

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

Weldon M, BS<sup>1</sup>, Holder, J M.D., Ph.D.<sup>1</sup>, Rumbaugh, G Ph.D.<sup>1</sup>, Smith-Hicks, C. MD PhD<sup>1</sup>, Boulanger V<sup>2</sup>, Seebald A<sup>2</sup>, Rossov S<sup>2</sup>, Milligan K<sup>3</sup>, Radtchenko J<sup>3</sup>

<sup>1</sup>Bridge the Gap – SYNGAP Education and Research Foundation, Cypress, TX, USA, <sup>2</sup>National Organization for Rare Disorders (NORD), Danbury, CT, USA, <sup>3</sup>TRIO Health Analytics, La Jolla, CA, USA

## 1. BACKGROUND AND AIM

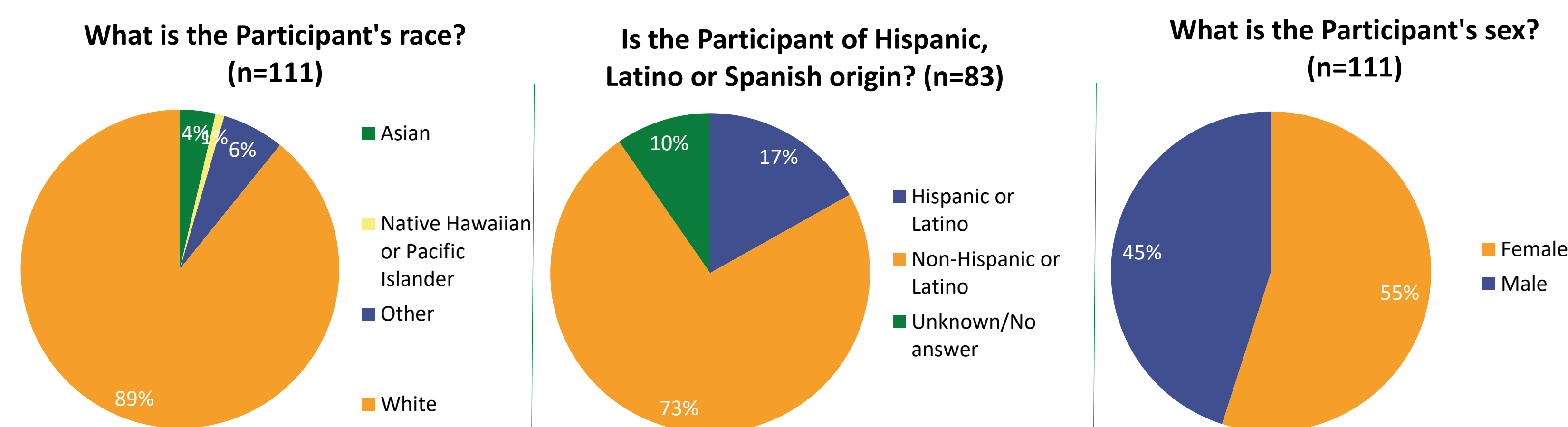
Predominantly affecting children, SYNGAP1 mutations lead to developmental delay, intellectual disability, and additional symptoms that are common with other causes. As such, confirmation of SYNGAP-related NSID is through genetic testing. To improve awareness and understanding of SYNGAP-related NSID and 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 (MRD5) patient registry in 2017. Here, we describe patient demographics, diagnoses, and quality of life in registry patients.

## 2. METHODS

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

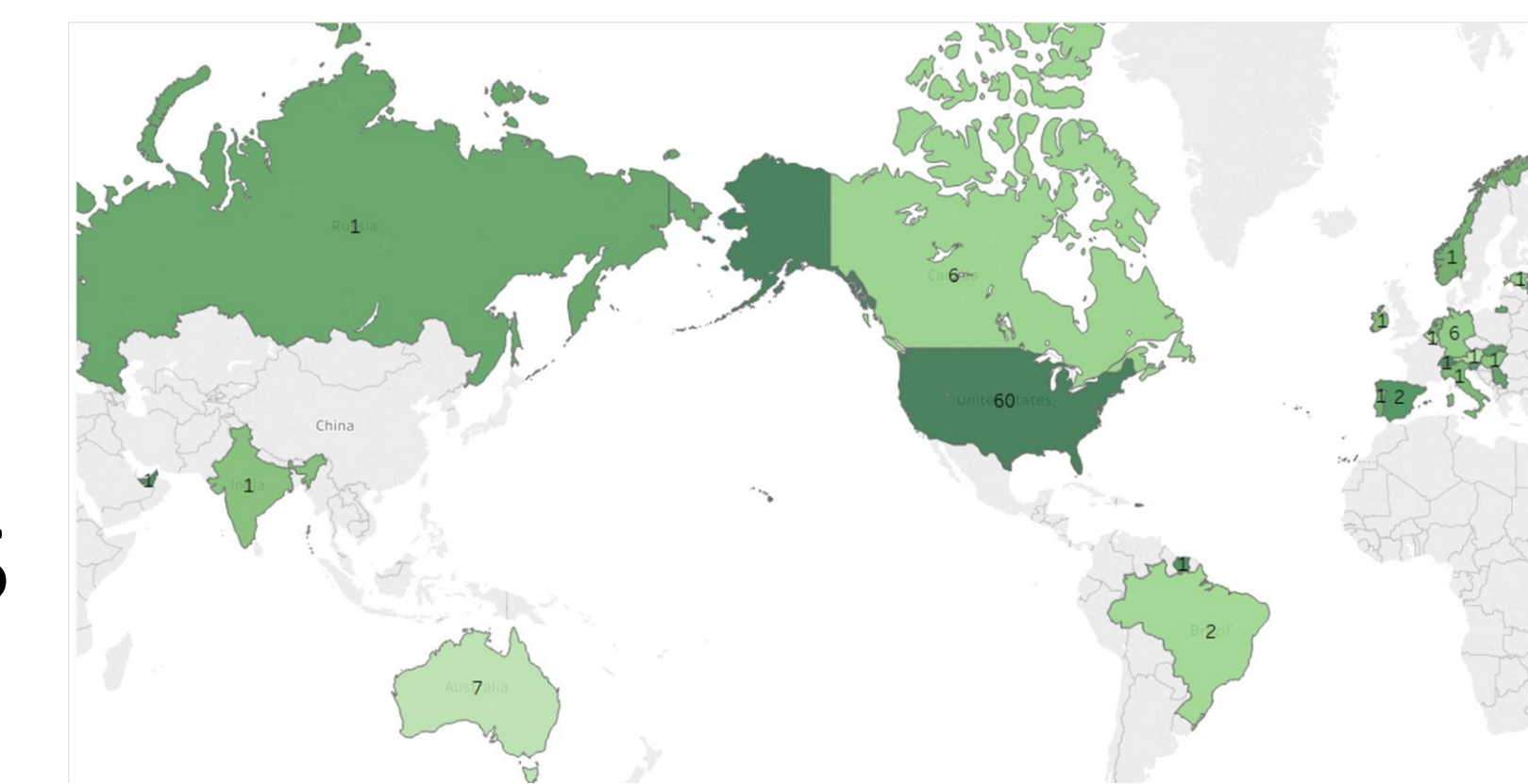
## 3. PARTICIPANT RACE AND SEX

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



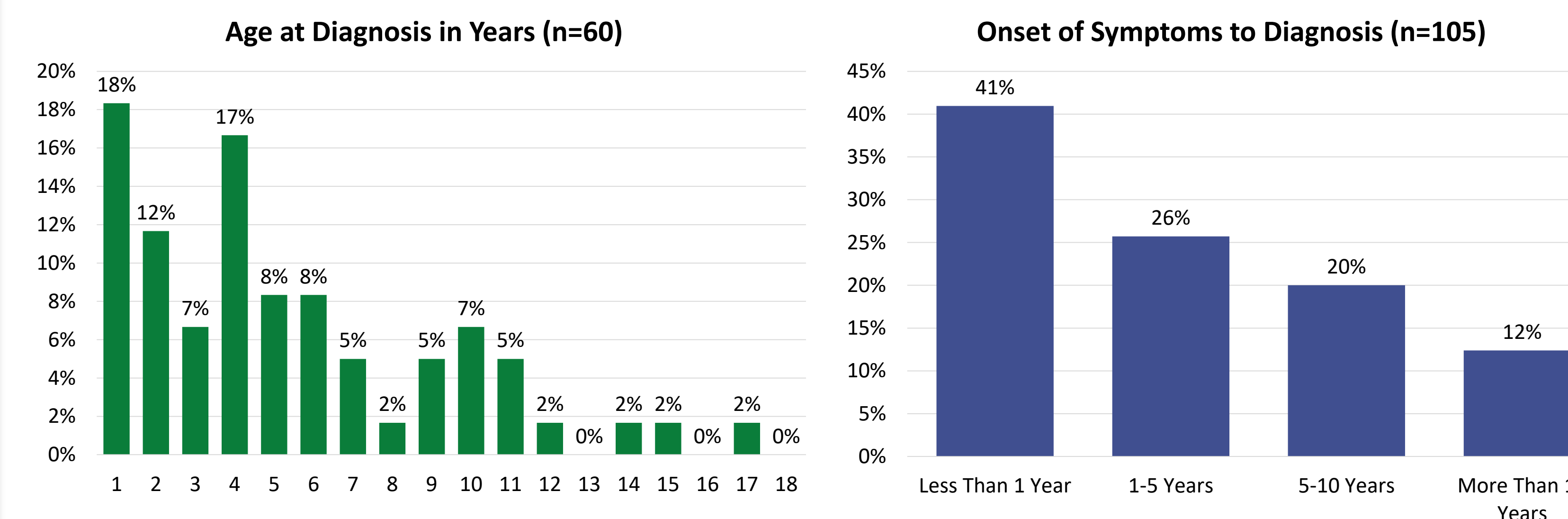
## 4. COUNTRY OF RESIDENCE

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



## 5. DIAGNOSIS

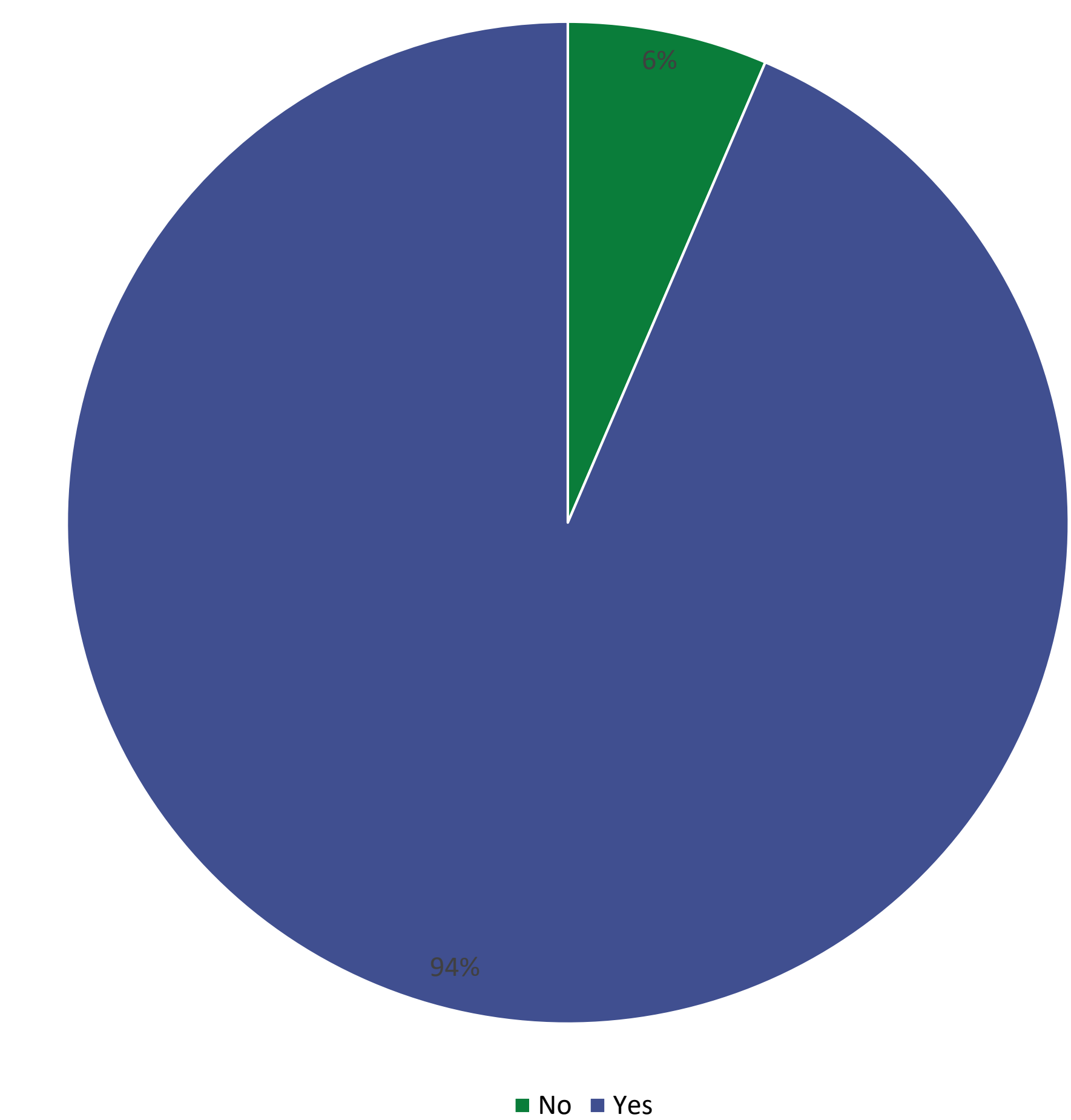
All respondents indicated diagnosis before age 18, with 54% diagnosed before age 5. 41% of participants were diagnosed within one year of symptom onset.



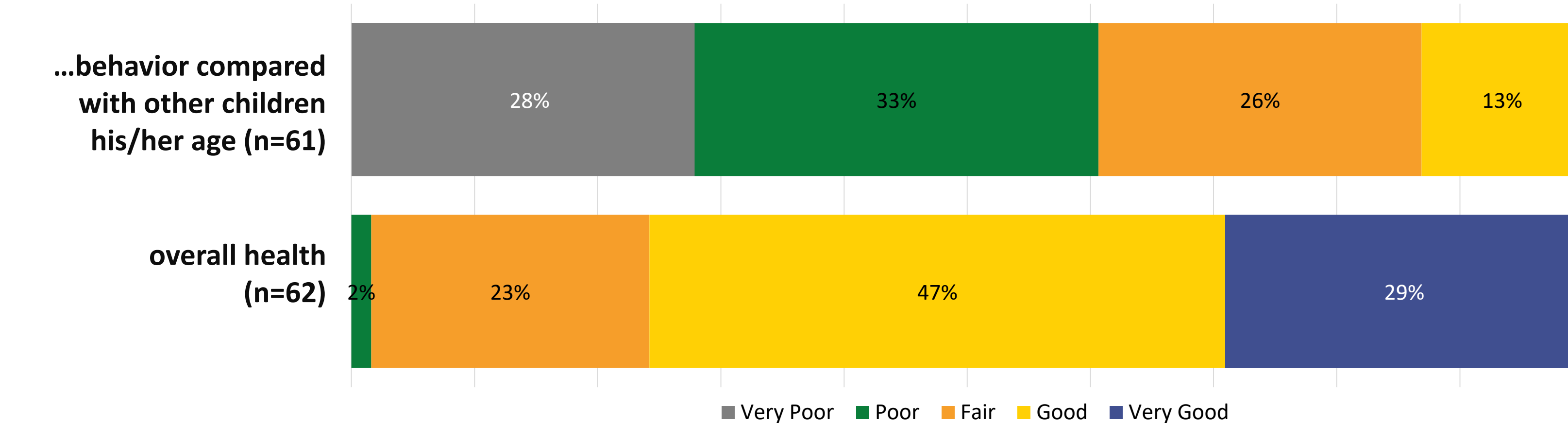
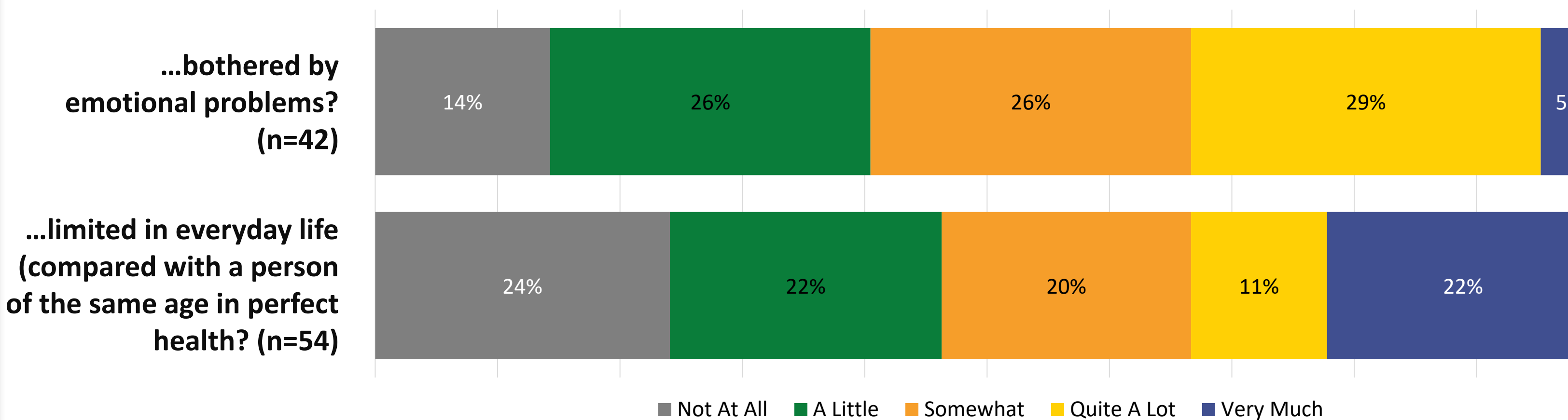
## 6. TREATMENT

Most participants (94%) indicated that they took medication to treat side effects.

Is/was the Participant taking the medication to treat his/her rare disease or rare disease related symptoms? (n=62)



## 7. QUALITY OF LIFE – DURING THE PAST 4 WEEKS



## 8. CONCLUSIONS

Patients in the registry have significant disease burden and impacted quality of life. Data collection through the SYNGAP1 (MRD5) patient registry continues with the intent of raising awareness of the disease and enabling the development of treatments.