About this Report

The report shows data collected in Simons Searchlight from PPP2R5D participants. Data from other published research are not included.

Data presented in this report come primarily from the medical history phone call. The data are current and may reflect information not yet available in an official researcher data release.

Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.

This report may look different from previous reports because we have streamlined our process and updated the variants.

**Variants**

Graphs include individuals with variants classified as pathogenic or likely pathogenic unless otherwise noted. Variants of uncertain significance (VUS) and less typical variants are not included.

We reassess variants classified as VUS annually.

**Ages in Years**

Individual participants: 61

- 0-4: 25
- 5-9: 19
- 10-14: 7
- 15-19: 3
- 20-24: 4
- 25+:

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**Participation & Registration**

Learn more at SimonsSearchlight.org

**PPP2R5D Progress**

Includes families registered with all variant types

- Registered Families: 101
- Person with Genetic Change Consented: 100
- Lab Report Approved: 83
- Medical History Phone Call: 67
- Data Released to Researchers*: 77

* Most recent data release - February 2021
**Developmental & Behavioral Diagnoses**
Individual participants: 61

- **Intellectual Disability/Development Delay**: 51
- **Autism**: 20
- **Language Impairment**: 31
- **ADHD**: 4
- **OCD**: 2
- **Anxiety**: 2

**Neurological Problems**
Individual participants: 61

- **Clumsy**: 15
- **Low Muscle Tone**: 5
- **High Muscle Tone**: 2
- **Large Head Size**: 7
- **Small Head Size**: 3
- **Movement Abnormalities**: 2
- **Cerebral Palsy**: 3

**Gastrointestinal Problems**
Individual participants: 61

- **GERD**: 19
- **Diarrhea**: 17
- **Constipation**: 21
- **Inflammatory Bowel Disease**: 2
- **Celiac Disease**: 1
- **Other**: 1

**Seizures**
Individual participants: 61

- **Myoclonic**: 9
- **Tonic**: 1
- **Complex Partial**: 6
- **Atonic Drop Attack**: 5
- **Infantile Spasm**: 2
- **Petit Mal**: 5
- **Grand Mal**: 7

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