

Neuromuscular diagnoses in a nutshell

a new way of visualizing clinical summaries for rare neuromuscular diseases

Geir Hevnskjel Ringvold, Andreas Dybesland Rosenberger, Tine Stokmo, Kjell Arne Arntzen

Norwegian Centre for Rare Diseases, Unit for Neuromuscular Disorders, University Hospital of North Norway, Tromsø, Norway

Background

Rare inherited neuromuscular disorders (NMDs) are heterogeneous and often complex conditions that are challenging to understand for patients, relatives and non-specialized healthcare professionals (HCPs). Available information is often extensive, text-heavy and scattered over several sources. We saw the need for concise, accessible and visually intuitive summaries that support communication, education and clinical decision-making.

Approach

We developed a standardized visual format presenting key information for rare neuromuscular disorders ("in a nutshell"). Each summary is based on a structured template including classification codes, muscle involvement, clinical features, symptom onset, genetics and inheritance, prevalence, available treatment options and critical considerations. Information is presented in a compact, single-page visual layout combining anatomical illustrations, icon-based elements and short text segments, allowing rapid orientation while retaining clinical relevance.

Content is derived from established sources such as Orphanet, GeneReviews, national guidelines and expert consensus.

Results

The project has resulted in a series of visual "flash card"-summaries for selected diagnoses (e.g. Limb-girdle muscular dystrophy (LGMD)(Fig.1), Charcot-Marie-Tooth disease (CMT), Myotonic dystrophy (DM) and Spinal muscular atrophy (SMA)), and is made to be applicable to all NMDs.

The format enables quick comparison of disease-specific patterns and key clinical features.

The illustrations are designed to work well on both desktop and mobile devices, as well as when printed on A4 paper in black and white. In the digital PDF version, relevant sections of the illustration contain embedded links to further information.

Preliminary feedback from clinicians and patient representatives indicates that the summaries are intuitive and educational.

Visual clinical summaries represent a useful tool for simplifying complex medical information in rare neuromuscular disorders. The format has potential applications in clinical practice, patient education and teaching.

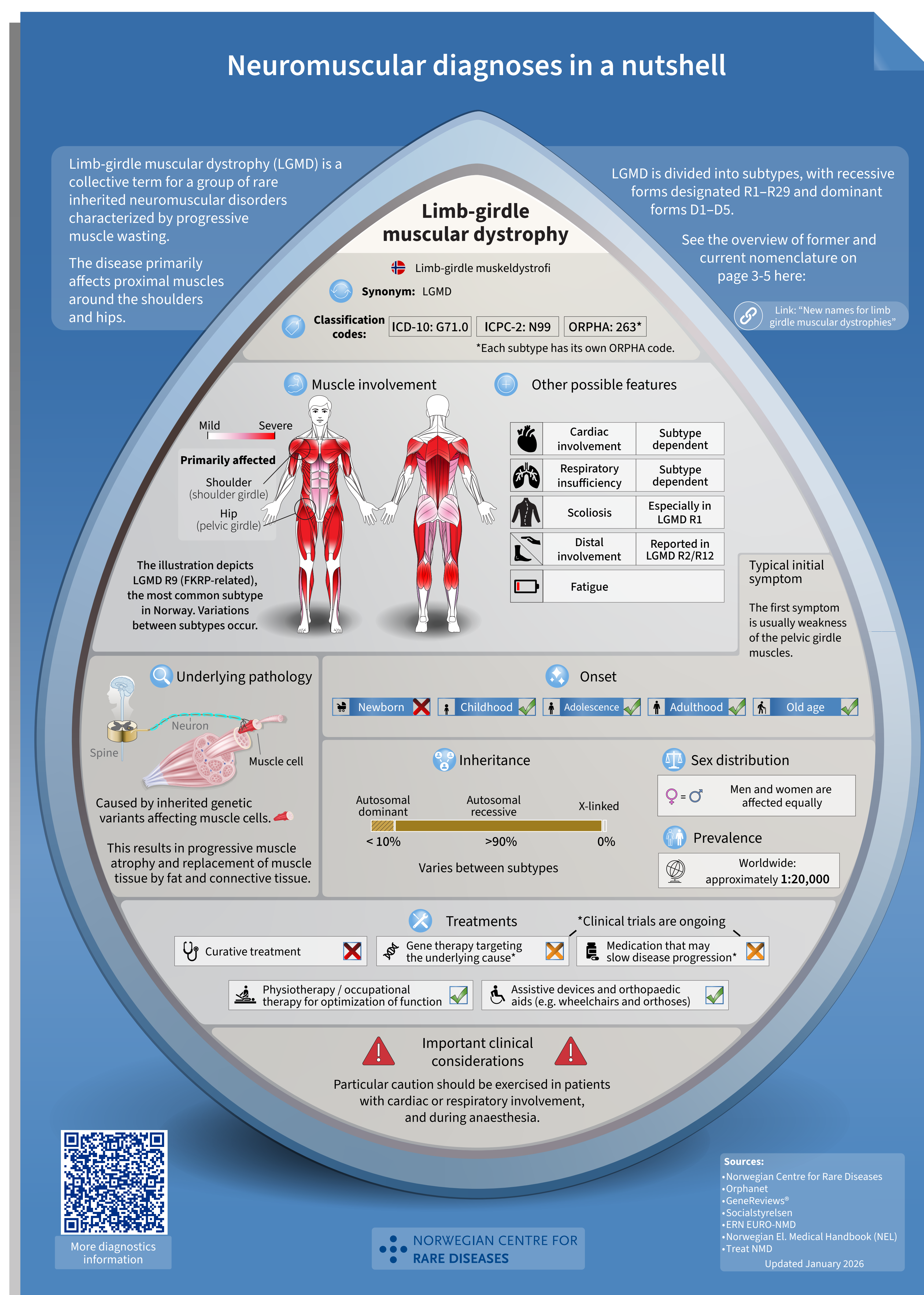
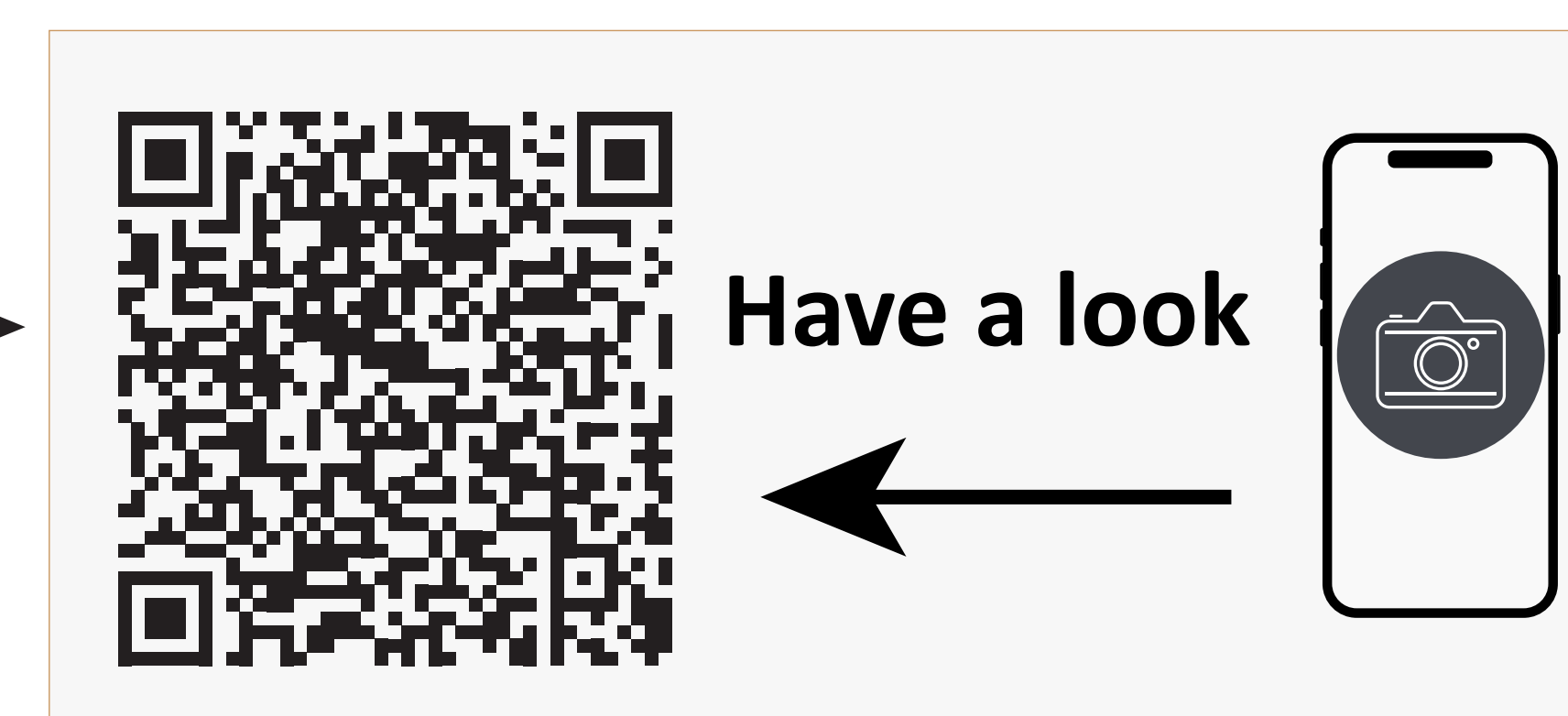


Figure 1. Example of a visual clinical summary for Limb-girdle muscular dystrophy (LGMD).



Further work will include expansion to additional diagnoses and user testing in patients and HCPs. Structured evaluation will assess the impact on general understanding and awareness of NMDs with non-specialized HCPs, such as GPs, local neurologists and medical students.