

The Natural History Study of Mitochondrial NeuroGastroIntestinal Encephalopathy (MNGIE)

NCT01694953

Status RECRUITING
Sponsor Columbia University
Enrollment 20 participants

Key Eligibility Criteria

Inclusion (7)

- Thymidine Phosphorylase (TP) defect:
- homozygous or
- compound heterozygous mutations in the TYMP gene, and/or
- TP enzyme activity of $\leq 20\%$ of normal.
- Increased plasma Thd > 3 micromole/L
- ... and 2 more (see full listing online)

Exclusion (4)

- Participation in an interventional (study medication or other experimental intervention) study (within 1 month of participation in this study).
- Unable to travel to site for research visits.
- Unwillingness to sign informed consent form.
- Substance abuser

Locations (1 total)

Columbia University, New York, New York, United States