Applications
Applications received: 5310
Applications under review: 294
Participants accepted: 2133

Evaluations
Participants evaluated: 1749
Participants diagnosed: 505
Participants with exome and/or genome sequencing complete: 1522
Participants with metabolomic analyses complete: 216
Participants with samples in the Central Biorepository: 1102
Variants evaluated by the Model Organisms Screening Center: 353

Data Sharing
Manuscripts published: 151
ClinVar submissions: 539
Participants with data available in dbGaP: 1319
Participants with data submitted to PhenomeCentral: 1585

Last updated: October 4, 2021
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 The number of applications received does not include applications that were withdrawn.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.