Applications
Applications received: 4631
Applications under review: 333
Participants accepted: 1817

Evaluations
Participants evaluated: 1439
Participants diagnosed: 434
Participants with exome and/or genome sequencing complete: 1274
Participants with metabolomic analyses complete: 172
Participants with samples in the Central Biorepository: 964
Variants evaluated by the Model Organisms Screening Center: 309

Data Sharing
Manuscripts published: 106
ClinVar submissions: 462
Participants with data submitted to dbGaP: 854
Participants with data submitted to PhenomeCentral: 1387

Last updated: October 26, 2020
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 The number of applications received does not include applications that were withdrawn.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.