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Short report

Survey of service needs to embed genome sequencing for motor neuron disease in neurology in the English National Health Service

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ABSTRACT

All people with motor neuron disease (pwMND) in England are eligible for genome sequencing (GS), with panel-based testing. With the advent of genetically targeted MND treatments, and increasing demand for GS, it is important that clinicians have the knowledge and skills to support pwMND in making informed decisions around GS. We undertook an online survey of clinical genomic knowledge and genetic counselling skills in English clinicians who see pwMND. There were 245 respondents to the survey (160 neurology clinicians and 85 genetic clinicians). Neurology clinicians reported multiple, overlapping barriers to offering pwMND GS. Lack of time to discuss GS in clinic and lack of training in genetics were reported. Neurology clinicians scored significantly less well on self-rated genomic knowledge and genetic counselling skills than genetic clinicians. The majority of neurology clinicians reported that they do not have adequate educational or patient information resources to support GS discussions. We identify low levels of genomic knowledge and skills in the neurology workforce. This may impede access to GS and precision medicine for pwMND.

INTRODUCTION

Within the English National Health Service (NHS), all people with motor neuron disease (pwMND) are eligible for genome sequencing (GS),[1] with panel-based reporting. In 20%–30% of apparently sporadic MND, and 60%–70% of familial MND, a potentially causal monogenic variant can be identified.[2 3] As genomic technology advances, more pwMND will be found to have a monogenic cause, leading to an increased demand for testing. GS for MND is delivered by specialist clinical genetics and MND services, who have expertise in supporting people to make decisions about GS for life-limiting conditions with multiple-cause aetiology. In the English NHS, neurology clinics are staffed by consultant neurologists, neurology specialist trainees (postgraduate doctors training to consultant level) and specialist nursing staff. Clinical genetic clinics are staffed by consultants in clinical genetics (a medical doctor trained in clinical and genomic diagnosis of genetic conditions) and genetic counsellors (a non-medical specialist trained to help people understand, and act on, their genomic test result). In the English NHS, most neurology clinics are based in separate institutions from the genetic services.

Key to the NHS 5-year Genomic Medicine strategy is the embedding of GS in mainstream medicine to facilitate the personalisation of care.[4] Currently, there are no clinical patterns to make a judgement about whether a pwMND is likely to have a monogenic cause.[2] The genomic basis of MND, and implications for treatment, is complicated.[3] Variants in more than one gene can contribute to disease in an individual, and there can be variability in age of onset and clinical manifestations (eg, MND or frontotemporal dementia) within a family.[5 6] pwMND will require information about MND genetics, the implications of GS test results for management of MND and the consequences of results for family members.[7] It is unclear what health professionals need to embed GS in current practice and support shared decision making about testing and treatment for pwMND.

We undertook a survey of the genomic knowledge and skills of health professionals in the English NHS who manage pwMND. This study is part of a project to develop a patient decision aid supporting pwMND to make decisions to have GS within neurology services. This project draws on the Medical Research Council (MRC) complex intervention development framework to guide the research studies needed to inform the development of this complex intervention (phase 1). Bekker's Making Informed Decisions Individually and Together framework[8] is used to provide the theoretical scaffolding to developing a decision aid for implementation within healthcare systems that represent the goals, needs and experiences of the different people involved in making GS decisions (see online supplemental figure 1).[9] The research objectives are to (a) describe current practice for GS across England and (b) identify resource needs for health professionals to integrate GS within their service.

MATERIALS AND METHODS

A cross-sectional questionnaire survey, to assess genomic knowledge and skills, was delivered via *qualtrics*, between January 2023 and 1 May 2023. We followed the consensus-based checklist for reporting of survey studies. Full methods are online: supplemental methods.

RESULTS

There were 245/268 completed surveys, including 160 neurology clinicians (106 consultants, 26 specialty registrars and 28 MND nurses) and 85 clinical genetic clinicians (20 consultants in clinical genetics and 65 genetic counsellors) (online supplemental table 1). The qualitative responses from the free text sections were categorised under two themes: (1) current practice and barriers to GS and (2) professional upskilling, patient resources

and service needs for future GS implementation (online supplemental figure 2). The survey's quantitative responses are synthesised under the headings below.

In neurology clinics, most MND genetic testing discussions are undertaken by consultant neurologists

A variable proportion of neurology clinicians reported having been involved in arranging GS for pwMND (63% of consultant neurologists, 83% of neurology trainees and 57% of MND specialist nurses). Of these clinicians, the majority of neurology consultants had both requested GS and discussed results with pwMND, while the majority of MND specialist nurses had only requested testing (online supplemental figure 3). The majority of neurology clinicians would refer to clinical genetics for further discussion of results if requested by pwMND, but only a minority discuss the possibility of predictive testing for unaffected relatives (online supplemental table 2). Neurology teams reported multiple, overlapping barriers to GS (online supplemental figure 4). Lack of time to discuss genomic testing (49%), paperwork (47%) and timescale to get results (37%) were the barriers to offering GS most frequently reported by consultant neurologists.

Neurology clinicians report low levels of familiarity with genetic testing guidelines and criteria

The majority of consultant clinical geneticists and genetic counsellors rated themselves as 'fairly' or 'very' familiar with each genetic testing guidelines question (online supplemental table 3). Only a minority of neurology clinicians rated themselves as

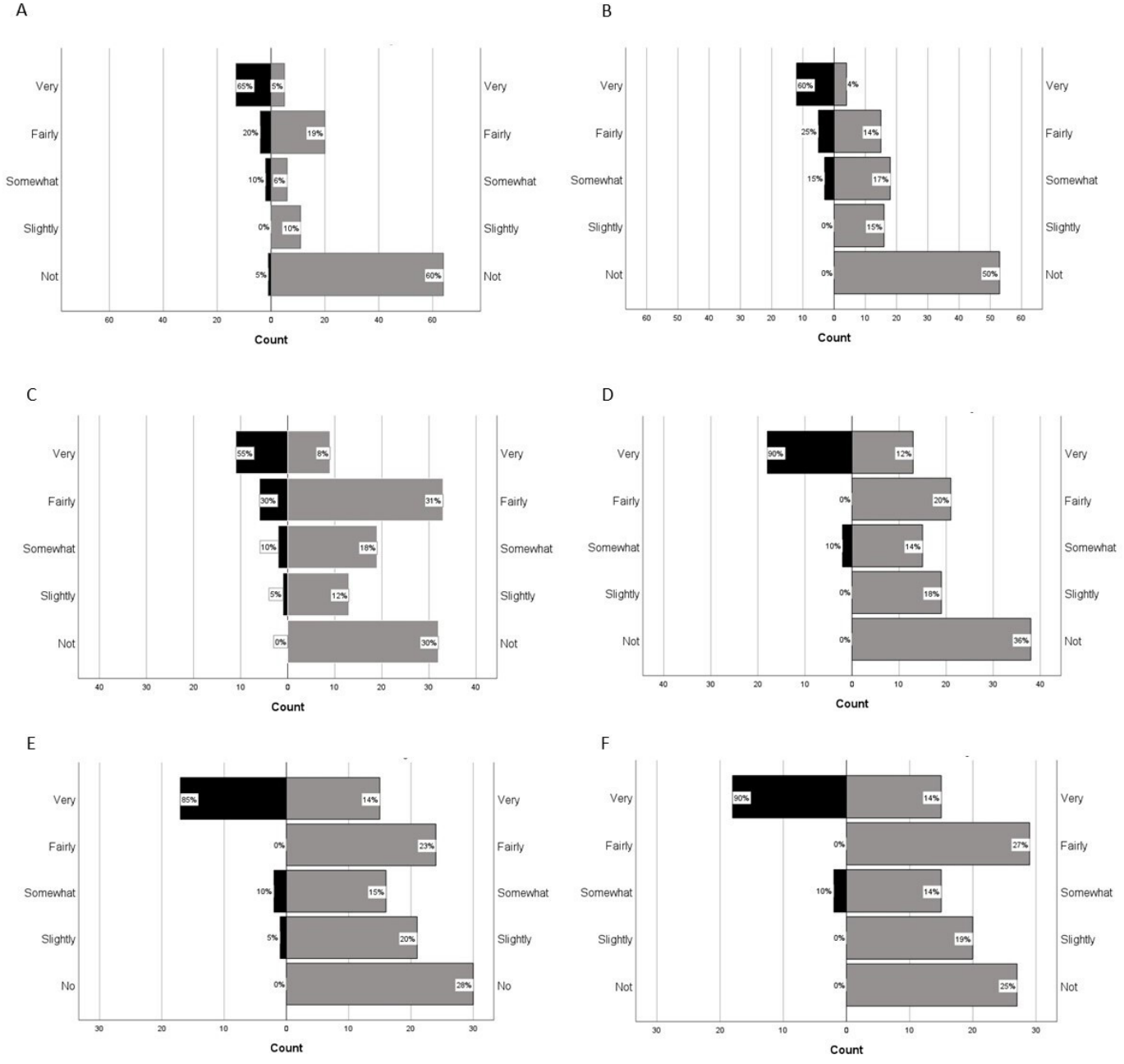

Figure 1 Self-reported genomic knowledge and understanding of predictive testing process for consultant neurologists. Pyramid blots illustrate consultant neurologists' (grey) and consultant geneticists' (black) responses on the 5-point Likert scale. (A) Knowledge of American College of Medical Genetics criteria. (B) Knowledge of Joint Committee on Genomics in Medicine statement on consent and confidentiality. (C) Knowledge of test directory. (D) Understanding of predictive testing process. (E) Understanding of implications of predictive test results. (F) Understanding of reasons for predictive testing.

'fairly' or 'very' familiar with the genomic test directory, American College of Medical Genetics Criteria or Joint Committee on Genomics in Medicine consent and confidentiality guidance (online supplemental table 3). A Wilcoxon-signed rank test demonstrated that neurology clinicians scored significantly lower in each item than genetic clinicians (figure 1, online supplemental table 4).

Neurology clinicians report low confidence in genetic counselling skills

The majority of consultant clinical geneticists and genetic counsellors rated themselves as 'fairly' or 'very' familiar with each genetic counselling skills question (online supplemental table 3). Only a relatively small proportion of neurology clinicians were fairly/very confident in explaining a variant of uncertain significance, oligogenic inheritance or variable clinical expression. In addition, only a small proportion reported being fairly/very confident in undertaking the clinical procedures to request GS of completing the 'Record of Discussion' form, interpreting a genomic laboratory report and communicating results to families (online supplemental table 3). A Wilcoxon-signed rank test demonstrated that neurology clinicians scored significantly lower in each item than genetic clinicians (figure 2, online supplemental table 4).

Genetic counselling training was associated with increased confidence in embedding GS in practice

We sought to understand the effect of genetic counselling training on neurology clinicians' knowledge and skills. We defined genetic counselling training for mainstream clinicians as courses such as continuing professional development courses, Master's degree programmes or a research doctorate. A higher

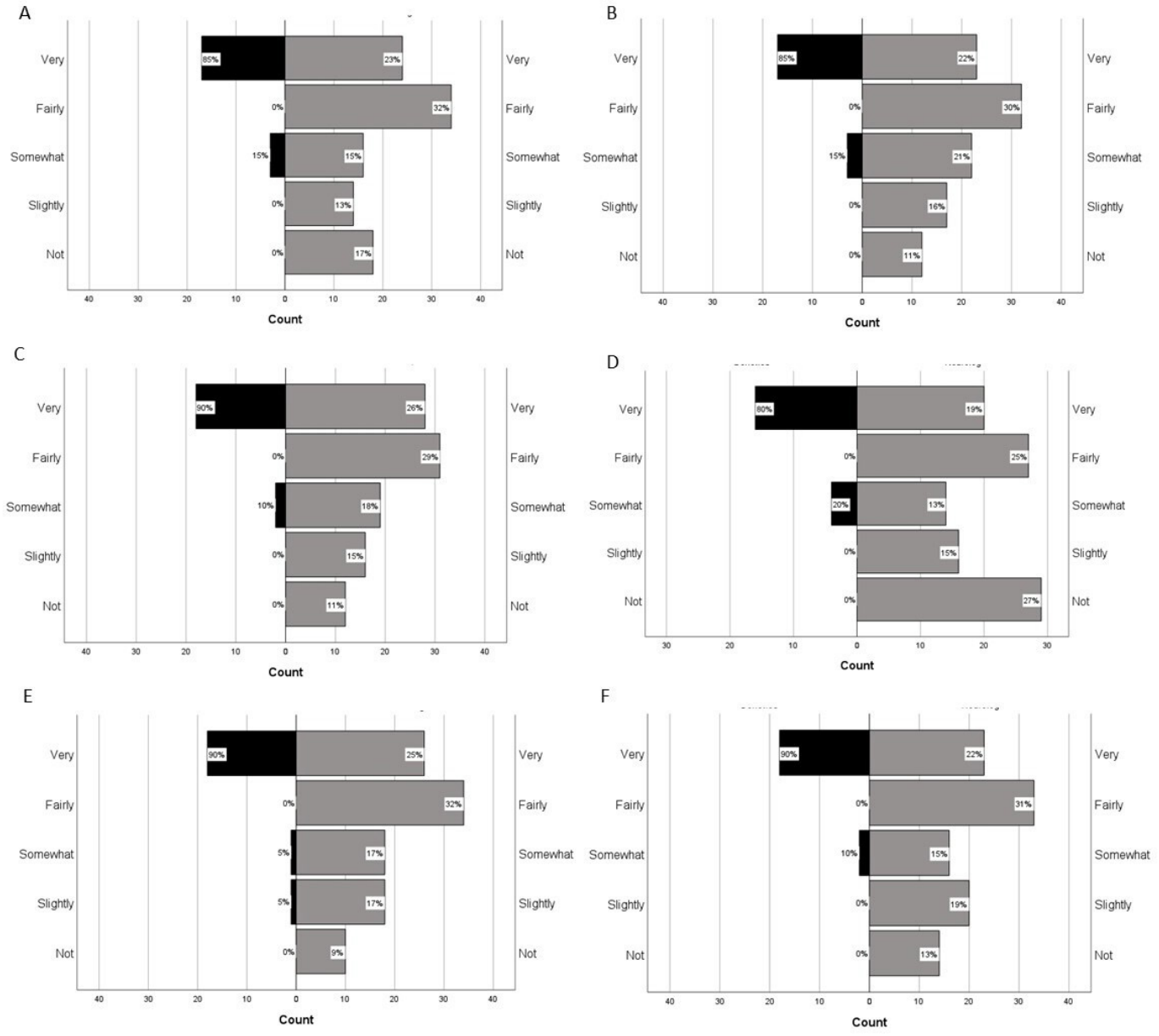

Figure 2 Self-reported confidence in procedures to request genome sequencing and confidence in genetic counselling skills for consultant neurologists. Pyramid blots illustrate consultant neurologists' (grey) and consultant geneticists' (black) responses on the 5-point Likert scale. (A) Completion of record of discussion form. (B) Interpreting a genomic laboratory report. (C) Discussing results with patients. (D) Explaining oligogenic inheritance. (E) Explaining variable expressivity. (F) Explaining a variant of uncertain significance.

proportion of consultant neurologists who had genetic counselling training had arranged MND genomic testing (12/13 vs 57/93, chi-squared p=0.028). Consultant neurologists with genetic counselling training did not rate themselves as 'fairly' or 'very' familiar on all genetic testing guideline questions more frequently than those without (1/13 vs 3/93, chi-squared p=0.4). There were no significant differences in these individual item scores between consultant neurologists with and without genetic counselling training. More consultant neurologists with training were likely to self-rate 'fairly' or 'very' confident for all genetic counselling (8/13 vs 19/93, p=0.0014), all clinical procedures (10/13 vs 32/93, p=0.003) and all predictive testing (7/13 vs 24/93, p=0.037) items than those without training. There were no statistically significant differences in genetic counselling skills, procedures to request GS or predictive testing individual item scores between trained and untrained consultant neurologists. There was no difference in any of the item scores for neurology consultants aged under or over 50 years. Suggesting that it is training in genetic counselling skills and not clinical experience which influences genomic knowledge and confidence. Overall, these findings support an influence of training in genetic counselling on confidence in genetic counselling skills among consultant neurologists (online supplemental figure 5).

Neurology clinicians lack adequate resources to support MND genetic discussions

We asked neurology clinicians about what resources would best support MND genetic discussions (online supplemental table 5). Only 50% of neurology consultants, 46% of neurology trainees and 19% of MND nurses felt that they currently have adequate resources to support such discussions. The most popular choice of resource was training materials on MND genetics (online supplemental figure 6).

DISCUSSION

We found that, in the English NHS, most GS for pwMND is requested by neurology consultants. A recent survey of English neurology consultants identified variability in offering GS for pwMND; less than 50% would discuss GS with newly diagnosed pwMND.[10] Our findings illustrate a low proportion of neurology clinicians discuss the possibility of predictive genetic testing. A recent global survey of neurologists found that only 48% discuss predictive testing.[11] It is crucial that neurology clinicians address predictive testing, where appropriate, given the potential role for presymptomatic treatments (eg, tofersen), noting the need for pretest genetic counselling (usually via a genetic counsellor).[12 13] Self-reported genomic knowledge and counselling skills were significantly lower in neurology clinicians than genetic clinicians. Only a minority of neurology clinicians rated themselves as 'fairly' or 'very' familiar/confident with core genomic knowledge and counselling skills. We found that training in genetics is associated with higher genomic knowledge and skills in neurology consultants, and greater likelihood of requesting GS for pwMND. Neurology clinicians reported multiple barriers to offering GS including a lack of time to discuss genomic testing in clinics with pwMND, and burdensome paperwork.

Our findings provide a potential explanation for variability in practice for GS and identify needs for changes to innovate genomic testing in neurology clinics. Our findings resonate with recent findings in the UK and globally suggesting these are important ingredients for interventions to integrate genomic testing in the NHS. North American primary care doctors reported low levels of confidence with requesting and interpreting genomic tests, and low understanding of ethical and legal frameworks.[14] A systematic review of barriers to offering GS, found lack of genomic knowledge, time and guidelines, as well as ethical concerns, were consistently identified as barriers.[15]

Our findings have implications for clinical practice and service innovation. Genomic testing for pwMND is being requested by neurology clinicians with low genomic knowledge and skills. Services must ensure that clinicians are trained appropriately. Training curricula for neurology clinicians need revision to include relevant aspects of genomics, and educational resources (eg, the NHS Genomics Education Programme) could be updated to include details on more complex aspects of MND genomic testing and clinician guidelines produced.[16 17] Additionally, neurology clinicians cited a lack of resources to support genomic testing discussions for pwMND, which suggests that pwMND may lack important information and guidance when considering genomic testing options. Resources such as information leaflets, videos or patient decision aids could be developed to fill this gap. In conclusion, we suggest that mainstream genomic testing for pwMND requires increased clinician training, streamlined processes and resources supporting shared decision making.

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Competing interests None declared.

Patient consent for publication Not applicable.

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