

Clinical and Genetic Evaluation of Individuals With Undiagnosed Disorders Through the Undiagnosed Diseases Network

NCT02450851

Status	RECRUITING
Sponsor	National Human Genome Research Institute (NHGRI)
Enrollment	20,000 participants

Key Eligibility Criteria

Inclusion (5)

- Ideal participants for tier 2-4 evaluations include individuals with:
- One or more objective findings pertinent to the phenotype for which a case was submitted.
- No diagnosis despite evaluation by specialists who assessed the patient for the objective finding(s).
- Agreement for the storage and sharing of information and biomaterials, in an identified fashion amongst the UDN centers, and in a de-identified fashion to research sites beyond the network.
- Participants unable to consent can be enrolled.

Exclusion (5)

- Individuals who are unlikely to be assigned to tier 2-4 evaluations include those with:
- Reported symptoms with no relevant objective findings.
- A diagnosis explaining objective findings.
- A diagnosis suggested on record review.
- Unwillingness to share data.

Locations (33 total)

University of Alabama at Birmingham, Birmingham, Alabama, United States
HudsonAlpha Institute for Biotechnology, Inc., Huntsville, Alabama, United States
University of California, Los Angeles, Los Angeles, California, United States
... and 30 more locations