Applications

Applications received: 4634
Applications under review: 371
Participants accepted: 1740

Evaluations

Participants evaluated: 1413
Participants diagnosed: 427
Participants with exome and/or genome sequencing complete: 1249
Participants with metabolomic analyses complete: 165
Participants with samples in the Central Biorepository: 885
Variants evaluated by the Model Organisms Screening Center: 291

Data Sharing

Manuscripts published: 94
ClinVar submissions: 388
Participants with data submitted to dbGaP: 854
Participants with data submitted to PhenomeCentral: 1337

Last updated: July 27, 2020
Note: only proband data is reported here