Supplementary material

Methods

Survey

A cross-sectional questionnaire survey was delivered on-line via *qualtrics,* between January 2023 - 1st May 2023. We followed the consensus-based checklist for reporting of survey studies (CROSS). Ethical approval was granted by a UK NHS Research Ethics Committee (22/SW/0047) and the University of Sheffield (050846). The study questionnaire was developed by the authors to capture current practice for MND WGS. Items were informed by the Medical Student Undergraduate curriculum from the British Society of Genomic Medicine, prior research, and current policy. In the UK, guidance on consent and confidentiality in relation to genomic medicine is given by the Joint Committee on Genomics in Medicine document *Consent and Confidentiality* in Genomic Medicine (2019). Genomic variant interpretation follows the American College of Medical Genetics criteria (sequence variants v3.0). Criteria for which patients can access genome sequencing are defined in the National Genomic Test directory. To request genome sequencing clinicians must complete a record of discussion form (in conjunction with the patient or consultee) and then a test order form to activate the genome sequencing test with the laboratory.

Consultant clinical geneticists, and genetic counsellors, were invited via the UK Predictive Genetic Testing Consortium email list, and contact with Lead Clinicians at each Regional Genetics Clinic. Consultant neurologists with a special interest in MND, and MND specialist nursing staff, were recruited via the MND UK Clinical Studies Group (CSG), and email contact with Lead Clinicians at each of the UK MND Care centres. Consultant neurologists and neurology trainees without a special interest in MND were recruited by email contact with Lead Clinicians in Neurology departments without an MND care centre.

The questionnaire was pilot tested with 2 Consultant Neurologists, 2 Consultant Clinical Geneticists and a Genetic Counsellor for content validity and item clarity.

Items assessed: Perceived awareness of UK genomic testing guidelines and criteria (3 questions), Self rated confidence in genetic counselling skills (10 question). Knowledge on predictive testing (3 questions). Self-rated confidence in clinical procedures to deliver WGS (3 questions). Responses were recorded using a 5-point Likert scale. Resources needed to support services offering MND genetic testing were enquired about using free text.

Statistical analyses

Scores on individual survey items were compared using a Wilcoxon-signed rank test. Proportions were compared using a chi-squared test. Significance was taken at the 5% level. All statistical analyses were performed in SPSS. Likert responses were compared between groups using a Wilcoxon-signed rank test. Hierarchical clustering was performed using

Clustergrammer (https://maayanlab.cloud/clustergrammer/). Free text responses were analysed using a framework analysis approach.

Framework analysis of free text survey responses

The analysis of the free text comments was based on a framework analysis approach. This was selected as it is a pattern-based approach to thematic analysis through which the data are presented in a framework. Themes and subthemes are presented in columns whilst cases are presented in the rows, which allows for comparison between and within cases, whilst maintaining a focus on the data.

The framework analysis involved a 5-stage process, starting with familiarisation through repeated reading of and immersion in the data, gaining an overview of the content and recording initial ideas and topics of interest. At this stage, the data were read as part of each survey response to maintain the context of each extract. Early notes and ideas on topics were then refined and expanded as the data were re-read to construct an initial thematic framework of themes and sub-themes. In the next phase, the data were indexed and sorted into this coding framework. Here, data were extracted into NVivo for ease of coding. This was carried out in conjunction with the fourth stage, where extracts were reviewed, and the framework was refined. This was an iterative and comparative process which involved looking at the data coded within and between each theme, re-coding, collapsing, subsuming, and renaming codes where appropriate. The fifth stage involved summarising and displaying the data, with the thematic framework used to develop the framework representing the key themes and subthemes in the data. Given the focus of the study, this was not developed into a more conceptual analysis. An analytic log was kept throughout this process with reflections, decisions, thoughts and ideas.

Given the large sample size of the survey and the significant number of participants who did not submit free text comments, clinician groups have been used as the cases displayed in the rows of the framework, as opposed to each participant individually. This was aligned with the comparative focus of the survey analysis which looks at patterns between clinician groups. The framework presented displays prominent themes but does not include all comments for relevance. Data are presented as submitted.

Data Availability

Anonymised data is available from the authors on reasonable request.