Applications
Applications received: 4791
Applications under review: 306
Participants accepted: 1894

Evaluations
Participants evaluated: 1482
Participants diagnosed: 445
Participants with exome and/or genome sequencing complete: 1349
Participants with metabolomic analyses complete: 179
Participants with samples in the Central Biorepository: 1062
Variants evaluated by the Model Organisms Screening Center: 325

Data Sharing
Manuscripts published: 117
ClinVar submissions: 470
Participants with data available in dbGaP: 854
Participants with data submitted to PhenomeCentral: 1429

Last updated: January 25, 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 The number of applications received does not include applications that were withdrawn.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.