CKDL5 deficiency disorder: caregiver's perception of clinical symptoms, disease management and its impact on quality-of-life

PCR44

carenity

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Introduction

Cyclin-dependent kinase-like 5 (**CDKL5**) deficiency disorder (**CDD**) is an ultra-rare **genetic condition** marked by developmental **epileptic encephalopathy** causing early-onset **seizures**, severe **motor** and **cognitive impairments** [1]. To this day, **no disease-modifying** treatments exist for this condition, and treatments focus mainly on symptoms management [2].

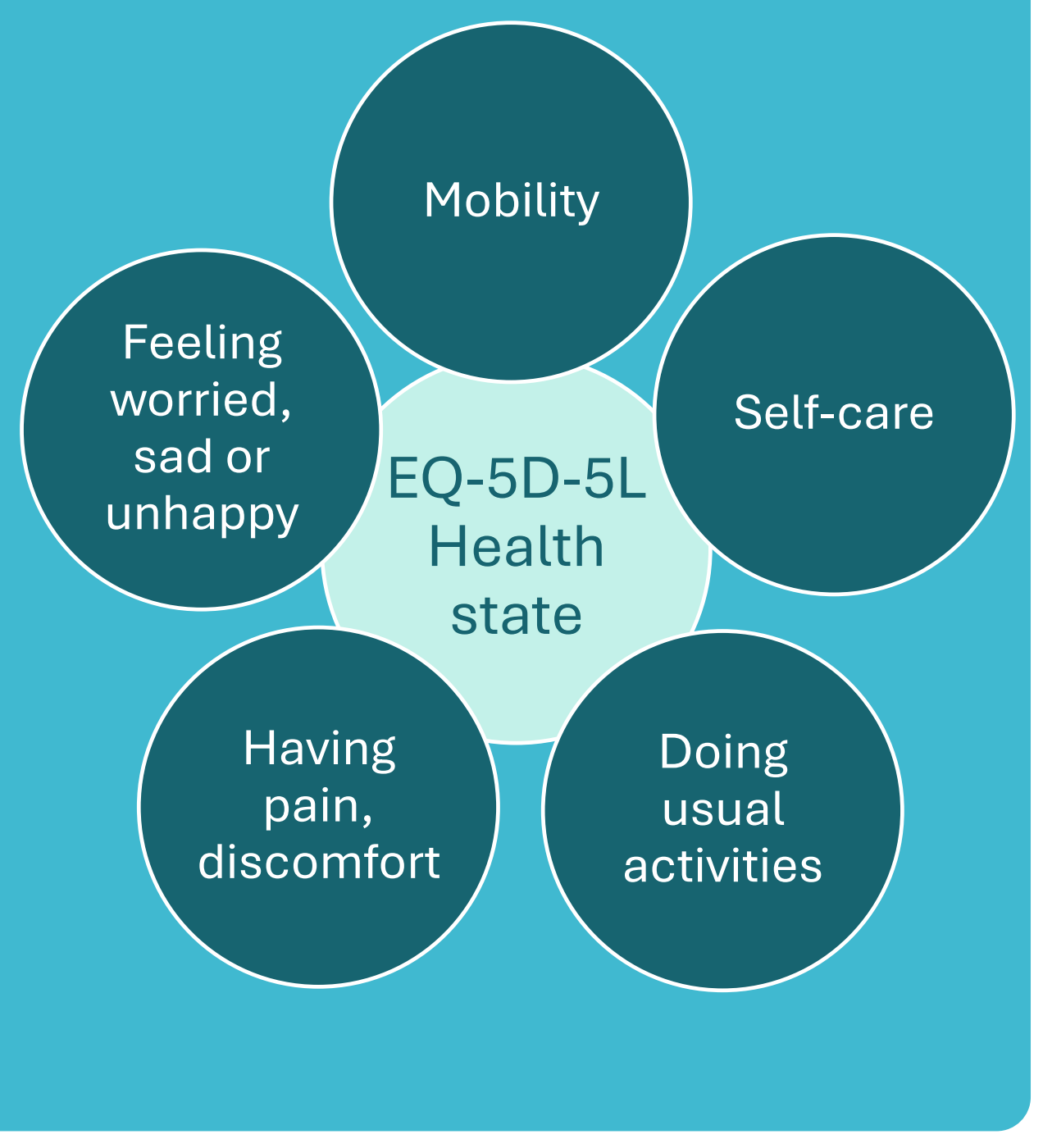
Objective

This study aimed to **assess the burden of CDD** for patients and their families, from the perspective of **caregivers**.

Methods

- An **international cross-sectional study** was conducted through **Carenity**, an online patient community, via a **web-based survey** composed of 40 questions for **caregivers of CDD patients.**
- An adapted version of the **EQ-5D-5L** (Proxy 1), a standardized tool developed to assess patients' health-related quality of life (**HRQoL**) based on 5 levels of severity and 5 dimensions, was used to report patient's HRQoL as perceived by their caregivers. It also included a **visual analogue scale** (VAS) evaluating the overall patient's health.
- Descriptive analysis was performed on responses from **132 caregivers**.

EQ-5D-5L dimensions



Results

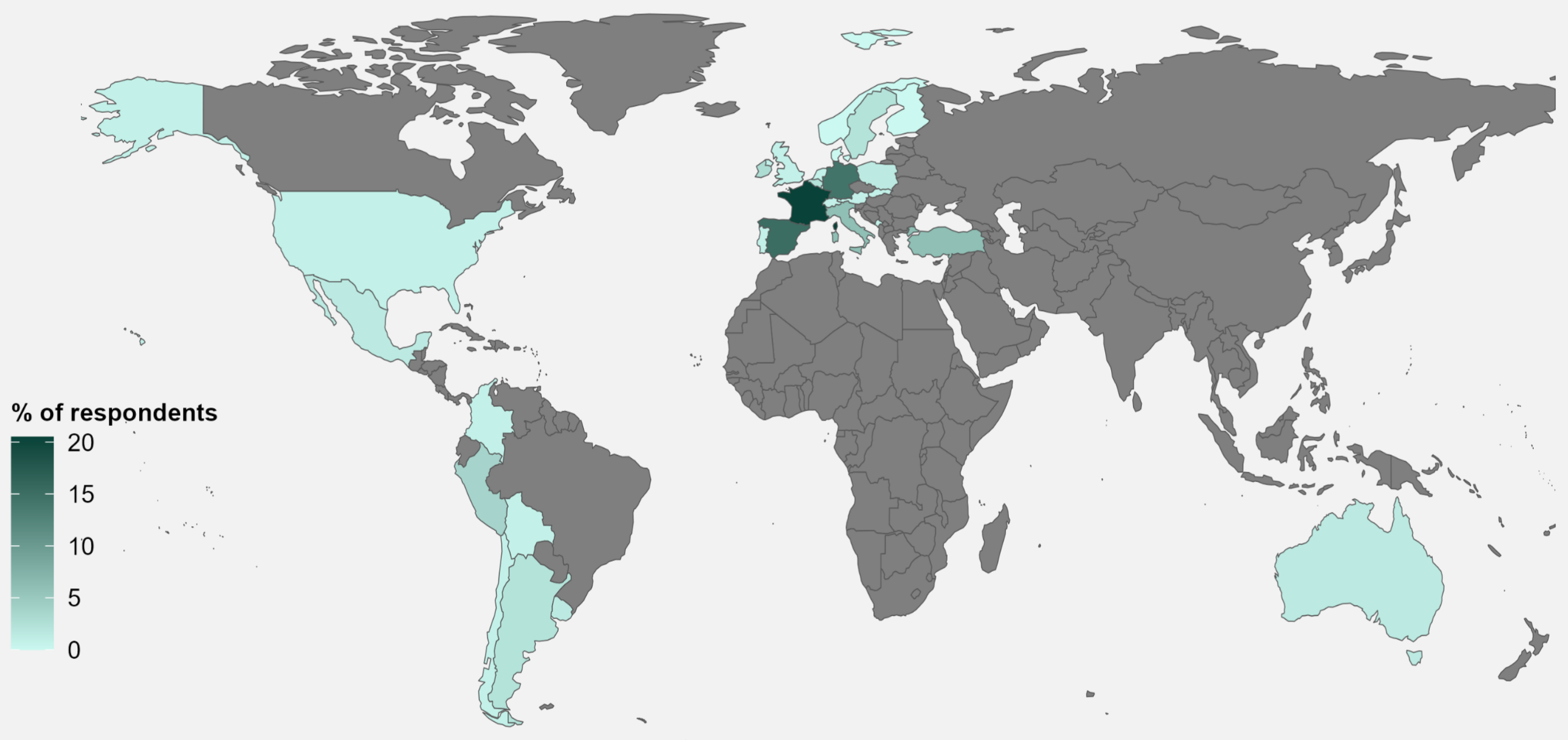
Sociodemographic profile

- All **132** respondents were **parents** of CDD patients
- Caregivers' median **age**: 41 y/o
- Caregivers' **gender**:

85% ♀ ♂ 15%

- Patients' median **age**: 7.6 y/o
- Patients' **gender**:

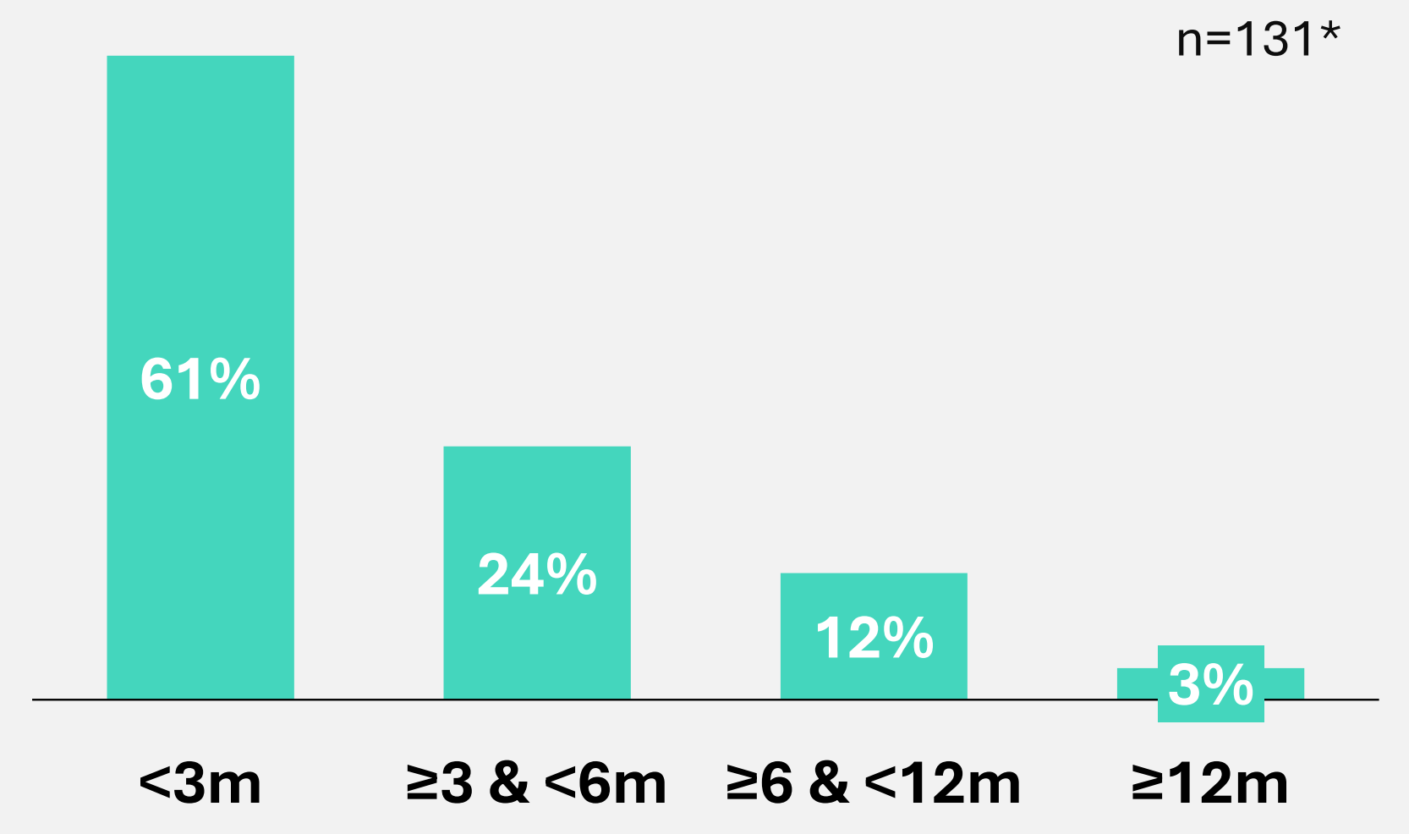
89% ♀ ♂ 11%



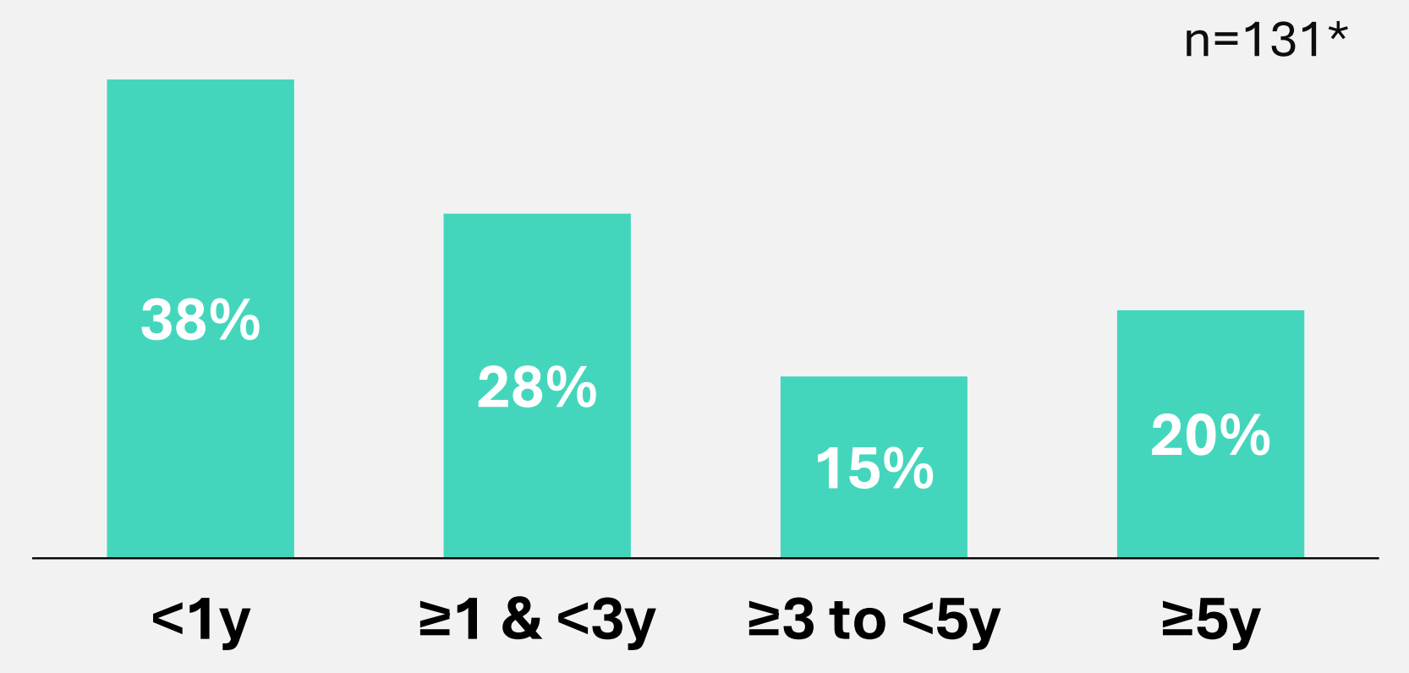
European (EU) countries (n=106)						Non-European Countries (n=26)
EU regions	**Central (n=21)**	**Eastern (n=4)**	**Northern (n=20)**	**Southern (n=29)**	**Western (n=32)**	• LATAM (15) • Turkey (8) • US (2) • Australia (1)
Countries (n=)	• Germany (19) • Austria (1) • Switzerland (1)	• Poland (2) • Slovakia (1) • Montenegro (1)	• UK (13) • Ireland (4) • Sweden (3)	• Spain (20) • Italy (8) • Portugal (1)	• France (27) • Benelux (5)	

Patients' medical characteristics

- Age at first seizure/symptoms in months:

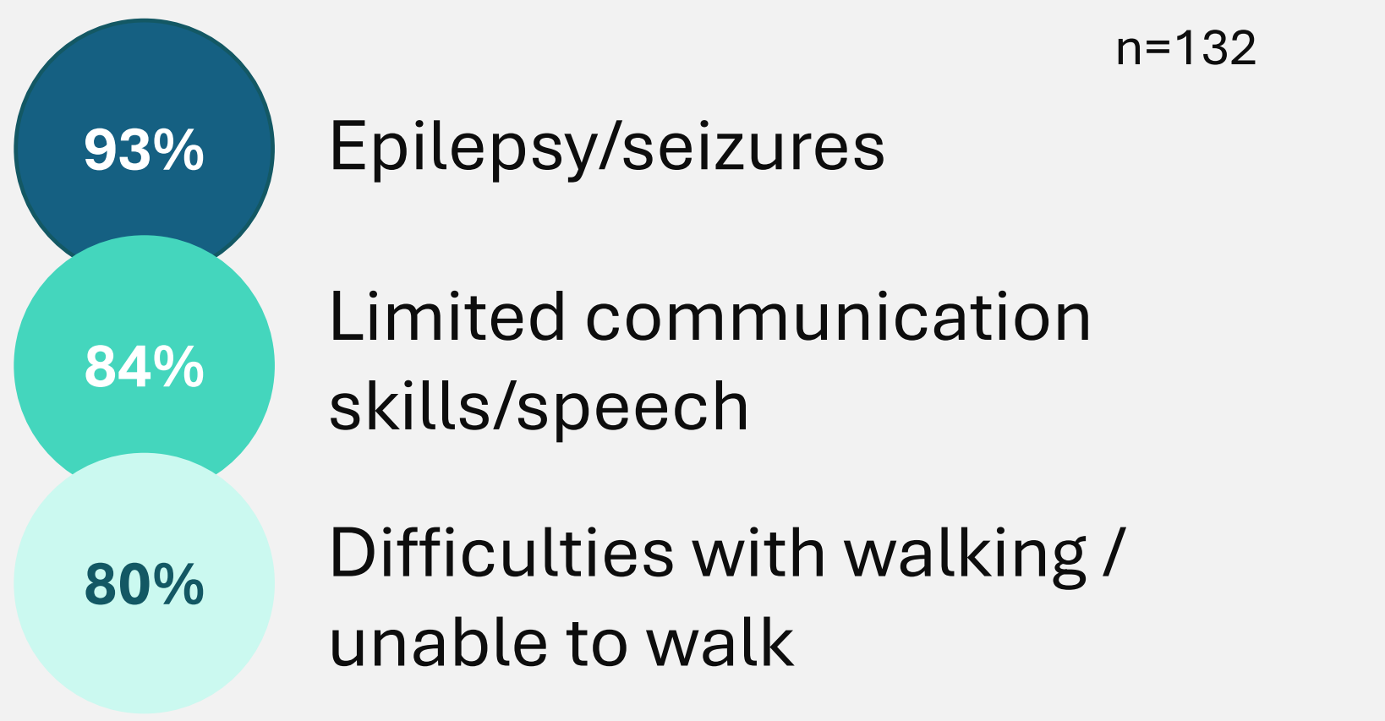


- Age diagnosis in years:



**n=1 caregiver did not provide a valid date of birth for one patient*

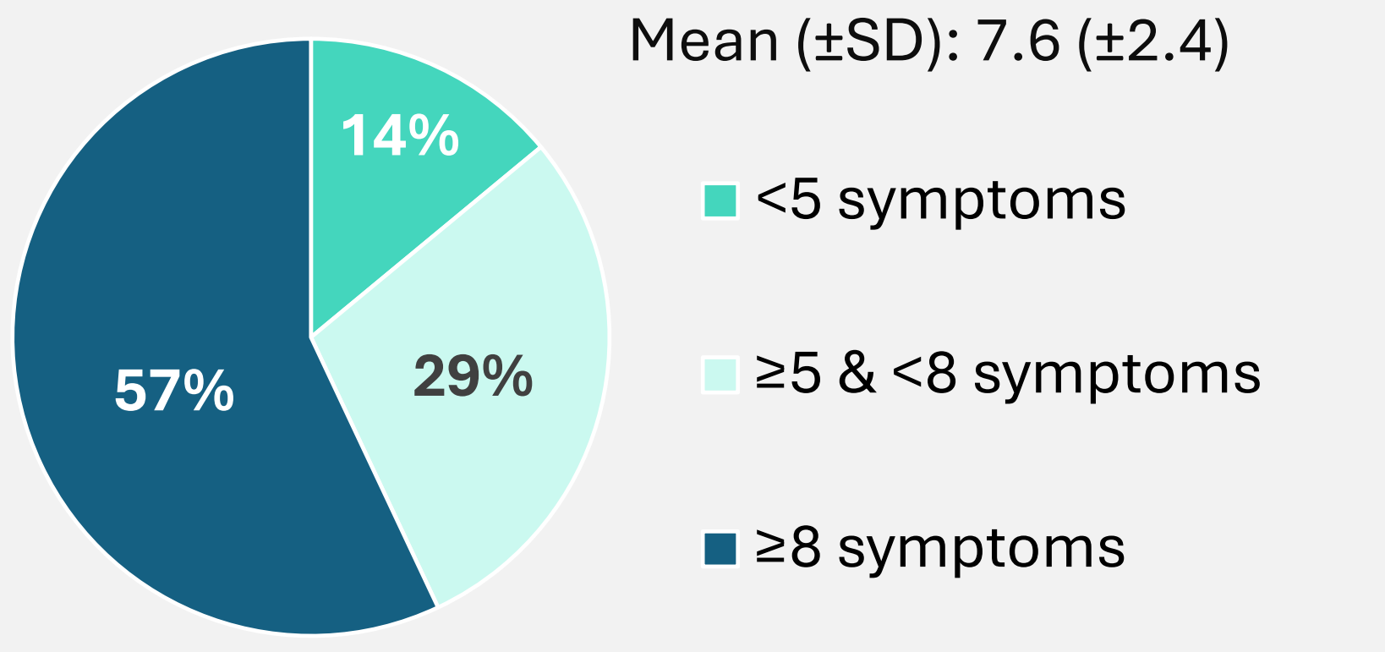
- Most frequent symptoms:



- Number of symptoms reported per patient:

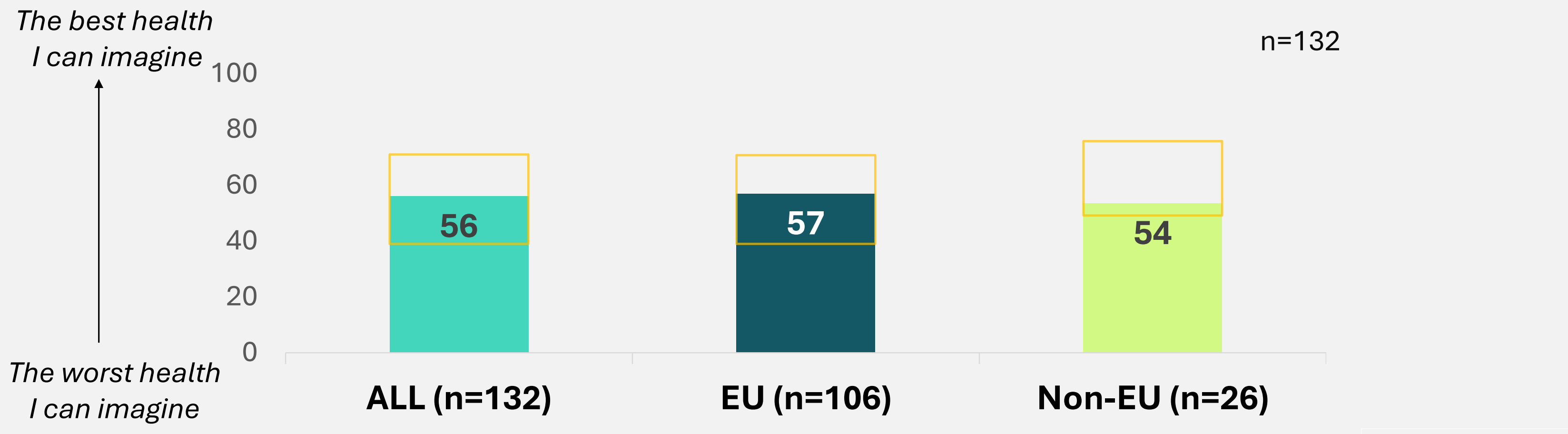
n=132

Mean (±SD): 7.6 (±2.4)



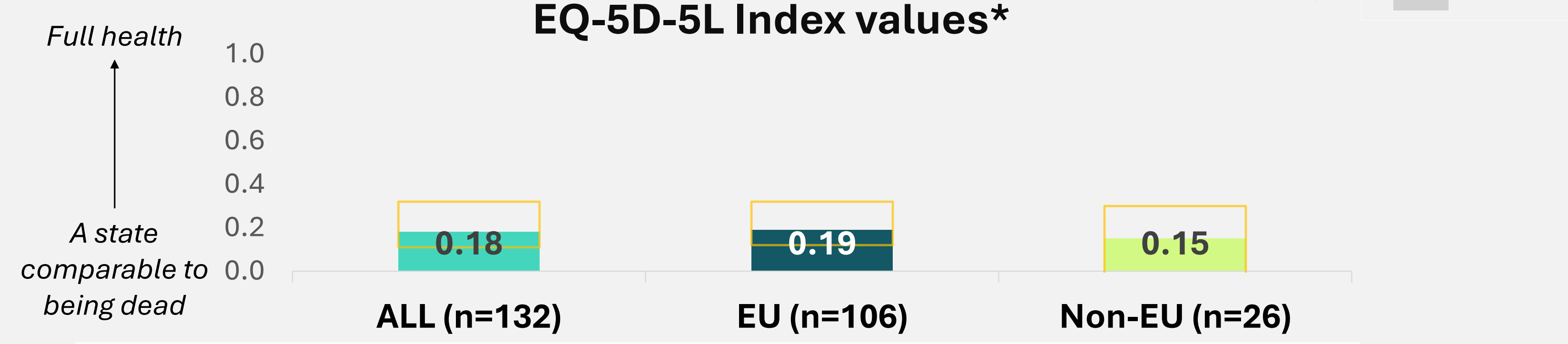
EQ-5D-5L Results: patient health-related quality of life

CDD patients' health as perceived by their caregivers on the EQ-5D-5L VAS



Mean (SD)	54.2 (22.9)	54.2 (22.6)	54.4 (24.6)

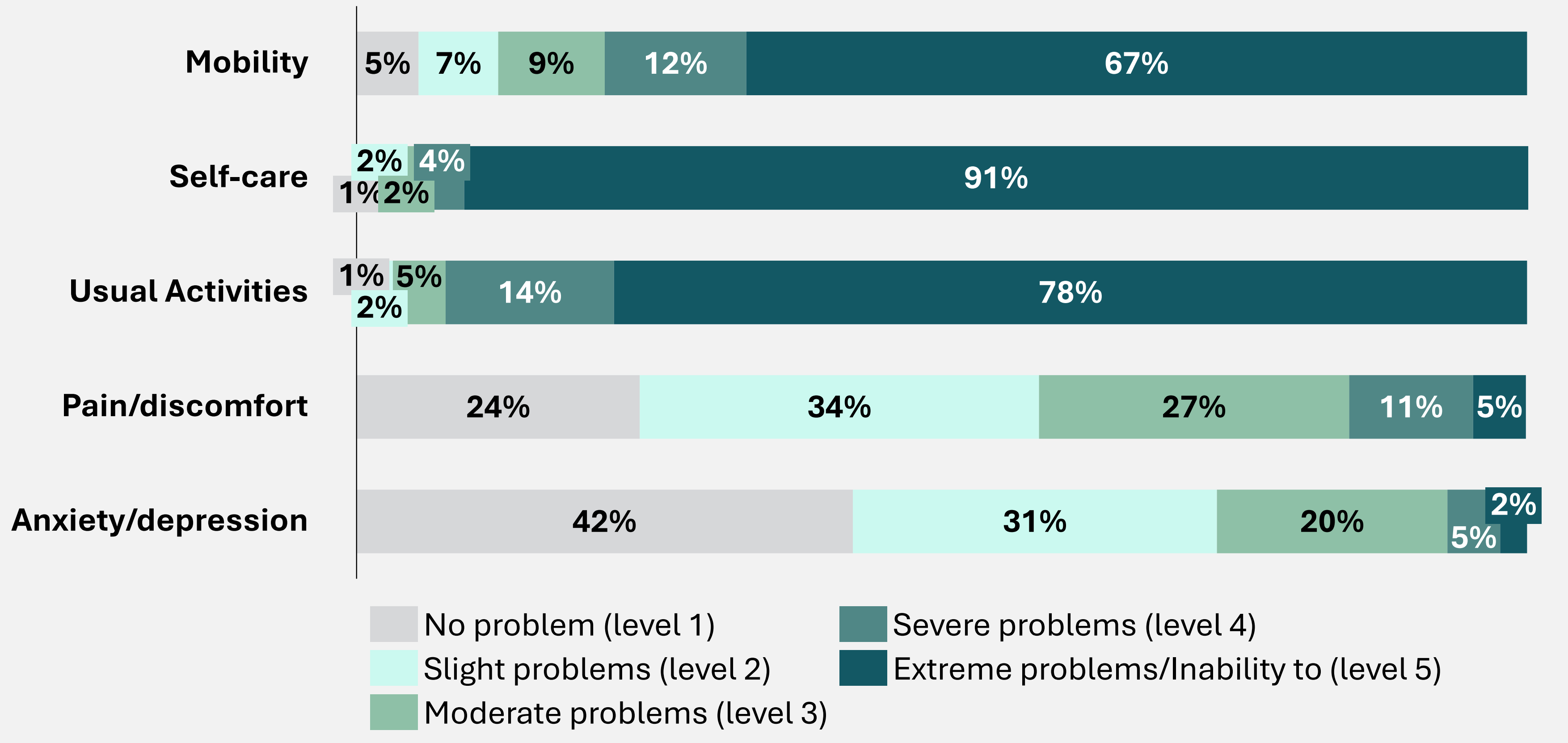
EQ-5D-5L Index values*



Mean (SD)	0.19 (0.29)	0.22 (0.25)	0.09 (0.42)

*Index values are obtained by combining the levels for each EQ-5D-5L dimensions and weighting them according to the preferences of the general population in the country considered.

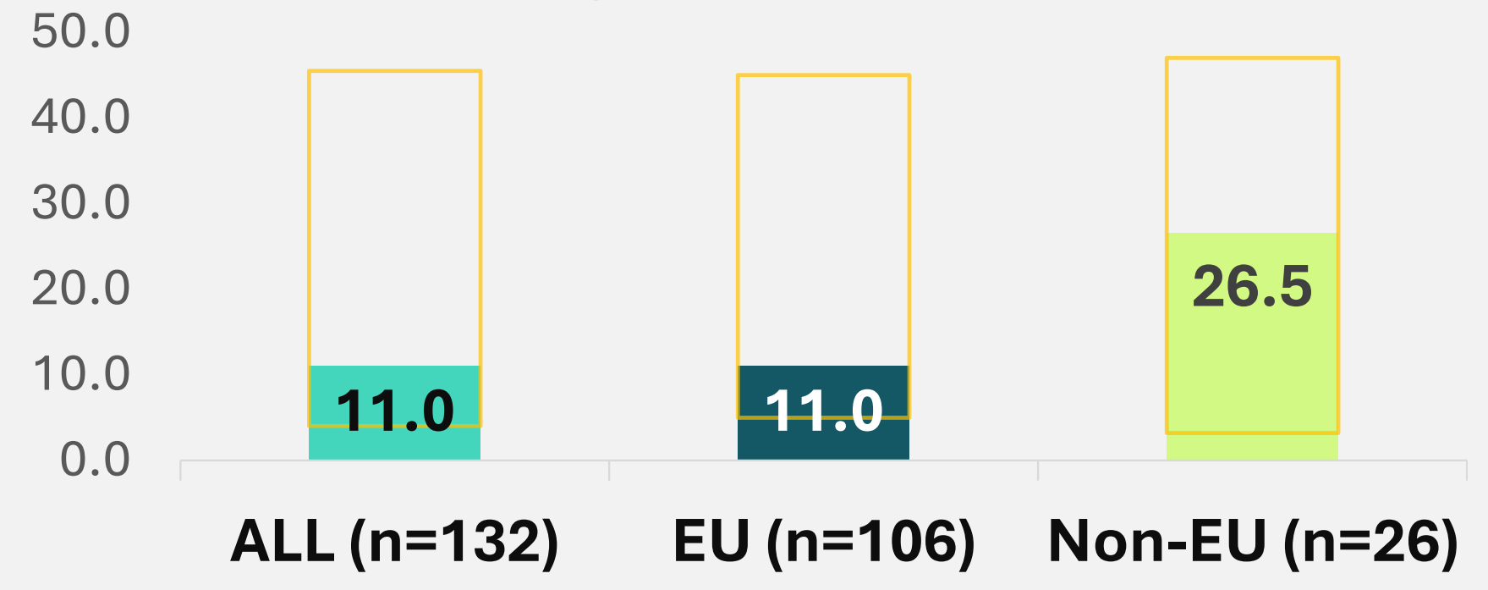
Severity (level) by EQ-5D-5L dimensions, as perceived by CDD patients' caregivers



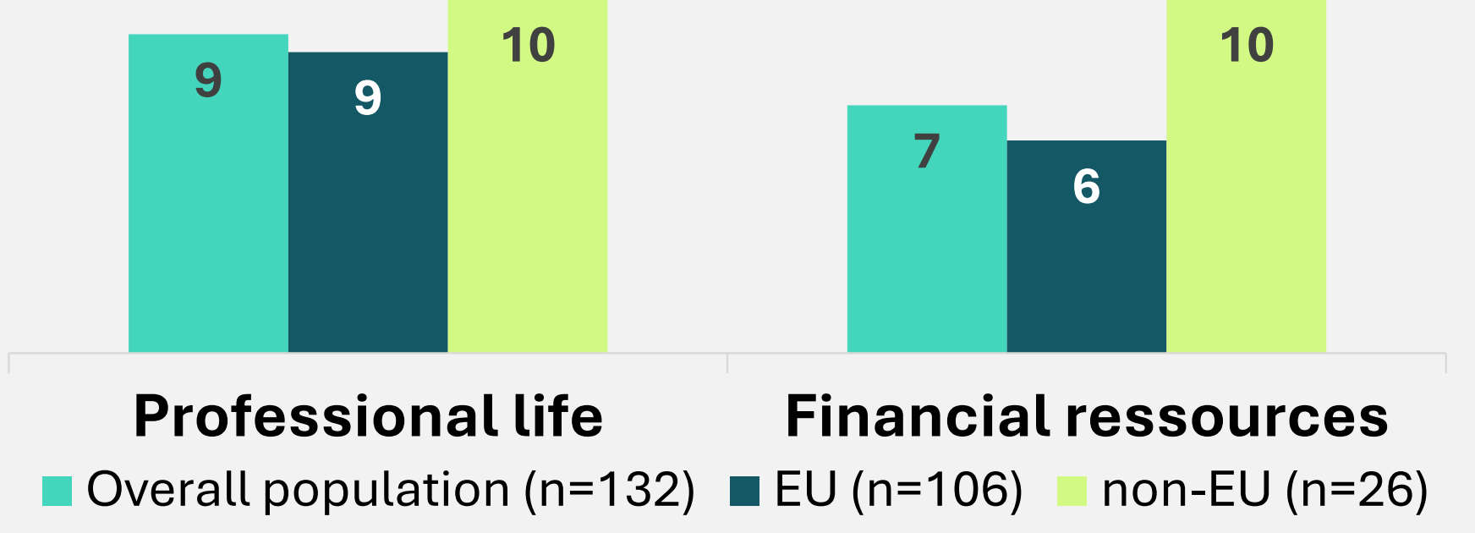
- **Outside EU**, caregivers reported a lesser level of severity for **self-care** (81% reporting level 5, vs 93% for EU), and **usual activity** (65% vs 81% for EU).
- Other dimensions were reported similarly across world regions

Geographic inequalities in care

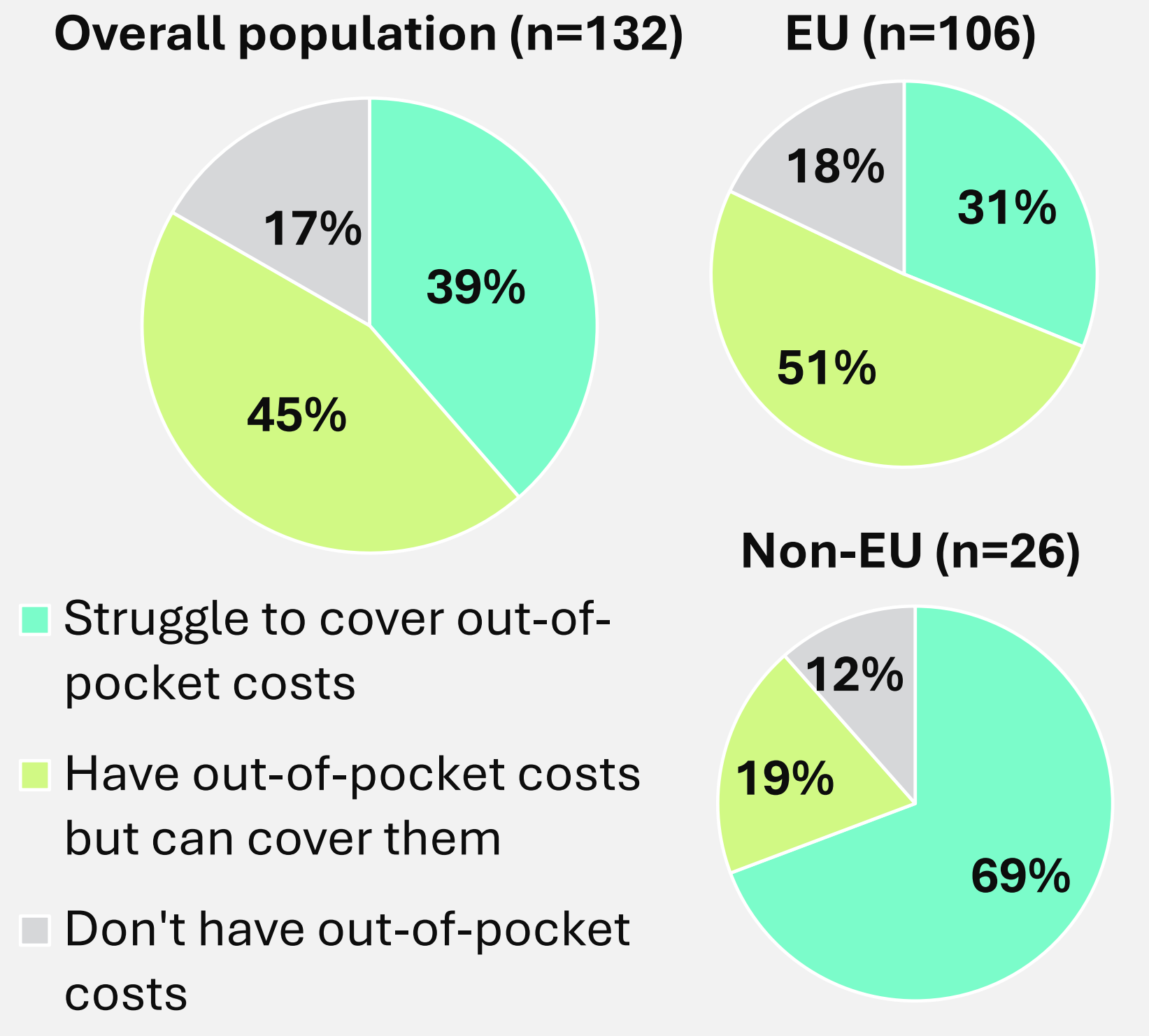
Median delay between first symptoms and diagnosis in months



Impact on caregivers' life

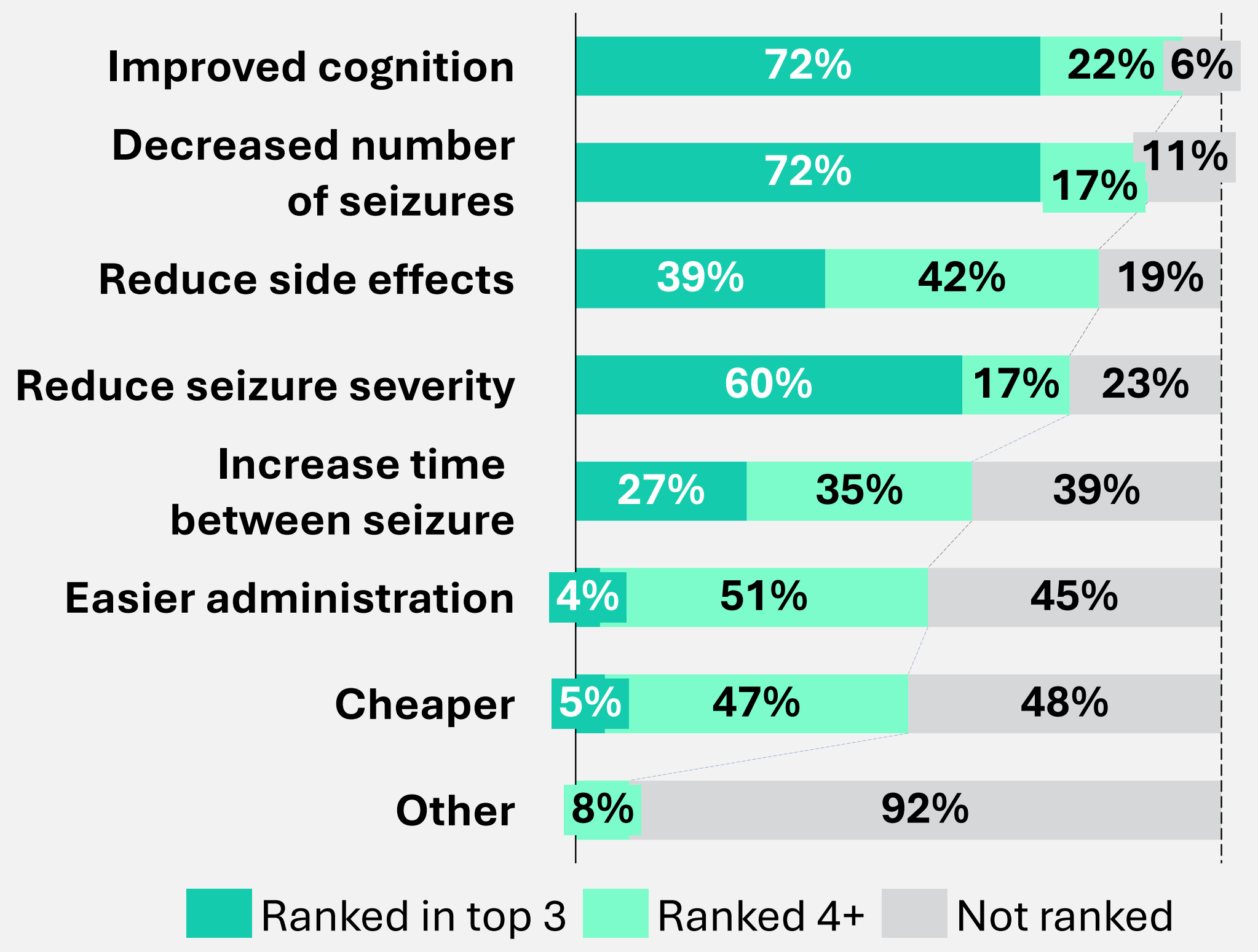


Out of pocket costs by geographic areas

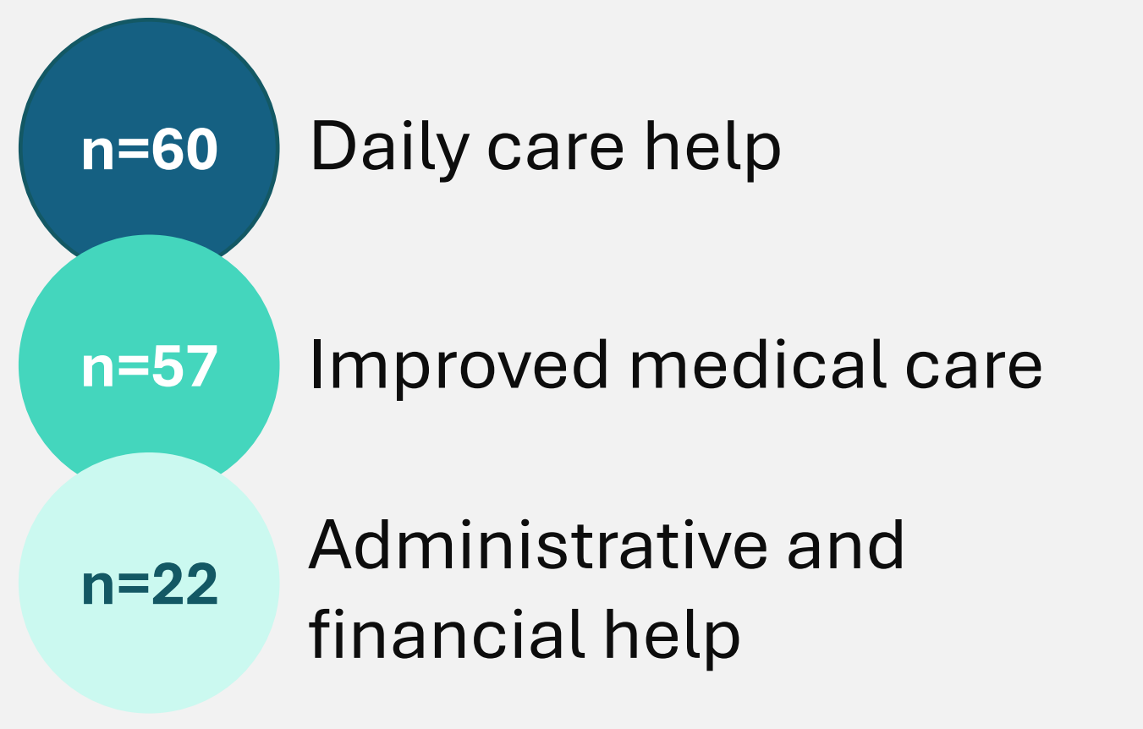


Caregivers' expectations and unmet needs

Caregivers' expectations for future treatment for epileptic seizures



- Most often mentioned unmet needs in free-text field:



The **need** for a more **complete** and **multidisciplinary care** for **adult patients** was also identified.

Conclusions

- This study highlights the **high burden of CDD** on both **patients** and **caregivers** using the **EQ5D-5L Proxy1** for the first time.
- **Regional disparities** in diagnosis and care were identified, as well as the need for **better support systems** for CDD caregivers and **effective treatments** for the persons they care for.

References & Funding statements

References: 1. Leonard H, Junaid M, Wong K, Demarest S, Downs J. Exploring quality of life in individuals with a severe developmental and epileptic encephalopathy, CDKL5 Deficiency Disorder. *Epilepsy Res*. 2021;169:106521. doi:10.1016/j.eplepsyres.2020.106521
2. Hong W, Haviland I, Pestana-Knight E, et al. Cdkl5 deficiency disorder-related epilepsy: a review of current and emerging treatment. *CNS Drugs*. 2022;36(6):591-604. doi:10.1007/s40263-022-00921-5

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