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

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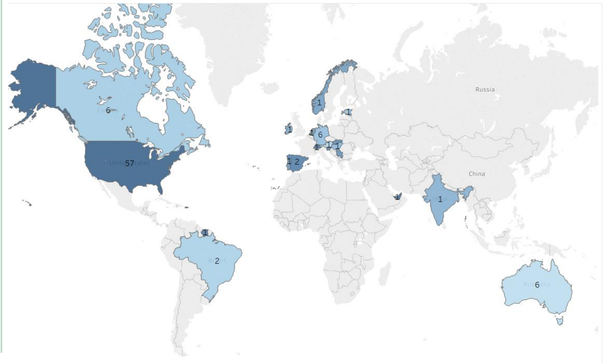
1. BACKGROUND AND AIM

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. 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 and diagnoses.

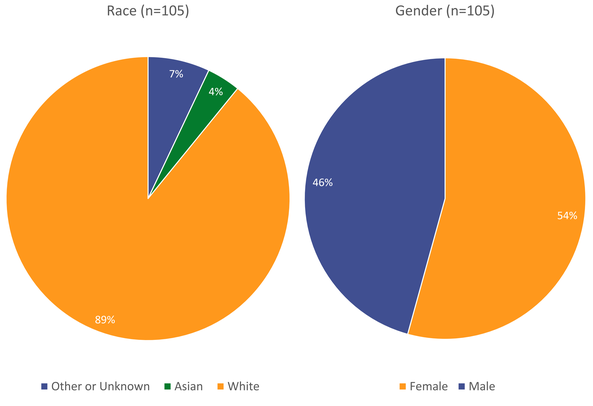
2. METHODS

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.

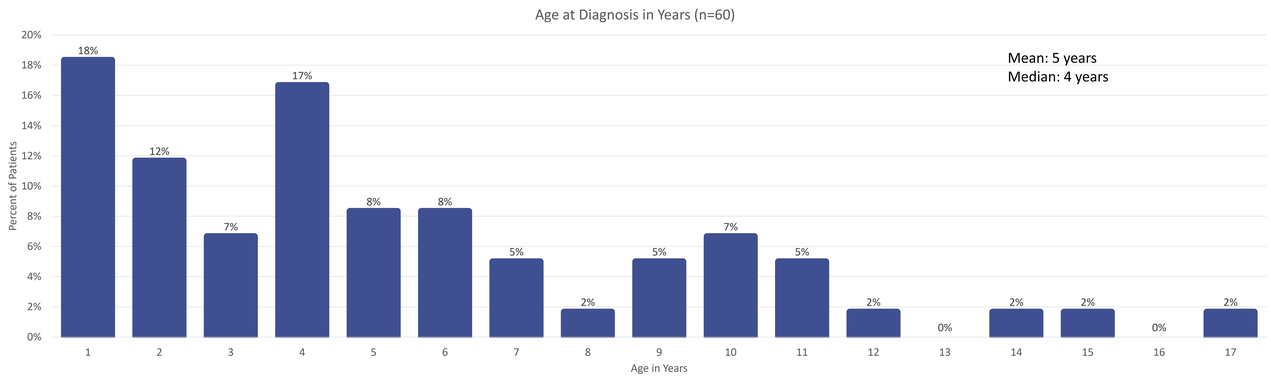
3. LOCATION



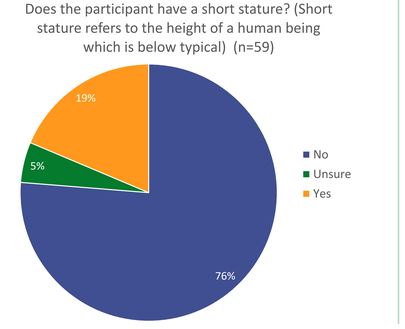
4. RACE AND GENDER



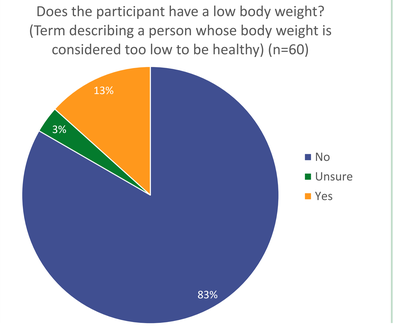
5. AGE AT DIAGNOSIS



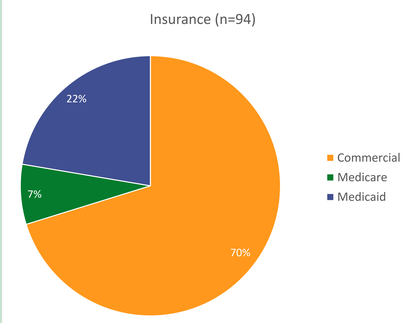
6. STATURE



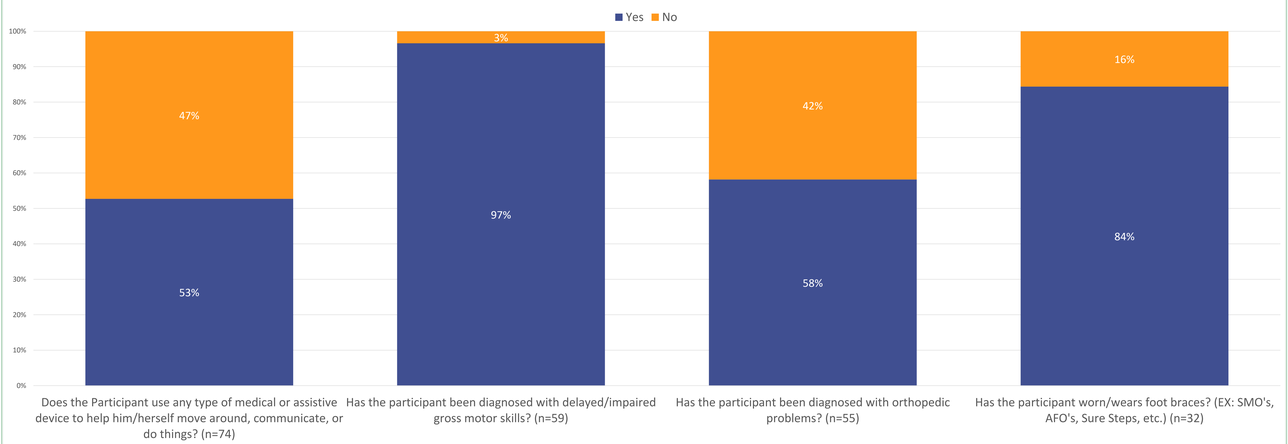
7. BODY WEIGHT



8. INSURANCE



9. PHYSICAL HEALTH AND MOVEMENT



9. SUMMARY

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 (MRD5) patient registry in 2017. Here, we describe patient demographics and diagnoses.

Location
- Participants are located in 21 countries, 54% (57/105) US based.

Race and Gender
- Participants are mostly white (89%, 93/105) and female (54%, 57/105).

Age at Diagnosis
- Participants were diagnosed at a mean age of 5 years and a median age of 4 years

Stature
- 19% (11/59) of participants have a shorter than average stature.

Body Weight
- 13% (8/60) of participants have a lower than average body weight.

Insurance
- 70% (66/94) of participants are covered by commercial insurance.

Physical Health and Movement
- Over half of respondents (53%, 39/74) indicated that they use some kind of medical device to assist in movement or communication.
- Nearly all participants (97%, 57/59) were diagnosed with delayed or impaired gross motor skills.
- 58% (32/55) of participants indicated that they had been diagnosed with orthopedic problems.
- Most participants (84%, 27/32) have worn or currently wear foot braces.

Conclusion

Data collection through the SYNGAP1 (MRD5) patient registry continues with the intent of raising awareness of the disease and enabling the development of treatments.