

Developing a Pipeline to Employ RNA-Seq as a Complementary Diagnostic Tool in Rare Diseases

NCT05996731

Status	RECRUITING
Phase	Not Applicable
Sponsor	Mario Negri Institute for Pharmacological Research
Enrollment	105 participants

Key Eligibility Criteria

Inclusion (9)

- Healthy subjects.
- Male and female adults
- Written informed consent
- Male and female adults
- Genetic diseases affecting RNA levels (frameshifts, stop, large deletions, alteration of canonical splicing sites)
- ... and 4 more (see full listing online)

Exclusion (9)

- Inability to understand the potential risk and benefits of the study
- Legal incapacity
- Validation cohort.
- Underage patients
- Inability to understand the potential risk and benefits of the study
- ... and 4 more (see full listing online)

Locations (1 total)

Centro di Ricerche Cliniche per le Malattie Rare "Aldo e Cele Daccò", Ranica, BG, Italy