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 of 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.

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 727 survey submissions.

3. LOCATION
The majority of patients (62%, 66/105) are Americans. In the United States, the patient registry is most heavily represented in California (28%, 29/105) and Texas (9%, 9/105). The registry also includes participants from 20 other countries, with the largest number of patients coming from France (5%, 5/105) and the United Kingdom (4%, 4/105).

4. RACE AND GENDER
Participants are mostly white (89%, 93/105) and female (54%, 57/105).

Racial distribution includes 9% (9/105) White, 7% (7/105) Black, 5% (5/105) Hispanic, and 3% (3/105) Asian.

5. AGE AT DIAGNOSIS
Most participants (84%, 27/32) have worn or currently wear foot braces.

Nearly all participants (97%, 57/59) were diagnosed with delayed or impaired gross motor skills. The median age at diagnosis was 4 years (Mean: 5 years).

6. STATURE
Does the participant have a short stature? (Short stature refers to the height of a human being which is below typical) (n=59)
- Yes: 13% (8/60)
- No: 87% (52/60)
- Unsure: 0%

7. BODY WEIGHT
Does the participant have a low body weight? (Term describing a person whose body weight is considered too low to be healthy) (n=60)
- Yes: 8% (5/60)
- No: 92% (54/60)
- Unsure: 0%

8. INSURANCE
Insurance (n=94)
- Commercial: 78% (74/94)
- Medicare: 10% (9/94)
- Medicaid: 1% (1/94)
- Other or Unknown: 9% (8/94)

9. PHYSICAL HEALTH AND MOVEMENT
The majority of participants (69%, 70/105) report that they device to help them move around, communicate, or do things.

Has the participant been diagnosed with orthopedic problems? (n=63)
- Yes: 43% (27/63)
- No: 57% (36/63)
- Unsure: 0%

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.