Applications
Applications received: 4688
Applications under review: 346
Participants accepted: 1774

Evaluations
Participants evaluated: 1417
Participants diagnosed: 429
Participants with exome and/or genome sequencing complete: 1266
Participants with metabolomic analyses complete: 165
Participants with samples in the Central Biorepository: 912
Variants evaluated by the Model Organisms Screening Center: 294

Data Sharing
Manuscripts published: 96
ClinVar submissions: 394
Participants with data submitted to dbGaP: 854
Participants with data submitted to PhenomeCentral: 1349

Last updated: August 25, 2020
Note: only proband data is reported here