COL4A1COL4A2: Study of Pathological Conditions Involving Multiple Organs Caused by Mutations in the COL4A1 and COL4A2 Genes

NCT07374913

Status	RECRUITING
Phase	Not Applicable
Sponsor	Meyer Children's Hospital IRCCS
Enrollment	120 participants

Key Eligibility Criteria

Inclusion (5)

- Individuals (pediatric or adult) with a pathogenic or likely pathogenic mutation in the COL4A1 or COL4A2 genes and a clinical phenotype consistent with small vessel disease.
- Adult first-degree family members (parents, siblings, or children) who are confirmed carriers or suspected carriers of the same COL4A1/COL4A2 mutation.
- Adult first-degree family members who are non-carriers of the pathogenic mutation and who agree to provide a blood sample to be used as controls for laboratory analyses.
- Ability to provide written informed consent; for minors, consent provided by a parent or legal guardian.
- Any condition that, in the opinion of the investigators, would preclude participation in study procedures or reliable data collection.

Exclusion (1)

- Refusal or inability to provide informed consent.

Locations (1 total)

Meyer Children's Hospital IRCSS, Florence, FI, Italy