Navigating SMA Treatment: What Parents Prioritise in Newborn Screening Decisions

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The advent of multiple approved therapies for spinal muscular atrophy (SMA) has transformed the prognosis for affected infants, particularly those identified through newborn screening. But this progress introduces a new complexity for clinicians and families: choosing among nusinersen, risdiplam, and onasemnogene abeparvovec. Understanding parental priorities in this decision-making process is essential for effective shared decision-making.

KEY TAKEAWAYS

- The Pivot: Parental perceptions, not just clinical data, now drive treatment choice in SMA following newborn screening.
- The Data: Parents prioritise efficacy (95%), long-term impact (90%), and administration route (85%) when selecting therapies.
- The Action: Clinicians should integrate discussions on treatment administration, long-term commitment, and potential side effects early in the counselling process.

Spinal muscular atrophy, a devastating neurodegenerative disease, once carried a grim prognosis, particularly for infants with Type 1. The market for treatments shifted dramatically with the approval of several disease-modifying therapies. These treatments, when initiated pre-symptomatically, offer the potential for significantly improved motor function and survival. But the availability of multiple options, each with distinct mechanisms, administration routes, and safety profiles, means that the choice is no longer straightforward.1

A study published in the Journal of Child Neurology explored the factors parents considered when making treatment decisions for infants diagnosed with SMA through newborn screening.1 Researchers surveyed 20 parents of infants with SMA (Type 1, n=12; Type 2, n=8) who had initiated treatment after newborn screening. The study, conducted across three US academic medical centers, aimed to identify the most influential elements guiding these early choices.1

What Parents Actually Prioritise

Parents overwhelmingly prioritised treatment efficacy, with 95% of respondents rating it as 'very important' or 'extremely important'.1 This reflects the profound impact of SMA and the desire for the most effective intervention possible to mitigate disease progression. The long-term impact of the treatment also ranked highly, with 90% of parents considering it 'very important' or 'extremely important'.1 This suggests a forward-looking perspective, where families are not just seeking immediate symptom control but also sustained benefit and quality of life for their child. Clinicians should be prepared to discuss the long-term data for each therapy, even when it is still maturing, as parents are clearly weighing

future implications.

The route of administration emerged as another critical factor, with 85% of parents rating it as 'very important' or 'extremely important'.1 Nusinersen requires intrathecal injections, risdiplam is an oral solution, and onasemnogene abeparvovec is a one-time intravenous infusion. These differences carry significant implications for family logistics, hospital visits, and the child's experience. For instance, the ongoing nature of intrathecal injections can be a considerable burden, while a single infusion, despite its initial intensity, may appeal for its finality. This highlights the need for a thorough discussion of practicalities, not just pharmacodynamics, when counselling families.

Parents also considered the potential side effects, with 80% rating this as 'very important' or 'extremely important'.1 While all three therapies have demonstrated favourable safety profiles in clinical trials, the specific risks associated with each (e.g., potential for thrombocytopenia with nusinersen, liver enzyme elevations with onasemnogene abeparvovec) require careful explanation. The financial burden of treatment, while substantial for healthcare systems, was rated as 'very important' or 'extremely important' by 65% of parents.1 This suggests that while cost is a factor, it often takes a backseat to efficacy, long-term impact, and administration when families are making personal decisions for their child's health.

The Details of Administration and Long-Term Care

The study found that parents often weighed the perceived invasiveness of the treatment against its perceived efficacy. For example, some parents found the idea of repeated lumbar punctures for nusinersen daunting, while others accepted it if they believed it offered the best chance for their child. The single-dose nature of onasemnogene abeparvovec was attractive to many, but the initial hospital stay and monitoring requirements were also a consideration. The convenience of daily oral risdiplam was a clear advantage for some families, particularly those with other children or significant travel distances to treatment centers. These are not minor details; they are fundamental to a family's ability to adhere to and manage a lifelong condition. For a deeper dive into the complexities of rare disease management, our previous coverage on rare disease patients facing hurdles beyond diagnosis and treatment offers further context.

The perceived long-term commitment associated with each therapy also played a role. Nusinersen requires indefinite, regular intrathecal injections, while risdiplam is a daily oral medication. Onasemnogene abeparvovec is a one-time gene therapy, but its long-term efficacy beyond several years is still being established, and the potential for re-dosing or additional therapies remains an open question. Parents are not just choosing a drug; they are choosing a lifestyle of ongoing medical care. This is particularly true for conditions like SMA, where early intervention is paramount, but the disease course can still be variable. Understanding the genetic basis of such disorders, as discussed in our article on Harlequin Ichthyosis, helps frame these long-term discussions.

But the study was small, enrolling only 20 parents. This limits the generalisability of the findings, and a larger, more diverse cohort would provide a clearer picture of parental preferences across different socioeconomic and cultural backgrounds. The study also relied on self-reported perceptions, which can be subject to recall bias. Still, the consistency of the themes across the interviews suggests these factors are genuinely important to families. The Oxford Handbook of Paediatrics offers a concise reference for managing such complex paediatric conditions.

The Role of Physician Counselling

Physicians play a central role in translating complex clinical data into understandable information for parents. The study highlighted that parents valued clear, unbiased information about each treatment option, including its mechanism of action, efficacy data, administration schedule, and potential side effects. They also appreciated physicians who acknowledged the emotional burden of the decision and offered support. This goes beyond simply listing pros and cons; it involves active listening and tailoring the information to the family's specific circumstances and values. For example, a family living in a rural area with limited access to specialised medical centres might prioritise an oral therapy over one requiring frequent hospital visits.

The timing of the discussion also matters. With newborn screening, parents often receive a diagnosis before their infant develops symptoms, which can be both a blessing and a curse. It allows for pre-symptomatic treatment, but it also means parents are making life-altering decisions under immense emotional stress, often without a clear understanding of what SMA entails. Early, comprehensive counselling that addresses not only the immediate treatment choice but also the long-term implications of living with SMA is essential for the child's well-being. This includes discussing the need for ongoing physical therapy, respiratory support, and nutritional management, regardless of the chosen pharmacological intervention. The choice of therapy is just one piece of a much larger, lifelong care plan.

CLINICAL IMPLICATIONS

The shift to pre-symptomatic SMA diagnosis via newborn screening has fundamentally altered the treatment paradigm. Clinicians can no longer simply present the 'best' drug based solely on efficacy data; they must engage in a discussion that incorporates parental values and practical considerations. The administration route, long-term commitment, and perceived invasiveness are not secondary concerns for families; they are often decisive factors.

For pharmaceutical companies, this means that the 'best' drug is not just the one with the highest efficacy numbers, but also the one that integrates most seamlessly into a family's life. The convenience of an oral medication or the finality of a one-time gene therapy holds significant appeal, even if the efficacy difference is marginal. This competitive landscape will likely drive innovation not just in drug mechanism, but also in delivery and patient support programs.

GPs and specialists involved in the ongoing care of these children must be aware of the chosen therapy's specific requirements and potential long-term implications. While the initial decision rests with specialist teams, the day-to-day management often falls to primary care. Understanding the rationale behind a family's choice can help tailor support and anticipate challenges, ensuring that the benefits of early treatment are maximised throughout the child's life.

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