The French National Registry of Calpainopathies (CALPA)

Romain Glandier[1], France Leturcq[2], Martin Krahn[3], Guilhem Solé[4], Tanya Stojkovic[5], Caroline Espil[6], Filnemus network[7], Mélanie Bordes[1], Juliette Peterka[1], CRAs Group[1], Caroline Randazzo[1], Julie Lejeune[1], Isabelle Richard[8], Edoardo Malfatti[9]

1.AFM-Téléthon, Évry (France), 2.AP-HP, Hôpital Cochin, Service de Génétique et Biologie Moléculaires, Paris (France), 3.AP-HM, Hôpital de la Timone, Laboratoire de Génétique Moléculaire, Marseille (France), 4.CHU de Bordeaux, CRMR Maladies Neuromusculaires AOC, Bordeaux (France), 5.AP-HP, Hôpital Pitié-Salpêtrière, Institut de Myologie, CRMR Nord-Est-Île-de-France, Paris (France), 6.CHU de Bordeaux, Service de Neuropédiatrie / CRMR AOC, Bordeaux (France), 7.Centres de référence du réseau national neuromusculaire FILNEMUS (France), 8.Généthon, Évry (France), 9.AP-HP, Hôpital Henri-Mondor, IMRB / Université Paris-Est Créteil (UPEC), Créteil (France)

Background

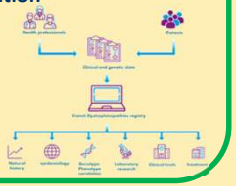
Calpainopathies are inherited myopathies associated with pathogenic variants in the CAPN3 gene, most frequently autosomal recessive (LGMDR1), the most common form of limb-girdle muscular dystrophy. The disease presents as a slowly progressive proximal myopathy beginning in childhood or adolescence, with loss of ambulation typically occurring after approximately fifteen years of disease evolution.

Objectives of the registry

The CALPA registry aims to centralize harmonized national clinical, biological and genetic data on calpainopathies. It supports documentation of natural history, facilitates clinical research, and contributes to clinical trials preparation by enabling standardized and early data collection.

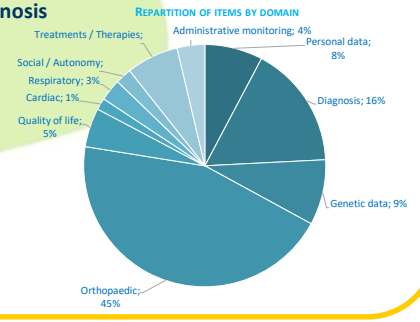
Registry Organization

- National registry supported by AFM-Téléthon and launched in December 2025
- Patients followed within neuromuscular expert centers of the FILNEMUS network
- Standardized data collection: sociodemographic, clinical and genetic information
- Quality monitoring performed by a Clinical Research Associate (CRA) Pool
- Registry compliant with GDPR and French data protection authority
- AFM-Téléthon is the registry data controller



Results (as of May 2026)

- 38 patients screened, informed as part of the consent pathway
- 3 patients included, all with a confirmed CAPN3-R1 diagnosis
- More than 350 standardized data items collected
- Data collected:
  - Clinical, genetic and sociodemographic domains
  - Motor, respiratory and cardiac assessments
  - Treatments



Uses and Perspectives

The CALPA registry supports harmonized documentation of disease natural history, strengthens collaboration across neuromuscular centers and improves data accessibility for research teams. It facilitates patient identification and represents a key instrument for the preparation of future Clinical Trials.

Conclusion

The CALPA registry is a key national tool for advancing knowledge on calpainopathies, supporting clinical research and improving patient management in LGMDR1.

Acknowledgements / Contact Information
- Registry CALPA
- Filnemus Network
- rglandier@afm-telethon.fr