GAPS & CONTROVERSY

'What's in a Name?' Naming Genetically Determined Movement Disorders: Gap and Controversy

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ABSTRACT: In 2016, the International Parkinson and Movement Disorder Society (MDS) Task Force for Genetic Nomenclature in Movement Disorders laid out a new proposal for naming genetically determined movement disorders. This proposal sought to address the difficulties arising from the practical usage of numbered loci (eg, DYT1, DYT2, DYT3, etc.) as names for disorders. The proposal incited commentary highlighting concerns of subjectivity, neglecting relevant non-movement features, and predicting the need for constant change as knowledge. An evaluation of the use of the nomenclature in the recent peer-reviewed literature revealed variable implementation across movement phenotypes. The nomenclature has strengths and weaknesses, which are discussed in this article. A consideration of opportunities for improvement is warranted, and weighing the options will be the task of the MDS Nomenclature in Genetic Movement Disorders Study Group.

Key Words: genetics; movement disorders; nomenclature

In 2016, the International Parkinson and Movement Disorder Society Task Force for Genetic Nomenclature in Movement Disorders laid out a new proposal for naming genetically determined movement disorders.[1] This proposal (hereafter referred to as the 'Task Force nomenclature') sought to address the difficulties arising from the practical usage of numbered loci (eg, DYT1, DYT2, DYT3, etc.) as names for disorders.[2] Problems arose because the assignment of numbers and phenotypic prefixes lacked consistent criteria and was uncurated. These lists contained unconfirmed and spurious associations, and duplicate entries.

The Task Force nomenclature set a level of evidence required to link a gene to a disease and set guidelines for how to determine genotype–phenotype relationships. The format of this nomenclature consisted of a prefix that could accommodate one or more dominant movement phenotypes (eg, DYT for dystonia or DYT/PARK for dystonia/parkinsonism) followed by the name of the gene (eg, DYT/PARK-*GLB1*). The goals of this nomenclature were to provide a standard way of referring to genetically determined movement disorders and to provide reliable groups of disorders that, by virtue of meeting the specified criteria,

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conveyed meaningful information regarding genotype–phenotype relationships.

While addressing an unmet need, the proposal incited commentary highlighting concerns of subjectivity, neglecting relevant non-movement features, and predicting the need for constant change as knowledge evolves.[3,4] Since its introduction, the system has grown to include over 300 genetic disorders manifesting dystonia, parkinsonism, ataxia, spastic paraparesis, and chorea.[5] Through this experience, challenges to the implementation have been recognized, and changes have been introduced to improve usability. As an index of the Task Force nomenclature's adoption by the community, we assessed its use in the English peer-reviewed literature from 2018 to 2024. We included clinical studies not authored by Task Force members related to each movement phenotype and its most common causative genes. Of 203 published papers sampled, 35 (17%) used the Task Force nomenclature. The proportion differed across movement phenotypes, being highest in dystonia (29/35, 64%) and lowest in parkinsonism (0/56). These results may reflect the longstanding engrained use of the simple (although defective) numbered system and the preferential adoption of a naming system where it helps in diagnosis and treatment (eg, in dystonia). The low adoption rate in other movement disorders indicates that the introduction of the new system has increased variability of nomenclature in the field, which hampers communication between and among researchers, health care providers, patients, and families. Thus a re-evaluation of the system is necessary.

The fundamental feature of this nomenclature is the connection of phenotype to genotype (ie, gene name). This connection was felt to be valuable, conveying important information about the disorder and standardizing terminology for the field. Around the same time, the two-axis classification of dystonia became an established approach for classifying other movement disorders.[6] A multi-axis structure, distinguishing clinical phenomenology from underlying etiology, was later extended to tremor[7] and myoclonus.[8] Bridging the axes through the Task Force nomenclature could facilitate diagnosis in the clinic, and guide genetic testing and interpretation. However, separating phenotypic and etiological axes has been a central feature for the classification of movement disorders because of the frequent lack of one-to-one correspondence between etiology and clinical features.

Given the operational challenges, in conjunction with the variable but overall low adoption in the community, the merits of connecting phenotype to genotype in a formal nomenclature deserve careful reconsideration. Thus, the lead developers came together with movement disorders experts not involved in the Task Force system to examine its strengths and weaknesses and propose directions for refinement.

What We Know

Naming of Genes and Disorders

For many decades, the Human Genome Organization (HUGO) has provided recommendations for naming genes.[9] Names for human genes should be comprised of uppercase letters and sometimes numbers in italics and, with rare exceptions, these guidelines have been adopted throughout most of human genetics. The Task Force nomenclature includes these HUGO-approved gene names.

However, the approach to naming disorders is less consistent, and the best strategy has become a matter of debate.[10] The historical approach of applying possessive or eponymous names (eg, Wilson's disease) is now discouraged.[11] Another historically popular strategy has been to name a disorder based on the initially described phenotype (eg, rapid-onset dystonia-parkinsonism). This approach has also been discouraged because it oversimplifies the phenotypic heterogeneity for many genetic disorders. After the causative gene for this disorder was found to be *ATP1A3*, other cases were shown to have very different phenotypes, such as insidious onset, and some had only dystonia or parkinsonism but not both. The same gene was ultimately linked with multiple strikingly unrelated phenotypes, including alternating hemiplegia of childhood, hemiplegic migraine, epilepsy, or CAPOS syndrome (cerebellar ataxia, areflexia, pes cavus, optic atrophy and sensorineural hearing loss). In fact, it is not infrequent in genetics that an initial specific phenotype later expands as more cases with the gene defect are found. Other disorders are named according to the biological consequence of the genetic change (eg, cerebrotendinous xanthomatosis). A major challenge is to address genetic heterogeneity and phenotype heterogeneity at the same time, which intermix variably in individual patients.

Some have proposed that genetic disorders follow the convention used in 'Online Mendelian Inheritance in Man' (omim.org), which assigns each disorder a unique six-digit number.[9] Indeed, some journals require this number in publications. Aside from challenges in remembering such numbers, the link from a gene to a disease is not unique, as different phenotypes, as well as increased susceptibility to various conditions, are often linked to the same gene (ie, the same number). Additional symbols preceding the number therefore specify relationships between phenotypes and genes/loci. To address the scenario of a phenotype having potentially different genetic causes, a progressive numbering system is sometimes used, including for various movement disorders (eg, DYT1, DYT2, etc.). Others have proposed that disorders should be named after the pathogenic gene (eg, *GCH1*-associated dystonia or parkinsonism). However, it has been argued that this strategy places undue emphasis on the gene name, when the phenotype may be more relevant to both clinicians and patients.[9] In short,

there is no widely accepted or uniform procedure for naming disorders.

The Task Force Nomenclature

The main features of the Task Force nomenclature for linking phenotypes (prefix) and genes (suffix) in monogenic movement disorders are summarized below.

1. The nomenclature only applies to disorders for which disease-causing pathogenic variants in a certain gene have been identified.
2. A movement disorder prefix is assigned when the phenotype (eg, PARK for parkinsonism) is a prominent feature of the disease linked to pathogenic variants in that gene in the majority of cases. Double (eg, DYT/PARK) prefixes can be assigned to accommodate two prominent phenotypes and mixed movement disorder (MxMD) prefix for more than two.
3. The suffix to the phenotype-driven prefix comprises the gene name (eg, DYT-*TOR1A*) for dystonia caused by pathogenic variants in the *TOR1A* gene.
4. The system only covers genes with disease-causing variants (monogenic disorders in contrast to genetic risk factors or polygenic risk).

Before including a gene in the list and assigning a prefix, criteria supporting pathogenicity of the gene's variation must be met, consistent with American College of Medical Genetics and Genomics (ACMG) recommendations.[12] In addition, two independent reports must be published describing the association between a movement disorder and the gene.

The relevant information is evaluated by Task Force (now Study Group) members to render decisions on naming. In cases where uncertainty remains, external experts are consulted.

Gaps and Controversies

Strengths and Weaknesses of the Current System

A nomenclature for genetically determined disorders should convey relevant information about a disorder, accurately reflect knowledge regarding the condition, be adaptable to changing knowledge, and, lastly, be easy to apply and align with nomenclature in the rest of medical genetics. These desired attributes are outlined in Table 1 and further described below.

A Nomenclature that Conveys Useful Information

Strengths

The Task Force nomenclature conveys a relationship between a given gene and the corresponding movement phenotype. Clinically, the importance of this relationship lies in guiding diagnostic testing by selecting phenotype-relevant genes. In research, lists of genes with particular phenotypic associations can provide clues to mechanisms of disease, by revealing previously unrecognized patterns. Naming disorders also has value as a communication tool and as a mnemonic aid for learners. There are precedents for the importance of linking phenotype to etiology, such as the recommendations issued by the World Health Organization (WHO) on best practices for naming new human infectious conditions.[13] These recommendations state that a disease name should consist of terms, based on the symptoms, who it affects, its severity or seasonality, and if the cause of the disease is known, it should be part of the disease name.

Recognizing the possibility of multiple phenotypic presentations, the Task Force nomenclature provides the option of including two movement phenotypes in the name when appropriate and acknowledging greater variation or complexity (more than two relevant phenotypes) with a specific prefix of MxMD. The process for assigning prefixes involves expert adjudication of the predominant phenotype in the majority of cases ensuring that there is meaning conveyed in the name. Despite these accommodations, it is difficult to properly reflect the high phenotypic variability and complexity of certain genetic movement disorders. Therefore, the prefix, in its current form, provides guidance to the most likely phenotype(s) but should not be regarded as an invariable or sole presentation.

Weaknesses

Connecting gene to phenotype comes with challenges. The main weakness is the information provided by the prefix. The MDS Task Force recommended that a phenotypic prefix be assigned only when the phenotype is observed as a *prominent* feature in the *majority* of cases. Two prefixes were to be included when two phenotypes occur roughly equally, and a mixed phenotype (MxMD) aimed to encompass greater phenotypic heterogeneity. However, the MxMD prefix conveys non-specific information about the phenotype. Where specific prefixes are provided, decisions regarding the prefix can sometimes be somewhat subjective and therefore subject to dispute; even among experts, 'predominant' phenotype has not been formally defined. Further confusion may arise when non-neurological phenotypes predominate (eg, common liver-predominant presentations of Wilson's disease) yet are assigned a movement prefix (DYT-*ATP7B*).

Only a minority of movement disorder genes are consistently associated with a single dominant phenotype. Applying a single or two prefixes oversimplifies the complexity of actual phenotype–genotype relationships.[14] Indeed, the lack of a one-to-one correspondence between

TABLE 1 *Strengths and weaknesses of the Task Force Nomenclature proposal based on important attributes of a nomenclature for health conditions*

Strengths	Weaknesses
Conveys relevant information	
• Links phenotype to genotype • Conveys the usual core phenotype • Conveys cause • Helps less experienced neurologists and learners by providing an overview of genetic causes of a given phenotype	• There is frequently no one-to-one link between movement phenotype and gene • Does not convey variant-specific information
• Allows more than one phenotype to be linked to a gene	• Challenging for complex phenotypes with many symptoms • Focus on predominant (movement) phenotype may reduce recognition of less common or non-motor phenotypes • Challenging to define the criterion of 'prominent' and frequency of movement disorder phenotypes that vary across individuals or with disease duration • Mixed movement disorder (MxMD) does not convey specific information regarding the actual phenotype(s)
• Transparency of thought process for assigning a phenotype–genotype link	• Identifying the predominant phenotypic features is subjective • Available evidence may be limited
Accurately reflects knowledge	
• Requires a minimum evidence base • Reduces the chance of assigning non-causal relationships	• Lack of consensus on criteria distinguishing risk factors versus causative genetic variants raises possibility of conveying inappropriate certainty as to causal/risk factor status • Simplification may compromise accuracy • Not all phenotypes are considered (eg, no prefix for tremor) • Knowledge changes quickly, raising possibility of inaccuracies if updates are delayed
Adaptable to changing knowledge	
• Stated intent to change names according to new knowledge • Updates periodically provided on MDS website	• Rapid evolution of knowledge makes timely updates challenging • Updating names if knowledge changes may cause confusion when comparing published names over time • Assigned names can become too quickly entrenched
Easy to apply	
• Short name aids memorization	• Could be simpler (eg, [gene]-related disorder) • Acronyms may be difficult to understand
• Focused on movement phenotype	• Focus on movement disorder neglects other (sometimes more) important aspects of phenotype • Unlikely to be adopted by non-movement genetics
• Conveys most relevant movement phenotype only	• Movement phenotype often oversimplified and may cause less common manifestations to be overlooked
Alignment with nomenclature used in related fields	
• Aligns with WHO recommendations for naming infectious diseases	• Does not align with nomenclature used in the rest of the field of human genetics

Abbreviations: MDS, International Parkinson and Movement Disorder Society; WHO, World Health Organization.

phenotypes and etiology has been a major factor driving trends to classify movement phenotypes using two separate axes as mentioned earlier. A nomenclature plan that attempts to merge phenotype and genotype conflicts with this trend. In the broader community of human genetics, it has also been advocated that names for phenotypes and genotypes should develop independently.[10]

Additionally, the appropriate prefix is subject to change over time as the disease progresses and/or as knowledge evolves. Another aspect of simplification is inherent in the gene name. For many if not most genes related to human diseases, different variants can be responsible for different phenotypes. The Task Force nomenclature does not include any information related to specific variants

or their relationship to phenotype. Admittedly, doing so would increase the complexity of the nomenclature and we are not aware of systems that have incorporated variant-specific information.

A Nomenclature System that Accurately Reflects Knowledge Regarding the Condition

Strengths

A major advantage of the Task Force nomenclature is the reliance on published and peer-reviewed evidence to guide the names of disorders. Assigning a prefix requires strong evidence that a gene has a disease-relevant relationship to the phenotype, based on criteria for pathogenicity as well as at least two independent publications linking the gene to a movement phenotype. This requirement minimizes incorrect nomenclature by reducing the chance of assigning non-causal relationships and reduces (although does not eliminate) instances of changing prefixes due to initial cases not being representative of the full phenotypic spectrum.

Weaknesses

Implementing the Task Force nomenclature requires decisions that are subjective. As an example, DYT11 (*SGCE*) was first named DYT-*SGCE*. However, myoclonus is often a more prominent aspect of the phenotype than dystonia.[15,16] After the introduction of the MYC prefix, the name was updated to MYC/DYT-*SGCE*. Another example relates to the absence of a prefix representing tremor. The recent classification of dystonia acknowledges the possibility that tremor is part of the phenotypic presentation of dystonia or alternatively a coincident additional movement disorder[6] Here, the DYT prefix would be used in the first case, whereas an alternative option would be DYT/TREM, but no prefix representing tremor has been established, since to date no monogenic isolated tremor disorders have yet been identified. Lesch–Nyhan disease provides another example. Under Task Force guidelines, this disorder has been designated DYT/CHOR-*HPRT1*. However, chorea is not a prominent feature in the majority of either classic[17] or atypical variant phenotypes[18] associated with pathogenic variants in *HPRT1*. The reasons for including CHOR could therefore be questioned.[19]

Subjectivity is also inherent in the decision to consider a variant causal rather than a risk factor for a condition because there is currently no consensus in the field as to the threshold for designating a variant causal. Therefore, assigning certain prefixes may convey inappropriate certainty that variants in the gene are considered causal. *GBA1* is an example of a gene that historically has been variably described as a risk factor or a causative gene, depending on the genetic variant. These examples highlight the subjective nature of prefix assignment under the proposed system and raise an even more difficult question regarding who has the authority to designate the most prominent phenotype(s), and who will arbitrate when there are disagreements.

A Nomenclature that Is Adaptable to Changing Knowledge

Strengths

In the face of rapid changes in knowledge, a system must be flexible and adaptable. In the field of movement disorders alone, the last Task Force nomenclature update of the literature from 2016 through 2021 identified over 80 new causal gene-phenotype relationships requiring additions to the nomenclature. In response to findings of recent systematic reviews, several prefix changes must now be considered. In this setting, the date of the last search of the literature on which the lists are based should be made clearly known in a public resource and updated as often as possible. With these goals in mind, publications of the latest lists are available on the website of the International Parkinson and Movement Disorder Society (MDS) (https://www.movementdisorders.org/MDS/About/Committees--Other-Groups/Study-Groups/Genetic-Nomenclature-in-Movement-Disorders.htm) and can be updated as necessary.

Weaknesses

This adaptability is both a strength and a weakness. The main weakness is that a disorder should preferably have a stable and consistent name over time. Some changes have already been proposed, such as inclusion of new prefixes (eg, MxMD) and elimination of some prefixes (eg, NBIA for neurodegeneration with brain iron accumulation). A changing name is confusing to patients and clinicians. In the research community, literature searches are much easier if the name is stable. The difficulty in eliminating eponyms as names for disorders as well as easy to remember numerical designations (eg, DYT1, etc.) demonstrates the difficulty in changing names once they come into common use.

A Nomenclature that Is Easy to Apply

Strengths

The Task Force nomenclature simplifies the clinical spectrum to yield a concise name. Although not a feature of the nomenclature per se, a decision was made to restrict the prefix(es) to describing the movement phenotype associated with a particular gene and only the most prominent movements. This approach necessarily omits information for more complex or heterogeneous phenotypes that include non-movement characteristics, or variations across individuals or over time. However, in doing so, it provides a more easily said and

remembered name that represents the core phenotype and is more likely to be adopted than a more complex name.

Weaknesses

In principle, the Task Force nomenclature is easy to apply. It links a relevant phenotype to a relevant gene. In practice, the plan may not be perceived as simple by individuals who are not experts in genetics. Gene names themselves are difficult to remember in comparison to numbers. The added requirement for linking them to a phenotypic prefix that may change over time adds further complexity. Furthermore, when the nomenclature varies according to different disorders, then the plan also becomes more difficult to learn and remember. For example, no prefix is assigned where a movement disorder is not usually prominent, even when a prominent movement disorder can indeed be observed in some individuals. Instead, inclusion in the respective phenotype list indicates this possibility.

A Nomenclature Plan that Aligns with Nomenclature of Genetic Disorders in Other Fields

Strengths

In the face of the lack of consensus on the naming of disorders, the Task Force nomenclature opted for a custom solution. In principle, it aligns with the WHO recommendations for naming infectious diseases, which include the major symptoms *and* the etiology (among other elements).[13] Nomenclature in genetically determined disorders is usually not formally established or consistent even within disease areas. In this context, a solution tailored to movement disorders introducing a systematic naming method could provide an example for other fields to adopt.

Weaknesses

Any approach to naming genetic movement disorders should be compatible with the approach in other fields of medicine. It is not likely that the proposed nomenclature will replace well-established names, such as possessive names or names based on phenotypes or pathobiology. Since existing prefixes are limited to movement disorders, the plan is not likely to be adopted by specialists in other fields, leading to confusion from multiple concurrent nomenclatures. It is also not likely that the Task Force nomenclature could be broadened to encompass non-movement phenotypic features to overcome this weakness, because the names would become too complex and unwieldy. In short, the Task Force nomenclature adds another naming option where multiple competing options for naming already exist. While a uniform plan for naming disorders may be desirable, it seems unlikely that a single plan will accommodate the remarkable heterogeneity already in place.

Moving Forward

The current Task Force nomenclature has both strengths and weaknesses that should guide how we move forward. It is important to acknowledge that the expansion of our knowledge of genetic determinants of disease has essentially outstripped human ability to systematically apply and remember complete lists of genes and their phenotypes. There are more than 100 genetic ataxias, and the number of genes for hereditary spastic paraplegia exceeds 80. Ultimately, solutions leveraging artificial intelligence that can be used to easily search databases for the phenotypic spectrum and provide possible genetic causes to consider based on *objective* (quantitative) criteria will serve the purpose of linking phenotype to gene. To date, inconsistent and insufficient phenotypic information in publicly available sources as well as disparities in access to genetic testing across the globe have handicapped efforts to understand phenotype–genotype relationships, and until this situation improves, artificial intelligence cannot provide reliable solutions. In the interim, options for the naming of genetically determined movement disorders include modifying the criteria or processes used to assign the names or modifying the format of the names themselves. These options are summarized in Table 2.

Considering the issues identified related to the subjectivity of naming disorders according to the nomenclature, assigning thresholds to define a *predominant* movement phenotype may increase objectivity. However, the challenges of incomplete or inaccurate clinical descriptions in the existing literature make implementation difficult.[20] Much of our current knowledge resides with clinical experts, remains unpublished, or is presented in formats that are challenging to extract in a systematic way. Seeking external advisors in the absence of expertise within the Study Group could access this knowledge, where available. A formal consensus process within the Study Group would increase transparency of decision-making.

Any modifications to the nomenclature itself should aim to improve its utility for the clinical and research communities, particularly emphasizing ease of use, and optimizing the information inherent in the name. The nomenclature was originally conceived to be applicable to all movement phenotypes and disorders; however, it is clear that it is challenging to apply to disorders with variable phenotypes across individuals and throughout the course of the disease. An option moving forward could be to apply the phenotype-gene naming format to those disorders with longitudinally consistent and simple phenotypes, or to assign prefixes based on the phenotype

TABLE 2 *Options for modification of the Task Force nomenclature*

Options	Pros	Cons/Limitations
Criteria modifications		
• Assign threshold criteria for 'prominent' or 'predominant' features • Assign names only after a sufficient number of cases have been described	• Increases interpretability/ meaningfulness of phenotypic prefix • Reduces the chances of unusual or inaccurate phenotypes influencing nomenclature • Reduces iterations of naming as the spectrum of the condition becomes known with additional cases	• Clinical features are often incompletely reported in the literature
Process modifications		
• Engage experts external to Study Group in the absence of internal expertise • Develop a formal process for resolving disagreements regarding prefix assignment • Review nomenclature periodically	• Accesses unpublished knowledge to improve accuracy of prefix assignment • Increases transparency of decision-making • Adapts prefixes to changing knowledge	• Experts may not be accessible for all disorders
Format modifications		
• (Gene)-related disorder	• Simple, avoids potentially misleading phenotype description	• Does not convey information on phenotype in the name itself
Scope modifications		
• Restricting to disorders with longitudinally consistent and simple phenotypes • Restricting to phenotypes with established two-axis classifications	• The same name will apply over the course of the disease • Names will be more consistently reflective of the dominant phenotype	• The naming system will apply to only a subset of disorders, limiting utility

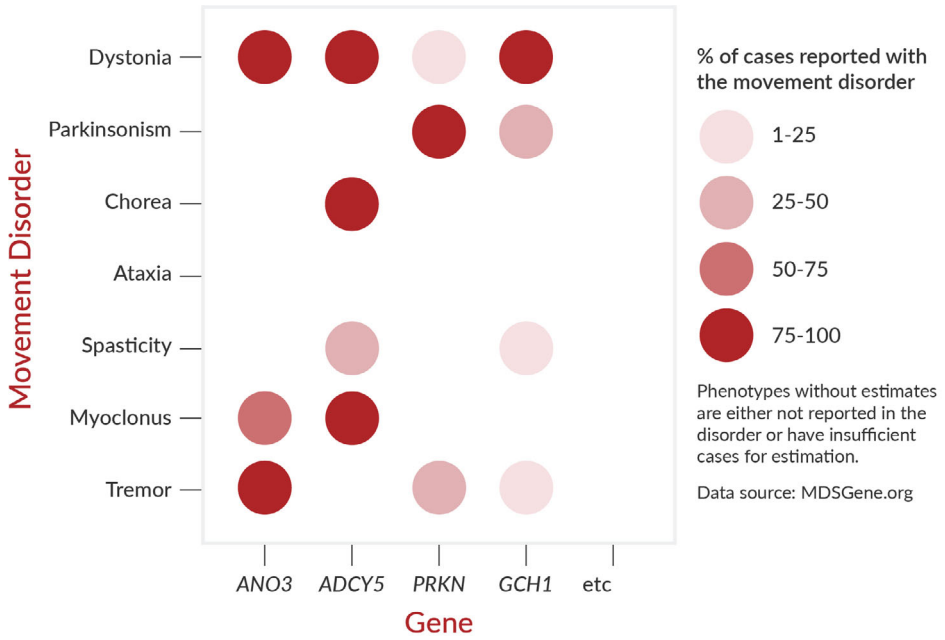

FIG. 1. Conceptual drawing of an online resource summarizing published data on gene–phenotype relationships for movement disorders. [Color figure can be viewed at wileyonlinelibrary.com]

present early in the disease. Alternatively, prioritizing disorders such as dystonia may provide a pragmatic way forward, as the two-axis classification for dystonia has been established for more than a decade and has gained wide usage. Admittedly, a system that cannot be systematically applied across our field to other movement phenotypes may not be widely accepted or useful.

Given these challenges, simplification of the nomenclature may offer a way forward. Simplifying the names to a format such as (gene)-related disorder (eg, *ATP1A3*-related disorder or, as has been suggested by experts in the complex field of genetic ataxias, [gene]-spectrum disorders[14]) could be a compromise. The current naming of genetic generalized epilepsies provides an example; these conditions are classified into major phenotypes (Juvenile Myoclonic Epilepsy, Juvenile Absence Epilepsy, Childhood Absence Epilepsy, and Epilepsy with Generalized Tonic–Clonic Seizures alone).[21] Etiology-based naming of the monogenic forms is handled separately with the gene name followed by '-related seizures' or '-related epilepsy' (eg, *SLC2A1*-related seizures).[22] This type of approach does not attempt to connect phenotype to genotype. More specific variations could be applied to individuals with different phenotypes, such as *ATP1A3*-related rapid-onset dystonia-parkinsonism or *ATP1A3*-related CAPOS syndrome. This naming format is already unofficially used in the literature (eg, Graziola et al.[23], Johansen et al.[24]). Such a solution does not specify the most common or predominant phenotype at the group level and therefore does not convey information beyond the individual's specific presentation, but it avoids many of the complications of the Task Force nomenclature.

Weighing these options (summarized in Table 2) will be the task of the Genetic Nomenclature Study Group. In this process, community consultation of a geographically diverse sample of individuals representing the basic and clinical sciences will be necessary to ensure we are meeting the needs of the movement disorders community. Meanwhile, the field would benefit from an online, publicly available resource showing associations between movement phenotypes and genes independent of a disease nomenclature. A draft example is shown in Figure 1. Such plots are valuable for showing the most common phenotype, but also less common phenotypes, and could incorporate the spectrum of phenotypic severity too. The data should be based on published literature, and one of the functions would be to highlight knowledge gaps. ●

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Data Availability Statement

Data sharing not applicable to this article as no datasets were generated or analysed during the current study.

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APPENDIX

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