Applications

Applications received: 4369
Applications under review: 548
Participants accepted: 1603

Evaluations

Participants evaluated: 1358
Participants diagnosed: 383
Participants with exome and/or genome sequencing complete: 1195
Participants with metabolomic analyses complete: 157
Participants with samples in the Central Biorepository: 885
Variants evaluated by the Model Organisms Screening Center: 278

Data Sharing

Manuscripts published: 81
ClinVar submissions: 356
Participants with data submitted to dbGaP: 450
Participants with data submitted to PhenomeCentral: 1208

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