DM family

Myotonic Dystrophy Patients' Group of Japan

We are DM-family

Annual Report, 2021

https://dm-family.net/

DM-family　Message from the President

My name is Akane Hatano and I am the President of DM-family, a myotonic dystrophy patient group. It is nice to meet you.

I am a patient with myotonic dystrophy type 1. It has been about 14 years since I developed the disease.

Before then, I was living a normal, healthy life, but since the onset, the disease has progressed so quickly. I started using a wheelchair, lost the ability to walk, and am now living in a hospital with assistance.

At first, I was frightened by the disease, despaired of my deteriorating body, and became desperate. One day, however, I read Professor Peter Harper's book "Myotonic Dystrophy-the facts" and was able to change my mind and be more optimisic.

I think every patient will initially feel a sense of unfairness and despair, asking "Why am I the only one to suffer this?" However, if they do nothing, they will not only deteriorate physically, but also mentally.

You may recall every single thing you can no longer do and grieve over it.

I don't think it is possible to be positive from the very beginning. It took me a good two years to sort out my thoughts, but after reading Professor Harper's book, I met a wonderful doctor, and am now able to even joke about my illness.

Can a patient do nothing? No, quite the opposite.

Just having hope for a cure can make a difference. And it is our patient group's role to work together with patients to find ways to express their feelings and provide them with various information.

We encourage all patients and their families to live cheerfully without giving up hope. A smile leads to a better quality of life.

It's important not to grieve but be thankful for the small blessings of what we take for granted every day. How many blessings do you have today? It should be more than the number of your tears.

Whether you spend the day crying or laughing while making the most of it, a day is a day.

So, I want to cherish this day.

And I will not give up hope. I believe that the next generation will beat myotonic dystrophy, if not my generation. I wonder every day if there is anything I can still do as a patient to make that happen.

I am sure there are many patients in the world who feel the same way. We may speak different languages, but our thoughts are the same.

Finally, I would like to thank all the people who are working hard to overcome myotonic dystrophy.

Thank you from the bottom of my heart. Let us continue to work hand in hand and together change the future of myotonic dystrophy.

Akane Hatano

Akane Hatano
President
DM-family

At National Hospital Organization Shimoshizu Natiaonal Hospital, President Akane Hatano, reading "Hamlet" aloud as rehabilitation for dysarthria

With Prof. Peter Harper during his visit to Japan

There Are Things You Can Do as Patients

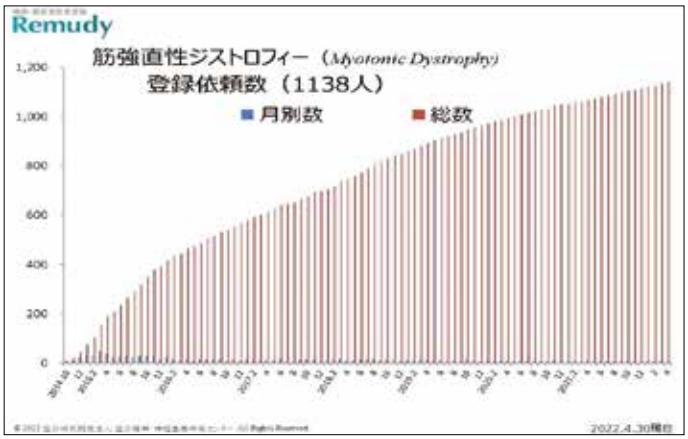
Patients Can Encourage Other Patients to Join the Patient Registry

Patients with neuromuscular diseases in Japan enroll in the patient registry of their own volition, without being forced to do so.

What is a registry? What are the benefits? Will I be discriminated against for having my disease diagnosed through genetic testing? These are the concerns that all patients and their families have. And the lack of motivation, which is one of the symptoms of myotonic dystrophy, makes the registry even more difficult.

The DM-family patients have been encouraging all patients to join the registry. In 2021, the registration had grown to more than 1,000 members, which is more than 10 percent of the estimated 10,000 patients in Japan.

Even today, the number of registrants continues to grow by 5 to 6 per month. Patient-to-patient encouragement of the registry has become an important part of DM-family's activities.



Results of “Remudy”, a patient registry system operated by the National Center of Neurology and Psychiatry and Osaka University
1138 registrants (as of April 2022)

Patients Encourage Others to Participate in Natural History Studies

In 2021, a natural history study of myotonic dystrophy was initiated in Japan at the Japan Agency for Medical Research and Development (AMED) research group.

Since the number of DM patients is small, it is essential to conduct research that records and observes accurate data on how patients progress (natural history) in order to develop treatments. While large-scale natural history studies are being conducted in many countries, it is highly significant to carry them out in Japan as well.

DM-family often receives inquiries about how the disease progresses. Simply explaining to them that progress varies from person to person does not alleviate the concerns of patients and their families.

DM-family has been conducting a nationwide webinar to encourage all patients to participate in natural history research.

For patients, natural history research means facing the progression of their own disease.

Akechi told patients to "have courage and participate in the research” and contributed to the increased number of participants.

Yuji Akechi,
DM-family Vice President

Participants in the natural history research

Without Knowing the Cause, It Is Impossible to Know How It Can Be Cured

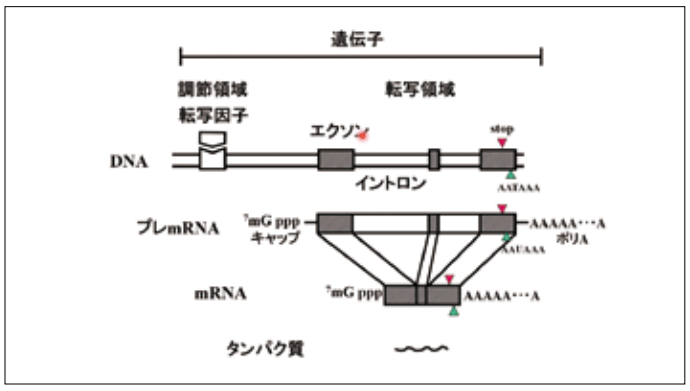
Dr. Shoichi Ishiura,
Professor Emeritus of the University of Tokyo

Dr. Ishiura at a conference in Canada in 2008

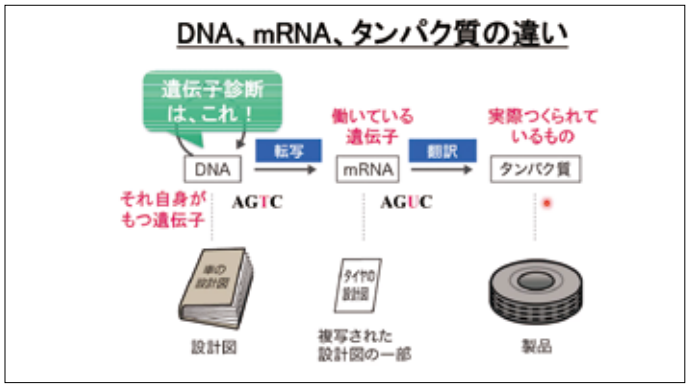
What Patients and Families Need to Know About Gene Mechanisms

On July 24, 2021, DM-family hosted a webinar "Understanding Genes and Therapeutics". About 70 people eagerly listened to the lecture by Professor Emeritus Shoichi Ishiura of the University of Tokyo.

A gene is the blueprint of the human body. There are exons, which are the blueprint for making proteins in the human body, and introns, which connect them.



Exons are combined to make multiple mRNAs. Splicing means cutting out the exons, and proteins are made from mRNA.



Causes of Myotonic Dystrophy

Myotonic dystrophy is caused by an abnormal expansion of DNA base repeats (C, T, G) at the end of the *DMPK* gene on chromosome 19. When the CTG repeat expands, the mRNA also expands, and the splicing regulator MBNL is adsorbed there. Abnormal splicing occurs in genes throughout the body, resulting in a variety of symptoms that afflict patients.

The Treatment is to Normalize MBNL

Three main strategies are considered by pharmaceutical companies and researchers.

1. Degrade long repeat RNA
2. Alter splicing
3. Make MBNL work

We Must Learn More

Prof. Ishiura then gave an easy-to-understand explanation of "antisense (antisense oligonucleotides)". It is difficult for antisense to reach throughout the body, and new technologies that make this possible are awaited.
Many patients and their families are unaware of the relationship between genes and treatment, and many expressed their appreciation after the lecture, saying, "Until now I have simply wished for the early approval of new drugs, but now I know more about the causes and genetic abnormalities."

Lack of Motivation, Fatigue...
What We Can Do at Home Is to Praise Each Other

Dr. Shugo Suwazono,
National Hospital Organization Okinawa Hospital

Dr. Yukihiko Ueda,
Okinawa International University

How Patients Can Have Self-Esteem

On July 11, 2021, DM-family hosted a Zoom meeting, "A Praise Practice".
DM patients, even without muscle weakness, can complicate their symptoms by central nervous system disorders. Due to a lack of motivation and apathy, it is difficult for the patients to have high self-esteem. Medical professionals recommend "praising the patient," but what is the best way to praise a patient who is lethargic and always sleeping?
Dr. Shugo Suwazono of the National Hospital Organization Okinawa Hospital and Dr. Yukihiko Ueda of Okinawa International University gave a lecture on "fostering self-esteem through praise," and in the latter half of the session, the participants practiced "praising" each other.

What Does It Mean to Praise?

In reality, it is not easy for patients and their families to praise each other. Living together every day; how can we praise the same thing over and over? But still, Dr. Suwazono said, "You should try it even if you get stuck in a rut."
Dr. Ueda indicated that when people receive praise, they develop feelings like "I am important" and "I am okay," and that it becomes a driving force for life and an opportunity to realize the goodness in themselves that they did not realize.
Patients and their families should also praise their own selves, which also has the effect of increasing self-esteem.
Furthermore, it is good to participate in society and receive praise from others from time to time.

Participants got excited with the Praise Practice

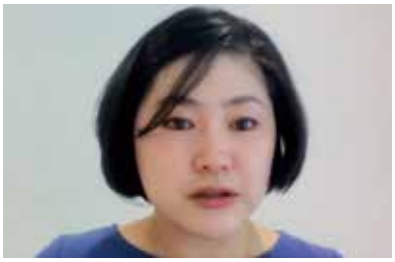
The participants took turns giving compliments to each other.
First, one person introduces himself/herself and talks about what made him/her happy recently. Another person listens and praises the other person's good points. How is the self-introduction and what makes one happy? The person giving the compliment listens intently and picks the other person's praiseworthy points quickly. Surprisingly, one will be able to see the other person's good points.

"I was able to have a reunion on Zoom, which I learned to do in the patient group." - "It's great to try something new and take on a leadership role."

"I made some potato chips for my child and he was delighted." - "The fact that you made potato chips by yourself is amazing."

The lesson was a great success, and everyone was filled with happiness at the end.

Research and Conversations about Children with Congenital Myotonic Dystrophy



Dr. Minobu Shichiji,
Tokyo Women's Medical University

What Information and Medical Care Do Families Want?

On Sunday, October 31, 2021, DM-family hosted a meeting, "Research and Conversations About Children", for parents of congenital myotonic dystrophy and childhood-onset myotonic dystrophy.

Twenty-eight people attended the meeting, where Dr. Minobu Shichiji, Department of Pediatrics, Tokyo Women's Medical University Hospital, reported the results of two surveys she conducted.

The results showed that in 54% of cases, mothers themselves were diagnosed as DM1 patients after giving birth. The children had intellectual disabilities, and many parents were working on language development, with a large number calling for appropriate social support.

After the lecture, both parents and children enjoyed interaction with each other.

Family Members Should Also Take Care of Each Other's Mental and Physical Health

How to stabilize the Caregiver's Emotional well-being

On Sunday, November 28, 2021, DM-family held a meeting for families, "Let's Get Together as Family Members in Charge of Care!" The anxiety that family caregivers face and the emotional problems that they cannot resolve on their own are serious.

At the meeting, Dr. Makiko Endo, who studies the psychological states of family caregivers at the National Center of Neurology and Psychiatry Hospital, gave a lecture. She reported that a survey of DM patient caregivers showed that about 30% of the families felt a significant burden.

How can caregivers increase resilience? Dr. Endo urged family caregivers to "practice physical and mental self-care themselves as well!"

Many caregivers may hesitate to provide support to others because of their strong sense of responsibility or guilt about taking care of themselves. However, support should be actively sought, and being able to do so is part of their ability.

If family members develop the habit of observing their own mental states objectively, it will be easier for them to take truly necessary actions.

Dr. Makiko Endo,
National Center of Neurology and Psychiatry Hospital

Pharmaceutical Companies Want a Real Picture of the Patient

We Need a Better Understanding of the Disease

On Sunday, September 12, 2021, the DM-family hosted a webinar, "Advancing New Drug Development for Rare and Incurable Diseases," with the help of the Development Department of Astellas Pharma Inc. In daily life, patients rarely have opportunities to meet pharmaceutical company employees directly. This webinar was an opportunity for them to hear frankly about what is needed in the development of therapeutic drugs.

Astellas spoke about the importance of how much reliable data they have on the target disease when making a clinical trial implementation plan (protocol). Rare diseases present the following difficulties: a small number of patients, lack of understanding of causes and disease progression.

How to Overcome Difficulties

To overcome these difficulties, we will need natural history studies to accurately measure how patients progress, and patient-reported outcomes (PROs) to observe what happens in their daily lives. Especially, natural history data may be used for a control group in rare disease trials and must be accurate.

Observational studies require the active participation of patients. Without the cooperation of patients, there will be no development of new drugs.

Ms. Midori Senoo, Executive Director of DM-family, with Astellas Pharma Inc. Representatives

国際筋強直性ジストロフィー啓発の日記念

INTERNATIONAL Myotonic Dystrophy Awareness Day SEPTEMBER 15

astellas × DM family

希少疾患・難病の新薬開発を進めるために

Members Undergo Dysarthria Rehabilitation with Other Members to Improve Conversation/ Articulation

On Sunday, December 12, 2021, a Zoom meeting entitled "What is Dysarthria Rehab?" was held.

DM patients often have dysarthria, such as having difficulty speaking out or not being able to make themselves understood.

They speak in a manner that is difficult for others to catch, and often stop the conversation, making them increasingly reluctant to continue.

What should be done in these situations? Dr. Chihiro Oda, a speech therapist at the National Center of Neurology and Psychiatry Hospital, gave us a lecture, including practical guidance.

Dr. Oda explained the delayed auditory feedback (DAF) method and a pacing board, and all the participants were amazed to watch a video which showed the dramatic change in a patient's speech manner after receiving the therapy.

Dr. Chihiro Oda,
National Center Hospital, National Center of Neurology and Psychiatry

There are also things that can be done in daily life.

Just trying to speak louder and slower can make a difference.

Mutual Compassion Is Important

Family members who listen to a patient also have some techniques to make it easier to catch the patient's voice. Before becoming irritated by patients who speak too fast, they should try to know the conversation topic beforehand, not miss the sign when patients show their desire to speak, and try to get as close to the speaker as possible.

Dr. Oda said, "In order to enjoy conversation, both the speaker and listener need to be considerate of each other."

Together, we can change the future of Myotonic Dystrophy

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