Written Testimony of:
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Mother of Eliza, age 16, with Sanfilippo Syndrome (MPS III)

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

Before the: U.S. Senate Special Committee on Aging, February 26, 2026

Chairman Scott, Ranking Member Gillibrand, and Distinguished Members of the Special Committee on Aging:

Thank you for your attention to the urgent, unmet needs of rare disease patients and to the 15 million children affected by rare diseases who are awaiting treatments that would allow them to grow up. I'm honored to provide testimony on behalf of our rare disease community regarding the impact that FDA processes and regulatory actions have on patients' timely access to safe and effective therapies.

My name is Dr. Cara O'Neill, and I am the Chief Science Officer and Co-Founder of Cure Sanfilippo Foundation. Cure Sanfilippo Foundation is a U.S. nonprofit dedicated to accelerating scientific development and access to a cure or therapeutic options for all affected by Sanfilippo Syndrome, driving advocacy to improve care and outcomes, and empowering families with information, guidance, and support to navigate the journey. My daughter, Eliza, is also personally affected by this ultra-rare, neurodegenerative disease, also considered a form of 'childhood dementia' or 'childhood Alzheimer's'.

While there have been advancements in rare disease policy and legal statutes over the past two decades, significant opportunities remain to address the desperate needs of 95% of the rare disease population, who still lack approved therapies. FDA review teams and leadership have tremendous responsibility and conduct complex work across thousands of disease states.

Recently, a renewed focus on FDA modernization has set forth a vision of positive change. Additionally, FDA leadership has announced several new policies and programs, including the Rare Disease Evidence Principles and, most recently, the Framework for Accelerating Development of Individualized Therapies for Ultra-Rare Diseases. We commend these efforts to add to existing expedited frameworks for drug development in rare diseases with small patient populations.

These emerging policies and programs, however, are not currently poised to help existing late-stage drug development programs and those under current review reach deteriorating patients within the urgent timeframe needed. Many rare disease communities, like ours, are at risk of losing therapies that are already demonstrating substantial benefit.

This is most evident for those with degenerative diseases, where time is the most-critical factor in accessing treatment. Due to the progressive and devastating nature of these conditions, regulatory practices must leverage every available flexibility authorized by Congress to prevent further irreversible disability and early death.

In the following testimony, you will find real-world stories about the impact of delayed time-to-treatment access in Sanfilippo syndrome (mucopolysaccharidosis type III or MPS III), a form of childhood dementia.

**Time: The Most-Critical Factor in Pediatric Neurodegeneration**

The impact of timely access can be seen in the lives of three children, all diagnosed with the same deadly form of childhood dementia, but with very different outcomes.

Izzy was 11 years old when we first met, shortly after our daughter, Eliza, was diagnosed with the same disease, Sanfilippo syndrome (MPS III). Izzy could no longer walk independently and had lost the ability to speak years earlier. She could no longer eat or drink by mouth without choking, requiring tube feedings for her nutrition. She suffered from seizures and movement disorders that twisted her arms and legs into painful positions.


Sanfilippo syndrome is caused by single-gene mutations that result in the accumulation of toxic levels of heparan sulfate, leading to progressive and irreversible brain damage.

During one of our visits, Izzy's mother shared that she had come to accept that the disease would take her daughter's life. But what she said next has always stuck with me:

*"I fear her suffering more than I fear her death."*

At that time, my daughter Eliza was around four years old and in the extremely hyperactive stage of the disease. But she still sang and talked with us. She played dress-up and clopped around the house in my high heels. She rode her tricycle everywhere. She looked healthy, but we knew that the disease was continuing to damage her developing mind and body.

Meeting Isabel put us face-to-face with Eliza's future: the concrete and cruel reality that lay ahead if she could not receive a treatment.

For Izzy, a treatment didn't come in time. Heartbreakingly, she did suffer a great deal before passing away just weeks before her 15th birthday.

We know exactly what causes the disease. We can precisely measure the levels of the toxic biomarker (heparan sulfate) to determine whether a treatment is working. And now, we have ways to treat it.

Thanks to NIH funding and support from non-profit foundations, including our own, a promising gene therapy was developed and propelled towards a clinical trial at Nationwide Children's Hospital in Ohio. Parents like us anxiously awaited news of the trial opening.

During this time, we noticed that Eliza's sentences were becoming shorter and her words were becoming less frequent. She was becoming more agitated and hardly slept. The disease was taking hold.

In May 2016, the trial finally began. Eliza, by then six-and-a-half years old, was able to receive the gene therapy at the first starting dose. We felt so lucky that she would have a chance at a future different from Izzy's; a chance to grow older and be healthy.

Now, at age 16, it's clear that the treatment changed Eliza's life. Surpassing average life expectancy, she can run on the beach and play in the water. She uses picture cards to tell us how she's feeling, and what show she wants to watch on TV. She can use a fork to feed herself and goes to school every day. Simple, but incredibly-meaningful abilities that have a positive impact on her everyday life.

Children, like Caroline, who were treated at an even earlier age and with a higher dose, have demonstrated even more remarkable outcomes. Now, at 10 years old, Caroline can read books, is on a softball team, has playdates with her friends, and even learned to ski on her family's vacation last week!

An entirely different future is in store for the few children who were able access treatment in the clinical trial. But despite these breakthroughs, and nearly a decade since the trial first began, children outside of the trial are still waiting for access - all while continuing to suffer more brain damage, month by month.

Last summer, patients' hopes were dashed when the drug was denied approval - not for safety issues or concerns about how the children were responding to the treatment, but because of questions about the manufacturing process. While an important issue, a flexible regulatory approach could have allowed the application review to proceed while addressing any outstanding questions in parallel.

Data early in the trial confirmed that the drug's mechanism was not just plausible; it is undeniably biologically effective. The levels of toxic biomarker (heparan sulfate) dropped significantly just six months after treatment in all the children. Earlier use of

Accelerated Approval based on the scientifically-sound biomarker would have changed the lives of so many children.

After being resubmitted, the clock was stopped again when the FDA requested additional paperwork before agreeing to proceed with the review.

Unfortunately, this is not an uncommon story across the rare disease community, with other diseases and conditions experiencing similar non-approvals despite strong evidence.

Congress's vision of speeding approvals for serious conditions through regulatory flexibility and the enactment of laws to enable this objective is simply not being realized for most rare disease patients and children suffering from progressive, debilitating diseases.

Transformative therapies are on the FDA's doorstep, and others are moving through the pipeline. Respectfully and sincerely, patients and their families ask decision-makers at the FDA to unlock the door.

**Sanfilippo Syndrome (MPS III)**

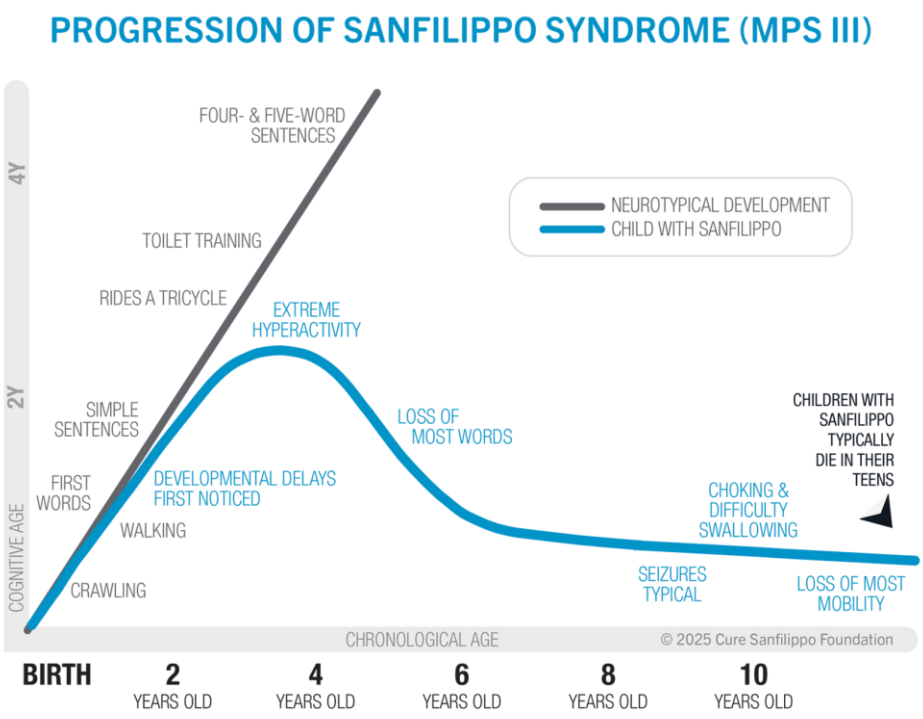


Figure represents studies of disease impact in rapidly-progressing forms of Sanfilippo Syndrome.

Sanfilippo syndrome, or mucopolysaccharidosis type III (MPS III), is a group of four devastating, rare genetic disorders causing progressive neurodegeneration and other debilitating multisystemic effects in children. Single-gene mutations lead to insufficient production of the enzyme needed to properly break down the naturally-occurring molecule heparan sulfate.

Toxic amounts of heparan sulfate accumulate, causing severe and irreversible brain damage, particularly during the sensitive neurodevelopmental periods of early childhood.

While seemingly typical during the first couple of years of life, children go on to experience broad cognitive regression and loss of basic skills such as the ability to talk, walk, and eat by

mouth. Over the ensuing years, these children suffer many other disease-related symptoms, including seizures, movement disorders, autism, significant behavioral and sleep disturbances, hearing and vision loss, and, in the most-common form of the disease, have an average life expectancy of just 15 years.

There are no FDA-approved treatments for any form of Sanfilippo syndrome.

**Regulatory Flexibility is Essential in Rare Disease Drug Development**

Over the past four decades, Congress has codified the FDA's authority and obligation to exercise regulatory flexibility for serious and life-threatening conditions and rare diseases. These regulations highlight several key provisions:

1) The development of medicines for rare diseases requires modified frameworks and innovative trial designs;
2) The FDA has the authority to use flexible evidentiary standards;
3) The FDA has been directed to consider disease severity and unmet need when evaluating benefit-risk; and
4) The FDA has been directed to incorporate patient perspectives.

In 2025, rare genetic neurodegenerative disease programs experienced a marked increase in Complete Response Letters (CRLs), an official communication from the FDA to a drug sponsor indicating that the review cycle for a marketing application (NDA, BLA, or ANDA) is complete, but the product cannot be approved in its current form.

Our analysis of publicly-available FDA data and sponsor disclosures identified seven CRLs and only two approvals for these rapidly-progressive conditions, compared with four approvals out of five applications in the prior year. Many of the affected programs were pursuing Accelerated Approval.

The deficiencies cited in the CRLs raise the question of whether the FDA is indeed using its regulatory flexibility in rare diseases. In 2025, CRLs increasingly emphasize trial design and evidentiary rigor over well-established, disease-causing biomarkers, such as heparan sulfate in MPS disorders. In ultra-rare, heterogeneous diseases with very small patient populations, trials will never resemble those conducted in common diseases. Requiring perfection in such settings risks denying progress altogether.

Increased Utilization of Accelerated Approval is Needed

Key Congressional legislation created the Accelerated Approval pathway, enabling regulatory flexibility in the evaluation of treatments for serious, life-threatening conditions. While the Accelerated Approval pathway is not exclusive to rare diseases, it is well-suited to address the needs of serious and life-threatening rare diseases. However, to date, 80% of drugs approved under Accelerated Approval have been for cancer

therapies. Based on an internal analysis of the last two years, only 12% of non-cancer rare diseases received approval via this accelerated pathway.

There remains a gap between therapies that demonstrate biologic plausibility and a regulatory system that struggles to translate that plausibility into timely, consistent decisions. Recent FDA statements and draft guidance acknowledge the mismatch between standards appropriate for common diseases and those feasible in ultra-rare conditions. Draft guidance is an important step. However, meaningful impact will depend on consistent implementation of both existing statutes and new initiatives. Regulatory flexibility must be applied not only in theory, but in practice.

The Accelerated Approval pathway, as codified in 21 CFR 314 Subpart H and 21 CFR 601 Subpart E, expressly permits FDA approval based on surrogate endpoints, such as biomarkers or intermediate clinical endpoints, that are reasonably likely to predict clinical benefit.

The use of biomarkers is an essential approach to achieving FDA approval for conditions that do not fit traditional drug-development frameworks.

Acceptance of heparan sulfate as the primary disease-causing biomarker for Sanfilippo syndrome (MPS III) and other forms of neurologically impairing MPS (MPS I, II, VII) reached consensus through a scientific convening of the Reagan-Udall Foundation for the FDA in February 2024. Key opinion leaders affirmed that:

- Elevated heparan sulfate is the primary pathologic cause of disease.
- Natural history data demonstrate a predictable pattern of cognitive decline.
- Changes in cerebrospinal fluid (CSF) heparan sulfate levels are reliably measurable and reasonably reflect brain tissue heparan sulfate accumulation.
- A reduction in heparan sulfate levels reflects the biological activity of drugs targeting the primary disease mechanism.

Manufacturing Process Flexibility

Regulatory flexibility must extend beyond clinical trial design to all aspects of the review, including Chemistry, Manufacturing, and Controls (CMC). CMC requirements for gene therapies are inherently complex and manufacturing processes evolve and require refinement over time.

In the case of a Sanfilippo syndrome gene therapy and a number of other rare disease treatments, letters denying approval (CRLs) or communications delaying regulatory processes cited only CMC concerns, not safety or efficacy concerns. Dosing of patients in the Sanfilippo gene therapy clinical trial was allowed to continue, reassuring that the agency continued to consider the drug product safe for use in children.
Despite established safety, the path towards approval is currently on hold again as the agency requested additional paperwork that is typically reserved for review during the

time of on site inspection. Timelines that are delayed due to administrative burdens further limit children's access to treatment, leading to more neurological damage.

Product safety and consistency are essential, however, Congress recognizes that therapies for life-threatening diseases may require a flexible, risk-based approach. This includes post-approval commitments to address manufacturing questions. When therapies demonstrate promise in progressive diseases, we must allow regulatory flexibility in CMC reviews to prevent delays in getting treatments to children.

Trial Designs that Delay Treatment Lead to Irreversible Brain Damage

Cognitive decline and neurodegeneration are central features of many pediatric rare diseases, including Sanfilippo syndrome. Brain development unfolds within well-defined, time-sensitive windows from early childhood through adolescence. During these critical periods, exposure to toxic substrates, such as heparan sulfate, has particularly harmful and irreversible effects. In Sanfilippo syndrome and other neurological MPS diseases, continued accumulation of heparan sulfate toxin directly damages the central nervous system.

The FDA has appropriately acknowledged that traditional, large, randomized-controlled trials are often infeasible in very small patient populations. Yet sponsors of ultra-rare pediatric programs continue to report regulatory requirements for randomized or "no-treatment" comparator arms, even in progressive neurodegenerative conditions. In practice, this means some children are knowingly assigned to sustained periods without therapy, during which irreversible brain injury will certainly occur.

When a primary disease-modifying therapy is responsive to a scientifically-sound biomarker for a neurologically progressive condition, innovative trial designs are an ethical necessity. External comparators, adaptive designs, patient-as-their-own-control, and biomarkers as surrogate endpoints can provide rigorous data for efficacy evaluations while minimizing preventable harm. In diseases in which neurons are lost daily, clinical trial design must adhere to both scientific standards and the biological reality of irreversible decline.

Consider a little girl from Pennsylvania named Liv. In a previous clinical trial, the protocol required an observation period of one year during which children were not given treatment, prior to them being switched over to the treatment arm of the trial. During that year while awaiting treatment, Liv suffered irreversible developmental regression.

Today, similar programs targeting neurocognitive disease symptoms face requirements for randomized control arms in small pediatric neurodegenerative disease populations.

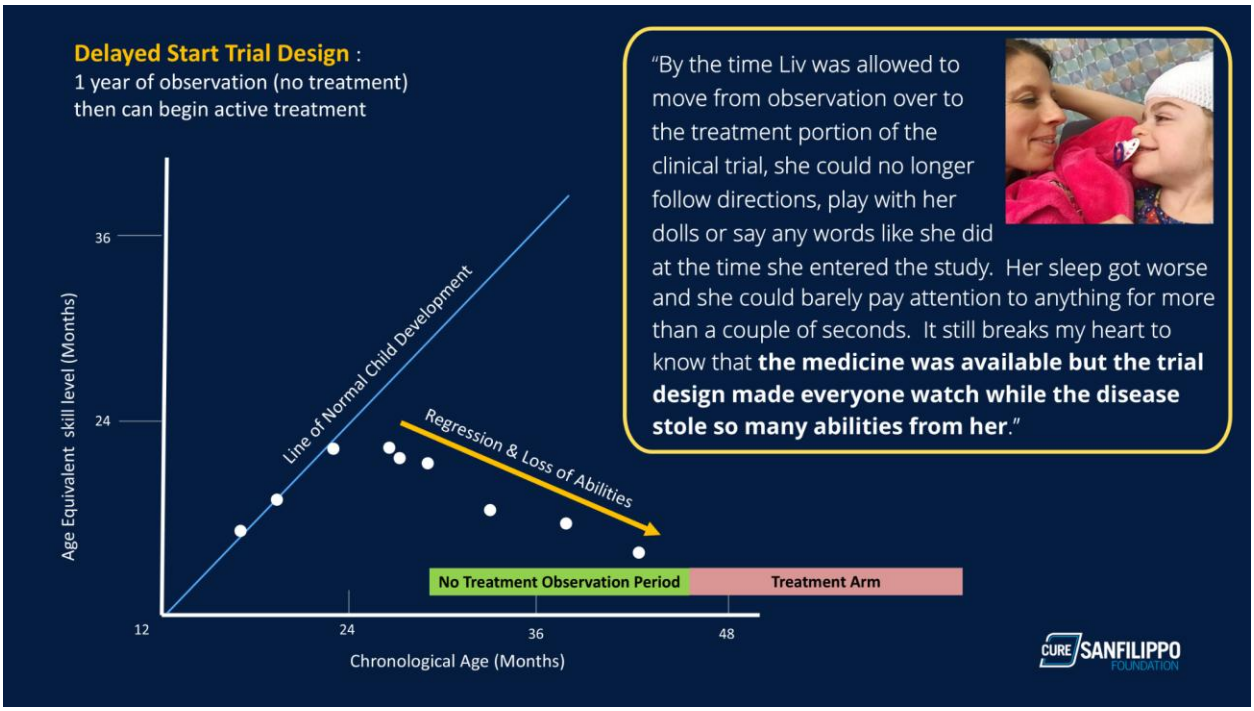



**Image depicts Loss of cognitive abilities during the delayed start period of an enzyme replacement clinical trial.*

**Importance of Patient Representation in Drug Development & Regulatory Review**

The 2012 reauthorization of the Prescription Drug User Fee Act (PDUFA V) formally advanced the concept of Patient-Focused Drug Development (PFDD), signaling Congress's intent that patient experience be systematically integrated into regulatory decision-making. In 2016, Congress enacted the landmark 21st Century Cures Act, which further codified PFDD into the FDA's statutory framework, reinforcing that patient input is not optional but core to the agency's mission. Since that time, patient organizations and advocates have become increasingly engaged in drug development activities, and the practice of PFDD continues to be adopted more widely.

Meaningful patient engagement strengthens regulatory science across the continuum of drug development. Patients and caregivers bring uniquely-valuable, disease-specific, lived experience necessary to inform study feasibility, treatment targets, degree of change constituting meaningful benefit, realistic risk tolerance in life-threatening conditions, and broader risk/benefit considerations.

FDA has made progress in developing and hosting patient engagement efforts such as the Patient Engagement Collaborative, internally and externally-led PFDD meetings, and Listening Sessions. It is unclear whether FDA's Patient Representative Program is still active, or to what extent representation is filled in rare disease review divisions. Further, a reduction and halting of product-specific advisory committee meetings has occurred.

Additional Opportunities to Integrate Patient Representative Insight

1) **Re-prioritize advisory committees:** Advisory committees provide a transparent venue for scientific dialogue among key stakeholders. These are typically public forums and provide a window into complex regulatory decisions. Last year, the number of ad-coms decreased substantially. Integrating patient/caregiver perspectives and real-world expertise into scientific discussions allows decision-making to be rooted in the primary goal of public benefit while also providing important case example learnings for the entire drug development community.

2) **Revive and/or expand FDA's Patient Representative Program or councils:** Revive and/or expand formal, standing councils of trained patient representatives to engage with FDA review divisions on an ongoing basis. These councils could provide rare disease-specific insight into lived experience, trial feasibility, acceptable risk, and what constitutes meaningful benefit from the patient perspective.

   Unlike one-time listening sessions, these councils may operate longitudinally, allowing for structured, bi-directional engagement through the development lifecycle (e.g., early trial design, endpoint selection, and post-approval or CRL). This would build on, but not replace, other PFDD initiatives by moving from one-time input to continual partnership. Working through the Rare Disease Hub would allow focused coordination to serve the rare disease community.

**The Economic Reality of Ultra-Rare Drug Development**

There are currently no approved treatments for any type of Sanfilippo syndrome, despite the disease being discovered in 1963. In ultra-rare diseases, early medical research and even clinical trials are often partially funded by family efforts - lemonade stands and 5K races - to support non-profit advocacy foundations funding research. NIH funding is also instrumental in supporting basic and translational steps needed to reach the point of a clinical trial. In recent years, NIH has created critical clinical trial funding sources, though these are limited.

Moving basic science to a clinical trial requires many millions of dollars. In rare diseases with very small populations, large pharma is often not interested due to the complexities, uncertainty, and limited returns involved in rare disease drug development. Therefore, companies involved in rare disease are often small, startup biotechs.

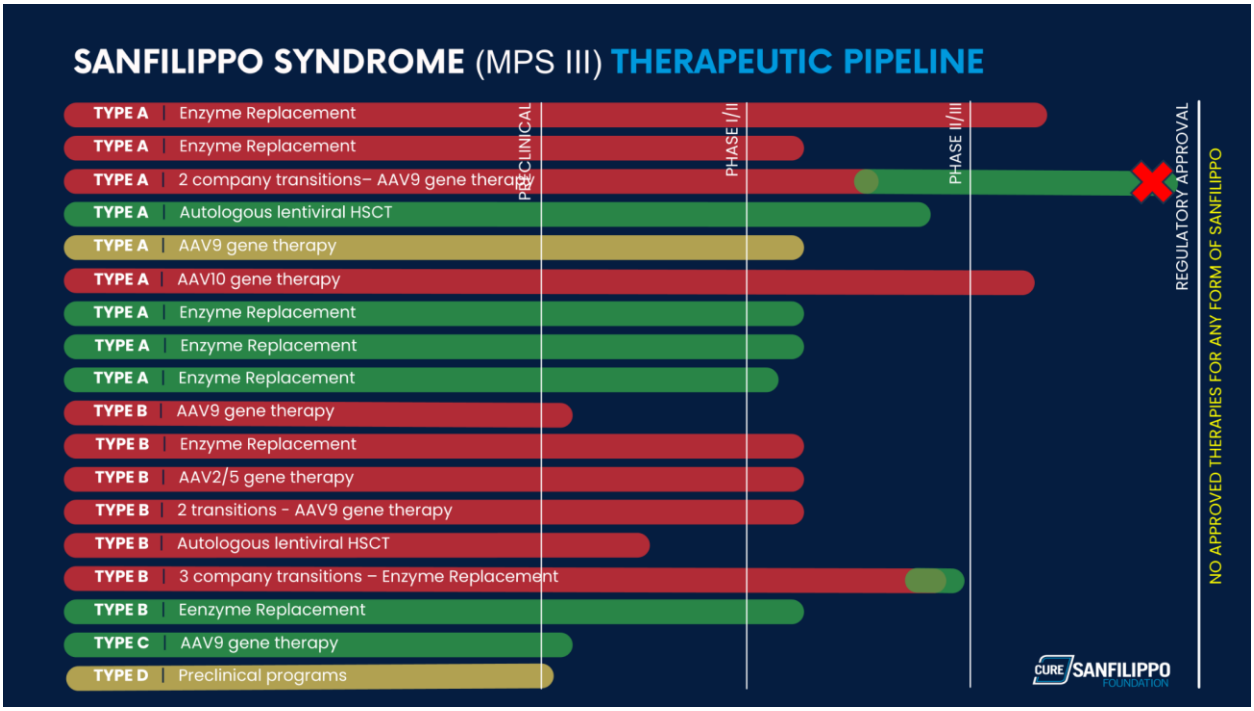
Over the last 10 years, we've seen eight promising Sanfilippo syndrome therapeutic programs abandoned. Many of the companies developing these drugs cited the financial burden of extended, uncertain regulatory timelines and pathway as reasons for shelving promising treatments. A number of these companies have gone out of business while attempting to maintain

their programs in Sanfilippo. Shifting regulatory opinion on study comparator arms, study design, and recognition of meaningful surrogate biomarker change lead to further uncertainty among investors and companies.

Parents have watched their children stabilize and continue to grow and learn while receiving treatment in clinical trials, only to have that promising treatment taken away. Most of these children never get access to another treatment.

Sanfilippo syndrome is not an isolated case. Many rare disease programs face similar obstacles. Small biotechnology companies, which develop the majority of ultra-rare therapies, often cannot withstand prolonged regulatory uncertainty, shifting evidentiary expectations, or extended review timelines.

Without FDA's greater use of flexibility, consistency, and transparency, we will continue to see companies turn their backs on rare disease drug development.




** Image depicts Sanfilippo syndrome therapeutic landscape*

## Proposed Actions and Conclusion

We commend FDA leadership for their numerous public statements supporting rare disease treatments and for developing new initiatives, such as the Rare Disease Evidence Principles and draft guidance for individualized therapies. While new programs are welcome, Congressionally codified programs already exist that offer the regulatory flexibility required to advance urgently-needed therapies and can be utilized to address the needs of patients today.

We respectfully request Congressional oversight to ensure consistent application of existing statutory flexibility:

1. **Use Accelerated Approval as Congress intended.** When a treatment clearly improves the underlying biology of a fatal rare disease, approvals should enable timely access before further irreversible disease progression.
2. **Increase acceptance of innovative trial design in rare diseases** where large or randomized trials are not feasible or ethically appropriate.
3. **Apply flexibility across all aspects of the rare disease review process**, from trial design to biomarkers to manufacturing.
4. **Provide clear, consistent expectations early in development** to build trust that regulatory opinion and guidance will not shift in later development stages.
5. **Increase transparency across the FDA review process** by integrating the patient representation into all aspects of drug review and reprioritizing advisory committees.

In children with progressive neurodegenerative diseases, biology does not wait for policy implementation, administrative refinement, or prolonged debates over biomarkers. These conditions move forward every day. Brain cells die. Abilities are permanently lost.

We are not asking for lower standards. We are asking that the standards and flexibility Congress has already authorized be applied consistently, transparently, and proportionately in the context of ultra-rare, life-threatening disease. Regulatory flexibility, including Accelerated Approval and use of biomarkers, are not concessions; they are tools deliberately created for situations where traditional development paradigms do not fit.

Please accept our gratitude to this Committee for elevating the unmet needs of patients suffering from rare diseases. We hope that you will continue your support in ensuring that the regulatory framework already in place is used with clarity, consistency, and urgency. For children facing irreversible neurodegeneration, time is the most precious and limited resource. Our shared responsibility is to close the gap between scientific possibility and regulatory execution so that no child is left waiting for a therapy that could alleviate suffering and save their life.

> *"You have the power to move with urgency. Please do not make families like mine wait while our children disappear in front of us. Because while you are deciding, I am watching my son disappear."* - Beckham's father from Georgia

More comments from families of children with Sanfilippo syndrome in the **Appendix** below.

## Appendix: Family Comments

Below are comments provided by parents of children with Sanfilippo syndrome (February 2026)

## What does time mean for you and your child with Sanfilippo syndrome?

A delay of 6 to 12 months means losing parts of my son that I will never get back.
Every single second matters for Beckham. 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 Beckham.
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 uses to take my son from me.

Brandon Hutcheson, Georgia
**Child: Beckham "Bex" Hutcheson**, age 4

Time is not on our side with Sanfilippo Syndrome. With each moment that passes, more brain damage occurs. Delays of six or 12 months or so means that Simon loses more words, more cognitive ability, and more memory. It means that Simon's neuromuscular scoliosis, avascular necrosis, and bilateral sensorineural hearing loss worsen. Resulting pain, agitation, and hyperactivity increase. Inflammation and oxidative stress continue to drain the light from our Simon's beautiful face. Sadness and confusion take over as he loses his abilities to count, spell, identify colors, dance, and run.

Alina Gorniak, Texas
**Child: Simon Croke**, age 9

Since FDA delay of UX 111 in July 2025, Lottie has already lost many more ASL signs and word approximations. Lottie no longer uses any ASL sign or sound consistently. Lottie has forgotten several songs that she used to get excited for. A delay of six more months will most likely mean that Lottie will need a G-tube, we anticipate seeing fewer laughs, and we believe we will continue to see a decline in her receptive skills as well.

Abby Milburn, IL
**Child: Lottie**, age 5

Delays mean that my daughter Veda can no longer speak. She doesn't color anymore or dance and sing to her favorite songs. Her body is starting to slow down and I see my daughter fading when I look into her eyes. I don't want this to happen to other families. Our children need a treatment when they are born, not when Sanfilippo has already started to steal them away.

Christin Ulrich, Florida
**Child: Veda Ulrich**, age 6

In just 6 months, Jacob has lost his ability to eat solid foods without struggling though each bite. It takes Jacob an hour to eat dinner. Eating is one of the things that Jacob has always enjoyed, and something as a family we have always celebrated. He is also much less mobile. While he attends our local high school in a special program, he is not as actively involved as he was just last spring.

Christine Moon, New York
**Child: Jacob Moon**, age 16

Time, for our family, is not measured in years. It's measured in windows.
Our son Emmett has Sanfilippo Syndrome, a progressive and terminal neurological disease. He has not started seizures yet. He is still gaining some skills. He still has some words. But we are seeing the slowing,  fewer words, subtle physical decline. We know what comes next.
A delay of 6 or 12 months is not a minor setback for children like Emmett.
Six months can mean lost brain cells that cannot be recovered.
Twelve months can mean the difference between treating a child who is walking and talking and one who no longer can.

Stephanie McCabe, Saskatchewan, Canada
**Child: Emmett Doucette**, age 6

My name is Ashley, and I am the mother of Sadie, who has Sanfilippo syndrome, a progressive and fatal neurodegenerative disease.
At this stage, Sadie is actively losing skills. Over the past 2 years, she has lost most of her speech, memory and had a significant cognitive decline. These losses are permanent.
A 6–12 month delay in treatment is detrimental—it results in irreversible decline. During that time, children like mine can lose communication, cognitive function, and the ability to safely engage with the world. By the time a treatment becomes available, it may be too late for it to provide meaningful benefit.
For our children, a delay is not just time—it is loss.

Ashley Haywood, NC
**Child: Sadie**, age 9

Further decline of cognitive abilities & functional abilities like speaking, eating/drinking, and ambulation

Krystal Cooley, SC
**Child: Dawson**, age 7

Time, for us, is not measured in years. It is measured in losses.
Sanfilippo Syndrome is progressive and unforgiving. Every 6 or 12 months of delay means more skills lost, more regression, more pain, more sleepless nights, and fewer abilities my children may ever regain. It means watching communication fade, mobility decline, and personality slowly disappear.
For most families, a year is time to plan. For us, a year can mean the difference between walking and not walking, speaking and never speaking again. Delays are not administrative, they are irreversible.

Soraya, Al Chouf Baakleen Lebanon
**Children: Sama Chaaban and Aram Chaaban**

They are just losing their capabilities, skills and health… until they get disabled or die after just because there is No cure for Sanfilippo type A syndrome

Khaled Chaaban, Lebanon
**Children: Sama Chaaban & Aram Chaaban**

With Sanfilippo syndrome, time is on our side. A six-month delay can cost a child the ability to speak, and a year the ability to move. For us, time isn't an abstraction, but rather concrete, lost skills that cannot be regained. Every month of waiting reduces the chances of effective therapy and increases the burden on the family. We don't ask for time; we fight for it, because for our child, every day counts.

Eugenia Sotnikova, Novosibirsk city, Russia
**Child: Alexandr Sotnikov**, age 7

FDS delays will cause further regression and cognitive delays for individuals with sanfilippo syndrome. The accumulation of toxic heparin sulfate continues to cause significant damage each and every day. The future for my two daughters is very uncertain. Seizures may return, language may be lost, motor skills will diminish and quality of life will deteriorate for Margaret, Bridget and our whole family.

Kathy Lindquist, New York
**Children: Bridget & Margaret Lindquist**

He will lose the ability to talk and can’t make any progress

Astrid, Germany, Bavaria
**Child: Charlie**, age 4

Bir aylık gecikme bile bizim için çok uzun bir sure (Even a one-month delay is a very long time for us.)

Mehmet vural, Türkiye
**Child: Zeren**, age 6

Leni is just two years old and full of love, laughter and life. She has not yet experienced the regression, brain damage, and very worst symptoms of Sanfilippo Syndrome. If she is able to access treatment before this begins (usually around 3 years old) she has the chance of a near normal life. Once the regression begins it cannot be reversed. Sanfilippo Syndrome is rapidly neurodegenerative and relentless. A delay of 6 months could be the difference between our sweet girl losing her ability to talk or not, and a delay of 12 months could be the difference between her losing her ability to walk or not. Sanfilippo Syndrome does not respect approval

timelines, these children need treatment now to give them the best possible chance at life - they can't wait. Delays also have a huge mental impact on the parents, families and loved ones of these children. We are advocating tirelessly for Leni to get access to treatment before it is too late. It is exhausting fighting for your child's right to live every single day and shifting timelines are absolutely heartbreaking when the impact of this is so catastrophic. Every day that Leni cannot access treatment toxic waste builds up inside her tiny body. We are watching her grow and develop into a beautiful little human with the haunting knowledge that if she is unable to access treatment and quickly she will lose everything, including her life. Time is precious, and time is our enemy.

Emily Forrester, Kent, UK
**Child: Leni Forrester**, age 2

Time is everything!  6-12 months could result in loss of skills, new medical issues, and the potential for a medication to now not be effective when it could have made a meaningful difference would it have been administered sooner.

Lauren Barber, Michigan
**Child: Autumn Barber**, age 7

Time is scarce with Sanfilippo. We know that the earlier Louisa can get access to a therapy or a clinical trial the better and greater the chances are for her to progress positively and not to lose skills. Time is also scary with Sanfilippo as since the diagnosis, the disease feels like a Sword of Damocles hanging over our family, it feels like a race against time. 6 months or 12 months feel like an eternity now.

Lennart Sieweke, Potsdam, Germany
**Child: Louisa**

Right now, Lydia has no regression. The next 6 months are crucial for her as it's the expected to begin to decline with far more brain damage taking away her quality of life. There are treatments & science out there that give these children a better quality of life, that will be filled with pain & suffering without it. It's our only chance of hope for our child.

Morgan Rachal, Louisiana
**Child: Lydia Rachal**, age 3

Frankie used to wake up every morning singing. She was full of joy, energy, humor, and affection. She had rich language, spoke in sentences, loved to sing, and even had funny jokes. She gave the most amazing running hugs. It only took Sanfilippo Syndrome 3 months to take it all. Over the course of 3 months, all of Frankie's words and songs were taken. She didn't know how to engage in play anymore, and she struggled to pick up a toy. She could no longer feed herself independently. She used to climb rock walls, and then she struggled to know what to do on a playground. At 5 years old, Sanfilippo Syndrome stole our daughter and in just 3 months. To us, 6 or 12 months means everything.

Gabrielle Price, Washington
**Child: Frankie Price**, age 6

Looking back 6 months is looking at a different child of mine. Looking back 12 months makes the difference even more stark. In 6 months, my child has lost her ability to communicate even more so than she did over the previous 6 months. She continues to grow in discomfort and what seems like pain as the disease continues to progress throughout her body. Delays of any kind mean simple experiences with her now could be taken away permanently.

Andrew Price, Washington
**Child: Frankie Price**, age 6

Living with Sanfilippo Syndrome means living against a clock that never stops ticking. While other families plan for the future in milestones gained, we brace ourselves for milestones slipping away. Time is not something we take for granted — it is something we fight for.
Six months may not sound like much in the world of drug development, regulatory timelines, or funding cycles. But for our child, six months can mean:
-the difference between speaking in sentences and struggling for words
-the difference between walking independently and needing support
-the difference between sleeping through the night and relentless exhaustion
-the difference between understanding us and slowly drifting away
Twelve months can mean even more. It can mean losing skills that will never come back. It can mean further cognitive decline. It can mean more seizures, more behaviors, more regression. It can mean watching your child change in ways no parent should have to witness.
For families like ours, delays in research, clinical trials, or funding are not abstract timelines. They are lived in real time. They are birthdays passed. Holidays changed. Abilities fading. Time is everything.

Christiane von Rosbitzki, Germany/Hessen/Rödermark
**Child: Theresa von Rosbitzki**, age 4

Time for a child with Sanfilippo is not abstract. It is physical. It is visible. It is measurable in lost words, lost abilities, lost moments. My daughter Payton was diagnosed at age five. She is eight now, and every day we watch pieces of her slip away. Time is not neutral for families like ours—it is a force that actively harms our children.

A six-month delay in access to a therapy for most conditions might be frustrating. For Sanfilippo families, six months can mean the difference between a child still being able to walk independently or not. A year can mean losing the ability to speak, to feed themselves, to sleep through the night, to recognize loved ones. These are not hypothetical possibilities; for us, they are the reality of the disease's trajectory.

When we hear "six months," we do not think of a bureaucratic interval. We think of six months of skills eroding. Six months of watching Payton's world shrink. Six months closer to losing abilities she will never get back. Six months of our family grieving in advance for what Sanfilippo will take next.

Twelve months feels like an entire chapter of her life that could have been lived differently—with more comfort, more connection, and more dignity—if safe and effective therapies were accessible when they are ready instead of after prolonged, unintended delays. For us, time is not just precious; it is life-altering. Delays take from our children what no therapy can restore.

Ally Geronzin, Arizona
**Child: Payton Geronzin**, age 8

I watched my daughter loss her ability to communicate in a 4 month span, -going from saying, "Mom, come on" to nothing. Currently, she loves food and is still able to eat and she loves to run and is still mobile. Those skills could easily slip away in a few months without treatment.

Rebecca Jordan, Ohio
**Child: Liv Jordan**, age 11

Loss of words, increasing headaches and aggression, loss of mobility, worsening gi issues, toileting accidents. His personality is fading with every passing day.

Nancy Rubino, Massachusetts
**Child: Merrick Rubino**, age 11

## What does a chance at a better quality of life look like for your child and your entire family? What would be different in their and your lives?

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 Beckham'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 Beckham 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.

Brandon Hutcheson, Georgia
**Child: Beckham "Bex" Hutcheson**, age 4

A clinically proven treatment would mean that the progression of his disease would slow. He'd be in less pain, experience less agitation, and hyperactivity. Simon would hold on to the skills that he worked so hard to achieve like playing basketball, putting together floor puzzles, and building with blocks. Our family's lives would be filled with more joy and happiness than stress, anticipatory grief, and heartbreak.

Alina Gorniak, Texas
**Child: Simon Croke**, age 9

Currently, Sanfilippo syndrome affects all parts of our lives. Lottie is the oldest of our four children. Her younger siblings even are one year-old are all more independent and less in need

of the constant monitoring that Lottie is in need of. In general simply syndrome hangs over our family like a dark cloud. Some days the grief is already so immense it makes doing any mundane activity astronomically hard.
Better quality of life for Lottie could mean so many things. One improvement would be getting to independently feed herself safely and for a longer period of time. Another improvement to her quality of life would be better sleep so that she experiences less daytime, exhaustion, and is able to have more fun with her younger siblings and to just be a kid. In general, if Lottie could receive treatment of some kind, I think that it would relieve some of the heaviness that we have felt since receiving lots of diagnosis and overall improving the quality of life for our entire family.

Abby Milburn, IL
**Child: Lottie**, age 5

Our daughter can't tell us where her pain is. With earlier treatment we might have been able to keep her speech so she could tell us where she is having pain. Now we have to rely on body language to figure out where she is hurting. Nothing is worse than the helpless feeling of hearing your child cry and not knowing what is wrong or how to help.

Christin Ulrich, Florida
**Child: Veda Ulrich**, age 6

Jacob was diagnosed on the "late" side. He was 7. I always say how ridiculous at 7, it was a late diagnosis, but in this world it was late. Our entire focus for Jacob has been maintenance. We are extremely lucky to have a support team that understands the importance of maintaining all of Jacob's skills. So for Jacob to continue to maintain his voice, his mobility, his ability to participate in everything his family and friends do are vital. If Jacob's sleep could improve everyone's quality of life would improve!

Christine Moon, New York
**Child: Jacob Moon**, age 16

It means fewer sleepless nights because his brain is calmer.
It means less pain and discomfort he cannot explain to us.
It means preserving the words he still has — or even gaining a few more.
It means holding onto his ability to walk, to laugh, to engage with his sister.

Right now, we live in anticipation of decline — seizures, loss of mobility, loss of communication. A better quality of life means slowing that progression. It means giving him more time as the child he is today.
For our family, it would mean breathing a little easier.

Stephanie McCabe, Saskatchewan, Canada
**Child: Emmett Doucette**, age 6

For my child living with Sanfilippo syndrome, a better quality of life would mean less pain, more consistent sleep, and the ability to hold onto the skills they still have.
It would look like being able to communicate basic needs instead of crying in frustration. It would mean fewer behavioral challenges driven by discomfort they cannot express. It would mean moments of connection that last longer—recognition, interaction, and engagement with the world around them.
For our family, it would mean stability where there is currently constant decline. It would mean fewer sleepless nights, less medical and safety crisis management, and more time simply being present together.
A chance at better quality of life does not mean expecting a cure. It means preserving what is still there for as long as possible—and reducing suffering in the time we have.
That would change everything.

Ashley Haywood, NC
**Child: Sadie**, age 9

It breaks my heart to see my child hurt or struggle in school and at home with ADLs so anything that helps would improve our families quality of life. We want our kids to have a normal life and experience all the things other kids get to experience

Krystal Cooley, SC
**Child: Dawson**, age 7

A better quality of life would mean less suffering, less neurological pain, better sleep, fewer behavioral crises, and the possibility of preserving the skills my child still has.
It would mean hearing their voice longer. Seeing them hold onto the ability to recognize us. Having moments of connection that are not overshadowed by decline.
For our family, it would mean living with hope instead of constant anticipatory grief. It would mean being parents again not just caregivers managing degeneration. It would mean stability, dignity, and the chance to build memories that are not defined by loss.

Soraya, Al Chouf Baakleen Lebanon
**Children: Sama Chaaban and Aram Chaaban**

Their life is essentially changing from a happy kid to sadness and dullness since they are losing almost everything. Walk, chewing, focusing, speaking and their senses.

Khaled Chaaban, Lebanon
**Children: Sama Chaaban & Aram Chaaban**

For us, improving our quality of life means restoring contact and communication. We would no longer have to guess why our child is crying. Understanding our child's needs would reduce our sense of helplessness.

Eugenia Sotnikova, Novosibirsk city, Russia
**Child: Alexandr Sotnikov**, age 7

For Margaret & Bridget a better quality of life means the difference better stabilizing their skills or losing them.  Margaret & Bridget would require increase level of care and increase of support. As working parents it would be life changing for my entire family.

Kathy Lindquist, New York
**Children: Bridget & Margaret Lindquist**

He can learn, he can play with his cousins and friends, he can dance and sing. All the things he loves to do right now

Astrid, Germany, Bavaria
**Child: Charlie**, age 4

Hereey çok güzel olurdu ama en önemlisi kızımız artık hayata daha güzel bakardı (Everything would be wonderful, but most importantly, our daughter would have a more positive outlook on life.)

Mehmet vural, Türkiye
**Child: Zeren**, age 6

Our family are faced with two totally different futures - one is extremely dark if Leni is unable to access treatment now, and the other is a near normal life. As parents it is our responsibility to protect our child and give them the best possible life - and without treatment options we are unable to protect our baby. If Leni is unable to access treatment she will lose the ability to walk, talk, run, play, laugh, eat and eventually die in her early to mid teens. She will suffer physical pain, mental distress, seizures and insomnia that can last for days. As a family we will have to cope with all this, whilst navigating a gruelling schedule of medical appointments and trying to give our little girl the best possible life. If she is able to access treatment in the next year Leni and our family will be able to lead a near normal life. The difference is dark and light, life or death.

Emily Forrester, Kent, UK
**Child: Leni Forrester**, age 2

Sanfilippo is a terrible disease.  As a family we dread watching Autumn suffer even more than her death.  Anything to help improve her quality of life could make a HUGE difference for our family regardless of if it extends her life or improves her cognitive function.  The sleep deprivation, pain, and frustrations from lack of communication result in extreme caregiver fatigue and suffering for Autumn and our entire family.

Lauren Barber, Michigan
**Child: Autumn Barber**, age 7

Less symptoms would mean for Louisa to have a more normal life, a life that every child deserves. A carefree life and a life where she doesn't stand out in a negative way because of her hyperactivity, impulsivity and lack of focus. For us as parents it is simply heartbreaking to see our child suffering, or being underdeveloped versus other same aged children in terms of speech or worse, to see Louisa being excluded sometimes due to behavioral issues, because she is perceived as being "too much".

Lennart Sieweke, Potsdam, Germany
**Child: Louisa**

Lydia can talk to us, laugh with us, play with us.. a typical toddler. With the science and drugs coming about, this wouldn’t take her childhood away from her. Her childhood wouldn’t be filled

with pain & suffering. She would be able to retain skills she has & keeping playing along side of her big sister.

Morgan Rachal, Louisiana
**Child: Lydia Rachal**, age 3

Learning that our daughter has a terminal diagnosis absolutely crushed us. To learn that not only is it terminal, but that you'll have to endure losing her piece by piece and face the extreme challenges of caregiving -- I still don't have the words for the cruelty of it all. Sanfilippo Syndrome causes great suffering for both the child and the family. A chance to ease that suffering and give a better quality of life is something we'd do anything to have. For us, it looks like less pain for Frankie, the chance to see her smile more, less distress for the whole family, better sleep for the whole family,  and her retaining any skills that she has. If our days with Frankie have to be numbered, we just want them to be as good as possible.

Gabrielle Price, Washington
**Child: Frankie Price**, age 6

Frankie isn't able to smile much these days. I hope that doesn't mean she doesn't feel joy or excitement. I hope that those feelings are happening inside herself just without a way to be expressed. A chance at a better quality of life could give her simple pleasure. It would mean freedom from restlessness, pain, and discomfort as well as the opportunity to express herself in ways that she isn't now.

Andrew Price, Washington
**Child: Frankie Price**, age 6

A chance at a better quality of life would change everything — not just for our child, but for our entire family.
For our child, it would mean comfort.
Less pain. Fewer restless nights. A body that feels calmer instead of constantly overwhelmed.
It would mean holding on to skills instead of losing them. Keeping words. Keeping understanding. Keeping the ability to connect with us.
It would mean more time where our child feels safe in their own body.
More laughter.
More connection.
More life — lived with less suffering.

Christiane von Rosbitzki, Germany/Hessen/Rödermark

**Child: Theresa von Rosbitzki**, age 4

Even small improvements would change everything.
A therapy that could lessen Payton's pain, help her sleep, or slow the daily decline would not only improve her life—it would reshape our entire family's reality. Better sleep means she could wake up more regulated, and so could we. Less pain means more comfort in her day, fewer moments of distress that she cannot communicate, and more opportunities for her to participate in family moments.
Retaining skills—communication, mobility, feeding—would extend her independence. It would allow her to keep engaging with the world, even in small ways. The ability to express a want, a feeling, or a need is something many families take for granted, but for us, even a little more connection would be life-changing.
For our family, improved quality of life doesn't mean a cure. It means preserving the joy she still has. It means fewer emergency interventions and less fear. It means being able to plan beyond the immediate crisis. It means allowing Payton to experience more of her childhood, instead of watching it disappear sooner than it should.
A therapy that could give us even modest improvements would not just lessen suffering—it would give us back moments we're losing far too quickly.

Ally Geronzin, Arizona
**Child: Payton Geronzin**, age 8

Spending quality time with your family is what life is all about. Feeling well and having a good nights rest are critical firsts.

Rebecca Jordan, Ohio
**Child: Liv Jordan**, age 11

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.

Nancy Rubino, Massachusetts
**Child: Merrick Rubino**, age 11

## How would you weigh the benefits and risks that could come with potential and promising therapeutic options?

I know what the outcome of Sanfilippo Syndrome is. I know this disease will take my son's life. I am already watching it take pieces of him every day. I have watched him lose his words. I have watched him lose his independence. And I know, if nothing changes, it will continue until it takes him completely.

So when I think about risk, I think about it differently. The greatest risk is doing nothing.

Doing nothing means accepting that my son will continue to suffer and that he will die from this disease. That is not something I am willing to accept without fighting for him.

Any potential treatment represents a chance. Not a guarantee—but a chance. A chance for him to keep what he still has. A chance for him to stay connected to his brothers. A chance for him to have more time with the people who love him.

I understand there are risks with any new or promising therapy. But there is also certainty in doing nothing. And that certainty is losing my son.

As his father, I am willing to accept reasonable risks if it means giving Beckham a chance at more life, more comfort, and more time with his family.

I cannot stand by and watch this disease take him without trying everything possible to help him.

Beckham deserves that fight. And I will never stop fighting for him.

Brandon Hutcheson, Georgia
**Child: Beckham "Bex" Hutcheson**, age 4

The natural progression of Sanfilippo Syndrome ensures that our son passes away by his 18th or 19th birthday, and since he's already 9, the latter years of his are going to be far from pleasant. Without treatment, his fate is to die of this disease before he completes his second decade of life. So, we are eager to try a promising treatment that could only improve his quality of life.

Alina Gorniak, Texas
**Child: Simon Croke**, age 9

As a family, we are very willing to try therapeutic options that come up. We are often asking Lottie's doctor about potential repurposed medication's, and are always looking into other types of holistic medicine when it seems that there is a lack of options in the medical world.

Abby Milburn, IL
**Child: Lottie**, age 5

Our children die a painful death. That is a certainty without any medical intervention. Parents have the right to try a drug, even if the risk is high and the outcome isn't certain. Our children deserve, at the very least, an opportunity to try a drug to ease their pain and possibly increase their quality and quantity of life.

Christin Ulrich, Florida
**Child: Veda Ulrich**, age 6

At 16, I think we have long come to terms with Jacob not qualifying for treatments because of his age. But I would sign Jacob up for anything that would keep Jacob as he is now forever!

Christine Moon, New York
**Child: Jacob Moon**, age 16

We understand that Sanfilippo Syndrome is progressive and ultimately fatal. The outcome, without intervention, is certain.
So when we weigh benefits and risks, we are not comparing treatment to a healthy future. We are comparing treatment to continued decline.
That changes the equation.
We educate ourselves. We ask questions. We consult specialists. We look at data. We understand that innovative therapies, especially in rare disease, come with uncertainty.
But doing nothing also carries risk.
The risk of seizures.
The risk of losing speech.
The risk of losing mobility.
The risk of losing our child far too soon.
We are willing to try promising therapies because the alternative is guaranteed progression.
If there is a chance to preserve brain function, to slow decline, to reduce suffering, that matters.
We are not asking for perfection.
We are asking for opportunity.
As parents, our job is to protect our child. In a disease like Sanfilippo, protection means being willing to take carefully considered risks for the possibility of more time, more skills retained, more quality of life.

Stephanie McCabe, Saskatchewan, Canada
**Child: Emmett Doucette**, age 6

As a parent of a child with Sanfilippo syndrome, I am making decisions in the context of a disease with a known and devastating outcome.
Because progression is certain and irreversible, I weigh risk differently. The risk of doing nothing is continued and predictable decline.
My child, Sadie, participated in a Phase 1 clinical trial for two years. That was not an easy decision—but it was an informed one. We accepted uncertainty because the alternative was guaranteed loss.
That experience reflects my willingness to pursue promising therapies, even when risks are present. I am not looking for perfection or certainty—I am looking for a chance to slow progression, preserve skills, and improve quality of life.
In diseases like Sanfilippo syndrome, even small benefits matter. More time with retained abilities, better sleep, or reduced discomfort can significantly impact both the child and their family.
Families like mine understand the risks. We live with the outcome of this disease every day. We should have a voice in determining what level of risk is acceptable when the alternative is already known.

Ashley Haywood, NC
**Child: Sadie**, age 9

We would be very willing to try any treatment to stop or delay the progression of this disease as it is inevitable. If it gives us more time and improves functional status, we feel it is worth it.

Krystal Cooley, SC
**Child: Dawson**, age 7

Sanfilippo Syndrome has a certain and devastating outcome. Without treatment, progression is guaranteed.
When the alternative is continued neurological decline, we are willing to consider reasonable risks associated with promising therapies. We understand that innovative treatments carry uncertainty  but the certainty of doing nothing is far worse.
We are not asking for reckless decisions. We are asking for urgency, flexibility, and compassion in evaluating therapies for children who do not have time to wait. We are willing to try treatments that offer even a chance to slow progression or preserve function.

Soraya, Al Chouf Baakleen Lebanon
**Children: Sama Chaaban and Aram Chaaban**

Since this disease is progressing day after day, promising therapeutic treatments will be the hope to find the proper cure, so the kids can retrieve their nice moments and abilities instead of going straight foreword to death

Khaled Chaaban, Lebanon
**Children: Sama Chaaban & Aram Chaaban**

We fully recognize that Sanfilippo syndrome is a progressive disease with a predictable, unfavorable outcome without intervention. The natural course of the disease guarantees loss of skills, a decline in quality of life, and a shortened life expectancy.
Since the natural course of Sanfilippo syndrome leads to severe disability and early death, the risk of inaction is unacceptable to us.
Even partial improvement (stabilization of skills, improved sleep) is a significant victory for us.
We are ready to try various treatment methods. Our motivation is extremely high, as we fight for every day of our child's quality of life.

Eugenia Sotnikova, Novosibirsk city, Russia
**Child: Alexandr Sotnikov**, age 7

Margaret and Bridget are 100% willing to try therapeutic medications and or treatments to maintain their level of independence. They understand their diagnosis is often called “childhood Alzheimer’s.” They know what Alzheimer’s looks like and that’s not something they want to happen to themselves.
They both have said themselves they are willing to participate in these types of treatments for the potential improvement-quality of life. They trust the science, the understand the data is strong. They trust the many pharmaceutical companies that have been working on this for years and years. There are no other options for them. They asked for your help today because today is their best day, tomorrow they are a little bit worse.

Kathy Lindquist, New York
**Children: Bridget & Margaret Lindquist**

We can see how he keeps his quality of life longer. We could see if he makes progress in language

Astrid, Germany, Bavaria
**Child: Charlie**, age 4

Biz her türlü ciddi çalışmanın içinde olmak istiyoruz (We want to be involved in all kinds of serious work.)

Mehmet vural, Türkiye
**Child: Zeren**, age 6

Without treatment our little girl will suffer the most cruel and catastrophic decline you can imagine, losing all physical and mental function and eventually her life to this horrific condition. When premature death after years of suffering is the reality we are facing without treatment it is worth any risks associated with experimental treatment options. It simply cannot be any worse than the outcome of Sanfilippo Syndrome, so the potential benefits far outweighs the risk. We are willing to try anything to save our little girl from the horrific future that awaits her.

Emily Forrester, Kent, UK
**Child: Leni Forrester**, age 2

As parents of children with Sanfilippo syndrome, we live with the reality that this is a progressive, terminal disease marked by profound neurological decline, suffering, and ultimately the loss of our children. Because of this, many of us — myself included — are willing to accept a high risk-to-benefit ratio when it comes to experimental therapies. When the alternative is watching your child gradually lose skills, awareness, mobility, comfort, and life itself, the calculus of risk changes. What might be considered “too risky” in other medical contexts feels different when time is limited and options are nonexistent.
Our family made the decision to travel internationally during a global pandemic so our daughter could participate in a clinical trial. I am a Nurse Practitioner, and my Husband is an Engineer. We chose to participate very educated on the scientific process and potential risk/benefits of the treatment. We knowingly sacrificed financial stability, physical safety, and emotional security. She ultimately did experience a complication from the treatment, and it is possible that the long-term benefit will be minimal. Even in hindsight, we do not regret the decision to participate.
We do not regret it because when faced with a disease that guarantees suffering and decline, doing nothing is not a neutral choice — it is simply accepting the inevitable. As parents, we needed to know that when an opportunity arose — one that so few families ever receive — we did everything within our power to try to help our child. Families affected by Sanfilippo understand the risks. We live the risks every day.

Lauren Barber, Michigan

**Child: Autumn Barber**, age 7

Since the outcome is certain and devastating, we would be willing to try everything that could give us more quality time with Louisa.

Lennart Sieweke, Potsdam, Germany
**Child: Louisa**

The disease is terminal. In a flat line, she will die early in life after pain & suffering. The risk I am willing to take for my child to have a better life outweighs anything. More happy days. More walking, more talking, more laughing, more playing. No suffering.

Morgan Rachal, Louisiana
**Child: Lydia Rachal**, age 3

We would do anything to try and help Frankie. Sanfilippo Syndrome is absolutely relentless and we'd give anything to ease its burden. Though we know therapies can have risks, we also know exactly what happens if we don't try. The potential to help our daughter is worth the risk.

Gabrielle Price, Washington
**Child: Frankie Price**, age 6

Doing nothing means guaranteed regression. Guaranteed loss of skills. Guaranteed suffering over time.
Yes, there may be risks. There may be side effects. There may be unknowns. But the disease itself is a known, relentless risk. Every month without intervention means more neurological damage that cannot be reversed.
We are willing to participate in clinical trials because we can only win!

Christiane von Rosbitzki, Germany/Hessen/Rödermark
**Child: Theresa von Rosbitzki**, age 4

Sanfilippo has a known and devastating progression. Without intervention, outcomes are certain, and they are heartbreaking. Because of that, families like ours think about risk differently than those facing conditions with stable or predictable futures.
For Payton, whose abilities decline every day, the risk of doing nothing is the greatest risk of all.

We understand that any therapy—especially innovative or first-in-class treatments—comes with uncertainties. But we also live with the certainty of a disease that will continue taking from her unless something interrupts its course. When the alternative is guaranteed loss, the potential benefits of a promising therapy, even one with some risk, carry enormous weight.
We are thoughtful, informed, and deeply invested in evidence and safety. But we are also parents watching our daughter lose skills she had only months before. That urgency is real, and it makes us willing to pursue therapeutic options that could give her a chance at a better future, even if they are not perfect. Payton has not yet had the opportunity to participate in a clinical trial—with her involvement so far limited to data-only research—but we would absolutely consider a treatment-based trial if it were available and scientifically promising.
When families like ours advocate for earlier access, it is not out of desperation—it is out of rational, informed hope grounded in the reality of the disease's progression. We are asking for the chance to try, before time makes trying impossible.

Ally Geronzin, Arizona
**Child: Payton Geronzin**, age 8

My daughter was the first in the US to receive enzyme replacement through a cranial port in the United States. I have zero regrets and would do it again. We need our attempts to gain access to treatment to become a reality.

Rebecca Jordan, Ohio
**Child: Liv Jordan**, age 11

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.

Nancy Rubino, Massachusetts
**Child: Merrick Rubino**, age 11

## If you could speak directly to the leaders at FDA, what would you say?

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 is taking more from Beckham. 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 Beckham do not have.
Please do not make families like mine wait while our children disappear in front of us.
His name is Beckham. He is four years old. He is a son. He is a brother. And he deserves the chance to fight for his life.
Because while you are deciding, I am watching my son disappear.

Brandon Hutcheson, Georgia
**Child: Beckham "Bex" Hutcheson**, age 4

Please approve clinically proven treatments TODAY. Ask for clarifying details about the manufacturing facility and production TOMORROW. Our kids' lives can't wait.

Alina Gorniak, Texas
**Child: Simon Croke**, age 9

If I could say one thing to FDA, it would be that my child’s life is in your hands. You have the power and the ability to save her life.

Abby Milburn, IL
**Child: Lottie**, age 5

The time is now! Our children are not a number on a page. They are real, they are here and they are worthy and deserving of an FDA approved treatment!!

Christin Ulrich, Florida
**Child: Veda Ulrich**, age 6

Please stop delaying treatments for our children! Please understand that families just want more time with their babies,

Christine Moon, New York
**Child: Jacob Moon**, age 16

Please remember that behind every data point is a child whose disease does not pause while decisions are being made.
Sanfilippo Syndrome is relentless and progressive. Our children are losing brain function every single day. We do not have the luxury of time.
We understand the responsibility you carry. We want therapies to be safe. We want them to be effective. But we also need urgency to reflect the reality of diseases where decline is certain and irreversible.

Stephanie McCabe, Saskatchewan, Canada
**Child: Emmett Doucette**, age 6

To the leaders at the U.S. Food and Drug Administration:
Please understand that for children with Sanfilippo syndrome, time is not without consequence. Every month of delay means irreversible loss—of skills, of connection, and of quality of life that cannot be regained.
I am not asking you to lower standards. I am asking you to match the urgency of your process to the reality of the disease.
Families like mine are willing to accept risk because we live with the certainty of what happens without treatment.
We need a system that moves with that same understanding—because our children do not have time to wait.

Ashley Haywood, NC
**Child: Sadie**, age 9

Our children are the most important thing in the world to us and the time we have is invaluable. Any delay in the approval of treatments leads to further progression of this disease.

Krystal Cooley, SC
**Child: Dawson**, age 7

Please see our children not as data points, but as lives on a clock that is moving too fast. Regulatory timelines that may seem reasonable in other contexts are devastating in rare, rapidly progressive diseases like Sanfilippo. We ask you to act with the urgency this disease demands. Every day, week and month matters. Every delay costs something that cannot be recovered. Our children do not have time. Please do not let the system take what the disease has not yet taken.

Soraya, Al Chouf Baakleen Lebanon
**Children: Sama Chaaban and Aram Chaaban**

This is a life-threatening, progressive condition with zero alternatives, which fits the criteria for urgent Fast Track or Accelerated Approval.

Khaled Chaaban, Lebanon
**Children: Sama Chaaban & Aram Chaaban**

I would say, "Look into my child's eyes." If you saw how this disease is stealing their smile and skills day by day, you would understand why we are begging for speed. We are not asking you to abandon safety; we are asking you to understand our urgency. Please stand with us in this race against time. Please hear the voice of patients: we are willing to take risks for the sake of hope. Don't expect perfection when children's lives are at stake. Speed up the approval process, because tomorrow may be too late. Your signature on the approval is more than just a document; it is a gift of life for children who no longer have hope.

Eugenia Sotnikova, Novosibirsk city, Russia
**Child: Alexandr Sotnikov**, age 7

I would ask the FDA to expedite the process for pediatric ultra rare diseases. We are not asking for shortcuts. Safety is a number one priority. But time is not on our side. These kids cannot

wait. We need the system to change for children with rare disease. Improving the quality of life is a significant change for parents and families with sanfilippo syndrome . It affects the entire body and the whole system. If a child's sleep could improve, digestion could improve, be seizure free, and less pain would result in happier days for everybody involved. Less hospital visits, less emergencies, less uncertainty. A person's quality of life is subjective and cannot be measured by the FDA or others who are not living with rare diseases. Having 2 daughters with Sanfilippo Syndrome causes enormous complications and tremendous suffering on our family and everyone who knows us and loves our girls.

Kathy Lindquist, New York
**Children: Bridget & Margaret Lindquist**

Please give our children a chance for a better life. They do not have an opportunity to have a normal life, to grow up and have their own families. So please help us make their short lives as happy as possible.

Astrid, Germany, Bavaria
**Child: Charlie**, age 4

Sanfilippo Syndrome is cruel, relentless, rapid and irreversible. It leaves no part of our children untouched and the impact is catastrophic for both the child and the families and people who love them. The science is there, treatment options exist, we just cannot access them - and that is absolutely devastating and incredibly frustrating. This is even harder to accept when it's lives that are at stake, and even more so when it is children. There is a window of opportunity to prevent this awful decline if a child receives treatment early enough, but that window doesn't remain open indefinitely and what has been lost cannot be recovered. Delays and regulatory processes are a matter of life and death - and I ask you to consider how you would feel if this was your child, and your reality. It is a race against time, and time is the one thing our children do not have. You must expedite drug approval protocol for rare diseases now, to save our children's lives. When what's at stake is a child's life, these issues cease to be political or administrative and become something profoundly moral.

Emily Forrester, Kent, UK
**Child: Leni Forrester**, age 2

We respectfully urge the FDA to ensure that regulatory processes and evolving evidentiary standards do not unintentionally delay access to promising therapies for rare, fatal pediatric conditions. In progressive neurodegenerative diseases with no approved treatments and no

options to improve quality of life, timing is critical and delay results in irreversible loss. Families facing certain decline and premature death are willing to accept substantially higher levels of risk and uncertainty, and regulatory frameworks should reflect the urgency and reality of these devastating diseases.

Lauren Barber, Michigan
**Child: Autumn Barber**, age 7

Please don't lose yourself in details and rigid processes - the speed of the process is critical and will save our children's lives. Evaluate wisely what the benefits of a treatment/clinical trial can mean for the children and their families.

Lennart Sieweke, Potsdam, Germany
**Child: Louisa**

As a mother, when my daughter was born.. I felt as if her life was all in my hands to take care of her well being. Now that I know she has a terminal disease with treatments on the way with the science working, I feel as if her life is also in the hands of the FDA. These are OUR children. It’s our duty to give children the absolute best quality of life we possibly can. Time is ticking, these treatments are my daughter’s ONLY chance at life.

Morgan Rachal, Louisiana
**Child: Lydia Rachal**, age 3

Frankie showed us exactly who she is before Sanfilippo Syndrome caused a sharp, fast regression. Smart, joyful, funny, loving, brave, the best big sister. Full of life. To see so much of her be taken is a pain we endure every day. To know that there's more pain to come is a fear we face every day. If it's too late for Frankie, we hope that it's not too late for others and for the children who will continue to be diagnosed with Sanfilippo Syndrome. We hope for a world where children can get access to therapies sooner. Time isn't on our side. 6 months can mean everything.

Gabrielle Price, Washington
**Child: Frankie Price**, age 6

If the system can be improved to expedite the rate of scientific discovery that can tangibly improve lives and experiences of families like mine, it should be done. I don't consider

experimental treatment to be an assured thing that could help my child. But I sure as hell want the option to try.

Andrew Price, Washington
**Child: Frankie Price**, age 6

Please see the urgency the way we live it.
For you, timelines are quarters, review cycles, regulatory pathways, and data sets. For us, timelines are skills lost, words forgotten, nights without sleep, and parts of our child slipping away that we will never get back.
Sanfilippo Syndrome does not pause while paperwork moves forward. It does not wait for the next meeting. Every month of delay means irreversible neurological damage. Time is brain — and our children are losing it in real time.
Our children do not have time for perfect.
They need thoughtful speed. They need regulatory courage. They need leaders who understand that in ultra-rare, rapidly progressive diseases, the cost of waiting is measured in childhood itself.

Christiane von Rosbitzki, Germany/Hessen/Rödermark
**Child: Theresa von Rosbitzki**, age 4

If I could say one thing to the leaders at U.S. Food and Drug Administration, it would be this: Please don't let the perfect become the enemy of the possible when children like my daughter Payton are losing abilities every single day.
For families living with Sanfilippo, time is not theoretical—we watch its consequences unfold in real time. Every delay, even when unintentional, has a human cost that can never be undone.
We are not asking you to lower standards of safety or effectiveness. We are asking you to recognize that for diseases with certain and devastating progression, inaction is not the safer option. The risk of doing nothing is guaranteed harm.
Please let urgency, compassion, and flexibility guide your decisions for rare diseases. Our children cannot wait. They don't have that luxury.

Ally Geronzin, Arizona
**Child: Payton Geronzin**, age 8

If it were your child, you would want treatment. It's here, it's available. Let us give it to our kids so they can live their best lives.

Rebecca Jordan, Ohio
**Child: Liv Jordan**, age 11

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.

Nancy Rubino, Massachusetts
**Child: Merrick Rubino**, age 11