Supplementary Patient Stories

Shared with the EveryLife Foundation for Rare Diseases to Inform the

United States Senate Special Committee on Aging Hearing on

"From Regulator to Roadblock: How FDA Bureaucracy Stifles Innovation"

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The EveryLife Foundation for Rare Diseases invited the rare disease community members to reflect on the personal impact of recent FDA therapeutic development regulatory delays. Submissions have been de-identified to omit personal information, such as name and location.

Section One – Represents submissions from communities who have been directly affected by regulatory delays over the last year.

Section Two – Represents submissions from across the rare disease community, reflecting additional experiences living with rare diseases.

Section One

MPS II (Hunter Syndrome)

Submitted from primary family member

When I was 15 years old, I watched my 13-year-old brother take his last breath and be taken away from our home in a hearse. He had Hunter Syndrome (MPS II). The weeks leading up to that moment started with trips to the hospital, watching him flatline and be revived, bringing him home on life support, and eventually watching my parents make the decision to remove him from life support. “Awesome God” still rings in my head and brings me to tears as we sang it to him as he breathed his last. The faces of my family members etched in my brain as a deafening silence crushed us. I watched my dad disappear while he tried to find distractions. My siblings fell into their own bad habits to cope. I stayed home from school for a week trying to plan my brother’s funeral while my mom crumbled in despair.

Now I am 32, and my only child was just diagnosed with the same disease that took my brother’s life. He’s not yet 2 years old, and I see the delays beginning. This sweet, energetic toddler has to go to the hospital once every single week for the rest of his life to be poked with a needle and sit still in a room for 4-7 hours while medication is pumped into his body. A medication that can’t stop the disease from getting worse, and it can’t fix the damage that’s already been done. All it can do is slow the progression of physical ailments.

The medication does not cross the blood brain barrier, so it does nothing to preserve his cognitive ability. He’s 19 months old. Any word he once said, he no longer says. I may never hear my son say, “I love you”. We gave up our hobby of off-roading because it’s not safe for him. He may never have a hobby of his own, or a job, or a girlfriend, wife, or kids. He may never even see adulthood. The life expectancy in severe cases is 10-20 years old.

Every time I look at my son, I see him dying in my arms like my brother did. We need better treatments and we need them now. My son is 19 months old, has been on the current available treatment for 6 months, and I’m seeing decline. Japan has a treatment that crosses the blood brain barrier, and a one-time gene therapy has been created and shown to be successful in clinical trials but was denied by the FDA. We don’t have time to wait. I’m already watching my son move closer to death every day.

Submitted from secondary family member

I met my future brother-in-law in 2008. He was in his later phases of life living with the rare disorder, Hunter's syndrome. He had lost the ability to walk and feed himself, so my future in-laws would move him around in his specially made wheelchair. They were also tasked with feeding, bathing, and cleaning him. His care was 24 hours a day, seven days a week as children with this condition rarely sleep.

My mother-in-law was a home maker which helped as I do not know how two working parents could care 24/7 for a child living with these symptoms. He used to speak several phrases before the condition worsened, though at the stage I met him I unfortunately never had the opportunity of hearing his voice. I never had the chance to see him use sign language as his limbs only moved when he experienced a seizure. I never saw him walk or play as he was secured to his specialized wheelchair for his safety.

Doctors could do little to help mitigate the painful symptoms associated with this debilitating condition. Back then, there were talks of new medications circulating, awaiting approval, in addition to rumored research for possible treatments. The science and approvals never caught up, nor were passed in time and, consequently, were unavailable to patients like my brother-in-law. He passed at just 13 years old in Aril 2009. I had only known him for those brief 9 months. During those months, I witnessed multiple hospital visits, seizures, and stress.

I am now married to his sister, and it is a tragedy that he was not at our wedding with the rest of our friends and family. Fast forward years later, my sister in-law's first and only child, has the same condition, Hunter's syndrome. She and her husband both work full time and care for him 24/7 with the assistance of my mother-in-law, as she helps the best she can with her schedule. We love my nephew dearly as we did my brother-in-law. When you watch someone lose the ability to speak, walk, and then eventually pass away, heart wrenching is not strong enough of a phrase. It is excruciating.

With news of a one-time treatment using a gene therapy called, RGX-121, we were optimistic that our nation will take the path forward and approve this treatment for our nephew. This hope, unfortunately, came to a halt in recent days with the news of the FDA not approving this groundbreaking gene therapy. We were so close to receiving approval for this life-changing treatment.

We always hoped and prayed that as time moved forward so would the treatment for this rare condition. Now, we have data, research, and the medical technology to help children with Hunter's syndrome by using this gene therapy. RGX-121 is a one-time treatment, so it will not be a continual process which makes it cost efficient and provides better care for patients. Other advanced nations, such as Japan, have begun treating their patients with gene therapy. The United States was once the medical center of the world. We had the best treatment facilities, cutting edge research, and people flocked from all over the world to bring their children here to receive the best healthcare. We have the financial success that most nations dream about, so the budgets are available. These funds need to be routed through the appropriate departments. This studied and reviewed gene therapy must be passed for these children's sake, their family's sanity, and for the future of this country's well-being. We, as a people, have a moral duty to fund all healthcare issues including lesser-known disorders. This medicine would also serve to help other children across the globe. With the United States' market and influence in the medical industry, once this gene therapy is approved and used for treatment here, it will become available to all children worldwide.

Every child deserves a chance to live a healthy and loving childhood.

Submitted by additional family member

To the Senate Aging Committee;

I am representing the community of those affected by the rare disease Mucopolysaccharidosis Type II (MPS II), otherwise known as Hunter's Syndrome, in support of the proposed gene therapy, RGX-121 (clemidsogene lanparvovec). In the following excerpts, I will discuss my family's experiences with losing my brother to Hunter's, along with the newfound journey of my sister who now has a son with Hunter's. Additionally, I will go further into detail about how this life-altering gene therapy will have positive outcomes not only for our family, but to all families that have a child with Hunter's Syndrome.

I have been both a sibling and now an aunt to an individual with Hunter's Syndrome. Growing up with my brother, and presently my 1.5 year old nephew, whom was diagnosed with the disease just this last summer in 2025. In a family of 5 children, I am the middle child with two older brothers, a younger sister and what was once our youngest brother. He was born with Hunter's Syndrome in 1996, and would have turned 30 this February. Unfortunately, his predestined disease only allowed him to make it to 13 years old, back in 2009.

My parents were unaware of my mother being a carrier of this genetic anomaly, seeing as Hunter's Syndrome is an X-linked genetic disorder. My brother's diagnosis was made known when he was 2-3 months old, with doctors making it significantly clear that this was a very rare disease & his lifespan would be cut notably short due to it. Without a treatment or cure that could cross the blood brain barrier, the missing link to his DNA would ultimately be his demise. At the time, only being 5 years old myself, I remember my parents telling us four kids that he was special & we didn't know how much time we would get with him on Earth. But that we would treat him just the same as any other child, and give him the same life experiences possible. And that we did.

Everyone knew him, mainly because in his younger more able days he loved to rip off his diaper and hightail it down the street with my mom chasing after him—as he laughed in a guttural, pure sort of way. He was a gentle bliss of a human, despite the downfalls of his condition. He knew no different, as did we. He had a twinkle in his eye that let you know he understood feelings and emotions, even if he couldn't speak it outwardly. Especially in his younger years, he could run with all of us kids along with surrounding neighbors, he could throw and kick a ball like a pro, and ride his big wheel tricycle into the sunset. My mom definitely had her hands full, but he had a way of making your heart even more overfilled with joy without even trying. Unfortunately this made his decline as he aged that much more apparent and heart wrenching to watch before our very eyes, as his spark of life slowly fizzled out as the disease progressed throughout his innocent body.

Some of the downfalls of my brother's disorder lasted the entirety of his life, not just during the swift decline in later years. He spent his life in diapers with messy bowel movements that were never solid. Even though I never heard my parents complain, he slept in my parent's bed for all 13 years of his life. This was primarily so my mom could keep watch over him 24/7, as he also did not sleep through the night in younger years. This even evolved to long nights of watching Barney in the living room, where my mom would lay half awake on the carpet with her arm around him as he sat up watching into the wee hours of the night. If I woke up in the middle of the night, I remember the soft glow of the television down the hallway & would come out to join them. I remember my mom giving him nightly breathing treatments, the sound of the machine humming is a heavy reminder of one of the many ways my mom did all she could to bring comfort to her child. I remember occupational therapists and speech therapists coming to our home to work one-on-one with him, some more patient with his diagnosis than others.

One thing was for sure, my mom and my brother were inseparable. She never let him out of her sight, aside from the few times she allowed him to be apart of some special needs school

programs in his younger years when he was active. Unfortunately, even many of the special education teachers were not proficient in dealing with such a specialized child, and it ended in him communicating with us that he was being abused by them. He could say few words, but did learn some basic sign language to help communicate. My mom never entrusted anyone with him after that.

As time went on, the more nonverbal he became as he aged. He went from being a lively wild child full of spunk and joy riding his big wheel, to a quiet sedentary teen as he lost all his mobility. He began losing his balance before the age of 10 and eventually had to have a specially made walker, and eventually a custom wheelchair where he resided most of his days. He suffered from debilitating seizures, heavy mucous production (some families opt for tube feeding), as well as breathing treatments to help reduce the potential for pneumonia (this would eventually be the means by which took his life). Given the limited knowledge on MPS II back then, my mom did all that she could for him. In his last week of life, we knew it was his time. It seemed like hundreds of people who had been impacted by my brother walked through our front door to pay their respects as he laid there calmly and quietly analyzing each one. He held on for his final visitor, his home helper. My mom told him teary eyed, “It’s okay, you can go now” as he took his last breathe. Then, as if the heavens lit up, the darkly lit bedroom was glowing with golden rays of sunshine and his sparkling blue eyes lit back up as he smiled his final goodbye.

Losing a family member leaves a heartache no one can heal, and outliving your own child is something most parents will never have to be concerned with. There is a feeling of hopelessness and deep sadness when you are told your child may only live a certain number of years. It is clear that my mom was his around-the-clock caregiver, so his absence struck her the hardest & she has never fully healed from his passing. Our entire family has a special place in their heart for my brother, and can easily get choked up talking about memories of him. There are numerous emotions that accompany this sort of loss, the “what if’s”, along with each family member feeling their own sense of guilt and regret, longing to see their loved one yet being thankful they are no longer suffering from such disease. It has been 17 years since his passing, and simply writing this while reflecting on his life has taken me weeks to finish due to the heavy hearted nature of this topic. With how gut-wrenching and painful the healing process has been after almost two decades, any sort of lasting solution to save children with MPS II is essential. Prevention by means of treatment of those diagnosed with Hunter’s is a must, as well as saving their families from having to go through this in the same way that my brother and we did. I believe we should stop at no obstacle, by all means necessary, to make this dream a reality.

Our family's life with my brother was not always negative or sad—he played a significantly important role in not only our lives, but with everyone he met. He touched the souls of everyone he laid eyes on, and in his younger years, when he still had facial expressions, he had a smile that could melt a stone cold soldier down to their boots.

His special happiness brought a peace to the challenges that his life presented. Adjustments usually had to be made to make life work better for our situation, but it never seemed like that much of an issue in the moment.

However, recalling past memories, it is apparent there were quite a few sacrifices we made to have him present in anything we did together as a family unit. Average households got to take flights on vacation, whereas we were advised not to take him on a plane. Our friends got to go out to dinner regularly, whereas it was a common occurrence that he had a build-up of mucus & choked violently anytime he ate—disrupting surrounding tables, and many people acting openly disgusted that we would bring him out in public. Some people even voiced their opinions to us, as if we didn't have the same rights and freedoms to be there. We never even were able to fulfill most childhood dreams to go on a family trip to Disneyland, because it was deemed as unsafe for him to go on any of the rides. All of this to say, even though our "normal" may have been different than the average family, we were able to conduct our lives in our own version of normal.

What had become our normal sometimes puzzled passerby's who didn't always understand his condition and ailments. As a child, I remember asking my mom why other children and more distinctly, why grown adults, would stare blatantly at him when we were out in public. To me, he was just my baby brother. But to the average person, he was… different. With Hunter's Syndrome there are features that go along with having the missing enzyme in their DNA. Some key physical attributes such as "coarse" facial features along with large heads and abdomens with a short stature to name a few.

Nevertheless, we didn't let any of this get in the way of having time with family and we knew no different than having him along for every memory while he was here. He will forever be a blessing in our life, and I wish the options available and, hopefully soon to be available, were around when he still had a chance. He may have lived the course of his life, but my baby nephew has just begun his. With the studied and successful treatments of RGX-121, this new therapy is able to cross large necessary enzymes across the blood brain barrier—something we waited for all of my brother's life. I believe there is a significantly higher possibility of my

nephew being able to live a long, prosperous & cognitively capable life—given that a treatment such as RGX-121 has achieved the unthinkable.

Understanding the specifics on how Hunter's Syndrome inhibits those affected may also help emphasize the importance of treatments available and the timeliness that this medicine can be made available. Hunter's Disease is a deficiency in the enzyme iduronate-2-sulfatase (I2S), meaning that this missing enzyme has the primary job to breakdown complex sugars (glycosaminoglycans). Without this important enzyme, the sugars build up & consequently wreak havoc in the affected individual's organs, bones, & joints. This build up leads to progressive physical and cognitive decline, including aforementioned coarse facial features, enlargement of organs (organomegaly), joint stiffness, and developmental delays, usually appearing between ages 2 and 4.

This is why the gene therapy RGX-121 is so important. It is a one-time AAV9-based treatment developed by RegenxBio to specifically treat Hunter's Syndrome. It aims to deliver a functional iduronate-2-sulfatase (IDS) gene to the central nervous system to treat neurological manifestations. This single dose treatment targets the cognitive and behavioral impacts of Hunter's, which the current available standardized enzyme replacement therapy cannot address. The IDS gene is delivered using the NAV AAV9 vector, enabling cells in the central nervous system to produce the missing enzyme that causes Hunter's Syndrome. NAV AAV9 (Adeno-associated virus 9) is a leading gene therapy vector from RegenxBio's proprietary platform, which is well-known for it's capability of crossing the blood-brain barrier (BBB). By crossing the BBB efficiently, genetic enzymatic material can be delivered to the central nervous system and peripheral tissues. NAV AAV9 is famously used in treating spinal muscular atrophy (SMA) and is widely adopted for treating various genetic diseases due to its high transduction efficiency and tissue-targeting capabilities.

Ultimately, the CAMPSIITE trial (NCT03566043) has been investigating safety and efficacy of RGX-121 therapy in patients with MPS II. Despite positive data, in February 2026, the FDA issued a Complete Response Letter (CRL) for the Biologics License Application (BLA), requiring further analysis, thus to our dismay has postponed the approval of the gene therapy. The primary target for the treatment are males aged 4 months to 5 years old.

Given that majority of children with MPS II have the life expectancy of 10-20 years, every minute counts. The sooner these children can receive treatment, the less damage they will undergo by improving the quality of their life while increasing the number of years they can live a healthier life. The cognitive and physical damage eventually costs these children their lives,

just like it did to my baby brother. Most do not make it into their twenties even with precautions such as special diets, which may lessen the build up of sugars. With the missing enzyme in their DNA, their bodies eventually lose the battle.

Until now. For the first time, there are new treatment options being discovered. Enzyme Replacement Therapy (ERT) is currently available in the United States, that of which my 1.5 year old nephew has been undergoing weekly. The reality of ERT is that an implanted medical port had to be put in place in his chest, we call this his “Iron Man”. Meaning that he had to be put under anesthesia as an infant, which already came with its own risks. Not to mention the detrimental risks that a port itself has, with infection being the most common concern especially with a toddler. The first signs of infection in or around the port would be a fever, meaning that any time he spikes a fever he must be rushed to the children’s hospital—which is an hour away. Sometimes toddlers get sick & have a fever for more than 24 hours. But if his fever persists, he must return to the hospital to check for infection of the port by means of culture sample—that of which takes 72 hours for the sample results to grow a bacteria culture if present. All of these precautions and tests, then add another hour long commuting time to receive his weekly enzyme infusion into his port. This includes keeping an active toddler busy in a hospital room, while his chest is connected to an IV. The visit all together usually is about 5-6 hours, along with the 2 hour round trip from home to the hospital.

It should be made known that children with Hunter’s do not have normal sleep patterns. Just to simply take a nap, my sister has to bounce on a yoga ball with her 33-pound son as a means of calming him down enough to fall asleep in her arms for an hour or so. Night time sleeping is just as erratic and typically ends with both my sister and him exhausted with less than a few combined hours of rest each night.

Between the lack of sleep, potential port infection complications, and weekly treatments— the need for an alternative upgraded treatment option for my nephew is more evident than ever, as is likewise for all the other families affected by a loved one with Hunter’s. Though the enzyme replacement therapy has been a decent option to begin with, unfortunately, it does not cross the blood brain barrier to halt the cognitive decline of Hunter’s Syndrome. However, this is what sets apart the treatment we are requesting be approved at the earliest convenience. The RGX-121 treatment does cross the blood brain barrier & with a single treatment can give children like my nephew, the best chance at the most normal life possible. His life is in the hands of your discretion to pass and approve this treatment. Please don’t let him down. Thank you for your time and consideration on this matter.

[New submission]

I have two sons with MPS II, Hunter Syndrome, the severe form. One of our sons was diagnosed at 2 years old when I was carrying our 3rd son. Both sons had unexplained respiratory distress at birth, staying in the NICU for 17 days each, starting the difficult and terrifying journey to diagnosis. Our first born was diagnosed at 2 saving his soon-to-be born brother from late diagnosis, even 2 years old is too late. Our youngest began the approved treatment at 3 months old and has enjoyed a fantastic quality of life. The disease has been much harder for our older son, especially in the beginning when development is so critical. He lost his ability to speak. Both boys participated in clinical trials on different timelines. The younger boy started a clinical trial at 2 years old to treat the central nervous system. This trial went on for years and did not meet its endpoint and therefore was never available to our older son, who did not qualify. Our younger son has enjoyed a fantastic life so far with a bit of joint pain but normal height and the opportunity to attend high school in our community.

Our oldest was selected for a trial at 12 years old that also treats the brain and CNS with a different technology that attaches the enzyme to a blood brain barrier penetrating molecule carrying much needed enzyme to the brain. During the trial, he was able to be potty trained, is still alive, attends a school for autism, and has a tranquil, happy life! His brother needs the treatment that our oldest is on, but it takes years to even get to the approval stage, often more than 5 years. Even then, these treatments have to meet impossible standards to be approved, which change depending upon the administration and mood of the approvers. If you think of it from a child's perspective, it is a huge time commitment. You endure PK blood draws around the clock, multiple tests you don't understand, especially if you are not verbal, all to try to compare you to the rest of the American population. It is asking too much, but the children comply because it saves their lives, and that the results will save their brother and the many other men and boys with Hunter Syndrome.

Our youngest has been in a trial for 90% of his life to date and thinks of the medical teams as his family. But this time could have been spent living if treatments were not withheld, delayed, extended, denied, canceled, etc. Please make helping them live and treat their disease easier so that they may enjoy life. We hold hope for their futures and innovation that leads to functional cures!

[New submission]

We have an absolutely adorable little 4-year-old boy in our family with Mucopolysacchaeidosis type 11, AKA MPS 11. This is a rare, progressive, life-altering, and life-limiting disease.

Children with MPS 11 are likely to begin losing basic functions before the age of 10. This could mean the ability to walk, talk, play, just to name a few.

Our little guy is currently receiving the benefits of being in a clinical trial. This has helped him develop more like a child without MPS. If his treatment is delayed, he may lose these skills which will leave him more isolated. This can impact his ability to enjoy the life healthy Children can take for granted. Most children with MPS don't make it to their 20th birthday, with some not even getting to see double digits. Not to mention, the quality of life during those years can be so limiting.

I could go on and on about the physical challenges facing Children with MPS, but I would be remiss if I didn't mention the emotional impact this disease has on his family. For his parents it is heart wrenching and requires Herculean effort to face each day with a smile for the sake of the child MPS along with any siblings they might have. The Grandparents' heart aches for the child but have to watch their own children walk through life with a heavy heart not knowing what each day will bring. For the siblings it is confusing and often times difficult for them to understand why they can't always get the attention they need because their parents are busy caring for a sibling with special needs. For the extended family, it is a feeling of helplessness because we don't always know how to help.

On paper our little guy can be a statistic, but to the family he is the whole world. He truly is a ray of sunshine, an inspiration because of how bravely he faces each day and the light of so many of our lives.

[New submission]

My godson was diagnosed with a rare genetic life altering and terminal disorder two years ago. At two years old he was diagnosed with MPS II. This diagnosis was terrifying for his parents and loved ones. MPS II comes with a host of symptoms and complications. When he got the diagnosis of short life expectancy, the clinical trials that he is in have been a beacon of hope. We have seen him progress and developed beyond what was expected. With continued access, my godson and other kids with MPS II have a chance and hope for a better life as they battle their condition which carries a limited life expectancy and devastating developmental disabilities.

[New submission]

A little over a year ago, my life looked completely normal. We had three thriving boys, and I was nine months pregnant with our daughter. No family history of genetic disease on either side. Our youngest needed hearing tubes and a small hernia repair, but nothing unusual. He had a little bit of a speech delay, and I suspected maybe a mild bit of ASD, but nothing severe enough to pursue testing. All and all, we were healthy, thriving, and living the American dream.

Then I saw a little girl on TikTok who looked exactly like my son. She was dying from a terminal neurodegenerative disease. Her parents had believed she was perfectly healthy, too — until social media connected the dots. Something in me quietly shifted that day.

I sought reassurance from six different physicians and a pediatric clinical geneticist. One by one, each of them told me my son was healthy and developing normally. That I was worrying about nothing. That if he started to regress, we would "deal with it then." But in neurodegenerative disease, "then" is too late. I could not ignore what I had seen. So, I ordered my own carrier screening test, and through that process, I unraveled his diagnosis myself.
There was no roadmap. Most of the doctors we encountered had never even heard of this disease. When the results came back, I was not handed a treatment plan. I was handed the responsibility to save my child's life.

I had to move immediately. With neurodegenerative disease, time is brain. Every month without treatment means irreversible damage. I had to locate specialists, find a clinical trial, and fight for access, all while knowing any delay could cost my son his future.

My son, now almost five years old, has Mucopolysaccharidosis II — MPS II (Hunter syndrome), a rare pediatric neurodegenerative disorder. Children with MPS are missing a critical enzyme responsible for breaking down toxic cellular waste called heparan sulfate. Without that enzyme, heparan sulfate accumulates in the brain and body, leading to progressive physical and neurological decline. Without treatment, life expectancy is typically in the mid-teens. There is one treatment approved by the FDA, but it cannot cross the blood-brain barrier. Despite receiving therapy, most children still pass away from neurological complications before the second decade of life.

I found a clinical trial for a brain-penetrating therapy that delivers a synthetic version of the missing enzyme and crosses the blood-brain barrier. This drug is already approved in Japan with robust data. But time was not on our side. The trial was almost full and projected to close at the end of the month. As an enrollment requirement, children had to receive 12 consecutive weeks of the FDA approved therapy, elaprase, before they could be screened. Against all odds, somehow a spot remained open as my son received his weekly elaprase treatments, and we made it into the trial. When I opened the email confirming he was accepted and randomized to be treated with the drug instead of the placebo, it was the greatest moment of my entire life. My son is now receiving this treatment, and he is thriving. Every week, I pack up my children and fly out of state to give him the chance to fight. The chance to save his life.

The difference in him is remarkable. He is not just holding steady; he is progressing. He is catching up to his neurotypical peers. For the first time since his diagnosis, his doctors have told us that with this treatment, a near-normal lifespan is within reach.

For a disease that once came with a teenage expiration date, that is nothing short of extraordinary.

But even after beating all the odds, we are living in fear that we could lose access. Not because the science failed. Not because companies stopped fighting rare disease.
But because of regulatory inconsistency.

For more than 30 years, the MPS community has fought for the FDA to recognize heparan sulfate as a valid biomarker — our only meaningful measure of disease progression. The FDA agreed. Trial designs were negotiated over a decade. Accelerated Approval pathways were discussed and aligned. Sponsors invested hundreds of millions of dollars. Then, after completion of trials, the FDA reversed course. In a recent Complete Response Letter, the agency questioned the reliability of heparan sulfate — the very biomarker it previously agreed to use — criticized trial designs they had accepted and rejected applications despite strong clinical data and visible patient benefit.

This is not an isolated case. Many rare pediatric neurodegenerative therapies are experiencing similar outcomes. Accelerated Approval was created precisely for:

- Small patient populations
- Serious and irreversible diseases
- Situations where traditional large, randomized placebo trials are infeasible

Yet ultra-rare pediatric genetic diseases have seen minimal utilization.

Meanwhile, standards appear to be shifting mid-stream:

- Biomarkers previously accepted are being reconsidered.
- Natural history comparisons are being rejected after prior agreement.
- Trial designs and expectations are changing after sponsors have already invested years and resources.

For small biotech companies, this unpredictability is existential. Companies developing life-saving therapies are beginning to withdraw from the space because they cannot absorb the regulatory risk.

The economic cost is staggering. Thirty million Americans live with a rare disease. The annual U.S. economic burden exceeds $1 trillion. Delaying treatment does not save money — it increases lifetime dependence on Medicaid, disability services, and eliminates future workforce participation.

But this is bigger than dollars.

Biotech companies that have invested millions into rare disease therapies are going bankrupt after these rejections. I personally know of ten companies with strong clinical data that have had to hand their programs to nonprofits and walk away because funding ran out.

If this continues, we will lose the very companies fighting to save our children.
Innovation will not disappear. It will move, and other countries are already preparing to receive it.

For families like mine, that does not just mean economic loss. It means losing access. It means watching breakthroughs happen somewhere else while our children decline.

In rare pediatric neurodegenerative disease, waiting is not neutral. It is watching your child slip away in front of you.

Last week, on a private call among MPS parents, a father's voice broke as he said something that will stay with me forever: "I wish my little boy had cancer. At least then we'd have a chance to fight."

We have submitted letters. Our physicians have met with FDA officials repeatedly. Advocacy leaders have walked the halls of Congress and even visited the White House. We have done everything asked of us. Yet rejection after rejection continues.

I am writing this from my son's hospital bedside. He will require weekly infusions for the rest of his life simply to survive. We have accepted that reality. What we cannot accept is losing access to the treatment that is keeping him alive because agreements are no longer honored.

We are not asking for shortcuts. We are asking for consistency. For enforcement of existing law. For regulatory reliability. We are asking for our right to try.

In the wealthiest, most innovative nation on earth, no child should lose their life because the system meant to protect them cannot keep its word.

We are closer than ever to rewriting the future of this disease, and all rare diseases. Please don't let my generation become the next group of MPS mothers who stand at gravesides instead of graduations.

[New submission]

Submitted by grandparent

I am a representative for the MPS II {Hunter Syndrome} community. This is a rare, progressive, and life-limiting genetic disease that mainly affects boys.

My grandson was a healthy baby. That changed when it was discovered that he had Hunter Syndrome. This is caused by a deficiency of the enzyme iduronate-2-sulfatase. This leads to an accumulation of glycosaminoglycans, especially heparan sulfate, throughout the body and the brain. Over time it continues to build up, damaging the organs, bones, airway, heart, and the central nervous system.

I want you to know our boy. Not just his disease. He dreams of amazing adventures. There are no mountains too high to climb, no ocean too far to sail across, no star he cannot reach. He is a Super Hero Boy! His Dreams are bigger than Life and his Imagination brings them all alive.

He shines bright - he wants to do it all. To meet every person he encounters and they in turn feel his glow. His personality is his gift, his laughter, his dancing, his stories and his joy no matter what he has gone through. He is my Hero and my Best Guy. How lucky am I to be his Gra!

Yet there is more to his story. Hunter Syndrome is relentlessly progressive in the neuropathic form, severe, children experience:

- Developmental delays than cognitive regression
- Speech and communication loss
- Behavioral changes
- Airway obstruction and recurring infections
- Cardiac valve disease and skeletal deformities

Many of these boys lose the skills they had. Their independence shrinks as their words fade away. Life expectancy is often in their teens. Time is a neurological function. In six months, language can disappear. In a year there can be serious cognitive decline and loss of mobility.

With so many days spent in a treatment bed for him, movies create a different world. I know someday he will direct and produce his own stories. I hope I will be a part of them. His imagination takes him into jungles, exploring mountains, and walking shorelines for treasures. So many steps are waiting to be taken. The World is waiting for him to take it by storm! To Fly, to soar, to create, to grow, to be Super Hero Boy!

Yet the world never just hands us something without a catch. His catch is so large and so unknown. My grandson has Hunter Syndrome, a genetic disorder causing Heparan Sulfate, what is known as a GAG, to accumulate in his body and brain. A simple explanation is that debris builds up within him. With the new medical trials that are now taking place, he would have the chance for the quality of life every child should have.

Currently there is an approved FDA therapy, but it has serious limits. Elaprase {idursulfase}, an enzyme replacement therapy given weekly by infusion. It is able to help with treating systemic symptoms. It can improve endurance, reduce organ enlargement, and stabilize some physical complications. This all matters, but it does not cross the Blood-Brain Barrier. It does not address the cognitive decline that comes with the most devastating form of Hunter Syndrome. Children can receive Elaprase for years and still lose their personality, language, and memory.

This is the central unmet medical need - a therapy that reaches the Brain.

Promising blood-brain barrier penetrating therapies have been developed to address neurological disease. Regulatory uncertainty and delays are holding this back. The questions about already established biomarkers like cerebrospinal fluid heparan sulfate and shifting expectations and the evidence needed to demonstrate neurological benefits. The impact is immediate when late in the development or approval process the standards are changed. This slows the trials and investment backs away. The real crisis is what happens to the children as they then decline. Delay leads to irreversible loss. In progressive neurological disease, success comes with stabilization.

In six months, he could lose his speech. In one year, his world could shrink away, measurable cognitive drop, worsening airway and cardiac disease, loss of independence with his life skills. Once he suffers these losses, his neurons will not regenerate and bring them back.

For Hunter Syndrome children and their families, Time is not an abstraction. It is the difference between preserving who our sons are or watching them disappear.
Regulatory delays are not administrative. It is Childhood, it is memories, it is their brain cells.

Will you support us in our quest to make this happen? Will you help us make sure that Super Hero Boy's Story has a happy ending? My grandson and the other Super Hero Boys are counting on you.

Submitted by additional family member

I represent families in the MPS II (Hunter syndrome) community — a rare, progressive, and life-limiting genetic disease that primarily affects boys, including my son.

Hunter syndrome is caused by a deficiency of the enzyme iduronate-2-sulfatase, which leads to the buildup of glycosaminoglycans — especially heparan sulfate — in cells throughout the body and brain. Over time, that accumulation damages organs, bones, the airway, the heart, and, in the severe form, the central nervous system.

Children with the neuronopathic form experience developmental delay followed by cognitive regression, loss of speech, behavioral changes, airway obstruction, cardiac disease, and skeletal deformities. Many boys lose skills they once had. Words disappear. Independence fades. Life expectancy is often in the teens.

There is an FDA-approved therapy, Elaprase (idursulfase), a weekly enzyme replacement infusion that treats systemic symptoms. It can reduce organ enlargement, improve endurance, and stabilize some physical complications. Those benefits matter. But Elaprase does not cross the blood–brain barrier. It does not address the cognitive decline that defines the most devastating form of Hunter syndrome. Children can receive treatment for years and still lose language, memory, and personality. That is the central unmet need: a therapy that reaches the brain.

Promising blood–brain barrier-penetrating therapies have been developed to target the neurological disease process. Yet our community has faced regulatory uncertainty, including disputes over biomarkers such as cerebrospinal fluid heparan sulfate and shifting expectations about what evidence is required to demonstrate neurological benefit. When approvals are delayed or standards change late in development, the disease does not pause. Children continue to degenerate while the FDA keeps moving the goalposts.

In a progressive neurodegenerative disorder, stabilization is success. Delay is irreversible loss. Six months can mean my son loses two-word phrases. A year can mean measurable cognitive decline, loss of toileting independence, worsening airway disease, or new cardiac complications. Neurons do not regenerate. Skills rarely return once lost.

For families like mine, time is not abstract. It is brain cells. It is memory. It is childhood.

[New submission]

When my son was two, my husband received a call from my son's ENT: "We need to see your wife and you in our office this week." Something was wrong. He had had routine ear tube surgery, and all had gone well. There was something that the doctor didn't think I was strong enough to handle. In his office, I heard those three letters for the first time: "MPS." And in a few seconds, my dreams and expectations for my family, for my son, were shattered. My baby boy was going to die.

As soon as we got home, I went for a long run. I ran so fast that I couldn't breathe, and then I fell on the ground and cried. I don't know how long I was there. But eventually, I picked myself up and headed home, determined to do what I needed to save my son's life.

As any parent would, I searched and found the best doctor in the country for MPS. We packed up the car and headed there. He said my son needed to start enzyme replacement therapy (ERT) immediately. But he cautioned us that the standard ERT would not cross the blood-brain barrier, so my son would continue to suffer brain damage until eventually he passed in his teenage years.

The doctor offered a sliver of hope. He told us that the only trial addressing the cognitive decline of severe MPS II was on hold by the FDA. We waited two long years – watching to see if my son was losing skills, sleeping next to him, so I could hear him breathe, wondering how I could continue to live when he passed away.

My son finally entered the Intrathecal Enzyme Replacement trial at age 4. I knew going into the trial that this wasn't a cure. I didn't expect him to regain the skills he had lost. All I hoped for was stability, slowing down the progression of the disease, and keeping my boy alive.

The FDA did not approve the intrathecal drug, but the pharmaceutical company and the FDA agreed that the boys in the trial could continue receiving it. Each month, for the past 143 months, he is sedated and receives the enzyme he is missing directly into his cerebrospinal fluid through a lumbar puncture.

At 15, my son can still do things I never imagined he could. He is stable. He and I go on three-mile walks together. He is still feeding himself and breathing without assistance. He completes 50-piece jigsaw puzzles and runs to his school bus by himself every morning.

The dose in the intrathecal trial was too low and not given often enough, but the drug did slow down his progression and has stabilized him. It gave him more time to run on the beach, chase the dog, and laugh at Curious George.

Other boys have not been as fortunate as my son. They did not get into the intrathecal clinical trial he was in, so their disease progression continued.

I will never forget the agonizing wait in the recovery room on that Friday in 2014 to see if he would be randomized to the drug or the control group. When the nurse came out and told me he was randomized to drug, I burst into tears. She gently touched my shoulder and said, "I don't think you heard me correctly. I said he would be getting the drug." I heard her correctly, but to me, her words came out, "your child is going to live."

I rushed out into the waiting room to tell my friend, "He is on drug! he is on drug!" As we hugged and cried, another mom sitting there asked me softly, "Is that for the MPS II trial?" I answered excitedly, "Yes, it is!" She replied, "My son has MPS II also." I asked her, "Is your son in the trial?" She said, "No, he is too old to qualify."

Her son would have to wait. But the drug was not available in time for him, and he died. Not because there was no treatment, but because he was too old. Imagine, as a parent, knowing there is a drug that can help your child, but not being able to receive it because someone who has never met your child decides your child is too old to receive treatment and isn't worth saving.

My goal for him was always to keep him alive long enough for something better to come along. Something better is here. For MPS II, we have two drugs up for approval. I have seen the kids taking the drugs, talking, singing, and laughing. They are stable. That's all I want for my son.

These drugs will change the trajectory of MPS II forever. We haven't had a new drug approved for this disease since 2006 -- twenty years without innovation. Our MPS II community is anxiously awaiting approval for tividenofusp alfa. We have been patiently trying to keep our kids alive, so they can experience the benefits and have more happy days. This drug needs to be available to every boy with MPS II, regardless of their age. Please don't discriminate against our older boys. They also deserve treatment and a chance at life.

[New submission]

Submitted by parent

My son is four years old. He is funny, determined, and full of life. He is also living with Mucopolysaccharidosis type II, or MPS II, a rare, progressive, and life-limiting disease.
When you receive an MPS II diagnosis, you are not handed a hopeful roadmap. You are told to enjoy what your child can do today… because there will come a day when he will lose those skills. A day when walking may stop. When words may fade. When pieces of him will be taken away right in front of you in just a few years. You're forced to embrace the mindset of packing in a whole lifespan in just a hand full of years before preparing to say goodbye forever.

That is what delayed access to treatment means to me.

My son is part of a clinical trial. As his mother, I fully understand the risks. I understand the unknowns. I understand that research carries uncertainty. But what people need to understand is this: the alternative is unfathomable.

Without intervention, MPS II had already begun to steal his development. Before treatment, the disease limited nearly every skill a typical two-year-old should be building. We watched him struggle to form words. To coordinate his movements. To keep up.

Since receiving treatment, I have watched something extraordinary happen. He can form words. He can talk. He can run. He can make friends. He can build meaningful connections. For most families, those milestones are expected. For us, they are everything.

This is what quality of life looks like for a boy with MPS II. It is not perfection. It is possibility. It is time. It is siblings getting to know their brother. Really know him, before the unthinkable day when we may have to say goodbye.

Delayed access to treatment is not an administrative issue. It is not paperwork. It is not policy. It is children losing abilities they may never regain. It is families watching time slip away while waiting. It is young brothers and sisters being asked to process more grief than many adults ever have to face.

Children with MPS II are losing their lives with every delay. And families like mine are fighting. We are sacrificing time, emotional strength, and countless hours in hospital because this is the only chance our child has to keep walking, to keep talking, to keep being here as fully as possible.

When we talk about access to treatment, we are talking about time. And for families like mine, time is everything.

Submitted by parent

Right now, he is four.

Right now, he is in the living room lining up his trucks like they're heading into some kind of mission. He runs, not fast, not steady, but with everything he has. When he falls, sometimes he laughs. Sometimes he just looks at me.

Confused.

Like I'm supposed to explain something.

Most of our days look normal from the outside. Cereal. Cartoons. Toys everywhere. "Dad, watch this!" shouted from the other room like it's urgent.

And I watch. I always watch.

But our mornings start earlier than most families.

Because his doctor is far. Far enough that we wake up in the dark to make it on time. We move quietly through the house. Pack bags. Load the car. Try not to wake the other kids too early before they go to their grandparents' house.

They know the routine now.

"Is it Liam's doctor day?"

They ask it casually. But they know something is different.

Once a week they see their grandparents while Mommy and Daddy take their brother to the doctor “to help him grow big and strong” That’s the phrase we use.

And I wish it were that simple.

There are nights we stay up later than we should, too. Talking. Researching. Watching him sleep. Trying to stretch time. Trying to control something. Trying to feel like we’re doing enough.

I’ve always believed hard work fixes things.

I’ve built things. Repaired things. When a toy breaks in our house, I look at my kids and say the same thing every time.

“Don’t worry. Anything can be fixed.”

I’ve said it for years without hesitation.

But this…

This can’t be fixed.

We wake up early.
We stay up late.
We drive miles.
We sit in waiting rooms.
We ask every question.
We do everything we’re told.

And there is still no fixing MPS II.

I can’t outwork a genetic disorder.
I can’t grind harder and beat it.
I can’t trade sleep for progress.
There is never enough time, not in the mornings, not at night, to rewrite what’s written in his DNA.

That realization humbles me in a way nothing else ever has.

During the day, though, he’s just four.

At the playground, he lines himself up next to kids his age. In his mind, he's exactly the same. He reaches for the same bars. Tries to climb the same ladders. His hands struggle to grip the way he wants them to. His legs work harder than they should have to.

He looks frustrated sometimes. Not defeated. Just confused.

He doesn't understand why his body won't do what he tells it to.

He's only four.

When he finally makes it up a step or jumps a little farther than last week, I cheer like he just won something huge. Because for him, it is huge.

Pride and heartbreak live side by side in those moments.

And then there was the day he looked at his grandfather and said, "Pop, you went to the doctor and he made you better."

He said it so simply. Like that's just how the world works.

You go.
You get fixed.
You come home better.

And then he looked at him.

It wasn't dramatic. It wasn't loud. Just a quiet look that asked a question he didn't fully know how to say.

Why am I not getting better?

I felt that in my chest in a way I can't really explain.

Because how do you tell a four year old that sometimes the doctor isn't fixing, he's managing? That sometimes "better" doesn't mean what we want it to mean?

At night, when the house is quiet, that's when the future tries to creep in.

Not as certainty.
Just as fear.

Fear of losing abilities.

Fear of watching something slip away.
Fear of outliving my child, a sentence that feels wrong even forming in my mind.

My wife and I don't always say it out loud. Sometimes we just look at each other when he struggles. There's love there. There's exhaustion there. There's strength we didn't ask to have.

We worry about his siblings. How this shapes them. Whether they feel the weight of it even when we try to shield them. Whether they'll someday understand the early mornings and the long drives differently than they do now.

But today, they're just kids together.
And today, he's just their brother.

Parenting a child with MPS II means living in real time.

It means celebrating what he can do while quietly grieving what might not come.
It means kneeling beside him when he's frustrated and saying, "We'll do it together," even when I wish I could do it for him.
It means learning that love doesn't fix everything, but it shows up anyway.

And here's the part that's hard to say out loud.

There are treatments in trial phases right now. There is research happening. There is real hope, hope that could mean more time, more ability, more life for children like my son.

But time is not theoretical for us.

Every delay is not paperwork.
It is not procedure.
It is not a line item on a calendar.

It is a child losing ground.

It is a father waking up early and staying up late knowing there may be something that could help, but it is still out of reach.

I understand safety.
I understand process.
I understand responsibility.

But progressive diseases do not pause while we wait.

My son does not get those months back.
He does not get those years back.

Right now, he is four.

Right now, he is laughing.
Right now, he is trying.
Right now, he reaches for my hand without looking because he trusts I'll be there.

I can't fix what's written in his DNA.

But you have the power to decide how quickly hope can reach him.

And right now, that matters more than anything.
Please don't let delay be the reason he runs out of time.

Submitted by aunt

My nephew and godson is four years old. Of the eight grandkids, he is the happiest of them all. He is also living with Mucopolysaccharidosis type II, or MPS II, a rare, progressive, and life-limiting disease.

When you receive an MPS II diagnosis, you are not handed a hopeful roadmap. You are told to enjoy what your child can do today… because there will come a day when he will lose those skills. A day when walking may stop. When words may fade. When pieces of him will be taken away right in front of you in just a few years. You're forced to embrace the mindset of packing in a whole lifespan in just a hand full of years before preparing to say goodbye forever.

That is what delayed access to treatment means to me.

My nephew is part of a clinical trial. I understand that research carries uncertainty. But what people need to understand is this: the alternative is unfathomable. Without intervention, MPS II had already begun to steal his development. Before treatment, the disease limited nearly every skill a typical two-year-old should be building. I have watched him struggle to form words. To coordinate his movements. To keep up.

Since receiving treatment, I have watched something extraordinary happen.

He can form words. He can talk. He can run. He can make friends. He can build meaningful connections.

For most families, those milestones are expected. For us, they are everything.

Delayed access to treatment is not an administrative issue. It is not paperwork. It is not policy. It is children losing abilities they may never regain. It is families watching time slip away while waiting. It is young brothers and sisters being asked to process more grief than many adults ever have to face.

Children with MPS II are losing their lives with every delay. And families like mine are fighting.

We are sacrificing time, emotional strength, and countless hours in the hospital because this is the only chance he has to keep walking, to keep talking, to keep being here as fully as possible.

When we talk about access to treatment, we are talking about time.
And for families like mine, time is everything.

Submitted by godparent

My godson was diagnosed with a rare genetic life altering and terminal disorder two years ago. At two years old he was diagnosed with MPS II. This diagnosis was terrifying for his parents and loved ones. MPS II comes with a host of symptoms and complications. When he got the diagnosis of short life expectancy, the clinical trials that he is in have been a beacon of hope. We have seen him progress and developed beyond what was expected. With continued access, my godson and other kids with MPS II have a chance and hope for a better life as they battle their condition which carries a limited life expectancy and devastating developmental disabilities.

Submitted by aunt

My nephew is a Rare Gem. He is so unique and so uncommon that he is one of only 500 little boys with the same enzyme deficient genetic disorder. Our family absolutely adores and treasures this gift of a spunky, fun loving, fierce little fighter. Please let me tell you why the FDA needs to approve lifesaving medication for him and the other 499 boys who will simply not survive without this drug.

His genetic makeup is such that he does not have the capability of creating an imperative enzyme that is used to break up waste in every single cell throughout the entire human body. As a result, every cell in his body retains waste and grows in size, eventually causing mobility issues, limited organ function and most especially decreased cognitive function. Almost 2 years ago, my nephew began receiving FDA approved enzyme infusions on a weekly basis to help

each cell break up that waste. He receives this drug out of state, so once a week my family must make the trip. We quickly found out that the FDA approved enzyme treatment did not cross the blood-brain barrier. So, while it did improve his mobility and organ enlargement, it did nothing to assist his speech processing, hearing loss, and general cognitive function. As a matter of fact, his general cognitive function only got worse and worse.

Not long after beginning the enzyme treatment, he qualified to partake in a trial of enzyme replacement drug that was not yet approved by the FDA. He has been in the enzyme replacement trial for just shy of a year and a half. This drug that we are begging for FDA approval on crosses the blood brain barrier and effectively administers the enzyme to brain cells, drastically improving my nephew's hearing, seeing, speech, and overall cognitive function.

This drug is imperative to my nephew's survival because without it, there is no other alternative to maintaining the same little boy he is growing to become. Without this drug, his brain function will only decline with no hope for any reprieve. Please approve the Denali therapeutics enzyme drug that crosses the blood brain barrier and keeps my nephew and all the rare gems alive and thriving.

Submitted by grandparent

My 4-year-old grandson has a rare disease known as MPS2, a life limiting, progressive disease that affects every system in the body for which there is no cure. The lifespan for children with this disease is usually until their second decade.

In some ways he is very much like a typical 4-year-old. He loves Hot Wheels and movie nights with his young siblings, playing in the snow, and making sandcastles on the beach. Currently the only available treatment for his condition does not cross the blood/brain barrier, so it does not address the cognitive decline that occurs with MPS2.

For the past two years, Liam has been in a clinical trial testing a treatment that does cross the blood/brain barrier. Since he started this trial, we have noticed great strides in his development. His speech and his physical capabilities have improved dramatically. He can run and jump, ride his bike, and play on the playground. He's full of wonder and questions about everything under the sun. This has given us great joy and hope. Joy that he is doing so well, hope that he will continue to do so.

But for children living with MPS2 that do not have access to this life changing treatment, abilities are slipping away, as cognitive decline sets in. Every delay in the FDA approval process results in lost abilities, lost lives, devastated families. Time is not on our side in this

battle against a relentless debilitating enemy, and the current FDA approved treatment falls woefully short.

Families have been watching, desperately waiting for access to meaningful, life-giving treatments for rare diseases - waiting for treatments that can give our children a fighting chance.

[New submission]

My son is diagnosed with MPS II, also known as Hunter Syndrome. He loves playing with his cars, hugging his friends, and has a laugh that shines through his entire body. He is joy. He is light. He is our whole world.

But every single day, I live with the fear that he will lose the skills he has worked so hard to gain — the ability to talk, to walk, to feed himself, to simply play like a child should. With this disease, time is not on our side. Every day is precious, and it is time we do not have to waste.

There are treatments and therapies currently in clinical trials that children like my son desperately need. Delaying approvals does not just delay paperwork — it costs children skills they will never regain. It steals milestones. It steals independence. And ultimately, it shortens lives.

Please listen to our stories. See our children. Remember that behind every policy decision is a child fighting to stay who they are, and a family fighting to give them the best possible quality of life.

Our children cannot afford to wait.

[New submission]

I am writing to you as a grandmother of a spirited four-year-old boy who is navigating the realities of Mucopolysaccharidosis type II (MPS II), a rare and progressive condition that is life-limiting. My grandson is not just a patient; he is a vibrant individual filled with joy, determination, and an unwavering zest for life. Nevertheless, his diagnosis has cast a shadow over our family, revealing the painful truth of a disease that steals skills and abilities, often far too soon.

When we received the diagnosis of MPS II, we were told to embrace every moment and enjoy what he can do today, knowing that there would come a day when he might lose those abilities. It is a crushing reality to confront: the prospect of watching my grandson possibly lose his ability to walk or talk. Delayed access to effective treatment means lost abilities and time – a heartbreaking reality for families like ours.

My grandson is currently involved in a clinical trial that carries risks and uncertainties involved, but the alternative is unthinkable. Before treatment, MPS II limited nearly every aspect of his development. However, since receiving treatment, we have been blessed to see him achieve milestones that we hold dear: he now talks, runs, and forms meaningful friendships. For our family, these aren't simply developments; they are lifelines.

What quality of life looks like for my grandson with MPS II is not a pursuit of perfection but rather striding towards possibility. It means allowing his siblings to know him in his entirety before we eventually face the unthinkable. Delayed access to treatment means watching him lose abilities we fear he may not regain. It means families like ours watching time slip away, with siblings grappling with a grief that few adults ever truly understand.

I implore the FDA to acknowledge that when we discuss access to treatments, we are discussing time—an invaluable currency for families battling rare diseases.

For us, time isn't just about years; it is about the quality of those moments, watching my grandson walk, talk, and grow.

Thank you for your attention to our story and the stories of countless other families like ours. We hope for your understanding and support as we advocate for expedited access to treatment for MPS II, allowing children the opportunity to thrive, live fully, and embrace the lives they deserve.

[New submission]

My friend's son is 4 years old and has MPS II. He is in clinical trials taking risks to himself so that others may have a brighter future. At only 4 years of age him and his family have made decisions, most of us cannot even fathom.

Delays to this treatment are risking the progress made and wasting time people do not have.

Please do not deny access or delay treatments

[New submission]

My son lives with the severe neuronopathic form of Hunter syndrome. Hunter syndrome is a rare, progressive, genetic condition that primarily affects boys. It occurs because the body cannot properly break down certain complex sugars (glycosaminoglycans). These sugars build up in cells and organs, causing damage throughout the body, including the brain.

How Hunter Syndrome Affects Our Life:

Hunter syndrome is not just one symptom. It affects nearly every part of my son's body and development. He receives weekly enzyme replacement infusions to help manage the disease in his body. But the most devastating part is that the current treatment does not cross the blood-brain barrier, meaning it does not protect his brain.

Hunter syndrome can cause:

- Developmental delays
- Cognitive decline
- Speech regression
- Behavioral challenges
- Progressive organ damage
- Hearing loss
- Joint stiffness and mobility limitations
- Shortened life expectancy in severe cases

For families like ours, the most painful reality is neurological decline. We watch milestones stall or disappear. We worry about skills being lost. We live with the fear that time is not on our side.

This disease is progressive. It does not pause while policies are debated.

Current Approved Treatments & Limitations:

There is an FDA-approved enzyme replacement therapy (ERT) for Hunter syndrome called Elaprase. It supports organ function, but it does not treat the brain.
For children with the severe form of Hunter syndrome, the unmet medical need is urgent. The neurological component continues to progress. Families are left knowing that while we are fighting for our children's bodies, their cognitive function may still decline.

There are gene therapies and brain-penetrating treatments in development, treatments that could change the trajectory of this disease entirely.

But they are not yet available.

Regulatory Challenges and Delays:

Our community is facing regulatory uncertainty, requests for additional data, and delays in trial progression and approvals, even when there is more than a decade of research behind some of these therapies.

Rare disease families understand the need for safety and evidence. We want safe treatments.

But rare diseases, by definition, have small patient populations. Waiting for perfect data in a small population can mean waiting while children lose skills, mobility, and cognitive function.

Some therapies have been:

- Delayed due to requests for more data
- Slowed by disagreements about endpoints
- Limited by narrow eligibility criteria
- Stalled after promising trial phases

For a progressive pediatric neurodegenerative disease, delays are not neutral.
They are not administrative. They are life-altering.

What Time Means to Us:

Time means everything. In six months, a child with neuronopathic Hunter syndrome can:

- Lose words
- Lose attention span
- Lose motor coordination
- Develop worsening behavioral symptoms
- Experience further cognitive decline

In one year, the difference can be profound. Regulatory delays are not abstract for us.

It is measured in:

- Words not spoken
- Skills not gained
- Memories that may fade
- Opportunities missed

Every month matters. Every quarter matters. Every year matters.

When a treatment that could potentially protect the brain is delayed, families like mine are left wondering if our children will still qualify or still benefit by the time it becomes available.

For rare disease families, time is not theoretical.

Time is neurological function. Time is mobility. Time is independence. Time is life expectancy. Time is our child's future.

And unlike policy timelines, our children's disease does not wait.

[New submission]

Dear Senate Special Committee:

Watching a child progress from being healthy, playful, smiling, and basking in love, to becoming immobile, requiring a feeding tube, multiple surgeries, or possibly even a tracheostomy, is heartbreaking for any parent, relative, or friend.

Our family has witnessed this with Hunter Syndrome in a beloved family group, as one child passed away at the age of 13 years. Another family member is affected by this disease.

[New submission]

I represent the Hunter syndrome community through both professional clinical experience and personal relationships with families affected by this devastating disease.

Hunter syndrome (MPS II) is a progressive, life-limiting disorder that robs people of their health, independence, and often their lives. Even with currently available treatments, many patients endure ongoing neurological decline, chronic pain, frequent infections, cardiac and orthopedic complications, and an immense treatment burden requiring lifelong medical care. Families live with the knowledge that existing therapies slow, but do not stop the disease from relentlessly damaging multiple organ systems.

A gene therapy that addresses the underlying cause of the disease represents more than incremental improvement, it offers the possibility of preserving cognitive function, preventing irreversible organ damage, reducing suffering, and fundamentally altering the trajectory of a child's life. For newborns and young patients especially, time is critical, as every day without effective intervention allows permanent harm to accumulate.

I urge the FDA to engage experts with deep, practical experience in Hunter syndrome, mucopolysaccharidoses (MPS), and other rare genetic disorders when evaluating investigational gene therapies. Rare disease trials inherently face small patient populations, clinical heterogeneity, and complex, multi-system manifestations. These realities should inform regulatory flexibility instead of becoming barriers to progress.

When evidence demonstrates meaningful improvement in quality of life, reduced disease burden, and strong support from informed patients and families, approval should not be delayed absent compelling safety concerns. For rare, progressive pediatric diseases, regulatory timelines have real human consequences.

Approving gene therapy in a timely manner could mean fewer hospitalizations, less joint pain and disability, reduced cardiac and respiratory complications, preserved cognitive abilities, and ultimately longer, fuller lives. It could also significantly reduce the long-term healthcare burden associated with unmanaged disease progression.

For families facing Hunter syndrome, time is precious and irreplaceable. I ask the FDA to act with urgency, scientific rigor, and compassion to ensure that promising gene therapies are evaluated efficiently and made available without unnecessary delay.

[New submission]

I am representing the Hunter syndrome (Mucopolysaccharidosis Type II) community, a rare genetic disorder that primarily affects young boys and progressively damages nearly every organ system. Children with Hunter syndrome are unable to properly break down certain complex sugars, leading to their buildup in the body and causing symptoms such as developmental delays, breathing and heart problems, joint stiffness, hearing loss, and, in severe cases, neurological decline. While enzyme replacement therapy is available and can help manage some physical symptoms, it cannot fully stop disease progression or effectively treat the neurological impacts, leaving families with significant unmet medical needs. Access to treatments and emerging therapies has been challenged by the rarity of the disease, the difficulty of conducting large clinical trials, and regulatory hurdles that can delay approval of potentially life-extending options. For families, this means navigating a devastating illness with limited tools, racing against time while hoping for advances that may come too late for their loved ones. I'm hopeful that signing this will help my friends in need.

[New submission]

Hunter Syndrome is a rare disease, and watching a child digress in their physical and mental health is heart-wrenching for any family member, or family friend. We saw our daughter in law's younger brother die from this disease, at age 13 years, and knew of his disease progression.

Knowing that there is treatment with the potential to stop the ongoing disease process is a gift for every family member and friend affected by this disease, but not being able to access the needed medication/treatment is beyond our ability to comprehend. Watching an infant,

a young child, a teenager, or a young adult progress to death from irreversible changes in their body is something no parent should have to witness.

Please, please consider allowing the treatment, a one-time gene therapy for Hunter Syndrome, to receive FDA approval. Another family member, a young child over one year of age, is also affected by this disease and desperately needs the treatment that is available, the treatment that could be life changing. Please do not allow another young baby to progress to not being able to walk, talk, eat normally, or to end up with developmental delays, severe cognitive impairment, continued respiratory infections, and progress to needing 24-hour a day total care. Thank you.

[New submission]

I have two sons affected by a rare genetic disease called Hunter Syndrome (Mucopolysaccharidosis Type II, MPS II). It's a systemic disease that affects every tissue and organ, which causes physical disabilities, developmental delays, and eventually leads to organ failures and death in early adulthood. With only about 400 diagnosed patients in the entire country, virtually no one knows about the disease, including many of their doctors. The standard therapy can only partially slow down the disease's progression in physical symptoms and have no effect on developmental delays. There was a clinical trial that was actively recruiting patients shortly after their diagnosis in 2011. However, the stringent enrollment criteria delayed their eligibility for a few years due to impractical trial design, and the endpoints were destined to fail - cognitive improvements are impossible after the onset of cognitive damages. Eventually, they were enrolled but all too late. They are currently both intellectually disabled and are in palliative care. As much as we love them, they present a heavy burden to our family in terms of daily living, financially, physically, and mentally.

In the past 15 years since my sons' diagnosis, science has made tremendous progress. New clinical trials include gene therapy and enzymes that can enter the brain and address brain development symptoms. Our community has engaged in multiple ways with the FDA to communicate what's important to the patient families, and the scientific community has provided undisputable evidence that the surrogate biomarker called Heparan Sulfate is clearly indicative of treatment effectiveness. Unfortunately, the FDA remained stuck in time and refused to grow with science. Their continued skepticism against surrogate biomarkers shows their arrogance and close-mindedness. The members of the current leadership team at the FDA are not fulfilling their duties to the American taxpayers and walked back their stance on accelerated approval based on surrogate biomarkers and chose to ignore the patient community's voice and scientific evidence.

We need the FDA to be held accountable for its actions. We need the FDA to listen to the patient community and understand that clinical science is helping patients at the end of the day, and the endpoints should be aligned with what the patients and families value with most. We need the FDA to be impartial and be science driven, rather than letting the biased view (or even political agenda) of individuals in leadership position to dominate its opinions and review results. It's too late for my sons, but new patients suffering from rare genetic diseases are born every single moment. We need actions from the FDA to give them a better start of life when the evidence of clinical trials demonstrates effectiveness.

[New submission]

Thirty years ago, we received a brutal diagnosis from a geneticist in Boston that our son had Hunter Syndrome. He was 2 years old. We had never heard of this rare disease and were basically told to "love him while you have him". At the time there were no treatments or therapies for MPS II. Our pediatrician read the diagnosis from a medical textbook and we began a journey we never expected to take.

When he was 8 years old, he became one of the youngest patients to be enrolled in an enzyme replacement therapy clinical trial. While this double blind, placebo study went on, we began to see transformations in him. We knew he was getting the "real deal". His liver and spleen reduced greatly as his stamina increased. We made the trip twice a week for three years...within a few months we were chasing him through the airport terminal as he was off on his own amazing race.

Our son has been getting ERT for over 25 years now. He graduated from college, got his license and landed a job at a pharmaceutical company. All milestones we never dreamed we would see.

The current therapy for Hunter Syndrome does not cross the blood brain barrier however, and we have seen the progression of this disease continue as time has gone on.

He was in a gene editing trial in 2019 that unfortunately did not give us the benefits he'd hoped for. At our most recent visit, we were told of a potential drug that was close to FDA approval that could reach his brain and spine. Those hopes have been dashed away, however, with regulatory hiccups at the FDA. We don't have time on our hands to wait for these approvals as it seems the FDA keeps kicking the can down the road.

We are forever grateful for the research, funding, doctors and families who have given their lives to making a better life for our Hunter boys. We seem to be at a crossroad with recent stalls. We implore your Committee to do whatever can be done to continue to give us hope for the future of our children and keep our dreams alive.

MPS I

My son is now seven years old. He was diagnosed with MPS1, a rare lysosomal storage disorder at the age of nine days. Fortunately, for MPS1, there is an approved treatment: a bone marrow transplant. He underwent chemotherapy and transplant at the unimaginable age of 3 months old. The complications of transplant landed him in the ICU, and he was on a ventilator for almost 2 weeks. Many times, I thought we would lose him.

So many kids do not survive the bone marrow transplant. Even if a transplant is successful, it is not a cure. Patients with MPS1 still undergo countless risky surgeries due to the effects on the skeletal system, heart, and lungs. Children often suffer tremendous developmental delays when diagnosis and treatment occur late. These delays affect their ability to be productive members of society as well.

We need better treatments. Better treatments = less complications. A large percentage of patients with MPS1 are covered under Medicaid insurance due to the debilitation of this disease.

Cost of care rises when better forms of treatment are delayed. Within just one year, my son went from needing no surgical intervention to requiring major hip surgery, back surgery, and now possible carpal tunnel surgery as well. All within a single year. This is because, despite the best treatment, MPS1 continues to progress quickly.

Delays in approval mean suffering for innocent children. Parents unable to work because of worsening symptoms in their children, excess surgeries requiring more parental care, and unfortunately at times, death of an innocent child. Children with MPS1 and other debilitating rare diseases deserve better.

[New submission]

My son was diagnosed with MPS1 Hurler Syndrome shortly after birth as a result of newborn screening - a testament to what scientific advancements can do to change the trajectory of a child with a rare illness if we are willing to commit to harnessing our scientific potential as a nation.

Due to early diagnosis, my son was able to almost immediately take advantage of the two available FDA approved treatments - he started Aldurazyme enzyme replacement therapy at just 5 weeks old and received a bone marrow transplant the day he turned 4 months old.

Each and every day I feel two incredibly opposing feelings about my son's life. I am eternally grateful for the scientists, advocates, and doctors who fought to develop treatments for

MPS1 despite it being an ultra-rare condition. His bone marrow transplant will significantly slow the progression of his disease, and ERT, while not protective of his brain, does help to shield his body from some of the effects of his disease. Neither is anywhere near a cure, but I can't help but be grateful he has an opportunity to receive treatment at all. I know all too well this is the exception and not the rule when it comes to rare diseases.

And in the same breath that I thank God for the ability to have some access to treatment to support for my son, I feel anger, frustration, and confusion around why, despite BETTER and SAFER treatments being technically possible (and even in development) on the part of biotech companies and researchers, we were still relegated to a the newest available treatment that, at the time of his birth, was approved by the FDA 20 years ago. How could we not have advanced by this point? Who are we as a country if this is how we support our most vulnerable children?

I lament the lost time I had with my newborn while we left his father and sister behind to live in the hospital and isolate for months to undergo his bone marrow transplant, quite literally risking his life for a shot to extend his life. His transplant course, like most children's courses, was not without complications and came with forever life-altering consequences. He survived, thank God, but the impact emotionally and physically will be with him forever. To add insult to injury, his donor percentage nearly 3 years post-transplant is failing. We're facing the possibility of a second transplant, a second round of isolation and toxicity, and a second test of his ability to survive being brought to the brink of death all because the FDA is limiting our ability to access safer and more effective treatments.

Despite obvious evidence that OTL-101 is both more effective and safer than traditional transplant, my son is not eligible as the FDA requires an unethical, double-blind study against traditional transplant that has ALREADY taken the life of a fellow MPS1 child we met while undergoing the transplant process when they were randomized into the BMT arm. We have decades of data to show us what happens when you put a child through a bone marrow transplant. We know the risks involved, and we have extensive documentation of its shortcomings when it comes to preserving the life and abilities of children with MPS1. To pretend we need a randomized trial is insane, and to withhold OTL-101 while newborns and toddlers continue going through BMT is just plain cruel.

Despite incredible potential for B-cell therapies like those developed by Immunosoft which require absolutely no toxic conditioning and are re-dosable, the FDA will not allow children to benefit from these clinical trials.

We could avoid a second transplant if this option were open to us but, due to his age, it's not.

And beyond that, we've watched as MPS treatments have been delayed or denied in this last year in our immediate and wider MPS community for absolutely unwarranted reasons (RGX-121, RGX-111, Tividenofusp alfa, UX111). In many cases these diseases have NO approved treatments whatsoever. We are killing children in full conscious when hope is dangling right in front of these families!

How can I possibly be standing here, watching my almost three-year-old walk, talk, play, run, and jump like any other toddler with a death sentence over his head? How can I possibly be standing here facing another transplant that could take him from us? How can I possibly be standing here knowing he will be subjected to dangerous and painful surgeries, all while his body slowly continues to poison itself until his eyes can no longer see through the clouding, his body can no longer hold him up, and his brain can no longer communicate the way it does in this moment, until eventually, his entire body can no longer sustain life?

How is that the case? In large part it's because the FDA is communicating to the researchers and biotech companies that could save my child's life that they are not valued. That regulatory technicalities matter more than children without hope. That death is a sacrifice we have to make so we can have just a little more time to sort through the research on biomarkers that's already been approved.

And if people don't matter to you, maybe dollars do. Newborn screening coupled with a definitive treatment could protect these children from years of Medicaid funded treatment, extensive disability, and an inability to contribute financially to society. If that's where your line is drawn, then it's not too late for my son. Get him the treatment he deserves, and he'll repay this society beyond your wildest dreams. At this moment he's still young enough that his potential is unlimited. Make him wait a few more years and you'll lose your future dollars while I lose my son.

What are you waiting for!? The choice is yours.

Sanfilippo Syndrome

My daughter has MPS Sanfilippo Syndrome. She is 4 years old and was diagnosed at age 2.5 with a rare genetic condition (1 in 70,000 births) causing debilitating effects on the body. But the most marked effect is progressive and fatal cognitive decline. Children with Sanfilippo will suffer Brain Damage Leading to loss of all their functional skills and is often times characterized as childhood Alzheimer's. Research has shown that brain damage becomes irreversible by the age of 3. There is currently no approved treatment.

Recently, Ultragenyx received an incomplete Response Letter from the FDA related to their BLA resubmission. Tough news after a year of unnecessary delays. The data show that the treatment is successful, and the earlier kids have access to it, the better. Time is crucial, and we pray that the FDA will keep their commitment to accelerate approvals."

[New submission]

My two daughters have MPS IIIB. There is no FDA cure or approved treatments. The rare disease community is urgently asking for your help and flexibility approving gene therapy and enzyme therapy for these kids. Their neurodegenerative diseases do not pause; it continues to cause regression and cognitive decline.

[New submission]

From a parent to a child with Sanfilippo Syndrome and an advocate with the Cure Sanfilippo Foundation

A delay of 6 to 12 months means losing parts of my son that I will never get back. Every single second matters for him. Not months. Not weeks. Seconds.

I have already watched him lose so much in such a short period of time. He used to sing in the car. Now there is silence. He used to talk. Now the only word he consistently says is "daddy." And I live with the fear every single day that one day, he will stop saying it. I won't know when the last time was. I won't be prepared for it. It will just be gone. That is what time does to children like mine.

A delay of 6 to 12 months means 6 to 12 more months of watching my son slip away from me. It means more moments where he seems lost. More moments where he cannot communicate. More moments where I see pieces of him disappear. It means his brothers losing their little brother piece by piece. It means our family living with the pain of watching someone we love fade in front of our eyes while we wait for help that may come too late. Every second matters

because every second is time I may never get back with my son as he is today. A delay is not just time on paper. It is time that this disease takes my son from me.

A better quality of life means I don't have to keep watching my son drift further away while I stand there unable to reach him. There are moments now where I look into my son's eyes, and I don't know if he fully understands or recognizes what's happening around him. I talk to him, and I don't know if he can respond the way he wants to. I see him struggling, and I see the frustration in him. And as his dad, there is nothing more painful than knowing he's still in there, but I can't fully reach him anymore.

A better quality of life means he doesn't have to live in that place of confusion. It means he can feel comfort instead of frustration. It means he can feel safe, peaceful, and connected to us instead of slowly slipping further away. It means his brothers don't have to grow up watching their little brother lose more of himself. They deserve to have real moments with him. They deserve to hear his voice, see his smile, and feel like their brother is still there with them—not just physically, but emotionally.

For our family, it would mean relief from the constant heartbreak. Right now, every day feels heavy. Every day feels like we are waiting for the next piece of him to be taken. A better quality of life would mean fewer of those losses. It would mean more peace for him and more peace for all of us. It would mean he could live with comfort, dignity, and love, without this disease continuing to take more from him than it already has. He has already lost so much. A better quality of life means he doesn't have to lose everything.

I need you to understand that this is my son's life, and I am the one who has to watch him die while waiting for decisions I have no control over. Every day that passes, Sanfilippo Syndrome takes more from him. I am not reading about it. I am living it. I see it in his silence where his voice used to be. I see it when he seems lost. I see it when he struggles with things that once came naturally to him. And while this is happening, I am forced to wait. Waiting for people who have never met my son, who will never know his laugh, his smile, or the way he says "daddy," to decide whether he gets a chance.

You are not the one who has to sit down with his brother, who is in kindergarten, and try to explain why his little brother is changing. You are not the one who has to answer questions no parent should ever have to answer. You are not the one who has to watch the confusion and sadness in his brother's eyes while trying to stay strong yourself. That choice should not belong to someone who has never looked into my son's eyes. That choice should belong to me.

I am his father. I am the one who loves him. I am the one who has to hold him, comfort him, and live with whatever happens. I am willing to accept the risks if it means giving him a chance. I am willing to fight for him with everything I have. What I am not willing to do is stand by and watch him die while waiting for permission to try to help him.

You have the power to give families like mine a chance. You have the power to move with urgency. You have the power to recognize that time is something children like my son do not have.

Please do not make families like mine wait while our children disappear in front of us. He deserves the chance to fight for his life. Because while you are deciding, I am watching my son disappear.

[New submission]

For me and my child with Sanfilippo Syndrome, time means loss of words, increasing headaches and aggression, loss of mobility, worsening gi issues, toileting accidents. His personality is fading with every passing day.

A chance at a better quality of life would mean the ability to live past his teenage years, retention of mobility and ability to continue eating all his favorite foods by mouth, remembering all the family members he loves by name.

When it comes to risk from therapeutic options, there are not many risks worse than this terrible progressive disease itself. We are hoping every day to hear that the therapy that could save Merrick's life will finally be accessible to him. We are willing to move to raise money, to go into debt—anything to get him this treatment.

We wish the FDA felt the same urgency to save my son's life that we do. Every day we hope to hear good news that could turn the tide for this disease. With AI tools, science breakthroughs will continue to progress more and more rapidly, and the FDA needs to figure out how to speed up their own processes for the good of everyone.

Duchenne Muscular Dystrophy

I am the mother of a 25-year-old young man with Duchenne muscular dystrophy. Diagnosed at 18 months, we were told that there was nothing they could do. No treatments other than steroids, and to just "open the door and let them be."

We did just that. My son has thrived. He has had amazing opportunities to be in the school band, played in a steel drum band, scouts, achieving Eagle Scout rank, graduating from college with a degree in hospitality management. We are extremely blessed to live in such a community that is so caring and inclusive.

If you are not aware, Duchenne is caused by the lack of dystrophin production in muscles, caused by a mutation in their DNA. What you may not be aware of is that there are many different mutations. My son has a duplication of exons 8-12. There are no research or trials for him.

I'm asking you to review the current legislation and reevaluate current guidelines for trials and research. For example, a trial that produces only a small amount of dystrophin for our boys that can maintain their pulmonary or cardiovascular health may not seem like a significant endpoint for the trial, but for these men/boys it can be life altering.

Our family appreciates your time and consideration of rare disease awareness. Just know that we are out here, as members of the community you serve. Hopefully thriving and always aware of what your voice could mean when combined with ours.

[New submission]

Submitted by parent

Our son has Duchenne muscular dystrophy (DMD), which is the number one genetic killer of boys worldwide, affecting 1/300 births boys with the disease, girls unknowingly carriers. This is the largest gene in the body with the highest spontaneous mutation rate, like in our case, and over 30% of all cases. Boys with DMD don’t have the structural protein dystrophin, so their muscles begin to deteriorate affecting all of their muscles including his heart, diaphragm, and eventually affecting all their muscles.

Boys with DMD usually die in their teens or early 20s. After over 2 decades of my son participating in a clinical trial for PTC 124, Ataluren, the FDA decided not to approve it this past week, even though it is safe and had better endpoints than Elevidys, the gene therapy drug the FDA approved last year even though several boys had died. Ataluren allowed my son to graduate from college, live on his own 5 hours from his family, work for the Governor, and as a HS social worker. At 34 1/2 he makes the 17 hour trek each year to a children’s hospital where he is the oldest DMD patient. This clinic has over 800 DMD patients.

There is no question this drug is responsible for him to breathe on his own. Unfortunately, there are no other drugs for a premature stop codon mutation, his mutation. Without this drug, he will decline and die. This oral drug is safe with hundreds who have been on the drug. This drug is approved in over 50 countries.

There are very different evaluation criteria at CDER, where Ataluren was evaluated, than CBER, where Elevidys was granted approval.

By denying this drug for our son, the FDA is creating a type II error, and boys will die needlessly.

Submitted by other family member

I am writing on behalf of the young men affected by muscular dystrophy, including my brother-in-law. The FDA is denying access to the drug Ataluren, and I am requesting they reconsider. This drug has allowed him to pursue a career as a social worker, live independently for a portion of time, and has given our family the beautiful gift of having him as “Uncle” to our two young sons who ADORE him. This medication has helped him to continue breathing on his own—a privilege many of us take for granted every day. DMD has taken so many things from my brother-in-law. Please don’t take the one thing we have that has helped him. Please reconsider continued access for all boys with muscular dystrophy as they too deserve the same chance at a fulfilling life.

Submitted by cousin

My cousin was diagnosed with Duchenne's muscular dystrophy when we were both young. I still remember seeing the heartbreak my aunt and uncle went through upon learning about his future as an individual diagnosed with the disease. Throughout nearly his entire life, he has persevered through trial after trial so that he could live as normal a life as possible. In spite of it all, he has been able to graduate from college with a master's degree in social work; giving him the opportunity to share his unique life experience with high school students.

None of this would have been possible without Ataluren. My cousin would not have survived or succeeded for the past 34 years; this is the only drug currently available that treats the decline associated with a premature stop codon.

Real-world data supports this drug's effectiveness over the past 20 years. By choosing to deny Ataluren for use by patients with Duchenne muscular dystrophy you are condemning my cousin and many others. I am pleading with you to reconsider this decision.

Submitted by cousin

My cousin has Duchenne muscular dystrophy. After over two decades participating in a clinical trial for Ataluren PTC 124, the FDA decided not to approve it this past week. This drug allowed him to graduate from college, live on his own, five hours from his family, work for the Governor, and as a HS social worker. At 34 1/2 he makes the 17 hour trek each year to a Children's Hospital where he is one of the oldest with DMD out of 800 patients.

There is no question this drug is responsible for him to breathe on his own. It is a travesty that the FDA considers that ad hoc data and not relevant. Other INVASIVE drugs in which boys have died with a similar p-value have been approved. This FDA ruling will have adverse effects on future DMD clinical trials. Unfortunately, there are no other drugs for a premature stop codon mutation. Without this drug, my cousin's health will decline. This is a safe oral drug approved in 50 countries.

Please consider the approval of this drug. It will extend the life of my cousin and maintain his improved quality of life. He has spent his life in service of others. He is an amazing man. He and others just like him deserve the opportunity to benefit from this drug. I humbly beg you to get approval for Ataluren.

Submitted by aunt and uncle

Our nephew was diagnosed with DMD as a preschooler and accepted into a trial for the drug Ataluren. It has been a life changing drug that enabled him to graduate from college with a master's degree in social work and share his skills while working with high school students. Without Ataluren, Jacob would not have survived or succeeded for the past 34 years. This is the only drug currently available that treats the decline associated with a premature stop codon.

We ask that you please reconsider the FDA denial of Ataluren for use by patients with DMD. Real-world data—our nephew' life story—supports this drug's effectiveness over the past 20 years.

Submitted by cousin

My amazing, intelligent, and funny cousin, was diagnosed with DMD as a preschooler and accepted into a trial for the drug Ataluren. It has been a life changing drug that enabled him to graduate from college with a master's degree in social work and share his skills while working with high school students. Without Ataluren, he would not have survived or

succeeded for the past 34 years. This is the only drug currently available that treats the decline associated with a premature stop codon.

I ask that you PLEASE reconsider the FDA denial of Ataluren for use by patients with DMD. Real-world data—my cousin's life story—supports this drug's effectiveness over the past 20 years.

[New submission]

Submitted by parent

We are writing today to tell you about our son's experience with Ataluren (Translarna) manufactured by PTC Therapeutics. This oral small-molecule treatment allows for stop-codon read through to produce dystrophin in patients, like our son, with nonsense mutations which leads to a diagnosis of Duchenne muscular dystrophy. Recently, the FDA was unable to resolve differences in data interpretations with the manufacturer, causing the new drug application to be withdrawn.

Duchenne muscular dystrophy is a genetic disorder that causes progressive muscle weakness and primarily affects boys. It is caused by a mutation in the dystrophin gene, which normally produces a protein that helps protect and strengthen muscle cells. Without sufficient dystrophin, muscle fibers become damaged and are gradually replaced with fat and scar tissue. Diagnosis usually occurs in early childhood, between the ages of 2 and 5. As the disease progresses, weakness spreads to more muscles, and many require a wheelchair during their early teenage years. Over time, Duchenne affects the heart and breathing muscles. Although there is currently no cure, treatments such as corticosteroids, heart and respiratory care, and newer gene-targeted therapies can help slow progression and improve quality of life.

Our son was diagnosed with Duchenne in November 2013, four months after his first birthday. We knew shortly after the time of diagnosis that Ataluren was the medication he needed for his type of mutation. Unfortunately, at the time they were only enrolling boys over the age of 7 for any trials. We were told that an FDA approval was hopefully around the corner. Three years came and went, and while there was no approval, PTC was starting a safety study for children under the age of five. We immediately started researching sites and were selected to enroll. Eleven days before his fourth birthday, he received his first dose of Ataluren. Within the first six weeks we saw a positive change in his strength and mobility and haven't looked back since.

Our son is still ambulatory at the age of 13.7 years. In the past year, his Cardiac MRI, Pulmonary Function tests, and bone density scan results were all within a normal range. All his physicians marvel at how incredible he is doing compared to his peers of the same age and diagnosis. They are all very encouraged about how Ataluren has delayed the onset of the typical pathology of Duchenne. Other than a medical stroller and mobility scooter (to use long walking distances, like at an airport or theme park), he has no assistive devices. Remarkably, our son participates in PE for 60 minutes 5 days a week. While some of the activities are modified for his safety (no pull-ups, push-ups, etc.), he is still able to claim that PE is his favorite time of the day. A few months ago, he came home excited to share that while playing flag football he caught a pass and ran 20 yards to score a touchdown!

When our son was diagnosed in 2013, we were told by one neurologist to go home and give him the best life possible because by the age of 7, his mobility would start to decline. Another neurologist told us not to give up hope that there were so many things in the pipeline for Duchenne, so much more than there had ever been before. During what was one of the most terrifying times in our lives we are grateful for that second neurologist's advice. We knew there was something out there to help slow this terrible disease. Ataluren was that "something" for our son. We have no doubt in our minds that Ataluren and the age at which he was able to be first dosed is the reason he is still running and jumping today. It is the reason his heart, lungs, and bones are still healthy. The safety profile of Ataluren is also a major benefit of the drug. He has never suffered any side effects while taking this drug for the last 9 years.

In addition to our own observations and experiences, results from the most recent clinical trial show a 3.5-year delay in loss of ambulation. Ataluren is also found to be safe and generally well tolerated. The most recently approved gene therapy Elevidys has shown promise biologically but its impact on motor function is still being investigated and safety concerns have led to liver complications and even death. This makes the denial of a new drug application for Ataluren even more confusing and frustrating. Our son does not qualify for Elevidys because his mutation is on exon 9 which is part of the exclusion criteria. Patients receiving Elevidys with mutations in exons 8 or 9 have the risk of severe immune-mediated myositis. This is a severe, life-threatening autoimmune reaction where the body's T cells mistakenly attack the skeletal muscle.

Currently, my son's access to a safe drug, Ataluren, is being denied. However, a drug that would potentially have a life-threatening outcome is approved. With the FDA's decision, he is in a lose-lose situation. There are no other options for him besides the standard of care.

Without Ataluren, his life will change dramatically. He will require more assistance with everyday tasks such as dressing, bathing, and eating. His favorite activities, like playing basketball in our driveway and football with his friends at school, will be stolen from him. His easily accessible middle school for an ambulatory teen will now be faced with daily obstacles. His future depends on a treatment that is safe and helps preserve his function. Ataluren is that treatment. Please help our son stay on Ataluren; his life depends on it.

Thank you.

Submitted by grandparent

I would like to address this committee about my feelings and opinions about my grandson, who has nonsense mutation Duchene muscular dystrophy. This is a very rare form of this terrible disease and he has been on Ataluren since age 4 with remarkable results. I am a retired physician.

He was diagnosed at an early age with Duchene muscular dystrophy and was not able to ambulate and perform other tasks requiring muscle strength. His treating pediatric neurologist informed his parents that he would be in a wheelchair by age 6. Since beginning Ataluren, not only has he been able to ambulate and perform normal tasks for his age (currently almost 14), but he attends junior high school and only uses a wheelchair for long trips. His quality of life has vastly improved since on Ataluren from what had been predicted.

The FDA has denied approval for this drug despite evidence from clinicians that it has definitely helped children with this rare genomic form of this disease. It is my opinion that the trial did not reach statistical significance because of the small number of participants.

Regulatory delay in the production and dispensing of Ataluren will definitely, in my medical opinion, quickly hasten the worsening of his condition, which will ultimately lead to an earlier demise. This would be unforgivable!

Submitted by grandparents

Our grandson was diagnosed with Duchenne muscular dystrophy at just two years old. We were told to expect a steady and devastating loss of strength, independence, and ultimately, life. Today, at 13, he continues to defy those expectations.

For over nine years, he has been taking Ataluren. His physicians consistently express surprise at his strength and function compared to untreated boys his age. He remains ambulatory, relying only partially on assistive devices during fatigue, and maintains strong pulmonary function and normal bone density. His doctors firmly believe Ataluren is delaying the progression of his disease.

Clinical trials involving nearly 800 patients have shown that Ataluren can delay loss of ambulation by approximately four years and slow pulmonary decline by two years. In a fatal disease with limited treatment options, those years represent precious time — time walking, breathing, and living with greater independence. The drug's safety profile has been remarkably favorable, and it is approved in more than 50 countries.

We have witnessed its benefit not as an abstract data point, but in the life of our grandson. Without access to this treatment, our grandson's decline will accelerate. The physical losses are inevitable in Duchenne — but the timing matters. Every preserved year of strength profoundly affects his quality of life and emotional well-being.

We respectfully urge reconsideration of approval for Ataluren and greater flexibility in evaluating therapies for rare, fatal diseases with significant unmet need. For families like ours, this is not policy — it is time, hope, and the chance for our grandson to remain a little stronger for a little longer.

Our grandson is not a statistic. He is a child fighting a relentless disease. We are asking for the opportunity to keep fighting with him.

Submitted by family friend

My friend's 13-year-old son has Duchenne muscular dystrophy. His mom and I are very close, we are co-workers and neighbors, and our kids are close in age. I'll never forget my daughter hitting her physical milestones and her son was not. He was diagnosed with DMD at one. My friend was told he would be in a wheelchair before the end of elementary school. She sought alternative treatments, and he was put on a study for Ataluren.

He began hitting his milestones, he played chase with the other kids, and he continued to thrive. Now at 13, he's still standing and thriving with the help of this drug, walking the halls of middle school with his peers. None of this was expected, but Ataluren is the cause. Please approve it for his special case. He has a bright future ahead of him and deserves to receive the treatment he needs. Ataluren is a necessity for him. Thank you for your time.

Submitted by teacher

The kids call me Coach. I have been my student's physical education teacher since he was 5 years old. He suffers from DMD (Duchenne muscular dystrophy). He is now in his 8th grade year, and I am witnessing each day how the drug Ataluren has halted the progression of this muscular disorder in this young man. Not only is he mobile, he is active! I was acutely aware to expect him to be wheelchair bound by the end of his elementary school days. I am here to tell you that I have not seen a decline in his physical abilities at all. Not one day! I have only seen improvements and gains in his physicality and motor skills.

We have recently learned that Translarna, the maker of the remarkable and effective medicine Ataluren, which my student takes, has ceased production because of FDA non-approval. This is outrageous. It works! Please come to our school and watch him run and catch a football thrown at him from 30 yards! And then watch him return the throw on target with a perfect spiral EVERY TIME!! More importantly, come and watch him WALK to and from class each day in the hallways with his friends like a normal kid. Please! Please come and see the reality of this kind of data. Those affected by this are too few, apparently, to obtain enough data for approval. This too is outrageous! Change your scope, please! This is my plea to those who make the big important decisions that affect the lives of everyone in this community... come and see us! We will show you!

Submitted by teacher

I am a teacher and one of my former students has DMD. He's now 13. Through his treatment with the drug Ataluren, he is defying the natural progression of this disease. He can still do PE class. He can throw and catch a ball and do things that all 13-year-olds should be able to do. My heart breaks at the thought of this life changing medication is now gone. That all of the progress he has been able to have is now threatened. Duchenne may be a rare disease but the boys and men who have it matter. They deserve to be able to have medication that makes their life better.

[New submission]

I am a 32-year-old man living with nonsense mutation Duchenne muscular dystrophy (nmDMD), a rare and progressive genetic disease. I have no financial relationship with the manufacturer of Ataluren and receive no compensation for sharing my story.

I was diagnosed with Duchenne muscular dystrophy just before my first birthday. Duchenne gradually weakens every muscle in the body–first the legs, then the arms, then the muscles that support breathing and heart function. It is the most common fatal genetic childhood disorder, affecting 1 in every 5,000 boys born each year. Currently, there is no cure. Steroids may slow progression but do not stop the disease and can have significant side effects. Fewer than half of individuals with Duchenne live beyond their late twenties.

Throughout my childhood, this illness shaped my daily reality. I couldn't run and jump with my classmates. After walking or standing for ten minutes, my muscles would become so tight that I would collapse into a chair, unable to bend my legs. Night splints, orthotics, physical therapy, and occupational therapy were part of my life for as long as I can remember. By sixth grade, I could only reach my bedroom by crawling up the stairs. My parents renovated our home to give me a ground-floor bedroom and a wheelchair accessible bathroom. By the end of grade school, I relied on a motorized scooter to navigate much of my world.

In ninth grade, I enrolled in the Translarna (ataluren) 007 study. My life changed in ways that my family and I never imagined possible.

After starting the medication, I began gaining strength and endurance. Though I had taken steroids for many years, they only really slowed the progression of my disease and caused negative side effects like weight gain. It wasn't until starting Translarna (ataluren) that I noticed that I was getting stronger. Halfway through high school, I stopped using my motorized scooter. My senior year, I completed a mile-long hike with my family–something that, just a few years earlier, had been unimaginable. After high school, I began exercising regularly, lost 35 pounds, and completed a 5K race. Today, I am fully ambulatory and live independently with my dog, a fifty-pound labradoodle whom I am physically strong enough to manage and routinely walk for a mile or more. I have experienced no negative side effects from medication.

In 2017, I traveled to Washington, D.C. to testify before the FDA Advisory Committee Meeting regarding ataluren. Ultimately, the FDA did not approve the medication, citing insufficient evidence of effectiveness. But in rare diseases like mine, the measures used in clinical trials do not always capture meaningful improvements in real people's lives. For me, this medication has been truly life changing.

Now, after further regulatory setbacks, the manufacturer of ataluren has stopped pursuing FDA approval. As a result, access for patients like me in the United States is uncertain. I recognize that I may be an outlier, but the benefits of this medication are undeniable. I worry what would happen to me if I'm unable to continue taking it.

Time is not abstract in Duchenne muscular dystrophy. Six months can mean the difference between walking and needing a wheelchair. One year can mean losing the ability to climb stairs, to stand from a chair, or to lift your arms. Once lost, those functions do not return. Delay is not neutral – it is decline. When I was little, I expected that having muscular dystrophy meant I would steadily lose the ability to walk, to breathe, and, ultimately, to live. I watched other boys in my muscular dystrophy support group follow that path. After starting this medication, my life trajectory changed. Instead of planning for decline, I began building a future. Instead of losing independence, I gained strength and stability.

My older brother is a physician who understands the natural course of Duchenne. He has watched my trajectory closely and has seen how, after beginning this treatment, my course diverged from what medicine predicts for this disease. He says this change is extraordinary.

I respectfully ask the Committee to consider how regulatory processes could better take into account the real-world experiences of patients with rare, progressive diseases. For patients

like me, access to treatment is not theoretical–it can mean the difference between walking and needing a wheelchair, between having hope and facing inevitable decline.

For individuals with Duchenne muscular dystrophy, time is muscle. And muscles, once lost, are gone forever.

[New submission]

I submit this testimony as an individual living with Duchenne muscular dystrophy, a rare muscle-wasting disease, and as a representative voice for countless Americans affected by rare illnesses and chronic conditions. Our community faces unique challenges in accessing life-changing therapies, and we urge policymakers to recognize and address these barriers.

Adding to these challenges, the FDA held the Ataluren file for approval for over 18 months and refused to include the STRIDE data in its review. Ultimately, the FDA indicated it would not review the application based on the totality of the data provided, forcing the drug company to withdraw it voluntarily. This decision has left patients and families in limbo, unable to access a therapy that has demonstrated safety and life-changing efficacy.

We are born into a world with the potential to offer us greater opportunities, yet we are too often marginalized. If the FDA is truly committed to supporting Americans with rare diseases, it must adapt its policies to recognize the unique realities of our population.

Specifically, the agency should reconsider how it evaluates drug efficacy for rare diseases, acknowledging that small cohort studies can still yield significant, life-changing results.

Without FDA approval or continued compassionate use, patients will face rapid decline, loss of essential abilities, and preventable suffering and deaths. I urge the FDA and lawmakers to act swiftly to secure ongoing access to Ataluren and similar therapies for all who need them. Hope for a better life should not be a fleeting platitude—it must be a tangible promise, backed by responsive policy and regulatory action.

Thank you for your attention and commitment to improving the lives of Americans with rare diseases.

[New submission]

My 26-year-old son has Duchenne muscular dystrophy. When he was diagnosed at age 2, life expectancy was mid-to-late to teens, but advancements in the standard of care allowed him to attend and graduate from college and survive well into his twenties.

Duchenne is all-encompassing, continuing to rob my son of his independence and agency. He lost his ability to walk at age 12. He uses a BiPAP machine to help him breathe at night. He

wakes multiple times nightly and calls me to reposition him. He lost his ability to raise his elbows and arms last year, so he is no longer able to feed himself. Recently, his hands have become weaker, making it difficult for him to use his wheelchair joystick for too long and interfering with using his computer mouse.

Although he earned a bachelor's degree in computer science, he is unable to obtain consistent employment because he is easily fatigued and needs to take frequent breaks to reposition in his chair. My son's ongoing, compounding losses in his physical abilities has led to much anxiety and depression–living at home with his mother, completely reliant on her for all daily cares–as his peers are thriving in careers, getting engaged and married, etc.

Since he was diagnosed in 2001, many therapies have entered the pipeline that potentially could benefit him. The most significant is Ataluren (originally named PTC-124 and later named Translarna when conditionally approved in Europe), developed by PTC Therapeutics. Ataluren was the first drug for Duchenne that saw clinical trials with Duchenne patients. He was in that trial in 2006. Participation required many 7-hour drives, countless blood draws and clinical testing, as well as two muscle biopsies.

Because of Duchenne's complexities and progressive nature–it affects ALL muscle groups and systems in the body–identifying data points to show the drug's efficacy has proved difficult. After FDA's initial denial, PTC continued to hone its trial design and allowed Nick to enter into an extension study, requiring additional invasive muscle biopsies and blood draws. FDA denied it again. PTC continued to offer the drug to patients in an extension study for two decades as they worked to develop the necessary data for the FDA. However, after 20 years, the FDA is still not persuaded, so PTC has ended its pursuit of approval.

Ataluren is not a miracle drug. It doesn't "cure" Duchenne. But I believe it has played a role in my son's pulmonary function. Yes, he does use a BiPAP and his cough strength has weakened; however, his rate of decline seems significantly delayed compared to others his age with Duchenne. Stopping Ataluren is devastating for him.

Obviously, PTC's struggle with approval for Ataluren is an extreme case in terms of time delay. But all time is precious with a disease like Duchenne that daily robs patients of their mobility, independence and self-direction. Clearing hurdles to drug approval is essential for ensuring maximum quality of life for our community.

Thank you for your help.

[New submission]

I am the parent of an 18-year-old son living with Duchenne muscular dystrophy (DMD). Duchenne is a rare, progressive, and fatal neuromuscular disease. I am submitting this statement to share what regulatory decisions and treatment delays mean to families like mine who live every day with the consequences.

Living with Duchenne

Duchenne causes ongoing and irreversible muscle loss. It is a genetic condition often diagnosed in early childhood due to gross motor delays. Patients gradually lose the ability to walk, then lose strength in their arms and hands, followed by declining heart and lung function. Life expectancy is significantly shortened, and quality of life steadily declines despite best supportive care.

My son was diagnosed as a young child. Over the years, we have watched him lose abilities one by one; walking, standing, climbing, lifting. Today, at 18, he depends on assistance for most daily activities. Every year brings noticeable decline. Time is not neutral in Duchenne.

Treatment Experience and Access Barriers

There is no cure for Duchenne. The few available therapies are meant to slow progression, not stop or reverse it. My son participated for five years in a clinical trial. During the trial, he tolerated the drug well and remained more stable than expected for someone with his disease progression.

After the FDA approved the therapy using the Accelerated Approval Pathway, our insurance continued to deny coverage—not because the drug was unsafe, but because my son is now older than the age group studied in the trial despite already having received the therapy for years. This is a common experience in the Duchenne community: patients who survive longer and need treatment most are often the ones who are denied or lose access.

Trial Design and the Reality of Older Patients

As boys with Duchenne get older, they are frequently excluded from clinical trials like my son, who currently does not qualify for any ongoing trials. This is not because patients cannot benefit, but because it becomes riskier and harder to show large, statistically significant changes to traditional endpoints.

What gets lost in this process is what matters most to patients and families. For my son, maintaining the ability to use the toilet on his own, feed himself, adjust his arm position and use his phone makes a real difference in his independence and dignity.

These changes may look “small” on a chart, but they are enormous in daily life.

When trials and regulatory decisions focus only on endpoints that favor younger patients, older individuals are left behind. Their benefits may be harder to measure, but they are no less real.

Regulatory Uncertainty and Its Impact

Recent regulatory actions including delayed reviews, complete response letters, and uncertainty around trial outcomes have created fear and instability for Duchenne families. Communications from the FDA regarding trials not meeting primary endpoints raise serious concerns about continued access to therapies that patients are already relying on.
These are not abstract policy decisions. Every delay has consequences. Duchenne does not wait for new trials, new endpoints, or regulatory alignment.

What Time Means for My Son

My son will graduate from high school soon and has been accepted at premier institutions to pursue his college education. In six months, my son could lose more function and struggle with basic self-care. In a year, further respiratory decline could permanently change his health and independence. These losses are irreversible, making achieving his dreams impossible.

For families like ours, regulatory delay is not just delay—it is loss of function, loss of ability, and loss of time we cannot get back.

I urge this Committee to consider regulatory approaches that better reflect the realities of progressive rare diseases: flexibility in trial design, recognition of patients' lived experience and continuity of access for patients who age beyond trial populations. Waiting for perfect data should not come at the expense of the people living with the disease today. For my son, time matters. Every decision made affects what he will still be able to do tomorrow.

[New submission]

I have a 21-year-old son who lives with progressive heart failure. He barely survived three life threatening episodes in 2025. Yet, between ICU stays, he is a thriving honor roll student who just wants to live like other college kids. Duchenne muscular dystrophy has stolen his arm and leg muscles, and now his heart and breathing muscles are in peril. Duchenne progressively

destroys skeletal, cardiac and respiratory muscles, leaving teenagers' breathing abilities weakened to the point of needing ventilators, and hearts weakened to needing pacemakers, and ultimately, stopping.

While some newer genetic therapies are showing promise in a select few younger patients with Duchenne, none have specifically rescued or stopped progression in the heart. There is an extremely high unmet need for teenagers and young adults with DMD. A therapy that helps the dystrophic heart is urgently needed by the entire Duchenne & Becker muscular dystrophy community, not specific to ages or mutations.

Our hopes have been pinned on Deramiocel (CAP-1002) from Capricor Therapeutics. We had hoped for an Accelerated Approval by FDA in August of 2025, based on very encouraging Phase 2 data. FDA was also reportedly impressed with the cardiac data from a Phase 2 trial and understood the incredible breakthrough that this therapy represents to the entire Duchenne community and guided the company as such. However, with delays at FDA, apparently new staff did a complete U-turn on the review of the investigational therapy, cancelled an Ad Comm meeting and issued a CRL, despite their earlier advice given to the company. We were shocked and devastated by this delay.

While the company has time to resubmit their data to the FDA, my son does not have time. He continues to lose cardiac and respiratory muscle function every day. Since the delay, this hard-working college student has now been prescribed to use a daytime ventilator attached to his wheelchair. We've gone over a cliff I had hoped we would never see. Can someone still attend college on a daytime ventilator, or even leave their house? Can they still talk, speak, breathe and laugh?

We live in fear of the flu season. Even mild colds can push DMD patients to the point of no return. Deramiocel (CAP-1002) represents the retaining of strength to stay alive. In December alone, my family lost 3 friends in their young 20s to DMD, dying suddenly due to cardiac failure -- young lives that could have potentially been saved by Deramiocel -- we will never know.

Heart function in Duchenne is a matter of life and death, and therefore the review and potential approval of Deramiocel is a matter of life and death. Delays and misdirection by FDA are costing young lives. Please help.

Tay Sachs

This is affecting the Tay Sachs community. My daughter qualifies for a trial, but we are being faced with a 1:3 placebo rate, which means I would have to take her off her compassionate-use medication to go on the trial, while risking a 1:3 placebo rate. My child is 13 and her life expectancy is 15 years old. I don't have 18 months to gamble with a placebo.

Eosinophilic Esophagitis

I am representing the eosinophilic esophagitis (EOE) community as the parent of a child living with this chronic immune-mediated disease. It has profoundly affected his life through daily abdominal pain, difficulty swallowing, food aversions, anxiety around eating, restrictive diets, and social isolation, all of which significantly lowered his quality of life even though the condition is not typically life-limiting.

For years we relied on elimination diets, steroids, and repeated endoscopies while waiting for better options, and those delays meant ongoing inflammation, fear of choking, emotional distress, and disrupted school participation. But now that dupilumab (Dupixent) has finally received approval from the U.S. Food and Drug Administration, he is in remission. His stomach pain and inflammation have improved, his allergies are also improving, and he can eat with less fear. This treatment is not a cure: access barriers and variable responses remain, and we still manage chronic symptoms and monitor his nutrition and mental health. Time in rare disease is not abstract because six months can mean prolonged pain, nutritional compromise, and worsening anxiety around food, and a year can mean cumulative physical and emotional loss during critical developmental years. So, regulatory delays translate directly into months of suffering and missed milestones, which is why this approval is so meaningful to our family and why my son, now finally feeling better, hopes to return to school soon and reclaim the normal childhood experiences that EOE once took from him.

Limb Girdle Muscular Dystrophy

Limb Girdle Muscular Dystrophy 2I/R9 is an ultra-rare, progressive muscle-wasting disease that ultimately leads to loss of ambulation, respiratory complications, and cardiac failure.

My 18-year-old daughter, was diagnosed with LGMD2I/R9 at the age of two. Since then, she has endured profound physical and emotional losses. She falls frequently and can no longer rise from the floor independently. She is unable to transition from sitting to standing without assistance, which means she cannot use a traditional bathroom or sit in a standard classroom or restaurant seating without someone physically lifting her. Recently, she shared that the constant vigilance and planning required just to get through each day is mentally and emotionally exhausting.

My daughter has already undergone two major surgeries, a spinal fusion, and an Achilles tendon lengthening, to address severe contractures caused by this disease. She experiences extreme fatigue and pain when she pushes herself beyond her limits. And yet, despite her strength and resilience, LGMD2I/R9 continues to take more from her each year.

For the past 15 years, our family and our foundation have worked tirelessly to support research and drug development. Despite this commitment and progress, there are still no FDA-approved therapies for LGMD2I/R9. Several clinical trials are underway, offering hope to our community. However, difficulties with funding have halted progress in a promising gene therapy program that could significantly impact patients like my daughter.

Time is not a neutral factor for our community. This disease is progressive. Every month without treatment means further muscle loss, decreased independence, and irreversible decline. Every day that passes carries the risk of a fall that could result in fractures, respiratory failure, cardiac complications, or permanent loss of remaining muscle function.

My daughter, and so many others like her, do not have time to wait.

Niemann-Pick

My son had Niemann-Pick type C (NPC) disease and died at the age of 20. NPC disease is a fatal, genetic, neurodegenerative disease that causes progressive deterioration of the nervous system resulting in dementia, seizures, disrupted sleep patterns, behavioral disturbances, fine and gross motor problems, and learning disabilities. The disease takes away your ability to walk, talk, swallow, eat, and breathe.

The NPC1 gene was discovered in 1997 after decades of research. Then it took 27 more years of research before the FDA approved the first treatment for neurological symptoms of NPC disease in September of 2024. There needs to be multiple drugs approved by the FDA to address all the symptoms of NPC disease.

There are multiple challenges and regulatory delays surrounding FDA approved treatments for NPC disease.

1. Drug trials for common diseases like high blood pressure and asthma can include thousands of participants. The size of the trial makes it easier to achieve better randomization and see statistical significance of the drug's efficacy
2. Trials are much more difficult for rare diseases, particularly because symptoms and the rate of disease progression in the patients is often highly variable
3. The cyclodextrin trial for NPC disease (my son was in this trial) included just 56 children and young adults with a broad range of disease symptoms, which limited the interpretation of the results
4. Rare disease drugs need another pathway to approval by the FDA. They cannot follow the same pathway to approval as common diseases
5. We need to modernize clinical trial designs
6. Include natural history controls and natural history comparison groups
7. Rely on surrogate or intermediate endpoints for rare genetic diseases
8. Allow Accelerated approval for ultrarare genetic diseases with primary disease biomarkers based on clear biology for treatments aimed at the underlying cause
9. Issue a guidance in compliance with original FAST bill (FDASIA) to ease the excessive burden on qualification of primary disease cause biomarkers
10. Accelerated approval shortens the time it takes to drug approval, while allowing for longer timeframes to collect confirmational data
11. Randomized placebo-controlled trials for a disease with irreversible brain or muscle disease are unethical. No placebo-controlled trials for these diseases.

Niemann-Pick type C disease relentlessly takes away your ability to walk, talk, swallow, eat and breathe as time marches on. Regulatory delays mean your ability to swallow or walk

slips away. Having longer timeframes to collect confirmational data after a drug is approved gives patients with neurodegenerative diseases access to medicines that can slow or stop the progression of their disease and buy them time with their families. Rare disease drugs need another pathway to approval by the FDA. They cannot follow the same pathway to approval as common diseases.

[New submission]

I am the mother and sole caregiver of my 4-year-old daughter.

I represent the Niemann-Pick disease type C community.

Niemann-Pick Type C is an ultra-rare, progressive, neurodegenerative genetic disease affecting approximately 1 in 100,000 individuals. In the United States, there are only about 943 diagnosed cases.

It is terminal.
It is fatal.
There is currently no cure.

Many children with early-onset forms do not survive into adolescence or early adulthood.

The disease is often described as “childhood Alzheimer’s.” Like Alzheimer’s and other neurodegenerative conditions, it causes gradual and irreversible loss of brain function. But in my daughter’s case, that decline began in early childhood.

My daughter was born healthy. She walked. She talked. She laughed. She said “Mom.”

Then she began to regress.

Niemann-Pick Type C is caused by mutations in the NPC1 or NPC2 genes, preventing the body from properly transporting cholesterol within cells. Toxic accumulation damages the brain and nervous system over time.

Symptoms vary widely, which frequently delays diagnosis. In our case, it took nearly two years to receive answers. During that time, we watched her lose skills while moving from specialist to specialist. Many providers had never encountered the disease.

By the time we received the diagnosis, progression had already taken hold.

Today, she can no longer walk, talk, or swallow independently. She has lost voluntary movement in her limbs. She is aware. She is present. But she is increasingly trapped inside her own body.

Last year, what was expected to be a brief hospital stay became a two-month hospitalization after she took a sudden turn for the worse.

This is what terminal neurodegeneration looks like in real time.

In six months, a child can lose speech.
In a year, a child can lose mobility.
Decline does not wait for process.

There is no approved cure to stop this disease. Supportive care may manage certain symptoms, but it does not halt progression. The unmet medical need is profound.

As her caregiver, I live with the reality that the clock is not on our side.

Time, for us, is measured in functions.

Time is whether she can still swallow safely.
Time is whether she can still move her hands.
Time is whether she still recognizes my voice.

When research funding slows, when regulatory pathways stall, or when advisory committee meetings decrease — as seen in 2025 with significantly fewer meetings for prescription drugs and biologics — families like mine feel that delay immediately.

Because regulatory delay is not neutral.

For a terminal disease, delay means decline.

The work of this Committee matters. Pediatric neurodegenerative diseases and adult neurodegenerative diseases share biological pathways. Investments in one advance progress in the other.

I cannot change her diagnosis.

But Congress can ensure that the pace of research and regulatory review matches the pace of disease.

For families living with terminal neurodegeneration, time is everything.

And once it is lost, it cannot be returned.

Thank you.

ALS

We lost Mom to ALS 29 years ago last month.

The prognosis seemed outrageous then, and it is all the more outrageous today. There is still no known cause, and the treatments are not very effective.

A dear friend I lost to ALS talked about the ALS Clock. It runs relentlessly fast. Healthcare delivery simply doesn't keep up. Research moves at the speed of concrete. Surely there are ways to speed up the deliverables without compromising the scientific truth.

We want therapies that work. We also want to stop this relentless destruction of vibrant human beings.

I was diagnosed with ALS mid-2025 and am now really starting to lose control of my body. Before the disease, I was an active equestrian, long walker, and generally in great health. Not only is it disheartening to have a terminal illness, but the lack of approve treatments to extend life without trach/feeding tube is horrifying. This disease really strips away who you are along with your dignity since ultimately you will have to rely 100% on others for care (not a good thing for a previously independent person who likes to be in control). I feel like there are far more people trying to live with this disease than I ever thought --- all relying on help to maintain their lives. Please help this community out by funding further research with a goal of curing this disease or at least converting it to a 'chronic' one.

[New submission]

I am paralyzed from ALS, a rare disease. This results in a large uncompensated caregiver burden on my family. While there are approved treatments, they are not effective at stopping the disease progression. The current clinical trials disqualify the vast majority of people living with ALS.

The FDA could be more involved in Expanded Access Protocol therapies, which recently have shown positive medical evidence in conjunction with clinical trials.The FDA provides minimal funding for ALS research, while many identified disease ideas go unfunded.

Please provide the FDA with directions and funding for ALS Research, CTs & EAPs.

Spinocerebellar Ataxia

I am 43 and living with Spinal Cerebella Ataxia. My sister died at 48 and my father died at 56. There are no approved treatments for my condition. The FDA regulations don't seem to reflect the essence of time, and what it means to me as a rare disease quickly degenerates my abilities. I need ends to justify the means in this situation.

My disease is harsh and hope is hard to find when my government organizations seem to be misleading research, so medicines are denied. The triruzole drug seemed like it had followed all the rules and was being fast tracked towards approval only to be denied. This is not about a company making money or not about the population of voters being denied access to the first drug that was proven in studies to slow the progression of my SCA3 rare disease. Please have compassion-based ideals and firmly understand that the disease is worse than the medicine, and we are all looking at a shortness of time.

[New submission]

I have Spinocerebellar Ataxia type 3 (SCA3), a hereditary degenerative disease. Each day my balance and coordination worsen, or I notice something new that I can no longer do. Recently, I noticed that I can no longer jump or coordinate to do a jumping jack. This disease has been known and researched for 30 years, yet there is still no treatment or cure. Rather, some medicine, Troriluzole has been proven to help, but we cannot get it because FDA red tape prevents its approval. The neurologists have nothing to offer us and tell us to exercise a lot, but our bodies are losing the ability to move well.

My mother who died from complications related to SCA3 always told me, "Even if you get it, a cure will exist by your time." Now, I fear that I will have to tell my children (who have a 50% likelihood of getting it) the same thing.

[New submission]

I have a SCA3. There is no cure for it. It is disheartening when you hear of a possible treatment for it, only to hear that the FDA has rejected the medication. Whatever their reasons for rejecting it, there is still no cure, so what is the harm. There will be a day when I am unable to type this. I am restricted to wheelchairs now. Unable to drive, traverse the lawn let alone woods. I used to work for Verizon installing and repairing communication lines. I was up and down ladders and utility poles every day. Now I can't stand being unassisted.

[New submission]

I was 42 years old and absolutely terrified. My body was doing something strange. I embarked on a four year journey to find a neurologist that was able to help me. In the interim years, I was subjected to countless blood tests, MRIs, spinal tap, and multiple nerve conduction studies. All this to find that I have Spinocerebellar ataxia. Unfortunately, there has not been enough research to specify exactly what I have. Although this is incurable, I would like to be able to pinpoint my diagnosis. This is not possible without more research being done.

[New submission]

I was diagnosed with SCA 6 Ataxia several years ago -- and have been in a trial program with Biohaven Pharmaceutical for the past year. The drug offered is to "slow down the progression" of the disease. The FDA has not approved the drug for distribution, however, to my knowledge this is the only drug available for those suffering with SCA 6 Ataxia. On my 70th Birthday my PGA handicap index was 12. I was enjoying life as a part-time employee, playing golf, riding bikes, traveling with our Airstream and being an active grandparent. Now I am preparing my home for a wheelchair ramp, adding a lift to our mini-van, and waiting for a cure! Help is REQUIRED! Life has radically changed for me, my wife, my children and our grands. WE Need ACTION! Thanks.

[New submission]

I have a form of Spinal Cerebellar Ataxia, specifically SCA5. I have a de-novo mutation of the gene which means that neither of my parents have the disease, but I have passed it to at least two of my children. My two girls, now in early adulthood (22 and 24), are experiencing gait and balance issues. My youngest son (21) has not presented at this time, and we have not tested for the gene because there are no cures or trials for this rare disease.

I currently use a walker because this disease causes issues with my balance and gait. I am beginning to experience some slight difficulties with speech as well. I am 54 and am much more concerned with finding a cure that can halt or slow down the progression for my children. My oldest is getting married and I'm concerned with navigating life as a young mom and being able to handle a child.

I am working with a researcher on the disease and am learning how archaic the process is. I have learned a bit about Alphafold and some of the tremendous benefits AI can have on research and drug development. Especially in reducing the time and more importantly the cost of drug development. However, I am concerned that bottlenecks in the FDA process will

negate these benefits. I am very concerned because my disease and the disease that affect my children is progressive and every delay means that they will face some of the things that I am experiencing as an adult. I am not as worried for me as I am for my children and urge the committee to do whatever is necessary to move this forward.

[New submission]

One year ago, I was diagnosed with Spinocerebellar Ataxia type 3 (SCA3), a rare, progressive, and currently incurable neurodegenerative disease. I am facing a future of steadily worsening balance, coordination, speech, and daily functioning. Without medical intervention, I am destined to end up in a wheelchair with complete reliance on caregivers.

I am a wife and a mother of two daughters, and I inherited SCA3 from my father. My father had no chance of treatment for his SCA3, as there was none available while he was alive. I witnessed how the disease stole his ability to walk, to feed himself, to speak intelligibly, and to swallow food without choking. His mind remained intact, but his body was failing. SCA3 eventually claimed his life. What echoes in my ears is one of the last understandable words that he spoke as he lay dying in the hospital. That word was “help.” Distressingly, there was no help to be had.

As a doctor who witnessed how SCA3 first affected my father, I understood the implications when I unexpectedly stumbled one day. Over the following year, frequent muscle cramps became alarming occasions when I would take an extra step to catch myself from falling. I became wary of uneven ground and fearful of walking up or down stairs without railings. I was only 51. I made a bucket list of all the things I wanted to do in the next 20 years or so before I lost the ability to walk, to hold a pen, or to communicate well. The thought that my daughters had a 50% chance of inheriting this crippling disease from me kept me up at night.

However, I was given new hope 10 months ago when I started taking VYGLXIA (troriluzole), an investigational drug developed by Biohaven, through the FDA’s Expanded Access (compassionate use) program. In clinical trials, Biohaven showed that troriluzole has the potential to slow disease progression by 50-70%. Astoundingly, within 3 months of starting the medication, my physical exam reflected improved balance and coordination. By 6 months, I was more confidently walking in a straight line and climbing up and down stairs without assistance. In the last 10 months, my symptoms have stabilized and I have many more good days than bad ones. I know troriluzole will not cure me, but it is my only lifeline to a better future — maybe even one without a wheelchair.

Disappointingly, in November 2025, the FDA issued a Complete Response Letter to Biohaven's New Drug Application for troriluzole. Fortunately, Biohaven remains committed to working with the FDA to find a path forward. I am fearful that I and many others will lose access to troriluzole once the Expanded Access program ends. This affects not only me, but the thousands of other spinocerebellar ataxia patients across the country who are on this medication. My biggest fear, however, is that my daughters will not have access to troriluzole if they should need it.

I respectfully ask that you contact the FDA and express your strong support for the expeditious review and approval of troriluzole for spinocerebellar ataxia. Your advocacy could make the critical difference in bringing the first disease-modifying therapy to those affected with spinocerebellar ataxia.

Thank you for your commitment to improving the lives of those living with rare and serious diseases. While there was no disease-modifying treatment for my father when he was dying from SCA3, you can help give me, and thousands of others who are fighting this dreaded disease, a chance to enjoy their best lives for as long as possible.

[New submission]

To whom it may concern:

I'm a 47-year-old male with SCA Type 1. There are currently no treatments for this disease. It is an extremely rare progressive, inherited neurodegenerative disorder caused by a CAG trinucleotide repeat expansion in the ATXN1 gene, typically inherited in an autosomal dominant pattern. It causes imbalance, lack of coordination (ataxia), slurred speech, and dysphagia, usually starting in the 3rd or 4th decade, with a 10–30-year lifespan post-onset.

Bottom line, there's no money in research/curing this disease, so I'm at the mercy of my government through a subsidy. The FDA recently denied approval for a drug called Troriluzole, which has very promising potential delaying the onset of symptoms by an average of 60% with real world data. Meaning I'd live long enough to die of something else. After the denial, the company developing the drug was forced to put it on the back burner so to speak, as they couldn't keep investing money into something that may not have a return. The drug is still available through the Expanded Access Program and may be my only hope for any kind of quality of life over the next 20 years. They have another hearing with the FDA soon to discuss a potential path forward. I have 3 children.

My hope, even if I don't get it, would be to have it available to them. Judging by how the disease is passed on, I would bet at least 2 out of the 3 are carriers. My Grandfather took his own life due to the disease. My Father is 70 and is very close to being in a wheelchair and needing a feeding tube. My children have witnessed his decline over the past 10 years and will likely have to deal with mine as well. I've begun experiencing symptoms, as have several cousins of mine. Time is of the essence. Thank you for taking my statement.

[New submission]

Good day, I was diagnosed with Ataxia last year. As you know this is a quickly degenerative inherited disease that impacts the quality of life (I have no balance, prone to falls, shrinking brain, no control of extremities, slur speech and more) I have left as a man of only 50 years old, a full time employee for the municipality I work for and a proud father of 3 school age children. My father and my son suffer from this disease severely and it hurts to know that there is no cure at the moment. However, a company by the name of Biogen just concluded some successful studies with a pill that the FDA did not approve even though their clinical trials showed success within the subjects they recruited. Unfortunately, I cannot get the pill Biogen designed because of all the regulatory decisions. I am imploring to please approve Biogen's treatment for Ataxia so that I may live longer and live a better-quality life. FDA—please approve Biogen's pharmaceutical treatment for Ataxia so that my father, who is now dying at 72 because of this disease, my son is now completely disabled, myself and others an opportunity at a slightly better life with treatments targeting Ataxia. Thank you and I hope you can bring some hope to those of us who are in the dark.

Chronic Inflammatory Demyelinating Polyneuropathy

I represent the rare disease community of people living with Chronic Inflammatory Demyelinating Polyneuropathy (CIDP), and I stand alongside those impacted by Guillain-Barré syndrome (GBS). These are autoimmune diseases where the immune system attacks the peripheral nerves — stripping away the protective myelin sheath and disrupting the body's ability to function.

For many people, CIDP is described as a "motor neuropathy" that causes weakness and sensory loss.

But for me, it did more.

CIDP did not just attack my ability to walk.
It attacked my autonomic nervous system — the system that controls the things your body is supposed to do automatically, like regulating heart rate and blood pressure.

My heart began misfiring. My rhythm became unstable. My body could no longer reliably control something as basic — and life-sustaining — as a heartbeat.

As a result, I now live with a pacemaker.

At 60% pacing, my device is not optional — it is supporting a heart that cannot consistently regulate itself. What started as an autoimmune neuropathy progressed into cardiac instability. This is something many people do not realize can happen in the GBS/CIDP spectrum.

CIDP affects quality of life in profound ways. Weakness. Fatigue. Nerve pain. Instability. Periods of paralysis. Loss of independence. And in cases like mine, autonomic dysfunction that can impact the heart, blood pressure, and overall safety.

CIDP may not always shorten lifespan directly — but untreated or poorly controlled disease can lead to permanent nerve damage, severe disability, and life-altering complications. Six months without adequate treatment can mean muscle atrophy. A year can mean irreversible nerve damage. Autonomic involvement can mean emergency interventions.

There are FDA-approved treatments, including IVIG, corticosteroids, plasma exchange, and subcutaneous immunoglobulin therapies. These treatments can slow progression and restore strength for some of us. They have given me stability at critical moments.

But they are not cures.

Many patients require lifelong infusions. Some do not respond to first-line therapies. Others lose response over time. Access can be delayed due to insurance barriers, prior authorizations, step therapy, or questions about diagnostic criteria. Because CIDP is rare and heterogeneous, clinical trials are small and complex. There have been ongoing debates about appropriate endpoints, biomarkers, and what level of evidence is “enough.”

For patients like me, those debates are not theoretical.

Regulatory delays are measured in nerve loss.
In muscle loss.
In independence lost.

When a decision is delayed by six months, that may mean six months of progression. In autonomic involvement, that can mean dangerous instability. Nerves do not regenerate quickly. Some damage cannot be undone.

Time, for me, means:

Six months — the difference between walking independently and needing assistance.
One year — the difference between working and permanent disability.
Delay — the difference between a stable rhythm and a life-threatening one.

I never imagined that a rare autoimmune neuropathy would lead to me needing a pacemaker. But rare diseases do not follow neat textbook boundaries.

I share my story because patients cannot afford prolonged uncertainty. We cannot afford regulatory gridlock. We cannot afford to wait while our nerves are under attack.

In rare disease, time is not just a policy variable.
Time is nerve function.
Time is autonomy.
Time is independence.
Time is life.

[New submission]

Do you like to wake up in the morning feeling pain free, full of energy, and ready to start your day? I wish that was my reality, but it is not. I have lived with a rare disease for 31 years since the age of 28. I have Chronic Inflammatory Demyelinating Polyneuropathy (CIDP). I am 1 in 100,000.

Each morning, I wake up sore, stiff, and in pain. I have been on IVIG - intravenous imuglobulin gammagloublin for 25 years. As a patient of a rare disease community, the FDA is important. We need you to allow research for these rare conditions to continue. Without research we will not develop new and improved medicine to improve our quality of life. Medicine is expensive. More research allows patients and doctors options. Without medicine, I would lose my quality of life. I cannot go longer than two weeks without my medicine. I rapidly declined. Walking, daily life skills, and being a productive member of society decrease rapidly. Regulatory delays mean that I will end up in a wheelchair sooner or even totally dependent on others for my care. How would you like to live with that prognosis?

Huntington's Disease

I am at risk of living with Huntington's disease and live with family members who tested gene positive. I am submitting this testimony to share my concerns about current FDA regulatory decisions and their impact on the Huntington disease (HD) community.

Huntington's disease (HD) is a rare, inherited, and ultimately fatal neurological disorder. Often striking in the prime of a person's life. It causes a progressive decline in a person's ability to think, move, and function independently. If one parent has HD, their children have a 50 percent chance of inheriting HD. There is currently no treatment that slows or stops the progression of HD. It progresses from symptom onset to death over approximately 15 years often beginning in the prime of life.

As a child, experiencing a parent with HD was traumatizing, tragic, and painful. Not only did I lack a maternal figure, I had to watch someone, someone who I should've known but instead as a stranger, deteriorate before my eyes. I couldn't explain what was happening or relate to any of the peers or adults in my life. It was isolating. Even after her passing, seldom do people understand the trauma I lived with as a child. In the present, the ever-impending doom of my brother's sickness as well as my own potential sickness, looms over my daily life like a thick fog. I can never be at peace. I never will be at peace. This is the experience of many children of HD parents like myself.

My brother participated in a clinical trial. This trial occurred hours away and was worrying to experience even from a distance. There was an overlying anxiety over his physical and emotional wellbeing.

People living with HD and their families are willing to take risks to stop the progression of the disease. Nearly three-quarters of respondents to the 2024 HDSA HD Symptoms and Treatment Impact Survey said they would accept treatment risks to gain five years with no disease progression, and almost 35% were willing to accept risks for even three years of halted progression.

When FDA policies are unclear, inconsistent, or unpredictable, it creates additional barriers for therapies that may represent our only hope. I am concerned about lack of transparency in decision-making for HD therapies, inconsistent application of regulatory standards, and failure to adequately account for the realities of small patient populations. I respectfully urge the Committee to direct FDA to:

- Consider benefit/risk tolerance of people living with Huntington's disease when making decisions about trials for HD treatments and cures.
- Utilize natural history studies for rare disease trials.
- Be clear and transparent about its decisions related to HD trail decisions, including recent statements about uniQure's AMT-130.

Thank you for the opportunity to submit testimony for the record. I appreciate the Committee's attention to the needs of rare disease patients and families. For our community, innovation is not optional — it is survival. It represents more time, more independence, and more life.

[New submission]

I am gene-positive for Huntington's disease. I cared for my father as he declined and eventually passed away from this disease in January 2021. I now live knowing that Huntington's disease is progressing inside me.

I am also a clinical research participant.
Over the years, I have participated in a therapeutic clinical trial and in several observational studies, including contributing to the natural history research that helps define how Huntington's disease progresses over time. That natural history data is not abstract to me - it represents blood draws, cognitive testing, travel, vulnerability, and time given freely in the hope that it will help build a pathway to treatment.
Patients like me enroll in these studies because we believe in science. We believe in partnership. We believe that if we do our part, the system will work with us.

Huntington's disease is a fatal, inherited neurodegenerative disorder with no disease-modifying treatment. Time is everything. Time is cognitive function. Time is independence. Time is the ability to be present for your children – to help with homework or teach them how to ride a bike.

Recent regulatory decisions affecting rare disease therapies - including incomplete response letters, refusals to file, and shifting expectations for trial design - have had destabilizing consequences across patient communities.

For diseases like Huntington's, traditional placebo-controlled trials are uniquely difficult. Our population is small. Our disease is progressive and irreversible. Natural history data and external controls are not shortcuts; they are tools built from years of patient contribution and scientific rigor.

When regulatory expectations shift late in development, it does more than delay a submission.

It calls into question whether the years patients have spent contributing to natural history databases will truly be recognized and utilized as intended.

We are not asking for lower standards or shortcuts. We are asking for:

- Consistent application of accelerated approval pathways
  Transparent and predictable regulatory expectations
- Scientific flexibility appropriate for fatal neurodegenerative disease
- And meaningful incorporation of patient experience into risk-benefit decisions

I have sat in research visits answering memory questions, knowing that one day I may not be able to answer them. I have contributed to natural history studies designed to map the decline of my own disease. That data exists because patients like me were willing to document our progression in the hope that it would shorten the path to treatment, not extend it.

When a delay happens in Huntington's disease, it is not neutral.
Delay is decline.
Delay is an irreversible loss of brain cells.
Patients have done their part. We show up to studies. We give our data. We accept risk. We build natural history datasets so that innovative trial designs can be possible.
We are asking that the regulatory framework meet us with the same urgency and consistency.
Thank you for holding this hearing and for examining how FDA processes affect real families like mine that are facing progressive, fatal rare diseases.

[New submission]

I represent the Huntington disease (HD) community, which is a rare, neurodegenerative disease that impacts motor, cognition, and behavior. There are no treatment options and the average lifespan is 10-20 years since symptom onset. I witnessed my mom battle this for 17 years before passing away over 10 years ago. Because of the genetic component, each child of a parent with the disease has a 50-50 chance of inheriting it. I decided to go through testing and found out I also carry the gene, and I'm guaranteed to get the disease unless there's a treatment in time. I tested over 15 years ago and now I'm racing against this invisible clock as I get closer and closer to potentially developing symptoms.

More recently, there's been some promising research from a company called uniQure who's working on an investigational gene therapy for HD. They demonstrated clinical benefit in outcome measurements that are meaningful to patients and families. They previously agreed with the FDA on using an external control arm through a natural history dataset instead of placebo since it's not ethical to make a patient go through a brain procedure. However, the FDA is no longer in agreement, and the HD community is running out of time as people will continue to get worse.

Every moment matters as the HD community specifically told the FDA in a recent PFDD meeting. We want to continue to build memories with our loved ones rather than having them lose their independence day by day.

Prader-Willi Syndrome

I am a constituent of West Virginia and have a 7-year-old daughter with Prader-Willi Syndrome (PWS). West Virginia has always been her home, and she received a diagnosis of PWS at 2 weeks old. PWS is a rare genetic disorder affecting approximately 1 in 20,000 individuals, and for decades families have navigated it with almost no FDA-approved treatment options. It begins at birth with failure to thrive and quickly becomes a lifelong medical and developmental challenge requiring intensive therapies, constant monitoring, and extraordinary vigilance. In 2000, growth hormone became the first FDA-approved treatment—a milestone that changed outcomes for physical development but did not address the full complexity of the disorder. Then, twenty-five years later, in 2025, a second approval came: Vykat for hyperphagia, the hallmark, and most life-threatening symptom of PWS. Hyperphagia is not overeating by choice; it is a relentless biological drive to eat without ever feeling full, leading to severe obesity, diabetes, hypertension, and premature death. Two approvals in twenty-five years is progress—but it is not enough. PWS is multifaceted, involving cognitive delays, behavioral dysregulation, anxiety, rigidity, and metabolic instability. Families cannot wait decades between breakthroughs. We need accelerated research, additional FDA approvals, and treatments that address the full spectrum of symptoms—not just one.

I see two realities and issues that policymakers can easily overlook. The first is that parents are taking steps for earlier interventions (therapies and these few approved treatments) and individuals with PWS are living longer than before. Secondly, their caregivers are also aging alongside them. Parents who have managed complex medical regimens, locked kitchens, prevented life-threatening food access, and de-escalated neurological behavioral episodes for decades cannot do so forever. There is not an individual with PWS who can live fully independently without structured support, yet as parents age, the system offers few sustainable options. Institutions have rightly closed, but closure without replacement leaves families in crisis. We need support both in-home for as long as our loved ones can remain there, and highly supported residential communities that preserve dignity, provide medical oversight, and ensure safety when parents can no longer serve as primary caregivers. It’s a both/and. The life expectancy for individuals with PWS still hovers in the low twenties, largely due to complications of hyperphagia. Twenty is too young. If we are serious about extending lifespan, FDA approvals must continue, treatments must expand, and long-term care infrastructure must evolve in parallel. Our loved ones deserve more than survival—they deserve longevity, stability, and the promise that when we age, they will still be protected.

Hereditary Xerocytosis

I'm a Registered Nurse. My mother-in-law, daughter and husband have Hereditary Xerocytosis. At this point, my daughter is in stable condition. My husband, however, has elevated liver enzymes, jaundice, fatigue, and iron overload as a result. My mother-in-law has daily severe fatigue, symptomatic anemia requiring transfusions, and jaundice. This affects them in completing activities of daily living.

Residing in a rural area, one of the biggest obstacles is finding a physician knowledgeable in the illness. I am unaware of any approved treatments for the disease. Time for us is everything. My loved ones live with a high risk of aplastic anemic crises. I hope for a cure or suppressing treatment & gene therapies for my daughter's lifetime. What a gift it would be for her to be able to become a mother knowing she can avoid passing on this hereditary illness.

Pyruvate Dehydrogenase Complex Deficiency

I am writing as a parent to a sweet, three-year-old girl who is currently living with Pyruvate Dehydrogenase Complex Deficiency (PDCD). On September 4, 2025, Saol Therapeutics shared a press release stating that the U.S. Food and Drug Administration (FDA) will not approve Saol Therapeutic's New Drug Application for SL1009 (DCA) for Pyruvate Dehydrogenase Complex Deficiency (PDCD) in its current form, outlining specific observations that must be addressed before potential approval can be reconsidered. We were fortunate to receive her diagnosis in utero. Because of that, she began the ketogenic diet on the first day of her life.

From birth, everything has been carefully measured and medically managed. We also knew time is a gift with her, as life expectancy with PDCD is early childhood. Despite early intervention and around-the-clock care, she is nonverbal and non-ambulatory. She lives with profound neurological and physical disabilities that impact every aspect of her daily life.

She cannot speak to tell me how she feels. She cannot walk to explore the world. She depends on us for everything. And yet, she is the sweetest, strongest little girl you could ever meet. She is joyful. She is determined. She deserves every possible chance at life.

I have spent years advocating for PDCD to try and help save our daughter and give her the best chance at life possible. We have been anxiously awaiting any type of therapy or drug for her for three years, and this news is devastating to say the least. Right now, someone living with PDCD has a life expectancy of early childhood, and we are in a race against time to get a drug approved to extend and improve their quality of life.

However, DCA has given us hope and my daughter was able to start the drug in November of 2025 through an Expanded Access Program.

Since starting DCA, we have witnessed meaningful changes:

- Increased energy
- Improved appetite
- More voluntary movement
- Greater engagement with her surroundings
- And for the first time, she began to wave

That wave may seem small. For us, it was monumental. For a child who is nonverbal, it was connection. It was communication. It was hope.

DCA remains under regulatory review. The possibility that Expanded Access could be restricted or that approval could be delayed due to another clinical trial requirement is frightening beyond words. For children with PDCD, time is not an option as disease progression does not pause.

In rare pediatric diseases, even the smallest milestones matter. A wave matters. Increased energy matters. Engagement matters. Traditional clinical endpoints may not fully capture what progress looks like for children like my daughter. For our community, perfection cannot be the enemy of progress. When therapies show safety and real-world functional improvement—even small ones—those gains should matter.

My daughter was not given a generous timeline when she was diagnosed. Every month she is stronger, more engaged, more connected is a gift. Removing access or delaying flexibility risks taking away progress that her fragile body fought so hard to achieve.

She cannot advocate for herself. I am asking you to help ensure that children like her are not left behind by a system that moves more slowly than their disease. We hope the right voices make it to the FDA and meetings with Saol to come up with a plan moving forward for full approval given there have been no safety concerns with the results.

Thank you for standing with rare disease families and children who cannot afford to wait.

[New submission]

I am writing on behalf of my son who was diagnosed with Pyruvate Dehydrogenase Complex Deficiency. He was diagnosed at the age of 5 after many years of testing and lack of answers. This mitochondrial disease affects his energy levels. At times he loses motor function in his

legs, leaving him unable to walk or stand. These episodes come with extreme fatigue at times leaving my son, a now 8-year-old boy unable to act as his peers at times.

We are blessed. Many patients with PDCD have cognitive delays, feeding issues, and some die from this condition. There is no cure for PDCD and as it is a rare genetic disease, it is not screened for with newborn screening. We spent 3 years and an unmeasurable amount of money and time to try and figure out what was causing Millers symptoms. There is not currently an approved treatment for PDCD, though DCA has been proven in some trials to lesson symptoms. The FDA would not approve DCA for the treatment of PDCD and closed further trials.

My son has been on a ketogenic diet since he was diagnosed to keep his symptoms at bay. He loves baseball and it is hard as a parent to watch him struggle with something he loves because of a condition that has no treatment. Since diagnosis, he has participated in speech, physical, and occupational therapies. His medical shakes that he requires as part of his ketogenic diet are not covered by Blue Cross Blue Shield insurance, therefore, we pay for those out of pocket. Funding and clinical trials for children like Miller are necessary. These kids deserve a life lived fully.

Other Mitochondrial Diseases

I am representing the mitochondrial disease community, specifically families of children with extremely rare genetic forms such as COXPD4, a life-threatening mitochondrial disorder that affects energy production in every organ system and has shaped my daughter's entire life through chronic fatigue, muscle weakness, neurologic symptoms, gastrointestinal challenges, and significant mental health impacts that fluctuate with her metabolic stability. Many children with mitochondrial disease do not survive to adulthood, and while our daughter is nearly there, her path has required constant vigilance, complex medical care, and extraordinary financial and emotional effort just to maintain stability and function.

With no FDA-approved cure available, treatment is limited to supportive care, metabolic supplements, and alternative therapies such as neurochiropractic care that have helped her tremendously but are expensive and often not covered by insurance. This leaves families to rely on grants, out-of-pocket costs, and relentless advocacy simply to preserve quality of life.

Regulatory and research challenges for ultra-rare conditions like COXPD4 include small patient populations, limited clinical trial feasibility, and disagreements about appropriate outcome measures, all of which slow the development and approval of targeted therapies while children continue to decline or face life-threatening metabolic crises. For us, time is everything because six months can mean the difference between stability and regression in cognition, stamina, or psychiatric symptoms, while a year without disease-modifying treatment can mean irreversible loss of function that can never be regained. So, regulatory delays are not theoretical—they represent lost abilities, increased medical fragility, and ongoing uncertainty about survival.

Although we feel deeply grateful and privileged that through hard work and grants we have mostly been able to afford the supplements that keep her stable, the reality is that many families cannot, and a true cure would mean the world to our family and to the many children with mitochondrial disease who are still fighting for the chance simply to reach adulthood.

Invasive Fungal Disease

I am representing the invasive fungal disease community, specifically families affected by CNS mucormycosis (Apophysomyces variabilis) -- a rare, aggressive fungal infection that can spread from the sinuses to the brain.

Mucormycosis is often misunderstood, under-recognized, and rapidly fatal if not treated immediately. Survivors are rare. Families navigating it often feel invisible.

My Story

What began as what appeared to be orbital cellulitis from a routine sinus infection progressed into a life-threatening brain infection.

Mucormycosis is an angioinvasive fungus-it invades blood vessels, cuts off blood supply, and causes tissue death. In my case, it spread from the sinuses to the skull base and brain.

The impact included:

- multiple hospitalizations
- prolonged ICU care
- five brain surgeries, including emergency neurosurgical intervention
- skull base osteomyelitis (bone infection)
- venous sinus thrombosis
- brain herniation
- six months of toxic antifungal therapy with amphotericin B
- permanent hearing loss caused by life-saving medication
- ongoing neurological recovery

Recovery is slow and uncertain. Brain healing takes months to years. Monthly PET scans still show osteomyelitis throughout my skull base. Fatigue, cognitive slowing, and emotional trauma are part of my everyday.

Mucormycosis carries high mortality when it reaches the brain. Survival is medically remarkable, and the aftermath reshapes daily life.

There is no FDA-approved therapy specifically for mucormycosis.

Treatment relies on:

- antifungal medications such as amphotericin B
- aggressive surgical removal of infected tissue
- prolonged hospitalization and intensive monitoring

These medications are toxic and difficult to tolerate. They can damage the kidneys, cause permanent hearing loss, disrupt electrolytes, and produce severe systemic side effects. Treatment is grueling and even with treatment, mortality remains high when the brain is involved.

There are significant unmet needs.

Current treatments:

- are decades old
- were not specifically developed for mucormycosis
- do not reliably penetrate necrotic or infected bone
- require prolonged IV administration
- carry substantial toxicity

There are limited clinical trials for rare fungal infections. I am dependent on compassionate use programs, investigator-initiated protocols, or extrapolated evidence from other fungal diseases.

Because mucormycosis is rare, research funding is limited. Diagnostic tools are also inadequate. There is no rapid, reliable blood test for early detection. Diagnosis often requires biopsy, for me, progression had already occurred by then. Earlier recognition and targeted therapies could have changed everything.

Rare invasive fungal infections face unique barriers:

- small patient populations make traditional clinical trials difficult
- lack of standardized endpoints for ultra-rare infections
- uncertainty around acceptable evidence thresholds

- limited commercial incentive for pharmaceutical development delays in diagnostic innovation

Patients with mucormycosis can deteriorate within days. Regulatory timelines operate in months or years.

For rapidly progressive infections, time is survival. When treatment development stalls, patients are left with outdated, toxic options. When diagnostics are delayed, infections are misidentified until irreversible damage occurs.

With mucormycosis, time is everything.
Six weeks of misdiagnosis allowed the infection to invade my brain.

Days can mean the difference between localized disease and catastrophic neurological injury.

Brain healing continues, but deficits are permanent.

For our family, time is measured in MRIs, surgical dates, antifungal infusions, and waiting for scans to show stability.

Regulatory delays mean another person may lose their hearing. Another patient may lose cognitive function.
Another parent may not survive.

We need faster diagnostics.
We need targeted antifungal therapies.
We need regulatory flexibility for ultra-rare, high-mortality infections.
We need recognition that invasive fungal diseases are not fringe...they are emerging global threats.

Every day matters! Because when a rare infection reaches the brain, time is no longer on your side.

Section Two

I was diagnosed with SCA7 (Spinocerebellar Ataxia Type 7). My mother and maternal aunties had this condition as well as my grandmother. In my generation, my brother and I were unlucky to have gotten it out of a 50% inheritance chance; we are very unlucky. Oh, less I forget, my cousin had SCA7 and died because she was not able to care for herself and since her mother was also battling it, she could not help and watched her daughter, my cousin, die.

The thing with this condition is that each generation is worse than the previous. My mom's symptoms started around 40 years; mine is starting around my late 20's; I'm 29 this year. I am very unstable when I walk; my vision is bad; I stumble a lot. I randomly walk into people. They find it weird and so do I; I try to avoid them but my body does the opposite. My cerebellum is shrinking; I get a jolt of shock when I stand for too long and I pass out. My brother, who is 36, has all my symptoms but worse. He says he is "shy." Why? Because you were strong and all of a sudden people come and help you; it's embarrassing. I completely understand him. He does not know of my diagnosis; he is not strong enough for his let alone mine. My mother does not know of my diagnosis either. The lifespan for this is maybe 20-30 years. Since it affects the muscle; eventually, breathing becomes difficult and the heart stops.

Currently, my mother is bed bound (she can't walk/stand/see/feed herself) and I am her caregiver, but I am also sick. I worry about myself. I do not have a husband nor kids to do what I am doing for my mom. I have no one. My brother forcefully got married, so no woman rejects him when he can't walk. Every year that goes by, we get worse; time has never been more precious to us than it is now. As I care for my mom, I see my future, and when I see my brother, I see my path to my future, and it is painful. It depresses me each time I think about it.

A treatment I heard about was Troriluzole. I have contacted the manufacturer to see if they can help. They pointed me to a doctor. She has been ignoring me and there is no one to help me. No neurologist can help. Every help points me to the same doctor who ignores me. I even joined an exercise group at the Ataxia Foundation, but the people there were seniors; I didn't belong. I pray to God for the approval of Troriluzole, so that I do not have to get it prescribed by this one doctor who I cannot seem to access. I am fighting to get better even if it means delaying symptoms. I have not enjoyed life. I wanted to become a doctor but chose to work at NIH to further research for SCA related neuropathies. I sacrificed my future; that is how bad I want this.

Troriluzole is 50-70% efficient. SCA7 is among the rare neurodegenerative diseases. When I look up SCA7, small information exists. I get other SCA related neuropathies instead, yet, because the backbone of all SCA neuropathies are similar, Troriluzole targets that. Troriluzole needs more FDA approval. I hope with complete FDA approval, more than 1 doctor can prescribe it, so I have a broader network of doctors to work with instead of the one at JHU who ignores me.

Troriluzole may not help my mother, nor aunties, but it can help my brother and I. We may have 10-20 years added because of this drug. It is my hope that it gets approval, so we can get the help we -- my brother and I-- need.

[New submission]

I have CRPS (Complex Regional Pain Syndrome). I have been fighting this war for around four decades. I have tried many treatments. However, the treatments that are offered are too late for me. It took me 20 years before I found out what my diagnosis was.
Doctors did not believe this condition exists. I am still told that I am making this up. No one can be at a pain scale of 10 and still smiling/laughing. But, crying and screaming gets a person nowhere and it hurts a lot less by smiling. Laughing is getting to the point of hurting too much. Research on this "disease" is basically not heard of in the US. However, countries overseas have come up with some positive treatment ideas. Those treatments cannot be done here. They are not approved by the FDA.

It gets very disgusting when the treatments that are allowed are for people who were diagnosed within the first 10 years of initial onset. I was already behind the eight ball, at 20 years. There are no treatments offered for someone like me. I struggle every day. I can't find an employer who is willing to take a chance on me.

Even with my scooter, I have huge issues with going out in rain/snow and cold/hot temperatures. Weather patterns, I feel a few days before one hits. How bad I hurt usually means it will be rough front. The deeper the low pressure is, the more I hurt. I am not making this up. It is not in my head. Please believe me. Is that too hard to do? I guess it still is. I can see it in your body language.

[New submission]

Please help patients with chronic illness requiring controlled medications matter again! Why are we the ones left paying a high price for actions of other people? My life is destroyed losing medication that gave me a productive life. Over policy, not my individual health needs! They don't matter at all anymore! Patient suicide jumped 400% since 2016, this

doesn't matter to you? Preventing addicts from getting a few "safer" pills matters more than patient lives?? It's pushed patients to turn to illegal drugs to relieve intolerable pain because Drs turn them away, is that what the goal is?? Kill off as many people with chronic illness as possible? Because that is what people think, genocide by denying vital medication! The added health issues from ongoing physical stress caused by pain cause easily 100,000 more deaths than overdose does!! And you could care less!!

[New submission]

In 2022, at the age of 66, my blood pressure suddenly became treatment resistant after a decade of blood pressure being controlled by one medicine. I also developed constant pulsatile tinnitus. After several months of trying different medicine combinations, a combination of 4 medicines stabilized my blood pressure. I had undergone a multitude of tests to determine cause, and an MRA showed bilateral renal artery stenosis. The goal was to keep my blood pressure in a healthy range and monitor kidney function via bloodwork.

Prior to 2022, I had always been a physically active person. I am a 3rd degree Black Belt in Taekwon Do and won a World Championship in weapons at age 51. When my blood pressure went haywire, I became extremely fatigued and lost almost 20 pounds of muscle mass because my appetite simply vanished.

I worked at regaining healthy weight and tried to stay active, but I would have drops in blood pressure during any kind of excursion. Loss of physical activity was, and still is, a daily mental challenge.

This year, 2025, I began retaining a lot of fluid and had a sharp uptick in shortness of breath and fatigue. My cardiologist referred me to a vascular surgeon who did a Doppler ultrasound on the renal arteries which showed significant stenosis, and it was determined that stenting was required. On February 17, the procedure was started and when the camera approached the left renal artery, the giant monitor showed the classic string of beads associated with Fibromuscular Dysplasia. The right artery presented the same structure. The vascular surgeon let me know that he preferred not to stent until he could consult with the nephrologist.

As it turns out, none of my local physicians have had any FMD patients, so I reached out to FMDSA to find a medical team that could help me. They told me about a specialist I could see. The doctor ordered additional scans and discovered FMD in my iliac, coronary, and carotid arteries in addition to the renal arteries. He also had me added to the FMDSA

registry, which is an important tool for gathering detailed information for use by researchers.

I have added a therapist to my medical team to help me learn new coping mechanisms for the anxiety I have about living with FMD. My old coping mechanism of good, hard work out in the gym is a thing of the past. 24/7 pulsatile tinnitus is also hard to live with. I miss silence. Therapy is helping and I am so grateful for FMDSA.

[New submission]

My friend's son is a real-life superhero. He is kind, funny, resilient, and lights up every room he is in. He is also fighting a rare genetic condition. He is part of a clinical trial that has shown incredible improvement in his day-to-day life. He needs this trial. My best friend needs her son. His brother and sister need him. This is more than devastating. The FDA needs to approve these treatments. These children deserve and chance.

[New submission]

ALS is 100% fatal and terminal with 0 treatment options and a prognosis of 2-5 years. It does not discriminate against age or gender. I am a 33-year-old stay-at-home mom of a 4 year old. It could happen to anybody, including you.

[New submission]

Please provide more funding for research, especially diagnosing, of the disabling disease of gluten ataxia.

[New submission]

I have lived with Dystonia and Ataxia all my life. I have other debilitating issues as well, but these 2 are the rare ones that cause havoc in my daily life. There is no cure, just a few things to try to get relief. Botox was designed for Dystonia but it's too expensive for us patients and our Dr's to have. Hollywood people enjoy the medicine because they have reduced their wrinkles but us who need it for relief can't afford it.

I hope this helps people understand that these rare diseases are not easily diagnosed or treated. Thank you for your time.

[New submission]

My time has passed, so has my children's, but my grandchildren and great grandkids have an opportunity of beating this awful disease. I urge each and every one to help our future generations to eradicate rare genetic conditions. Your unborn child or grandchild or great grandkids could suffer from a rare disease. Wouldn't it behoove you to help in so many ways to help them.

[New submission]

I have had Spinocerebellar Ataxia for 4 years. It's been a great struggle living with and doing daily activities. I've been going from doctor to doctor and because this is, we're not a lot of people who are educated and have ways to help me out. All I can do is exercise and physical therapy. I just hope there will be a cure approved soon for all of us that have this.

[New submission]

I got disabled with this rare disease called Ataxia. I am using a study medication called TRORILUZOLE, that is helping me a lot. Please get this FDA approved.

[New submission]

My son was diagnosed with a rare disease, Langerhans cell histiocytosis, when he was 4 years old. There is no known cause or cure. His treatment was considered experimental because it was considered an orphan disease and had very little funding for research. It is vital that rare diseases get funding at a similar ratio as more well-known diseases, as the victims should be considered no less important and need hope. Thank you.

[New submission]

Being in the sixth generation of a family which has been plagued by this horribly debilitating disease, whose mom (2014) & brother (2017) passed away. As well as grandparents, great grandparents , many aunts, uncles and cousins over the decades.

I think the FDA has to be open to new drugs for not only SCA3, but all the related ataxias.

I currently have been taking Troriluzole for at least 6 months, under an expanded access protocol EAP.

I was part of the original trial for this drug back in 2020.

While it is not a cure, I certainly have had a slow progression of the disease.

Please reconsider the drug use and trials for all of us.

[New submission]

My son has DMD. It is a devastating disease we live everyday. Every day is a day we deal with fear. Fear that he will fall and break his hip, fear that he won't be able to live past his 20's and a fear that limits your capacity to live a life with hope. Currently, he is in a clinical trial

that is helping him. My son wouldn't be able to walk at 19 if he wasn't on the drug. Please consider all outcomes from real people that are dealing with this devastating disease. If a drug helps just one person then it's a success. I urge you to really listen to the DMD community with compassion and love. Thank you for taking the time to read this.

[New submission]

My 10-year-old son has Batten Disease CLN3. There is no FDA-approved treatment or cure. There have been drugs that showed promise but were never able to make it through the requirements to become approved. My son's disease causes waste to build up on the cellular level, and that waste causes cell death. That cell death causes a progressive loss of skills, blindness, cognitive decline, seizures, until all abilities are lost and the cellular death causes early death in the late teens to early 20s. Every day that passes without a treatment or cure is another day closer to losing my son.

[New submission]

Ataluren
It is literally needed to save my nephew's life!
I am a physician, educator, and have seen what can happen when people fail to get needed meds first hand.
Let's move forward to help people, not hurt them by denying.

[New submission]

I am 41 years old. I have 2 healthy children, and I've been diagnosed with muscular dystrophy. I've been to many MDA clinics to usually hear the same thing, that there is no cure, and you don't qualify for any treatments. But the only problem I have with that is that I'm 41 years old and I would like to see my children graduate. It's horrible for the children to see their parents died from this miserable disease. Just because the doctor won't give you Compassionate Use. Or Right to Try.

We already have CRISPR-Cas9 for 41 years. I've been chasing a cure with my parents since I was a child. I have been to many MDA association clinics I've been to... flying back and forth, wasting a lot of money and getting harder every time.

[New submission]

I was a kindergarten teacher. Teaching was what I was meant to do.

I had serious sinus infections, fatigue, and pain in my hands and feet. I went to the doctor that did a CT scan that revealed a mass in my left lung. This hospital discharged me with an appointment to see a Rheumatologist in six months. I went home, but a week later was

much worse. My family drove me to a university medical center. It was during the covid pandemic so I was alone in the hospital. No family allowed. Doctors did a kidney and lung biopsy to diagnose me with Granulomatosis with Polyangiitis. My fingers changed colors due to the lack of blood flow. The nurses could not get readings on the oximeters. They would use every finger. In June 2021, my index finger on my left hand and my pinkie finger on my right hand were amputated.

First, doctors gave me Viagra to increase my blood flow so I would not lose more fingers. Insurance would not cover this since I was a female. It cost my mom $800 for a 15 day supply. I started with 60 MG Prednisone which caused me to have mobility problems. It also caused me to gain 60 pounds. This was tapered to end before my finger amputations. Cytoxan infusions were every month for six months to kill my white blood cells. In my disease, the white blood cells attack normal organs by stopping blood flow. I have stage three kidney disease because blood was stopped to my kidneys. After my amputations, Rituxan infusions were scheduled every six months. My insurance does cover some of the cost of these infusions. I would be dead if it did not.

The infusions are $27,000 every six months. Rituxan was too expensive. My cost after insurance paid was $800. I asked my doctor to switch me to Ruxience which has a total cost of $20,000. My cost is about $400. These infusions are every six months for the rest of my life. There is no cure for my disease.

I did enter remission in 2021 during my Cytoxan infusions. If I do not get these infusions, I would flare causing my disease to become active again. Blood flow will stop going where it is suppose to go in my body. Who knows what part of the body I would lose if I did not have my infusions? I may have to be put on dialysis in the future. Many people with my disease have to get nose surgeries due to build up in their nose. I hope to stay in remission to prevent that from happening. I have my nose cleaned out by my ENT once a year. It cost $800 for that procedure.

I take medications for high blood pressure, reflux, and depression. Since my purpose in life has been taken away due to this disease, my mental health does need help. I have found a new purpose in advocating for myself and others with my disease. In the future, I hope there will be other treatments for this disease. I hope for a cure.