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Essential genetic testing in movement disorders – results from a Delphi study

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ABSTRACT

Background: While genetic testing in Movement Disorders (MD) has expanded enormously, access to genetic testing and genetic counseling remains asymmetric at the global scale. Guidance on efficient testing strategies for clinicians, governments and stakeholders is crucial.
Objectives: Establish a list of genetic movement disorders considered essential as determined by a group of MD experts.
Methods: All genes associated with MD were searched using the OMIM and MDS Gene database. We collected all additional tests available at 4 different laboratories from the EuroGentest database. The results were compiled in 6 questionnaires. A genetic test was considered essential if molecular testing had a direct impact in the management of the patient, including treatment of the disease or its comorbidities, or genetic counseling of the patient and family members. Two Delphi rounds were conducted asking MD experts which specific tests they considered essential in an adult MD clinic.
Results: Fifty-nine disorders were considered essential to genetically identify by the MD experts. This included 25 genes associated with ataxia, 15 with parkinsonism, 14 with dystonia, eight with chorea, five with paroxysmal disorders, four with myoclonus, four with hereditary spastic paraparesis, and one with tremor. Sixteen disorders reached 100% consensus among experts: Huntington's disease, PxMD-PPRT2, Wilson's disease, DYT-*SGCE*, DYT-*THAP1*, DYT-*TOR1A*, DYT/PARK-*GCH1*, Fragile-X Tremor-ataxia syndrome, PARK-*GBA*, PARK-*LRRK2*, PARK-*PINK*1, PARK-*PRKN*, PARK-*SNCA*, Cerebrotendinous Xanthomatosis, Ataxia-Telangiectasia, and Niemann-Pick disease type C.
Conclusion: This study provides a list of genetic MD that should be molecularly tested in adult centers with a compatible phenotype according to a group of MD experts.

The advance of DNA sequencing techniques, collectively called next-generation sequencing (NGS) techniques, has improved our understanding of genes involved in the pathogenesis and phenotypic presentation of neurological disorders, including movement disorders [1–3].

Molecular diagnosis can provide an explanation to the patient of the symptoms, ending often long series of ancillary investigation that frequently precedes a conclusive diagnosis, and improves genetic knowledge that may inform life choices and family planning. Furthermore, genetic testing can facilitate patient inclusion in clinical trials of potentially disease-modifying drugs and in research studies to advance treatments and provide better understanding of disease mechanisms [4]. Finally, although treatment is more often aimed at alleviating symptomatic manifestations of the disorder, in a subset of patients, disclosure of a genetic diagnosis will lead to a therapeutic action, either specific treatment decisions or tailored monitoring [5].

However, and even thought the cost of NGS is declining, these techniques are still prohibitively expensive in many regions of the world, often not available or only available through the private sector. Several barriers to accessing genetic testing and clinical exist worldwide [6]. Considering the practical impact of genetic confirmation and the asymmetric distribution of resources and availability of testing worldwide, guidance for clinicians working with genetic movement disorders is crucial. Implementing an essential movement disorder list could reduce the cost of testing and improve access to these valuable diagnostic techniques. We conducted a Delphi study among Movement Disorders' experts of three study groups and Task Forces of the Movement Disorders Society to identify which disorders were considered essential by experts.

1. Methods

1.1. Search strategy

We searched the Online Mendelian Inheritance in Man (OMIM) platform (https://www.omim.org/) using the term "ataxia", "tremor", "Parkinson's disease", "parkinsonism", "mirror movements", "myoclonus", "spastic paraplegia", "chorea", "dystonia", and "dyskinesia" for genes associated with movement disorders phenotypes. Additionally lesser-known variants associated with movement disorder phenotypes were searched for in the MDS Gene database (https://www.movementdisorders.org/MDS/Resources/MDSGene.htm). Both databases were accessed in January 2020.

To identify other possibly available genetic tests (such as karyotype, mitochondrial DNA analysis or whole genome sequencing) and the most common techniques used, we searched all genetic tests available at 4 different laboratories in the EuroGentest database in January 2020 (http://www.eurogentest.org/index.php?id=219). The EuroGentest is a project funded by the European Commission to harmonize the process of genetic testing, from sampling to counseling, across Europe, and this search was intended to obtain a list of available tests as extensive as possible. Data was retrieved on the genetic testing available online for four certified laboratories from four different countries (IBMC-CGPP in Oporto, MGZ – Medizinisch Genetisches Zentrum in Munich, the Genome Diagnostics in Nijmegen, and The Salpêtrière Laboratory in Paris) in January 2020. If the disorder was not present in EuroGentest, the "Genetic Testing Registry" (https://www.ncbi.nlm.nih.gov/gtr/) was used to confirm the laboratory methodology.

The phenotypic description was reviewed before the disorder was included in the final list. If insufficient phenotype data was present, the Medline database was used to review the phenotype. Pure sensory ataxia was excluded since these patients are not usually seen in Movement Disorders clinics.

1.2. Definition of essential genetic testing and clinical relevance

We defined essential genetic testing as for disorders/genes that should be molecularly tested and confirmed in the setting of a movement disorder clinic. To be considered essential, the disclosure of the genetic diagnosis should have direct and crucial implications for the patient's clinical management. The clinical management could include not only the treatment of the disease and associated co-morbidities, but also clinical and genetic counseling of the patient and families, and should be available at any center, regardless of setting (university/tertiary/non-university) or geographic location.

1.3. Delphi method

The Delphi method was used to define the essential genetic testing in the setting of an adult Movement Disorders Clinic.

The questionnaire was sent to all members of the Task Force on Recommendations for Clinical Genetic Testing in Parkinson's Disease, the Rare Movement Disorders Study Group, and to the Task Force on Genetic Nomenclature in Movement Disorders Study Group.

Two Delphi rounds were performed. Task Force members were asked

to use a Likert scale (completely disagree to completely agree) to determine how essential they found the testing of a particular gene/genetic technique when considering a specific phenotype (dystonia, parkinsonism, tremor, chorea and hyperkinetic movement disorders, mirror movement disorders, paroxysmal movement disorders, ataxia, myoclonus and startle syndrome, or spastic paraparesis).

The first Delphi round lasted from March to July 2021. A second Delphi round was sent to the responders of the first Delphi round and was performed from May to July 2022. In both rounds, we also asked members to provide further tests and comments they found relevant to add to the final list.

1.4. Data analysis

The responses of both Delphi rounds were evaluated using structured spreadsheets.

An answer was considered positive and added to the next list if the genetic test had an agreement rate (agree or completely agree) equal or superior to 75% in the first Delphi round and equal or superior to 80% in the second Delphi round.

Considering the global availability of and access to broad genetic testing and genetic counseling, whole exome sequencing and whole genome sequencing were excluded from the analysis.

2. Results

A total of 885 possible genetic test orders were compiled (820 disorders plus broad genetic testing techniques (Supplementary Table 1). The final list of genetic tests can be accessed in the supplementary data. The final list was further divided into 6 questionnaires according to the most frequent phenotypic manifestation ("chorea and paroxysmal disorders", "dystonia", "tremor and parkinsonism", "myoclonus, startle and mirror movements", "ataxic disorders" and "spastic paraparesis") (Supplementary data).

2.1. First Delphi round

Twenty-one experts answered the first Delphi round. Twenty members responded to the chorea/paroxysmal disorders/hyperkinetic questionnaire, 20 to the dystonia questionnaire, 21 to the tremor/parkinsonism questionnaire, 17 to the myoclonus questionnaire, 15 to the hereditary spastic paraparesis (HSP) questionnaire, and 14 to the ataxia questionnaire.

Of the 885 results of the first Delphi round, 13 (out of 52) genetic tests in chorea disorders met the agreement cutoff, nine out of 18 in paroxysmal movement disorders, 32 out of 174 in dystonia, 18 out of 63 in parkinsonism, five out 20 in myoclonus, 27 out of 391 in ataxia, and five out of 192 in HSP. None of the testing initially included in the lists of tremor disorders, startle, or mirror movements met criteria for inclusion in the second Delphi.

Furthermore, we asked experts to provide further comments/specific genes that might have been missed from the initial list. Two additional tests in chorea were suggested, as well as three in parkinsonism, one in myoclonus disorders, one in startle disorders, 66 in ataxia disorders and one in HSP.

Therefore, a final list of 184 tests (109 from the first Delphi round and 75 newly suggested) was included in the second Delphi round (Supplementary Table 2). Furthermore, two suggestions were excluded due to the absence of the suggested gene in OMIM or PubMed databases and one because the phenotype was consistent with sensory ataxia due to Charcot-Marie-Tooth.

2.2. Second Delphi round

A second Delphi round was sent to the responders of the first round. Likewise, the results were compiled in the six questionnaires mentioned above. Fourteen experts responded to the second Delphi round: 13 to the chorea/paroxysmal disorders/hyperkinetic questionnaire; 14 to the dystonia questionnaire; 12 to the tremor/parkinsonism; 11 to the myoclonus; ten to the ataxia; and ten to the HSP. Additionally, following suggestions from responders, DYT-*SCGE* was added to the myoclonus list, and PxMD-*SLC2A1* to the paroxysmal movement disorders list.

2.3. Final list

A total of 59 disorders meet the inclusion criteria (Table 1Table 2). Of these, 25 genes were associated with ataxia, 15 with parkinsonism, 14 with dystonia, eight with chorea, five with paroxysmal disorders, four with myoclonus, four with hereditary spastic paraparesis, and one with tremor.

Some of these disorders such as neuroacanthocytosis or

Table 1
Final list of essential genetic testing and associated disorders associated with chorea, paroxysmal movement disorders, dystonia, tremor and parkinsonism. **HDL:** Huntington-like disease; **FTD**: frontotemporal dementia; **Px-MD**: paroxysmal movement disorder. *: Suggested in the second Delphi round.

Final list of essential genetic testing	% agreement
Chorea	
HTT (CHOR-*HTT*)	100%
ADCY5 (MxMD-*ADCY5*)	92.3%
C9orf72 (HDL-disease and FTD-parkinsonism or Frontotemporal dementia and/or amyotrophic lateral sclerosis with parkinsonism)	92.3%
PRNP (Gerstmann-Straussler disease)	92.3%
TBP (ATX-*TBP*)	92.3%
VPS13A (CHOR-*VPS13A*)	92.3%
ATN1 (ATX-*ATN1*)	84.6%
FTL (CHOR-*FTL)*	84.6%
Paroxysmal Movement Disorders	
PRRT2 (PxMD-*PPRT2*)	100%
ATP1A3 (DYT/PARK-*ATP1A3*)	92.3%
KCNA1 (PxMD-*KCNA1*)	84.6%
MR1 (PxMD-*PNKD*)	84.6%
SLC2A1 (PxMD-*SLC2A1*)	*
Dystonia	
ATP7B (DYT/ATX-*ATP7B*)	100%
SGCE (DYT-*SGCE*)	100%
THAP1 (DYT-*THAP1*)	100%
TOR1A (DYT-*TOR1A*)	100%
GCH1 (DYT/PARK-*GCH1*)	100%
CYP27A1 (HSP/ATX-*CYP27A1)*	92.8%
DDC (DYT-*DDC*)	92.8%
KMT2B (DYT-*KMT2B*)	92.8%
TUBB4A (DYT-*TUBB4A*)	92.8%
VPS13A (CHOR-*VPS13A*)	92.8%
ATP1A3 (DYT/PARK-*ATP1A3*)	85.7%
HPRT1 (DYT/CHOR-*HPRT*)	85.7%
PANK2 (NBIA/DYT-*PANK2*)	85.7%
PLA2G6 (NBIA/DYT/PARK-*PLA2G6*)	85.7%
Tremor syndromes	
FMR1 (Fragile X Tremor-ataxia syndrome)	100%
Parkinsonism	
GBA1 (PARK-*GBA*)	100%
LRRK2 (PARK-*LRRK2*)	100%
PINK1 (PARK-*PINK1*)	100%
PRKN (PARK-*parkin*)	100%
SNCA (PARK-*SNCA*)	100%
PLA2G6 (NBIA/DYT/PARK-*PLA2G6*)	91.7%
VPS13C (PARK-VPS13C)	91.7%
VPS35 (PARK-VSP35)	91.7%
ATP13A2 (PARK-ATP13A2)	83.3%
C9orf72 (HDL-disease and FTD-parkinsonism or Frontotemporal dementia and/or amyotrophic lateral sclerosis with parkinsonism)	83.3%
DJ1 (PARK-*DJ1)*	83.3%
FBXO7 (PARK-*FBXO7*)	83.3%
GCH1 (DYT/PARK-*GCH1)*	83.3%
MAPT (Frontotemporal dementia and/or amyotrophic lateral sclerosis with parkinsonism)	83.3%
SNCA (Lewy Body disease)	83.3%

Table 2
Final list of essential genetic testing and associated disorders with myoclonus, spastic paraparesis and ataxia. **CANVAS**: cerebellar ataxia, neuropathy and vestibular areflexia syndrome. *: Suggested in the second Delphi round.

Final list of essential genetic testing	% agreement
Myoclonus	
DNAJC5 (MYC-*DNAJC5*)	90.9%
EPM2A (MYC/ATX-*EPM2A*)	81.8%
PRICKLE1 (adult myoclonic epilepsy type 1B - Unverricht and Lundborg type)	81.8%
SGCE (DYT-*SGCE*)	*
Spastic paraplegia	
CYP27A1 (HSP/ATX-*CYP27A1*)	100%
NPC2 (ATX-*NPC2*)	90%
NPC1 (ATX-*NPC1*)	80%
ABCD1 (Adrenoleukodystrophy)	80%
Ataxia	
ATM (ATX-*ATM*)	100%
CYP27A1 (HSP/ATX-*CYP27A1*)	100%
FMR1 (Fragile X Tremor-ataxia syndrome)	100%
NPC1 (ATX-*NPC1*)	100%
ATP7B (DYT/ATX-*ATP7B*)	90%
ATXN1 (ATX-*ATXN1*)	90%
ATXN2 (ATX-*ATXN2*)	90%
ATXN3 (ATX-*ATXN3*)	90%
ATXN7 (ATX-*ATXN7*)	90%
CACNA1A (ATX-*CACNA1A*)	90%
FXN (ATX-*FXN* – expansions analysis)	90%
FXN (ATX-*FXN*– gene sequencing)	90%
GBA1 (ATX-GBA)	90%
NPC2 (ATX-*NPC2*)	90%
TBP (ATX-*TBP*)	90%
SPG7 (HSP/ATX-*SPG7*)	90%
ATN1 (ATX-*ATN1*)	80%
ATXN10 (ATX-*ATXN10*)	80%
CP (DYT/PARK-*CP*)	80%
HEXA (ATX/HSP-*HEXA*)	80%
MTTP (Abetalipoproteinemia)	80%
PHYH (ATX-*PHYH*)	80%
PLA2G6 (NBIA/DYT/PARK-PLA2G6)	80%
RFC1 (CANVAS)	80%
TTPA (ATX-*TTPA*)	80%

cerebrotendinous xanthomatosis, appear in more than one phenotype list, showing both the heterogeneous and often overlapping phenotypes. Pleiotropy and phenotypic variation represents one of the diagnostic challenges neurogenetic disorders pose to clinicians. Therefore, a table with the most frequent phenotypes associated with the genes/disorders, inheritance mode, and the existence of gene-specific treatment is provided (Supplementary Table 3).

3. Discussion

The final list after two Delphi rounds includes 59 disorders. These were considered by a group of MD experts to be essential to be tested in adult patients presenting with a potentially genetic movement disorder. The genetic diagnosis was considered essential either for treatment, management of complications, prognosis, or family counseling.

This list comprises a small fraction of the potential genetic disorders that people living with MD can be diagnosed with. Although ideally every patient should receive as broad genetic testing as possible, a survey on barriers to genetic testing in Low and Middle-Income countries (LMIC) stressed not only the lack of availability of these tests but also the accessibility barriers, namely financial constraints, the scarce human resources and also health literacy and genetic knowledge among patients [7]. Another one focusing of PD, found similar results, with cost (79%), access (64%), and knowledge (60%) being perceived by responders as the most frequent barriers [8].

Regarding cost-effectiveness, although we found no study in movement disorders, a recent metanalysis in epilepsy testing compared three techniques – chromosomal microarrays (CMA), epilepsy panel (EP) with deletion/duplication testing and whole exome sequencing (WES). Regarding the diagnostic yield, this was highest for WES: 0.32 (95% CI: 0.22–0.44)), followed by EP: 0.23 (95% CI0.18–0.29), and CMA: 0.08 (95% CI: 0.06–0.12) [9]. Meanwhile, in the cost analysis, and considering prices practiced in the United States, the most cost-effective test was WES, with an incremental cost-effectiveness ratio (ICEP) of $15,000/diagnosis, followed by EP with an ICER of $15,848/diagnosis and CMA with $17,888/diagnosis. Furthermore, NGS has the advantage of possible reanalysis without the need of repeating sequencing, and whole genome sequencing (WGS) had the advantage of covering also intronic regions and detect structural variants and repeat expansions disorders [10]. However, the authors stress that cost-effective analysis often does not consider the phenomenology as a variable that might influence outcome. This is true also for movement disorders. For instance, expansion disorders, which are not included in WES, are the most common causes of autosomal dominant spinocerebellar ataxias, which might be underdiagnosed by this technique, as WGS is still not widely available, which might decrease the cost-effectiveness of these tests.

We must consider not only financial constraints, but also resources. Data from Gatto et al., showed that only 46% of the institutions surveyed offered genetic testing and these were not available in 13% of the countries [6]. Testing for chorea disorders was the most frequently available worldwide, while other phenotypes such as parkinsonism, dystonia, HSP, or ataxia had significantly more limited access in several regions. Regarding specific tests, another study, showed that the most frequently ordered genetic tests were multigene panels (52.9%), followed by single-gene testing (41.0%), whole-exome sequencing (29.7%), and whole-genome sequencing (14.9%), with variable results across the different geographic regions. Furthermore, even in the same country there can be asymmetry, and access to genetic testing can be a source of inequity in care [11]. Considering this, a list of tests that could be deemed essential could help local physicians and stallholders to advocate for local availability of single tests or even targeted genetic panels.

The final list contains not only disorders associated with single mutation variants but also microdeletions and duplications (such as *Parkin*-PD) and repeat disorders (such as HD and many spinocerebellar ataxias). Many of these disorders will not appear in broad genetic testing, such as NGS panels or in WES, highlighting the need to promote education in genetics in physicians treating people living with genetic movement disorders. In this context, targeted testing of "essential diagnoses" might be a feasible option, and a list of essential diagnoses might help clinicians and other stakeholders to push locally for the availability of these techniques. Furthermore, this list was obtained after extensive research of both disorders' phenotypes and testing techniques and achieved through a consensus among Movement Disorders Experts actively engaged in MDS working groups working with genetic disorders.

However, this list and should not be interpreted as the sole guidance for a genetic diagnostic work-up in patients with MD and we acknowledge several limitations.

Although this questionnaire was sent to a diverse group of movement disorders experts, and phenomenology was considered, we did not consider epidemiology. Furthermore, both the composition of the panels to which the questionnaires were sent and the final list of responders is highly geographically biased, including mostly European and American (both North and South) responders, which might have implications in the list of disorders selected, since burden, priorities, and impact of genetic disease can vary between geographical location [12]. Although this list aims limited constrained settings, most of the responders to the Delphi study came from high income settings, which might introduce bias into the answers. Future endeavors should promote a greater geographic diversity in the panel.

We also obtained a small number of responders, and that the prevalence of the disorders was not specified in the Delphi questionnaire. Therefore, the results might be biased by the responders' personal

experience and geographic location. Even though in most categories, the most common genetic causes of the phenotypes are included in the final list, there are some exceptions that are worth mentioning. In chorea syndromes, although ADCY5, a relatively new and rare form of chorea was included, Huntington-like disease (HDL) type 2, caused by CTG expansions in *JPH3* was not included, even though is it responsible for 15% of cases in South Africa [13], and up to 10% of cases of HDL cases in Brazil [14]. Likewise, in spastic paraparesis, SPG4, one of the commoner genetic causes is excluded, while rarer disorders were included [15]. This bias is likely to reflect the epidemiological context where experts work, and may also be influenced by the lack of specific treatments for certain phenotypes, such as spastic paraparesis.

It should also be considered that not every potentially treatable disorder highlighted by a previous paper by the Rare Movement Disorders Study Group (RMDSG) [16] appears in this recommendation. However, most of these disorders usually present in infancy with severe phenotypes, rarely seen or diagnosed in adulthood, and therefore we re-iterate that this is a recommendation for movement disorders with adult onset. Furthermore, we acknowledge that many childhood-onset movement disorders may reach adult movement disorders clinics undiagnosed, thus it is important for adult movement specialists to be aware of them, and view this as a limitation of this guidance.

Finally, as genetics is an ever-evolving field, since the end of the search and the final draft of the consensus, other genes have been associated with the included phenotypes. However, in most cases, these additions refer to rare disorders, with only single case or limited case series reported, and therefore these would be less likely to be included by experts as disorders essential to be tested, considering the lack of knowledge on prevalence, treatment, or therapeutic guidance in most cases. An exception is the recently identified ATX-*FGF14* (SCA27B), which seems to explain 10-60% of cases in various populations of inherited and sporadic ataxias. Moreover, patients with ATX-*FGF14* have a good response to treatment with 4-aminopyridine, which makes their identification crucial [17,18]. This gene had not been identified at the time of the first Delphi round and therefore was not considered in this study. Additionally, considering the rarity of some of the genes and related reports in the literature, it is possible that some genes associated with MD were not included in the initial list, nor in the diagnosed added to the second Delphi round. The questionnaire also did not consider the well-established phenotypic variability that underlies many genetic disorders nor the possibility of the presentation of complex movement disorders.

This is not a list that should be adopted blindly: phenotype, age of onset, family history, epidemiology, and patient's preferences should always be considered when investigating the genetic etiology of a patient's disorder, nor it should be interpreted as a minimal required list of all centers, but as a guidance for centers trying to implement testing in MD clinics.

And finally, this list should not be considered alone, as to promote implementation of testing in low-resource incomes, several steps should be considered. Building of working capacity at local level, and the promotion at the national and international level of collaborative efforts [10,19,20], both in healthcare and research, such as the GP2 initiative [19] are pivotal. In LMIC, a major impediment to the implementation of genetic medicine training is the lack of health professionals trained in genetics, genomics and bioinformatical analysis. Furthermore, the education of clinicians, using digital platforms to democratize access to knowledge, and the development of best-practice and value-based guidance for genetic testing in MD are also crucial. The scarcity of cytogenetics and molecular genetics laboratories and disparities in health priorities, might also prevent or challenge the allocation of funding [21]. Fostering national and international networks between neurological and genetic centers, public-private partnerships, and advocacy for governmental funding are also needed in many settings to promote implementation of laboratories, and promote testing in unserved regions [7,22]. The establishment of well-characterized cohorts, including all the key stakeholders would be helpful to provide insight in the epidemiology of different genetic disorders in underserved populations and to prioritize the allocation of health resources and testing [22]. Finally, patient's education, patients' advocacy and public awareness of genetic disorders are crucial, especially in populations where genetic disorder is accompanied by heavy social stigma and discrimination [7,22].

In conclusion, we report a list of genetic disorders that were considered essential to be available for testing in adult Movement Disorder centers by Movement Disorders experts. This list is a first effort to help clinicians worldwide advocate for implementation of genetic testing. This could be replicated in the future, with larger groups in smaller geographic regions, and considering local epidemiology, to help clinicians improve local diagnosis, counseling and treatment.

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The authors confirm that the approval of an institutional review board was not required for this work.

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