

# Investigating the real-world experience of patients with Niemann-Pick disease type C (NPC) and their carers: Preliminary results

Comparing adult self-report, adult caregiver-report, and pediatric caregiver-report across functional domains



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## Background / Rationale

NPC is a rare, progressive, neurodegenerative lysosomal storage disorder characterized by substantial phenotypic heterogeneity. Clinical manifestations span a broad spectrum, ranging from early visceral involvement in infancy to predominantly neurological decline presenting in childhood or adulthood. Age at onset and the rate of disease progression vary considerably across individuals, creating meaningful challenges for prognosis, patient and family counseling, and the design and interpretation of clinical trials. In this context, patient- and caregiver-reported perspectives provide essential insight into functional outcomes and disease burden, though reported severity and impact may differ depending on whether outcomes are self-reported or captured through proxy respondents.

## Primary Objective:

To describe demographic and functional differences across NPC patient subgroups and characterize differences in disease progression patterns across functional domains.

## Survey Design

This analysis used data from a cross-sectional, international NPC patient and caregiver survey designed to characterize demographic and functional differences across subgroups and capture real-world disease burden. The survey collected demographic information and assessed functional status across key domains, including mobility/walking ability, swallowing, speech, fine motor function, cognition, and neurologic symptoms, with outcomes summarized descriptively using means and proportions.

## Survey Respondents

	Adult (>18y) (Self) (n=12)	Caregiver of Adult (n=41)	Caregiver of Minor (<18y) (n=49)
Age of Individual with NPC	38.2y	35.2y	9.8y
Age at Diagnosis (mean, IQR)	29, IQR=30.3	24.3, IQR=10.8	4.2, IQR=6.2
Sex of Individual with NPC	50% F	46% F	41% F
Sex of Respondent	---	73% F	82% F
Age of Respondent	---	61.4	41.1
Marital Status of Respondent	42% S, 50% M, 8% D	S 7%, M 78%, D 10%	S 14%, M 90%, D 2%
Relationship to Individual with NPC	---	Mom 63%, Dad 22%, Spouse 5%, Sibling 5%, Other 5%	Mom 78%, Dad 22%
Ave Household Size	2.3 (25% have children)	2.55	3.5
Diagnosed before Birth	0	0	1
Neurologic Symptoms	92%	100%	92%
Currently on Miglustat	9/12	23/41	38/49
Mypllyffa	6/12	8/41	14/49
Aqneursa	6/12	14/41	34/49

## Future Directions

Phase II of survey coming in 2026. Expansion of this study into other languages (Phase 2) will allow for broader data collection to better understand disease burden and its management.

## Survey Analysis

All analyses were descriptive in nature. Summary statistics were calculated using means for continuous variables and proportions for categorical variables. Comparisons across survey cohorts (adult self-report, adult caregiver-report, and pediatric caregiver-report) were presented descriptively to highlight differences in demographic characteristics and functional outcomes without formal hypothesis testing. Functional impairment across domains was reported using standardized severity categories (none, mild, moderate, and severe) to facilitate consistent interpretation of patient- and caregiver-reported outcomes.

## Survey Respondents

Responder Type	<2mo	2mo-<2y	2y-<6y	6y-15y	<15y
Adult (Self)	1 (8%)	0	1 (8%)	3 (25%)	6 (50%)
Adult (Caregiver*)	2 (4%)	2 (4%)	2 (4%)	15 (37%)	19 (46%)
Child/Minor (Caregiver)	14 (29%)	9 (18%)	12 (18%)	11 (22%)	4 (8%)

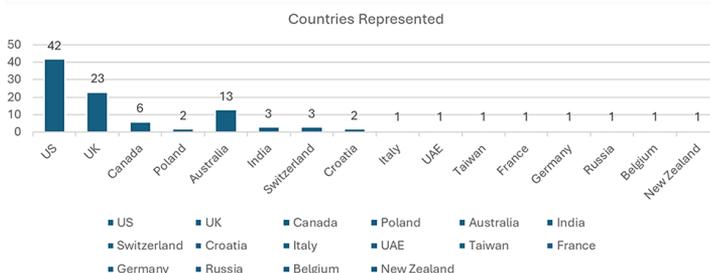
Responder Type	Symptoms Before Dx
Adult (Self)	8 (75%)
Adult (Caregiver*)	38 (93%)
Child/Minor (Caregiver)	41 (84%)*

\*3 Not Sure

NPC survey responses demonstrate distinct demographic and functional profiles across lifespan cohorts, with pediatric caregiver-reported cases showing a broader distribution of moderate-to-severe impairment. Differences between self-reported and caregiver-reported outcomes were substantial, reinforcing the importance of multi-informant approaches to accurately characterize NPC progression and disease burden.

## Results and Conclusions

Functional outcomes demonstrated clear differences across lifespan and respondent groups. Adults completing the survey for themselves reported greater preservation of mobility, swallowing, speech, and cognition compared with caregiver-reported cohorts, including 41% indicating no walking difficulty, whereas fewer in caregiver groups reported this level of function. Pediatric patients more frequently required assistance or wheelchair use and showed greater severity of swallowing and speech impairments, as reflected in caregiver reports. Fine motor and cognitive impairments were prevalent across all cohorts, with pediatric caregiver-reported outcomes shifting toward moderate-to-severe categories, suggesting earlier functional decline. Nearly all respondents reported neurologic symptoms, underscoring the pervasive burden of NPC. Differences between self- and caregiver-reported outcomes may reflect both true variation in disease severity and systematic reporting effects, including greater caregiver sensitivity to impairment or proxy-report bias. Overall, the findings highlight substantial heterogeneity in NPC progression across the lifespan and support the value of incorporating multi-informant perspectives to more accurately characterize functional trajectory and disease burden in NPC research.



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