

UNDERSTANDING DISEASE AND BURDEN IN SYNGAP1-RELATED NON-SYNDROMIC INTELLECTUAL DISABILITY (NSID) PATIENTS USING A PATIENT REGISTRY DATABASE

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1. BACKGROUND AND AIMS

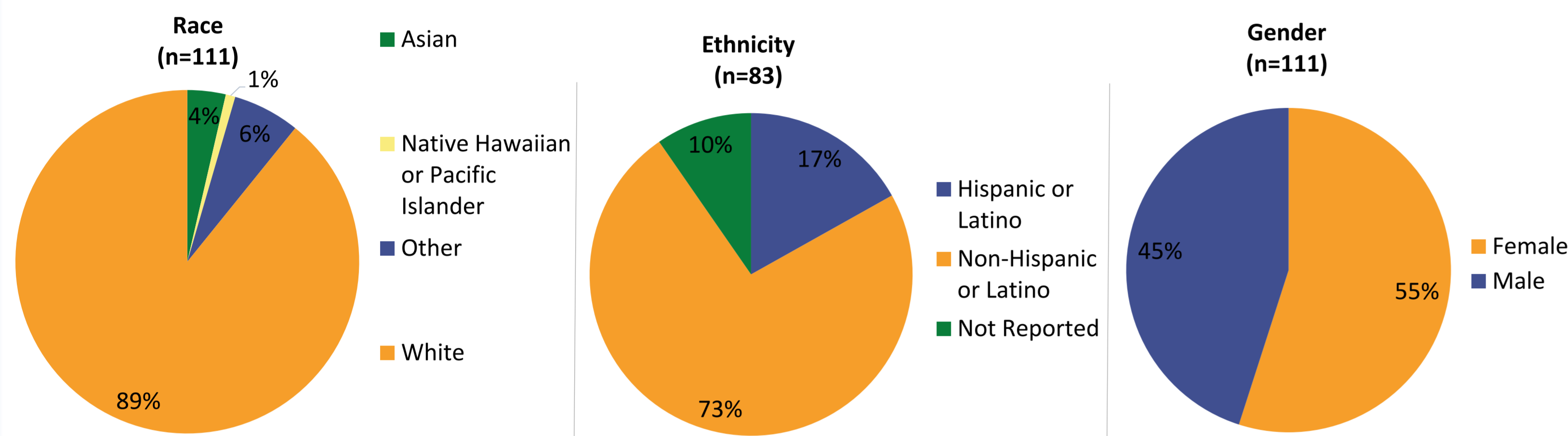
SYNGAP1-NSID is thought to result from limited functional levels of SynGAP protein, a protein critical in proper brain development and function. Predominantly affecting children, SYNGAP1 mutations lead to developmental delay, intellectual disability, and additional symptoms that are common with other causes. As such, confirmation of SYNGAP-related NSID is through genetic testing. To improve awareness and understanding of SYNGAP-related NSID and better inform treatment development, 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 (Mental retardation, autosomal dominant 5 [MRD5]) patient registry in 2017. In this analysis we describe patient demographics, diagnoses, and quality of life of the registry patients.

2. METHODS

The registry contains 13 surveys covering diagnostics, disease, treatment, care management, and quality of life. As of March 2019, 209 patients were enrolled in the registry, 112 patients have provided data via 808 survey submissions.

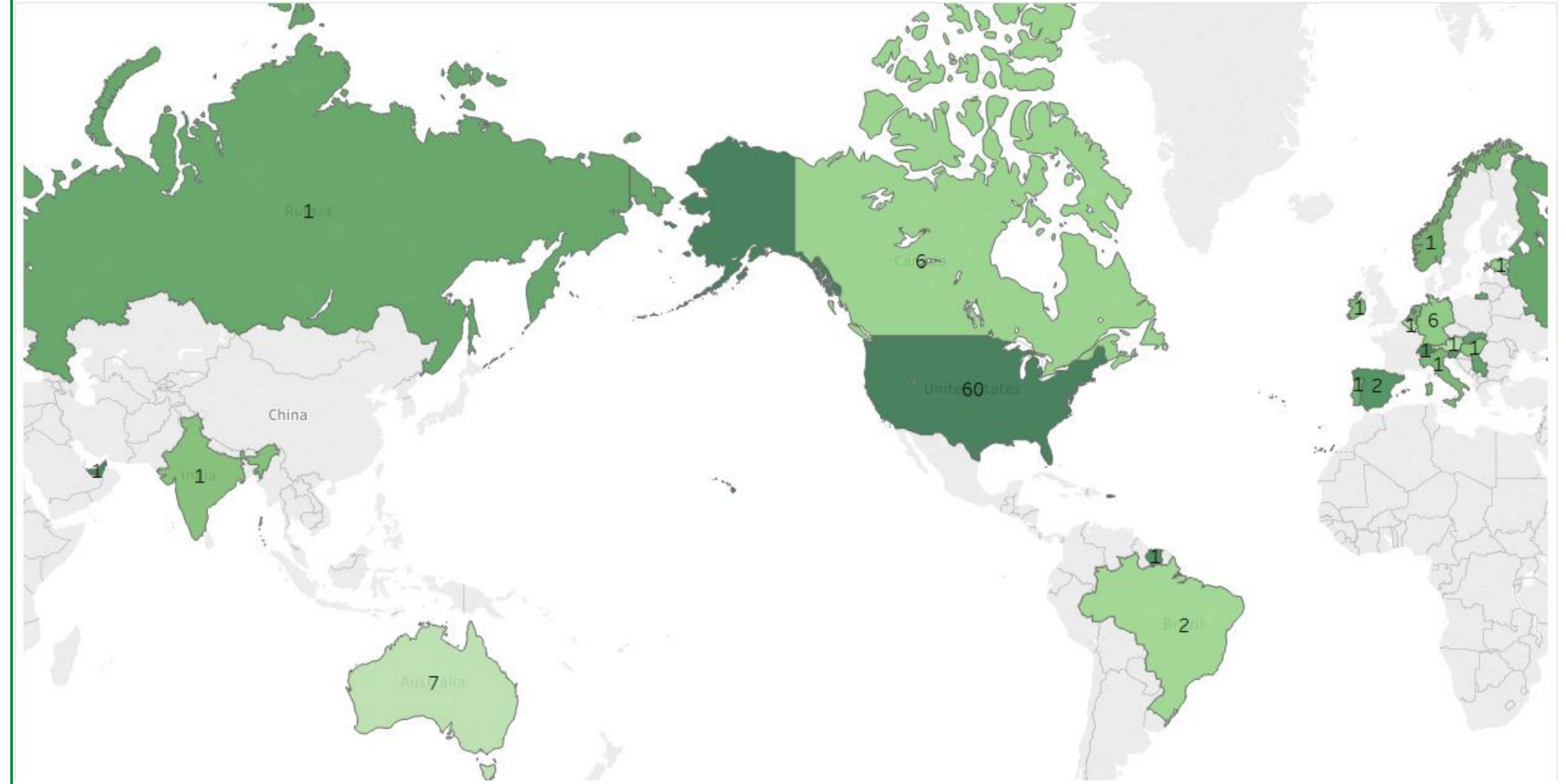
3. DEMOGRAPHICS: RACE AND GENDER

Participants in the registry were mostly white (89%, 99/111), non-Hispanic or Latino (73%, 61/83), and over half of them were female (55%, 61/111).



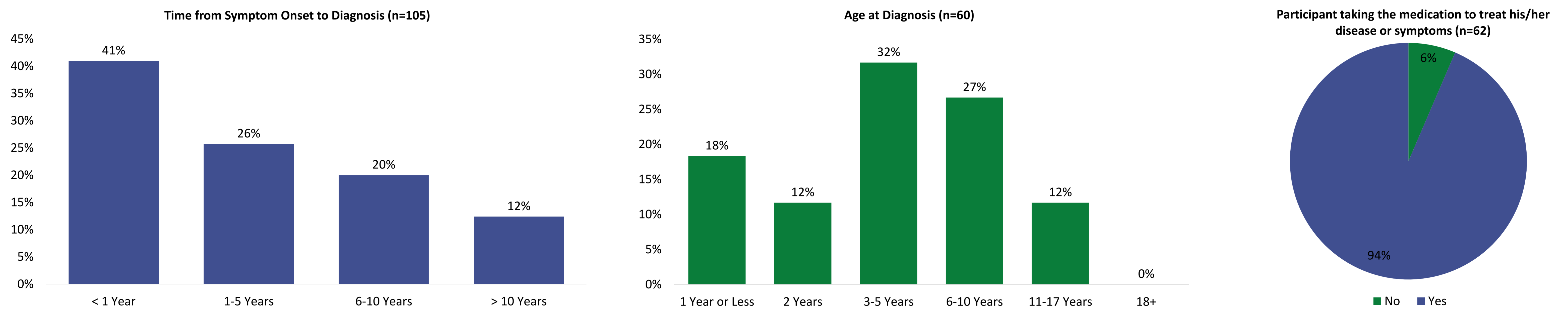
4. DEMOGRAPHICS: LOCATION

Participants in the registry are located in 24 countries, with the majority of participants in the US (54%, 60/112).



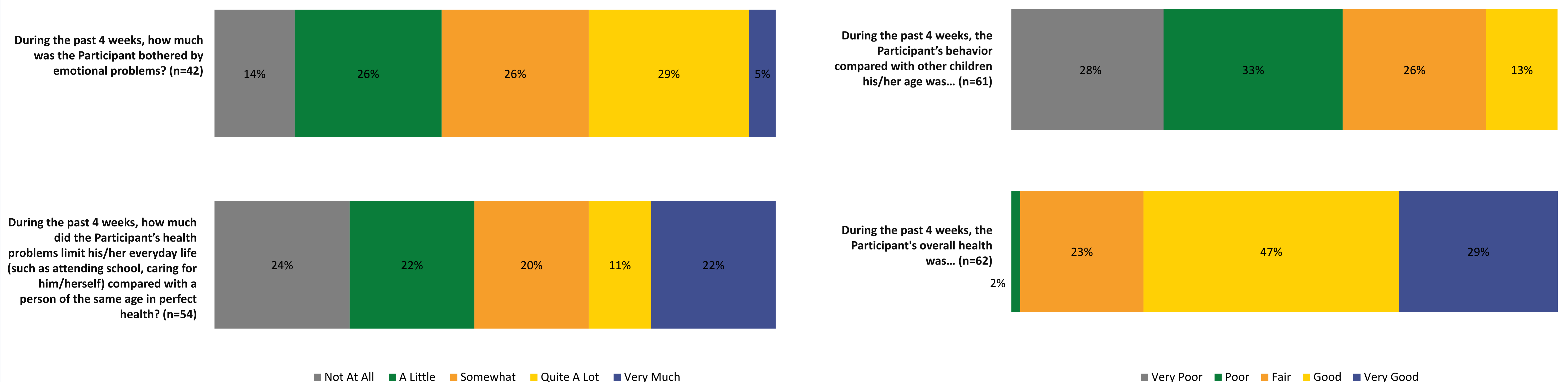
5. DIAGNOSIS AND TREATMENT

- While 41% of the participants were diagnosed within one year of the symptom onset, 32% of the participants were diagnosed after 6 or more years since the symptom onset.
- All registry participants were diagnosed below the age of 18 and 52% were diagnosed by the age of 5.
- Most participants (94%) reported taking medications for management of their disease.



6. QUALITY OF LIFE

While 76% of the patients reported having good or very good overall health, only 46% had little or no limitations of their every day activities due to their disease, 40% reported having few emotional problems, and only 13% reported good behavior compared to their peers.



7. CONCLUSIONS

Patients in the registry reported a significant disease burden impacting their quality of life physically and emotionally. Data collection through the SYNGAP1 (MRD5) patient registry continues with the intent of raising awareness of the disease and enabling clinical trial recruitment to support new treatment discovery.