

Applications

Applications received: 4791

Applications under review: 306

Participants accepted: 1894

Evaluations

Participants evaluated: 1482

Participants diagnosed: 445

Participants with exome and/or genome sequencing complete: 1349

Participants with metabolomic analyses complete: 179

Participants with samples in the Central Biorepository: 1062

Variants evaluated by the Model Organisms Screening Center: 325

Data Sharing

[Manuscripts](#) published: 117

[ClinVar](#) submissions: 470

Participants with data available in [dbGaP](#): 854

Participants with data submitted to [PhenomeCentral](#): 1429

Last updated: January 25, 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 The number of applications received does not include applications that were withdrawn.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.