Applications
Applications received: 5882
Applications under review: 268
Participants accepted: 2354

Evaluations
Participants evaluated: 1946
Participants diagnosed: 576
Participants with exome and/or genome sequencing complete: 1799
Participants with metabolomic analyses complete: 247
Participants with samples in the Central Biorepository: 1162
Variants evaluated by the Model Organisms Screening Center: 418

Data Sharing
Manuscripts published: 180
ClinVar submissions: 595
Participants with data available in dbGaP: 1319
Participants with data submitted to PhenomeCentral: 1845

Last updated: July 8, 2022
Please note: family members are not included in these calculations. The number of applications received does not include applications that were withdrawn. Some of the participants diagnosed have not been evaluated. These participants are not included in the number of participants evaluated.