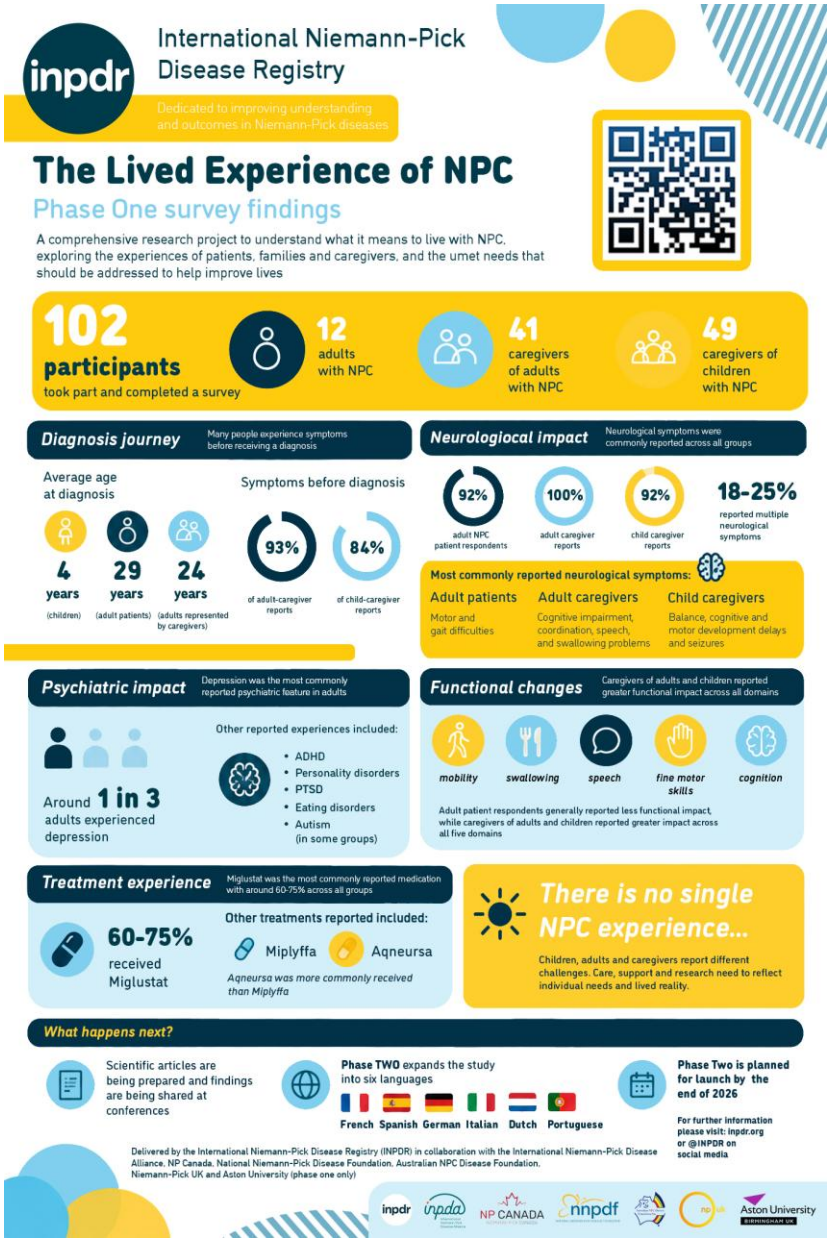
QUALITY OF LIFE IN NIEMANN-PICK DISEASE TYPE C

A lay summary produced by INPDR

In one sentence

This research explored what living with NPC means for people and families, and tested NPC-specific questionnaires designed to measure quality of life and disease severity more meaningfully.



Executive summary

Niemann-Pick disease type C (NPC) affects far more than physical or neurological symptoms. It can affect independence, communication, emotional wellbeing, relationships, education, employment and participation in everyday life. It also has a substantial effect on caregivers and wider families.

The central message

Disease severity and quality of life are connected, but they are not the same. A person may experience significant symptoms while still finding enjoyment, connection and meaning. Clinical measures therefore need to be used alongside measures of everyday quality of life.

What the thesis achieved

- Explored the lived experiences of adults with NPC and caregivers through in-depth interviews.
- Tested and refined an NPC-specific quality-of-life questionnaire for adults.
- Tested and refined an NPC-specific quality-of-life questionnaire for children.
- Improved and validated a lay-reported NPC Clinical Severity Scale that can be completed by people with NPC or caregivers.
- Identified how these tools could support clinical care, research, treatment evaluation and rare-disease registries.

Why this matters

The research provides more meaningful ways to capture both how NPC is progressing and what that progression means in daily life. These tools may help clinicians and researchers look beyond symptoms alone and understand outcomes that matter to people and families.

The research at a glance

Study area	Participants	Main output
Lived experience interviews	17 interviewees, representing the experiences of 20 people with NPC	Five themes describing diagnosis, independence, caregiving, coping and wider family impact
Adult quality of life	64 respondents	A refined 27-item adult questionnaire with three quality-of-life areas
Child quality of life	54 caregivers	A refined 14-item child questionnaire with two quality-of-life areas
Lay-reported disease severity	12 cognitive interview participants and 124 survey respondents	A clearer, reliable five-domain severity scale for patients and caregivers

1. What was the thesis trying to find out?

The thesis asked two linked questions:

- What is everyday life like for people living with NPC and for those who care for them?
- Do the available questionnaires accurately capture quality of life and disease severity in ways that are meaningful to the NPC community?

NPC may affect movement, balance, swallowing, speech, thinking and independence. However, symptom scores alone cannot fully describe the effect on a person's identity, relationships, emotional wellbeing, daily activities or family life. The research therefore examined both clinical impact and lived experience.

2. How was the research carried out?

A mixed-methods approach was used, combining personal accounts with statistical testing. This allowed the researcher to listen to experiences first and then test whether the questionnaires performed as intended.

Approach	What it contributed
Qualitative interviews	Detailed accounts of diagnosis, symptoms, treatment, caregiving and the COVID-19 pandemic.

Questionnaire surveys	Evidence about whether the adult and child quality-of-life measures were consistent and valid.
Cognitive interviews	Direct feedback about confusing wording or response choices in the lay-reported severity scale.
Psychometric analysis	Statistical testing of reliability, structure and expected relationships between severity and quality of life.

3. What did people say about living with NPC?

Interviews produced five connected themes. Together, they show that NPC affects the whole course of family life, from the search for a diagnosis to changes in independence, relationships and future expectations.

1. The journey to diagnosis

People and families described long, frustrating routes to diagnosis, including misdiagnosis and repeated assessments. A confirmed diagnosis could bring relief because it provided an explanation, but it could also bring distress and uncertainty about the future.

2. Challenges to independence

Changes in movement, speech, cognition and other abilities could reduce control over everyday decisions. Children might be unable to join peers in activities, while adults could experience difficulties with employment or independent living.

3. The demands of caregiving

Caregiving could be physically and emotionally intensive, with limited respite. Parents described uncertainty, worry, grief and the effect of caring responsibilities on work, retirement and wider family life.

4. Coping and support

People used a range of coping strategies, including focusing on the present and maintaining routines. Patient organisations were important sources of understanding, connection and community.

5. The wider family

NPC could affect siblings, partners, grandparents and other relatives. Families experienced practical restrictions, changes in relationships and concern about the future.

Impact of the COVID-19 pandemic

Restrictions increased isolation, disrupted routines and reduced access to family support, education, services and some clinical trials. These experiences reinforced how important connection, routine and

support are to quality of life.

4. Measuring quality of life in adults with NPC

The adult NPC Quality of Life Questionnaire was tested using responses from 64 participants. After analysis, the measure was refined from 30 to 27 items. The items grouped into three broad areas:

Independence and self-efficacy
Control, confidence, motivation, hope and the ability to act independently.

Social connections
Friendships, relationships, isolation and feeling connected to other people.

Emotional impact and acceptance
Emotional wellbeing, support, feeling understood and adapting to life with NPC.

The questionnaire showed that its questions worked well together to measure different aspects of quality of life (internal consistency). People's responses also showed expected relationships with scores from an established, general quality-of-life questionnaire. These findings provide encouraging early evidence that the NPC-specific questionnaire is useful.

5. Measuring quality of life in children with NPC

The child questionnaire was completed by caregivers. Following analysis, it was refined to 14 items, covering two broad areas:

Wellbeing and social participation
Enjoyment of daily life, play, friendships, family routines, special occasions and coping with change.

Disease burden and limitations
Mobility and fine motor difficulties, frustration, sadness, changed relationships and being unable to do things other children can do.

This two-part structure is important because it shows that quality of life is not simply the absence of disability or symptoms. Positive experiences can continue alongside significant disease burden.

6. Making disease severity easier to report

The lay-reported NPC Clinical Severity Scale focuses on five areas: walking, fine motor skills, swallowing, cognition and speech. Cognitive interviews showed that some original wording was difficult or inconsistent.

Issue identified	How it was improved
The terms “slight” and “mild” were difficult to distinguish.	Response levels were changed to clearer steps such as mild, moderate, severe and very severe.
“Learning delay” sounded child-specific.	The wording was changed to “cognitive ability” so it could apply across ages.
Swallowing questions focused on eating.	Drinking was added so difficulties with fluids could also be reported.
“Occasionally” and “frequently” were interpreted differently.	Frequency descriptions were defined more explicitly.
The impact question referred to the family.	The wording was changed to focus on the daily life of the person with NPC.
Respondents sometimes used experiences outside the intended period.	The “typical day over the past week” timeframe was repeated in each item.

The revised scale was tested using responses from 124 participants. It showed good internal consistency, and more severe symptoms were associated with a greater reported impact on daily life. Higher severity was also associated with poorer quality of life, particularly in the paediatric sample.

7. What are the main implications?

For people and families

The work gives greater weight to patient and caregiver perspectives. It recognises that outcomes such as participation, independence, connection and emotional wellbeing matter alongside clinical symptoms.

For clinical care

The questionnaires could help structure conversations, identify areas requiring support and complement clinical assessment. They are not replacements for clinical judgement.

For research and clinical trials

The tools could help researchers assess whether a treatment produces meaningful changes in everyday life, not only changes in clinical signs.

For registries

Routine collection of quality-of-life and lay-reported severity data could enrich longitudinal datasets, natural-history research and preparation for future studies.

For services and policy

The findings support coordinated, family-centred care that includes psychological, educational, social and respite support as well as medical care.

Key principle for future use
The most complete picture comes from combining disease severity, quality of life and lived experience. No single score can describe the whole person or family.

8. Limitations and next steps

The findings are promising, but the thesis identifies important limitations. NPC is rare, so sample sizes were modest. Most data were collected at one point in time, and the work was conducted in English across varied cultural and healthcare settings.

Recommended future work

- Follow participants over time to understand changes in quality of life and severity.
- Test short-term stability when a person's health has not changed.
- Establish what size of score change is meaningful to patients and families.
- Use larger international datasets for more advanced psychometric testing.
- Evaluate how the tools work in routine clinical care and remote monitoring.
- Integrate the measures into rare-disease registries where appropriate.
- Translate and validate the questionnaires across languages and cultures.

Conclusion

This thesis makes an important practical contribution to understanding and measuring life with NPC. It shows that NPC affects independence, relationships, participation, emotional wellbeing and whole families, not only physical or neurological functioning. By evaluating NPC-specific quality-of-life tools and improving a lay-reported severity scale, the research offers a stronger foundation for person-centered care, research and treatment evaluation.

Final take-away

To understand whether care or treatment is truly helping, we need to measure not only what is happening clinically, but what that change means for the person's everyday life.

About this report

This is a plain-English summary of:

Pountney, J. (2025). Quality of life in patients with Niemann-Pick disease type C and their carers. Doctoral thesis, Aston University.

The report paraphrases the thesis for accessibility. It does not replace the full thesis, clinical guidance or the original validated questionnaires. Any quotation or further publication should appropriately acknowledge the thesis author and source.

Terminology note: this report uses "Niemann-Pick diseases" when referring to the wider group of conditions and "Niemann-Pick disease type C" or "NPC" when referring specifically to the condition studied in the thesis.