Applications
Applications received: 5045
Applications under review: 319
Participants accepted: 2004

Evaluations
Participants evaluated: 1563
Participants diagnosed: 479
Participants with exome and/or genome sequencing complete: 1420
Participants with metabolomic analyses complete: 200
Participants with samples in the Central Biorepository: 1080
Variants evaluated by the Model Organisms Screening Center: 342

Data Sharing
Manuscripts published: 137
ClinVar submissions: 493
Participants with data available in dbGaP: 1216
Participants with data submitted to PhenomeCentral: 1485

Last updated: May 27, 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 The number of applications received does not include applications that were withdrawn.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.