

# Modifying Factors in Striated Muscle Laminopathies

NCT05394506

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| <b>Status</b>     | RECRUITING  |
| <b>Phase</b>      | Not Applicable  |
| <b>Sponsor</b>    | Institut National de la Santé Et de la Recherche Médicale, France |
| <b>Enrollment</b> | 40 participants   |

## Key Eligibility Criteria

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### Inclusion (5)

- Patient with an LMNA mutation that has led to the diagnosis of laminopathy affecting striated muscle
- Presenting the symptoms of the disease, whether they are index cases or related to this index case (muscle weakness, tendon retractions with or without respiratory or cardiac involvement)
- Have no contraindication to muscle or skin biopsy, i.e., 1) presence of a history of allergy to latex, antiseptics, local anesthetics and adhesive dressings, 2) Current oral or parenteral anticoagulant therapy (anti-vitamin K, heparins, anti-platelet agents, anti-factor X, anti-thrombin), 3) History of inherited (haemophilias, platelet diseases) or acquired (vitamin K deficiency, liver failure) coagulation disorders.
- Patients (adult participant) or both holders of parental authority (minor participant) must sign a free and informed consent. If a minor has only 1 legal representative, the latter must attest to this on the consent form.
- Patients affiliated to the general French social security system, to the French Universal Medical Coverage (CMU) or to any French equivalent scheme.

### Exclusion (2)

- Pregnant or breastfeeding women
- Adult subject to legal protection measures (safeguard of justice, curatorship and guardianship).

## Locations (8 total)

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Centre de référence maladies neuromusculaires, Hôpital Femme Mère Enfant, CHU Lyon, Bron, Auvergne-Rhône-Alpes, France  
Centre de référence maladies neuromusculaires, Institut de myologie, Hôpital Pitié-Salpêtrière, Paris, France, France  
Service de Neuropédiatrie, Centre de Référence Maladies Neuromusculaires, CHU de Montpellier, Montpellier, Hérault, France  
... and 5 more locations