Applications
Applications received: 4713
Applications under review: 351
Participants accepted: 1845

Evaluations
Participants evaluated: 1457
Participants diagnosed: 434
Participants with exome and/or genome sequencing complete: 1299
Participants with metabolomic analyses complete: 177
Participants with samples in the Central Biorepository: 1062
Variants evaluated by the Model Organisms Screening Center: 316

Data Sharing
Manuscripts published: 115
ClinVar submissions: 468
Participants with data submitted to dbGaP: 854
Participants with data submitted to PhenomeCentral: 1414

Last updated: December 10, 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 The number of applications received does not include applications that were withdrawn.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.