Muscular MRI-based algorithm to differentiate inherited myopathies presenting with spinal rigidity

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Abstract

Objectives

Inherited myopathies are major causes of muscle atrophy and are often characterized by
rigid spine syndrome, a clinical feature designating patients with early spinal
contractures. We aim to present a decision algorithm based on muscular whole body
magnetic resonance imaging (mWB-MRI) as a unique tool to orientate the diagnosis of
each inherited myopathy long before the genetically confirmed diagnosis.

Methods

This multicentre retrospective study enrolled 79 patients from referral centres in France,
Brazil and Chile. The patients underwent 1.5-T or 3-T mWB-MRI. The protocol
comprised STIR and T1 sequences in axial and coronal planes, from head to toe. All
images were analyzed manually by multiple raters. Fatty muscle replacement was
evaluated on mWB-MRI using both the Mercuri scale and statistical comparison based
on the percentage of affected muscle.

Results

Between February 2005 and December 2015, 76 patients with genetically confirmed
inherited myopathy were included. They were affected by Pompe disease or harbored
mutations in RYR1, Collagen VI, LMNA, SEPN1, LAMA2 and MYH7 genes. Each
myopathy had a specific pattern of affected muscles recognizable on mWB-MRI. This
allowed us to create a novel decision algorithm for patients with rigid spine syndrome by
segregating these signs. This algorithm was validated by five external evaluators on a
cohort of seven patients with a diagnostic accuracy of 94.3% compared with the genetic
diagnosis.
Conclusion

We provide a novel decision algorithm based on muscle fat replacement graded on mWB-MRI that allows diagnosis and differentiation of inherited myopathies presenting with spinal rigidity.

Key Points

• Inherited myopathies are rare, diagnosis is challenging and genetic tests require specialized centres and often take years.

• Inherited myopathies are often characterized by spinal rigidity.

• Whole body magnetic resonance imaging is a unique tool to orientate the diagnosis of each inherited myopathy presenting with spinal rigidity.

• Each inherited myopathy in this study has a specific pattern of affected muscles that orientate diagnosis.

• A novel MRI-based algorithm, usable by every radiologist, can help the early diagnosis of these myopathies.

Keywords

Whole body imaging  Myopathies, structural, congenital  Muscular diseases
Muscular dystrophies  Spinal curvatures

Abbreviations

MAM

Mean of percentages of affected muscle

mWB-MRI

Muscular whole body magnetic resonance imaging

RSS

Rigid spine syndrome
Electronic supplementary material

The online version of this article (https://doi.org/10.1007/s00330-018-5472-5) contains supplementary material, which is available to authorized users.

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Notes

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Compliance with ethical standards

Guarantor

The scientific guarantor of this publication is Mickael Tordjman

Conflict of interest

The authors of this manuscript declare no relationships with any companies whose products or services may be related to the subject matter of the article.

Statistics and biometry

Nathaniel Bern kindly provided statistical advice for this manuscript. One of the authors has significant statistical expertise: Moustafa Biyoukar.
Informed consent

Written informed consent was obtained from all subjects (patients) in this study.

Ethical approval

Institutional review board approval was obtained.

Methodology

- retrospective
- diagnostic or prognostic study/observational
- multicentre study

Supplementary material

330_2018_5472_MOESM1_ESM.docx (16.6 mb)
ESM 1 (DOCX 17032 kb)

References

   CrossRef (https://doi.org/10.1016/0966-8966(91)90039-U)

   CrossRef (https://doi.org/10.1016/j.nmd.2013.11.003)
3.
PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC5258110)
Google Scholar (http://scholar.google.com/scholar_lookup?title=Dia.pngstic%20approach%20to%20congenital%20muscular%20dystrophies&author=CG.%20B%C3%B6nnemann%20et%20al
4.
CrossRef (https://doi.org/10.1086/342719)
PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC378532)
5.
CrossRef (https://doi.org/10.1016/j.nmd.2012.08.003)
PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3930922)
6.
CrossRef (https://doi.org/10.1007/s00330-010-1799-2)
7.

CrossRef (https://doi.org/10.1002/mus.24634)


CrossRef (https://doi.org/10.1002/mus.25018)


CrossRef (https://doi.org/10.1016/j.nmd.2008.01.009)


CrossRef (https://doi.org/10.1053/ejpn.2002.0617)

CrossRef (https://doi.org/10.1097/WCO.0000000000000364)

PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1644828)

CrossRef (https://doi.org/10.1212/01.wnl.0000251268.41188.04)

15. Wu S, Ibarra MCA, Malicdan MCV et al (2006) Central core disease is due to RYR1 mutations in more than 90% of patients. Brain 129:1470–1480
CrossRef (https://doi.org/10.1093/brain/awl077)
Google Scholar (http://scholar.google.com/scholar_lookup?title=Central%20core%20disease%20is%20due%20to%20RYR1%20mutations%20in%20more%20than%2090%25%20of%20patients&author=S.%20Wu&author=M.%20Ibarra&author=MCV.%20Malicdan&journal=Brain&volume=129&pages=1470-1480&publication_year=2006)

CrossRef (https://doi.org/10.1002/ana.22119)


CrossRef (https://doi.org/10.1093/hmg/ddi025)


CrossRef (https://doi.org/10.1002/ana.21417)


CrossRef (https://doi.org/10.1016/j.jns.2010.09.011)


CrossRef (https://doi.org/10.1038/ng713)

CrossRef (https://doi.org/10.1016/j.jocn.2015.04.016)


CrossRef (https://doi.org/10.1002/ana.10693)


CrossRef (https://doi.org/10.1016/j.nmd.2012.06.005)


Google Scholar (http://scholar.google.com/scholar_lookup?
title=Selective%3andomuscle%20involvement%20in%20magnetic%20resonance%20imaging%20in%20autosomal%20dominant%20Emery-
Dreifuss%20muscular%20dystrophy&author=E.%20Mercuri&author=S.%20Counsell&author=J.%20Allsop&journal=Neuropediatrics&volume=33&page=10-
14&publication_year=2002)


CrossRef (https://doi.org/10.1037/h0031619)

Google Scholar (http://scholar.google.com/scholar_lookup?
title=Measuring%20nominal%20scale%20agreement%20among%20many%20raters&author=JL.%20Fleiss&journal=Psychol%20Bull&volume=76&pages=378-
382&publication_year=1971)


CrossRef (https://doi.org/10.1177/00131644600200104)

Google Scholar (http://scholar.google.com/scholar_lookup?
title=A%20coefficient%20of%20agreement%20for%20nominal%20scales&author=J.%20Cohen&journal=Educ%20Psychol%20Meas&volume=20&pages=37-
46&publication_year=1960)


CrossRef (https://doi.org/10.1080/01621459.1967.10482916)

Google Scholar (http://scholar.google.com/scholar_lookup?
title=On%20the%20Kolmogorov-
Smirnov%20test%20for%20normality%20with%20mean%20and%20variance%20unknown&author=HW.%20Lilliefors&journal=J%20Am%20Stat%20Assoc&vol-
ume=62&pages=399-402&publication_year=1967)

28. Mann HB, Whitney DR (1947) On a test of whether one of two random variables is stochastically larger than the other. Ann Math Stat 50–60

Google Scholar (https://scholar.google.com/scholar?q=Mann%20HB%20%20Whitney%20DR%20%281947%29%20On%20a%20test%20whether%20one%20of%20two%20random%20variables%20is%20stochastically%20larger%20than%20the%20other.%20Ann%20Math%20Stat%2050%20%E2%80%9360)


CrossRef (https://doi.org/10.2307/3001968)

Google Scholar (http://scholar.google.com/scholar_lookup?

CrossRef (https://doi.org/10.1002/ana.21846)
Google Scholar (http://scholar.google.com/scholar_lookup?
title=Muscle%20magnetic%2oresonance%2oimaging%2oinvolvement%2o
muscular%2odystrophies%2owith%2origidity%2othe%2ospine&author=E.


CrossRef (https://doi.org/10.1016/j.nmd.2004.08.006)
Google Scholar (http://scholar.google.com/scholar_lookup?
title=Magnetic%2oresonance%2oimaging%2of%2omuscle%2oin%20congenital%2omyopathies%2oaassociated%2owith%2oRYR1%20mutations&author=H.%20Jungbluth&author=MR.%20Davis&author=C.%20M%20C%20Müller&journal=Neur
muscul%20Disord&volume=14&pages=785-790&publication_year=2004)


CrossRef (https://doi.org/10.1016/j.nmd.2015.10.001)
Google Scholar (http://scholar.google.com/scholar_lookup?
title=Muscle%20imaging%2oin%20muscle%20dystrophies%2oproduced%2oby%2
omutations%2oin%2othe%2oEMD%2oand%2oLMNA%2ogenes&author=J.%2
0D%20D–
Manera&author=A.%20Alejaldre&author=L.%20Gonz%C3%A1lez&journal=Neur
muscul%20Disord&volume=26&pages=33-40&publication_year=2016)


CrossRef (https://doi.org/10.1016/j.nmd.2011.06.748)
Google Scholar (http://scholar.google.com/scholar_lookup?title=Whole-body%2omuscle%20MRI%2oin%2opatients%2osuffering%2ofrom%2olat
ome%2onset%2oPompe%2odisease%3A%2oinvolvement%2opatterns&author=RY.
%20Carlier&author=P.%20Laforet&author=C.%20Wary&journal=Neuromuscul%2
0Disord&volume=21&pages=791-799&publication_year=2011)


CrossRef (https://doi.org/10.1016/j.nmd.2006.09.013)
cmd=Retrieve&db=PubMed&dopt=Abstract&list_uids=17134899)
Google Scholar (http://scholar.google.com/scholar_lookup?


CrossRef (https://doi.org/10.1016/j.nmd.2012.12.009)
cmd=Retrieve&db=PubMed&dopt=Abstract&list_uids=23394783)
PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3594745)
Google Scholar (http://scholar.google.com/scholar_lookup?


CrossRef (https://doi.org/10.1016/0960-8966(94)90028-0)
cmd=Retrieve&db=PubMed&dopt=Abstract&list_uids=7919974)
Google Scholar (http://scholar.google.com/scholar_lookup?


CrossRef (https://doi.org/10.1016/0960-8966(95)90039-X)

CrossRef (https://doi.org/10.1038/ejhumgenet2014.169)


CrossRef (https://doi.org/10.1093/brain awp236)
PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4038491)


CrossRef (https://doi.org/10.1016/j.yexcr.2007.03.028)
PubMedCentral (http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2964355)

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