

# Genetic Analysis of Uncommon Disease Presentations in Non-US Populations

NCT06595940

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<b>Status</b>	RECRUITING
<b>Sponsor</b>	National Human Genome Research Institute (NHGRI)
<b>Enrollment</b>	400 participants

## Key Eligibility Criteria

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### Inclusion (6)

- To be eligible to participate in this study, an individual must meet all of the following criteria:
- Stated willingness to comply with all study procedures and availability for the duration of the study.
- Probands aged  $\geq 2$  years at enrollment or first-degree relatives of probands (age  $\geq 2$  years).
- Suspicion of genetic etiology of illness due to strong family history, precocious onset, severity or mildness of phenotype, or all factors being present.
- Affected individuals and unaffected family members, determination of clinical criteria for inclusion will be determined by medical record review prior to participation.
- ... and 1 more (see full listing online)

### Exclusion (5)

- An individual who meets any of the following criteria will be excluded from participation in this study:
- Anyone unwilling to provide informed consent (for themselves as adults, on behalf of their children as minors, or on behalf of an adult who is unable to provide consent for themselves) or assent.
- Individuals who have undergone diagnostic testing for a genetic condition AND the test results were positive.
- Evidence that symptoms are secondary or caused by an undiagnosed condition that is unlikely to have a genetic cause.
- In the opinion of the investigator, participant has a condition that would preclude participation in the study by interfering with the participant's ability to engage in the required protocol evaluation and testing.

## Locations (1 total)

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University of Mauritius, Moka, Mauritius