

Beyond the Infusion: The Caregiver Experience of Gene Therapy

Gene therapy has transformed the landscape of SMA. For many families, onasemnogene abeparvovec (OA, also known as Zolgensma®) represents a treatment that can alter the course of a condition that was once associated with severe disability and early death.

We know a great deal about the clinical outcomes of OA treatment. We know that earlier treatment can improve survival and motor outcomes. We know that many children achieve milestones that would not previously have been expected. But what is less well understood is what the experience of treatment is like for families.

As part of my MSc in Genetic and Genomic Counselling at Cardiff University, I worked with SMA UK to explore the experiences of caregivers whose children had received OA treatment in the UK. Through in-depth interviews with eight caregivers, I sought to understand how families navigated diagnosis, treatment decisions, expectations, and life after gene therapy. While every family's experience was unique, several common themes emerged.

Racing against time

One of the strongest findings from the study was the importance of time. Many caregivers described delays in diagnosis and feeling that they were fighting to have their concerns taken seriously. Several parents spoke about relying on their instincts long before a diagnosis was made.

One caregiver recalled: "I think it's just mother's gut intuition... I just had this inkling, like he's going to die."

Another described the frustration of repeatedly raising concerns: "Nobody seems to be taking me seriously... I knew there was something wrong."

The feeling of being dismissed by healthcare professionals was found amongst most participants.

One participant reported: "No one was taking it seriously and that was horrendous".

Another reported: "Our world had shattered ... he'd been seen like by eleven health professionals before this point."

For many families, receiving a diagnosis was only the beginning. Parents often found themselves taking on the role of advocate, pushing for referrals, seeking answers, and ensuring their child accessed treatment as quickly as possible.

The urgency surrounding OA treatment was a recurring theme. Caregivers understood that treatment effectiveness depends on early intervention, creating immense pressure at an already overwhelming time.

"It could be amazing if someone is getting that treatment from two weeks, three weeks, a month old, even two months old, it could change their lives forever and they could be living a life without it."

These findings reinforce the importance of timely diagnosis and highlight the urgent need for newborn screening for SMA to be implemented consistently across the UK.

Making decisions in the face of uncertainty

The decision to pursue gene therapy was rarely straightforward. Caregivers described making treatment decisions while processing a life-changing diagnosis, often within a very short timeframe. Many felt there was little opportunity to fully absorb information before being asked to make significant decisions about their child's future. For some families, OA felt like the only realistic option available.

One parent reflected: *"We've got to go for the gene therapy."*

Another recalled being told: *"You've got kind of three choices. You can let her die. You can take palliative care. You can have a go at Zolgensma."*

Although families recognised that there were uncertainties surrounding long-term outcomes, most felt the potential benefits outweighed the unknowns.

The appeal of a one-time treatment was also important. Parents described wanting to avoid repeated hospital procedures and the ongoing burden associated with other treatment options. For some, OA represented not only hope for their child, but also an opportunity to move beyond the immediate crisis and begin rebuilding everyday family life.

More than a one-time treatment

One of the most interesting findings was the contrast between expectations of OA as a 'one-time treatment' and the reality families experienced afterwards.

The infusion itself was often described as straightforward. However, treatment did not end when families left the hospital.

Parents spoke about side effects, repeated blood tests, steroid management, appointments, monitoring, and ongoing care needs. Some described needing to continue advocating for their child within healthcare systems long after treatment had been administered.

Many caregivers also reported gaps in care coordination and inconsistencies in support. Rather than feeling that treatment marked the end of their healthcare journey, parents often described it as the start of a new phase that brought different challenges.

Learning to live with uncertainty

Another key finding was the emotional adjustment that followed treatment.

Caregivers frequently described a shift in their expectations over time. Initial hopes often evolved into a more nuanced understanding of what OA could and could not achieve. Importantly, they also described managing the expectations of others, who sometimes viewed gene therapy as a ‘wonder drug’ or cure. For families whose children remained significantly affected, these assumptions could feel challenging and highlight the gap between public perceptions of treatment and the realities of living with SMA.

“There is there is this assumption that it is a wonder drug ... Our expectations of what normal looks like has been the biggest movements in our head.”

“I think people think, ‘oh, he's cured’. It's like, no, he's not cured. And actually, he was so late diagnosed. He's actually severely affected for the rest of his life.”

“And I think it's really hard because I think sometimes people are like, ‘oh, you know, well, he's had gene therapy now, like, you know, everything's all right’ ... And you're like, yeah, cheers. That's pretty helpful.”

Many parents moved away from focusing on specific milestones and instead celebrated individual progress, while also discussed developing different ways of coping with uncertainty.

One caregiver explained: *“We are more focused on trying to enjoy every day and not think too much about the future.”*

Others described becoming highly practical and focusing on immediate challenges rather than distant uncertainties.

This process of adaptation was ongoing. Even when children were making progress, many parents continued to live with unanswered questions about the future.

The emotional impact doesn't disappear

Perhaps the most important message from this research is we should not expect this treatment to eliminate the emotional burden of caregiving.

Although many caregivers reported positive clinical changes following OA treatment, they also described ongoing anxiety, uncertainty, and emotional strain.

One parent reflected:

"It's always a bit in our mind... we don't know how long she'll live."

Another described the experience as:

"A very lonely process to go through and very confusing because all you want to do is do what's right for your child."

Another parent described:

"The first two years was really, really rough ... we had lots of hospitalised missions and everything and every time you kind of like think, 'Oh my God, is this the last time, is this is this the end?'".

Many caregivers spoke about remaining constantly vigilant, unable to fully relax into typical parenting experiences because they were always aware of potential health concerns. Importantly, advocacy continued beyond diagnosis and treatment, becoming an ongoing aspect of caregiving.

"We are now having to always advocate wherever we go ... if we don't do it, we won't hear"

"For me the tricky part was yeah, just keeping advocating and we just didn't even have time to, you know, grieve or anything about, like, the diagnosis".

Families also described the wider impact of SMA on daily life, relationships, education, housing decisions, employment, and future family planning. These challenges often continued despite successful treatment.

One parent summarised:

"He's actually severely affected for the rest of his life. And so are we."

What can we learn from these experiences?

The findings from this study suggest that while gene therapy offers important clinical benefits, treatment alone does not remove the practical and emotional challenges associated with caring for a child with SMA.

Families need more than access to innovative therapies. They also need timely diagnosis, clear information, coordinated healthcare services, and ongoing emotional support.

As genomic therapies become increasingly available for rare conditions, it is essential that healthcare professionals understand the experiences that exist beyond clinical

outcomes. Listening to caregivers helps us better understand the realities of living with SMA and identify where support is still needed.

I am incredibly grateful to the caregivers who generously shared their experiences for this research. Their openness, honesty, and willingness to discuss both the challenges and positives of their journey have provided valuable insights into life before, during, and after gene therapy treatment.

I would also like to thank SMA UK for supporting this research and for their continued work advocating for individuals and families affected by SMA.