

Transcriptomic Analysis to Put an End to Misdiagnosis in Patients With Rare Muscle Diseases

NCT06833489

Status	RECRUITING
Phase	Not Applicable
Sponsor	Assistance Publique Hopitaux De Marseille
Enrollment	50 participants

Key Eligibility Criteria

Inclusion (4)

- patients with rare genetic muscle diseases who have benefited from high-throughput sequencing analysis (panel of 200 genes defined by the FILNEMUS Rare Neuromuscular Disease Network) carried out at the Molecular Genetics Laboratory, Medical Genetics Department, Timone Enfant Hospital since 2017.
- This criterion is necessary to limit the analysis to patients with muscular diseases among all the patients analysed by the Molecular Genetics Laboratory.
- this genetic analysis did not identify pathogenic variants explaining the patient's phenotype This criterion is necessary in order to include only patients in diagnostic error.
- a muscle biopsy of the patient is available in the Biological Resources Centre (CRB) at the AP-HM.

Exclusion (3)

- Patients with no muscle biopsy available in the CRB.
- Patients with an established molecular diagnosis.
- Patients for whom RNA extraction from a muscle biopsy sample did not yield RNA of sufficient quality (INR ≤ 7) will be excluded from the study. A maximum of two extraction attempts will be performed.

Locations (1 total)

Hopital Timone, Marseille, France