Applications  
Applications received: 5124  
Applications under review: 321  
Participants accepted: 2035

Evaluations  
Participants evaluated: 1618  
Participants diagnosed: 484  
Participants with exome and/or genome sequencing complete: 1468  
Participants with metabolomic analyses complete: 204  
Participants with samples in the Central Biorepository: 1080  
Variants evaluated by the Model Organisms Screening Center: 345

Data Sharing  
Manuscripts published: 143  
ClinVar submissions: 527  
Participants with data available in dbGaP: 1216  
Participants with data submitted to PhenomeCentral: 1505

Last updated: July 12, 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 The number of applications received does not include applications that were withdrawn.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.