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ARTICLE

Investigating the role of genetic counseling in neuromuscular disease considering life events

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Abstract

Genetic diagnoses are becoming a routine in the medical practice of neuromuscular diseases. Many diagnoses, however, can have an influence on relatives and family members and thus must be handled carefully by genetic counseling (GC). Here, we aimed to assess the purpose of undergoing GC to verify the utility of collaborations between clinical and genetic divisions. We investigated consecutive GC cases of neuromuscular disease and examined the role of GC. Our study included 102 cases who underwent GC in our hospital from July 2005 to March 2018: 86.3% were women and 45.1% were in their 30's. Disease explanation was the most common reason for attending GC (29.4%), followed by prenatal diagnosis (25.5%), pre-symptomatic diagnosis (17.6%), and carrier diagnosis (14.7%). Clients typically visited the hospital for GC when some kind of life event occurred, such as marriage, had a desire to bear a child, or a change in the condition of the proband. Clinicians should be conscious of such life events from the perspective of both the client and their relatives, and guide the GC at an appropriate time. Overall, the degree of recognition of genetic risk by clients differed; thus, it is important for GC to determine the status of each unique situation and respond individually.

Introduction

Genetic testing is an effective approach to gain a definitive and differential diagnosis in neuromuscular diseases. A survey conducted in 2013 in > 1400 certified neurologists in Japan reported that 82.6% had some experience with genetic testing [1]. Because genetic information does not change in our lifetime and is shared with the relatives, the impact of obtaining genetic information is large. Although genetic testing is performed in daily clinical practice, special attention should be paid to these shared characteristics of genetic information when a genetic test is performed.

Genetic counseling (GC) aims to help people understand and cope with the medical, psychological, and familial implications of genetic contributions to diseases [2]. Saskia et al. [3] indicated that GC is essential before and after genetic testing because the result of genetic testing affects not only the patient but also his/her family. On the other hand, Valente et al. [4] indicated that GC prior to diagnostic testing is optimal, but not mandatory for pediatric neurological disorders. According to the 2011 guidelines on genetic testing published by the Japanese Society of Medicine [5], the attending physician should give informed consent to patients who are symptomatic before genetic testing. If necessary, the physician should consider referring his/her patient to receive expert GC so that the patient receives appropriate support when the results are available. Meanwhile, in cases in which the relatives are contacted for genetic testing (e.g., to make a non-progressive carrier diagnosis, pre-symptomatic diagnosis, or prenatal diagnosis), they should also undergo GC. GC is typically carried out in multidisciplinary teams due

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to the wide range of often rare disorders that can be identified by genetic testing. Unfortunately, GC is currently difficult to be performed in the daily clinical setting because many doctors don't have enough time to gain the appropriate skills and knowledge to implement GC in busy daily practice. Therefore, it is necessary to train GC experts and provide resources for GC program development [6]. General practitioners, instead, must understand the value of GC, identify those who require urgent GC, and then refer them to a specialized GC facility [1, 7]. In Japan, expert clinical geneticists (physicians) and certified genetic counselors (non-physicians) take charge of GC. Both experts must be certified by the Japan Society of Human Genetics [8] and the Japanese Society for Genetic Counseling [9]. As of January 2019, there are 1387 clinical geneticists in Japan [10]. Since 2002, these clinical geneticists have been permitted to respond to consultations from attending physicians from any department and conduct the appropriate genetic service (including GC). As of December 2018, there were 243 certified genetic counselors (CGCs) in Japan [11]. Since 2005, these CGCs have been supporting clients with clinical geneticists from an independent stand point. CGCs must study medical genetics and obtain a master's degree in counseling, and can then take employment in many different contexts, including hospitals, universities, and companies. At present, 115 institutions in Japan have a clinical genetics department [12] that is available for consultations with patients or relatives affected by hereditary neuromuscular diseases.

Many refractory neuromuscular diseases, such as Huntington's disease, have been proposed to require special consideration for "refractory" disease during presymptomatic diagnosis [13–15], as well as during non-progressive carrier and prenatal diagnosis [16]. The role of GC without limiting its purpose to only genetic tests needs to be examined to help understand the best methods of collaboration between medical and genetic departments and determine the needs of the genetic division. Such studies will improve genetic literacy as well as national welfare.

Here, we investigated the role of GC in patients with neuromuscular disease who were referred to our hospital and examined the motivations of clients to attend. To the best of our knowledge, no report to date has comprehensively examined the role of GC specifically in clients affected by neuromuscular disease. All GC for relatives seeking non-progressive carrier testing, pre-symptomatic testing, or prenatal testing and preimplantation genetic testing are available in our hospital. However, because some genetic tests requested by the relatives are not available in our hospital, some clients were referred to other institutions after attending GC in our hospital.

Materials and methods

Study design

This exploratory, retrospective, observational study used the information described in the GC medical record.

Patient inclusion

A total of 102 consecutive cases of neuromuscular disease in which GC was performed were initially recruited to this study between July 2005 and March 2018. Patients were excluded if they or their proxy refused to participate (i.e., did not provide informed consent) in this study. No patients refused to participate.

Survey items

Information regarding age, gender, family relationship, target disease, and the presence of accompanying person at GC was taken for each patient. The purpose of GC visit, the motivation of the visit, and the outcome were extracted from individual patient medical records. We extracted the main purpose and motivation for GC visit from medical charts of the cases. The extraction was made by discussion in multiple authors.

Results

Background of the clients attending GC

Our retrospective study included 102 clients who visited the department of clinical genetics, of which 86.3% were women (Table 1). Age distribution ranged from < 20 to > 70 years, with the majority of patients aged 30–39 years (45.1%), followed by the patients aged 20–29 years (30.4%). Most of the individuals who were undergoing GC were relatives (75.5%), of which 74.0% were first-degree relatives (i.e., parents, children, and siblings), and 18.6% were patients with a neuromuscular disorder. Of the neuromuscular diseases covered by the patients, relatives, and other clients, the most common was Duchenne muscular dystrophy (DMD; 22.5%), followed spinocerebellar degeneration (SCD; 17.7%) and myotonic dystrophy (DM1; 13.7%) (Table 2).

Purpose of the visit

Most of the clients requiring GC (29.4%) were looking for an explanation of the disease in question (Fig. 1): here, 26.7% were patients, 60% were relatives, and 13.3% were non-relatives. During GC, we found that the patients themselves (particularly those affected by DM1) also

Table 1 Client characteristics

Total ($N = 102$)		
	N	%
Gender		
Male	13	12.7
Female	88	86.3
Unknown	1	1
Age (years)		
Under 19	2	2.0
20–29	31	30.4
30–39	46	45.1
40–49	7	6.9
50–59	5	4.9
60–69	5	4.9
Over 70	3	2.9
Unknown	3	2.9
Relationship		
Patient	19	18.6
Relatives	77	75.5
First-degree relatives	57	74.0
Parents	28	49.1
Child	17	29.8
Sibling	12	21.1
Second-degree relatives	14	18.2
Aunt/niece	12	85.7
Grandmother/grandchildren	2	14.3
Third-degree relatives	3	3.9
Cousin	3	100
Other relatives	3	3.9
Non-relatives	6	5.9
Partner	3	50
Partner's family	3	50
Companion		
Yes	70	68.6
No	31	30.4
Unknown	1	1.0

Table 2 Eligible disease included in this study

Total ($N = 102$)		
	N	%
DMD	*23*	*22.5*
SCD	*18*	*17.6*
MJD/SCA3	5	
SCA6	5	
SCA1	1	
SCA2	1	
DRPLA	1	
Unknown	5	
DM1	*14*	*13.7*
PMD	*7*	*6.9*
FCMD	*5*	*4.9*
BMD	*4*	*3.9*
XLH	*4*	*3.9*
SMA	*4*	*3.9*
SMA1	3	
SMA2	1	
MtD	*4*	*3.9*
MELAS	2	
MELAS and MERRF	1	
LHON	1	
SBMA	*3*	*2.9*
HD	*3*	*2.9*
ALS	*2*	*2.0*
Others	*11*	*10.8*

DMD Duchenne muscular dystrophy, *SCD* spinocerebellar degeneration, *MJD* Machado–Joseph disease, *SCA* spinocerebellar ataxia, *DRPLA* dentatorubralpallidoluysian atrophy, *DM1* myotonic dystrophy, *PMD* Pelizaeus–Merzbacher disease, *FCMD* Fukuyama congenital muscular dystrophy, *BMD* Becker muscular dystrophy, *XLH* X-linked hydrocephalus, *SMA* spinal muscular atrophy, *MtD* mitochondrial disease, *MELAS* mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, *MERRF* myoclonus epilepsy associated with ragged red fibers, *LHON* Leber's hereditary optic neuropathy, *SBMA* spinal and bulbar muscular atrophy, *HD* Huntington's disease, *ALS* amyotrophic lateral sclerosis

requested for a more detailed explanation about the disease (Supplementary Fig. 1a). DMD was the most common disease (38.5%) for which the affected clients wanted GC prior to a prenatal diagnosis (38.5%). Here, two women had suspected DMD based on high creatine kinase values and muscle weakness and were diagnosed as symptomatic carriers. There were two diseases (amyotrophic lateral sclerosis and SCD) for which prenatal diagnosis was not performed in our hospital (Supplementary Fig. 1b). Of the 18 clients (17.6%) who underwent GC for pre-symptomatic diagnosis, the majority were suspecting to have SCD (44.4%). Only two clients (11.1%) with HD have been reported by many previous studies as having undergone a pre-symptomatic

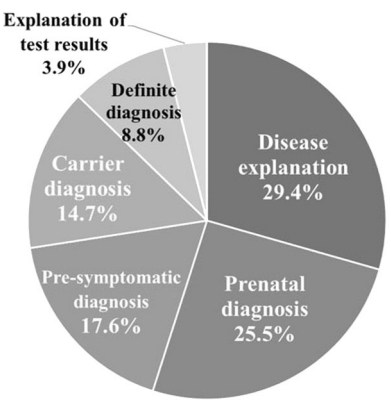

Fig. 1 The purpose of genetic counseling

diagnosis. Only one client required consultation for a disease (namely, familial amyloid polyneuropathy) in which a disease-modifying therapy was established. Because of the degree of onset, mitochondrial disease was difficult to be predicted from genetic testing results and was thus not applicable for obtaining a pre-symptomatic diagnosis (Supplementary Fig. 1c). Finally, 15 (14.7%) clients underwent GC for a non-progressive carrier diagnosis: all of these cases were with regard to X-linked diseases (Supplementary Fig. 1d). Of the remaining clients, nine wanted to obtain a definitive diagnosis by genetic testing and four wanted a deeper explanation of the test results.

Motivation to attend GC

Patients themselves

We next examined the opportunities due to which the clients had to attend the hospital for GC. The affected patients themselves formed the majority (36.8%) of the group that had the most opportunity for GC because they had considered genetic testing. Among them, there were patients who had been given a clinical diagnosis of SCD despite not carrying pathogenic variants with a high frequency for this disease. In addition, there were patients who were suspected symptomatic carriers of DMD but exhibited rare clinical symptoms that could not be managed at local hospitals. One patient attended GC to discuss the timing of genetic testing as they were considering the consequential effects on their relatives. Three clients (15.8%) visited after receiving a genetic diagnosis, seeking a more detailed explanation of the disease and to discuss about the best timing to communicate the results with their relatives. Five clients (26.3%) attended GC as a result of pregnancy and/or with a desire to bear a child. Here, the clients wanted to discuss the genetic influence on the next generation and their options for a prenatal diagnosis. Two clients (10.5%) attended because relatives wanted a pre-symptomatic diagnosis, one client (5.3%) consultation from the attending physician and another client (5.3%) attended because she wanted to obtain hospital information where she should be examined (Supplementary Fig. 2a).

Relatives of patients

The majority of relatives (51.9%) attended GC on the basis of pregnancy and/or desire to bear a child: 21 clients (52.5%) considered prenatal diagnosis, but some considered pre-symptomatic diagnosis or non-progressive carrier diagnosis for their own risk assessment. For 20 clients (26.0%), either a genetic diagnosis had recently been made on the proband or the proband showed a worsening condition. In these cases, the purpose of the GC visit ranged from requests to explain the disease to requests for further genetic testing. The motivation greatly depended on the relationship with the proband and the amount of prior access to disease information. Many clients themselves and/or their relatives went through a life event (such as marriage (10 clients), becoming adults (two clients), or retirement (one client)) and thus they wanted to determine whether or not they were in an "at-risk state" before informing their relatives about their genetic condition (Supplementary Fig. 2b).

Non-related clients

Receipt of an affected spouse's genetic diagnosis (two clients), impending pregnancy, desire to bear a child (three clients), and consideration of marriage (one client) were all triggers to attend GC for unrelated individuals. Here, some clients underwent GC without informing their partners about the visit (Supplementary Fig. 2c).

Case presentation by purpose

Explanation of the disease

Case 1 Twenty-seven-year-old female (Fig. 2a; III-2).

Purpose of GC To understand more about DMD.

History prior to GC consultation This client's younger brother (III-4) was diagnosed with DMD at 3 years of age. His parents had reported only the disease name to the client when she was a junior high-school student. The client's older sister (III-1) was a carrier, but lived far away and had no opportunities to talk with the client. The client recently learned that DMD was a hereditary disorder and received a positive carrier diagnosis at another facility. She visited our hospital with her husband (III-3) to gain a detailed explanation of the disease.

Contents of GC session We provided the patient with an outline of the disease and how it is inherited. We explained that there was a possibility that women might have mild symptoms and that the recent developments for new therapies based on gene therapy were advancing. We also explained the options for prenatal and preimplantation diagnosis, and if she liked to have a baby, we would schedule further GC with obstetricians. The client later requested for a prenatal diagnosis and returned for GC.

Prenatal diagnosis

Case 2 Nineteen-year-old female (Fig. 2b; IV-6).

Purpose of GC To discuss the implications of a prenatal diagnosis of DM1.

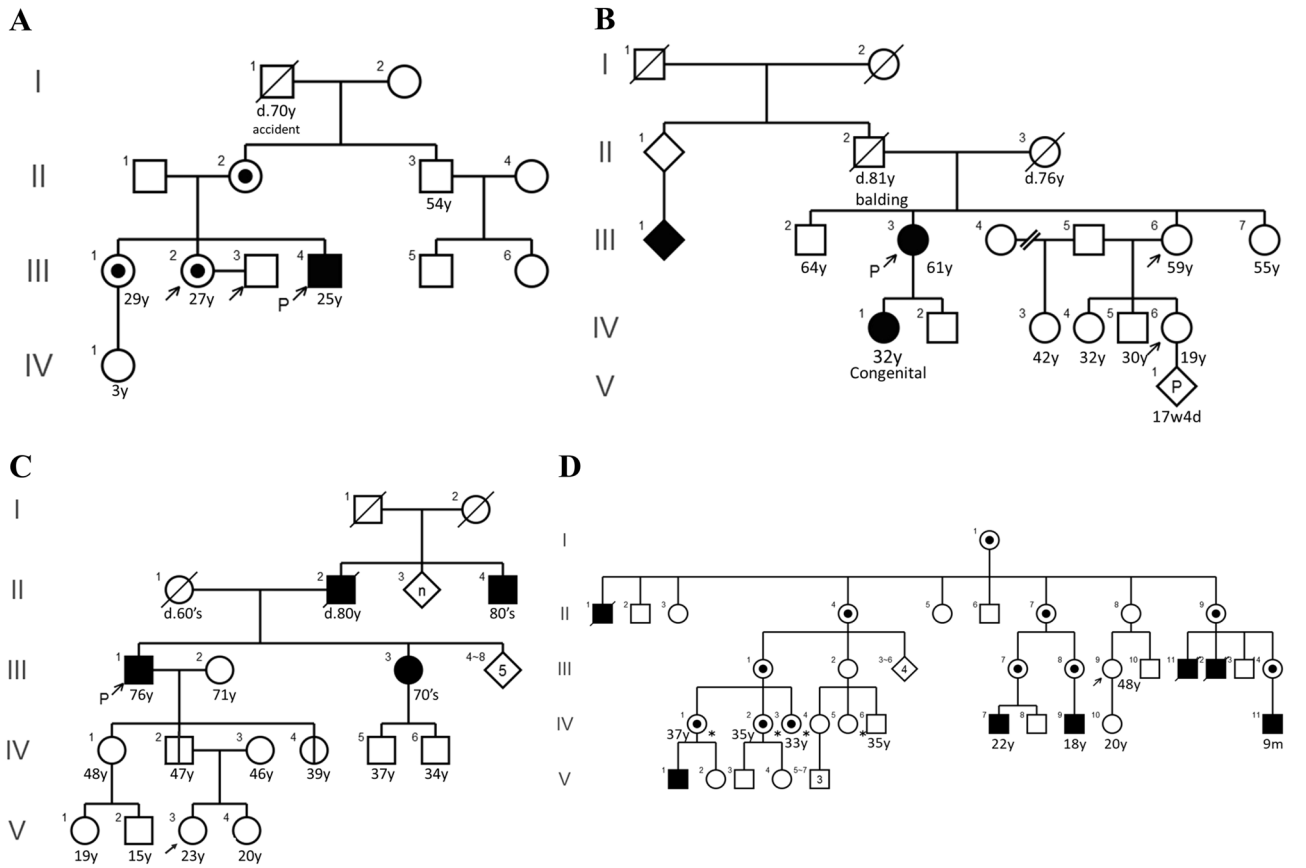

Fig. 2 GC client family trees. Ages are shown under each symbol where available. **a** DMD cases. The proband is III-4. The client is III-2, who is accompanied by III-3. **b** DM1 cases. The proband is III-3. The client is IV-6, who is accompanied by III-6. **c** SCA6 cases. The proband is III-1. The client is V-3. **d** Pelizaeus–Merzbacher disease cases. The client is III-9. Document evaluation (*) was used for someone who underwent genetic testing. DM1, myotonic dystrophy; DMD, Duchenne muscular dystrophy; GC, genetic counseling; SCA6, spinocerebellar ataxia 6

History prior to GC consultation The client was 17 weeks pregnant when she consulted the department of clinical genetics. Her maternal aunt (III-3) and cousin (IV-1) were diagnosed with DM1, but genetic testing was not conducted. The client knew that DM1 was hereditary, but she did not understand the details and was worried about the implications for her unborn baby. The clinical geneticist made the judgment that the client or her mother (III-6) had no evident symptoms indicative of DM1.

Contents of GC session We explained that there was no possibility that the fetus would develop the disease if the client or her mother was not a carrier of DM1. However, we could not eliminate the possibility that the client or her mother was a DM1 carrier only from subjective symptoms as the symptoms may not be noticeable when mild. We gave our opinion that it was necessary for her maternal aunt to have a genetic diagnosis to warrant a prenatal diagnosis. The prenatal diagnosis needed an application to be approved by the Institutional Review Board of our hospital, but it took time to acquire. The client understood the situation and decided against a prenatal diagnosis. Because there was a chance that the client had DM1 without overt symptoms, we decided to manage her pregnancy at our hospital. We also referred the client's mother to a neurologist to confirm whether she had any clinical symptoms and if she should consider genetic testing.

Pre-symptomatic diagnosis

Case 3 Twenty-three-year-old female (Fig. 2c; V-3).

Purpose of GC Consultation for obtaining a pre-symptomatic diagnosis of spinocerebellar ataxia 6 (SCA6).

History prior to GC consultation This client's paternal grandfather (III-1) had a clinical diagnosis of SCA6. A genetic diagnosis was made when her paternal aunt (IV-4) wanted a pre-symptomatic diagnosis. After several GC sessions, genetic testing was conducted and it revealed a positive result. The patient's father (IV-2) understood that it was possible that he was SCA6 positive and also requested for a

pre-symptomatic diagnosis. Again, genetic testing was performed after several GC sessions, and the result was positive. Her paternal aunt (IV-1) also underwent GC, but she did not tell her children that the family had a history of a hereditary disease, and pre-symptomatic diagnosis at that time was not conducted. Knowing that her father was positive, the client considered the possibility that she may also have inherited the condition.

Contents of GC session We confirmed the history of SCA6 and explained the impact of genetic testing. The client described that she had learned from her parents at a young age that her grandfather's illness was hereditary, and she assumed that she would also develop the disease. She was not worried about the genetic influence on the next generation at that time because she had no desire to marry at that time. If the result was positive, the client explained that she would consult with her employer about her future work, and if it was negative, she would undertake nursing care for her affected family members, including her father. After several GC sessions, genetic testing was performed and the result was negative. Her sister (V-4) also had GC, but she did not want a pre-symptomatic diagnosis.

Non-progressive carrier diagnosis

Case 4 Forty-eight-year-old female (Fig. 2d; III-9)

Purpose of GC Consultation for a non-progressive carrier diagnosis of Pelizaeus–Merzbacher disease (PMD).

History prior to GC consultation The client's family included several sufferers of PMD, including her maternal uncle and cousin. When the client's daughter reached 20 years of age, she considered the possibility that her daughter might marry and have children and thus underwent GC in the hope of obtaining a carrier diagnosis by genetic testing.

Contents of GC session The client understood the outline of the disease as there were multiple persons diagnosed as carriers in her family. She had not talked to her daughter about PMD in detail. We advised her that if she was a carrier, she should consider the best timing for genetic testing and discussion of the consequences of her diagnosis with her daughter. Her husband also agreed to take a carrier diagnosis. Genetic testing was performed and the result was negative.

Discussion

This study is the first to comprehensively investigate the needs of GC in the context of neuromuscular disease without limiting its purpose as only a requirement for a genetic test. Previous reports from other countries have indicated that most neurologists have no training in genetic diagnostics, but that education on clinical genetics is becoming more important as the complexity of genetic testing is increasing [17, 18]. Our clients had a variety of needs, identifying that neurologists need to provide not only the knowledge of the neuromuscular disease but also the knowledge on clinical genetics and refer the client to a higher-order GC at an appropriate time.

As a standard, we consider it necessary to confirm the amount of knowledge and concept of each disease held by the client. For some neuromuscular diseases, disease-modifying therapies are available: we consider it important to provide up-to-date information about treatment options.

Overall client characteristics and GC purpose

Our analyses identified that the majority of clients attending GC were women (86.3%). This gender bias may explain why pregnancy and desire to bear a child comprised most of the motivations for GC sessions. Most clients were aged between 20 and 30 years (75.5%), which is consistent with the previous reports that the predominant age range of clients undergoing GC for a pre-symptomatic diagnosis is between 20 and 39 years [15, 19, 20]. This young age encompasses the time when most individuals consider marriage and try for a child. Regardless of the precise purpose for visiting, we found that many patients themselves or their relatives were motivated to attend GC due to a recent or impending life event (case 4).

Many clients attended GC to gain a clear explanation of the genetic disease. Despite the patients or their relatives themselves being affected by a particular disease, some clients were not aware of the precise pathophysiology (case 1), and the clinical geneticists and/or CGCs needed to organize the relevant information. DM1 was the most frequent disease that required explanation by the clinical geneticists and/or CGCs, perhaps because the clinical symptoms of DM1 are diverse. Anticipation in genetic disorders is the phenomenon in which the signs and symptoms of a genetic condition become more severe and present from an earlier age with each subsequent generation. With this in mind, we considered that repeat expansion was extended to the next generation and different clinical symptoms were exhibited due to genetic anticipation. Several neuromuscular diseases exhibited by the clients showed genetic anticipation, suggesting that inputs from the neurological department, and other relevant medical departments, would be beneficial for GC.

Those considering a prenatal, pre-symptomatic, or carrier diagnosis had typically learned of the presence of a

hereditary disease in the family line. However, some clients were diagnosed as symptomatic carriers without any family history and they later considered a prenatal diagnosis (Supplementary Fig. 1b).

GC for patients

Approximately 20% of the clients attending GC were patients, of which many wanted clarity on the disease and to obtain a definitive diagnosis. A genetic diagnosis of relatives who are non-symptomatic can only be performed when a pathogenic variant is observed in the proband. Some patients requested a genetic diagnosis and attended GC (case 3: III-1) but some relatives wanted a genetic diagnosis so that genetic testing of the proband was not enforced.

In the 2013 survey of neurologists in Japan, 43.2% of the neurologists were reluctant to make a genetic diagnosis for DM1 and 30.4% were reluctant to make a genetic diagnosis for HD [1]. The reasons for this opinion were that it is "possible to diagnose without genetic testing," "not linked with effective prevention or therapy," and "difficult to support after disclosure of the results." Based on these numbers, there will be a considerable number of patients with a clinically diagnosed neuromuscular disease who have not been genetically diagnosed. When relatives desire a genetic diagnosis, the person in charge of GC may create an opportunity to re-examine the genetic diagnosis in cooperation with the physician of the proband.

Some of the diseases presented at our hospital that required a genetic diagnosis were rare; here, it was hoped that the patient would consent to participate in research using whole-exome sequencing and whole-genome sequencing after GC. In Japan, a genetic diagnosis following suspected clinical symptoms is usually made by Sanger sequencing technique. Many neuromuscular diseases, however, exhibit genetic and phenotypic heterogeneity. As the demand for comprehensive genetic testing is high, and as the costs and timing of next-generation sequencing are decreasing, it is expected that comprehensive genetic diagnoses made by next-generation sequencing will increase [21]. Whole-exome sequencing and whole-genome sequencing are required to interpret the results and respond to secondary findings, making it difficult to conduct in general practice. We expect, therefore, that the number of cases of neuromuscular disease requiring involvement from clinical genetics departments from the confirmed diagnosis stage will increase.

GC for relatives

Most relatives who underwent GC were first-degree relatives (74.0%), of which half were parents. Many relatives had a first child with an inherited disease, and they wanted to discuss about the potential outcomes for a second baby when pregnant. Many relatives also wanted a prenatal diagnosis, but a prenatal diagnosis that involved an invasive examination (such as chorionic villus sampling or amniocentesis) was only granted when the requirements proposed by the Japan Society of Obstetrics and Gynecology were met [22]. In cases in which it is unclear whether one or both parties in a couple are carriers of a genetic disease, a prenatal diagnosis cannot be granted; an ethics committee must judge whether a case is serious or not.

Of the 26 relatives (25.5%) who wanted a prenatal diagnosis, 19 (73%) visited after pregnancy was established. There were four cases (15.4%) in which the examination could not be performed due to time constraints (case 2). Neurologists attending to patients with a hereditary disease must understand the process of prenatal diagnosis, and relatives should be properly guided when they consider genetic testing.

In cases in which a child attended GC, it was typically motivated for wanting a pre-symptomatic diagnosis. A study conducted in Japan in 2007 found that DM1 was the most frequent neuromuscular disease for which a pre-symptomatic diagnosis was requested [13]; rather, we found that SCD was the most frequent disease in our hospital. This discrepancy may be because three of those who wanted a pre-symptomatic diagnosis of SCD were from the same family (case 3). As a result of one family member's pre-symptomatic diagnosis, other family members might consider a simultaneous examination, making it necessary to carefully proceed with GC while confirming the situation of each family member. We found that many second-degree relative referrals considered a carrier diagnosis: some clients underwent GC with little information on the disease because the proband had already died. As required for patients, it was necessary to confirm the amount of knowledge held for each disease.

One case at our hospital involved a proband who had just been genetically diagnosed and another case in which the deterioration of the symptoms triggered a GC inquiry. A previous report has described the burden on caregivers and how the level of distress increases when symptoms deteriorate in the proband [23]. We should, therefore, consider the possibility that changes in the proband's symptoms might trigger caregivers to consider themselves also to be at risk and seek GC.

GC for non-relatives

GC is available to address all genetic concerns and worries, and so clients unrelated to an affected patient may also have concerns that warrant undergoing GC. In this study, non-relatives accounted for 5.9% of the clients. Unlike patients and relatives, non-relatives had only few chances to consult

with the department of clinical genetics. There are some topics that GC cannot solve, but supporting autonomous decision making by providing clear disease understanding and advice to help address the current problem is the key role of GC.

Study limitations

Our hospital has pediatric and obstetric departments, and is the core medical facility in the area. A high proportion of clients were motivated to undergo GC due to pregnancy and/or a desire to bear a child, but in Japan there are limited institutions in which a prenatal diagnosis is possible, and the possibility that cases are gathered in one of only a few facilities should be taken into consideration. In fact, a prenatal diagnosis was possible for 57.8% (59/102 diseases) of the diseases in our hospital. Thus, our dataset may have a bias towards prenatal diagnoses. Finally, our hospital has been conducting SCD genetic testing as part of a long-term ethics committee-approved study; thus, we must consider a potential bias towards SCD cases in our dataset.

Future tasks

Most of the clients had a knowledge of genetic risk and were seeking GC when a life event had occurred and had experienced psychosocial changes. However, while some clients in a family chose to undergo GC following a life event, some did not (case 3). This effect may be due to the type of event and the level of understanding of genetic risk exhibited by each individual. A future study should examine the method of GC intervention according to the client's cognitive level. Ours is the first study restricted to hereditary neuromuscular diseases, and the characteristics of these diseases may be reflected in the background of the visit. Future studies should consider that the role of GC in neuromuscular diseases will become clearer as GC is conducted in other areas, such as in the context of cancers or cardiovascular diseases.

Conclusion

In summary, this study has examined the nature of GC required by patients with hereditary neuromuscular diseases and their relatives/non-relatives. We found that many clients were motivated to undergo GC in response to a life event. Practitioners should, therefore, be aware of the events that affect not only the patient but also their relatives and guide GC at an appropriate time. Because of the diverse requirements of each client, neurologists should be equipped with the standard knowledge on clinical genetics and neurological diseases. Depending on the client, the type and the level of recognition of genetic risk will differ, meaning that the clinical geneticists and/or CGCs should thoroughly understand the present situation of the client and provide a tailored treatment.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Ethics approval This study was conducted with the approval of the Hokkaido University Hospital Institutional Review Board.

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