



Family Priorities Survey Report - Based on 46 responses – April 2026

Informing Research Direction in CASK-Related Disorders

Introduction

To better align research efforts with the needs of families affected by CASK-related disorders, CASK Research, alongside the Patient Led Research Hub conducted a global survey asking parents and caregivers to rank key areas of research by priority.

Participants were asked to score each topic on a scale of 1 to 5:

- 5 = Highest priority; requires further research and support
- 1 = Lower priority; sufficient knowledge already exists or less relevant to their family
- Respondents were able to assign the same score to multiple categories

This report summarises key findings from the dataset, highlighting:

- Areas of highest shared priority
- Topics with the greatest variation in opinion
- The influence of the child's age on responses

Key Findings

1. Clear, Shared Priorities Across Families

The survey revealed a strong and consistent alignment among families on several core areas. These domains received the highest average rankings and showed relatively low variation, indicating broad agreement across the community.

Top priority areas:

- Communication (verbal and non-verbal)
- Long-term outcomes (including independence, life expectancy, and regression)
- Patient quality of life
- Mobility and movement

Communication emerged as the single highest-ranked priority, with both a high average score and minimal variation between responses. This suggests a near-universal need for improved understanding and interventions in this area.

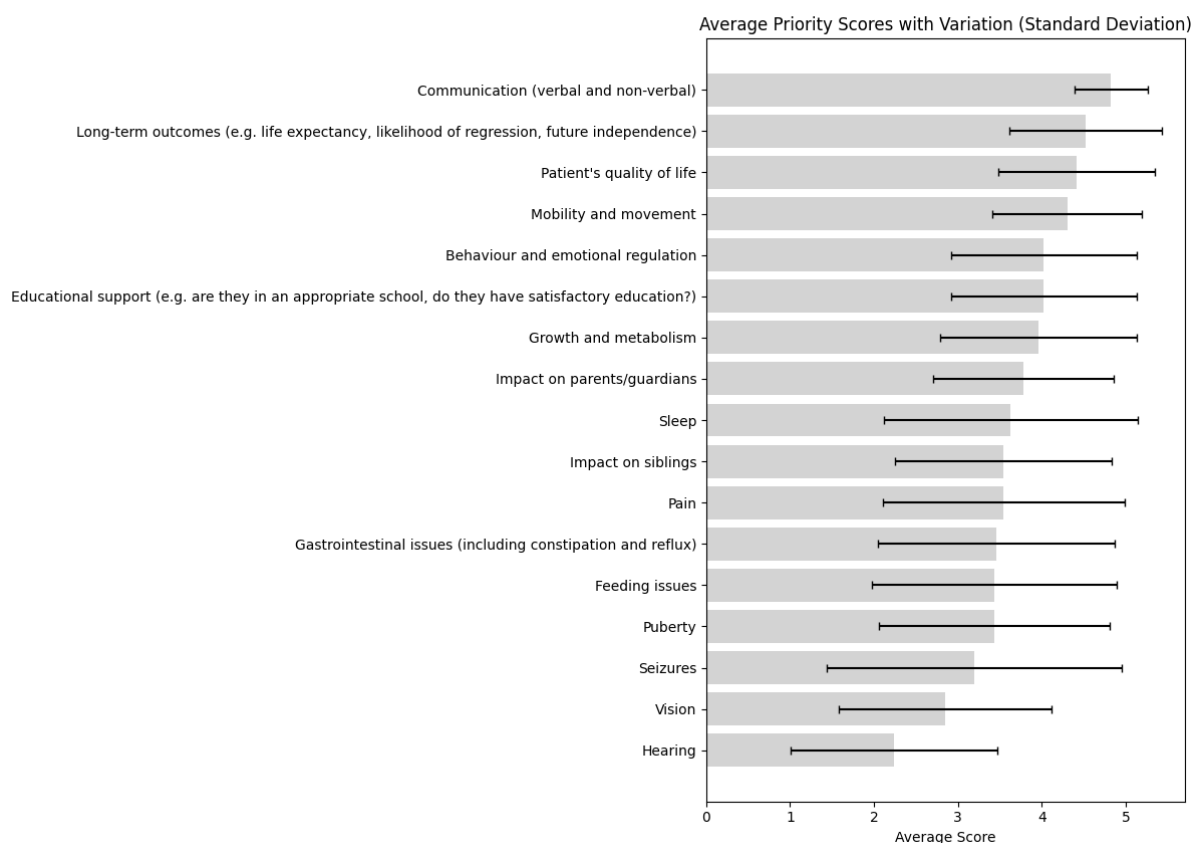
Similarly, long-term outcomes and quality of life reflect a strong desire among families to better understand the future trajectory of the condition and to maximise wellbeing over the lifespan.

2. Important but More Variable Priorities

A second group of topics was consistently rated as important, though with slightly more variation between respondents:

- Behaviour and emotional regulation
- Educational support
- Growth and metabolism
- Impact on parents and caregivers

These areas remain significant but may reflect differing experiences depending on the child's age, severity, and family circumstances.



3. Highly Variable and Individualised Priorities

Several topics showed substantial variation in responses, with some families rating them as critical (5) and others as low priority (1–2).

Most variable areas:

- Seizures
- Sleep
- Feeding issues
- Gastrointestinal issues
- Pain
- Puberty
- Impact on siblings

This pattern likely reflects the clinical heterogeneity of CASK-related disorders. For example, seizures may be a dominant and urgent issue for some individuals, while not present or well-controlled in others.

4. Lower Average Priority Areas

Some areas received lower average scores overall, including:

- Vision
- Hearing

However, it is important to emphasise that lower averages do not imply lack of importance. In many cases, these results reflect relevance to a subset of families rather than the entire population.

Influence of Age on Priorities

All groups had communication as their highest priority whilst there was variation shown in priorities for all other symptoms. Patient's quality of life was always ranked second or third; long-term outcomes and mobility and movement were consistently ranked as high priorities but with more variability.

With respect to mobility and movement the age groups 0-2yrs and 22+yrs had an increased concern for this symptom. This potentially reflects that the parents of infants had concerns for future development due to knowledge of how CASK presents or they may have noticed initial physical developmental delays whilst parents of older children were noticing a regression/reduction in movement and mobility, leading to increased concern.

Pain as a priority symptom varied greatly amongst age groups. It was a lesser priority in the age groups 0-7, increases a priority in age groups 12-16, drops in age group 17-21 and then becomes the second biggest priority in age group 22+.

As one would expect, puberty is more of a priority for families whose children are entering puberty (8-11yrs and 12-16yrs) than those who are yet to enter puberty or those who have finished puberty.

Seizures were found to be the joint second highest priority for age group 8-11 whilst for other age groups it was one of the least prioritised symptoms on average.

While age influences priorities to some extent, there is no clear pattern of certain symptoms gradually increasing or decreasing in priority as age increases. It's important to note that the small number of responses in some age groups may sway results (e.g. only 3 responses for 12-16yrs).

Conclusion

This survey provides clear evidence that families are aligned on several key priorities whilst also highlighting significant variability in other domains due to the heterogeneous nature of the condition.

Communication issues are a significant issue for the majority of the families surveyed. In our scoping literature review, we found only one publication ([Wallace et al, 2024](#)) about communication techniques or aids which might benefit CASK families. [RARE-X](#), an international family-reported research platform managed by Global Genes, does collect some surveys that may help interpret communication and cognitive ability, but further work is required to understand family experiences and explore if/what existing interventions and tools could benefit this cohort.

Research efforts are already underway to determine long-term outcomes for people living with a CASK-related disorder. There are currently a number of studies, on-going, collecting current and retrospective data on specific co-morbidities in addition to the RARE-X long-term global data collection programme.

Mobility and movement issues can be addressed by improved access to NHS physical therapy and occupational therapy. One paper found that motor skills such as trunk control, gross reaching abilities and fine motor skills can be improved by intensive therapy in some children ([Wallace et al, 2024](#)). Future medical interventions may include treatments that target the cerebellum and the degeneration of the cerebellar granule cells which has been observed in mice models (although its prevalence or even presence is not determined in human patients). A number of research groups have projects concentrating on this particular therapeutic target.

Data on quality of life for patients and the impact of caring for an individual with a CASK-related disorder has never been published for CASK disorders. RARE-X collects quality of life data but it is yet to be analysed and published.