

Implementation of an artificial intelligence module on the online imaging portal MYO-Share for guiding the diagnosis of muscle diseases

Submission date	Recruitment status	<input type="checkbox"/> Prospectively registered
11/05/2023	No longer recruiting	<input type="checkbox"/> Protocol
Registration date	Overall study status	<input type="checkbox"/> Statistical analysis plan
06/06/2023	Completed	<input type="checkbox"/> Results
Last Edited	Condition category	<input type="checkbox"/> Individual participant data
02/06/2023	Musculoskeletal Diseases	<input type="checkbox"/> Record updated in last year

Plain English summary of protocol

Background and study aims

Genetic muscle diseases are a group of over 200 inherited disorders that cause progressive muscle weakness and wasting due to fat replacing muscles. Clinicians use muscle magnetic resonance imaging (MRI) to identify fat replacement, which helps to diagnose the disease. The pattern of muscle involvement accurately describes muscles progressively replaced by fat in a specific disease. Researchers use a score called the Lamminen-Mercuri score to quantify the amount of fat in muscles to identify patterns of muscle involvement. Genetic diagnosis is the gold standard for diagnosing and categorizing muscle disease. A machine learning-based software called MYO-Guide has been developed to analyze muscle MRIs and predict high-accuracy diagnoses of 10 muscle diseases. This software could help clinicians who are not specialized in identifying muscle disease types from MRI images and those working in busy and resource-limited health centres to help with selecting genes for analysis or verifying candidate gene variants. This tool could suggest the genes that should be analyzed using sequencing in hospitals, speeding up the diagnosis of patients. This study has two main aims:

1. To use artificial intelligence, specifically machine learning, to analyze muscle MRIs of patients with confirmed neuromuscular diseases to develop an algorithm to predict the diagnosis.
2. To create an automatic segmentation tool that can delineate muscles of the pelvis, thigh and leg and automatically quantify skeletal muscle fat replacement using the Lamminen-Mercuri scale.

Who can participate?

This is a data archive study and no patients will be recruited into the study. This study will be using historical muscle MRI scans as well as limited patient data (i.e. age, sex, and genetic diagnosis of muscle disease)

What does the study involve?

The Newcastle University research team plans to use muscle MRI images for developing a machine-learning model to predict neuromuscular diseases. They will score the images and

collect data for the algorithm. To aid clinicians with the scoring process, an automatic segmentation tool using neural network technology will be developed. This tool will identify and score individual muscles. The team will collect MRI images to inform both the diagnostic tool and the automatic segmentation tool. Anonymized MRI images and patient data will be obtained from NHS sites and healthcare settings worldwide via an online platform or from data archives and Newcastle University. The MRI scans for the automatic segmentation tool will be stored in a folder on the Newcastle University server and viewed using specialized software.

What are the possible benefits and risks of participating?

None

Where is the study run from?

Newcastle University (UK)

When is the study starting and how long is it expected to run for?

September 2021 to December 2025

Who is funding the study?

1. AFM Telethon (France)
2. Muscular Dystrophy UK

Who is the main contact?

Prof. Jordi Diaz Manera, jordi.diaz-manera@newcastle.ac.uk

Contact information

Type(s)

Principal investigator

Contact name

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Additional identifiers

Clinical Trials Information System (CTIS)

Nil known

Integrated Research Application System (IRAS)
313309

ClinicalTrials.gov (NCT)

Nil known

Protocol serial number

NU-009732, IRAS 313309

Study information

Scientific Title

MYO-Guide: a machine learning approach to the analysis of MRI

Study objectives

The diagnosis of muscle diseases is typically based on clinical examination, blood analysis, muscle biopsy, and/or muscle MRI, which direct genetic diagnosis performed using DNA sequencing. Next-generation sequencing (NGS) has made genetic diagnosis easier and earlier for patients with inherited muscle diseases. However, NGS has limitations, and a tool such as MYO-Guide could help clinicians in the diagnosis process by automatically analyzing the amount of fat present on each muscle MRI using machine learning and suggesting a list of potential diagnoses. The piloted version of MYO-Guide used T1 weighted imaging to score the amount of fat in muscles from zero to four and applied random forest-supervised machine learning to develop an algorithm that could predict the correct diagnosis with 95.7% accuracy. The tool could facilitate the selection of genes to be analyzed or the verification of candidate gene variants identified in panels or exomes, thus speeding up the diagnosis of patients with rare diseases, such as neuromuscular diseases.

Ethics approval required

Old ethics approval format

Ethics approval(s)

Approved 29/04/2022, South West - Central Bristol Research Ethics Committee (Ground Floor, Temple Quay House, 2 The Square, Bristol, BS1 6PN, UK; +44 (0)207 104 8029; centralbristol.rec@hra.nhs.uk), ref: 22/SW/0065

Study design

Observational machine learning using MRI data

Primary study design

Observational

Study type(s)

Diagnostic, Screening

Health condition(s) or problem(s) studied

Neuromuscular diseases

Interventions

The Newcastle University research team will score muscle MRI images already obtained for diagnosis in clinics using the Lamminen-Mercuri scale (Diaz-Manera 2015). The numerical data from these scores will be input into a machine learning algorithm to generate a model that is able to predict a diagnosis. To help clinicians to apply the Lamminen-Mercuri score the research team will develop an automatic segmentation tool using neural network methodology that will recognize and delineate the muscles and provide the Lamminen-Mercuri score of each muscle.

Intervention Type

Other

Primary outcome(s)

1. To develop an artificial intelligence tool using machine learning that can guide the genetic diagnosis of muscle disorders based on the analysis of muscle MRIs.
2. To develop an artificial intelligence tool using a methodology called neural network, which will automatically identify and segment pelvic and leg muscles to quantify the amount of fat present in the skeletal muscles.
3. To collect many muscle MRIs of patients with different genetically confirmed muscles diseases.
4. To score fat replacement of all muscles of the pelvis and legs of the new cohort of patients included in the study.
5. To generate a new version of the MYO-Share platform containing MYO-Guide and the automatic segmentation tool.

The MRI images of muscles from patients who have a neuromuscular disease will be included in this study. The purpose of this image data collection is twofold: 1) to inform the artificial intelligence tool used for diagnosis and 2) to inform the artificial intelligence tool used to automatically segment MRIs. For the diagnosis tool, the anonymised MRI images and patient data will be obtained either via an online platform (MYO-Share) uploaded by NHS sites and health care settings around the world or from data archives and Newcastle University. The automatic segmentation software will be able to identify and delineate each single muscle in the pelvis, thigh, and leg. To build the automatic segmentation algorithm, anonymised MRIs will be obtained from Newcastle University. We will use a neural network approach to identify the muscles on the MRI and quantify the amount of fat present in the muscles. On a first step, we will delineate manually all muscles of the lower limbs using an imaging delineation tool and assign a label of each muscle creating what is known as masks. On a second step, we will use all the masks generated to train a neural network that will automatically delineate muscles on the MRIs. We will test the tool on MRIs already manually delineated and test the accuracy of the tool. We will estimate that we will need a minimum of 200 MRIs to train the automatic segmentation tool, but this will vary depending on the accuracy obtained.

The data obtained from MYO-Share, as well as from Newcastle University, will be used to train a machine-learning model. The number of images needed for each disease will vary according to the homogeneity of fat replacement exhibited by disease type. A greater homogeneity of fat replacement requires fewer MRI images to train the model whereas a greater heterogeneity of fat replacement requires more MRI images. Seventy percent of the images will be used to train the model, 25% will be used to validate the accuracy of the model and 5% will be reserved to test the model with never seen before data. Splitting up the data in these proportions for training and validation is a standard technique employed by data scientists and was used our pilot study (ref Verdu-Diaz 2020). A minimum of 2000 models will be run to determine the one with the best accuracy for prediction purposes.

Key secondary outcome(s))

There are no secondary outcome measures

Completion date

31/12/2025

Eligibility

Key inclusion criteria

This is a data archive study and no patients will be recruited into the study. This study will be using historical muscle MRI scans as well as limited patient data (i.e. age, sex, and genetic diagnosis of muscle disease)

Participant type(s)

Other

Healthy volunteers allowed

No

Age group

All

Sex

All

Key exclusion criteria

This is a data archive study and no patients will be recruited into the study. This study will be using historical muscle MRI scans as well as limited patient data (i.e. age, sex, and genetic diagnosis of muscle disease)

Date of first enrolment

01/09/2021

Date of final enrolment

31/12/2025

Locations

Countries of recruitment

United Kingdom

England

Canada

Chile

Denmark

France

Italy

Korea, South

Spain

Study participating centre

Great Ormond Street Hospital for Children
Great Ormond Street
London
United Kingdom
WC1N 3JH

Study participating centre

University College London Hospitals NHS Foundation Trust
250 Euston Road
London
United Kingdom
NW1 2PG

Study participating centre

Leeds Teaching Hospitals NHS Trust
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United Kingdom
LS9 7TF

Study participating centre

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Study participating centre

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464

Study participating centre

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Study participating centre

Neuromuscular Clinic & Copenhagen Neuromuscular Center

Section 8077

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Righospitalet

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Study participating centre

Instituto de Investigación Hospital 12 de Octubre

Fundacion Investigacion Biomedica

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Study participating centre

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Study participating centre
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Study participating centre
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Study participating centre
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Study participating centre
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Study participating centre
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Study participating centre
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London
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SW17 0QT

Sponsor information

Organisation
Newcastle University

ROR
<https://ror.org/01kj2bm70>

Funder(s)

Funder type
Charity

Funder Name
AFM-Téléthon

Alternative Name(s)
French Muscular Dystrophy Association

Funding Body Type

Private sector organisation

Funding Body Subtype

Associations and societies (private and public)

Location

France

Funder Name

Muscular Dystrophy UK

Alternative Name(s)

Muscular Dystrophy UK London, Muscular Dystrophy Group, Muscular Dystrophy Campaign, MDUK

Funding Body Type

Government organisation

Funding Body Subtype

Trusts, charities, foundations (both public and private)

Location

United Kingdom

Results and Publications

Individual participant data (IPD) sharing plan

The protocol for the MYO-Guide study involves the collection and analysis of MRI images and data for patients with genetic diagnoses. All sites involved in the study must gather and upload their patients' MRI images and data to the MYO-Share platform, and share their anonymized patient MRI images and data with Newcastle University. NHS sites must use a password-protected Excel file to store patient data and upload the MRI images and patient data to MYO-Share. The research team at Newcastle University will collate data into a spreadsheet with no personally identifiable data. Patient data will be removed from the dataset if the patient has requested it or if they have opted out of the National Database for research or planning purposes. The anonymized data from patients, including MRI images, age, sex, and genetic diagnosis, can be used in research studies. The data can be uploaded to MYO-Share, managed by the University of Ottawa, Canada, and used to store MRI scans from NHS sites and other collaborators. The images are automatically anonymized, and no other clinical data is included. Researchers from Newcastle University can view the MRI images shared by all sites, but they cannot be downloaded from MYO-Share. The MYO-Share investigators must follow the rules outlined in the MYO-Share Governance Policy as well as their local and national governance guidelines.

IPD sharing plan summary

Stored in publicly available repository

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
Participant information sheet	Participant information sheet	11/11/2025	11/11/2025	No	Yes
Study website	Study website	11/11/2025	11/11/2025	No	Yes