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ORIGINAL ARTICLE

Decision-making processes behind seeking regular cardiac checkups for individuals with Marfan syndrome: A grounded theory study

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Abstract

Patients with Marfan syndrome (MFS) present with various symptoms, such as aortic aneurysm/dissection, tall stature, and lens deviation. Among them, acute aortic dissection is a complication that leads to sudden death. Some individuals with MFS are reluctant to see a cardiologist and discontinue regular checkups until they develop life-threatening complications. We conducted a grounded theory study to investigate how individuals with MFS decided whether to adhere to healthcare recommendations, specifically to attend cardiology appointments. The study recruited individuals with a clinical or genetic diagnosis of MFS from a Japanese university hospital and individuals from a support group. Semi-structured interviews were conducted with 28 consenting participants. In this study, we identified the decision-making processes of individuals with MFS concerning their cardiology visits. We extracted "perception of the gap between their health status and medical recommendations" as the central category. This decision-making process consisted of three parts: (A) the process by which an individual with MFS sees a cardiologist for the first time, (B) the process by which an individual with MFS keeps up with cardiology checkups, and (C) the process by which parents bring their children with MFS to the cardiologist. Individuals who learned of the possibility of MFS decided whether to adhere to medical recommendations depending on how they perceived the gap between their health status and the medical recommendations. In addition to medical information and treatment experience, adaptation to MFS, which changed through interactions with others, influenced the perception of the gap. This study suggests the role of genetic counseling and molecular genetic diagnosis as factors that may facilitate adaptation to MFS. The involvement of genetic counselors is important for helping individuals with MFS keep up with regular checkups while affirming their own experiences. These results provide insight into adherence to medical recommendations for individuals with MFS.

KEYWORDS
decision-making, genetic counseling, health behavior, Marfan syndrome, patient adherence, risk management

1 | INTRODUCTION

Marfan syndrome (MFS) is an autosomal dominant hereditary connective tissue disease caused by mutations in the *FBN1* gene (Dietz et al., 1991). Its incidence is approximately 1 in 5000 people. The Japan Intractable Diseases Information Center ("Overview of Marfan syndrome, Diagnostic Criteria, etc.," 2015) has estimated that there are 15,000–20,000 individuals with MFS in Japan. The Ghent criteria, revised in 2010, are used worldwide as the main clinical diagnostic criteria (Loeys et al., 2010). Japan also follows the Guidelines on Diagnosis and Treatment of Aortic Aneurysm and Aortic Dissection (Japanese Circulation Society, Japanese Society for Cardiovascular Surgery, Japanese Association for Thoracic Surgery, and Japanese Society for Vascular Surgery, 2020) regarding the treatment of MFS. In Japan, MFS has been designated as an intractable disease and a specific pediatric chronic disease since 2015, and therefore, individuals with MFS are eligible for public support. *FBN1* genetic testing has been covered by the National Health Insurance of Japan since 2016.

MFS presents with various age-related symptoms, including cardiovascular, skeletal, and ocular symptoms, requiring the involvement of many clinical departments. Managing aortic dissection requires lifelong surveillance, lifestyle modifications, drug therapy, and multiple surgical procedures. Advances in diagnosis and management have significantly improved the prognoses of affected individuals (Pearson et al., 2008). MFS is an autosomal dominant condition and multiple mutation carriers may exist in a family; if one family member is diagnosed with MFS, early diagnosis and treatment of other relatives is recommended.

However, diagnosis may be delayed until the development of life-threatening complications (Caglayan & Dundar, 2009; Summers et al., 2006). Many patients return to medical institutions after the interruption of continuous medical care only when the aortic aneurysm/dissection becomes severe (Takeda, 2018). Individuals with MFS and their families require continual support after diagnosis to understand and adapt to medical and genetic information.

It was reported that adherence in patients with genetic aortic disorders is related to psychological adjustment (Connors et al., 2015). However, the ways that interactions between individuals with MFS and others affect adaptation have not been investigated. Additionally, in Japan, previous reports have only referred to physical and psychosocial situations in daily life (Sato & Teramachi, 2010; Shimizu et al., 2017), and there is a lack of research on the decision-making processes regarding adherence to healthcare recommendations by individuals with MFS.

Genetic counseling is the process of helping people understand and adapt to the genetic involvement of disease, and genetic counselors are responsible for assisting individuals who have genetic traits, as well as their families. Genetic counseling is recommended as an important part of multidisciplinary care for individuals with hereditary cardiovascular diseases and their families (Ahmad et al., 2019). An increasing number of cardiovascular clinical guidelines recognize the importance of the cardiac genetic counselor in patient care (Hershberger et al., 2018; Ommen et al., 2020; Towbin et al., 2019). However, Japanese guidelines do not mention the role of genetic counselors for patients with cardiovascular disease.

It is important to understand how Japanese patients with genetic cardiovascular disease make decisions about receiving medical care and what factors influence the decision-making processes. Therefore, this study aimed to (1) determine how individuals who recognize the possibility of MFS, while interacting with others in their lives, decide to seek healthcare and maintain cardiovascular visits and (2) examine the role of genetic counselors and genetic medicine departments in Japan.

What is known about this topic

Regular cardiovascular examinations are important for individuals with MFS to prevent sudden death from acute aortic dissection. Some people are reluctant to seek medical care despite recognizing the possibility of having MFS. They opt to stop receiving it once it starts or seek medical attention at a medical institution only after experiencing a severe cardiovascular event.

What this paper adds to the topic

The involvement of the healthcare team, family, and community influences the understanding and adaptation of individuals with MFS, which influences their adherence to healthcare recommendations. Genetic counselors can help affected individuals continue attending checkups for MFS while affirming their experiences.

2 | METHODS

2.1 | Participants

The inclusion criteria were as follows: age 16 years or older, a self-reported clinical or genetic diagnosis of MFS, and the ability to complete an interview in Japanese. Participants were recruited in two phases. Phase I: Between July 2020 and September 2021, physicians of the MFS treatment team at Kyoto University Hospital provided explanatory materials to patients or their families who met the eligibility criteria. Those who contacted us to participate were provided information about the study by SH, who is responsible for this study, and a genetic counselor who studied genetic counseling during their postgraduate training and has been certified since November 2021. Those who agreed to participate in the study were interviewed between September 2020 and October 2021. Phase II: Between February 2022 and November 2022, research participants were recruited through the mailing list of the Japan Marfan Association (one of the MFS support

groups in Japan), its website, and social events. The interviewer explained the study to those who contacted us to participate. Those who agreed to participate in the study were interviewed between February 2022 and November 2022. Participants of Phase I had similar attributes (age, medical history, etc.), and the data were below theoretical saturation. Therefore, we initiated a Phase II recruitment effort to recruit patients with different demographic characteristics. To enrich the variations in Phase II, we recruited for individuals who were diagnosed at a younger age and those who had discontinued regular cardiology checkups. And we determined that theoretical saturation was achieved and no further recruitment was necessary.

The interview duration ranged from 44 to 95 min, with a median of 66 min. Interviews were conducted by telephone or online (Zoom Cloud Meetings or Google Meet) depending on the participants' preferences. Each interview was conducted one-on-one between a participant and the interviewer. The interviewer used a private room where third parties could not hear the conversation. Each interview was recorded with a digital audio recorder and transcribed verbatim. All interviews were conducted by SH, who received training in qualitative research from a senior researcher (MT, a certified genetic counselor).

2.2 | Interviews

The main topics can be outlined as follows: (1) history of medical visits and treatment, (2) thoughts on continuing medical care, (3) thoughts on health management, and (4) expectations of medical care and medical professionals. In our interviews, we focused on participants' behavior after learning about the possibility of having MFS and their motivations and obstacles in continuing medical care. SH took field notes during and after the interviews.

2.3 | Procedures

This study was approved by the Ethics Committee at the Kyoto University Graduate School and Faculty of Medicine (approval number: R2501-3). All the participants provided informed consent to participate in the interviews. Before starting the interview, the interviewer explained the purpose of the study and verbally reaffirmed their consent to participate and their right to withdraw at any time.

2.4 | Data analysis

This study used grounded theory (Strauss & Corbin, 1998), which is "an approach that aims to capture the diversity of processes involved in how people perceive and respond to a particular situation, the actions or interactions that arise, and how the situation changes as a result" (Saiki-Craighill, 2016). Grounded theory is based on symbolic interactionism (Glaser & Strauss, 1967) and focuses on clarifying social change through social interactions and processes, considering the conditions, actions/interactions, and consequences (i.e., paradigms) (Saiki-Craighill, 2016; Strauss & Corbin, 1998).

The grounded theory method involves generating verbatim transcripts, reading data carefully, categorizing them based on meaning, abstracting concepts, identifying properties and dimensions, labeling categories based on properties and dimensions, and comparing and organizing them. Through these preliminary stages, we examined a paradigm of the decision-making process involved in cardiology visits using paradigm conditions, actions, interactions, and consequences. Once the relationships between categories were identified within the paradigm, we created a Category Relationship Diagram (CRD) to visualize the rationale behind their connections (Saiki-Craighill, 2016). CRD involves linking multiple categories using properties and dimensions to illustrate the interdependencies that drive the processes between these categories. After following these steps for a single interview dataset, we looked at what data was missing variations. From that information, we considered who we should recruit as the next participants and what questions we should ask in the next interview. In grounded theory, CRDs related to the same phenomenon (in this study, the "decision-making process in cardiology visits") are stacked together to eventually form a unified Category Relationship Integration Diagram (CRID). For instance, we got one CRD from the first dataset, and another CRD from the second dataset, and we merged the two CRDs to create one CRID. This process was repeated for each additional dataset, culminating in a final CRID (Figure 1), which included the decision-making processes of all 28 participants.

SH conducted all the labeling, categorization, and associations, and MT checked their validity. The analysis results were reviewed by four researchers with experience in genetic medicine and MFS, namely SH, MT, and two other clinical geneticists (TW and HK). They discussed any disagreements until a consensus was reached. MAXQDA2020 (VERBI GmbH, Japanese version) was used for data management. Categories and quotes were translated into English after completing the analyses in Japanese. SH and TW reviewed the accuracy of these translations. Participants were not asked to review transcripts or comment on the findings.

3 | RESULTS

3.1 | Participants' demographics

At Kyoto University Hospital, 31 individuals who met the inclusion criteria were recruited by their attending physicians, and 14 consented to participate in this study (Phase I). An additional 14 participants were recruited through the Japan Marfan Association (Phase II). Educational level and socioeconomic status were not obtained. All participants were interviewed once. Of all the

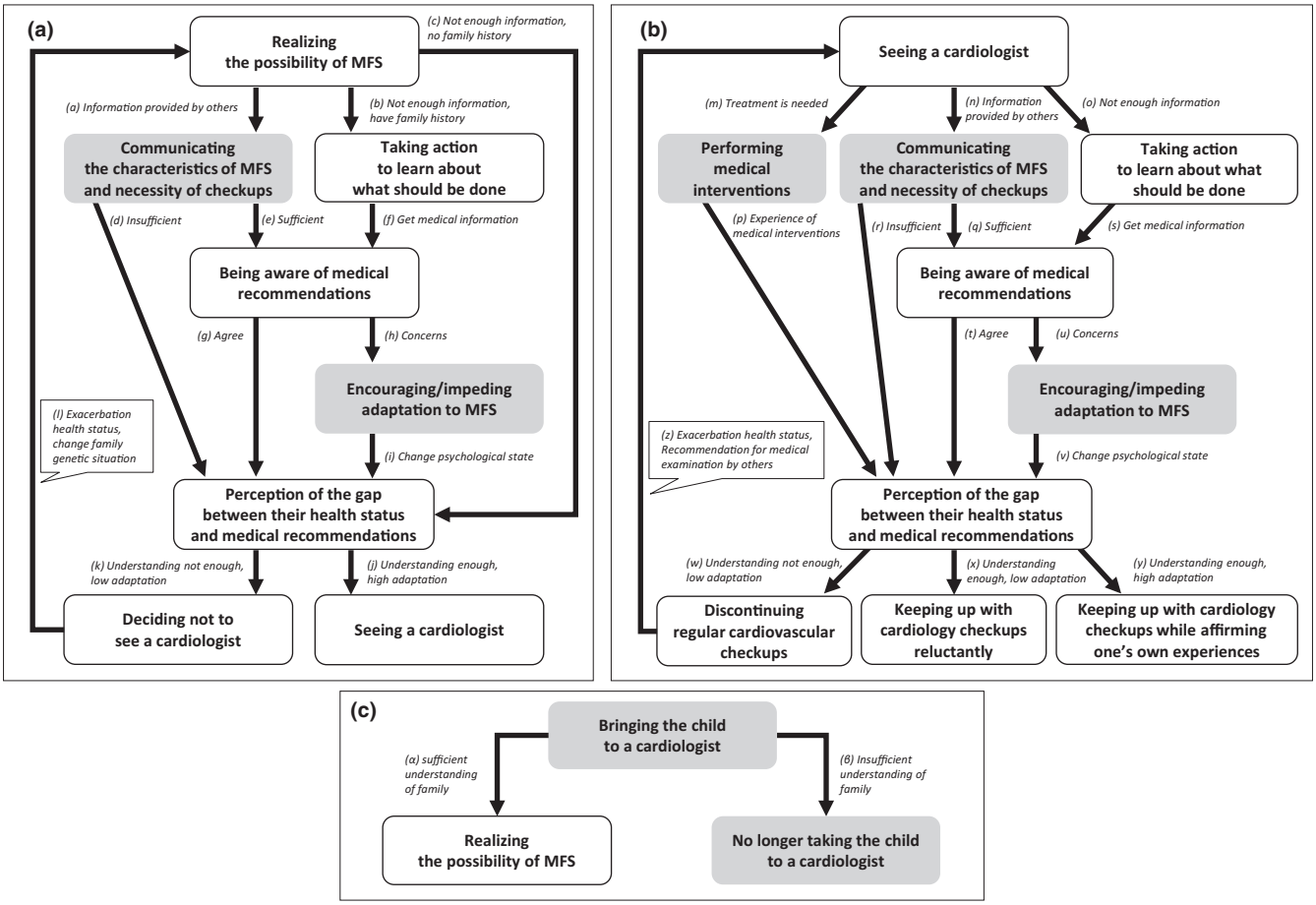

FIGURE 1 The decision-making processes for individuals with MFS during cardiology visits. (a) The process by which an individual with MFS sees a cardiologist for the first time (process a). (b) The process by which an individual with MFS keeps up with cardiology checkups (process b). (c) The process by which parents bring their children with MFS to the cardiologist (process c). Each box represents each category. The subject of the white box is an individual with MFS; the subject of the gray box is others. The arrows between boxes represent how categories are related through specific properties and dimensions. Generally, the thematic diagrams relate categories using multiple properties and dimensions (Saiki-Craighill, 2016). However, in the diagrams in this paper, only a summary of the main dimensions (in italics beside arrows) is provided due to space and visibility.

interviews, 11 were conducted by telephone, and 17 were conducted by online teleconferencing, depending on the participants' preferences. Among the participants, 12 (42.9%) underwent genetic testing, all positive for *FBN1* mutations, and 21 (75%) had undergone cardiovascular surgery. More participant demographic information is described in Table 1 and Appendix S1.

3.2 | Categories and processes descriptions

The decision-making process (phenomenon) related to cardiology visits for individuals with MFS was identified by analyzing the interview data. In grounded theory, a central category represents and explains the phenomenon under study. In addition, the name or phrase used to describe the central category should be sufficiently abstract that it can be used to research other substantive areas, leading to the development of a more general theory (Strauss & Corbin, 1998). This study identified the central category as "perception of the gap between their health status and medical recommendations."

The phenomenon involves 14 categories consisting of 1645 labels (codes). Among the 14 categories, 9 are related to individuals with MFS and 5 are related to others (see Appendix S2). The term "others" refers to all people surrounding an individual with MFS, comprising cardiologists and other physicians, genetic counselors, medical staff, family members/relatives, friends, and other MFS patients.

The phenomenon consisted of three parts: (A) the process by which an individual with MFS sees a cardiologist for the first time, (B) the process by which an individual with MFS keeps up with cardiology checkups, and (C) the process by which parents bring their children with MFS to the cardiologist. Each part is described below, according to the categories (in bold). Several translated quotes from the interviews are shown in the block text. Lowercase letters in the discussion below correspond to those in Figure 1 and Appendix S2.

TABLE 1 Participants' demographics ($n = 28$).

Demographic variable	*n* (%)
At the time of learning the possibility of having MFS	
Age	
<20 years	12 (42.9)
20–29 years	5 (17.9)
30–39 years	4 (14.3)
40–49 years	4 (14.3)
50–59 years	2 (7.1)
60–69 years	0 (0)
>70 years	1 (3.6)
At the time of survey	
Age	
<20 years	0 (0)
20–29 years	1 (3.6)
30–39 years	5 (17.9)
40–49 years	10 (35.7)
50–59 years	6 (21.4)
60–69 years	4 (14.3)
>70 years	2 (7.1)
Sex	
Female	16 (57.1)
Cardiovascular symptoms present[a]	24 (85.7)
History of cardiovascular surgery present	21 (75)
Experience of stopping medical visits present	6 (21.4)
Current department for medical visits	
Cardiology	26 (92.9)
Ophthalmology	12 (42.9)
Orthopedics	7 (25)
Departments where medical visits were previously conducted	
Cardiology	28 (100)
Ophthalmology	22 (78.6)
Orthopedics	13 (46.4)
Family history present	16 (57.1)
FBN1-positive	12 (42.9)
Genetic counseling experience present	13 (46.4)

[a]People for whom cardiovascular disease was previously indicated include dilatation tendencies of the aortic root (self-reported).

3.2.1 | The process by which an individual with MFS sees a cardiologist for the first time (process A)

Realizing the possibility of MFS

Many individuals were made aware of the possibility of MFS by their doctors or family members. Most had never heard the name "Marfan syndrome" and could not imagine what the condition was.

P12: At that time, my cardiothoracic surgeon told me, "You probably have Marfan syndrome." This was the first time I realized that such a thing could happen to me.

At the same time, participants and family members sometimes learned about the characteristics of MFS and the need to see a cardiologist (letter a, in Figure 1a).

When information from physicians or family members was unavailable, the ability to perceive MFS depended on personal experience and family history. Individuals who were aware of their genetic condition or had a family history were likelier to take further action to learn what to do (b). However, if they did not recognize their genetic condition or had no family history, they may decide to see a doctor without any knowledge of MFS (c).

P7: I remember the doctor who conducted the retinal detachment surgery and mentioned "Marfan syndrome." However, he did not recommend seeing other specialists or taking a CT scan. As a result, I believed that MFS was not a severe illness. Since no one in my family had MFS, my family and I were slightly confused.

Communicating the characteristics of MFS and the necessity of checkups

Physicians and family members informed individuals with possible MFS of the risk of cardiovascular symptoms and the need to see a cardiologist. However, the way or degree to which the risk of cardiovascular symptoms was communicated varied.

P14: The doctor said the most dangerous thing is having an aortic dissection like my father. He recommended that I should have regular checkups on my heart anyway.

P8: I was told I did not have to worry about cardiovascular symptoms while I was still young.

P18: The doctor did not seem to understand MFS well. He only said, "Oh, that is a serious illness." He explained that my physical condition may change suddenly, and that's all.

If the medical information communicated by others was insufficient or inaccurate, the individual with MFS sought medical attention without fully understanding the medical recommendations (d). When medical information was adequately correct, they understood the need for cardiology visits (e).

Taking action to learn about what should be done

Participants who were identified as having a possible MFS diagnosis gathered information on their own to determine how likely it was to be MFS and what should be done.

P3: Before my surgery, I was curious about what the doctor meant by "Marfan syndrome," so I looked it up on the internet. Almost all the physical features of MFS that I read about matched my own. Therefore, I was certain that I had MFS.

Those who obtained correct information were more likely to learn about the general cardiovascular risks of MFS and medical recommendations (f).

Being aware of medical recommendations

Participants learned that MFS requires regular visits to a cardiologist through information provided by others or gathered by themselves. Upon learning of their cardiovascular risk, several individuals felt fear and anxiety.

P8: If there is a way to delay the onset or reduce the risk of aortic dissection, seeing a cardiologist for checkups is necessary.

P23: I studied MFS by reading household medical books. So, I only knew about aortic dissection. However, I never imagined that I would suffer from it.

P10: I understood that not everything written online was necessarily true. However, when I read descriptions such as "individuals with MFS cannot live beyond 40 years old," I felt that I would not be able to live for long.

After learning and agreeing with the general medical recommendations, they considered whether the recommendations also applied to them (g). Some asked others for advice before seeking a doctor (h).

P13: After discovering that I might have MFS, I got to know someone with MFS through social networking. I asked them for advice on many things.

Encouraging/impeding adaptation to MFS

Sometimes, participants consulted others who helped them with acceptance, encouraged them, and supported them in seeing a doctor, but sometimes they did not. The involvement of others changed the psychological state of the participants. In this study, "adaptation" refers to the process by which participants accurately assessed and addressed their cardiovascular risk.

P10: When I looked it up on the internet, I was despairing. However, by consulting with staff in the genetic medicine department, I was able to choose a positive view of my future. Even with MFS, I felt like I could live normally.

P18: My husband opposed the idea of diagnosing me with MFS. He said that if MFS was a genetic condition, there was probably no cure or benefit in determining the name of the disease. Rather, he said, it would be detrimental to my unmarried children to have me diagnosed with MFS.

Sometimes, the genetic diagnosis encouraged participants to adapt to their condition.

P2: If I am diagnosed genetically, I must have regular cardiovascular checkups even if I have no symptoms.

As described above, individuals' adaptation to MFS was influenced by their interactions with others, and the degree of adaptation to MFS influenced their decision to seek medical care (i).

Perception of the gap between their health status and medical recommendations

Participants considered the extent of the gap between their perceived health status and those of individuals with imagined MFS using their knowledge, experience, and surroundings to determine if the medical recommendations applied to them.

P2: I have not considered health management until now. Now, I know I have MFS, and I want to get regular cardiovascular checkups.

Participants who correctly understood the general medical recommendations involved with MFS accepted that they had MFS and allocated time and effort for medical visits with a cardiologist (j). Conversely, some individuals had an insufficient understanding and could not accept that they had MFS, could not arrange an environment for seeking medical care, and did not consult a cardiologist (k).

Deciding not to see a cardiologist

Some participants did not see a cardiologist despite knowing the medical recommendations. In several cases, decades had passed with no consultation.

P23: I had never felt any symptoms before, so I did not go to a cardiovascular checkup and just went on as usual.

However, when their health deteriorated, they became aware of the potential for their children to inherit MFS or recognized that other family members had been diagnosed with MFS, and they reconsidered the possibility of having MFS (l).

Seeing a cardiologist

Participants who recognized the importance of regular cardiovascular checkups and aimed to adapt to MFS decided to see a cardiologist.

3.2.2 | The process by which an individual with MFS keeps up with cardiology checkups (process B)

Seeing a cardiologist

Participants' cardiovascular status was confirmed at their first appointment with a cardiologist. Individuals who identified as having progressive symptoms and required treatment received medical intervention (letter m, in Figure 1b).

P19: I had no symptoms, but my aortic root had dilated to 3.5 cm. The doctor said, "Actually, it is quite wide for a thin person like you, although it depends on the patient whether it is wide or not."

Even if treatment was not immediately necessary, participants were sometimes asked to continue with follow-up care (n). Participants continued gathering information to determine what they should do after they started seeing a cardiologist (o).

Performing medical interventions

When medical visits indicated that surgical treatment was needed, participants underwent cardiovascular surgery. In some cases, physicians prescribed anticoagulants and antihypertensive medications.

P3: At the time, I had no choice but to undergo surgery. I already had an aortic dissection when I went to see the doctor.

The experience of receiving medical intervention influenced participants' decision-making concerning the need for medical attention (p).

Communicating the characteristics of MFS and the necessity of checkups

Participants had the opportunity to obtain information about MFS from cardiologists and other medical staff.

P8: I had a good primary doctor. He gave me much information about MFS, including cardiovascular risks during pregnancy and childbirth.

They reaffirmed the significance of regular visits to a cardiologist based on the information they received from others (q). Some participants decided to continue seeing a cardiologist without understanding MFS correctly, either due to misinformation provided by others or their misunderstanding (r).

Taking action to learn about what should be done

Some individuals inquired about new concerns that arose after seeing a cardiologist, and some participated in patient associations of MFS or received genetic counseling to collect information.

P7: When I need advice, I ask my acquaintances with MFS on social networking services.

They recognized the need for regular checkups from the information they obtained (s).

Being aware of medical recommendations

Participants learned that they should continue to see a cardiologist even if they have no symptoms and the condition does not require immediate treatment, or if the treatment ends in surgery.

P24: I strongly felt that I needed to do regular checkups even when I had no symptoms. This is because I can receive treatment earlier at a lower cost if symptoms are found at a mild stage.

Once the individuals understood and agreed with the medical recommendations, they decided to continue seeing their doctor (t). Sometimes, they disclosed to others that they had MFS or discussed their MFS-related concerns (u). Some individuals had already received a clinical diagnosis but wished for a more definitive diagnosis and requested molecular genetic testing.

P16: When I came to the university hospital, I saw a posting about genetic counseling or something similar. I wanted a molecular genetic test to clarify if I had MFS, so I told my doctor I wanted genetic counseling.

Encouraging/impeding adaptation to MFS

Others influenced some participants.

P4: I was told by people without Marfan syndrome that "you can probably live longer and healthier than us because you get your whole body checked regularly."

P14: The genetic counselor said, "Has MFS made you unhappy? If you have been happy up to now, then that is you!" Maybe her personality made her say that, but she encouraged me.

The words and attitudes of those around them affected the attitudes of participants toward seeing a doctor and their adaptation to MFS (v).

Perception of the gap between their health status and medical recommendations

The decision by participants to continue seeing a doctor was based on their understanding of medical recommendations, their adaptation to MFS, and their judgment of the environment in which they could see a doctor.

P6: If I quit seeing my cardiologist, sudden death or something similar could occur. I don't want to inconvenience my family, coworkers, or friends, so I want to do everything possible (i.e., regular checkups) to prevent any trouble.

P26: I must visit the hospital regularly because I am taking Warfarin, right?

P14: For adults, beta-blockers are just a medicine that lowers blood pressure and not a cure for MFS. Therefore, some people may wonder if there is any point in going for regular checkups.

Those who could not understand the medical recommendations or adapt to having MFS discontinued their cardiology visits (w). Individuals who understood the medical recommendations but were slow in their adaptation to MFS continued cardiology checkups, but reluctantly (x). Some individuals who understood the medical recommendations and adapted well to MFS were positive about continuing cardiology checkups (y).

Discontinuing regular cardiovascular checkups

Some individuals with less understanding of the need to see a cardiologist and poor adaptation to MFS discontinued seeing a cardiologist.

P19: At that time, I had no symptoms and I didn't understand the need to visit the hospital regularly. I decided not to visit the cardiology department until I experienced any symptoms.

P18: I was afraid the doctor might tell me I needed heart surgery. So, I told the doctor, "I don't need regular checkups until I notice any significant symptoms. So I don't need to book the next appointment."

Those who had stopped seeing a cardiologist became aware of MFS again when symptoms appeared when they found that family members had signs or symptoms of MFS, or when others recommended that they see a cardiologist (z).

P20: I felt a slight pain in my back, so I thought it would be better to see a cardiologist, even if I was pushing myself a bit.

P19: My aunt, who had experienced aortic dissection, told me, "You must see a cardiologist."

Keeping up with cardiology checkups reluctantly

Individuals with a high understanding of the need to see a doctor but a low adaptation to MFS were reluctant to see a cardiologist.

P14: I frequently have a strong aversion and I feel reluctant to go to the hospital. However, I do not think that means I should not go.

P7: I think of my regular visits as my job. I reluctantly accept the necessity of going for checkups.

P4: I recognize that MFS is an incurable disease, and I must live with it for the rest of my life. Going for regular checkups is unavoidable. I try to remind myself that being able to see a doctor regularly is a form of happiness.

Keeping up with cardiology checkups while affirming one's experiences

Individuals with a high level of understanding of the need to see a doctor and a high level of adaptation to MFS continued to see a cardiologist.

P3: It is good to check my health through regular checkups every six months or once a year because it makes me happy if I find out my cardiovascular symptoms are not progressing. Even if some cardiovascular symptoms develop, I would feel lucky to catch them early. I sincerely hope other individuals with MFS can consider regular checkups more positively.

Continuing the cardiology checkups forced participants to face their diagnosis. They had chosen to "live with MFS" after accepting both positive and negative aspects.

P5: MFS has many negative aspects, such as needing cardiovascular surgery, but there are also positive aspects, depending on how I think. How we perceive MFS depends on how we communicate with those around us and how we grasp it.

P9: There may be various mental conflicts when accepting MFS, but we must accept it someday. We must consider how to deal with, overcome, or live with MFS.

3.2.3 | The process by which parents bring their children with MFS to the cardiologist (process C)

Bringing the child to a cardiologist

Minors with MFS who were diagnosed in childhood were unaware of their condition, and the responsibility for seeing a doctor depended primarily on the parents. When parents recognized that their child might have MFS, they took the child to a cardiologist or pediatrician.

P4: I could continue going for regular checkups because my mother took me to the hospital, regardless of my feelings. At that time, I was not aware that I was going for checkups because of MFS.

If the parents understood the need for a medical examination, they continued to have their children undergo regular checkups. This led to the children's awareness of the possibility of MFS (letter α, Figure 1c). If the child already had signs of cardiovascular symptoms, the parents considered it necessary to take the child to a cardiologist, even if they did not fully understand MFS.

P4: Because my mother did not know the name "Marfan syndrome," she seemed to feel that the doctors were talking about something strange. But, since I had cardiovascular symptoms, my mother took me to the pediatrician regardless of whether she realized those symptoms were related to MFS.

Parents who did not fully understand the need to see the doctor stopped taking their children for regular checkups (β), especially if their children had no cardiovascular symptoms.

Realizing the possibility of having MFS

Many minors were not aware of the possibility of MFS when their parents began taking them for checkups in childhood. However, as they continued to receive regular checkups, they became aware of the possibility of MFS.

P5: I first became aware of having a medical condition in elementary school. I found "Marfan syndrome" on the health card submitted to the school and realized I had such a condition.

Some individuals found it difficult to be different from their classmates because they had to miss school for treatment or skip physical education to protect their hearts. However, none of them had negative feelings about their parents taking them to the hospital.

No longer taking the child to a cardiologist

Parents stopped taking their child to the cardiologist/pediatrician if they did not fully understand the characteristics of MFS and the need for cardiovascular checkups.

P15: My parents thought taking me for regular checkups was a hassle. They believed I did not need to go to the hospital because I could go to school and perform like other students. As a result, they stopped taking me for regular checkups.

As a result, children with MFS (or with the possibility of MFS) did not have the opportunity to fully recognize that they had MFS or that they needed regular visits to a cardiologist. Later in adulthood, when others pointed out the possibility of MFS, some understood why their parents had taken them to the hospital when they were young.

4 | DISCUSSION

This is the first qualitative study in Japan to interview 28 people with MFS. Although the participants were recruited from a single facility and a single patient association, our study ensured variation in the data by using a grounded theory approach. Specifically, we considered which participants to recruit next based on the results of data analysis, adjusted interview questions to gather missing information, and ensured comprehensive analysis. People with MFS judged the need for medical visits through interactions with other people, and we clarified the process by which these interactions influence their medical visit behavior (Figure 1). These processes were related to decision-making behaviors and the psychological status in which the patient underwent medical visits. This study is also the first to demonstrate that genetic counseling and molecular genetic diagnosis are useful for promoting adaptation. Furthermore, the research has shown that adaptation to disease is an essential element for enabling individuals with MFS to choose medically recommended behaviors and continue their medical visits (Connors et al., 2015).

4.1 | Connecting individuals with possible MFS to cardiologists

Those who recognized the possibility of MFS also recognized and accepted their health and genetic conditions and decided to attend their cardiology appointments. A family history of cardiovascular symptoms may lead participants with MFS to perceive the severity of their symptoms and seek medical attention. Meanwhile, those with few subjective symptoms or no family history did not perceive the possibility of having MFS or the severity of cardiovascular symptoms and tended to postpone their medical visits.

A family history of cardiovascular disease is significantly associated with recognizing the disease (Bekke-Hansen et al., 2014; Frich et al., 2006). For example, individuals with a family history of cardiac disease were significantly more likely to believe that coronary artery disease was a chronic disease with a long duration and high recurrence rate than those without a family history of cardiac disease (Noureddine et al., 2013). Similarly, for type-2 diabetes, which also requires long-term and regular checkups, incorrect estimation of genetic factors may reduce the willingness for treatment (Habuka, 2012). Medical staff should consider the family history, communicate the risks of MFS with individuals, and ensure their understanding. Genetic counselors can play a significant role in gathering family history information and making assessments of genetic risks.

In cases where a minor was suspected of having MFS and there was a family history, especially if a parent has an MFS diagnosis, information was often shared within the family. A pediatrician or cardiologist typically conducts follow-ups. Medical staff should encourage parental understanding, especially when minors with MFS have no family history or no signs or symptoms of MFS, because parents may consider it unnecessary to take them to the hospital. The involvement of medical staff from departments other than cardiology and pediatrics, such as ophthalmology, orthopedics, and clinical genetics, can also play a key role in ensuring proper cardiovascular checkups.

4.2 | Preventing individuals with MFS from avoiding regular cardiovascular checkups

Patients who continue to visit doctors tend to have better outcomes (Ashour et al., 2020). Therefore, whether they feel positive or negative, it is important to continue medical visits.

Notably, medical visits sometimes continued if participants were prescribed medication, regardless of whether they had undergone surgical treatment or their degree of understanding about MFS. According to Japanese clinical practice guidelines (Japanese Circulation Society, Japanese Society for Cardiovascular Surgery, Japanese Association for Thoracic Surgery, & Japanese Society for Vascular Surgery, 2020), beta-blockers are a first-line treatment for preventing aortic dilatation. The prescription may prompt patients to continue their medical visits. However, no evidence has been established for their preventive effects against aortic dissection or aortic rupture.

Medical professionals should focus on those who reluctantly keep up with cardiology checkups and address the reasons for this. Although all the reasons are not shown in the text (See Appendix S2), our study indicated several factors that impede the adaptation of individuals with MFS, including a lack of understanding by family members without MFS and an attitude of healthcare providers who are more interested in the disease than individuals. We also identified several environmental factors that deter individuals from going to the hospital, including financial burden, concerns about the impact of medical visits on work or education, prioritizing caregiving and childcare over one's health, difficulty accessing medical care, and insufficient information-sharing

among medical departments. These factors contributed to the hesitancy of participants to see their doctor regularly, even if they had a high understanding of the medical recommendations and were accepting of their diagnosis.

Moreover, MFS is designated as an intractable disease and confirmed progression of cardiovascular symptoms is required for patients to receive coverage for medical expenses in Japan. As a result, the costs of each visit can become a burden for asymptomatic individuals. Medical professionals should strive to minimize barriers to access by supporting individuals and providing information on social resources. In addition, it is necessary to take measures within the related departments and hospitals to coordinate appointment dates and share information more effectively.

4.3 | Adaptation to disease and the role of genetic counseling

Because MFS is a genetic disease that cannot be cured even with regular checkups and medication, some patients felt that "it is useless to undergo checkups." Thus, participants reluctantly continue with cardiology checkups and feel uncertain because of the lack of evidence regarding the usefulness of drug therapy (Ammash et al., 2008; Elefteriades & Farkas, 2010). Moreover, individuals experience a strong fear of death after recognizing the possibility of MFS or having confirmed aortic symptoms, leading to a feeling of "wanting to be normal" and psychological distress due to "fear of the unknown." These feelings have all been linked to decreased patient adherence (Connors et al., 2015). Sharing experiences with other individuals who have MFS may dispel the "fear of the unknown" and encourage adaptation (Connors et al., 2015).

Genetic counselors are not involved directly in treating the person with the disease, but in helping them understand what MFS means in their lives. Participants who were asymptomatic or mildly ill and unaware of MFS in their daily lives often found that regular checkups were their only opportunity to think about having MFS. Since the physician was in the position of treating the individual with MFS, their primary role in the routine checkup was communicating clinical facts to participants. Each visit to the cardiologist was a source of emotional distress, as they were forced to remind themselves that they had MFS and were anxious about the possibility that the physician would point out the progression of their condition. In this context, genetic counselors can play a significant role in supporting patients with MFS by helping them perceive their future positively and providing emotional support.

Based on our results, we recommend that genetic counselors should organize the correct information (e.g., natural history, family history, appropriate treatment, the need to see a doctor even when there are no symptoms, and its heredity) and encourage clients to understand and adapt to MFS. Cardiac genetic counselors are valued for their role in promoting patient empowerment and instilling a set of beliefs that allows people in families affected by genetic disorders to feel some control and hope for the future (McAllister et al., 2008). Patient engagement and empowerment have been shown to increase medical adherence and promote healthy behaviors; thus, empowerment is an important outcome of genetic counseling (Rutherford et al., 2014; Zakas et al., 2019). However, the Japanese clinical practice guidelines do not mention the role of genetic counseling or certified genetic counselors in patient care. It is necessary to promote the role of genetic counselors in the cardiology field in Japan.

A genetic diagnosis may facilitate adaptation to the disease, particularly in patients with few subjective symptoms, resulting in continued medical visits. In cardiovascular diseases, symptoms often significantly impact prognosis and motivate individuals to attend medical checkups. At the same time, remaining asymptomatic or having mild symptoms is the most crucial factor for survival. The finding that a definitive diagnosis of MFS with genetic testing increased self-awareness and encouraged adaptation to the disease was a significant result that could be applied to other hereditary diseases with few subjective symptoms.

Moreover, genetic diagnosis not only promotes disease adaptation and medical visitation of adults but also encourages the medical visitation of children and their parents. Manifestation of the clinical features of MFS depends on age (Loeys et al., 2010). For children and young adults with a risk of MFS who do not fully meet the clinical criteria, periodic evaluation, and *FBN1* testing should be considered. (Faivre et al., 2007). Recently all children of parents newly diagnosed with MFS are recommended to undergo screening for manifestations of MFS (Tinkle et al., 2023). Our study suggests that genetic diagnosis in childhood can positively influence medical visit behavior later in life. However, the psychosocial impact of making a definitive diagnosis during childhood cannot be ignored, and there is a need to support adaptation in these individuals by providing regular genetic counseling and considering not only medical aspects but also school, work, and lifestyle.

Finally, the psychological statuses of individuals undergoing cardiology checkups were on a continuous spectrum and fluctuated depending on their situation. At times, they affirmed their own experiences and thought positively, but at other times, they faced their situations reluctantly. Medical staff should be aware not only of the physical status but also of the psychological status of individuals with MFS. In some cases, referring individuals with MFS to a clinical genetics department may be useful.

4.4 | Study limitations

Participants were recruited through one university hospital in Japan and one MFS support group. They are currently receiving medical care and are considered to have a good understanding of MFS. Therefore, their experiences are only partially representative of the wider population. Although all participants self-reported fulfilling the criteria for MFS, the possibility of other genetic diseases, such as collagen disease, cannot be ruled out. Additionally, educational level

and socioeconomic status may influence adherence to healthcare recommendations, but this study did not collect this information.

Interviews were conducted either by phone or online. Although it is not possible to check the facial expressions and gestures of the participants over the phone, other aspects, such as tone, speaking speed, intonation, and the duration of pauses, were noted in the interview and post-interview, and considered during the analysis.

This study is a qualitative study using grounded theory. The experience and knowledge of SH, the researcher who was heavily involved in both the acquisition and analysis of the data, may have contributed to bias in the results. However, all verbatim transcripts were sliced into fine sections before analysis to minimize bias. This technique of data slicing is unique to grounded theory. It involves temporarily disconnecting data from their original context, allowing for a thorough and multifaced examination that avoids being constrained by initial impressions. Furthermore, the validity of the analysis was reviewed by multiple researchers. The behavioral process described in this study represents an interpretation of the available data; other interpretations may emerge in larger datasets of different populations.

4.5 | Practice implications

Our findings suggest the need for several intervention strategies and may help identify those who have decided not to see a cardiologist or are at risk of ending their medical visits.

Physicians may need to consider communicating information and risks more carefully when telling their patients without a family history of MFS about the possibility of MFS. When indicating the possibility of MFS to minors, it is especially important to encourage understanding by the parents. Individuals with MFS who have stopped their visits to a cardiologist in the past or those who are thought to have a negative psychological condition toward medical visits may require more careful engagement from medical staff. In some cases, a proactive approach from the healthcare organization may be necessary, for example, contacting patients from the hospital.

Adaptation to MFS should be encouraged in all individuals with MFS to motivate them to continue their medical visits. Genetic counseling and genetic testing for definitive diagnosis may be useful in this regard. Particularly, asymptomatic individuals who may be hesitant to see a cardiologist should be referred to the genetic services department.

All genetic counselors need to help individuals with MFS and their families adapt on an ongoing basis, not just before and after diagnosis; consultation with other departments may be possible, as well as education for the individual, to ensure that minors with MFS receive appropriate medical care as adults. A detailed family history should be obtained to identify relatives who may have MFS and encourage them to seek medical care; the experiences of other individuals living with MFS and information about support groups may be an incentive to continue medical visits. As of 2023, there are approximately 350 certified genetic counselors in Japan. Due to the limited number, genetic counselors are working across multiple disease areas. Consequently, the number of genetic counselors specialized in the field of cardiology, known as "cardiac genetic counselors," is restricted. In the future, it will be necessary to further invest in the training and education of cardiac genetic counselors in Japan. However, it's important to note that any genetic counselor can provide counseling on MFS.

Surveillance recommendations for individuals with MFS who have not developed cardiovascular symptoms need to be established in Japan. Additionally, the roles of genetic counselors should be more explicitly outlined in Japanese clinical practice guidelines, emphasizing active collaboration as part of a team.

Our results may help individuals with other cardiovascular diseases or hereditary conditions adhere to regular lifelong medical visits.

4.6 | Research recommendations

Large-scale quantitative studies in broader populations are required to validate whether the processes identified in our study can be applied in clinical practice in Japan. It is necessary to survey individuals with MFS throughout Japan with various attributes and in various healthcare settings. It is also necessary to clarify the perceptions of medical professionals involved in MFS treatment in Japan and the status of the medical system.

Genetic counselors can assume the role of serving as a hub that connects various medical departments and the genetics department. Examining realistic challenges and prospects for how each medical and genetic department can work together is important.

5 | CONCLUSIONS

This qualitative study aimed to clarify how medical recommendations by cardiologists are judged and dealt with by people with MFS based on their own experiences. Our results provide insights into how those with MFS adopt medical visit behaviors based on interactions with others. Our findings suggest that appropriate interventions lead to better medical visit behavior.

AUTHOR CONTRIBUTIONS

SH planned, designed, collected, and wrote the study. MT, HK, and TW analyzed and interpreted the data, and reviewed and approved the manuscript. SK provided oversight and review of the study. SH confirmed responsibility for data accuracy and integrity. All authors approved the final version for publication and took responsibility for questions regarding accuracy and integrity.

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CONFLICT OF INTEREST STATEMENT

SH, MT, HK, TW, and SK declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

The data cannot be shared due to privacy and ethical concerns.

ETHICS STATEMENTS

Human studies and informed consent: This study was approved by the Kyoto University Graduate School and Faculty of Medicine and the Kyoto University Hospital Ethics Committee (approval number R2501-3). Participants' consent was obtained through the completion of a consent form, which was approved by the institutional review board. The study adhered to the ethical standards set by the responsible committee on human experimentation, both institutionally and nationally, and was conducted following the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all the participants for inclusion in the interviews.

Animal studies: No animal studies were conducted by the authors for this research.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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