

Simons Searchlight Registry Update

MED13L

March 2021

About this Report

The report shows data collected in Simons Searchlight from MED13L 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.

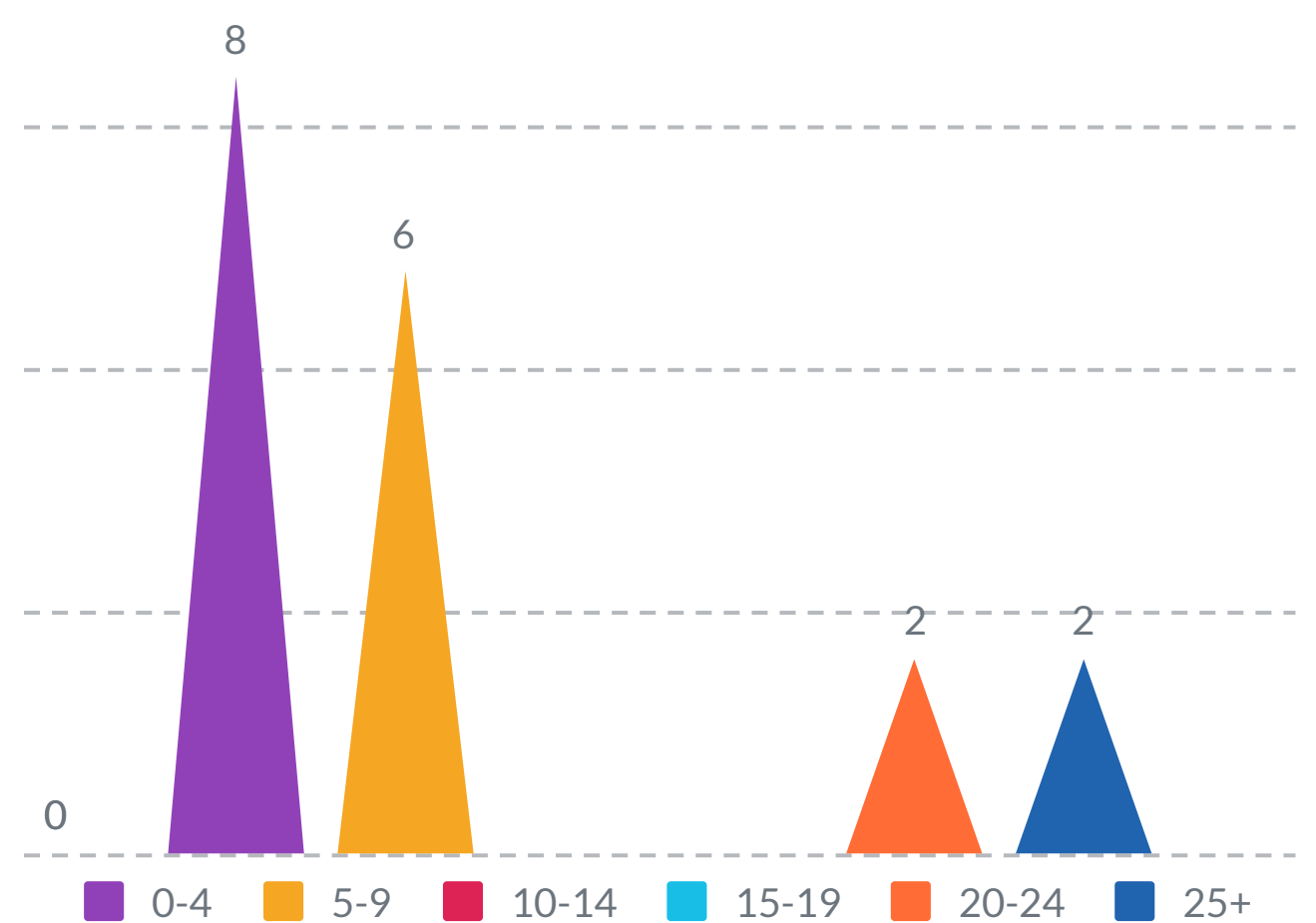
Variants

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.

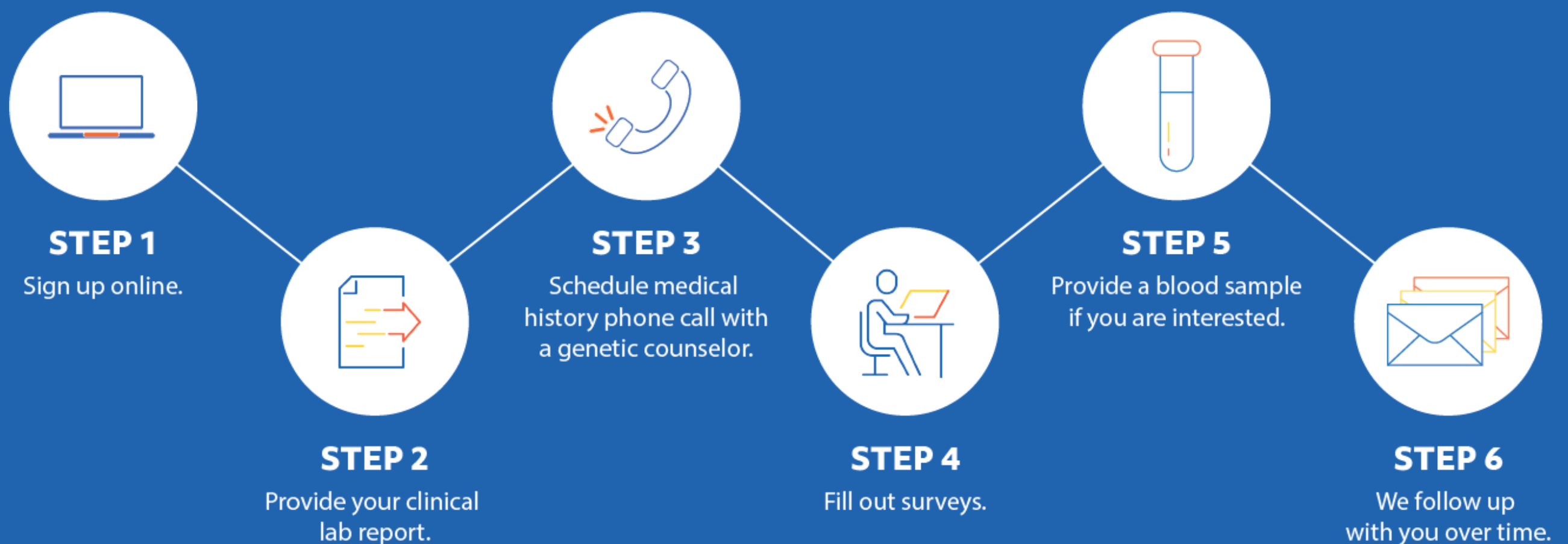
Ages in Years

Individual participants:18



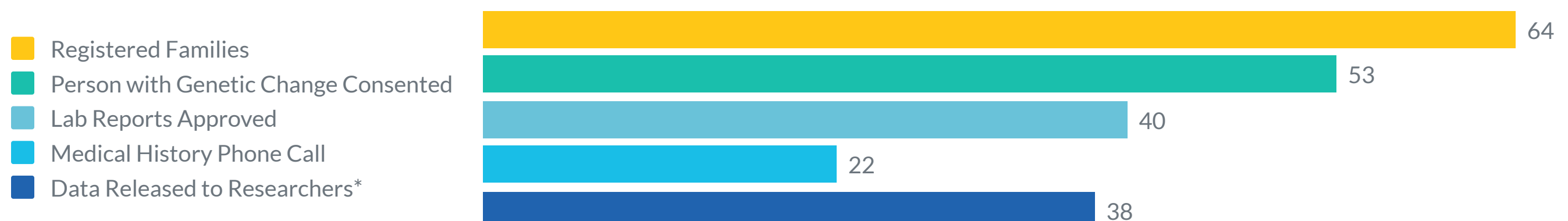
Participation & Registration

Learn more at SimonsSearchlight.org



MED13L Progress

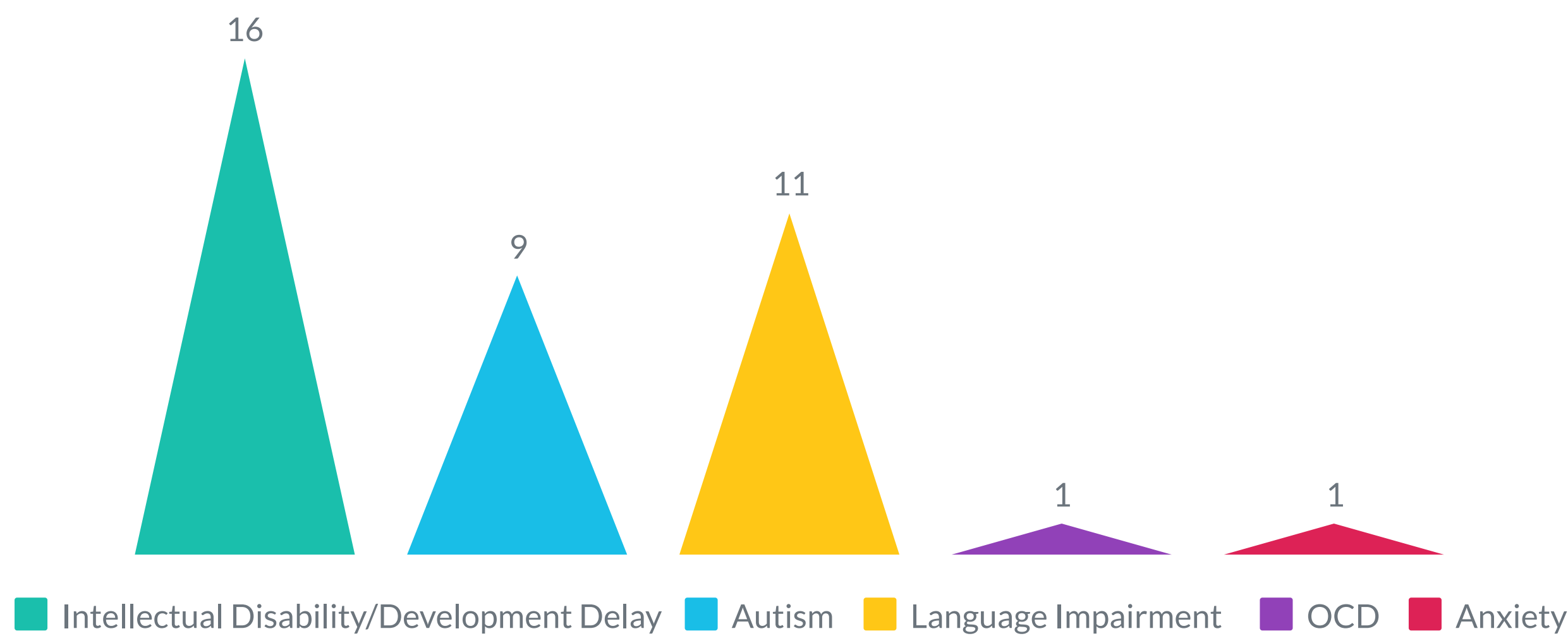
Includes families registered with all variant types



* Most recent data release - February 2021

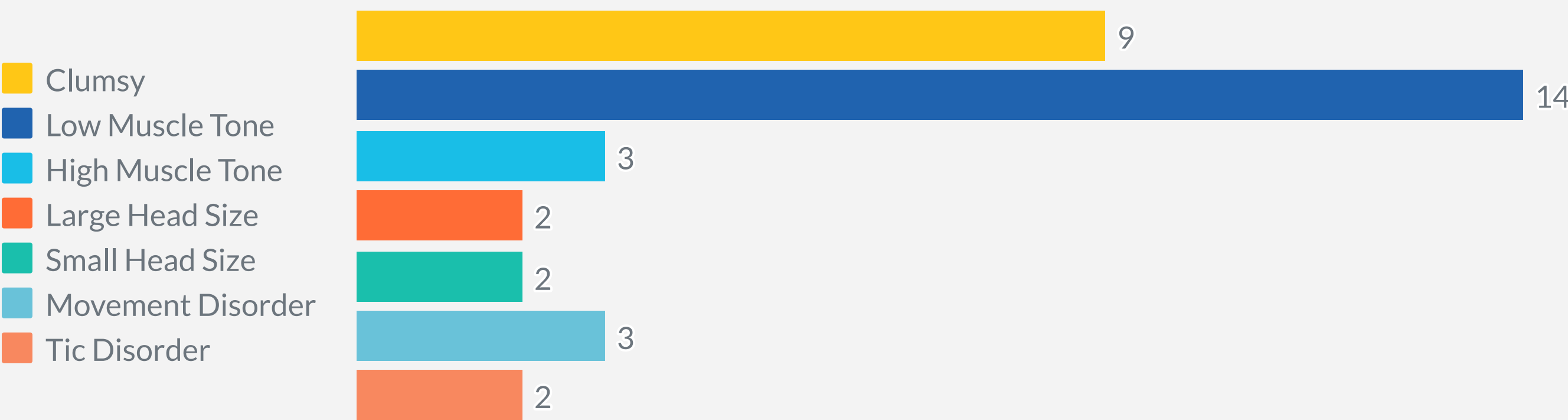
Developmental & Behavioral Diagnoses

Individual participants: 18



Neurological Problems

Individual participants: 18



Gastrointestinal Problems

Individual participants: 18

