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

Submission date
11/05/2023

Recruitment status
Recruiting

Registration date
06/06/2023

Overall study status
Ongoing

Last Edited
02/06/2023

Condition category
Musculoskeletal Diseases

[ ] Prospectively registered
[ ] Protocol
[ ] Statistical analysis plan
[ ] Results
[ ] Individual participant data
[ ] 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?
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Contact information

Type(s)
Principal investigator

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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
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WC1N 3JH

Study participating centre
University College London Hospitals NHS Foundation Trust
250 Euston Road
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NW1 2PG

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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Study participating centre
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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