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

MYO-GUIDE: A MACHINE LEARNING APPROACH TO THE ANALYSIS OF MRI

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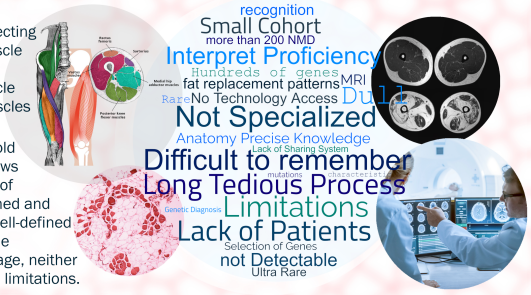
BACKGROUND: Daily Clinic Diagnosis

Neuromuscular diseases (NMDs) are a group of more than 200 inherited muscular disorders affecting more than 6 million individuals worldwide. They typically present with weakness, progressive muscle wasting and often lead to severe disability and premature death.

Fat replacement in muscles is characteristic of NMDs. For many years, clinicians have used muscle MRI on patients with muscle diseases because it easily identifies fat replacement in skeletal muscles (Diaz-Manera 2020).

These patterns help guiding the selection of genes tested using DNA sequencing, currently the gold standard for diagnosing and categorizing muscle disease. Next generation sequencing (NGS) allows reaching a genetic diagnosis earlier and easier thanks to the simultaneous analysis of hundreds of genes. An earlier diagnosis means patients benefits from genetic counseling, access to well defined and tailored care based in the diagnosis, clear definition of expectations about clinical progression, well-defined follow-up strategy and inclusion in natural history studies or clinical trials. However there are some drawbacks as most clinicians are not specialised in identifying muscle disease type from MRI image, neither they have the precised knowledge of body anatomy. Moreover, NGS has several well-documented limitations.



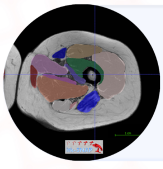
OBJETIVES and METHODS

1. To create an automatic segmentation tool which can delineate muscles of the pelvis, thighs, and legs to aid the automatic quantification of the skeletal muscle fat replacement using Lamminen-Mercuri scale.

2. To create within a new version of the MYO-Share Platform (MYO-Guide V2.0), a machine learning algorithm in order to apply an artificial intelligence for the analysis of the lower limbs muscle MRIs, which alongside the automatic segmentation tool will obtain an automatic quantification of fat replacement and hence an automated prediction of one specific diagnosis from a list of 25 neuromuscular diseases.

Data Collection:

There are 2 sources of data (anonymised):
- Worldwide MRIs shared via MYO-Share platform
- MRIs and other data from own Archive



Segmentation Tool:

(from Own Archive Data source)

On a first step, manually delineation of all muscles of the lower limbs creating *masks*. After, trained by those *masks*, the creation of a *Deep Neural Network – Artificial Intelligence* tool, that will be able to automatically segment muscle regions on any MRI.

Fatty Replacement

(from MYO-Share and Own Archive Data sources)

Creation of an Artificial Intelligence tool which benefits from the previously learned muscle recognition skill, that can automatically quantify the skeletal muscle fat replacement using the Lamminen-Mercury scale on each muscle of the pelvis, thighs and legs.

Genetic Diagnosis of Muscle Disorders Guide MYO-Guide V2.0

On this new version of MYO-Guide investigators and clinicians will be able to upload patient MRIs in Dicom format.

MYO-Guide V2.0 will execute the following automated actions:
- Automatic de-identification of MRIs
- Automatic delineation/segmentation of the muscles
- Automatic fat replacement analysis
- Automatic pattern recognition
- Automatic generation of potential diagnosis

This diagnosing guide will therefore aid the selection of genes to be tested using DNA sequencing. The outcome of this means an earlier and faster diagnosis with the many benefits that it brings for the patients.

To this extent, a long and tedious process for the clinicians which required them to be specialized in identifying muscle disease from MRI and to remember the many different patterns of muscle involvement for each neuromuscular disease gets fully automated with no need of access to any specific technology but internet connection.

Furthermore, this version will expand the number of diseases that MYO-Guide could predict from 10 to 25.

RESULTS

In proof of concept, we created a machine-learning algorithm, MYO-Guide, which predicted muscle disease diagnosis with an accuracy significantly higher when compared with the diagnoses obtained by four clinical experts in muscle MRI analysis. (Verdu-Diaz, 2020)

For this purpose, 976 muscle MRIs were collected focusing on pelvis, thigh and leg muscles of patients with a confirmed diagnosis of 10 different muscular diseases. Fat replacement in every muscle was quantified using the Lamminen-Mercuri scale to score the amount of fat in muscles. 70% of the images were used to train the model: 25% to validate its accuracy; and 5% to test the model with never seen before data. 2000 different algorithms were generated and selected the one with higher accuracy.

- 95.7% accuracy
- 92.1% sensitivity
- 99.4% specifity

CONCLUSION

Our present study aims to create an improved version of MYO-Guide to analyse a larger number of NMDs; supporting an easier genetic diagnosis process in the daily clinics (helping to identify characteristic patterns that can guide genetic testing), hence an earlier and faster diagnosis of patients. This will have a big clear impact in the medical and patient communities.

Furthermore, this platform will remain as an open access portal and one of the largest cohort of patients with NMDs internationally, free and publicly available as an anonymised MRI sharing platform, open atlas and equipped with the Artificial Intelligence and automatic segmentation diagnosis guide technology that promotes robust collaboration research and educational tool for investigators.

more NMDs analysis | automated diagnosis | free and open access

Verdú-Díaz J, Alonso-Pérez J, Nuñez-Peralta C, Tasca G, Vissing J, Straub V, Fernández-Torrón R, Llauger J, Illa I, Díaz-Manera J. Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology. 2020 Mar 10;94(10):e1094-e1102.