The French National Registry of Sarcoglycanopathies (SARCO)

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Background

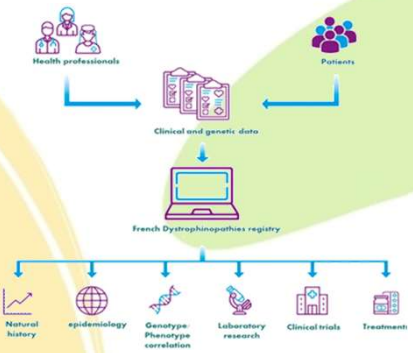
Sarcoglycanopathies are autosomal recessive limb-girdle muscular dystrophies caused by defects in α-, β-, γ- or δ-sarcoglycan genes. These ultrarare conditions are characterized by symmetric proximal muscle weakness with variable cardiac and respiratory involvement. Despite ongoing gene therapy trials, their natural history remains insufficiently documented.

Objectives of the registry

The SARCO registry aims to centralize harmonized national data on sarcoglycanopathies. It supports documentation of disease natural history, facilitates clinical research, and contributes to the preparation of clinical trials through standardized data collection across expert centers.

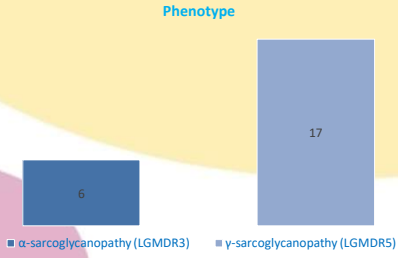
Registry Organization

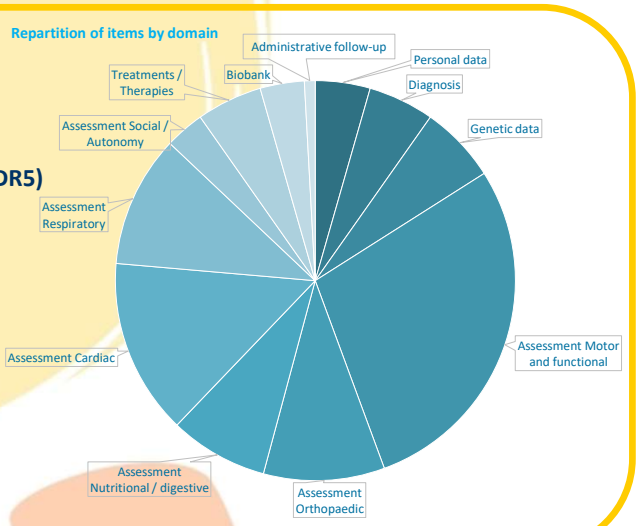
- National registry created in May 2025 with the support of AFM-Téléthon
- Patients followed in neuromuscular expert centers of the FILNEMUS network
- Standardized collection of clinical, biological and genetic data
- Quality monitoring performed by Clinical Research Associate Pool
- Registry compliant with GDPR and French data protection authority (CNIL)
- AFM-Téléthon is the data controller of the registry



Results (as of May 2026)

- 37 patients screened, informed as part of the consent process,
- 23 patients included with a confirmed phenotype
- 6 α-sarcoglycanopathy (LGMDR3) & 17γ-sarcoglycanopathy (LGMDR5)
- More than 500 standardized data items collected across clinical, genetic and sociodemographic domains





Uses and Perspectives

The SARCO registry strengthens documentation of natural history, improves data accessibility for research teams and enhances collaboration between neuromuscular centers. It facilitates patient identification and supports the preparation of future clinical trials, contributing to improved long-term care.

Conclusion

The SARCO registry provides a unified national resource for advancing knowledge on sarcoglycanopathies and for supporting future therapeutic research.

Acknowledgements / Contact Information
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