

STUDY OVERVIEW: Co-existing Facioscapulohumeral Muscular Dystrophy (FSHD) and Myasthenia Gravis (MG): International Case Series

NMJ Working Group, ERN EURO-NMD

Background

Our recently published UK multicentre study (McMacken et al., JNNP 2026) identified a fivefold higher than expected co-occurrence of AChR-positive myasthenia gravis (MG) in patients with genetically confirmed FSHD, suggesting a non-random biological association. MG may be difficult to recognise in patients with pre-existing FSHD due to overlapping phenotypic features, and timely diagnosis is critical as MG is treatable. This international case series aims to characterise the clinical phenotype of this rare combination in a larger international cohort.

Study Design

Retrospective international observational case series. No direct patient contact is required from participating clinicians beyond completion of the data collection form using existing clinical records.

Eligibility Criteria

- Confirmed diagnosis of FSHD (genetic confirmation or clinical diagnosis with supportive features).
- Co-existing confirmed diagnosis of MG (clinical, serological and/or neurophysiological criteria).

What Participation Involves

- Complete a standardised data collection form for each eligible patient using existing clinical records.
- Pseudonymise all data before submission by assigning each patient a unique coded identifier.
- Sign the Participation Schedule of the Collaboration Agreement confirming local compliance.

Estimated time: approximately 15 to 20 minutes per patient. No additional patient investigations, clinic visits or biological samples are required.

Governance and Consent

This study is an official study of the Neuromuscular Junction Working Group of ERN EURO-NMD. It has received ethical approval from the Research Ethics Committee at Queen's University Belfast. Participating clinicians are responsible for ensuring compliance with consent requirements at their institution before submitting data. The Participation Schedule of the Collaboration Agreement is the mechanism by which local compliance is confirmed.

How to Participate

If you have one or more eligible patients and wish to contribute, please contact:

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