

QUALITY OF LIFE IN PEDIATRIC PATIENTS WITH SYNGAP1-RELATED NON-SYNDROMIC INTELLECTUAL DISABILITY (NSID); DATA FROM THE SYNGAP1 (MRD5) PATIENT REGISTRY



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1. BACKGROUND AND AIM

SYNGAP1-NSID is thought to result from limited functional levels of SynGAP protein, a protein critical in proper brain development and function. Predominantly affecting children, SYNGAP1 mutations lead to developmental delay, intellectual disability, and additional symptoms that are common with other causes. Here, we describe patient-reported quality of life as collected through the SYNGAP1 (MRD5) patient registry.

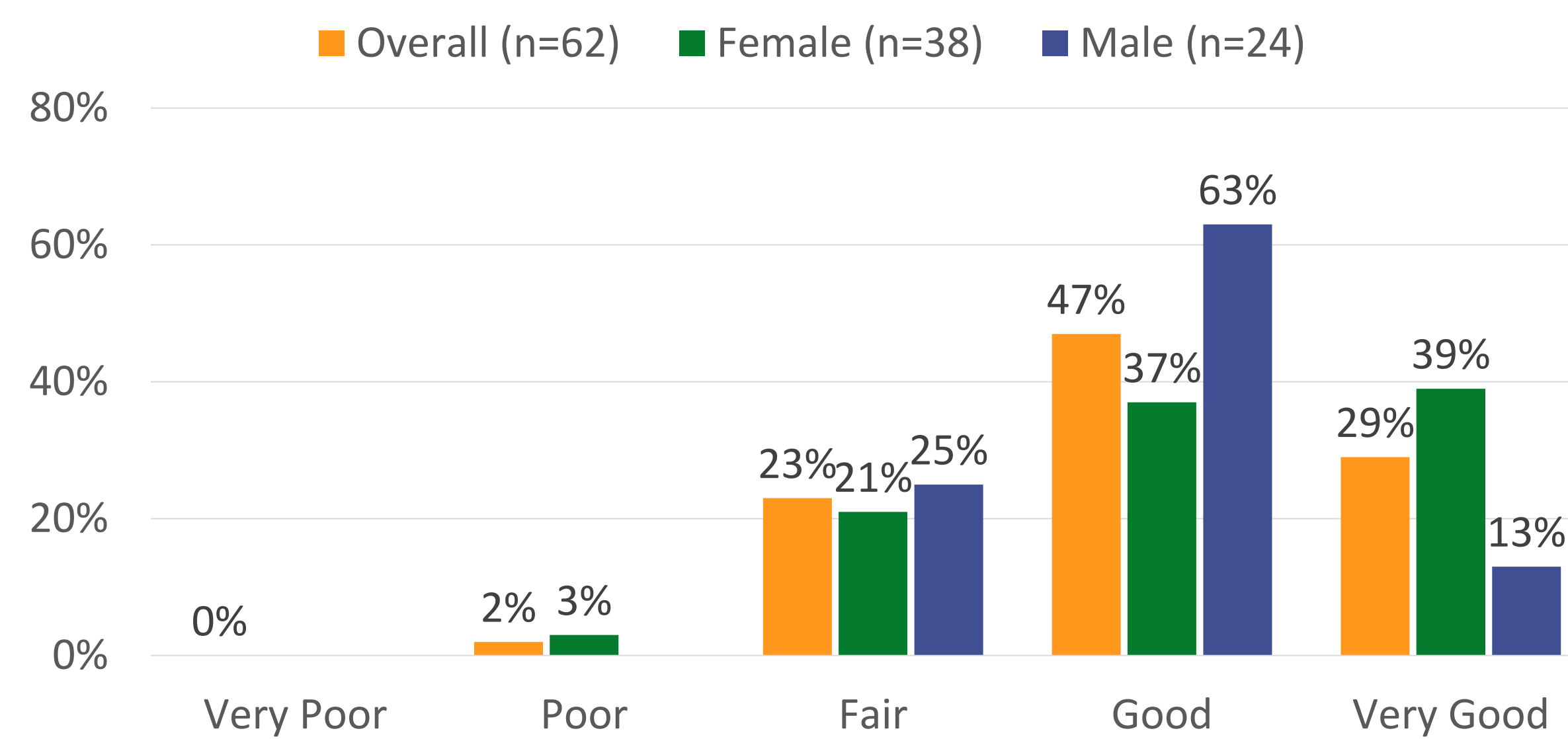
2. METHODS

The Bridge the Gap Education and Research Foundation, in partnership with the National Organization for Rare Disorders and support from the US Food and Drug Administration, launched the SYNGAP1 (MRD5) patient registry in 2017. The registry contains 13 surveys covering diagnostics, disease, treatment, care management, and quality of life. As of December 2018, 105 patients have provided data for 717 survey submissions.

The pediatric quality of life survey has 62 respondents and questions that cover the impact of the disease on the participant's life in the 4 weeks preceding their responses. Respondents in the survey were less than 18 with a median age of 4 years at diagnosis (mean of 5.18 years), 61% (38/62) female, and mostly resided in the US (56%, 35/52).

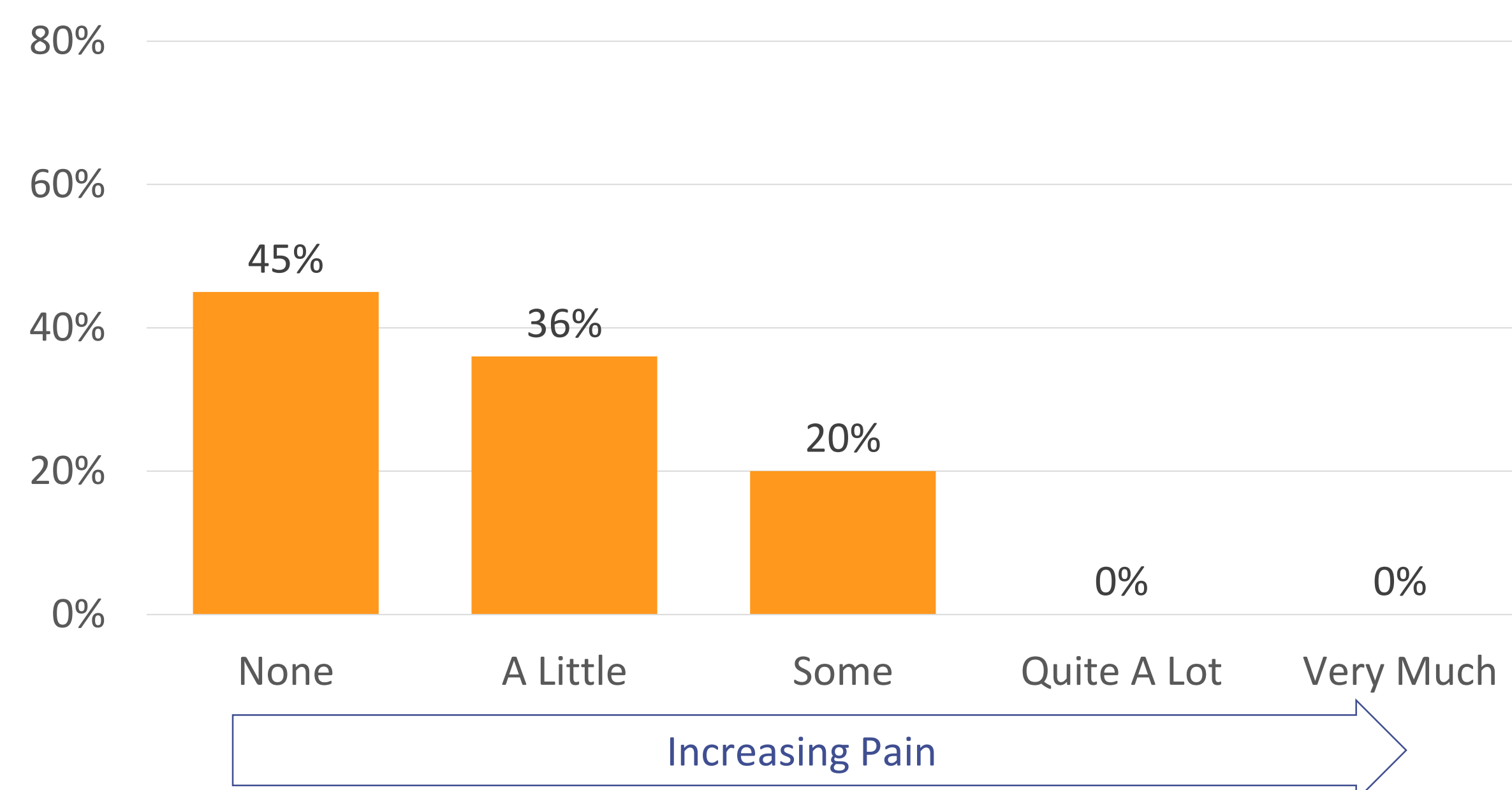
3. OVERALL HEALTH IN PAST FOUR WEEKS

During the past 4 weeks, how would the Participant describe their overall health?



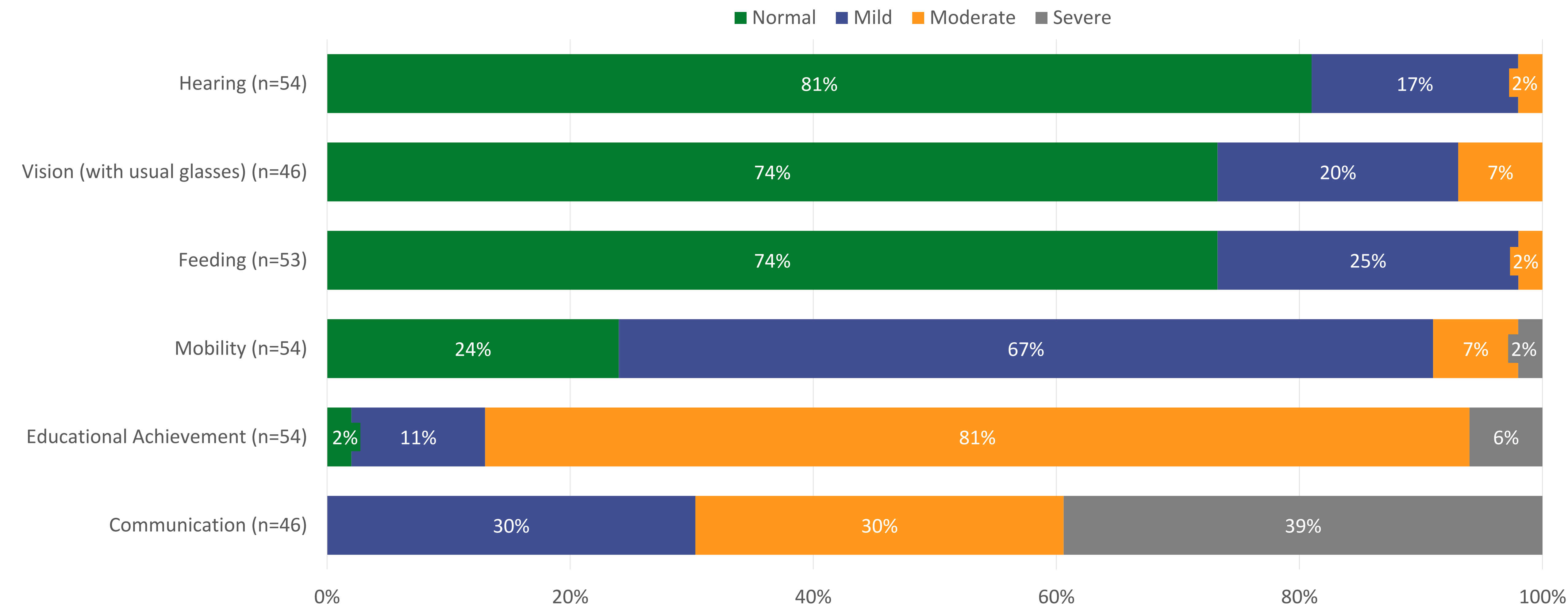
4. PAIN LEVEL IN PAST FOUR WEEKS

During the past 4 weeks, how much bodily pain/discomfort did the Participant have? (n=56)



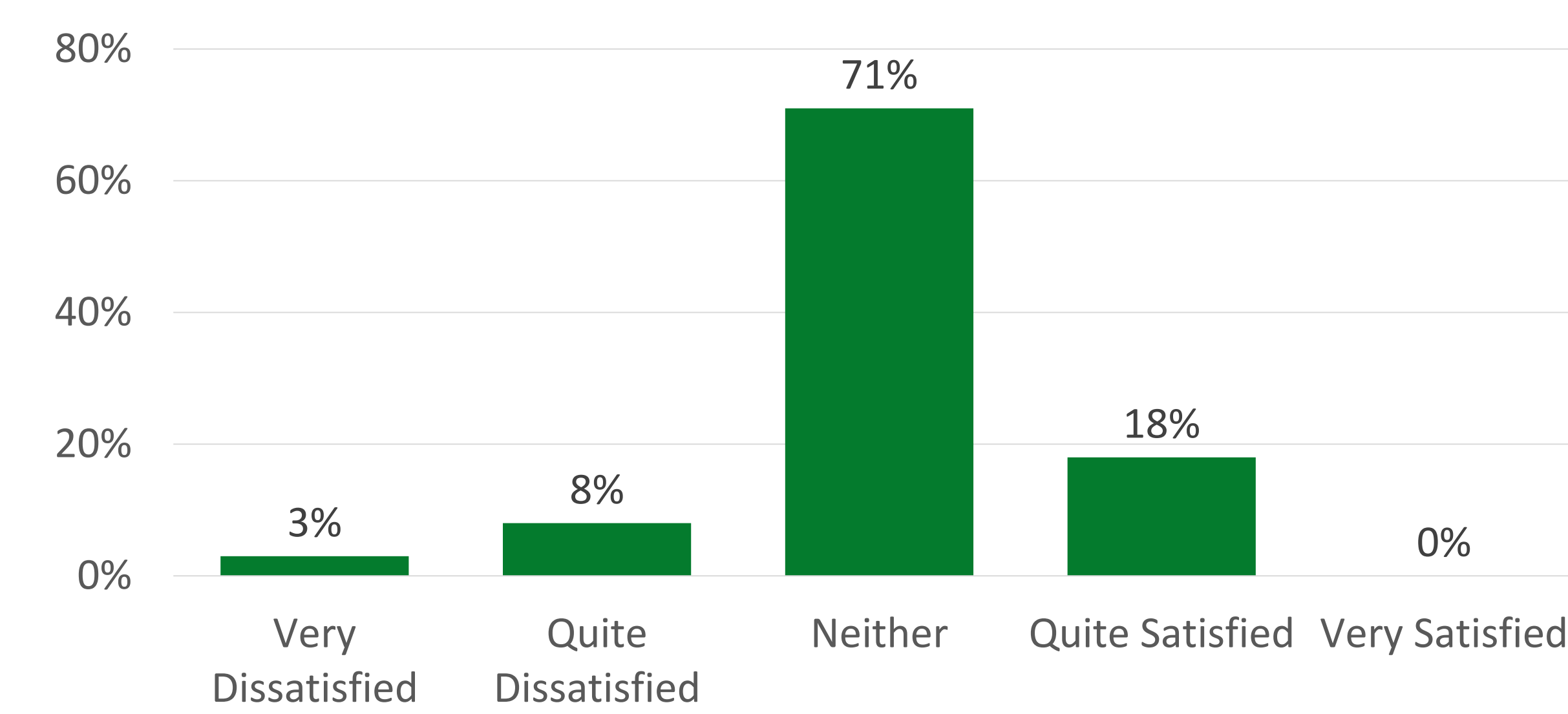
5. FUNCTIONAL IMPAIRMENT IN THE PAST FOUR WEEKS

During the past 4 weeks, to what degree were the following functions impaired?



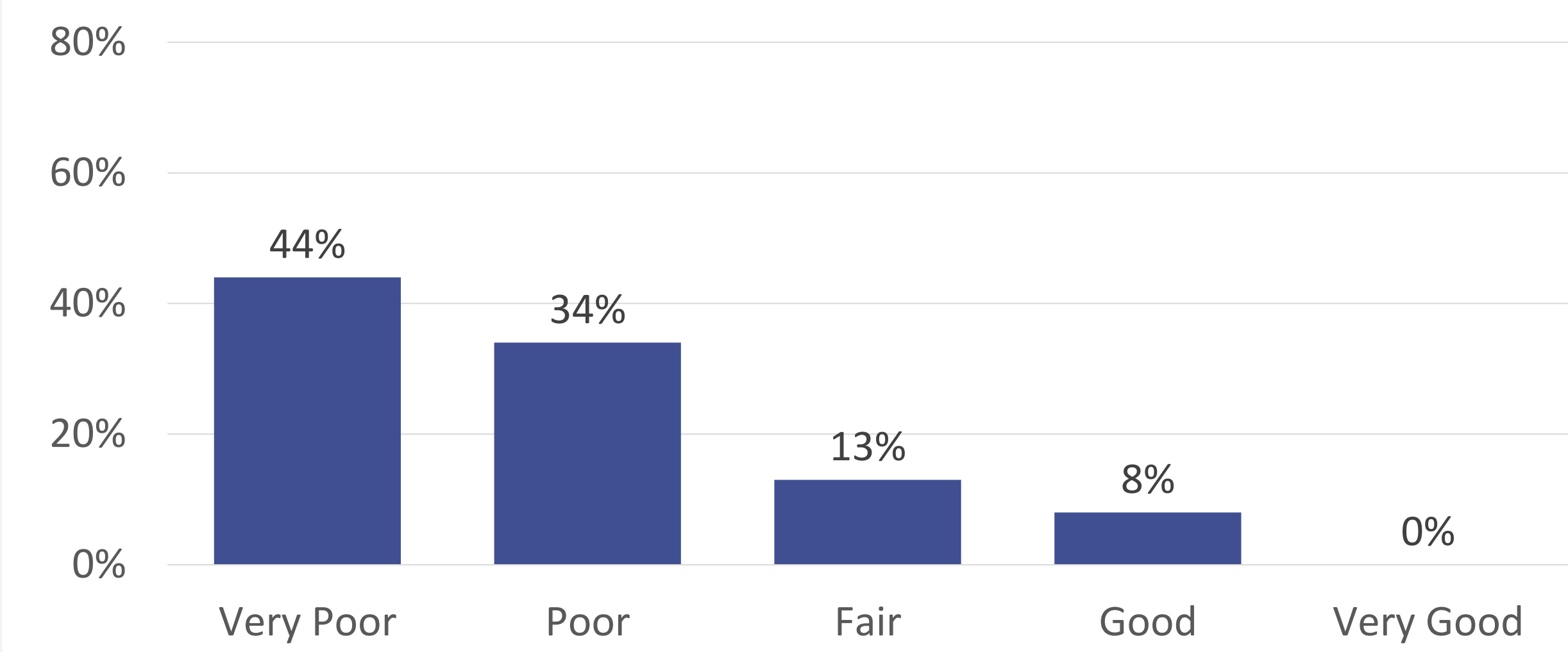
6. OVERALL QUALITY OF LIFE IN PAST FOUR WEEKS

During the past 4 weeks, how satisfied did the Participant feel with his/her abilities, looks, relationships with other people and life in general? (n=38)



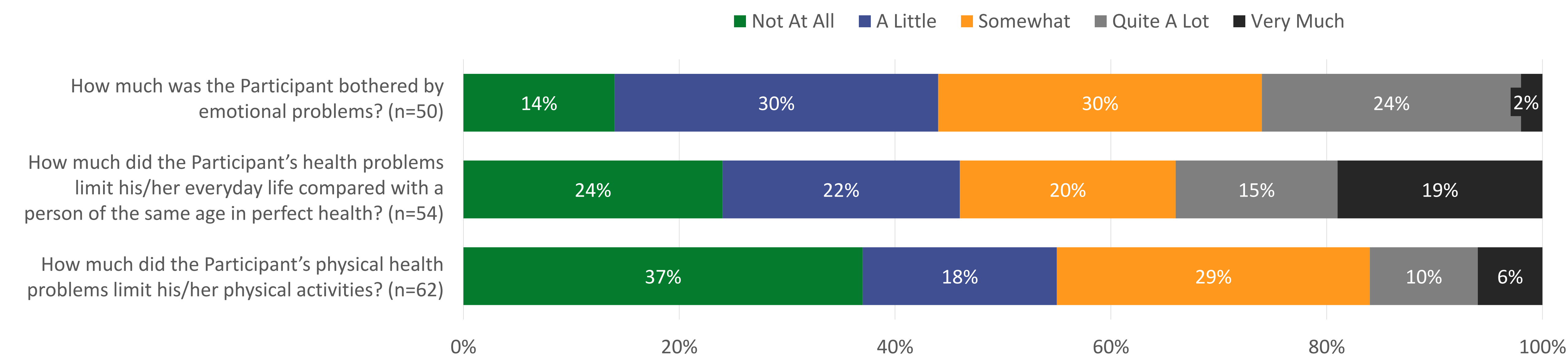
7. SOCIAL ABILITY SATISFACTION

During the past 4 weeks, the Participant's ability to interact with other people compared with other children his/her age was: (n=61)



8. PATIENT PERCEPTION OF HEALTH IMPACT

During the past 4 weeks, to what extent did the following impact the Participant?



9. SUMMARY

The Bridge the Gap Education and Research Foundation, in partnership with the National Organization for Rare Disorders and support from the US Food and Drug Administration, launched the SYNGAP1 (MRD5) patient registry in 2017. Here, we provide results gathered as of Feb 2019 from the pediatric quality of life survey (n=62). Respondents in the survey were less than 18 with a median age of 4 years at diagnosis (mean of 5.18 years), 61% (38/62) female, and mostly resided in the US (56%, 35/52).

Overall Health

- Patients were fairly positive about their overall health, with 76% (47/62) reporting it to be good or very good.
- By gender, more females (39%, 15/38) reported that their overall health was very good relative to males (13%, 3/24).
- 24% (9/38) of female patients and 25% (6/24) of male patients reported their health to be fair or poor.

Pain

- Patients reported relatively low pain levels with nearly half (45%, 25/56) reporting no pain or discomfort in the past four weeks.

Functional Impairment

- Communication was indicated as the functional area most severely impacted (39%, 18/46). None indicated normal communication functioning.
- Education achievement was severely impacted for 6% (3/54) patients though 98% (53/54) indicated less than normal functioning.
- Mobility was indicated as less than normal for 76% (41/54) patients though only 2% (1/54) reported severe impairment.
- Normal functioning for hearing (81%, 44/54), vision (74%, 34/46), and feeding (74%, 39/53) was reported for the majority of respondents.

Overall Quality of Life

- Patients were mostly neutral on their satisfaction with life in general, with 71% (27/38) reporting that they were neither satisfied nor dissatisfied.

Social Ability

- Patients reported low levels of ability to interact with others, with 78% (48/61) reporting their ability in the past four weeks to be poor or very poor.

Patient Assessment of Health Impact

- Over half of respondents (54%, 28/52) indicated that their health problems affected every day functioning.
- 86% (43/50) of respondents indicated some burden due to emotional problems, with nearly a quarter (26%, 13/50) responding emotional problems impacted them "quite a lot" or "very much."
- 62% (31/50) indicated that health issues limited the ability to perform activities s/he enjoys most.

Pediatric patients in the registry have impacted quality of life. Data collection through the SYNGAP1 (MRD5) patient registry continues with the intent of raising awareness of the disease, enabling treatment development, and improving patient Quality of Life.