has an IV in his arm, he does really well. However, the anxiety that leads up to the blood draw is pretty extensive.”- Amy, Caregiver of a six-year-old patient

“The impact of POI, secondary to having galactosemia, was very difficult. In my twenties, I just wanted to be like any other woman my age, socializing and having fun. However, I was depressed and had a difficult time making friends and meeting new people. There were many nights where I cried myself to sleep.” - Maureen, Patient, 50 years old

Many caregivers discussed how their children sometimes felt like they were a “burden” because of the accommodations that they require to deal with galactosemia symptoms.

Trouble eating, vomiting

Trouble eating is a challenge for many individuals living with galactosemia. This can sometimes be related to apraxia or to sensory processing disorders. Some experience frequent stomach aches as well as cyclical vomiting syndrome, which one caregiver described as “a migraine that affects the stomach.”

“My child does have some issues with foods. He hates textures that he has to chew on quite a bit. He’ll chew and chew and finally give up and usually spit it out.”

- Amy, Caregiver of a six-year-old patient

“My 10-year-old son has a vomiting issue as well. This is something we have been working on since he was a baby...he struggles to move his food around in his mouth due to his apraxia and often swallows his food whole. Then it comes back up within a couple hours. I also notice that he vomits when he is constipated. My very underweight child can’t afford to have his food coming up.” – Cadence, Caregiver of a 10-year-old patient (via chat)

“My daughter has been having the vomiting since she was about two. And I actually did not know that others were having that until I saw it on the chat. So every night she sleeps with a container next to her because she will wake up between three and five in the morning and vomit. It is a major issue for school. Once she starts it, she will vomit every two hours or so until about noon that day.” - Megan, Caregiver of a seven-year-old patient

Many caregivers discussed how their children vomited frequently and needed to have barf bags or bowls nearby.

Low muscle tone (hypotonia) and motor skill impairment

Low muscle tone, hypotonia, or impaired motor skills impairment are important galactosemia symptoms. These symptoms can cause mobility challenges, a lack of coordination, and balance issues which can lead to falls.

“Alenna would get into a crawling position and then just stay there. Or, while standing, she would look so determined to take a step, yet some unknown force was holding her back.”

- Cassidie, Caregiver of a 13-year-old patient

“Apraxia is more than just a speech disorder. It is a movement disorder that we can see affects Jake every day in his life, especially when it comes to doing sports. He would love to be able to do some of those fine motor sports, but it involves him extra practicing more than any other child to try to master those skills.”

– Kristine, Caregiver of a 15-year-old CG patient (via chat)

“High levels of galactose during the newborn period caused a domino effect of secondary issues. Jacob has cerebral palsy. This means damage to areas of the brain affecting motor movement like walking, writing or eating.” ... Balance issues have caused falls, trips to the ER, and several sets of stitches.” - Natalie, Caregiver of a 25 and a 22-year-old patient

Parents discussed how a lack of fine motor skills and poor hand-eye coordination make handwriting, using scissors and getting dressed a challenge, especially if shoelaces, snaps and buttons are involved. Low muscle tone also affects the bladder so often children living with galactosemia need to go to the bathroom frequently.

Other galactosemia health concerns

Sensory processing disorder, low bone density/osteoporosis and cataracts are galactosemia-related health concerns selected in the polls. A large number of other galactosemia-related symptoms were mentioned in the meeting, discussed in the chat, and mentioned in the submitted comments. These include: GI issues such as Crohn’s disease/IBD, chronic constipation; dental issues such as cavities, low enamel, demineralization; pain and fatigue; bleeding disorders, anemia and abnormal blood cell counts; cerebral palsy; cardiac issues; delayed adolescent development; impaired kidney function; alopecia; urinary tract infections; osteoarthritis; Chiari malformation; and dysmorphisms. Some of these are illustrated below.

GI issues including chronic constipation. This can be related to low muscle tone and is often painful. Parents pointed out how disruptive constipation can be to daily life and how it affects mood and sleep.

“Day in and day out if they’re constipated, and they’re not going to the bathroom regularly, it can really affect the tone in our whole home.” - Tara, Caregiver of two CG patients, 10 and 8 years old

“You think of it as such a small thing, and it’s not. It’s just not. It will be days of pain. She will be in pain, crying on the floor in a fetal position. ... We’ll be on a trip and it just will destroy the trip. ... It’s definitely something that really affects our day-to-day lives.”

- Gillian, Caregiver of a five-year-old patient

Dental issues. Underdeveloped baby teeth were discussed during the meeting along with frequent cavities, missing adult teeth, demineralization, and hypoplasia (thin or no enamel on certain baby teeth).

“Dental issues are common. I had lots of cavities when I was younger, even though I would brush my teeth and I would floss. I felt like I took care of my oral health, but I would usually get bad news at the dentist office.” – Shelby, Patient, 31 years old (in the chat)

Pain and fatigue. Although mentioned throughout the meeting, pain and fatigue seemed to particularly affect older patients.

“Even at age 25, Jacob suffers from pain in his hips, legs and joints.”

- Natalie, Caregiver of a 25 and a 22-year-old patient

“As he’s older now, fatigue has really kept him so he can only work two days a week.”

- Elaine, Caregiver of a 62-year-old patient

GREATEST IMPACTS

Patients and their families described the many lifelong impacts of galactosemia.

"I've been told my son is an extremely hard worker. Sometimes our kids work so hard to overcome this, but it does not mean that their needs are being met."

– Scott, Caregiver of a 15-year-old CG patient

"Galactosemia will forever impact Garrett's life. He will always have to work harder to complete a job that comes easier to most people. He will forever have to verify the food that he is eating is safe. His delays will continue to be something that he will have to work on a daily basis to continue to strive for a better life."

– Heather, Caregiver of three CG patients, 24, 21 and 16 years old

"Jacob will never be able to live independently, have a job or make life decisions on his own. While diagnosed earlier, Anna will still experience challenges with communication, anxiety, problem-solving, social situations and fertility."

– Natalie, Caregiver of a 25 and a 22-year-old patient

During polling, the top three galactosemia impacts identified were: communication impacts, having meaningful friendships and attending social events. Poll results are in **Appendix 5, Q3** and described below.

Communication impacts

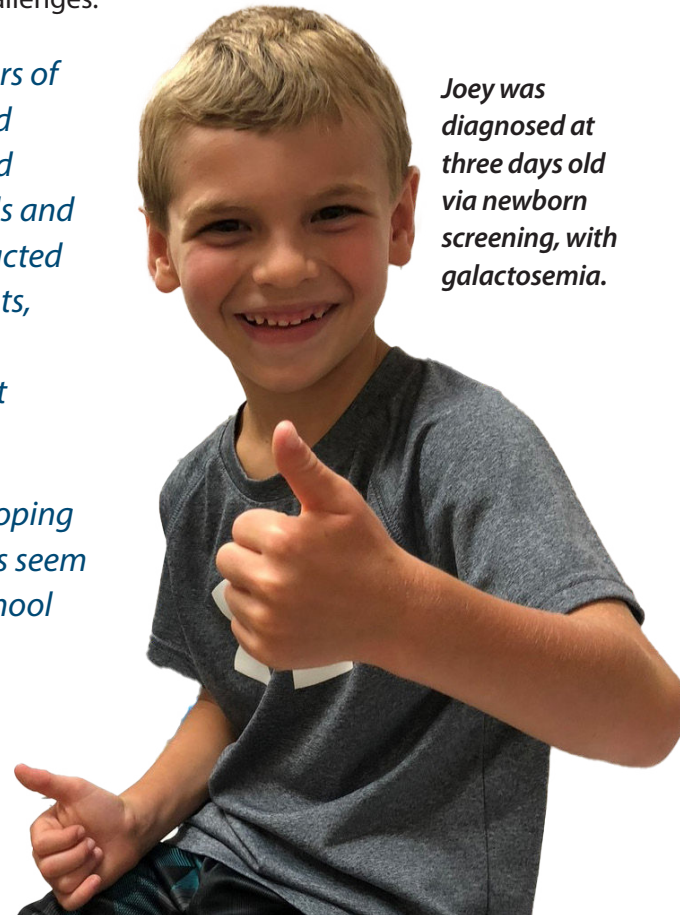
Communication is an important activity of daily life impacted by galactosemia, and this affects other important things such as having friendships, socializing, school and employment. Many parents spoke about how frustrated their children become due to communication challenges.

"Then there are the speech deficits, which cause errors of pronunciation, awkward pauses while speaking, odd substitutions of sounds or syllables. This then created barriers to communication not only between the kids and me, but also with their friends and family who interacted with them. It created frustration, behavioral outbursts, and made it difficult to make friends."

– Natalie, Caregiver of a 25 and a 22-year-old patient

"A lot of times I've noticed that she does seem to be searching for her words, and she has difficulty developing friendships as a result. Simply because conversations seem to be difficult for her. As she's entering the middle school years, I have a lot of concerns for her."

– Anne, Caregiver of a 10-year-old CG patient



Joey was diagnosed at three days old via newborn screening, with galactosemia.

"He has to practice and practice and practice just to be able to speak in front of people. He is a hard worker though. I don't think if I had half his issues, I would be strong enough and brave enough to stand in front of a class. Despite his issues, he does his best."

– Kelley, Caregiver of a 12-year-old patient

Biking or playing sports

Activities like biking, playing sports, skipping, and climbing, can be a struggle for individuals living with galactosemia. Cognitive issues, mobility challenges, hand-eye coordination, and fine motor skills can create difficulties for activities.

"I also have some issues with hand eye coordination. So things like playing sports are hard for me because of the hand eye coordination." – Kimberly, Patient, 47 years old

"She can't ride a bike yet. She's seven. She can't skip."

– Megan, Caregiver of a seven-year-old patient

"She was terrible at sports and anything that involved gross motor skills. She tried dance, tried gymnastics but she wasn't good at it and the other kids just never accepted her because she just couldn't catch on and had no coordination." – Janet, Caregiver (via chat)

Having meaningful friendships

In addition to communication issues, some individuals living with galactosemia have challenges understanding boundaries and interpreting social cues. Many spoke about feeling isolated and lonely and are all too aware of their social issues and limitations. Some children spend so much time in speech therapy and other therapies that they don't have time left to play with others.

"Meaningful friendship, that's another part that is hard for me. ... In middle school, I was the odd one out, as I did not make eye contact when speaking and had tough time with making friends. My peers ridiculed me and ostracized me for most of my middle school days. ... To make small talk or have eye contact is very, very difficult for me." – Shelby, Patient, 31 years old

"One of the hardest things for Jerry, ... is being on the edge of life. He's almost average, but he has lots of small difficulties. And learning, greeting, making friends, sleeping and living independently, always in special ed." – Elaine, Caregiver of a 62-year-old patient

"I think that the social impairment of this disability is almost like someone of autism. Aiden is in first grade and he's just now starting to make friends. He's just now starting to understand the aspects of needing people in your life. I don't know if anybody else has that issue with their kids, but mine just doesn't really care about other people, or he doesn't understand other people's needs or emotions or the things that... It's always one sided, if that makes sense." – Amy, Caregiver of a six-year-old patient

“Socially, slow processing leaves her out of a conversation. By the time she’s had time to process the information, people have already moved on, this frustrates her. She feels left out and in constant need to catch up.” - Cassidie, Caregiver of a 13-year-old patient

Attending social events at someone’s home or in public

Not only can patients be socially excluded for their communication and relationship challenges, but they can also be excluded with regards to the type of food being served.

“Many people do not understand that saying your child is ‘dairy-free’ is not a choice we are making for him -- it is to save his life.” - Allie, Caregiver of a two-year-old patient

“Although the diet may have isolated me from social events, my mom always made sure that I had a special treat to bring with me, not just enough for me, but enough for everyone to share.” - Maureen, Patient, 50 years old

*“Recently he has struggled with not being able to consume the same foods as his friends at school, parties, and other social settings. It is hard to watch your child cry about not being able to have the pizza or cupcake. I try my best to always make sure there is a dairy free alternative for Kaden anywhere we go because I do not want him to feel left out.”
- Kayla, Caregiver of a six-year-old patient*

Other galactosemia impacts

Living independently, eating independently, sleeping and being intimate with a partner were poll options that were selected by some as one of their top three impacts. Galactosemia can exact a heavy emotional toll on caregivers and family members and it can financially impact the family.

Emotional impacts on caregivers and siblings. Many caregivers spoke about their mental health challenges including depression, anxiety and PTSD. Some parents experience guilt about neglecting other siblings because their children living with galactosemia require so much additional care and attention.

“As a mother of a young child battling this rare disease, my anxiety has greatly increased due to constantly reading labels, trying to decipher conflicting information about what is allowed and what is not, juggling appointments and blood draws, and managing the fear of going out to eat because you don’t know how a restaurant prepares their food and may not understand the questioning that comes from a parent.” - Allie, Caregiver of a two-year-old patient

“The fact that my daughter has so many medical, therapy, and tutoring appointments also means that our son who does not have classic galactosemia is affected as well. There simply is not enough time to bring him to the extracurricular activities he wants to attend. It is a difficult position for our family.” - Megan, Caregiver of a seven-year-old patient

The emotional impacts on caregivers and siblings were discussed at great length in the chat.

WORRIES FOR THE FUTURE

Poll results are presented in **Appendix 5, Q4**.

Worries that symptoms will worsen

This was the top worry, selected by most of the poll respondents. Many worry about their children’s hard-earned skills deteriorating, disease progression, while others worry when seizures will start. Some worry that any treatments being developed would be too late to reverse the damage already done.

*“It worries us that it’s going to get worse, the older that he gets. ... Our biggest fear is that he has worked so hard to master his skills and to shrink his gap between his peers that, he’s going to regress in all of it, and lose his confidence, and not be able to have his independent life. Not be able to have a family, kids of his own. And that his whole body, sunny disposition, personality’s just going to disappear and he’ll lose all of his confidence.”
- Kayla, Caregiver of a six-year-old patient*

“She’s having seizures at two and tremors. What’s going to happen 10 years from now. What are her seizures going to be?” - Gillian, Caregiver of a five-year-old patient

“I am terrified to think of what the future could bring too, with tremors and seizures and more impairments along the way.” - Megan, Caregiver of a seven-year-old patient

*“And then just overall, I think just the fear of the unknown. There’s just so many things that we just don’t know about this disease, and we don’t know what it’s going to look like.”
- Tara, Caregiver of two CG patients, 10 and 8 years old*

Inability to live independently, to work or pursue a career

The second and third most selected worries in the polls were the inability to live independently, and the inability to work and pursue a career. Some worry that their children would be dependent on them for their whole lives and others worry about who will care for their child when they can no longer do so.

*“Even though our son does pretty well, it’s still a concern for the future about living independently, cooking, driving, maintaining a job, all of that. Because you do see struggles with him with organizational skills. And I’ve heard that from a lot of galactosemia families with older children, around Jake’s age, about thinking about that future, and what that looks like, and will they have to be taken care of them, and will they have to live with them.”
- Kristine, Caregiver of a 15-year-old CG patient*

*“She has big dreams. She wants to be a veterinarian when she grows up. Sadly, she has mentioned her hesitation about being a vet, knowing just how much school they have to go through. I hate that galactosemia could rob her of her dreams.”
- Cassidie, Caregiver of a 13-year-old patient*

Some parents expressed worries about how their children would make the transition to adulthood, and if they would continue to access the services that they needed. One parent worried that her son would never understand how to manage his illness by himself.

Inability to have children and a family

As mentioned in a previous section, many patients are unable to have families of their own because of POI. For caregivers, worries about their child's inability to have children and a family was an extremely emotional issue as well.

"The inability to have children. ... That is something that has weighed on her for her entire life. Even as a two-year-old, she would put stuffed animals in her shirt to pretend that she was pregnant. This has been a dream of hers since I'm sure she can recollect memory. And I know that she would pick that as one of her worries."

- Cassidie, Caregiver of a 13-year-old patient

In the chat, caregivers pointed out that they believe that this worry is much bigger than indicated in the polls. Patients and caregivers discussed approaches to fertility and shared stories of successful conceptions as well as adoptions.

Other worries

Other worries include the inability to communicate needs, that relationships will suffer, the inability to finish education, the inability to find galactosemia-safe food, worries about staying healthy while living with galactosemia, and worries about falling.

“

I also have a really hard time with the fact that my girls probably won't be able to have children. ... Both of them, when they were in kindergarten, had to give a speech on what they wanted to be when they grow up and they both chose 'a mom'. ... And we're at the point right now where they're 8 and 10 years old, and I haven't shared that information with them yet."

- TARA M.

Voice of the Patient - Topic 2: Perspective on Current and Future Treatments

Galactosemia has a very high unmet treatment need. There are currently no FDA approved therapies for galactosemia. Although a strict galactose-free diet is absolutely necessary for all individuals living with galactosemia, this diet alone cannot prevent continuing neurological damage and galactosemia-related symptoms.



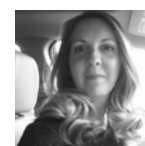
"The only management option for galactosemia is a restricted diet, and this will not prevent disease progression."

- Amber, Caregiver of an eight-year-old CG patient



"We were devastated to learn that even though we could remove galactose from her diet, her body would produce its own galactose, which would get in her bloodstream and damage her nervous system. For Charlotte, the effects of this damage include cognitive impairment, motor and speech issues, fine motor delays, and a poor memory."

- Tiffany, Caregiver of a nine-year-old patient



"Joey has followed a restricted diet since diagnosis day but is still affected by the neurological symptoms of this disease. ... His liver is functioning normally, and his galactose levels are in the acceptable range. However, the neurological impairments of this disease become more evident year over year."

- Tara, Caregiver of an 11-year-old patient

"My mom and dad were given a 32-page booklet entitled, 'A parent's guide to a galactose-restricted diet.' They were instructed to follow this booklet as I would need to be on a lactose-free dairy-free diet for the rest of my life, at that time, the only treatment for galactosemia."

- Maureen, Patient, 50 years old

"Anything that is safe and has some degree of effectiveness is better than what we have now."

- Pat, Caregiver of a 62-year-old CG patient (via chat)

CURRENT TREATMENTS

Poll results are presented in **Appendix 5, Q5** and illustrated with quotes, below.

Vitamin D and calcium supplements

Vitamin D and calcium supplements help to ensure that the bones, teeth and joints of those living with galactosemia are healthy. Some have their vitamin D and calcium levels tested on a regular basis and have DEXA scans to test for bone density.

"I've been taking calcium supplements pretty much all my life. When I was a kid, the chewies, the Flintstones, and then now I'm doing the pills and vitamin D supplements."

- Brett, Patient, 35 years old

“He’s on very high dosages of Vit D and Calcium (we monitor with doctors). His vit D levels have not improved the joint pain.” - Kelley, Caregiver of a 12-year-old patient (via chat)

A downside is that despite taking these medications for their entire lives, many living with galactosemia still experience vitamin D deficiencies, low bone density and fractures.

MiraLAX or other laxatives

Many reported taking MiraLAX on a daily basis, as well as probiotics, suppositories, and adding fiber and prune juice to their diets.

“I also need MiraLAX almost every other day. And I especially need it when I’m going on trips. Like, when we go on family vacations, that’s when I really need it.” – Erin, Patient, 27 years old

A downside of MiraLAX and other laxatives is that they are not equally effective for all individuals and some caregivers mentioned that MiraLAX can cause mood challenges.

Community members shared recommendations for solutions that had worked for them, including gluten-free diets, added dietary fiber, and MiraLAX.

Vitamin K and blood plasma transfusions were used to address the initial metabolic crisis, along with phototherapy for jaundice.

When he was a recently diagnosed newborn, “Joey underwent treatment by putting him under the bili lights. He received multiple vitamin K shots and numerous blood plasma transfusions. - Tara, Caregiver of an 11-year-old patient



*“Doctors performed two blood transfusions to clean his blood of the excess toxin. He had one seizure during this process. The NICU docs then gave us the sobering news that our baby may not survive the night, and we waited through what is still the worst night of my life.”
- Natalie, Caregiver of a 25 and a 22-year-old patient*

Medications for anxiety and depression including fluoxetine (Prozac) or busporin.

“I take medicine for my anxiety and it helps with my sleep.”– Erin, Patient, 27 years old

*“My son takes an oral dose of anxiety medicine every day and he was evaluated by a psychologist several years ago and diagnosed with OCD, all connected through the anxiety that comes from having galactosemia. ...His anxiety medicine can cause sleepiness.”
- Amber, Caregiver of an eight-year-old CG patient*

Hormone replacement medication

Many adult females living with galactosemia are required to take hormone replacement medications for POI.

*“At age 13, she began hormone replacement therapy, which continues indefinitely. Most female patients experience pre-ovarian insufficiency.”
- Natalie, Caregiver of a 25 and a 22-year-old patient*

Anti-seizure medications

Although just over a quarter of galactosemia patients reported experiencing seizures, those who do require lifelong anti-seizure medications. A downside for some, is that can take a long time to titrate the appropriate quantity of medication. A clear downside of rescue medications is that they cause depressed respiration which requires medical resuscitation.

“We did multiple trials trying to figure out what medications would help. And after three to four years, that they finally found the correct measure of medication that works, and that has been controlling my seizures.” - Jamie, Patient, 26 years old



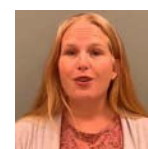
Garrett was prescribed anti-seizure medication after having a cluster seizure event in elementary school. “We are very fortunate that the first medication that was prescribed to him has worked well in controlling his seizures.” He was later prescribed additional anti-seizure medications for breakthrough seizures. “He also still takes this medication and will have to take it for the remainder of his life.” - Heather, Caregiver of three CG patients, 24, 21 and 16 years old

Other medications including investigational medications

Investigational medications. Several patients have participated in trials of an investigational medication that inhibits aldose reductase activity, lowering the levels of galactitol in the blood.

*Amber’s son participated in this trial. “While I can’t prove that Holden is on the drug instead of placebo, it’s clear that I can see his hand tumors are gone. He met his IEP goals for the first time since 2018. He learned to read and his teachers and therapists have taken notice of how well he’s doing as he masters grade level educational standards.”
- Amber, Caregiver of an eight-year-old CG patient*

Although the results were promising for some, the downsides of investigational medications and participation in clinical trials include the safety risks, uncertainty of outcomes especially as patients are not guaranteed to be on the active drug as opposed to placebo, the large amount of blood draws, tests and evaluations. Despite this, parents felt that it was important for their children to participate.



*“Adrian has been in the trial for over a year now. ... It is difficult to see my child have so much anxiety over blood draws and a clinic visit, especially when we don’t know if he benefits from the drug. ... Our family chooses to continue in this trial because we have great hope for Adrian and for other patients like him.”
- Amy, Caregiver of a six-year-old patient*

“

The trial has not been easy on Charlotte. She's had to endure a lot of blood draws and testing, and when she's on the drug, she has even more dietary restrictions than normal because there's a whole list of foods that make the drug less effective. ... These hardships are very small compared to how much she struggles in life in school, and I would gladly make these sacrifices to alleviate her struggles and improve her quality of life.”

- TIFFANY, CAREGIVER OF A NINE-YEAR-OLD PATIENT

“We have been participating in the only pediatric drug trial from its inception in 2021. It is hard. The participants in this trial are tugged from sleep to endure long days of testing, too many blood draws to count and absolutely too much stress than a child shouldn't have to take on; especially when they are either too young and/or don't have the cognitive ability to fully understand what is being asked of them.” - Jenny, Caregiver of two patients

In the chat, many galactosemia patients shared their motivation to help others in the future as their reason to participate in clinical studies. Caregivers discussed their difficult decisions to enroll their children in trials, the many challenges they encountered, and some shared the small changes and improvements they observed in their loved ones as a result of trial participation.

Other medications include sleep medications including natural supplements, and weight gain medications or supplements, myo-inositol, essential fatty acids (including fish oil), medications to help with cyclic vomiting syndrome, allergy medications, ADHD medications, OCD medications, probiotics, CBD, osteoporosis medications including Fosamax and Prolia, and biologics (Remicade) for Crohn's disease.

A small percentage selected they or their loved one had not used any medications or medical treatments. Michael's child is still young, but he fears that no effective treatments will be available.

*“My son who has CG is 16 months. At this point many of the medications are not required for him. However, knowing that there is currently no treatment or cure the constant stress of knowing that he will one day most likely need many of these drugs and supplements is very scary. As a parent it's an awful feeling knowing that he will probably get worse and there is no treatment I can provide him to help him get better.”
- Michael, Caregiver of a 16-month-old CG patient (via chat)*

OTHER THERAPIES USED

The poll results are presented in **Appendix 5, Q6** and illustrated with patient and caregiver quotes, below.

Speech therapy, occupational, and physical therapy

Speech therapy can help with apraxia and speech. Many had an early intervention approach, starting when they were only a few months old. The amount of speech therapy can be intensive, and many different approaches were described.

“When I was in school, my speech therapist had an office in the school building so I would see her once or twice during the week. ... And she helped when I was in elementary school and in middle school.” – Erin, Patient, 27 years old

“She had 11 years of speech therapy, which corrected sound substitutions and improved the ability for others to understand her speech.” - Natalie, Caregiver of a 25 and a 22-year-old patient

“In his early academic years, the speech therapy focused on improving Joey's speech so he could be understood by others and able to express himself. ... Now that he is older, “The

language component of speech is where Joey needs the most help now. Speech therapy improves Joey's ability to understand and express thoughts, ideas, and feelings while increasing his ability to problem solve." - Tara, Caregiver of an 11-year-old patient

Many patients also receive ongoing occupational and physical therapy to deal with hand strength, tremors, and coordination.

"They advised us to begin early intervention at three months old to start various types of physical, speech and occupational therapy to attempt to combat the disease progression. ... His occupational therapist noticed hand tremors, making it difficult for him to write in kindergarten. To this day, Holden continues to receive the same weekly therapy."

- Amber, Caregiver of an eight-year-old CG patient

"She also had years of occupational therapy to improve hand strength, finger coordination used for writing, gripping and feeding." - Natalie, Caregiver of a 25 and a 22-year-old patient

"All we can do is make the symptoms less profound by treating them with therapy, which we've done aggressively starting at a very young age. She started physical therapy at 12 months, speech therapy at 18 months, and we enrolled her in a developmental preschool as soon as she was eligible for early intervention. Currently, Charlotte still receives speech therapy, occupational therapy, and special education." - Tiffany, Caregiver of a nine-year-old patient

Speech, occupational and physical therapy downsides include difficulties in finding qualified therapists, long waiting lists for evaluation and to be accepted as a patient, the slow rate of improvement, and the fact that speech therapy only partially solves the problem and does not close the gap between people with galactosemia and their peers.

"Adrian has been in speech therapy and occupational therapy since he was 18 months old. These interventions have helped, but they're a very slow process. Because galactosemia is such a rare disease, most therapists have not even heard of it and don't always know the best methods for kids with galactosemia. This makes it difficult to find the ideal therapist. Currently, we are in a new city and it's already proving to be complicated to find a therapist who are best suited for Adrian's needs." - Amy, Caregiver of a six-year-old patient

"He probably had even more speech and language simulation than my other two children. And although I think that it has helped, obviously it certainly hasn't completely "fixed" his issues. So he struggles with sequencing in just about every possible format."

- Kelley, Caregiver of a 12-year-old patient

"Even when Jake was receiving speech therapy, early intervention wasn't enough for apraxia. We had to find somebody who was skilled in apraxia."

- Kristine, Caregiver of a 15-year-old CG patient

Nutritional/dietetic counseling

Half of polled individuals stated they receive nutritional or dietetic counseling. During the meeting parents described how important it was for their loved ones to recognize what they could eat on their restricted, dairy-free diet.

"At first, the entire family was fed his restricted diet, but we decided that wasn't in Jerry's best interest. We needed to teach him what he could and could not eat when he was out into the world. His mother would not be at his side forever. If he didn't know about a food, our motto became, 'When in doubt, go without.'" - Elaine, Caregiver of a 62-year-old patient

Some patients discussed some of the dietary changes that they had successfully incorporated, including a gluten-free diet.

Additional support for school, including tutoring, as well as assistive technologies

Some reported requiring additional scholastic support including one-on-one educational aids in a regular classroom, extra assistance in specific topics, and tutors. Many receive their speech therapy in the school system.

"When I was in school, I was in a lot of special education and classes to help with my math and my English therapy. Another teacher helped me with the subjects in the classroom, and then sometimes, I'd be pulled out of the main classroom and in a smaller class to just do those subjects and just learn with a smaller class of students." - Erin, Patient, 27 years old

"Joey was in a general education class setting for kindergarten and first grade and had one-on-one aids helping him stay to the course of the general education curriculum. However, we didn't see Joey progress and he fell behind his peers. In second grade, we made the decision to place Joey in a smaller special education setting with other children learning at his pace. This change was very beneficial and we started to see Joey slowly progress. Now, in fifth grade, he receives speech therapy, occupational therapy, and individualized reading services numerous times a week." - Tara, Caregiver of an 11-year-old patient

"Garrett was included in a general education classroom, but was pulled for extra assistance in math, reading, and speech. His speech services were received through the school and also through private speech therapy that we have obtained for him. He was taught skills to assist him in improving his speech and language skills and troubleshooting skills to help others understand what he's trying to say."

- Heather, Caregiver of three CG patients, 24, 21 and 16 years old

Some patients use technology supports ranging from simple visual aids including charts, pictures, and color-coded text, all the way to sophisticated assistive technologies including text-to-speech technologies. Downsides include the disruption of having to leave the regular classroom for special classes, and the extra costs for the tutoring and assistance. In the chat, a caregiver lamented how many different modifications were required in order for those living with galactosemia to function like the rest of the population.

Counseling or psychotherapy

Many galactosemia patients require counseling or psychotherapy to help them adapt to change, manage anxiety, and to help interpret and respond to social cues. Some also described using behavior modification approaches to help their children deal with frustration and anger.

"I've been seeing a therapist and she's been just helping with the daily anxiety. ... We were discussing better ways to handle those changes that you can't be prepared for, and she was trying to give me other things to help me with that because that was hard for me to handle."
– Erin, Patient, 27 years old

"Galactosemia comes with many symptoms and complications, one of them being depression. I do put a great deal of effort into my mental well-being. I practice meditation and I am a big advocate for self-care. I see a therapist and I do what I can to help myself. I do journaling and I have tried Cognitive Behavioral Therapy, which has proved helpful."
– Shelby, Patient, 31 years old

After Garrett started to act out his frustrations when he was in grade school, "We started using behavior modification with him. This would assist him in his frustrations and also making it safe for us to handle these breakdowns. ... I truly believe this therapy has allowed Garrett to become a responsible adult who is accepted in his workplace and by his peers."
– Heather, Caregiver of three CG patients, 24, 21 and 16 years old

Other treatment approaches not mentioned

Other therapy approaches and strategies include specialist referrals, exercise, moving the family, feeding therapy to deal with sensory issues, breathing exercises to help with speech, ankle foot orthosis (AFOs) to help address low muscle tone.

"Alenna has to see many specialists just to make sure her body is functioning, to the best of its ability. A geneticist, specifically for galactosemia, a dietician for nutrition, an endocrinologist to ensure her hormones are where they need to be, an ophthalmologist to keep an eye on cataracts, an orthopedist to ensure bone health, a GI doctor to keep her digestive tract healthy, a reproductive endocrinologist for reproductive health, a nephrologist to make sure her kidneys are okay. A pediatrician for other health related issues, and now lately, diagnostic tests to watch for any bodily changes that are common with galactosemia."
– Cassidie, Caregiver of a 13-year-old patient

"She's only five, but I swear to you, swimming helps her with her speech. The more she exercises, the more it helps with her speech, but that's only by trial and error that we've worked on this." – Gillian, Caregiver of a five-year-old patient

"We made the decision to move from the city to the country, thinking that'd be better environment for him, which it was." – Elaine, Caregiver of a 62-year-old patient

Currently not doing anything to help manage symptoms

A small proportion of patients received no treatment or interventions at all.

"My parents didn't learn a lot when I was younger about galactosemia. They didn't realize that there would be any type of speech problems or any learning problems, things like that. I never had any therapy." – Kimberly, Patient, 47 years old

EFFICACY OF CURRENT TREATMENTS

Poll questions are shown in **Appendix 5, Q7 and Q8** are consistent: current treatment approaches for galactosemia are not very effective and the majority reported that their current regimen controls symptoms "very little", "somewhat", or "not at all".

Not very effective or only treats some and not all of the symptoms

Despite all the therapy that individuals living with galactosemia endure, they still experience significant gaps between themselves and their peers. The caregivers of older patients reported that they observed progressive declines in skills and cognition in their loved ones.

"We started working with speech therapists, physical therapists, and a play therapist. We would see growth in his skills, but never enough to have him at the average level for his age. Since the only treatment for galactosemia is following a restricted diet, Garrett is proof alone that we need more for our kids." – Heather, Caregiver of three CG patients, 24, 21 and 16 years old

"He has to take MiraLAX every day and that is not an effective treatment for us. He still struggles with constipation regularly and that affects so many other aspects of his life. Not just using the bathroom, he has trouble sleeping. So we have tried some natural supplements for that, not super effectively. And before having access to a clinical trial drug, that was all we had. ... I don't really feel like any of them are effective. They just make it a little easier or a little better." – Amber, Caregiver of an eight-year-old CG patient

"We see very little progress from Joey with the treatments that we currently receive. ... It's important to note that even though we're doing these treatments, we don't see progress. In fact, sometimes we see regression. ... Even though they are getting these treatments in special education settings, they're not really that effective." – Tara, Caregiver of an 11-year-old patient

A few individuals did report in the polls that their treatment regimen controlled symptoms "to a great extent". It was hard to find many examples, except for a few caregivers who mentioned that their child seemed to be benefitting from enrollment in a clinical trial.



The reality is that the current treatments we use (occupational and speech therapy, tutoring, vitamin D, MiraLAX, supplements and vitamins) are not working as none of these target the root cause which is the daily damage to her central nervous system that is happening as a result of her body making endogenous galactose despite being on a dairy free diet. These treatments are not “fixing” anything. They are merely trying to manage the symptoms that result due to the neurological damage this condition causes, like putting a band aid on the problem versus targeting the root of the problem.”

- MEGAN, CAREGIVER OF A SEVEN-YEAR-OLD PATIENT

Limited availability or accessibility

Even the most fundamental galactosemia treatments, including speech and occupational therapies, are limited. Many caregivers described how challenging it is finding someone who has training in apraxia. Many patients live far away from the specialized therapists.

*“For us to get assessments here in Canada, it takes us almost two years to even just get one assessment. Then you’re re-training and re-teaching the medical staff, the professionals, about galactosemia, that you’re having to almost start the whole process over again.”
Debby’s grandson fell through the cracks. “Because he was not disruptive like most kids, they never gave him an EA or someone to work with him.” Debby had to fight hard to obtain resources including an educational assistant for her grandson.*

- Debby, Caregiver for a 14-year-old patient

“One of the biggest frustrations is that very, very rarely do any of these therapists have any idea what galactosemia is. We had to take the time to educate them about galactosemia and the specifics of why our kids are having these issues. Not only is it difficult to get any therapists, but specifically a therapist who knows galactosemia is almost unheard of. Very frustrating because they don’t understand the specific needs of that child.”

- Amy, Caregiver of a six-year-old patient

Some caregivers have worked hard to find alternatives and to provide the services that their loved ones need.

“I’m constantly looking up, trying new tools. Like with feeding therapy, we don’t have a feeding therapist that was open, so our occupational therapist kind of helped us and guide us along. ... I have bought swings, I have bought tools, I have bought chewing things. I have a collection, a pile of stuff to trial and error with her because there’s so many gaps.”

- Gillian, Caregiver of a five-year-old patient

“He needs OT, but we don’t have any occupational therapists out here where we live, so I watch YouTube videos and sit there and try to teach him how to have better fine motor skills and finger strength.” - Kayla, Caregiver of a six-year-old patient

A few patients were unable to obtain the assistance that they require.

“I took a test in second grade to see if I qualified for special education, and I passed by one question, and so the school district said, ‘Oh, she doesn’t need any help’. So, I never really got any extra help with any of the speech or the school, and I did have a lot of struggles in school.” - Kimberly, Patient, 47 years old

Requires too much effort and/or time commitment

Many caregivers reported the fatigue and overwhelm associated with the excessive amounts of speech and occupational therapies that galactosemia requires. They also mentioned the impacts on other family members.

“Last summer, I just remember my daughter falling asleep in the therapy room, and I was like, ‘there has to be an end point to this.’ She’s working so hard. We’re working so hard, but there’s no end point. We both had to take a breather from therapy because we weren’t getting anywhere.” - Gillian, Caregiver of a five-year-old patient

“She makes progress, but she continues to fall further behind her peers every year and the neurological damage continues to progress. This has taken a lot of time, coordination, and energy. And although I assume it’s made her deficits less profound, it takes up all of her time after school and it doesn’t do anything to prevent possible tremors or other complications in the future.” - Tiffany, Caregiver of a nine-year-old patient

“Going to all of those appointments takes a huge toll on the family. We have to find someone to bring her there. ... We do have another child that wants to be in sports and other activities, but when you’re running to therapy appointments or tutoring almost every night, there’s very limited time for other activities with the family.” - Megan, Caregiver of a seven-year-old patient

Parents reported feeling stressed from all the management required.

“Managing his healthcare and his educational services is a challenge, from attending IEP meetings, advocating for special equipment and services and navigating medical insurance.” - Natalie, Caregiver of a 25 and a 22-year-old patient

“Coordination of care is super frustrating. I have to call in any changes to each specialist, to make sure one order doesn’t affect the other. There are whole days of traveling for EEG at the hospital and labs.” - Gillian, Caregiver of a five-year-old patient

Side effects

Potential side effects include depressed respiration from rescue medications, mood changes from laxatives, sweating, weight gain and headaches from anxiety/OCD medications.

“They also come with spiraling issues of other side effects from each one of those medicines. ... The MiraLAX medicine can actually cause mood issues for him. So, there’s not really anything that actually works.” - Amber, Caregiver of an eight-year-old CG patient

Medication errors can cause unintended side-effects, which can be potentially deadly for those living with galactosemia.

“Penelope was given lactulose for constipation, which is literally milk sugar. The doctor, the nurse, the pharmacist, the computer system, her medical alert band, and the signs in her room, could not prevent this error.” - Gillian, Caregiver of a five-year-old patient

Other downsides

Other downsides include route of administration and finding physicians and therapists who are familiar with galactosemia. Those who are involved with clinical trials mentioned the downsides of frequent blood draws, lengthy assessments, travel and the chance of potentially being randomized to a placebo arm of a clinical trial.

HOPE FOR FUTURE TREATMENTS, SHORT OF A CURE

Appendix 5, Q9, and illustrated below with patient quotes.

Improvement in learning and memory

An improvement in learning or memory, was the top hope for a future treatment. This would include improved comprehension and addressing neurological deficits.

“If we can help them with their learning and their memory issues and just having a clear brain function, I think would alleviate a lot of the other trickle-down effects.” - Charlotte, Caregiver

“If I could improve something, it would just be his ability to understand. I feel like sometimes he just doesn’t have the ability to understand things.” - Amy, Caregiver of a six-year-old patient

“I hope that over time, she wouldn’t struggle so much in school. And I hope that one day, instead of spending all of her time after school and therapy, that she could participate in something she enjoys, like dancing or theater or sports.” - Tiffany, Caregiver of a nine-year-old patient

Prevention of new disease symptoms

Patients and caregivers expressed their concern about the progressive nature of galactosemia throughout the EL-PFDD meeting.

“I hope that if there’s a treatment available, she won’t go on to develop tremors as she gets older.” - Tiffany, Caregiver of a nine-year-old patient

“We’re hopeful for future treatments to limit the symptoms of the disease, but worry that those treatments will be presented too late in Joey’s life to reverse the neurological damage already done and that will continue to be done.” - Tara, Caregiver of an 11-year-old patient

Improvement in speech

Improvements in speech would contribute to better relationships, employment and independence.

“I wish for a medicine to improve my apraxia and classical galactosemia.” - DJ, Adult patient

“I hope that a treatment would enable her language and communication to improve so that she can sustain deeper, more meaningful relationships with her peers.” - Tiffany, Caregiver of a nine-year-old patient

"I would want Alana to be able to communicate well. I want her to be able to function in society. I want her to be able to have gainful employment."

- Cassidie, Caregiver of a 13-year-old patient

"Just having all of those metabolites out of their system and just having a healthier body in general would help them with their speech and would help them with their language processing, because the communication is such a big key... so that they can develop those relationships with friends and family." - Charlotte, Caregiver

Gainful independence

Many wish for gainful independence, employment and a more normal life.

"My hope, if an improved treatment becomes available, is that Charlotte can grow up and live a normal life. ... Charlotte deserves a shot of normal life, and an approved treatment would give her that shot." - Tiffany, Caregiver of a nine-year-old patient

"Thinking about your children - your goal is to set them on their way in life and make them contributing members of society and to find a good job and something they love and people they love to be around them, and I think any parent's fear is not being there to care for a child who needs cared for. So, living independently would be very, very important." - Scott Saylor

"I want to know that I will be able to live independently when I am older with no other problems." - Jamie, Patient, 26 years old (via chat)

"Our hopes for her in the future, is that she can be empowered to do anything she wants. I want her to have all the opportunities any other child has, with less medical worries and more options for help." - Gillian, Caregiver of a five-year-old patient

"I just feel so very passionately that I want my child to be taken care of when I'm gone, and it would be awesome if he could just simply be able to do that for himself rather than having to rely on other people." - Amy, Caregiver of a six-year-old patient

Other galactosemia treatment goals

Other goals include control of tremors and/or seizures, improvement in fertility, improvement in social skills, improvement in anxiety or depression. A small number of individuals selected improvement in muscle tone and coordination or balance.

"I hope that her ovarian function is saved, and one day she can have children of her own."
- Tiffany, Caregiver of a nine-year-old patient

Parents and patients described other treatment targets including addressing the underlying metabolic disorder, which would likely resolve most of the galactosemia symptoms. They also asked for treatments to increase bone density, better treatments for constipation, and therapies that were neuroprotective.

ADDITIONAL CLINICAL TRIAL AND RESEARCH IDEAS SHARED AT THE GALACTOSEMIA EL-PFDD MEETING

Many people expressed an interest to participate in clinical trials in order to make things better for their community. Some had specific recommendations for clinical trials including using the data from the natural history studies to mitigate or shorten the placebo arm of a trial.

I believe the placebo is such an important part of this trial. However, because we are dealing with children, the length of time that the child is on the placebo should be taken into consideration. The kids are suffering a great deal from this trial and all of them deserve to be on an actual drug in a timely manner. - "Amy, Caregiver of a six-year-old patient

Parents wished to have more research into understanding and enhancing the secondary metabolic pathways involved with galactose metabolism.

"My recommendation to researchers, NIH, and FDA, is to try to characterize those secondary pathways and look for ways to enhance them. As long as the diet is well controlled, it may not take much to reverse the course of this disease." - Brian, Caregiver of a 29-year-old patient

Tiffany Platt is a mechanical engineer who works for Johnson & Johnson Robotics and Digital Solutions. Tiffany lives in the San Francisco Bay Area with her husband Cliff and two daughters, Avery (11) and Charlotte (9). Her daughter Charlotte has classic galactosemia.



“

I'm constantly looking up, trying new tools. Like with feeding therapy, we don't have a feeding therapist that was open, so our occupational therapist kind of helped us and guide us along. ... I have bought swings, I have bought tools, I have bought chewing things. I have a collection, a pile of stuff to trial and error with her because there's so many gaps.”

- GILLIAN, CAREGIVER FOR HER FIVE-YEAR-OLD DAUGHTER WITH GALACTOSEMIA

Incorporating Patient Input into a Benefit-Risk Assessment Framework

The FDA's Benefit-Risk Assessment Framework 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 describes the challenges of having a lifelong disease burden that patients living with galactosemia endure. It serves as the proposed introductory framework for the Analysis of Condition and Current Treatment Option that could 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 galactosemia. The data resulting from this meeting may help inform the development of galactosemia-specific clinically meaningful endpoints for current and future clinical trials, as well as encourage additional researchers and industry to investigate options for treatments.

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

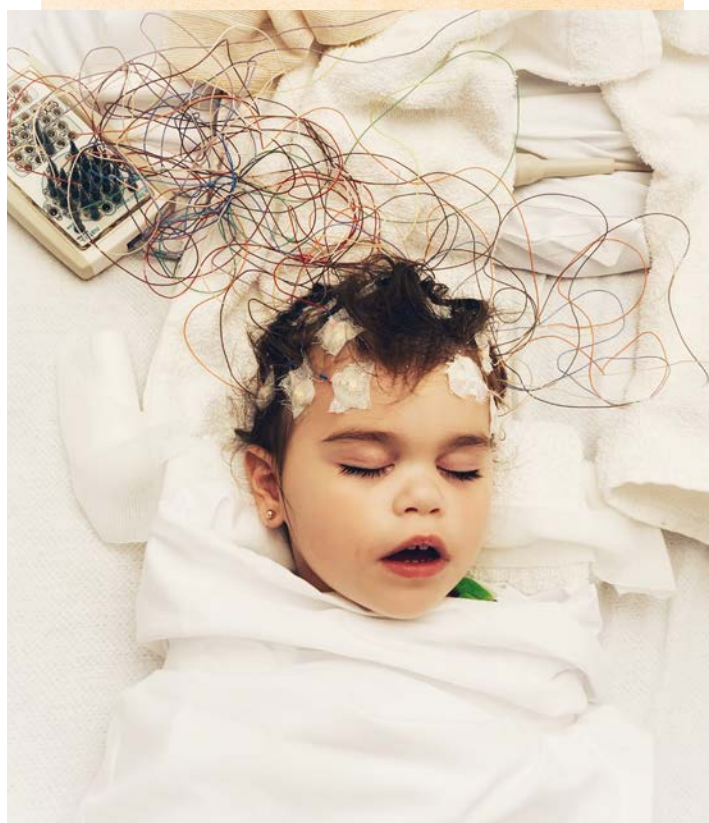


TABLE 1: Galactosemia Benefit-Risk Table

	EVIDENCE AND UNCERTAINTIES	CONCLUSIONS AND REASONS
ANALYSIS OF CONDITION/ IMPACTS ON ACTIVITIES OF DAILY LIVING	<p>Newborn screening is critically important yet even with early intervention, galactosemia-related neurological damage may develop. The first few days of life may be traumatic as patients experience metabolic crisis, struggling with liver failure, lethargy and failure to thrive before diagnosis is confirmed.</p> <p>Individuals living with galactosemia experience a variety of symptoms and have variable outcomes. Neurological complications include learning disabilities, cognitive delays, apraxia, speech impairments, and memory problems. Pre-ovarian insufficiency in female patients, anxiety and depression, tremors and seizures as well as many other health effects impact life on a daily basis.</p>	<p>The impacts and challenges of galactosemia persist for a lifetime. Communication challenges impact relationships and the ability to socialize. Individuals living with galactosemia experience many challenges with education, employment and sports and they have to work much harder than others at school, to obtain a job and to maintain employment. Some are unable to live independently. The social and financial impacts on families are enormous.</p> <p>Patients and families worry about disease progression and the emergence of new symptoms including seizures. Individuals living with galactosemia worry that their symptoms will get worse and that they will be unable to live independently in the future.</p>
CURRENT TREATMENT OPTIONS/ PROSPECTS FOR FUTURE TREATMENTS	<p>The leading galactosemia treatment strategy is to eliminating galactose from the diet, which does not prevent long-term challenges. Endogenous galactose production continues despite a galactose-free diet.</p> <p>Families try many types of treatments and therapies to help their children learn, to thrive socially, and to have their best chance at a full life. Treatments are mostly targeted towards symptom management. Many take vitamin D and calcium, along with probiotics and other medications. Many engage with speech, feeding, occupational and motor therapies, however despite their persistent hard work, individuals with galactosemia still fall behind. Some treatments come with side effects, therapy can be incredibly costly, and many individuals and their families feel exhausted and overwhelmed.</p>	<p>Galactosemia has tremendous unmet treatment needs. There are no FDA-approved treatments to address the underlying cause of galactosemia. Throughout life, neurological damage continues to accumulate, seizures are uncontrolled, and muscle strength deteriorates.</p> <p>Families are desperate for more effective treatments to prevent or minimize the long-term complications of galactosemia. Research and development in this area is desperately needed. Many would like to participate in clinical trials to ensure better outcomes for themselves and for others.</p>

A more detailed narrative is included throughout the VOP report.

Appendix 1: Galactosemia EL-PFDD Development Meeting Agenda

September 1, 2022

11:00am	Opening Remarks <i>Peter Saltonstall, President & CEO, NORD</i> <i>Nicole Casale, President, Galactosemia Foundation</i>
11:10am	Clinical Overview of Galactosemia <i>Judith Fridovich-Keil, PhD, Professor, Department of Human Genetics, Emory University School of Medicine</i>
11:30am	Welcome Remarks <i>Sheila Farrell, MD, Reviewer, Division of Rare Disease and Medical Genetics, U.S. Food and Drug Administration (FDA)</i>
11:40am	Discussion Format and Meeting Overview <i>Debbie Drell, Director of Membership, NORD</i> <i>Scott Saylor, Board Member, Galactosemia Foundation</i>
11:45am	Demographic Polling Questions
11:55am	Topic 1: Living with Galactosemia—Burdens and Symptoms
12:25pm	Polling Questions and Facilitated Audience Discussion on Topic 1 <i>Debbie Drell, Director of Membership, NORD</i>
1:25pm	Break
1:45pm	Topic 2: Current & Future Treatments
2:15pm	Polling Questions and Facilitate Audience Discussion on Topic 2 <i>Debbie Drell, Director of Membership, NORD</i> <i>Scott Saylor, Board Member, Galactosemia Foundation</i>
3:15pm	Summary Remarks <i>Brittany Cudzilo, Vice President, Galactosemia Foundation</i>
2:25pm	Wrap Up and Next Steps <i>Scott Saylor, Board Member, Galactosemia Foundation</i>
3:30pm	Adjourn

Appendix 2: Speaker Biographies and a List of Meeting Panelists

Speaker Bios

Presented in order of meeting appearance.



Peter Saltonstall, President and CEO, NORD

Peter joined NORD in 2008 after having served for more than 30 years as a senior executive in both for-profit and not-for-profit health care environments. Under his leadership, NORD has maintained the integrity of the Orphan Drug Act while forging new relationships between the patient community and the executive branch, Congress, HHS, FDA, NIH, Social Security Administration and CMS, as well as with drug and device companies and with the medical, academic and investment communities. His efforts to build collaborations stem from his view that advances for the rare disease patient can be achieved best through joint efforts. Today he continues to be one of the country's leading voices on rare

disease issues to industry, FDA, Congress and the federal government. Peter is also committed to globalization of the rare disease patient community, as diseases do not recognize geographical boundaries and research can be expedited when patients from many countries are involved. He has helped established collaborative programs with patient communities throughout Europe, Australia, Japan, Asia and South America. Under Peter's leadership, NORD has grown to be the global reference site for the rare disease community, with NORD's website now receiving more than one million requests a month for information. He has also overseen the expansion of NORD's US-based Patient Assistance Network, which works with manufacturers and patients to provide assistance to patients in need of medications they cannot afford. He has also played a major role in building the NORD Longitudinal Natural History System, which is recognized by the FDA as one of the tools of choice for Patient Organizations collecting data on their disease.



Nicole Casale, President, Galactosemia Foundation

Nicole joined the board of the Galactosemia Foundation in 2014 and now serves as the President of the organization. She and her husband Nick live in Latham, NY, and have three children, Noah, Joseph, and Reagan. Joseph was born in March of 2012 and was diagnosed with classic Galactosemia at five days old. Like many he had a rough start, becoming septic and contracting meningitis at nine days old which resulted in unilateral deafness. He currently receives speech, occupational therapy and special education services while enjoying both taekwondo and gymnastics for fun! Nicole's other two children do not have Galactosemia, they may or may not be carriers.

Nicole is a financial coach and homeownership counselor at Troy Rehabilitation and Improvement Program. She received her Bachelor's degree in Elementary Education from University at Plattsburgh, and her Masters of Science in Special Education from the University at Albany. Her hobbies include running, biking and swimming, as well as fundraising.



Judith (Judy) Fridovich-Keil, PhD

Dr. Fridovich-Keil is a Professor of Human Genetics at Emory University School of Medicine, in Atlanta GA. Judy was born and raised in Durham NC. She completed her undergraduate training at Princeton University where she majored in biochemistry. From there, she moved to Cambridge, MA where she completed a PhD in biology at MIT, followed by postdoctoral and ABMG fellowship training at Harvard Medical School. Galactosemia has been the focus of her research for more than 31 years. Nicole Casale, President, Galactosemia Foundation



Sheila Farrell, MD

Sheila Farrell M.D., M.P.H., is a medical officer in the Division of Rare Diseases and Medical Genetics (DRDMG) in the Office of New Drugs (OND) at the Food and Drug Administration's (FDA's) Center for Drug Evaluation Research (CDER). Prior to joining the FDA, Dr. Farrell held several academic appointments; most recently she was an associate professor of pediatrics at the Louisiana State University Health Sciences Center (LSUHSC) in Shreveport. She received her M.D. from Georgetown University School of Medicine and completed a residency in pediatrics at Children's Hospital National Medical Center in Washington, DC. After residency she received her M.P.H. from the Johns Hopkins

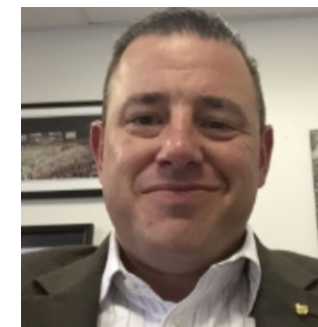
Bloomberg School of Public Health and completed a pharmacoepidemiology fellowship at the FDA through the Epidemiology Training Program.



Debbie Drell, Co-Moderator

Debbie serves as the Senior Director of Membership at NORD. In this role, she oversees NORD's membership programs, which support the collective and individual needs of rare disease patient organizations, patients and advocates through education, research, advocacy and mentorship. She brings to the organization over 22 years of leadership in nonprofit public health education, awareness and advocacy. Prior to joining NORD, Debbie spent 13 years with the Pulmonary Hypertension Association, a NORD member organization. During that time, she led the growth of the organization's network of support groups from 80 to nearly 300, developed new services personalized to the diversity

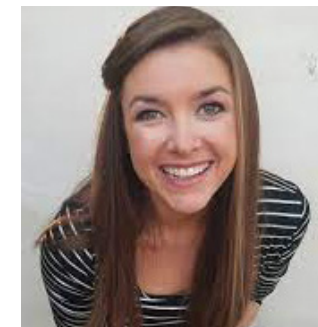
of patients and caregivers, and convened the largest gathering of pulmonary hypertension patients in history. Debbie has represented the patient perspective on several national platforms, including as a guest on National Public Radio's Kojo Nnamdi Show. She has served as a member of the board of trustees of the American Thoracic Society, a 115-year-old medical society with a global membership of 16,000 pulmonologists, critical care and sleep disorder researchers, clinicians and other medical professionals. An accomplished public speaker, she has presented extensively at colleges and universities on women's health issues, delivered speeches on caregiving across the country, including at Johns Hopkins University events, and moderated panels at the World Orphan Drug Congress European and American meetings. A graduate of the University of California, Irvine, and the University of Kent, Debbie's dedication to the rare disease community is rooted in a deeply personal connection. She was inspired to enter the field after her older sister, Alex, was diagnosed with pulmonary hypertension.



Scott Saylor, Co-Moderator

Scott joined the Galactosemia Foundation Board in 2010 and has held many roles and positions, he is currently the treasurer. Scott is also Director of Pharmacy and Retail for Walgreens Drug Stores overseeing Southern Virginia. Scott has been an advocate for those living with Galactosemia, wanting to improve treatments and resources.

Scott resides in Chesterfield, Virginia with his wife Kristine, daughter Brooke and son Jake. Jake was born in 2007 with classical Galactosemia. Soon after Jake was born the Saylor's started "Fore the Cause" and have raised more than \$300,000 for Galactosemia Foundation. Scott and Kristine devote a lot of time to Brooke and Jake's sports interests. Brooke is captain of her high school volleyball team and plays travel. Jake is a freshman competing in cross country and track. Besides rooting for his kids, Scott enjoys physical fitness including running and playing various sports.



Brittany Cudzilo, Vice President of the Galactosemia Foundation

Brittany currently serves as the Vice President of the Galactosemia Foundation. Brittany and her husband Ben live in Knoxville, TN with their four daughters, two of whom have Classic Galactosemia. They have been making connections with those around the area since their daughter Ansell was born. They are committed to reading the latest research and have a desire to understand Galactosemia to their greatest ability.

Brittany's background is in secondary education, and she taught high school for 5 years. Since leaving, she has been at home raising their girls and is now working to build the GLOW fundraiser to connect Galactosemia families and raise awareness and support for the Galactosemia Foundation.

Patient and Caregiver Speaker Bios

Presented in order of meeting appearance.



Gillian Sapia

Gillian Sapia is a registered nurse and a rare disease patient advocate. She works with the Galactosemia Foundation, Haystack Project, and Everylife Foundation to advocate our needs to Senate and Congress. She is currently working on the Heart Act and accelerated approval revisions.



Elaine Olson

Elaine Olson is the mother of Jerry Olson age 62, who was diagnosed at a few weeks of age with galactosemia, therefore making us pioneers of Galactosemia. Elaine birthed six children, two passed away from undiagnosed galactosemia in infancy. She has lived in central Minnesota all her life as part of a large, loving family. She and her husband, Melvin, have been married for 67 years and raised five children and fostered many others. She loves to travel and spend time with family and friends.



Cassidie Archuleta

Cassidie Archuleta began her advocacy work in 2009 when her first and only child, Alenna, was diagnosed with classic galactosemia. From advocating for her daughter in medical and school settings, being a resource to other parents, and speaking with lawmakers across the U.S., she has gained a passion for improving the lives of people with galactosemia. Cassidie and Alenna live in Salt Lake City, Utah where they enjoy spending time with friends and family, exploring the many trails around their house, and are always up for new adventures.



Natalie Whittington

Natalie Whittington is a Music Educator and Owner of Sweet Music Studio. She has 25 years of experience teaching, mentoring and advocating for students of all ages and abilities. Natalie currently resides in Tampa, Florida with her husband and two children. She enjoys traveling, cooking, and gardening.



Maureen Bell

Maureen (Lindenhofen) Bell was diagnosed with classic galactosemia (Q188R mutation) seven days after her birth in 1972. Maureen has experienced many of the challenges those living with galactosemia have. Maureen recently turned 50 years old and resides in Chalfont, Pa with her husband of over 20 years, Bill. Maureen is an advocate for those living with galactosemia and a helpful resource for new parents with their newly diagnosed children.



Amber Jones

Amber Jones is a teacher living in Oklahoma with her husband and two children. Holden, her eight-year-old son, is diagnosed with classic galactosemia.



Heather Scharff

Heather Scharff is a mother of three children with classic galactosemia. She lives in Ottumwa, IA with her husband, children and the family dog Gable. Heather loves spending time outdoors, crafting and hot air ballooning with her father.



Tiffany Platt

Tiffany Platt is a mechanical engineer who works for Johnson & Johnson Robotics and Digital Solutions. She also manufactures and sells a children's product called Shampoo Buddy. She was born and raised in Chicago and now lives in the San Francisco Bay Area with her husband Cliff and two daughters, Avery (11) and Charlotte (9). Her daughter Charlotte has classic galactosemia.



Tara Tanella

Tara Tanella is a wife to her husband Joe and a mom of three children, 11-year-old Joey, eight-year-old Victoria and one-year-old Adriana. She lives in New York with her husband, children and dog (Rudy). As a professional, Tara works at Morgan Stanley and as a Rare Disease Caregiver, she is the biggest support and advocate to Joey, who was diagnosed at three days old via newborn screening, with the rare disease, galactosemia.



Amy Morrow

Amy Morrow currently lives in Clarksville, Tennessee. She is the mother of five, with one child who suffers from classic galactosemia. She is a former English and Public Speaking teacher.

List of Panelists and Meeting Discussants

Topic 1: Living with Galactosemia—Burdens and Symptoms

Panel

- Gillian, Caregiver of a five-year-old patient
- Elaine, Caregiver of a 62-year-old patient
- Jerry, Patient, 62 years old
- Cassidie, Caregiver of a 13-year-old patient
- Natalie, Caregiver of a 25 and a 22-year-old patient
- Maureen, Patient, 50 years old

Topic 2: Current & Future Treatments Panel

- Amber, Caregiver of an eight-year-old CG patient
- Tiffany, Caregiver of a nine-year-old patient
- Heather, Caregiver of three CG patients, 24, 21 and 16 years old
- Tara, Caregiver of an 11-year-old patient
- Amy, Caregiver of a six-year-old patient

Appendix 3: Demographic, Polling and Discussion Questions

Demographic Questions

1. Are you:

- A. Someone living with Classic Galactosemia
- B. Someone living with Clinical Variant Galactosemia
- C. Someone living with Duarte Galactosemia
- D. A caregiver of someone with Classic Galactosemia
- E. A caregiver of someone with Clinical Variant Galactosemia
- F. A caregiver of someone with Duarte Galactosemia

- E. 31-50 years of age
- F. 51-70 years of age
- G. 71 years of age or older

2. Where do you currently reside?

- A. US Pacific time zone
- B. US Mountain time zone
- C. US Central time zone
- D. US Eastern time zone
- E. US Alaska time zone
- F. US Hawaii time zone
- G. Europe
- H. Middle East
- I. Asia
- J. Canada
- K. Mexico
- L. Other

3. Are you or your loved one with Galactosemia:

- A. Female
- B. Male
- C. Prefer not to identify
- D. Other

4. How old are you (the patient) or your loved one living with Galactosemia?

- A. 0-5 years of age
- B. 6-10 years of age
- C. 11-18 years of age
- D. 19-30 years of age

5. Do you or your loved one have a genetic diagnosis?

- A. Yes, I/they have diagnosed Classic Galactosemia via genetic testing
- B. No, I/they do not have a diagnosis from genetic testing (but have received the diagnosis from newborn screening)
- C. Unsure

Polling Questions

Topic 1 - LIVING WITH GALACTOSEMIA: SYMPTOMS AND DAILY IMPACT

1. Which of the following GALACTOSEMIA health concerns have you or your loved one ever had? Select ALL that apply

- A. Failure to thrive, lethargy (infants)
- B. Liver failure, Jaundice, or Sepsis
- C. Cataracts
- D. Low muscle tone and motor skill impairment
- E. Pre-ovarian Insufficiency
- F. Low bone density
- G. Trouble eating or Sensory Processing Disorder
- H. Anxiety and Depression
- I. Learning Disability, Cognitive delay and/or Speech impairment
- J. Impaired memory
- K. Tremors and/or Seizures
- L. Other

2. Select the TOP 3 most troublesome GALACTOSEMIA-related health concerns that you or your loved one have ever had. Select up to 3

- A. Failure to thrive, lethargy (infants)
- B. Liver failure, Jaundice, or sepsis
- C. Cataracts
- D. Low muscle tone and motor skill impairment
- E. Pre-ovarian Insufficiency
- F. Low bone density
- G. Trouble eating & Sensory Processing Disorder
- H. Anxiety and Depression
- I. Learning Disability and/or Cognitive delay and/or Speech impairment
- J. Impaired memory
- K. Tremors and/or Seizures
- L. Other

3. What specific activities of daily life that are important to you (or your loved one) that you/they are NOT able to do or struggle with due to GALACTOSEMIA? Select TOP 3

- A. Biking or playing sports
- B. Communicating
- C. Driving
- D. Eating independently
- E. Living independently
- F. Working/Maintaining a job
- G. Sleeping
- H. Having meaningful friendships
- I. Attending social events at someone's home or public
- J. Being intimate with a partner
- K. Other

4. What worries you most about you or your loved one's condition in the future? Select TOP 3

- A. That my/their symptoms will get worse
- B. Inability to live independently
- C. Inability to communicate needs
- D. Inability to find galactosemia-safe food
- E. Inability to finish education
- F. Inability to work or pursue a career

- G. Inability to have children
- H. That my/their relationships will suffer
- I. Other

Topic 2 - PERSPECTIVE ON CURRENT AND FUTURE APPROACHES TO TREATMENT

1. What medications or medical treatments have you or your loved one used (currently or previously) to treat symptoms associated with GALACTOSEMIA? Select ALL that apply

- A. Anti-seizure medications
- B. Hormone replacement medication
- C. Weight gain medication or supplements
- D. Vitamin D supplements
- E. Calcium supplements
- F. Sleep medications
- G. MiraLAX or another laxative
- H. Other medications or supplements
- I. I have not used medications or medical treatments recently
- J. Other

2. Besides medications and treatments (currently or previously), what have you or your loved one used to help manage the symptoms of GALACTOSEMIA? Select ALL that apply

- A. Physical therapy
- B. Occupational therapy
- C. Speech therapy
- D. Counseling or psychotherapy
- E. Nutritional/Dietetic counseling
- F. Additional support for school (such as tutoring)
- G. Other
- H. We are currently not doing anything to help manage symptoms

3. How well does your current treatment regimen treat the most significant symptoms of GALACTOSEMIA?

- A. Not at all
- B. Very little
- C. Somewhat
- D. To a great extent
- E. Not applicable because they not using anything

4. What are the biggest drawbacks of your or your loved one's current approaches? Select up to 3

- A. Not very effective at treating target symptom
- B. Only treats some not all symptom(s)
- C. High cost or co-pay, not covered by insurance
- D. Limited availability or accessibility
- E. Side effects
- F. Route of administration
- G. Requires too much effort and/or time commitment
- H. Other
- I. Not applicable as I am not using any treatments

5. Short of a complete cure, what TOP 3 specific things would you look for in an ideal treatment for GALACTOSEMIA? Select up TOP 3

- A. Control of tremors and/or seizure
- B. Improvement in speech
- C. Improvement in learning or memory
- D. Improvement in fertility
- E. Improvement in muscle tone and coordination/balance
- F. Improvement in anxiety or depression
- G. Improvement in social skills
- H. Gainful independence
- I. Prevention of new symptoms of disease
- J. Other

Meeting Discussion Questions

Topic 1 - LIVING WITH GALACTOSEMIA: SYMPTOMS AND DAILY IMPACT

1. Of all the symptoms and health effects of GALACTOSEMIA, which 1-3 symptoms have the most significant impact on you or your loved one's life? Which symptoms most affect you or your loved one now?

- A. Which symptoms were the most significant at other times in your or your loved one's life?
- B. What were the first symptoms that you detected (after being put on a Galactosemia-safe diet, such as developmental delays, speech delays, etc...)

2. How does GALACTOSEMIA affect you or your loved one on best and on worst days? Describe your best days and your 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 GALACTOSEMIA?

- A. How does GALACTOSEMIA affect you or your loved one? What are the challenges you face?
- B. How does GALACTOSEMIA affect life activities (school/work, learning abilities, self-sufficiency, living situation, activities, etc.)?
- C. If you or your loved one could do one activity that you are currently unable to, what would it be?

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?

- A. What capabilities are you most concerned about you or your loved one potentially being unable to do while growing older?
- B. What frustrates you or your loved one most about their condition?

Topic 2 - PERSPECTIVE ON CURRENT AND FUTURE APPROACHES TO TREATMENT

3. What are you currently doing to manage your or your loved one's GALACTOSEMIA symptoms?

- A. Which specific GALACTOSEMIA symptoms do the treatments address?
- B. How has this treatment regime changed over time and why?

2. How well do these treatments treat the most significant symptoms and health effects of GALACTOSEMIA?

- A. How well do these treatments improve the ability to do specific activities that are important in daily life?

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 GALACTOSEMIA? What factors would be important in deciding whether to use a new treatment?



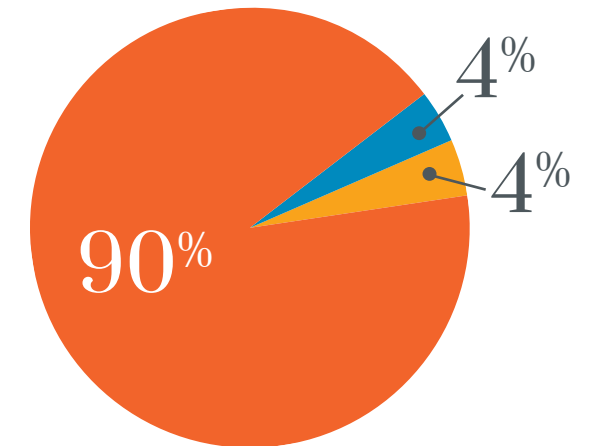
Maureen Bell was diagnosed with classic galactosemia (Q188R mutation) seven days after her birth in 1972 and has experienced many of the challenges those living with galactosemia have. Maureen recently turned 50 years old and is an advocate for those living with galactosemia as well as a helpful resource for parents with their newly diagnosed children.

Appendix 4: Meeting Demographics

The graphs below include all attendees who chose to participate in online voting. While the response rates for these polling questions is not considered scientific data, it provides a snapshot of those who participated in the Galactosemia EL-PFDD meeting and is intended to complement the patient comments made during and after the meeting. "N" indicates the number of individuals who voted in each of the online poll questions.

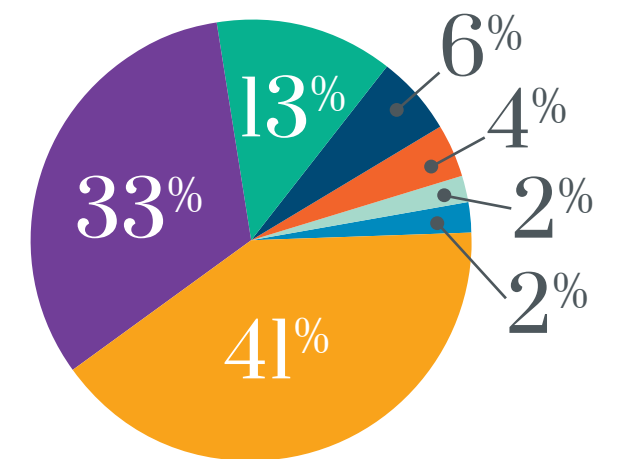
1. Are you:

- A caregiver of someone with Classic Galactosemia
- A caregiver of someone with Duarte Galactosemia
- Someone living with Classic Galactosemia



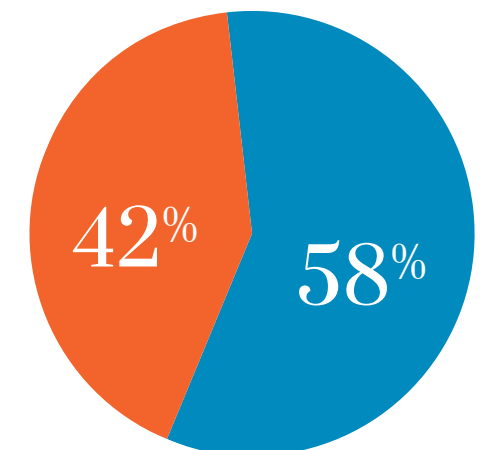
2. Where do you currently reside?

- US Eastern time zone
- US Pacific time zone
- US Mountain time zone
- Mexico
- US Central time zone
- Canada
- Other

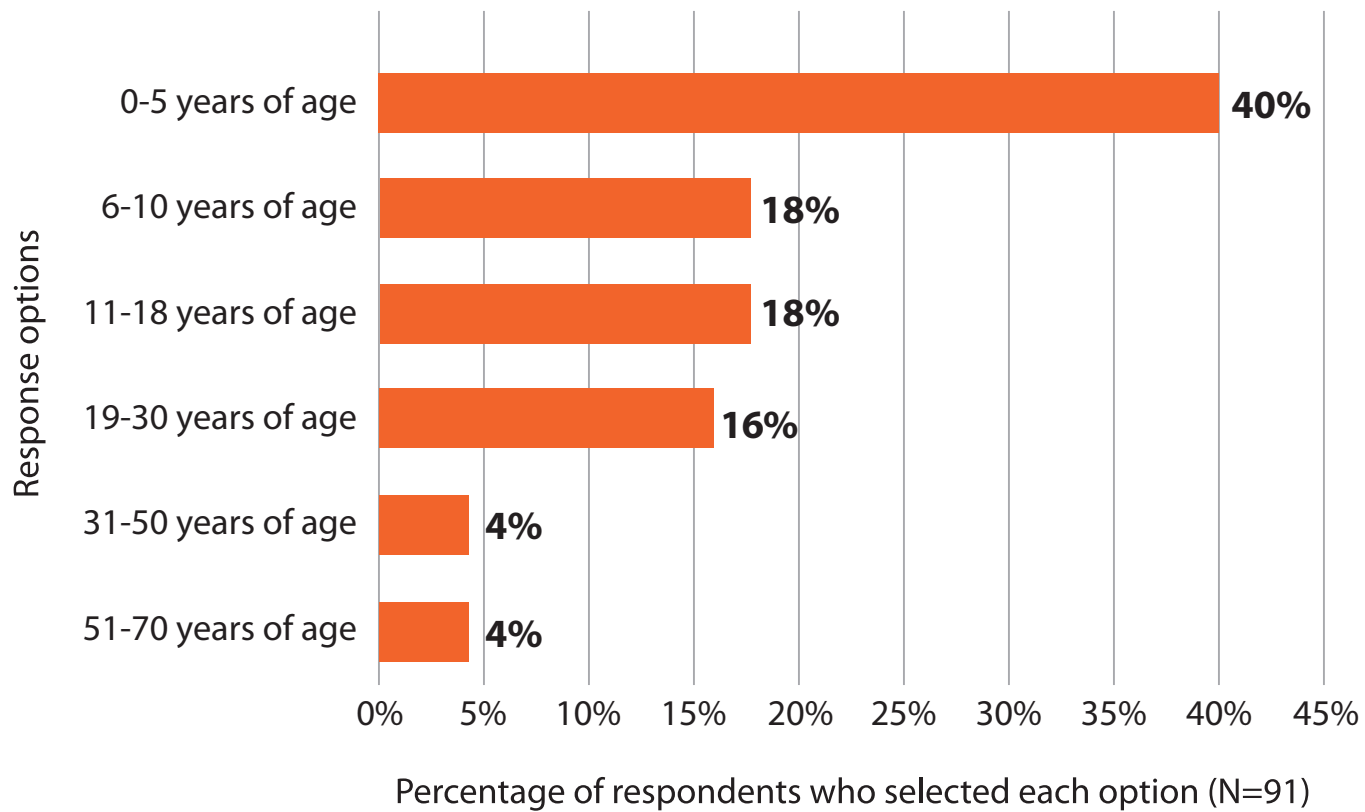


3. Are you or your loved one with Galactosemia?

- Male
- Female

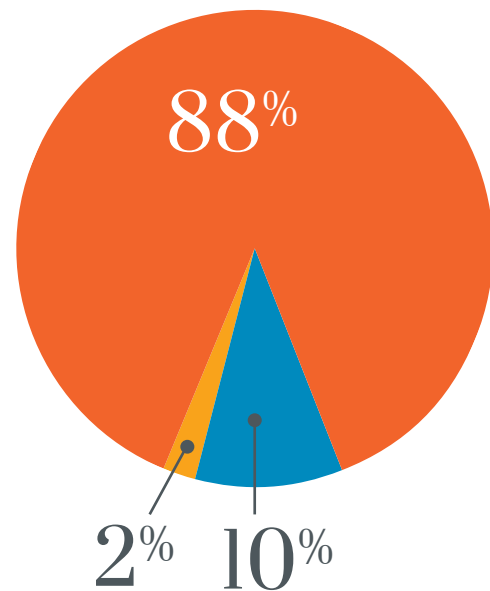


4. How old are you, (the patient) or your loved one living with Galactosemia now?



5. Do you or your loved one have a genetic diagnosis?

- I/they have diagnosed Classic Galactosemia via genetic testing
- I/they do not have a diagnosis from genetic testing (but have received the diagnosis from newborn screening)
- Unsure



Appendix 5: Meeting Poll Results

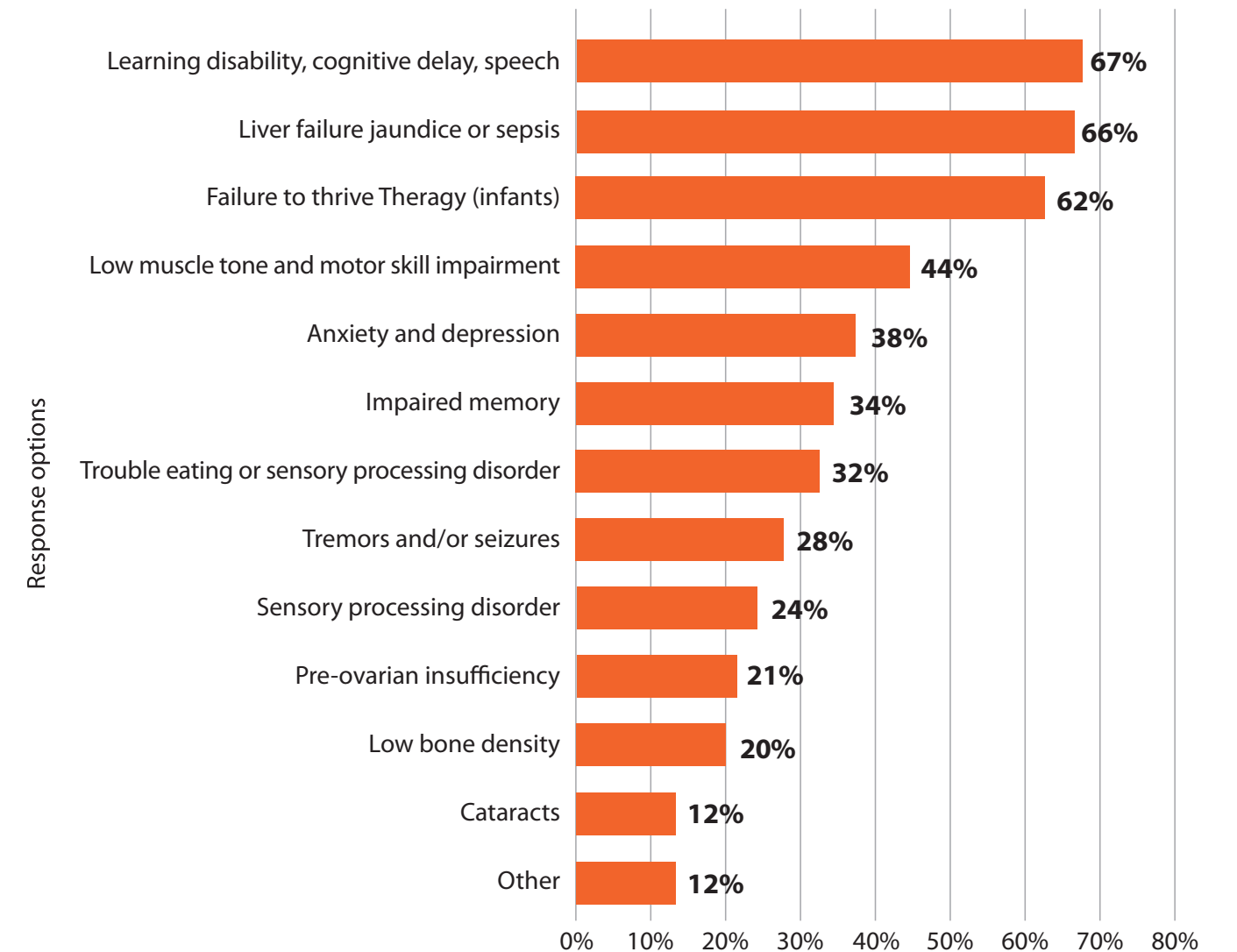
The graphs below include all patients as well as parents and caregivers who chose to participate in online polling during the EL-PFDD meeting. The number of affected individuals and caregivers who responded to each polling question is shown below the X axis (N=x).

The responses for these polling questions are not considered scientific data, but are provided to present a snapshot of those who participated in the Galactosemia EL-PFDD meeting. This is intended to complement the patient comments made during the meeting.

Note that the results of the meeting poll and the post-meeting survey were similar, so only the meeting poll results are shown.

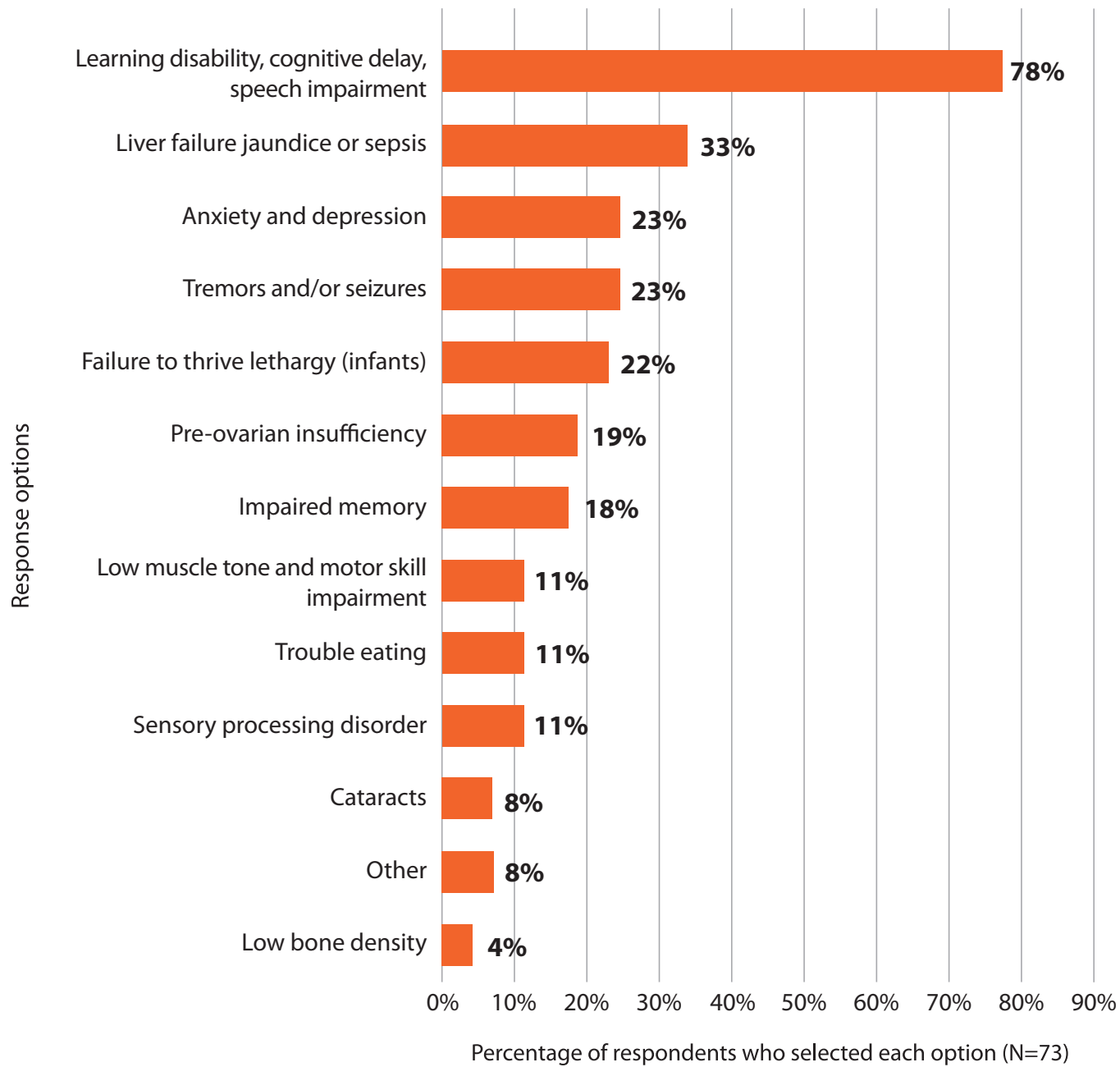
Results of Topic 1: Living with Galactosemia—Burdens and Symptom

1. Which of the following Galactosemia health concerns have you or your loved one ever had? Select all that apply

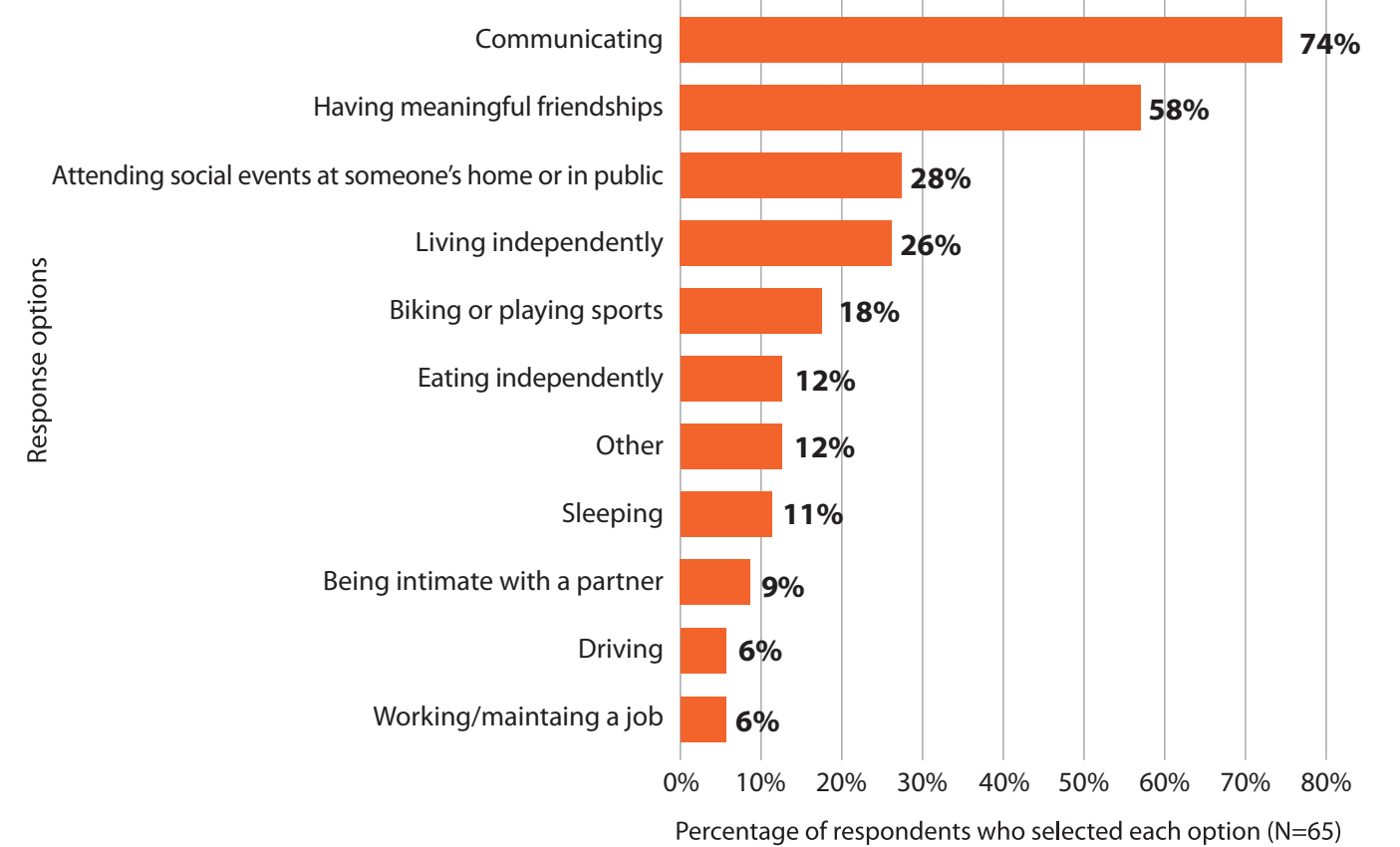


Percentage of respondents who selected each option (N=82) Each respondent selected an average of 4.6 responses

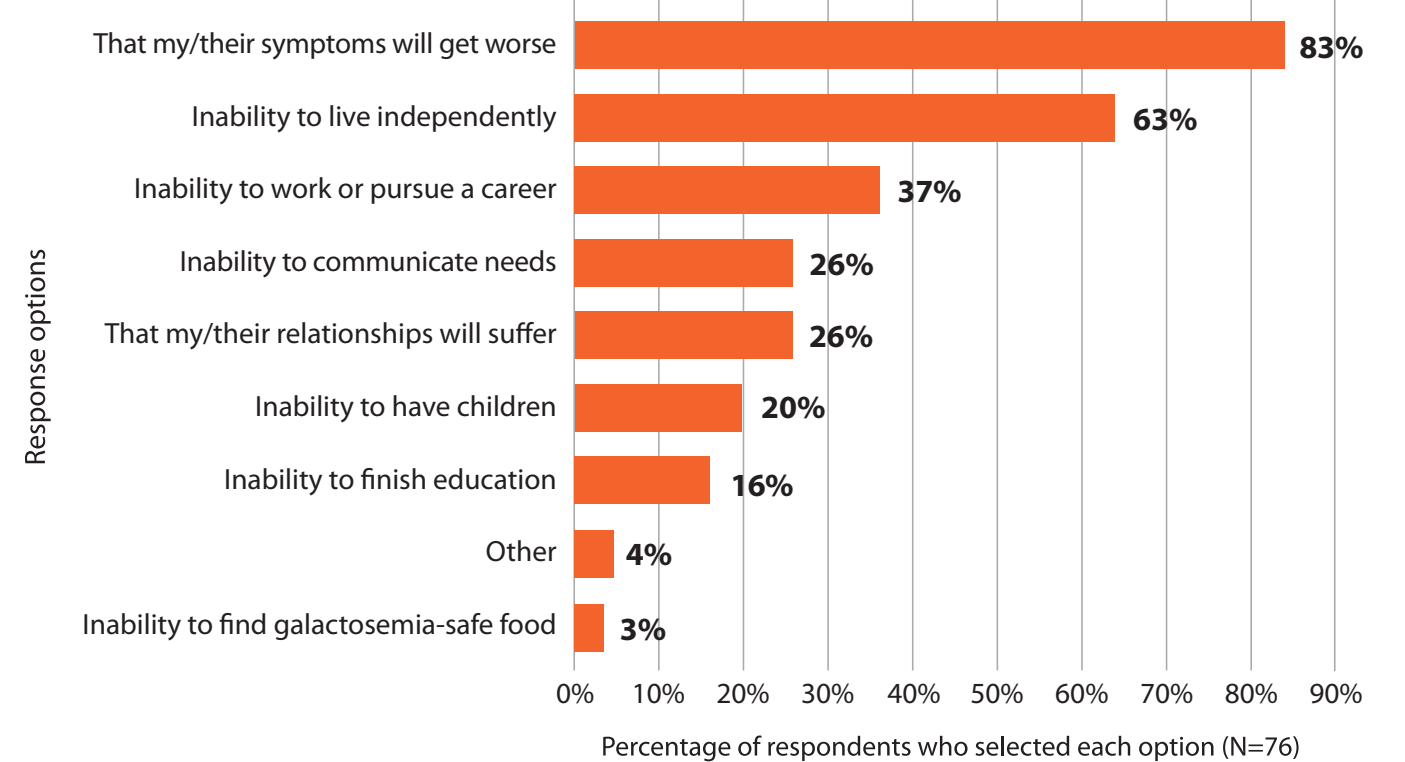
2. Select the TOP 3 most troublesome Galactosemia related health concerns that you or your loved one have ever had.



3. What specific activities of daily life that are important to you (or your loved one) are you/they NOT able to do or struggle with due to Galactosemia? Select top 3.

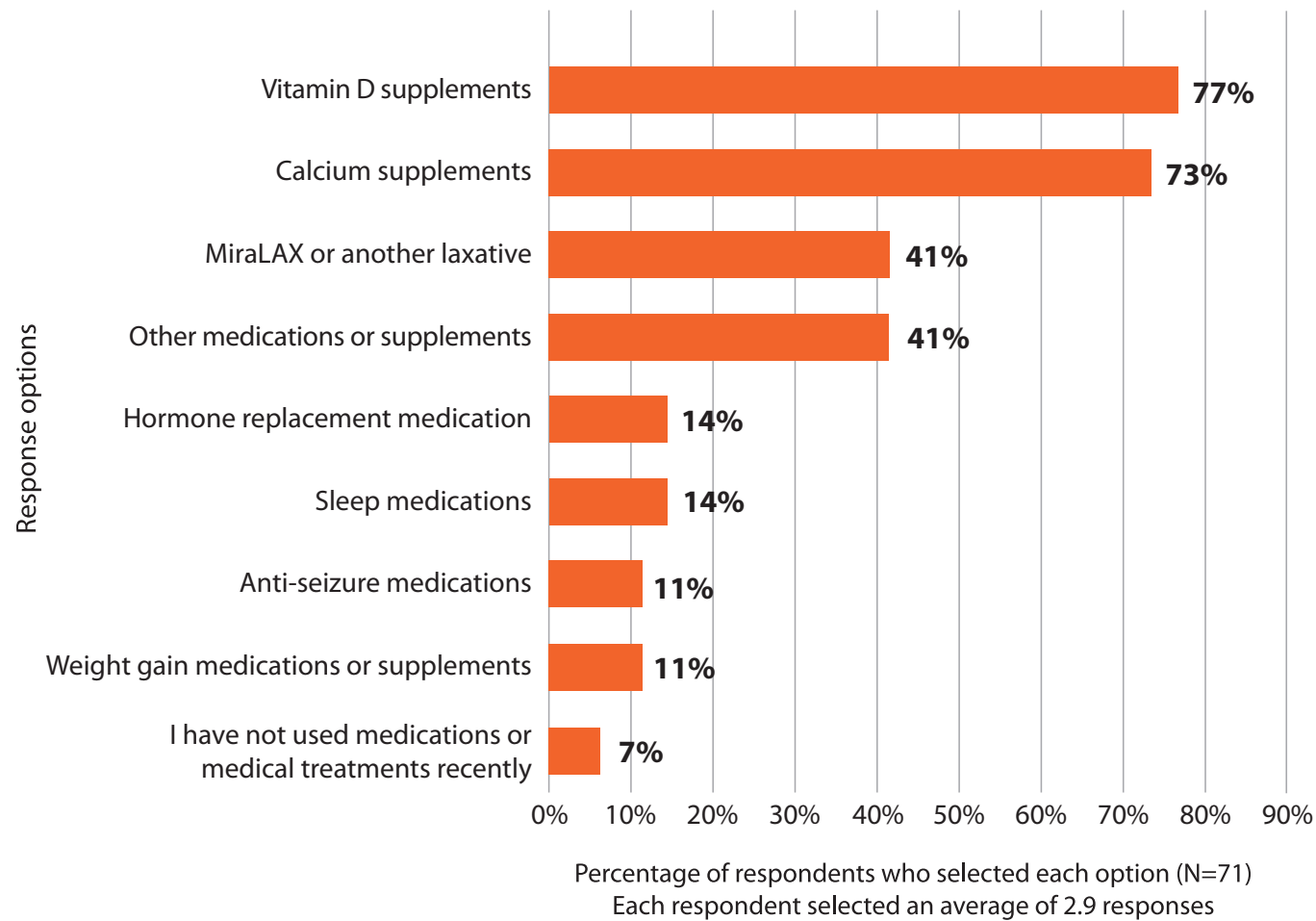


4. What worries you most about you or your loved one's condition in the future? Select top 3.

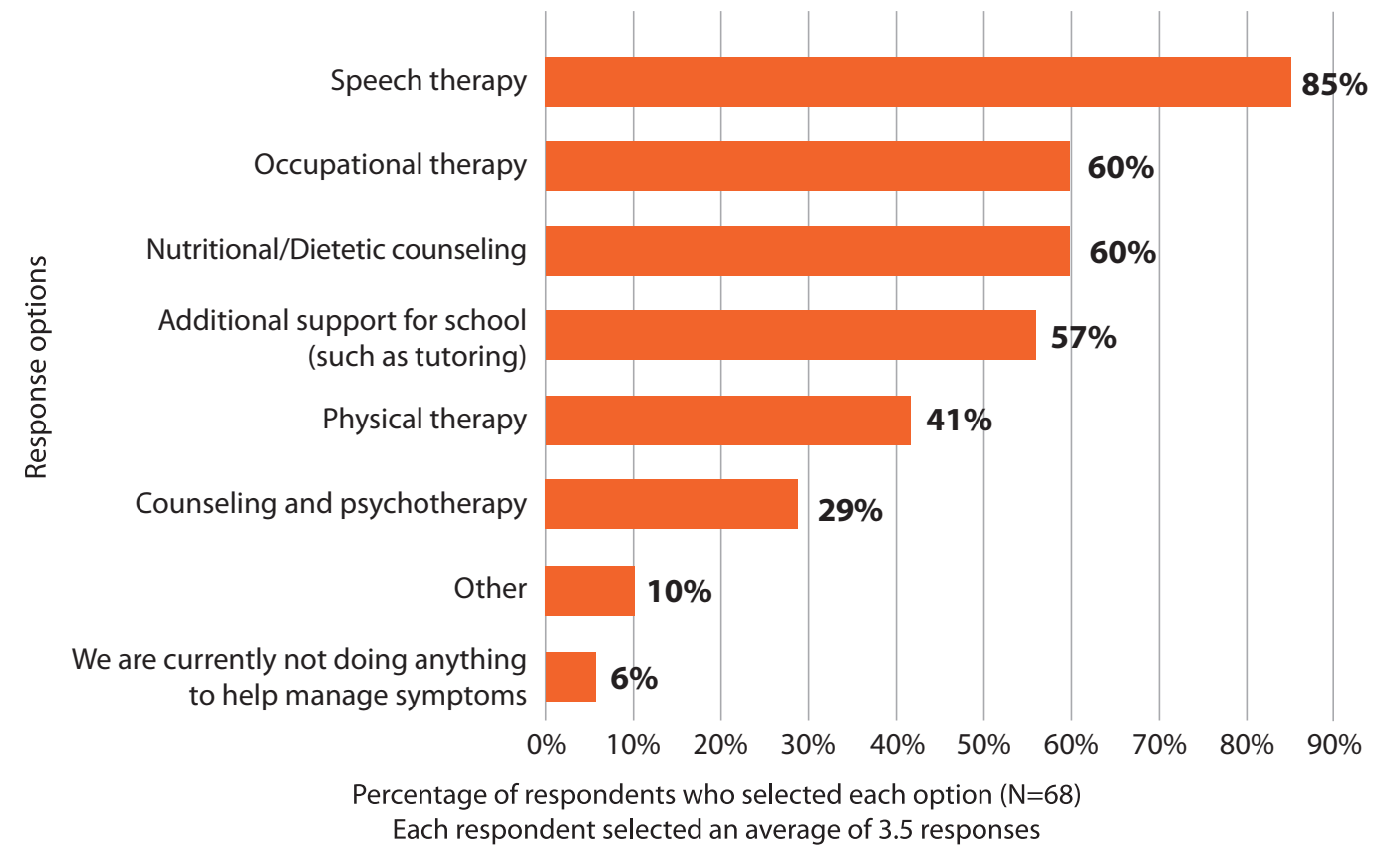


Results of Topic 2: Perspective on Current and Future Treatments

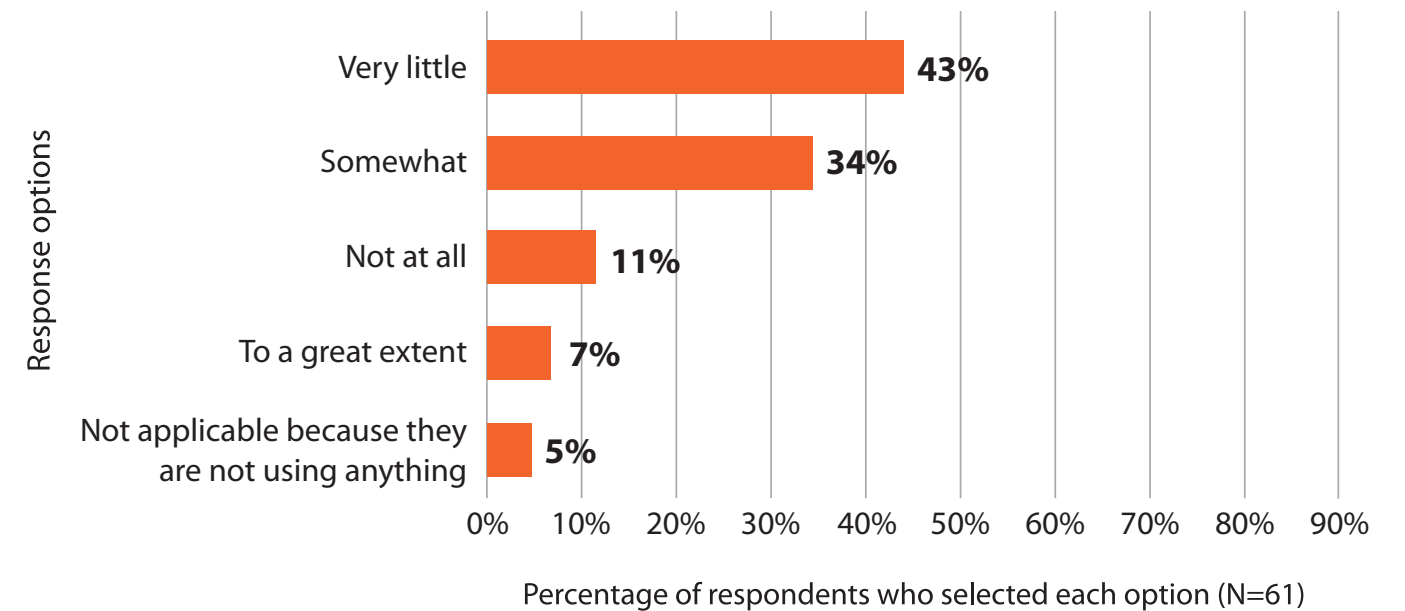
5. What medications or medical treatments have you or your loved one used (currently or previously) to treat symptoms associated with Galactosemia? Select all that apply



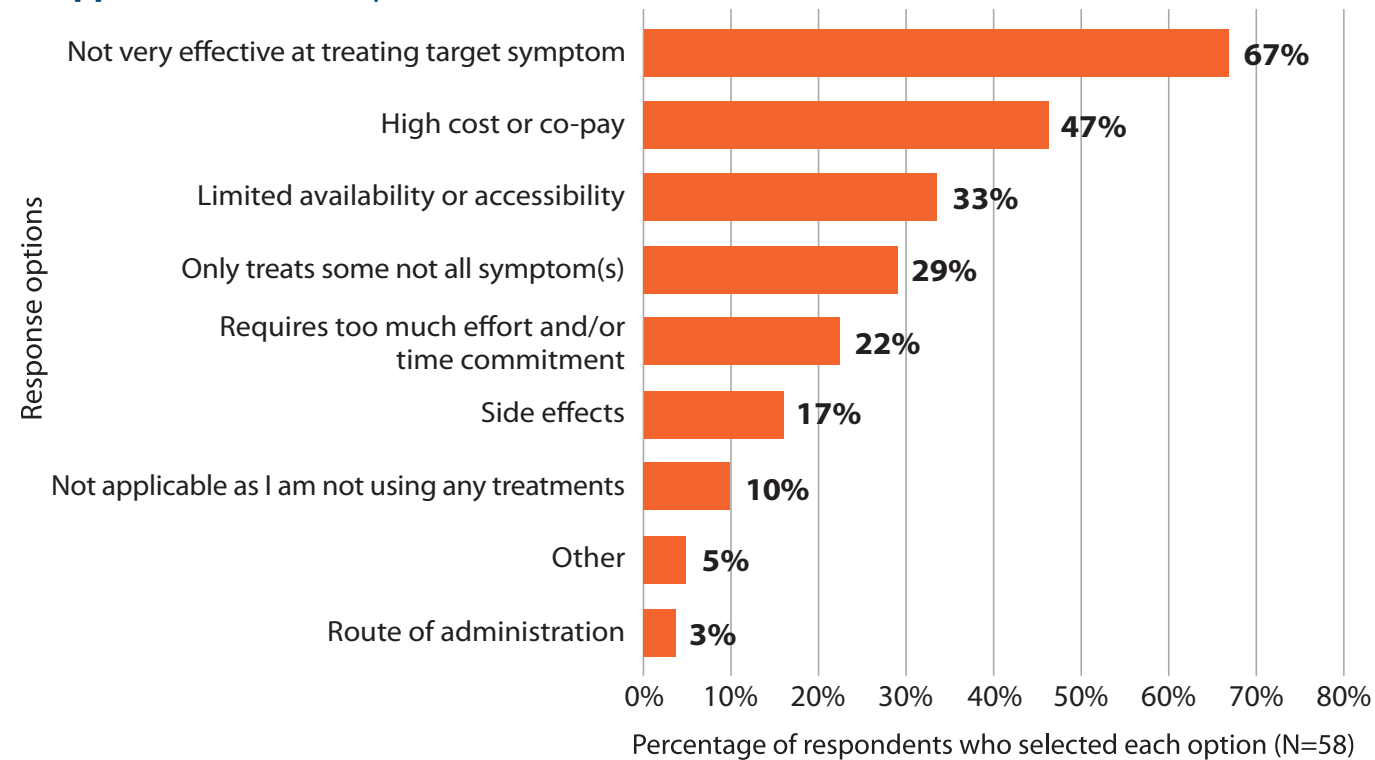
6. Besides medications or treatments (currently or previously), what have you or your loved one used to help manage the symptoms of Galactosemia? Select all that apply.



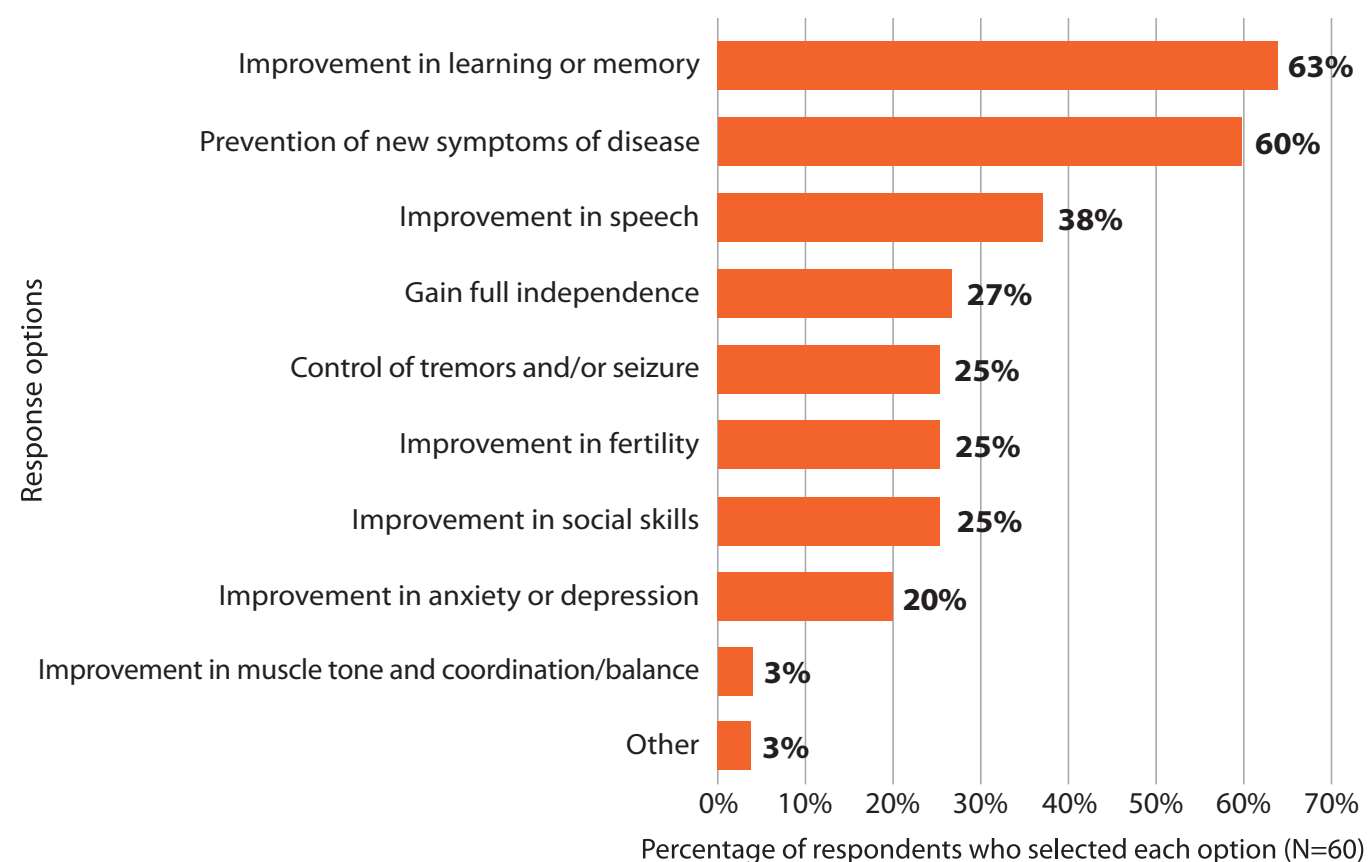
7. How well does your current treatment regimen treat the most significant symptoms of Galactosemia?



8. What are the biggest drawbacks of you or your loved one's current treatment approaches? Select up to 3.



9. Short of a complete cure, what TOP 3 specific things would you look for in an ideal treatment for Galactosemia?



Conclusion & Acknowledgements

Galactosemia Foundation and NORD dedicate this report to all of the families, patients, and caregivers in our community. Galactosemia is a rare, severe, and highly variable condition, but we are inspired by the strength and courage of our galactosemia patients and families.

The Galactosemia EL-PFDD meeting would not have been possible without the generous support and the hard work of many organizations and individuals. NORD and the Galactosemia Foundation wish to thank the many FDA representatives who were in attendance on the day of our EL-PFDD meeting to listen to our community speak about the challenges of living with galactosemia. We thank Dr. Sheila Farrell, Division of Rare Disease and Medical Genetics, FDA, for providing welcome remarks on behalf of the FDA. We also acknowledge the help and support of Will Lewallen from the FDA's PFDD staff for expertly guiding us through the EL-PFDD process.

Thank you Judith Fridovich-Keil, PhD, Professor, Department of Human Genetics, Emory University School of Medicine, for presenting such an enlightening description of galactosemia at our EL-PFDD meeting. Thank you to Brittany Cudzilo, Galactosemia Foundation's Vice President Information for providing such a concise summary at the end.

Thank you to Debbie Drell, Senior Director of Membership, NORD and Scott Saylor, Board Member, Galactosemia Foundation, who together moderated the meeting and facilitated such productive discussion. We thank the wonderful teams at Galactosemia Foundation and NORD who worked so hard to make our EL-PFDD meeting a success.

This report was prepared and submitted on behalf of NORD by Debbie Drell, Senior Director of Membership, Amanda Thomas, Sr. Events Manager, Alexa Moore, Vice President of Development; the Galactosemia Foundation by Nicole Casale, President, Brittany Cudzilo, Vice President and Scott Saylor, Treasurer, and by Chrystal Palaty, medical writer.

The Galactosemia EL-PFDD meeting was made possible, in part, with the generous support provided by Applied Therapeutics and Jaguar Gene Therapy. We wish to thank these organizations as well as all the clinicians, patient advocates and all those involved in advancing research and hope for the galactosemia community.

Finally, we wish to thank all of our community members who attended the EL-PFDD meeting and contributed in so many ways. Thank you to the many panelists and speakers for their vulnerability and courage in sharing their stories. Thank you to our community members who aided in discussion and polling and made the effort to connect and support each other in the chat. Our hope is that your courage in sharing your insights and perspectives will aid in the development of critical therapies and interventions for galactosemia.

NORD'S MISSION

We improve the health and well-being of people with rare diseases by driving advances in care, research, and policy.

GALACTOSEMIA FOUNDATION'S MISSION

To educate, support and provide advocacy for those affected by Galactosemia. Our goal is to network with professionals to inspire the treatment and advanced research of Galactosemia.



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