Applications
Applications received: 4983
Applications under review: 306
Participants accepted: 1988

Evaluations
Participants evaluated: 1532
Participants diagnosed: 470
Participants with exome and/or genome sequencing complete: 1400
Participants with metabolomic analyses complete: 195
Participants with samples in the Central Biorepository: 1080
Variants evaluated by the Model Organisms Screening Center: 339

Data Sharing
Manuscripts published: 129
ClinVar submissions: 493
Participants with data available in dbGaP: 1216
Participants with data submitted to PhenomeCentral: 1461

Last updated: April 22, 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 The number of applications received does not include applications that were withdrawn.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.