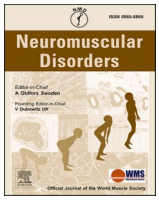
Neuromuscular Disorders 60 (2026) 106304

Contents lists available at ScienceDirect

Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd



286th ENMC international workshop: Muscle imaging: artificial intelligence, automatic segmentation and imaging data sharing in neuromuscular disease. Hoofddorp, The Netherlands, 7-9 March 2025

Jodi Warman-Chardon [a,b,*], Volker Straub [c], John Vissing [d,e], Sarah Schlaeger [f], Hermien E. Kan [g], MRI workshop study group

[a] Department of Medicine (Neurology), The Ottawa Hospital/Research Institute; University of Ottawa, Canada
[b] Department of Genetics, Children's Hospital of Eastern Ontario/Research Institute, Canada
[c] John Walton Muscular Dystrophy Research Center, Newcastle University Translational and Clinical Research Institute. Newcastle University, UK
[d] Copenhagen Neuromuscular Center, Copenhagen University Hospital, Rigshospitalet, Denmark
[e] University of Copenhagen, Denmark
[f] Department of Radiology, LMU University Hospital, LMU Munich, Munich, Germany
[g] Department of Radiology, C.J. Gorter MRI Center, Leiden University Medical Center, Leiden, the Netherlands

ARTICLE INFO

Keywords
Neuromuscular disease
Quantitative magnetic resonance imaging
Automatic segmentation
Machine learning

ABSTRACT

Quantitative muscle MRI (qMRI) has emerged as a promising non-invasive biomarker for assessing neuromuscular diseases (NMDs). However, clinical implementation is limited by the significant time required for manual muscle segmentation, which restricts analysis to limited muscle regions rather than comprehensive whole-muscle assessment. The 286th European NeuroMuscular Centre (ENMC) workshop brought together 18 international participants from 10 countries to establish consensus on optimal qMRI acquisition protocols and automated analysis tools, revealing that while most centers utilize qMRI techniques, barriers to manual segmentation include limited expertise and excessive time requirements. Automated segmentation methods using machine learning architectures, particularly 3D U-Net models, have demonstrated promising results for individual muscle segmentation. Multi-center studies are starting to implement standardized protocols, while machine learning approaches can distinguish among many NMDs with higher accuracy than human experts. Data sharing platforms and federated learning approaches address the need for larger NMD cohorts with standardized and vendor-agnostic data formats, while maintaining patient privacy. The integration of automated 3D muscle segmentation tools integrated into clinical workflows represents a transformative advancement to revolutionize diagnosis, disease monitoring, and therapeutic assessment in NMDs. This consensus workshop provides a roadmap for accelerating the translation of qMRI from research tools to clinically implemented biomarkers for NMD management.

1. Introduction and background

Multiple novel therapies are in development for neuromuscular diseases (NMDs). However, assessing the efficacy of these promising treatments is limited by the lack of sensitive, standardized and reproducible methods to assess subtle disease progression or therapeutic response [1,2]. Magnetic resonance imaging (MRI), and particularly quantitative muscle MRI (qMRI), has emerged as a non-invasive imaging biomarker to distinguish important structural changes like fat replacement, muscle volume and edema in NMDs [3–8]. Qualitative and semi-quantitative MRI studies have demonstrated characteristic imaging patterns using visual and signal-density assessment of fat replacement to provide a diagnostic pattern for several NMDs [8–11]. Although qualitative imaging analysis permits subjective categorising of disease features, such as the extent of signal intensity on T_1 weighted and T_2 weighted MRI protocols, qMRI techniques are more sensitive in detecting subtle changes and offer a more objective assessment [5,12,13]. QMRI techniques, including chemical-shift-based-fat-water (Dixon) imaging techniques, transversal relaxation time (T2) mapping (either water T2 or global T2), and diffusion tensor imaging, are more sensitive

* Corresponding author.
E-mail address: jwarman@toh.ca (J. Warman-Chardon).

https://doi.org/10.1016/j.nmd.2025.106304
Received 1 December 2025; Accepted 7 December 2025
Available online 8 December 2025

to the variable pathological changes in muscle tissue, compared to traditional semi-quantitative visual assessment of muscle fat/edema [13–21]. Beyond diagnostic applications, qMRI approaches are emerging as sensitive and reproducible biomarkers, and provide evidence of disease progression, which is critical for use in clinical trials [22,23]. Yet, the challenges of the scarcity of annotated qMRI scans, the variable NMD progression in each muscle and the high number and rarity of different NMDs, all provide barriers to using qMRI to identify diagnostic patterns and to assess disease progression or therapeutic response in research settings. The current necessity of time-consuming manual segmentation and the inaccuracy of automated segmentation algorithms hinder clinical implementation.

For qMRI, muscle segmentation is required to identify muscle regions of interest (e.g., to extract quantitative parameters of muscles) and to distinguish them from subcutaneous and perimuscular adipose tissues and bone. Manual muscle segmentation is operator dependent and extremely time-consuming, which has limited the clinical implementation of qMRI and evaluation of these biomarkers in large cohorts [1]. Consequently, qMRI analyses are typically performed on part of a limb only using a limited number of slices in the center of the limb [5,12,24,25]. This also limits the ability to fully assess the entire length of the involved muscle, which may be differentially affected in regions of the proximal-distal axis [26–28]. A full whole-muscle fat assessment would therefore provide a better way of identifying disease progression. There is a critical need for the integration of reliable automatic 3D segmentation methods over the whole limb and throughout the length of the muscle to improve diagnosis and disease progression for clinical trials.

More recently, machine learning strategies have been able to distinguish >10 NMDs based on MRI scans, depicting fat replacement with higher accuracy than human experts in the field. This provided a critical proof of concept demonstrating that artificial intelligence (AI) can be applied to the field of muscle MRI in NMD [29,30]. However, given the burden of time and expertise required for scoring images across a large range of hundreds of NMDs, automatic segmentation methods and feature extraction are required [31]. Several AI-enabled automated segmentation methods aim to segment individual muscles (or muscle groups), using data from numerous acquired MRI slices to permit more complex analyses, to reduce processing times and to eliminate interobserver bias [1,31,32]. However, automation of muscle segmentation in MRI is very challenging in NMD where muscle borders are obscured by severe fat replacement, given the poor contrast between different muscles and the large variability of muscle shapes [1,32].

The 286th ENMC international workshop was held in Hoofddorp, The Netherlands, from March 7th-9th, 2025 to discuss barriers and strategic opportunities for implementation of qMRI techniques and assessment tools. The workshop assembled 18 participants from 10 countries, including France, Italy, The Netherlands, Denmark, Belgium, Switzerland, Germany, Canada, United States of America (USA) and United Kingdom (UK), comprising clinicians and researchers from NMDs, MRI and machine learning disciplines. The aims for this workshop were to establish international consensus for optimal muscle qMRI acquisition protocols, data storage and post-processing and analysis tools to increase clinical trial readiness internationally; review manual and automated imaging segmentation methods and discuss their reliability, reproducibility, and limitations in the context of NMD; review machine learning diagnostic approaches in MRI assessment and international imaging sharing platforms to support building larger cohorts for machine learning while ensuring imaging security, patient privacy in qMRI.

2. Preworkshop questionnaire: Evaluating quantitative use of muscle MRI, muscle segmentation and data sharing

A preworkshop questionnaire was completed by attendees to assess current implementation of qMRI, muscle segmentation and data sharing (Fig. 1). Manual segmentation was widely used for natural history and therapeutic studies, but faced barriers to implementation, such as limitations in anatomical knowledge of the vast number of muscles across

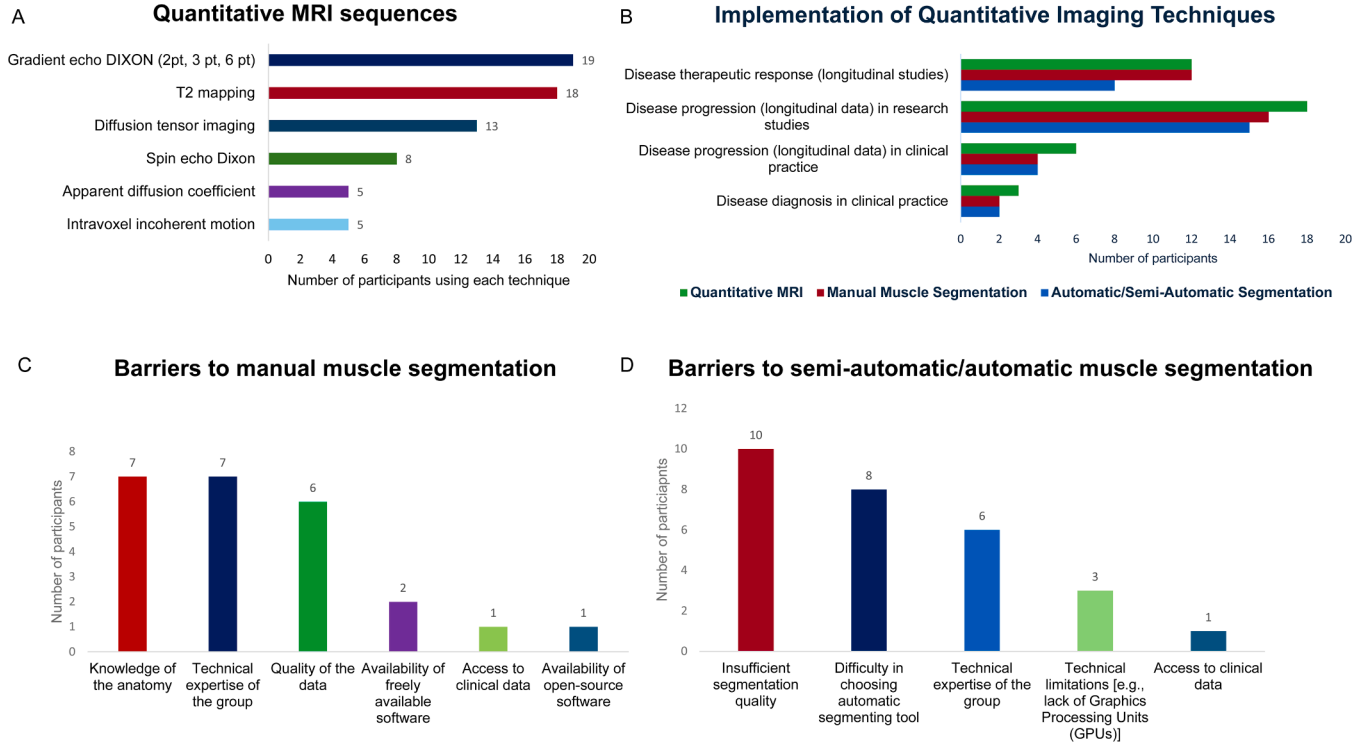


Fig. 1. Pre-workshop survey responses regarding muscle MRI utilization within participants' respective academic institutions. (A) Currently implemented muscle qMRI sequences; (B) Anticipated applications for expanded qMRI implementation; (C) Identified barriers limiting manual muscle segmentation adoption; (D) Obstacles to implementation of semi-automatic and automatic segmentation methodologies.

their length, limited technical expertise, poor image quality, and time demands required for segmenting each muscle at different regions. Automatic or semi-automatic segmentation was employed by many centres, though challenges included insufficient tool quality, tool selection difficulties, and technical limitations, with most analyses focusing on the lower extremities. Automatic/semi-automatic segmentation was being used to assess thighs (17/19), lower legs (16/19), upper extremities (9/19), trunk and pelvis (2/19) and no participant assessed head muscles, skull or brain, reflecting the use of a longstanding focus of MRI for lower extremities. However, imaging solely the lower extremities will miss some critical muscles for pattern recognition of specific disorders, such as tongue involvement in oculopharyngeal muscular dystrophy [21,33], and trapezius involvement with subscapularis sparing in facioscapulohumeral muscular dystrophy (FSHD) [34]. In addition, it is important to assess both sides individually given potential asymmetry of fat replacement or edema between limbs [28,35,36]. Data sharing relied on tools like MYO-Share and XNAT but was hindered by strict privacy regulations, incomplete consent, and technical constraints. For this survey, it is important to note that survey respondents comprised ENMC workshop attendees who were specifically invited based on their expertise in muscle segmentation and therefore may not be representative of the broader muscle MRI research community.

3. Muscle imaging: Artificial intelligence, automatic segmentation and imaging data sharing in neuromuscular disease workshop sessions

Following introductory remarks by Patricia van Dongen, Programme Manager of ENMC, Volker Straub, (UK), opened the 286th ENMC workshop by outlining the importance of assembling the required expertise to establish international consensus for optimal muscle qMRI acquisition protocols, data storage, and post-processing and analysis tools to increase clinical trial readiness internationally. The workshop goals were highlighted, including optimizing and standardizing qMRI assessments, comparing segmentation methods, exploring machine learning applications, reviewing international imaging platforms, and addressing imaging security and patient privacy in NMD.

3.1. Current state of integrating qualitative MRI muscle segmentation: implications for clinical practice

Pierre Carlier (France) provided an overview of the standard muscle qMRI techniques. Under the hypothesis that a “standard” quantitative skeletal muscle imaging protocol exists, it can comprise Dixon imaging sequences, the multi-TE spin echo sequence (MESE) and the diffusion tensor imaging sequences (DTI), by decreasing order of popularity and use. With the Dixon sequences, muscle trophism and fat replacement can be determined. With the MESE sequence, water T2 maps are generated, which evaluate the disease activity while DTI sequences provide information on myocyte orientation, dimensions and permeability. Whole-body qMRI is possible with modern scanners even in a clinical environment, thanks to a variety of acceleration techniques combined with AI denoising. For instance, whole body 3D Dixon with isotropic millimetric resolution is currently obtained in 4 to 10 minutes at 1.5T.

Hermien Kan (The Netherlands) presented an overview of manual segmentation, including the technical factors influencing segmentation, the gold standard and quality metrics often used. Manual segmentation is used to delineate individual skeletal muscles, usually on transverse MR images. Segmentation aims to provide muscle or region specific qMRI values, which can, for instance, be volume, fat fraction (FF) or water T2, per slice and per muscle. This can be used to aid in the differential diagnosis of NMD, as these have different patterns of muscle involvement, and to assess disease progression over time. For any quantitative parameter to be used for these purposes, is the value obtained reproducible and repeatable. Quality metrics that are commonly used are the Dice-Sorensen coefficient (DSC), or the intra-class correlation coefficient (ICC). Especially if the purpose of segmentation is to provide biomarkers to assess disease progression or response to therapy, an important quality metric is the sensitivity to change and the relation to a functional outcome. The former can be assessed using the standardized response mean (SRM), and the latter by association to function – either cross-sectional or longitudinal. Dr. Kan explained that there are many factors influencing segmentation, which can be divided into parameters at the acquisition level and in post-processing. At the scan acquisition level, it is necessary that the field-of-view covers a large enough proportion of the muscle, preferably the whole muscle, as many NMDs have proximo-distal differences in muscle involvement within the muscles [27,37–39]. Also, scan resolution needs to be sufficiently high to be able to delineate muscle borders. In post-processing, it is important that there is either landmark with fixed distance to a bone, or the insertion of a muscle, to make sure that assessments are done at the same level of the muscle between subjects, and over time. Finally, there are several practical considerations when drawing the regions of interest (ROIs) when segmenting muscle. This includes which type of images are used for drawing ROIs, whether muscles are delineated at the muscle border, whether to include all acquired slices in the analysis, whether single muscles are reported or muscle groups, and whether a single reader should draw all muscles within a study.

Harmen Reyngoudt (France) described the challenges for incorporating manual muscle segmentation in multi-site analysis in the International Clinical Outcome Study for Dysferlin (COS experience). QMRI including Dixon-type sequences and water T_2 mapping by MESE/Multi-Slice Multi-Echo (MSME) was part of the natural history Clinical Outcome Studies (COS) in dysferlinopathy funded by the Jain Foundation, in 14 different centers across the world (COS1 ran between 2012 and 2018) [24,40,41]. Successful manual muscle segmentation requires high-quality and reproducible MRI data across multiple visits (acquisition made with same central slice, volume, field-of-view, in-plane resolution). Since there were 14 different centers acquiring qMRI data and two different centers analyzing these qMRI data in COS, the analysis plan was documented in detail in several standard operating procedures (SOPs). To ensure validity between different centers, an essential step was that both analysis sites segmented on the same slices, so that all qMRI-based outcome measures were from exactly the same anatomical location. ROIs were drawn twice but differently on the first-TE MESE/MSME images on 5 slices in thigh and lower leg (Fig. 2), by analysis team 1 delineating nicely the muscle contours for FF values but especially for precise assessment of contractile cross-sectional area (cCSA), and by analysis team 2 drawn inside the ROIs to avoid inter-muscular/subcutaneous fat and fasciae, for water T_2 [24]. In both segmentations, visible blood vessels and tendons were avoided, and ROIs were eroded when including subcutaneous fat. A third rater verified coherence between both segmentations visually inspecting the ROIs drawn on the FF maps and the water T_2 maps paying attention to (i) similarity of ROIs drawn by both teams and (ii) major errors made in segmentation of the smaller or more difficult muscles to draw, before all qMRI results were merged into a single file with a FF, cCSA and water T_2 value per muscle per visit per subject. For COS2 (which ran between 2019 and 2023), the upper limb (arm and forearm) was also added to the qMRI protocol, with identical instructions for data analysis [42].

Francesco Santini (Switzerland) reviewed the imaging data formats and established ‘best practice’ standards in NMD imaging. Data sharing is key for modern research, especially in the current era of pervasive AI [43]. This is especially crucial in the field of NMD research, where the rarity of many NMD necessitates data collection from multiple centers. However, a high level of standardization is required for the efficient development of postprocessing and analysis tools, both in terms of acquisition modalities and in terms of data formats. The format supported by most medical imaging platforms is the “Digital Imaging and Communications in Medicine” (DICOM) standard, which is a detailed and flexible description of image data and metadata. However, this inherent generality and flexibility made it less than ideal as a proper

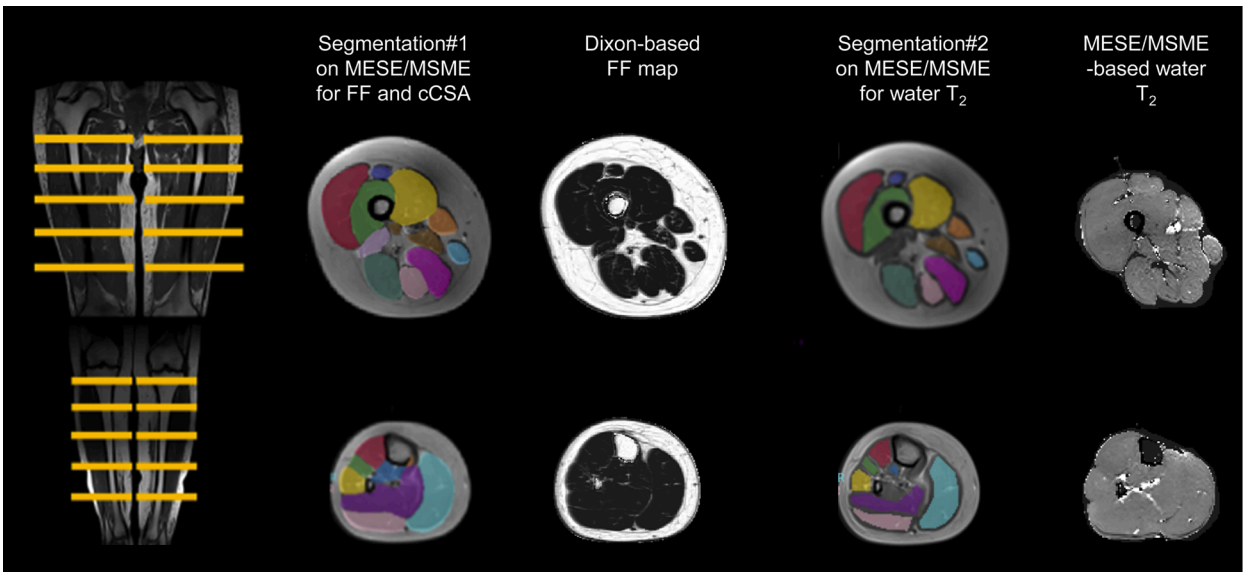


Fig. 2. qMRI in Jain COS in dysferlinopathy included two independent manual muscle segmentations performed on 5 slices of MESE/MSME data (first TE). Examples in one slice of thigh and lower leg are shown. Segmentations were slightly different depending on the qMRI-based outcome measure (FF/cCSA vs. water T_2). The corresponding Dixon-based FF maps and MESE/MSME-based water T_2 maps (with the subcutaneous fat masked) are also shown.

standard, as each device manufacturer practically implements its own proprietary "flavor" within the general framework of the DICOM standard, thus making the localization of relevant metadata for a specific purpose difficult in a reliable way [44]. For this reason, more specialized standards have been developed, starting with the Brain Imaging Data Structure (BIDS), a standardized way of organizing Neuroimaging Informatics Technology Initiative (NIfTI) imaging files in a folder structure and pairing it with metadata in Javascript Object Notation (JSON) format [45], subsequently extended to other districts and modalities, where it is sometimes referred to as MIDS (Medical Imaging Data Structure) [46]. For musculoskeletal imaging, the Open and Reproducible Musculoskeletal (MSK) Imaging Research (ORMIR) community (https://ormir.org) further developed the ORMIR-MIDS standard and associated support software [47], building on a previous initiative by muscle MRI researchers (https://muscle-bids.github.io/). ORMIR-MIDS strives to be bidirectionally convertible with DICOM, inherently anonymous, and to describe the relevant metadata to be included in the JSON header for each common contrast used in muscle and MSK imaging. ORMIR-MIDS also provides a command-line tool for the conversion of DICOM data, automatically recognizing various contrasts from the main MR scanner vendors, and thus converting each proprietary DICOM implementation into a common format, which can then be relied upon for the implementation of vendor-agnostic pipelines.

3.2. Implementing quantitative MRI and segmentation strategies in clinical practice

John Vissing (Denmark) reviewed challenges for incorporating manual muscle segmentation. The largest obstacle is the highly time-consuming workload of manual muscle segmentation, which precludes segmenting multiple sections along the whole length of the muscle. By just evaluating one or a few muscle sections, some muscle abnormalities may be missed between the two insertion points and the CSA can be underestimated. Also, identifying the pattern of proximal-distal involvement of a muscle can be missed. In longitudinal studies, segmentation of just one or a few slices also creates problems in finding the same location for the section at follow-up. The question is also whether to segment directly at the edge of the fascia or just within the fascia to ensure that proper muscle tissue volume is assessed. For CSA estimates, measuring on the fascia is preferred. A common issue is manual and automatic segmentation of almost end stage muscle, where muscle boundaries are difficult to define. However, end stage muscles do not change over time and do not respond to currently known therapeutic interventions. Therefore, this technical issue of assessment is less clinically relevant. Automatic segmentation techniques are increasingly being utilized, and this removes variability in interrater segmentation. Lastly, he briefly mentioned the development plan for a Danish AI muscle segmentation tool, which is currently in the final deep learning round using an iterative training and correction process.

Glenn Walter (USA) discussed manual segmentation for a Duchenne Muscular Dystrophy (DMD) cohort and regulatory challenges using MRI as a recognized biomarker for trials. MR imaging has become a critical tool for monitoring disease progression and evaluating therapeutic interventions in DMD. This study highlights the role of manual segmentation in standardizing MRI analyses, ensuring reproducibility, and improving biomarker sensitivity across clinical trials. The ImagingDMD initiative has expanded significantly, facilitating multi-center studies aimed at accelerating therapeutic development. Standardization efforts include detailed standard operating procedures (SOPs), training of image readers, use of multiple contrast techniques, and strict quality assurance measures. MRI and MRS, including maximal CSA (CSAmax), global T2, and FF, were highly reproducible across sites, with coefficients of variation (CV) ranging from 2 % to 7 %. Landmark-based segmentation methods enhance consistency in morphometric analysis, particularly in the thigh and lower leg muscles, improving the detection of disease progression. Data acquisition and adjudication protocols incorporate blinded assessments to mitigate bias, ensuring robustness in clinical trial applications. Their findings support the implementation of standardized MRI protocols as reliable biomarkers for assessing muscle degeneration and therapeutic efficacy in DMD. The continued refinement of segmentation techniques and automated tools will further improve imaging-based outcome measures in NMDs.

Mauro Monforte (Italy) discussed the different imaging features in FSHD, and how they related to the well-known phenotypic heterogeneity of the disease, ranging from the classical to the most complex and atypical clinical patterns. The Italian Clinical Network for FSHD is used to categorize patients according to a standardized Comprehensive Clinical Evaluation Form (CCEF) [48], that also incorporates uncommon features identified during clinical evaluation. The main patterns of preserved vs affected muscles and their combinations have been

described by MRI studies assessing large cohorts in FSHD, and further diagnostic value can be clearly added by scanning the upper girdle [34]. A simple pattern (trapezius involvement and bilateral subscapularis sparing) has been found to identify FSHD with high accuracy, also in atypical or cases with incomplete phenotypes [30]. Complex phenotypes in FSHD can also arise from a severe and predominant paraspinal muscle involvement, or, in some cases, due to double-trouble, like the co-occurrence with a distinct genetic NMD [49] or an acquired inflammatory myopathy [50].

Jasper Morrow (UK) presented his experience of muscle segmentation in inherited neuropathies. Using manual segmentation, calf muscle FF using Dixon MRI has consistently proven the most responsive outcome measure in this group of conditions [51]. Single slice analysis of all grouped muscles has been the most commonly used metric. Precise slice localization in the proximal-distal direction is crucial for improving reliability, which is facilitated by 3D Dixon acquisitions with a maximum of 5 mm slice thickness. Careful selection of patients and/or anatomical level for analysis can markedly increase outcome measure responsiveness by avoiding floor and ceiling effects [52]. Appropriate training of segmenters is also crucial and he outlined the training programme used at University College London, UK [18]. The quality of the segmentation can be assessed by reference to gold standard segmentations, or through assessing test-retest or longitudinal datasets. Segmentation is now performed using automated methods [53] requiring minimal changes during a quality control step [19]. Automated segmentation is more time efficient and allows for more detailed analysis of datasets.

3.3. Automatic muscle segmentation in NMD – current techniques, overcoming challenges in clinical practice

Lara Schlaffke (Germany) reviewed automated segmentation challenges in muscle MRI. In an ideal world, clinicians would routinely acquire whole-body quantitative images from all patients, which would then be automatically segmented, analyzed, and reported to neurologists for diagnosis and disease progression monitoring. These images would be uploaded to a centralized server, making them accessible to researchers. However, several challenges must be overcome to achieve this vision. One key challenge is improving communication among the main stakeholders—radiologists, neurologists, and scientists—to establish a shared understanding of clinical requirements and research objectives. Theoretical challenges include reconciling the differing optimal approaches for image acquisition, processing, and segmentation when used for diagnostic purposes versus longitudinal follow-up. Practical challenges involve ensuring sustainable system maintenance, addressing personnel and financial constraints, and defining responsibilities for outcome validation. For automated segmentation to be effectively used in reporting whole-body quantitative outcome measures in patients suspected of having NMDs, it is essential to establish clear communication regarding clinical and research needs, technological possibilities, and future objectives.

Francesco Santini (Switzerland) spoke about centralized federated learning in automatic segmentation. While the prevalence of NMDs, collectively, is roughly equivalent to other better-studied disorders such as multiple sclerosis or Parkinsons disease [54], each of them classified as less common diseases, making any type of data-driven modeling challenging, unless data from multiple centers are collated. However, in healthcare, legal and practical hurdles make sharing patient data difficult and labor-intensive [55], and it is therefore attractive to train models in a decentralized fashion, in so-called federated learning [56]. In this context, multiple institutions keep the data private from each other, and each of them independently train a model; the models are then centrally collected by a server and aggregated, before being redistributed for another round of training. The performance of such a system has been found to be similar to centralized learning [57]. A variant of this system (termed continuous collaborative learning) is present in the free software Dafne [58] (Deep Anatomical Federated Network, https://dafne.network/), in which a graphical interface is provided to the end user. The user can use a segmentation model, which is downloaded from a central server, to provide an automatic segmentation of the desired anatomical region. The user can then use manual tools to refine the proposed segmentation, and these modifications are then used to retrain the model locally on the user's data. The model is then sent back to the server, validated on server-stored data, and, if successful, merged with the baseline model and made available to the next user. While this approach allows the model to improve its generalization capabilities, the lack of a-priori data curation might make the performance of the model unstable over time. In fact, centralized, federated, and collaborative learning each fit different and complementary needs. Centralized learning provides the highest control over the input and is ideal when a model needs to be applied to homogeneous data, but it also requires high computational resources. Traditional federated learning requires lower resources, but also all data to be available at the same time, although not in the same place, to perform the federated rounds; input control is more limited but can still be coordinated. The Dafne approach, on the other hand, is the least resource intensive, but does not allow a-priori control over the data quality, and it therefore allows for the highest generalizability potential, at the cost of slower convergence and potential performance instability.

Martijn Froeling (The Netherlands) outlined AI-based segmentations and analysis for muscle MRI with standards for image acquisition and automated data processing. Dr. Froeling described the MOTION study at UMC Utrecht, where bilateral lower extremities of 162 healthy participants will be scanned using Dixon-based imaging, water T2 mapping, and DTI with fiber tractography [59]. To analyze this data, automated processing is employed, relying on the ORMIR-MIDS data structure and the QMRITools processing toolbox [60]. An essential part of this study is per-muscle analysis. To facilitate this, a lightweight 3D U-Net for automated segmentation was created. Since the U-Net architecture was first proposed [61], many variants have been developed; however, the most successful adaptation is nnU-Net [62]. This framework focuses on data fingerprinting and proper configuration of the network rather than tweaking the network itself. Since its introduction, it has become a useful tool for muscle segmentation, even in muscle diseases [63]. Training a U-Net, especially in 3D, can require heavy computational resources. Therefore, focus has been on developing a U-Net optimized for the segmentation of either lower or upper leg muscles. Because the network is optimized for one specific data type and task, the computational resources required for training are minimized. Furthermore, the use of heavy data augmentation to reduce the amount of data needed for training is essential. The segmentation network is fully integrated into the automated processing software. Applying a trained neural network to data is possible in most programming environments. However, integrating such a network into existing processing software and allowing it to be automated typically requires more effort. To facilitate the use of tools on other datasets, it is recommended that tools are made compatible with the BIDS data structure, and all commands can be run from the command line so that they can be easily integrated into existing processing scripts. To accelerate automated data processing, it is important that data preprocessing is considered when designing a study and the data is well curated. Standardized acquisition protocols are used where possible while still allowing sufficient freedom for customization where needed. It is unrealistic to expect that any type of data can be used for any processing pipeline. Most tools will have limitations or require specific data formats to function properly. Most free tools are a community effort and benefit from user input, feedback, and even contributions to development when needed.

John Thornton (United Kingdom) described work aiming to move qMRI with AI enabled segmentation towards implementation in clinical radiology practice. QMRI outcome measures have been developed to improve trials of new treatments for people with a wide range of NMD. There is now a need to make these methods practically available to

support diagnosis and treatment decisions, and improve follow-up, for individual patients. One model for this is the Quantitative Neuroradiology Initiative [64], which aims to make available to reporting radiologists, within their routine workflow, graphical summaries of key qMRI readouts. The MRI readouts must have a clear evidence base for their clinical value, healthy population reference data must be available, and a visual report format should be designed with technical and clinical validation prior to clinical deployment. A significant challenge is the integration within hospital information systems, so that qMRI reports are available at PACS workstations following automated analysis and automated data routing, with software developed under quality management satisfying regulatory requirements. He described the implementation of a model software infrastructure on this basis [65], allowing them to deploy, as a first exemplar application, a qMRI radiology report for epilepsy patients now used routinely in the hospital [66]-. They recently established the MuscleQuant project to adapt this approach to benefit patients with NMD. AI deep learning enabled automatic image segmentation is a core enabling technology [19,53]. Developing an appropriate graphical muscle qMRI report is the focus of current research – a pressing open question is to resolve which are the most important MRI readouts to include in the quantitative radiology report to inform patient management and treatment decisions for specific NMDs.

Kristl Claeys (Belgium) presented on automated MRI muscle segmentation in patients with NMD. She discussed the study of her research group on automated MRI quantification of volumetric per-muscle FF values in the proximal leg of patients with muscular dystrophies [67]. This study presents and evaluates a clinically relevant approach for the automated 3D segmentation of 18 individual muscles of the proximal leg from knee to hip in healthy individuals and in patients with muscular dystrophies and mild to severe fat replacement, using deep learning models based on a 3D convolutional neural network (CNN) with U-Net architecture [68,69]. To deal with pathology, a separate model was first trained for healthy and mildly affected subjects (low level of fat replacement (LI)) and subsequently retrained and finetuned for more severe cases (high fat replacement group (HI)). She demonstrated the feasibility of quantifying FF automatically in 3D in individual muscles over a broad range of per-muscle FF values (4–92 %) with clinically acceptable accuracy compared to manual analysis. She reported good segmentation results of all 18 muscles individually in terms of overlap (DSC) with the manual ground truth delineation for images with low fat replacement (mean overall FF: 11.3 % [6–16.6]; mean DSC: 95.3 % per image, 84.4–97.3 % per muscle) as well as with medium and high fat replacement (mean overall FF: 44.3 % [18.6–82.1]; mean DSC: 89.0 % per image, 70.8–94.5 % per muscle). Results from a Bland-Altman analysis for quantification of FF and muscle volume in LI and HI cases showed that the FF per muscle obtained using the automated segmentation agrees well with the FF obtained using the ground truth delineation: for LI mostly <1 % (except for a few outliers due to gluteus minimus muscle); and HI: mostly <5 % [67]. The automated segmentation model has meanwhile been extended to the distal lower limbs and is currently being trained for the shoulder and upper limb muscles.

3.4. Integrating machine learning approaches in data analysis

Anna Pichiecchio (Italy) presented bridging the qualitative and quantitative gap in clinical radiomics. QMRI provides crucial insights as a non-invasive tool in assessing disease involvement and progression in NMD. However, qMRI is currently limited to specialized centers for the need of specific sequences and post processing expertise. In contrast, conventional MRI sequences such Short Tau Inversion Recovery (STIR)-based sequences are more widely available in radiological departments, with the limit of being qualitative sequences. Radiomics, a powerful tool for extracting quantitative information from images offers the potential to identify disease patterns by analyzing pixel intensity distributions and spatial relationships in conventional MRI sequences. We investigated the possibility of obtaining quantitative inferior limb muscle biomarkers from conventional STIR and water T2 mapping sequences by combining feature extraction techniques with machine learning methods [70]. The results show that the best model (k-nearest neighbours algorithm, KNN) is a powerful predictor of qMRI parameters, achieving a mean absolute error of $\pm$ 5 percentage points for FF and $\pm$ 1.8 ms for water T2, supporting the potential of using conventional MRI for disease assessment in NMD, even though outcomes have to be better delineated in larger cohorts and longitudinal studies.

Pierre Carlier (France) presented MYOWEB, a web service with a graphic interface for the automatic segmentation of thigh and leg muscles and for the generation of water T2 maps from any segment of the body. The automatic segmentation algorithm makes use of a convolutional neural network and performs either a global or a per-muscle-group segmentation of out-of-phase Dixon images. The water T2 maps are created by the separation of the water and fat components of multi-TE spin echo images either by tri-exponential fitting or with the extended phase graph algorithm. Image processing in batch mode is also possible using command lines. MYOWEB access is provided for free for non-profit use. Requests are to be sent to info@cris-nmr.com

3.5. Data sharing in international imaging platforms to integrate machine learning

Giorgio Tasca (UK) presented challenges and opportunities for building large cohorts for machine-learning based diagnostics and on an ongoing project carried out at the John Walton Muscular Dystrophy Research Centre in Newcastle upon Tyne called MyoGuide. Distinctive patterns of muscle involvement have been identified as characteristic markers for various NMDs and recognising them is helpful in the diagnostic workup [71–73]. However, the complexity and heterogeneity of these patterns make their identification challenging and knowledge is restricted to a limited number of experts in the field. MyoGuide addresses this issue by aiming to provide an automated solution for identifying and analysing patterns of intramuscular fat replacement through custom muscle segmentation, a quantification pipeline, and a diagnostic model. These tools, which are made available through the MyoGuide web portal (www.myoguide.org), have the potential to transform the analysis of muscle MRI by automatically detecting the most distinctive patterns of muscle involvement, facilitating differential diagnosis, and significantly reducing the analysis time. Previously published results on a dataset of 10 muscular dystrophies were promising [29], and the disease range has now been expanded to 20 different NMDs, confirming the strong performance of the model [74].

Jodi Warman-Chardon (Canada) outlined the progress in the development of NMD imaging cohorts and international MRI data sharing platforms. She discussed the risks and benefits of sharing muscle MRIs for clinical assessment and clinical trials. She reviewed MYO-Share, a secure, online imaging portal to collect and view anonymized patient muscle MRIs that was established to build large, rare NMD imaging cohorts to help delineate disease-specific imaging patterns [75]. MYO-Share was developed based on recommendations of the MYO-MRI consortium [76], which brings together top international specialists (neuromuscular neurologists, radiologists) (www.myo-mri.eu). MYO-Share is now being leveraged to build large international rare NMD patient cohorts in 20 countries with 100 investigators to increase MRI use as a diagnostic imaging biomarker, to monitor disease progression and response to therapy [29].

4. Discussion

4.1. Workshop overview

This 286th ENMC Workshop on AI and Muscle MRI brought together an interdisciplinary group of experts to identify clinical standards for qMRI acquisition for diagnosis and longitudinal assessment (whole body

vs lower extremity) and imaging storage. Jasper Morrow summarized recommendations based on workshop discussions. He reviewed that successful deployment of machine learning in diagnostic muscle MRI depends on developing large, well-defined patient cohorts to validate quantitative parameters and diagnostic algorithms, ensuring robust clinical translation and evidence-based implementation of this advanced imaging methodology for clinical translation. A standardized, rapid, and reliable muscle qMRI protocol is essential for incorporating automated segmentation and AI-driven diagnostic tools to identify characteristic imaging patterns. To ensure broad accessibility and utility, a centralized platform should support remote analysis, standardized uploads, and ongoing model refinement, with potential for local deployment, full-body imaging, and pharmaceutical trial applications.

4.2. Standardized reporting guidelines

Based on the preworkshop questionnaire outlined above, clinical implementation of muscle MRI remains limited by a lack of knowledge of the anatomy, time required to analyze all muscles and lack of readily available reporting guidelines. Although the long-term objective of integrating qMRI into routine clinical practice is widely endorsed due to its superior sensitivity for detecting subtle pathological changes and capacity for objective assessment, structured reporting guidelines and interdisciplinary education for radiologists and neuromuscular specialists are essential to support consistent implementation in clinical practice. Sarah Schlager outlined the recently created German neuroradiology guidelines for reporting for MRI [77,78]. Based on those guidelines, previous recommendations [8] and the input during and after the ENMC workshop, we drafted a reporting outline (Appendix 1) for clinical radiologists.

4.3. Protocol recommendations

The lack of standardization of qMRI protocols in clinical use and research limits multicentre comparison for clinical studies. Many centres are currently acquiring muscle MRI using turbo spin-echo (TSE)-based T1 weighted images to assess the extent of fat replacement and fat-suppressed STIR TSE-based T2 weighted images to assess hyperintense signal related to disease activity (such as edema, inflammation). These qualitative scans miss the opportunity to collect quantitative data for clinical analysis and future studies. Moreover, STIR can be prone to artifacts such as surface coil artifacts, causing nonuniformity in the signal. Therefore, when possible, centres should move towards routinely integrating clinical NMD imaging with quantitative imaging techniques. Consensus was reached that the imaging protocol should preferably contain a whole-body Dixon for FF, and if needed either a spin-echo Dixon or MESE for water T2. These sequences can be used for qualitative diagnosis and may require less scan time than a typical T1 weighted image. As well as assessed qualitatively for routine clinical use, qMRI scans can be assessed by manual or automatic segmentation for natural history studies or baseline for response to therapy.

4.4. Segmentation

Improved methods for both individual and group muscle segmentation are essential to enhance the accuracy and consistency of qMRI analysis. While manual delineation of ROIs varies by disease and anatomy, maintaining internal consistency is critical. For example, muscles may be traced around their full contour or using a minimal area threshold. A common strategy discussed was tracing the muscle edge followed by a one-pixel erosion to reduce partial volume effects, which is particularly important for small muscles. Post-processing choices include selecting appropriate image types (e.g., Dixon-derived in-phase, Dixon-derived out-of-phase images or acquired water-fat in-phase and out-of-phase) and using both baseline and follow-up scans in a blinded fashion. For CSA measurements, tracing at the muscle edge and also eroding by one acquisition voxel may be implemented, while acquiring the maximal possible muscle volume during imaging allows for more precise alignment and analysis during post-processing.

Training for manual muscle segmentation should incorporate clear benchmarks for reliability, including reference standards and validated metrics [18]. Individual segmenters require rigorous training, supported by structured resources such as training manuals, direct instruction, and annotated MRI scans covering both large and small regions of interest (ROIs). While multiple observers may share the workload to allow inter-rater comparisons, consistency is best maintained when the same segmenter analyzes baseline and follow-up scans for each subject. ROI delineation should use all available image types, tracing full muscle borders and aligning volumes retrospectively using both baseline and repeat scans. Ideally, a single reader should segment the muscle MRI images for each subject's data in longitudinal studies, although multiple trained readers may segment across different subjects if needed. Whereas single-muscle values remain important for diagnosis, averaging FF values across muscle groups can enhance sensitivity for detecting disease progression in trials [25,37,51,79]. Developing standardized atlases and training materials will be crucial to improve segmentation reproducibility and diagnostic reliability.

There are multiple different automatic segmentation algorithms in development [32,58,74,80,81]. The potential for comparing automatic muscle segmentation tools was discussed, including the development of a standardized "segmentation challenge," analogous to Hackathons, to compare algorithm performance. Such an initiative would involve the use of standardized MRI datasets from individuals with various NMDs and healthy controls. Evaluation metrics, including Hausdorff Distance (HD), SRM and DSC, should be employed to assess segmentation accuracy and robustness across common technical variables, including slice gap variation and disease-specific muscle pathology. Importantly, the workshop emphasized the need for strategic investment in infrastructure to support the clinical implementation of automated segmentation, including funding from the European Union and other governmental support, as well as partnerships with industry leaders such as Philips, GE, and Siemens. Developing clinic-ready solutions has the potential to significantly advance diagnostic precision and longitudinal monitoring in both routine care and NMD clinical trials.

4.5. Data sharing

Robust data sharing practices for MRI are essential to support multicentre clinical trials and longitudinal studies, as well as testing and training of automated segmentation algorithms. Standardization of acquisition protocols and post-processing methods is necessary to ensure comparability across sites. Ideally, a centralized facility should oversee offline post-processing to maintain consistency. MRI data should be stored in a standardized data storage and sharing format such as ORMIR-MIDs format (https://github.com/ormir-mids). Adherence to a common data saving structure, such as ORMIR-MIDs, is crucial for enabling downstream analyses, including post-processing, harmonization, and cross-centre comparisons. Discussions emphasized the importance of harmonizing data sharing practices to increase the availability of datasets, particularly for natural history studies. There was strong consensus that imaging data collected during natural history studies and clinical trials should be made publicly available, as is already being done within some NMD communities, such as in FSHD [82]. To encourage pharmaceutical and biotech companies to contribute data, workshop participants suggested that data-sharing clauses be incorporated into trial contracts.

Establishing a reference database, including age- and system-specific normative datasets, is essential for comparative studies. For example, the NIH has created a nuclear magnetic resonance database of over 400 healthy individuals, which can serve as a comparative baseline. Imaging biomarkers, such as FF and water T2, should be interpreted in the context of age-related changes and scanner-specific variability, akin to

how laboratory values are interpreted with respect to reference ranges and acceptable margins of error following upgrades or protocol changes. Also, developing a centralized repository that supports multiple scanner types (e.g., Siemens, Philips 3T) and field strengths would be similar to existing frameworks for systematic analysis and would help account for platform-specific differences.

The discussion further highlighted current limitations in muscle qMRI techniques, particularly regarding the interpretation of FF values. Reliable use of FF measurements requires normative data for each muscle, as certain muscles—such as the gluteus maximus and lower paraspinals—naturally exhibit higher FF with age. Reference values (presented as mean ± standard deviation) are needed across scanner platforms, but existing literature is insufficient to establish comprehensive site-specific norms. This variability is analogous to clinical laboratory testing, where acceptable ranges may differ by machine but retain clinical validity. Technical constraints also include floor and ceiling effects; for instance, Dixon-based methods have a noise floor of approximately 5–6 %, limiting the detection of very low FF values. Accurate quantification requires control datasets, ideally including at least 50 healthy subjects per site, to establish local reference databases for software validation. Although FF differences of 1–2 % may not be clinically meaningful, changes of $\geq$10 % could significantly impact trial outcomes.

Privacy concerns surrounding MRI data sharing were also discussed, as they pose a significant challenge for collaborative rare disease research. The General Data Protection Regulation (GDPR) in Europe provides robust safeguards for personal data but also places strict constraints on how imaging data can be stored, processed, and shared across borders. Because muscle MRI scans can inadvertently capture identifiable features, such as facial structures or unique body characteristics, there is a growing risk of re-identification through advanced facial recognition algorithms. To mitigate this, certain MRI protocols have been adapted to exclude the head or facial regions altogether, focusing only on relevant muscle groups to reduce privacy risks. However, completely eliminating identifying features while maintaining diagnostic quality can be technically challenging, especially in diseases involving cranial or bulbar muscles. Moreover, differences in privacy legislation between countries add further complexity, as data transfers must comply with local regulations while supporting international research collaborations. Workshop participants emphasized the importance of developing standardized anonymization pipelines and secure, encrypted data storage solutions to protect patient confidentiality. In addition, clear consent procedures must be implemented to inform patients about how their data will be used, shared, and protected throughout multi-centre studies. Achieving a balance between strict privacy compliance and open, FAIR (Findable, Accessible, Interoperable, Reusable) data sharing is essential to advance qMRI applications in NMD. Future efforts should include cross-border agreements and technical safeguards that enable secure data exchange while maintaining the highest standards of patient privacy and ethical research practice.

In summary, this ENMC workshop underscored the urgent need for standardized, robust, and accessible muscle qMRI protocols to advance diagnosis and monitoring in NMD and eventually the implementation of qMRI in the clinical routine. Participants highlighted key challenges, including limited anatomical expertise, inconsistent reporting, and technical constraints that hinder clinical adoption, longitudinal assessments and multicentre comparability. Consensus recommendations emphasized implementing whole-body imaging where feasible, developing rigorous training resources for manual segmentation, and expanding data sharing through harmonized platforms and privacy-conscious frameworks. Strategic investment in infrastructure, industry partnerships, and clear data standards will be critical for integrating automatic segmentation and AI-driven tools to more effectively incorporate quantitative imaging biomarkers into routine practice and clinical trials. Ultimately, these collective efforts aim to accelerate evidence-based use of muscle MRI, improve objective assessment, diagnostic accuracy, and strengthen the design and execution of future therapeutic studies.

5. Workshop participants

Pierre G. Carlier, St Luc University and Erasme University Hospitals, Brussels, Belgium.

Kristl Claeys, Department of Neurology, University Hospitals Leuven, Belgium.

Martijn Froeling, University Medical Center Utrecht, Belgium.

Hermien E Kan, Department of Radiology, Leiden University Medical Centre, The Netherlands.

Mauro Monforte, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy.

Jasper Morrow, Queen Square Centre for Neuromuscular Diseases, University College London, UK.

Anna Pichiecchio, University of Pavia, Italy & IRCCS Mondino Foundation, Italy.

Susanne S. Rauh, Department of Radiology, Leiden University Medical Centre, The Netherlands.

Harmen Reyngoudt, Neuromuscular Investigation Center, Institute of Myology, Paris, France.

Francesco Santini, Department of Biomedical Engineering, University Hospital of Basel, Switzerland.

Sarah Schlaeger Department of Radiology, LMU University Hospital, LMU Munich, Germany.

Lara Schlaffke, Department of Neurology, BG-University Hospital Bergmannsheil, Ruhr-University Bochum, Germany.

Volker Straub, John Walton Muscular Dystrophy Research Centre, Newcastle University, UK.

Giorgio Tasca, John Walton Muscular Dystrophy Research Centre, Newcastle University, UK.

John Thornton, University College London Queen Square Institute of Neurology, London, UK.

John Vissing, Department of Neurology, Rigshospitalet, University of Copenhagen, Denmark.

Glenn Walter, Department of Physiology and Aging, University of Florida, Gainesville, USA.

Jodi Warman-Chardon, The Ottawa Hospital/Children's Hospital of Eastern Ontario, Canada.

CRediT authorship contribution statement

Jodi Warman-Chardon: Conceptualization, Data curation, Formal analysis, Writing – original draft, Writing – review & editing. **Volker Straub:** Conceptualization, Data curation, Formal analysis, Writing – original draft, Writing – review & editing. **John Vissing:** Conceptualization, Data curation, Formal analysis, Writing – original draft, Writing – review & editing. **Sarah Schlaeger:** Conceptualization, Writing – original draft, Writing – review & editing. **Hermien E. Kan:** Conceptualization, Data curation, Visualization, Writing – original draft, Writing – review & editing.

Declaration of competing interest

The authors declare the following financial interests/personal relationships which may be considered as potential competing interests:

Jodi Warman-Chardon, Volker Straub, John Vissing, Sara Shlager, Hermien Kan and the co-authors of the ENMC #286 conference reports travel and conference support was provided by ENMC. There are no other known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Acknowledgements

The organizers warmly acknowledge the staff of ENMC: Patricia van

Dongen, Programme Manager, Wilma Hinloopen, Operational Manager and Tamara van Esch, Workshop Assistant for organizational support for this workshop. The workshops and next generation programme are made possible thanks to the financial support of the European Neuromuscular Centre (ENMC) and its Full Partners: Association Française contre les Myopathies (France), Deutsche Gesellschaft für Muskelkranke (Germany), Muscular Dystrophy Campaign (UK), Muskelsvindfonden (Denmark), Prinses Beatrix Spierfonds (The Netherlands), Schweizerische Stiftung für die Erforschung der Muskelkrankheiten (Switzerland), Spierziekten Nederland (The Netherlands), Telethon Foundation (Italy). In addition, we would like to thank the Associated Partners: Österreichische Muskelforschung (Austria), SMA Europe, TREAT-NMD, World Duchenne Organisation, World Muscle Society (WMS), and the members of the ENMC Company Forum: Amicus Therapeutics, Dyne Therapeutics, Lupin Neuroscience, Novartis, Roche, Sanofi, Santhera and Sarepta.

Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:10.1016/j.nmd.2025.106304.

References

[1] Ogier AC, Hostin M-A, Bellemare M-E, Bendahan D. Overview of MR image segmentation strategies in neuromuscular disorders. Front Neurol 2021;12.
[2] Warman-Chardon J, Diaz-Manera J, Tasca G, Straub V. 247th ENMC International Workshop: muscle magnetic resonance imaging - implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts. Hoofddorp, The Netherlands. September 2019 Neuromuscul Disord 2020;30:938–47.
[3] Domínguez-González C, Fernández-Torrón R, Moore U, de Fuenmayor-Fernández de la Hoz CP, Vélez-Gómez B, Cabezas JA, et al. Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis. J Neurol 2022.
[4] Schiava M, Ikenaga C, Villar-Quiles RN, Caballero-Ávila M, Topf A, Nishino I, et al. Genotype-phenotype correlations in valosin-containing protein disease: a retrospective muticentre study. J Neurol Neurosurg Psychiatr 2022.
[5] Melkus G, Sampaio ML, Smith IC, Rakhra KS, Bourque PR, Breiner A, et al. Quantitative vs qualitative muscle MRI: imaging biomarker in patients with oculopharyngeal muscular dystrophy (OPMD). Neuromuscular disorders: NMD; 2022.
[6] Burakiewicz J, Sinclair CDJ, Fischer D, Walter GA, Kan HE, Hollingsworth KG. Quantifying fat replacement of muscle by quantitative MRI in muscular dystrophy. J Neurol 2017;264:2053–67.
[7] Strijkers GJ, Araujo ECA, Azzabou N, Bendahan D, Blamire A, Burakiewicz J, et al. Exploration of new contrasts, targets, and MR imaging and spectroscopy techniques for neuromuscular disease - A workshop report of Working Group 3 of the Biomedicine and Molecular Biosciences COST action BM1304 MYO-MRI. J Neuromuscul Dis 2019;6:1–30.
[8] Warman Chardon J, Díaz-Manera J, Tasca G, Bönnemann CG, Gómez-Andrés D, Heerschap A, et al. MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscul Disord 2019;29:827–41.
[9] Barp A, Laforet P, Bello L, Tasca G, Vissing J, Monforte M, et al. European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). J Neurol 2020;267:45–56.
[10] Tasca G, Monforte M, Diaz-Manera J, Brisca G, Semplicini C, D'Amico A, et al. MRI in sarcoglycanopathies: a large international cohort study. J Neurol Neurosurg Psychiatr 2018;89:72–7.
[11] Peric S, Maksimovic R, Banko B, Durdic M, Bjelica B, Bozovic I, et al. Magnetic resonance imaging of leg muscles in patients with myotonic dystrophies. J Neurol 2017;264:1899–908.
[12] Lehmann Urban D, Mohamed M, Ludolph AC, Kassubek J, Rosenbohm A. The value of qualitative muscle MRI in the diagnostic procedures of myopathies: a biopsy-controlled study in 191 patients. Ther Adv Neurol Disord 2021;14:1756286420985256.
[13] Vandenborne K, Walter GA, Straub V, Willcocks RJ, Forbes SC, Mercuri EM, et al. Quantitative muscle Magnetic resonance outcomes in patients with Duchenne muscular dystrophy: an exploratory analysis from the EMBARK randomized clinical trial. JAMA Neurol 2025.
[14] Revsbech KL, Rudolf K, Sheikh AM, Khawajazada T, de Stricker, Borch J, Dahlqvist JR, et al. Axial muscle involvement in patients with limb girdle muscular dystrophy type R9. Muscle Nerve 2022;65:405–14.
[15] Paoletti M, Monforte M, Barzaghi L, Tasca G, Bergsland N, Faggioli A, et al. Natural history of facioscapulohumeral muscular dystrophy evaluated by multiparametric quantitative MRI: a prospective cohort study. J Neurol 2025;272:306.
[16] Kroon R, Kalf JG, de Swart BJM, Heskamp L, de Rooy JWJ, van Engelen BGM, et al. Muscle MRI in patients with oculopharyngeal muscular dystrophy. Neurology 2024;102:e207833.
[17] Schlaffke L, Rehmann R, Froeling M, Güttsches AK, Vorgerd M, Enax-Krumova E, et al. Quantitative muscle MRI in sporadic inclusion body myositis (sIBM): a prospective cohort study. J Neuromuscul Dis 2024;11:997–1009.
[18] Morrow JM, Shah S, Cristiano L, Evans MRB, Doherty CM, Alnaemi T, et al. Development of an initial training and evaluation programme for manual lower limb muscle MRI segmentation. European Radiol Experiment 2024;8:85.
[19] O'Donnell LF, Pipis M, Thornton JS, Kanber B, Wastling S, McDowell A, et al. Quantitative MRI outcome measures in CMT1A using automated lower limb muscle segmentation. J Neurol Neurosurg Psychiatr 2024;95:500–3.
[20] Engelke K, Chaudry O, Gast L, Eldib MAB, Wang L, Laredo J-D, et al. Magnetic resonance imaging techniques for the quantitative analysis of skeletal muscle: state of the art. J Orthop Translat 2023;42:57–72.
[21] Melkus G, Sampaio ML, Smith IC, Rakhra KS, Bourque PR, Breiner A, et al. Quantitative vs qualitative muscle MRI: imaging biomarker in patients with oculopharyngeal muscular dystrophy (OPMD). Neuromuscul Disord 2023;33:24–31.
[22] Lilien C, Reyngoudt H, Seferian AM, Gidaro T, Annoussamy M, Chê V, et al. Upper limb disease evolution in exon 53 skipping eligible patients with Duchenne muscular dystrophy. Ann Clin Transl Neurol 2021.
[23] Dahlqvist JR, Widholm P, Leinhard OD, Vissing J. MRI in neuromuscular diseases: an emerging diagnostic tool and biomarker for prognosis and efficacy. Ann Neurol 2020;88:669–81.
[24] Reyngoudt H, Smith FE, Caldas de Almeida Araújo E, Wilson I, Fernández-Torrón R, James MK, et al. Three-year quantitative magnetic resonance imaging and phosphorus magnetic resonance spectroscopy study in lower limb muscle in dysferlinopathy. J Cachexia Sarcopenia Muscle 2022.
[25] Alonso-Jiménez A, Nuñez-Peralta C, Montesinos P, Alonso-Pérez J, García C, Montiel E, et al. Different approaches to analyze muscle fat replacement with Dixon MRI in Pompe disease. Front Neurol 2021;12:675781.
[26] Heskamp L, Ogier A, Bendahan D, Heerschap A. Whole-muscle fat analysis identifies distal muscle end as disease initiation site in facioscapulohumeral muscular dystrophy. Commun Med (Lond) 2022;2:155.
[27] Hooijmans MT, Niks EH, Burakiewicz J, Anastasopoulos C, van den Berg SI, van Zwet E, et al. Non-uniform muscle fat replacement along the proximodistal axis in Duchenne muscular dystrophy. Neuromuscul Disord 2017;27:458–64.
[28] Ansari B, Salort-Campana E, Ogier A, Le Troter Ph DA, De Sainte Marie B, Guye M, et al. Quantitative muscle MRI study of patients with sporadic inclusion body myositis. Muscle Nerve 2020;61:496–503.
[29] Verdu-Diaz J, Alonso-Perez J, Nunez-Peralta C, Tasca G, Vissing J, Straub V, et al. Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology 2020.
[30] Monforte M, Bortolani S, Torchia E, Cristiano L, Laschena F, Tartaglione T, et al. Diagnostic magnetic resonance imaging biomarkers for facioscapulohumeral muscular dystrophy identified by machine learning. J Neurol 2022;269:2055–63.
[31] Hostin MA, Ogier AC, Michel CP, Le Fur Y, Guye M, Attarian S, et al. The impact of fatty infiltration on MRI segmentation of lower limb muscles in neuromuscular diseases: a comparative study of deep learning approaches. J Magnet Resonan Imag 2023;58:1826–35.
[32] Agosti A, Shaqiri E, Paoletti M, Solazzo F, Bergsland N, Colelli G, et al. Deep learning for automatic segmentation of thigh and leg muscles. Magnetic resonance materials in physics. Biol Med 2022;35:467–83.
[33] Alonso-Jimenez A, Kroon R, Alejaldre-Monforte A, Nunez-Peralta C, Horlings CGC, van Engelen BGM, et al. Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. J Neurol Neurosurg Psychiatr 2019;90:576–85.
[34] Tasca G, Monforte M, Iannaccone E, Laschena F, Ottaviani P, Leoncini E, et al. Upper girdle imaging in facioscapulohumeral muscular dystrophy. PloS One 2014;9:e100292.
[35] Jenkins BM, Dixon LD, Kokesh KJ, Zingariello CD, Vandenborne K, Walter GA, et al. Skeletal muscle symptoms and quantitative MRI in females with dystrophinopathy. Muscle Nerve 2024;70:988–99.
[36] Giacomucci G, Monforte M, Diaz-Manera J, Mul K, Fernandez Torrón R, Maggi L, et al. Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. Eur J Neurol 2020;27:2604–15.
[37] van de Velde NM, Hooijmans MT, Sardjoe Mishre ASD, Keene KR, Koeks Z, Veeger TTJ, et al. Selection approach to identify the optimal biomarker using quantitative muscle MRI and functional assessments in Becker muscular dystrophy. Neurology 2021;97:e513–22.
[38] Chrzanowski SM, Baligand C, Willcocks RJ, Deol J, Schmalfuss I, Lott DJ, et al. Multi-slice MRI reveals heterogeneity in disease distribution along the length of muscle in Duchenne muscular dystrophy. Acta Myologica 2017;36:151–62.
[39] Mul K, Vincenten SCC, Voermans NC, Lemmers R, van der Vliet PJ, van der Maarel SM, et al. Adding quantitative muscle MRI to the FSHD clinical trial toolbox. Neurology 2017;89:2057–65.
[40] Moore U, Caldas de Almeida Araújo E, Reyngoudt H, Gordish-Dressman H, Smith FE, Wilson I, et al. Water T2 could predict functional decline in patients with dysferlinopathy. J Cachexia Sarcopenia Muscle 2022;13:2888–97.
[41] Bolano-Diaz CF, Reyngoudt H, Wilson IJ, James MK, Smith FE, Caldas de Almeida Araujo E, et al. Modeling of dysferlinopathy (LGMDR2) progression: a longitudinal fat fraction analysis. Neurol Genet 2025;11:e200283.
[42] Wilson I, Reyngoudt H, de Almeida, Araujo EC, Baudin P, Marty B, Bolano-Diaz C, et al. P125 Quantitative MRI in upper limb muscles of patients with dysferlinopathy: 6-months and 12-months longitudinal data from the natural history Jain COS 2 project. Neuromuscul Disorder 2023;33:S78–9.
[43] Bell LC, Shimron E. Sharing data is essential for the future of AI in medical imaging. Radiol Artif Intell 2024;6:e230337.

[44] Larobina M. Thirty years of the DICOM standard. Tomography 2023;9:1829–38.
[45] Gorgolewski KJ, Auer T, Calhoun VD, Craddock RC, Das S, Duff EP, et al. The brain imaging data structure, a format for organizing and describing outputs of neuroimaging experiments. Sci Data 2016;3:160044.
[46] Saborit-Torres JM, Nadal-Almela S, Montell-Serrano JA, Oliver-Garcia E, Carceller H, Gómez-Ádrian JA, et al. Beyond the brain: MIDS extends BIDS to multiple modalities and anatomical regions. Stud Health Technol Inform 2022;295:116–7.
[47] Bonaretti S, Hernandez M, Chiumento F, Founas Y, Froeling M, Hirsvaniemi J, et al. ORMIR-MIDS: an open standard for curating and sharing musculoskeletal imaging data; abstracts of the 24th quantitative musculoskeletal imaging workshop. J Bone Mineral Res 2025;9. i1–i128.
[48] Ricci G, Ruggiero L, Vercelli L, Sera F, Nikolic A, Govi M, et al. A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. J Neurol 2016;263:1204–14.
[49] Puma A, Tammam G, Ezaru A, Slioui A, Torchia E, Tasca G, et al. Double trouble: a comprehensive study into unrelated genetic comorbidities in adult patients with facioscapulohumeral muscular dystrophy type I. Europe J Human Genet 2025.
[50] Lauletta A, Allenbach Y, Béhin A, Evangelista T, Léonard-Louis S, Garibaldi M, et al. High prevalence of facioscapulohumeral muscular dystrophy (FSHD) and inflammatory myopathies association: is there an interplay? J Neurol Sci 2025;470:123400.
[51] Morrow JM, Sinclair CD, Fischmann A, Machado PM, Reilly MM, Yousry TA, et al. MRI biomarker assessment of neuromuscular disease progression: a prospective observational cohort study. Lancet Neurol 2016;15:65–77.
[52] Morrow JM, Evans MRB, Grider T, Sinclair CDJ, Thedens D, Shah S, et al. Validation of MRC Centre MRI calf muscle fat fraction protocol as an outcome measure in CMT1A. Neurology 2018;91:e1125–9.
[53] Kanber B, Morrow JM, Klickovic U, Wastling S, Shah S, Fratta P, et al. Musclesense: a trained, artificial neural network for the anatomical segmentation of lower limb magnetic resonance images in neuromuscular diseases. Neuroinformatics 2021;19:379–83.
[54] Deenen JC, Horlings CG, Verschuuren JJ, Verbeek AL, van Engelen BG. The epidemiology of neuromuscular disorders: a comprehensive overview of the literature. J Neuromuscul Dis 2015;2:73–85.
[55] van Panhuis WG, Paul P, Emerson C, Grefenstette J, Wilder R, Herbst AJ, et al. A systematic review of barriers to data sharing in public health. BMC Public Health 2014;14:1144.
[56] McMahan B, Moore E, Ramage D, Hampson S, BAy Arcas. Communication-efficient learning of deep networks from decentralized data. In: Aarti S, Jerry Z, editors. Proceedings of the 20th international conference on artificial intelligence and statistics. Editors. PMLR: Proceedings of Machine Learning Research; 2017. p. 1273–82.
[57] Czeizler E, Wiessler W, Koester T, Hakala M, Basiri S, Jordan P, et al. Using federated data sources and Varian learning portal framework to train a neural network model for automatic organ segmentation. Phys Med 2020;72:39–45.
[58] Santini F, Wasserthal J, Agosti A, Deligianni X, Keene KR, Kan HE, et al. Deep anatomical federated network (Dafne): an open client-server framework for the continuous, collaborative improvement of deep learning-based medical image segmentation. Radiol Artif Intell 2025:e240097.
[59] Schlaffke L, Rehmann R, Rohm M, Otto LAM, de Luca A, Burakiewicz J, et al. Multi-center evaluation of stability and reproducibility of quantitative MRI measures in healthy calf muscles. NMR Biomed 2019;32:e4119.
[60] Froeling M. QMRTools: a Mathematica toolbox for quantitative MRI analysis. J Open Source Software 2019;4.
[61] Ronneberger O., Fischer P., Brox T., U-Net: convolutional networks for biomedical image segmentation. Vol. 9351. 2015. 234–41.
[62] Isensee F, Jaeger PF, Kohl SAA, Petersen J, Maier-Hein KH. nnU-Net: a self-configuring method for deep learning-based biomedical image segmentation. Nat Methods 2021;18:203–11.
[63] Chow BVY, Morgan C, Rae C, Warton DI, Novak I, Davies S, et al. Human lower leg muscles grow asynchronously. J Anat 2024;244:476–85.
[64] Goodkin O, Pemberton HG, Vos SB, Prados F, Das RK, Moggridge J, et al. Clinical evaluation of automated quantitative MRI reports for assessment of hippocampal sclerosis. Eur Radiol 2021;31:34–44.
[65] Moggridge J, Vos S, Prados F, Grech-Sollars M, Matthews D, Schmidt P, et al. Clinical deployment of novel quantitative neuro-image analysis techniques using containerisation technology'. In: Proceedings of the International Society for Magnetic Resonance in Medicine annual Meeting and Exhibition. Abstract 3304; 2023.
[66] Goodkin O, Pemberton H, Vos SB, Prados F, Sudre CH, Moggridge J, et al. The quantitative neuroradiology initiative framework: application to dementia. Br J Radiol 2019;92:20190365.
[67] Huysmans L, De Wel B, Claeys KG, Maes F. Automated MRI quantification of volumetric per-muscle fat fractions in the proximal leg of patients with muscular dystrophies. Front Neurol 2023;14:1200727.
[68] De Wel B, Huysmans L, Peeters R, Goosens V, Ghysels S, Byloos K, et al. Prospective natural history study in 24 adult patients with LGMDR12 over 2 years of follow-up: quantitative MRI and clinical outcome measures. Neurology 2022;99:e638–49.
[69] De Wel B, Iterbeke L, Huysmans L, Peeters R, Goosens V, Dubuisson N, et al. Lessons for future clinical trials in adults with Becker muscular dystrophy: disease progression detected by muscle magnetic resonance imaging, clinical and patient-reported outcome measures. Eur J Neurol 2024;31:e16282.
[70] Colelli G, Barzaghi L, Paoletti M, Monforte M, Bergsland N, Manco G, et al. Radiomics and machine learning applied to STIR sequence for prediction of quantitative parameters in facioscapulohumeral disease. Front Neurol 2023;14:1105276.
[71] Tasca G, Monforte M, Iannaccone E, Laschena F, Ottaviani P, Silvestri G, et al. Muscle MRI in female carriers of dystrophinopathy. Eur J Neurol 2012;19:1256–60.
[72] Tasca G, Udd B. Hereditary myopathy with early respiratory failure (HMERF): still rare, but common enough. Neuromuscul Disord 2018;28:268–76.
[73] Giacomucci G, Monforte M, Diaz-Manera J, Mul K, Fernandez Torrón R, Maggi L, et al. Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. Eur J Neurol 2020.
[74] Verdu-Diaz J, Bolano-Diaz C, Chamorro-Gonzalez A, Fitzsimmons S, Kocak G, Alvim D, et al. Myo-Guide: a Machine Learning-based web application for neuromuscular disease diagnosis with MRI. J Cachexia Sarcopenia Muscle 2025. Accepted: JCSM13815.
[75] ten Dam L, van der Kooi AJ, van Wattingen M, de Haan RJ, de Visser M. Reliability and accuracy of skeletal muscle imaging in limb-girdle muscular dystrophies. Author response. Neurology 2013;80:2276–7.
[76] Straub V, Carlier PG, Mercuri E. TREAT-NMD workshop: pattern recognition in genetic muscle diseases using muscle MRI: 25-26 February 2011, Rome, Italy. Neuromuscul Disord 2012. 22 Suppl 2:S42-53.
[77] Zeng R, Schlaeger S, Türk M, Baum T, Deschauer M, Janka R, et al. [Expert recommendations for magnetic resonance imaging of muscle disorders]. Radiologie (Heidelb) 2024;64:653–62.
[78] Zeng R, Schlaeger S, Türk M, Baum T, Deschauer M, Janka R, et al. Expertenempfehlung zur magnetresonanztomographie bei Muskelerkrankungen. Die Radiologie 2024;64:653–62.
[79] Reyngoudt H, Marty B, Boisserie JM, Le Louër J, Koumako C, Baudin PY, et al. Global versus individual muscle segmentation to assess quantitative MRI-based fat fraction changes in neuromuscular diseases. Eur Radiol 2021;31:4264–76.
[80] Baudin PY, Balsiger F, Beck L, Boisserie JM, Jouan S, Marty B, et al. A minimal annotation pipeline for deep learning segmentation of skeletal muscles. NMR Biomed 2025;38:e70066.
[81] Martin S, André R, Trabelsi A, Michel CP, Fortanier E, Attarian S, et al. Importance of neural network complexity for the automatic segmentation of individual thigh muscles in MRI images from patients with neuromuscular diseases. Magma 2025;38:175–89.
[82] Monforte M, Attarian S, Vissing J, Diaz-Manera J, Tasca G, Attarian S, et al. 265th ENMC International Workshop: muscle imaging in Facioscapulohumeral Muscular dystrophy (FSHD): relevance for clinical trials. 22–24 April 2022, Hoofddorp, The Netherlands. Neuromuscul Disorder 2023;33:65–75.