Article:
Howard, J., Bekker, H.L., Mcdermott, C.J. orcid.org/0000-0002-1269-9053 et al. (1 more author) (2024) A report of resources used by clinicians in the UK to support motor neuron disease genomic testing. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration. ISSN 2167-8421

https://doi.org/10.1080/21678421.2023.2299204

Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration

ISSN: (Print) (Online) Journal homepage: https://www.tandfonline.com/loi/iafd20

A report of resources used by clinicians in the UK to support motor neuron disease genomic testing

Jade Howard, Hilary L. Bekker, Christopher J. Mcdermott & Alisdair Mcneill

To cite this article: Jade Howard, Hilary L. Bekker, Christopher J. Mcdermott & Alisdair Mcneill (02 Jan 2024): A report of resources used by clinicians in the UK to support motor neuron disease genomic testing, Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, DOI: 10.1080/21678421.2023.2299204

To link to this article: https://doi.org/10.1080/21678421.2023.2299204

Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2024; 0: 1–3

BRIEF REPORT

A report of resources used by clinicians in the UK to support motor neuron disease genomic testing

JADE HOWARD[1], HILARY L. BEKKER[2], CHRISTOPHER J. MCDERMOTT[1,3] & ALISDAIR MCNEILL[1,4]

[1]Division of Neuroscience and Neuroscience Institute, The University of Sheffield, Sheffield, UK, [2]Leeds Unit for Complex Intervention Development, The University of Leeds, Leeds, UK, [3]Academic Directorate of Neuroscience, Royal Hallamshire Hospital, Sheffield, UK and [4]Sheffield Clinical Genetics Service, Sheffield Children's Hospital NHS Foundation Trust, Sheffield, UK

Abstract

Genetic testing is a key decision-making point for people with motor neuron disease (MND); to establish eligibility for clinical trials, better understand the cause of their condition, and confirm the potential risk to relatives, who may be able to access predictive testing. Given the wide-reaching implications of MND genetic and predictive testing, it is essential that families are given adequate information, and that staff are provided with appropriate training. In this report we overview the information resources available to people with MND and family members around genetic testing, and the educational and training resources available to staff, based on information obtained through a freedom of information request to UK-based NHS Trusts. MND Association resources were most commonly used in information sharing, though we highlight distinctions between neurology and genetics centers. No respondents identified comprehensive training around MND genetic testing. We conclude with practice implications and priorities for the development of resources and training.

Keywords: *Genetics, training, patient information*

Introduction

Genetic testing is a key decision-making point for people with MND (pwMND), to establish their eligibility for clinical trials, better understand the cause of their condition and the potential risk to relatives. Family members of gene carriers may be able to access predictive testing to determine whether they have an increased chance of developing MND. In both cases, these are complex and multifaceted decisions with a range of implications for the individual and family. It is essential that families are provided with adequate information and resources to make informed decisions about genetic testing, and that staff are given appropriate training (1). Here, we survey resources available to clinicians in the United Kingdom to support MND genetic testing.

Methods

Ethical approval was granted by the University of Sheffield (056228). A freedom of information (FOI) request was submitted to NHS Trusts hosting MND care centers ($n = 23$) or regional clinical genetics centers ($n = 23$). The request sought confirmation as to whether the service offered MND genetic testing and predictive testing; what leaflets and websites are provided or signposted to pwMND and relatives on MND genetic testing and predictive testing; and what letters, leaflets or websites are provided to facilitate family communication. Trusts were also asked what educational resources or training they have to support staff with MND genetic testing and predictive testing. The request was sent in August 2023, with responses received within 20 working days.

Correspondence: Alisdair McNeill, Division of Neuroscience and Neuroscience Institute, The University of Sheffield, 385a Glossop Road, Sheffield S10 2HQ, UK. E-mail: a.mcneill@:sheffield.ac.uk

(Received 5 December 2023; accepted 18 December 2023)

ISSN 2167-8421 print/ISSN 2167-9223 online
DOI: 10.1080/21678421.2023.2299204

Table 1. Resources for people with MND and family members around genetic testing.

	What leaflets and websites do you provide (or signpost) to people with MND on genetic testing?		What leaflets and websites do you provide (or signpost) to at-risk relatives of people with MND on predictive testing?		What letters, leaflets or websites do you provide to people having MND genetic testing or predictive testing to facilitate family communication?	
	MND/ Neurology centers	Genetics centers	MND/ Neurology centers	Genetics centers	MND/ Neurology centers	Genetics centers
MND Association resources	15	7	13	8	9	1
MND Scotland	0	2	0	2	0	1
US based charities/ patient organizations (ALS.org; MDA.org)	0		1	0	0	0
Locally produced leaflets	3	1	2	2	3	0
Genereviews	0	2	0	2	0	0
Genetic alliance	0	0	1	0	0	0
Healthtalk.org section on inherited MND	1	0	1	0	1	0
Genetic testing decision tool Mnddecisiontools.com	1	0	1	0	1	0
NHS care centre/ department websites	2	0	2	0	1	1
Other NHS resources (leaflet on WGS)	1	1	0	1	0	1
Tailored summary/ clinic letter	0	0	0	1	2	4
Tailored family letter/ To Whom It May Concern letter	0	0	0	0	0	7
Support groups	0	1	0	1	0	1
Signpost/ referral (to genetics/ neurogenetics/GP)	2	0	3	1	5	2
Dementia resources (GENFI; Alzheimer's society; Dementia UK)	0	3	0	3	0	2
Referral to research groups/ networks/ related resources	0	0	1	1	0	1
Clinical appointments (to support family communication)	0	0	0	0	0	1
None	1	2	2	1	2	1
N/A	0	2	0	2	0	2

Results

A total of 33 responses were received (from 17/23 [74%] MND care centers and 16/23 [69%] regional genetics centers). There were differences in resources reported by MND care centers and regional genetics centers (Table 1). The most common information resources provided to pwMND, or relatives considering predictive testing, were those produced by the UK MND Association. These included webpages and information sheets on inherited MND, genetic testing and insurance. Locally produced resources covered general information (e.g., around inheritance patterns and genetic testing in neurodegenerative diseases) and were not MND-specific. The most common resources used to facilitate family communication by MND clinics were MND Association resources or referral to clinical genetics, while genetics clinicians supported information sharing with bespoke clinic letters. Three trusts reported no resources to share with pwMND around genetic testing, relatives around predictive testing, or to facilitate family communication respectively.

Ten trusts reported having no educational resources or training to support staff with MND genetic testing, increasing to twelve for predictive testing (with an even split between neurology and genetics centers). No respondents identified comprehensive training or educational resources for MND genetics/genetic testing (Table 2).

Discussion and conclusions

Most services signposted pwMND and family members to national MND charities for information around MND genetic testing and predictive testing; almost no services reported training staff to support their delivery of information about genetic or predictive testing. Although MND professionals often access continuing professional development training on genomics (e.g. Genomics Education Programme), these courses do not cover MND-specific issues, e.g., oligogenic inheritance and implications for treatment effectiveness.

Given these findings, it seems unlikely pwMND, and family members, are making informed decisions about testing with professionals managing their MND (2). This highlights the need for services to provide accessible, written information to support pwMND and family members to understand the consequences of genetic and

Table 2. Educational resources and training for staff around genetic testing.

	What educational resources or training do you have to support staff with MND genetic testing?		What educational resources or training do you have to support staff with MND predictive testing?	
	MND/ Neurology centers	Genetics centers	MND/ Neurology centers	Genetics centers
MDT/ peer/ case discussion and support	2	2	2	2
Via genetics service	1	1	2	1
Study days and Continuing Professional Development (CPD)	0	3	0	3
Conferences, symposiums and network meetings	1	3	2	3
Clinical supervision	0	1	0	1
Shadowing and observation opportunities	1	0	1	2
NHS genomics training (Genomics Education Programme; NHS Genomic Medicine materials)	2	2	1	0
Local training (annual training day; clinical support and letter sign off)	0	1	0	1
Decision support resources (Skilled helper decision counseling; decisional balance sheet)	0	0	1	1
Anxiety and depression assessment tools	0	0	1	1
MND Association virtual faculty / professional resources	3	1	1	0
Other MND Association resources	4	0	2	0
Journal access	0	2	0	0
Guidelines and policy documents (e.g., national genomic test directory, Huntington's disease adapted protocol)	1	1	1	4
Locally developed pathway	0	0	1	1
None	5	5	6	6
N/A	0	2	1	2
Not answered	0	1	0	1

predictive testing (3,4), and suggests a role for developing patient decision aids (PDAs) to support shared decision making about treatment and testing with MND preofessionals (5).

Whilst this report describes snapshot of information provision, it underscores that there is a dearth of resources for pwMND, relatives and staff around genetic testing. The current public-facing and staff resources are a starting point, yet a standardized approach is needed to ensure equitable access to tailored information and training. The research landscape around MND genetic testing, predictive testing and personalized medicine is rapidly increasing (6). Staff require upskilling to support proactively the conversations around these complex genetics of MND, and their consequences for pwMND and their family members.

Declaration of interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of this article.

Funding

This work was supported by the motor neuron disease association under Grant McNeill/Oct21/967-794 Motor Neurone Disease Association.

ORCID

Christopher J. Mcdermott http://orcid.org/0000-0002-1269-9053

References

1. Crook A, McEwen A. Genetic counselling and testing for neurodegenerative disorders using a proposed standard of practice for ALS/MND: diagnostic testing comes first. Eur J Hum Genet. 2022;30:394–5.
2. NICE. Shared decision making, NICE guideline. June 17, 2021. Available at: https://www.nice.org.uk/guidance/ng197. Accessed November 27, 2023.
3. Salmon K, Ross JP, Bertone V, Gobbo M, Anoja N, Karamchandani J, et al. The value of testing for ATXN2 intermediate repeat expansions in routine clinical practice for amyotrophic lateral sclerosis. Eur J Hum Genet. 2022; 30:1205–7.
4. Crook A, Jacobs C, Newton-John T, McEwen A. Genetic counseling and diagnostic genetic testing for familial amyotrophic lateral sclerosis and/or frontotemporal dementia: a qualitative study of client experiences. J Genet Couns. 2022;31:1206–18.
5. Bekker HL, Winterbottom AE, Gavaruzzi T, Finderup J, Mooney A. Decision aids to assist patients and professionals in choosing the right treatment for kidney failure. Clin Kidney J. 2023;16:i20–i38.
6. De Oliveira HM, Soma A, Baker MR, Turner MR, Talbot K, Williams TL. A survey of current practice in genetic testing in amyotrophic lateral sclerosis in the UK and Republic of Ireland: implications for future planning. Amyotroph Lateral Scler Frontotemporal Degener. 2023; 24:405–13.