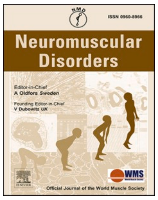
Neuromuscular Disorders 55 (2025) 106218

Contents lists available at ScienceDirect

Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd



Marion or the metamorphosis: a journey of self-reconstruction and hope in the face of FSHD

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ARTICLE INFO

Keywords:
Patient perspective
Living life to the fullest
Acceptance
FSHD

ABSTRACT

This Patients' Forum centers on the testimonial of visual artist Marion Sellenet, who lives with facioscapulohumeral muscular dystrophy (FSHD). In her film *Marion or the metamorphosis*, co-created with Laëtitia Moreau, she documents her dual journey: coping with the physical manifestations of FSHD and overcoming a "second illness" of fear, despair, and stigma induced by the diagnosis. By relinquishing the hope of cure, she found renewed meaning and a sense of wholeness, advocating for collaborative approaches that integrate medical, philosophical, and experiential knowledge. Her account is complemented by reflections from Alexandra Belayew, who underscores the importance of patient education and accessible communication of research; Baziel van Engelen, who highlights the need to bracket biomedical knowledge to address lived experience; and Ria de Haas and Nicol Voermans, who emphasize patient engagement in care and research. Together, these perspectives illustrate the transformative potential of patient narratives and underline the value of patient-centered, holistic approaches in FSHD.

1. Introduction *(Ria de Haas and Nicol Voermans)*

In this Patients' Forum, Marion Sellenet, a patient living with facioscapulohumeral muscular dystrophy (FSHD), shares the personal transformation she underwent in her effort to heal from a second, less visible illness. She describes how this second affliction—marked by fear, despair, and shame—emerged alongside her diagnosis. It was, she writes, like a sword of Damocles hanging over her. While medicine was unable to alleviate her physical symptoms, it also offered little guidance for living well with the disease. To reclaim a sense of wholeness—fractured by confusing medical terminology and a stigmatizing societal view of her condition—she created a film "**Marion or the metamorphosis**" as a means of self-reconstruction. And this film was created with the help of her talented co-director and accomplice Laëtitia Moreau. Paradoxically, it was only by relinquishing the hope of a cure that she rediscovered hope in life itself. Today, she advocates for collaboration among patients, clinicians, researchers, and philosophers—not only to treat symptoms, but to heal the person and cultivate knowledge on how to live well with chronic illness.

This testimonial is followed by three reflections. The first is of Alexandra Belayew, a Belgian molecular biologist, now professor emerita at the University of Mons, known for her pioneering work in the molecular mechanism of FSHD. The most significant contributions of her research group include the identification and characterization of the DUX4 gene—whose inappropriate activation causes expression of a potent transcription factor driving muscle pathology in FSHD. She is an expert in explaining complex concepts to a broad audience, such as the FSHD patient community. The second reflection is of Baziel van Engelen, a Dutch neurologist, philosopher and researcher, specialized in neuromuscular disorders. He is a retired professor at the Radboud University Medical Centre (Radboudumc) in Nijmegen, the Netherlands, where he led the translational research bridging patient care and basic science. The Forum article is concluded by a reflection of Nicol Voermans, a professor in Neurology at the same university, who leads the FSHD European Trial network.

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https://doi.org/10.1016/j.nmd.2025.106218
Received 21 July 2025; Received in revised form 19 September 2025; Accepted 21 September 2025

2. The metamorphosis of Marion Sellenet *(Marion's story in her own words)*

Hello, I am Marion. I am a visual artist born in France and living in Brussels, Belgium. I am practicing my passion through visual arts, collage, press illustration, communication, animation . But I have another practice that I also consider an art, the art of living with FSHD.

I never thought I would make a movie, let alone expose my genetic peculiarity or my intimacy to the world. But it felt like a necessity. I was inspired to make this film when, looking back on my life, I was moved to see how far I had come. I had become a different person with a different mindset. And today, I think that in some ways, my experience with FSHD has been a real blessing. And this is what I wanted to convey through this film. With Laëtitia, I also wanted to show the beautiful, unsuspected, poetic and crazy side of adventure which the general public often ignores, entrenched behind its own fears. That is why we have chosen this hybrid, artistic and whimsical form.

The symptoms of FSHD may set in slowly, but the diagnosis comes fast and hard, one that many of us may have experienced as a shock. A moment when life turns upside down. Because we understand that what we've lost, in this case our muscles, we've lost for the rest of our lives and will continue to do so. In my case, FSHD had not yet been detected in the family, so we had no "know-how" about how to live with this disease. For other families, the experience can be different when you already know about FSHD because family members are affected.

I was diagnosed in 2003, when I was fifteen and I could not lift my right arm. At the time, not much was known about FSHD. My father and I left the hospital empty-handed: there was nothing we could do, we hadn't been given any explanations or tools. Medicine could not cure me, but it could anticipate my condition, condemning me to certain deterioration. It was at this point that I caught a second disease, the disease of fear and despair that you get from certain tactless doctors. We also get it because, for most of us, our collective imagination and unconscious is negative about illness and disability. And then, of course, there is our character, our personality, our innate dose of optimism or pessimism. And according to me, my life was over, and I could be happy.

A year later, I went for my annual follow-up appointment, with the same tactless neurologist. This time, he brought in some interns to observe my body and my shoulder blades sticking out behind my back: young men. I was 16, and it was very embarrassing. After that, I stopped the follow-ups. But I started physiotherapy and psychotherapy. These therapists were not all good, some were bad, others, excellent.

At the time, I saw my life as a dead end. I did not want to be that Marion, I wanted to reset myself. I fantasized about another life without this disease, wasting my energy and not taking advantage of the present moment that was offered to me. There is this sentence I read in a book that sums up this state very well: "My mind was busy hoping rather than living, rather than being".

I was becoming a young adult and began to feel symptoms in my legs. I could not find the answers to my questions, questions that I had trouble formulating clearly: how do you manage, when you are so young, to live with these symptoms, and to live with the idea of losing? Is it possible to live with this sword of Damocles hanging over the muscles of my body? These are difficult questions to ask explicitly to my loved ones and even to my therapists and I've left to deal with them alone. I think medicine is the least gifted!

What medicine did not tell me was that it could not anticipate the curve of my happiness the way it could for the state of my muscles. I belatedly realized that these two curves were not necessarily going to follow each other or did medicine tell me that a human being is more than just a body and its biological functions. It shapes and reinvents itself at every stage and event of life. It has a great capacity to adapt. And then there are solutions, however small, technical or otherwise, that can improve our daily lives. Medicine does not know what we're going to do with this illness, or what we are going to get out of this experience, and that leaves a lot to be desired.

And yes, I can say I am a lot happier today than when I was running on two legs and could catch the bus. I have more symptoms today, I do fewer things. But I know exactly what is needed to feel good (arrange things at the right height at home, choose a place outside where there are chairs and put a cushion under my bottom, or wear comfortable shoes, take a walk limited to a few kilometres, …). I don't care if I can't wear heels anymore, because what is important is my physical well-being. And above all, to surround myself with interesting and caring people. Today I really enjoy taking care and take responsibility of myself.

During those hard years, even though life went on the outside, inside I felt quite depressed and alone. With lots of negative ideas floating around. And this despite a loving family and caring, supportive friends.

Little by little, I learned to manage the difficulty of my symptoms by looking for the techniques and therapists that best suited me. I met some extraordinary therapists who developed a personalized approach and were inventive. But the link with the outside world remained complicated. Here too, little by little, I entered the world of work with some positive experiences and that made me feel better. But I still felt that something was blocking me, because I was still clinging to this desire to heal, and I scrutinized every change in my body. I often heard people who did not have my disease say that you have to accept it, or that you have to stop identifying with the disease. But when the symptoms are there 24 h a day (like problems with posture, pain in the neck, the feeling that your head weighs a ton on your neck, and that nothing is in the right place in your body), it takes time to know how to put distance between ourselves and our invading symptoms.

For me, all these years have been a work of composition, a bit like a painting. We learn to deal with new data, in all areas of our lives, social relationships, work, health. We do no always succeed, we make mistakes, we move forwards and backwards, and it is in these moments that we can test what is right for us, figure out how to organize ourselves, how to recompose ourselves in our daily lives, and how to find balance.

It is more complex than the theory of mourning, according to which, in summary, we go through several successive states: denial, anger, bargaining, depression, acceptance, reconstruction. Moreover, the word "denial" seems to me to have a negative connotation. I find it a harsh way of talking about what people are going through, even though it is an essential moment of transformation and management with a new body.

A writer from the Dingdingdong collective compared this to the caterpillar in its chrysalis, in the middle of the mud. This process is slow, invisible, singular and sometimes not understood by others. The challenge is that there are many metamorphoses in a single FSHD life! Nevertheless, my own metamorphosis has been greatly nourished by encounters that I recount in the film. I would like to thank our French patient association Amis FSH for facilitating these encounters. It was also thanks to them that I met researcher Alexandra Belayew, but I will leave you discover the film.

At some point, I realized that if I did not have the power to change reality, it was time to act where I did have power, and that was in the way I looked at the situation. There is no manual, no ready-made recipe for feeling good with this disease. No easy way to transform the constraint and to take the disease as an opportunity to give another meaning to one's life. But neither is there an injunction to do so. Everyone has their own path and their own way of doing things.

For me, the big leap was confronting myself with what my future life with a chronic illness might look like and whose ultimate downfall would be the wheelchair. I met several members of Amis FSH and people who have other diseases such as multiple sclerosis. And there, to be honest, I was able to see the mountain of prejudice I had about disabled people, the unconscious superiority we can feel towards them. I realized that you could have a life, a job, a lover, a family, a hobby, all that without having all your limbs functional..

When I was a teenager, social networks were not that developed yet. It was not until quite late that I started to take advantage of all the testimonials we can see today thanks to the Internet. I remember when I came across a picture (Fig. 1): it was Nico who also has FSHD, and who was posing for a photographer. I said to myself: “Wow, it is beautiful, he dares to expose the particularity of his body”. It blew me away. Thanks to those damned algorithms, that have replaced cutes cats with people with singular bodies and good vibes. I came across other profiles: positive, dynamic people who assume their life with good humour and joy. It is very important to counter the stubborn idea of drama and unhappiness.

I took my interest in disability culture a step further, through the writings of militant collectives that added a social and political dimension to my thinking. I discovered the concept of “ableism”. Ableism means, for concrete example, valuing the use of tools (AI, digital, etc.) when they are used by able-bodied people, but stigmatizing people who use these same tools to speak with a synthesized voice, for example, to compensate for their inability to speak with their own voice. Or, authorizing massive home-office during Covid but refusing this measure for disabled workers in normal times. It is a hierarchy of lives.

I’ve seen some great films that challenge these notions, such as:

- “Crip Camp, a disability revolution” (Netflix) : In the seventies, a group of disabled teens find the means to mobilize thanks to an innovative summer camp that helps them organize a movement for greater equality.
- “Take a Look at This Heart” – A documentary about love and sexuality in the disabled community. (Prime video)

I think this is one of the best things FSHD has given me: to be generally aware about the prejudices I may have about people’s situations. Some people believe themselves to be superior to others in these respects, but are they really happier with their two arms and two legs, or their two eyes? Do they have a good relationship with their family? Do they have a job they enjoy?

I would add, of course, that we can be happy only so long as society allows us to live with dignity, to have access to suitable work and to all areas of civil life. Inaccessibility is an obstacle to happiness, to work and to relationships. I am not going to teach you anything about this. It should not be and that is why it is important to support these struggles.

Another important point I wanted to make, paradoxically enough, is that I regained hope in my life when I came to terms with the fact that I could not be cured. It were the written words, essays, podcasts and literature that helped me. The thinkers behind these texts - I like to call them “thought artists” - enabled me to redefine words and concepts to think about what I was going through. Some of their proposed framings were astounding and acted like medicine. Some of them I included in the film. I learned that good health is not a fixed standard, a perfect state to be achieved, beautiful curves to marry, good rates to have. Rather, it is above all a complex balance in which each person has their own standard.

When you are ill, you have to let go of the concept of what people call “good health”, and you lose a good part of your freedom and the multitude of choices you had. But this is an illusion, even for those in perfect health. Because when it comes down to it, we all have our little problems, or will soon, and we will all be faced with decrepitude at one point or another. Whatever those who dream of eternity may say (and in saying that I cannot help but salute Bryan Johnson), we are a physical body made of matter, which undergoes accidents and changes. The ordeal of illness can help us realize that we are not omnipotent. Illness is a fact of life. Life is a complex balance between the beautiful and the less beautiful, the pleasant and the painful. But we tend to live in a civilization that divides things into good and bad. We see happiness as the absence of unpleasant things, of defects. But harmony is rather a relationship and a balance between these things.

I also came across a wonderful little book written by Carlos Bustamante, who has multiple sclerosis, and which reminds me of our experience. For him, “it is all about exploring your body and its capacities, trying to take into account pain or difficulty without dwelling on the

Fig. 1. Nico (Instagram @acko_fshd), two pictures of the project "La Difference" of Veronique Droulez, photo by Jean-Francois Alois. This project is a powerful photographic series that explores visible facial or bodily difference with both aesthetic sensitivity and clinical relevance. The project foregrounds individuals who diverge from normative appearance and presents them with dignity, humanity, and quiet strength. Aloisi’s visual framing—carefully composed, restrained in colour and light—eschews sensationalism in favour of empathy, inviting the viewer to encounter the subject’s personhood rather than pathology. From a medical humanities perspective, *La Différence* offers a rich interdisciplinary platform: it reinforces the importance of narrative medicine, reminds clinicians to engage with patients’ psychosocial experiences, and prompts reflection on stigma, identity, and the embodied experience of living with difference. In doing so, it aligns with current approaches to patient-centred care by humanizing conditions often defined in strictly biomedical terms.

feelings it produces". Living with FSHD is exactly this challenge. It means constantly dealing with unpleasant, even downright painful sensations, with fatigue and daily frustrations. Personally, it taught me to understand how discomfort and pain turn into mental ruminations. We need to practice brain gymnastics, through meditation, breathing and other means. I think it is a very useful skill in life, to understand the mechanisms that link the body to the mind.

There is also this other quote by Carlos Bustamante that I like: "When life is reduced, in fact it becomes concentrated." Finally, he says: "When a new frame appears and the search for compatibility with one's environment can begin. One can begin to explore the world, the possibilities with this transformed body." I think there are many people on this earth, sick or not, who have no meaning in their lives, who do not know how to take care of themselves, who do not know how to look for an environment suited to their needs. Who can spend an entire lifetime with their certainties, without moving one iota in their functioning, in their beliefs, and being completely immobile in their minds. FSHD can be the opportunity for this inner movement, to move intellectually and philosophically. It is not an obligation, it is a desire.

Of course, I am aware there are situations that are sometimes unbearable, seriously ill patients. And that is why I hope a treatment should be found as quickly as possible. I salute all the extraordinary researchers who are working on this and who are gathered here. I thank them all. We need you!

For me, scientific and medical research is perfectly compatible with our inner quest as patients. We each have our own job to do. And if we can do it together, and understand each other, it will be even more effective. The common perspective is to treat people, not symptoms. In my opinion, it is necessary to invent for ourselves medicines that complement those of modern medicine.

Even while waiting for a hypothetical future treatment, and supporting research in every possible way, we need to live in the present, and look for our own resources. We cannot pin all our hopes on research only, which is dependent on funding, market, labs, politics, human relationships and so on. Because there will always be people we can't treat, and those lives will be worth living. And because what kind of shared human experience would we have if diseases were entirely eradicated? Personally, I think it is possible to live a healthy life with an illness, and it is up to us, as patients who are experts in our own situation, to create an environment around us that is conducive to our well-being, and to surround ourselves with positive, open-minded, ingenious people, whether in medicine, art, sports or other fields.

One great example of creating such an environment is the DUX Foundation in France. Its mission is to help and support children with FSHD; by funding therapeutic projects aimed at healing them and by promoting the best possible care. To carry out these missions, it has created a new, central community bringing together healthcare teams, research/biotech groups, and also a large, caring, contributing, and independent "family". The DUX Foundation thus brings together all children with FSHD in France through digital and independent exchange platforms, informational or charity events, and wellness workshops. I think it is absolutely essential to support children with FSHD, as they do. It is in these crucial turning points that they need the most support to regain their self-confidence and grow into happy adults.

3. The importance of patient education (*Alexandra Belayew*)

As Marion and Laëtitia show in their film, the way the diagnosis is announced is often experienced by the patient as a severe blow in itself. Part of the violence issue is probably linked to the mindset of medical doctors. Indeed, their purpose in life is to help fellow humans and it can be very frustrating to meet a patient affected with a rare and mysterious disease for which there is no cure. Since Marion's diagnosis 20 years ago, the understanding of FSHD has strongly improved and an important help for medical doctors is the European reference network for neuromuscular diseases (EURO—NMD) that provides easy access to centralized knowledge for these rare pathologies. The empathy issue is now more frequently addressed in medical education, and Marion's film about her personal experience could be part of medical staff training in empathy.

For Marion, the diagnosis also brought a huge wave of anxiety because of the mystery surrounding FSHD. The only certainty was irreversible evolution of muscle loss: like all her fellow patients, she would have to adapt over and over again as disease progresses, not knowing which muscle would be hit next and when, but always with the wheelchair in sight.

Those who have not experienced such trauma might recall the early days of the COVID-19 pandemic, when little was known about the disease beyond the harrowing reports from Bergamo, Italy—where dozens of healthcare workers were infected and died while trying to save lives, and military trucks were used to transport the overwhelming number of deceased to cemeteries in other cities. Many of us were eagerly trying to harvest data about the causal virus, its variants, their transmission, the disease pathophysiology and putative drugs, as a way to regain control on their life.

Patients recently diagnosed with FSHD need a similar knowledge-based empowerment: they want to understand their disease, its complex cause, its evolution, be able to anticipate the next muscle loss, and desperately surf internet in search of a putative remedy a fellow patient might have used. This is when science popularization becomes an absolute survival requirement. In addition, now that clinical trials are starting, patients are eager to discover the tested drugs: what are their modes of action, what could go wrong with them, could they block DUX4 expression/toxicity or at least stop disease progression, how will their impact on disease progression be evaluated, could the muscles still have the capacity to regenerate after years of DUX4 burden? Communication from pharmaceutical companies must be further clarified through public outreach efforts, to ensure that all patients gain a clear understanding of the development process and are better equipped to make informed decisions about trial participation.

My research group unexpectedly discovered *DUX4*, a gene in "junk" repeated DNA at the FSHD locus 25 years ago, at the KULeuven (Belgium) and I took the project in my own lab (UMONS) in 1999 till I became emerita in 2015. Our investigations of DUX4 putative role as "the" FSHD culprit gene and its targeting for drug development were mostly funded by international patient associations. As a sense of duty towards patient families I requested graduate students or post-doc researchers in my group they could explain in lay terms what they were doing in the lab with such grants. I had borrowed this lab tradition from Philip Leder (1934–2020), an outstanding early molecular genetic expert who was at the NIH when he trained both my PhD and post-doc mentors (Jacques Gielen at ULiège and Shirley M. Tilghman then at Fox Chase Cancer Center, Philadelphia). He told his post-doc researchers '*If you can't explain what you are doing at the bench to the person cleaning the lab, you haven't understood your project!*' Indeed, it is highly instructive for young scientists to engage in popularization, as it compels them to distil their work to its essential core. I later practiced science outreach to the general public when I was in Joseph Martial's lab (ULiège), and we had to give tens of talks on genetic engineering to very heterogeneous audiences.

This experience proved very useful when I started popularization for patient associations in the FSHD field. For instance, a trick I found that really helped listeners understand the DUX4 protein toxicity was its personification as a mad duck (Fig. 2A). Patients liked the metaphor of a duck that wakes up genes involved in embryonic development, and hated the mad duck which showed up in their muscle fibres to screw up their fine-tuned gene activation cascade and kill them. At the 2016 FSHD society meeting in Boston, I had bumped into a giant inflated duck that advertised for harbour tours: this gave the idea to punch DUX4 with a pin as a drug strategy! I have recently found pictures of giant ducks floating in different harbours, made by Dutch artist Florentijn Hofman, and one that had blown up because of extreme heat in Hong Kong in 2023: there was the drug-killed DUX4 (Fig. 2B)!

DUX4c has normal functions in skeletal muscles

DUX4 is toxic in muscle:
- it activates genes normally only used in embryo
- it is very similar to DUX4c

DUX4 is a gangster: it takes DUX4c
- target genes
- protein partners

DUX4 kills muscle cells and interferes with muscle regeneration

Fig. 2. 2A: A slide from the presentation of Alexandra at the FSHD Europe Connect meeting in Amsterdam in June 2025, explaining the competition of nasty DUX4 with nice DUX4c duck functions in FSHD muscle cells.
2B: One of the two monumental inflatable rubber ducks in Hong Kong's harbour developed a leak just one day after their unveiling (June 2023). The eighteen-meter-high *Double Ducks* were created by the Dutch artist Florentijn Hofman, originally from Delfzijl. *Photo of ANP.*
2C: An image from the film, depicting how Marion incorporated a duck in her composition, accompanied by the text : 'You express yourself too much. A gene must also be able to keep quiet".

Marion attended a public lecture I gave for patients in Belgium. For the first time since her diagnosis, she felt truly relieved to understand the nature of her disease—many years on, she finally saw a light at the end of the tunnel. She later asked whether I would agree to answer her questions in the film she was preparing with Laëtitia about her life as an artist with FSHD. Marion quickly adopted the DUX4 duck concept and it became part of her poetic creation, of her attempts to make the disease bearable; Marion even fancied she could calm it in one of her cell nuclei (Fig. 2C). Learning that drugs targeting DUX4 were in development was encouraging for Marion, yet of limited relevance to her daily life, as such treatments remained years away. She then embarked on the transformative journey depicted in her remarkable documentary, supported by compassionate individuals—including Vincent Tronel, who, despite living with FSHD and being wheelchair-dependent, has cultivated an inspiring philosophy of life. Following this personal transformation, Marion is now able to focus on the abilities she retains rather than those she has lost.

4. Marion's two metamorphoses: the importance of bracketing biomedical knowledge in patient education (B*aziel van Engelen*)

Marion and Laëtitia's impressive film shows two metamorphoses. The first one occurred at the moment of the diagnosis when she left the hospital empty-handed because "medicine could not cure me, but it could anticipate my condition, condemning me to certain deterioration". Marion concluded that there was nothing she could do: "I thought my life was over, that I could not be happy." In fact, receiving the diagnosis of FSHD in this manner subjected Marion to a secondary condition characterized by fear and despair.

The second metamorphosis—adjusting to life with FSHD, which forms the central theme of the film—unfolded at a significantly slower pace. There is no quick fix, no medical protocol or recipe to deal with FSHD in one's life. Marion gradually managed "to stop identifying with the disease". It took her considerable time to learn how to create psychological distance from her overwhelming symptoms. "What medicine did not tell me was that it could not anticipate the curve of my happiness the way it could for the state of my muscles. I belatedly realized that these two curves were not necessarily going to follow each other." She gradually understood that medicine often fails to convey that a person encompasses more than merely their body and its biological functions.

By setting aside the medicalized perspective of the disease, Marion created space for practical knowledge and lived experience on managing life with FSHD: "…paradoxically enough, is that I regained hope in my life when I came to terms with the fact that I could not be cured." At some point, she realized that if she did not have the power to change reality, it was time to act where she did have power, and that was in the way she looked at the situation. Marion concluded: "I think it is possible to live a healthy life with an illness". She even expressed that FSHD allowed her to find a new meaning in life, stating,: "FSHD can be the opportunity for this inner movement, to move intellectually and philosophically."

In 1951, Parsons delineated the concepts of disease, illness, and sickness to capture distinct perspectives: disease as the physician's clinical diagnosis ("You have a disease"), illness as the patient's subjective experience ("I feel ill"), and sickness as the societal recognition of being unwell ("You look sick") (Fig. 3). At the pivotal moment of diagnosis, the physician imparts the medical definition of disease to the patient. From that point forward, the diagnosis is made by and medically defined by the doctor, while the lived experience of that disease—how the patient perceives and endures it—is called illness.

The disease conveyed by the physician differs from the illness experienced by the patient. While the disease represents a scientific biomedical model, the illness constitutes the patient's personal, subjective experience. As Marion writes, there is "No easy way to transform the constraint and to take the disease as an opportunity to give another meaning to one's life." Medicine is usually not helpful to patients in this respect, only rehabilitation medicine specializes in tools and help to offer for living well *with* FSHD. Indeed, there is "no easy way to transform the constraint and to take the disease as an opportunity". Marion needed something else: time, "brain gymnastics", and "thought artists" – they enabled her to redefine words and concepts to think about what transformation she was going through.

The popularization of the scientific model of FSHD can be valuable, particularly when it is accompanied by a conscious bracketing of the typically dominant and exclusionary biomedical perspective that emphasizes the pursuit of a life free from disease. Conversely, a citizen science perspective may foster the exchange of experiences among patients and create space for practical knowledge on how to live with FSHD. By adopting this perspective at the time of diagnosis, clinicians—not only rehabilitation specialists—can help mitigate the "second illness" of fear and despair, and reduce the nocebo effect associated with the medicalization of the condition. In doing so, they may

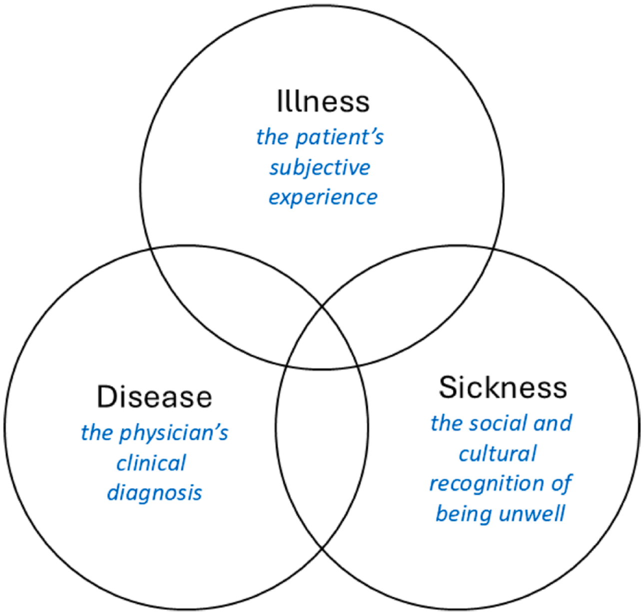

Fig. 3. Distinctions among disease, illness, and sickness as defined by Parsons (1951). Disease refers to the biomedical condition diagnosed by the physician, representing an objective, scientific model. Illness denotes the patient's subjective experience of symptoms and suffering. *Sickness* reflects the social and cultural recognition of an individual as being unwell by others in their community. These overlapping but distinct perspectives highlight the complexity of understanding health conditions in clinical and social contexts.

positively support the patient's transformative journey following the diagnosis.

To help patients metamorphosis directly, we propose to start a Patient Academy, where the patients are the educators, the 'knowers'. By setting aside the medical disease framework, we seek to create space for the individual's lived experience of illness directly at diagnosis. Rather than applying a predefined model of illness to educate patients, the aim is to support the co-creation of a personalized approach that enables individuals to construct their understanding of their condition and learn to live with it.

Our proposal is in line with the approach in the *Phenomenology of Illness* (Oxford University Press, 2016) by Havi Carel. The author, philosopher as well patient with a chronic disease, proposes a structured, three-step approach to understanding the lived experience of illness. This approach is not intended to replace medical knowledge, but to supplement it. It can aid patients who are trying to make sense of their illness, as well as clinicians caring for them. As the first step, Havi Carel encourages patients and clinicians to bracket the "natural attitude"—-that is, to temporarily set aside biomedical assumptions in order to focus on subjective experience, shifting the focus away from the disease entity toward the various features of the experience. Second, the illness experience is thematized by identifying key disruptions such as loss of bodily trust, control, and continuity. This thematizing may include attending to the cognitive, emotive, moral or aesthetic aspects of a phenomenon. Finally, the patient interprets these experiences within the context of their broader life-world, reflecting on how illness shapes identity, relationships, and meaning. This method enables patients to move away from a narrow understanding of disease as biological process, and allows for a richer account of illness as a new way of being in the world. Such a person-centered understanding of illness, supports a clinical practice that is attentive to individual narratives and addresses the risk of epistemic injustice in healthcare.

Carel's approach is compellingly illustrated through Marion's patient journey and its portrayal in the film. Marion's metamorphosis: real hope for patients with chronic diseases, such as FSHD.

5. Patient engagement in care and research *(Ria de Haas and Nicol Voermans)*

The testimonial of Marion and her film underscore a very important aspect of the current research of FSHD: That is that scientific and medical research is perfectly compatible with the inner quest of patients to live their life with FSHD to the fullest. That is why FSHD Europe was very keen to organize the first FSHD Connect Europe meeting in Amsterdam in June 2025. The program provided comprehensive information on the pathophysiology of the disease and the development of treatment options, while also offering workshops on various strategies for optimizing quality of life with FSHD, including the testimonial of Marion as introduction to her film which was shown in the evening.

Patient meetings for individuals with rare diseases, such as those organized by FSHD Europe, offer an invaluable space for patients to connect, share experiences, and build supportive communities. These gatherings provide not only emotional reassurance but also a sense of solidarity among people facing similar challenges. By coming together, patients gain access to critical information about their condition, learn about the latest research, and engage in open discussions with clinicians, researchers, and advocates. Such meetings enable patients to actively engage in their own care, enhance their understanding of the disease, and contribute to a unified voice that advances research, treatment options, and the development of strategies to optimize living with a chronic condition. Ultimately, these patient-centric forums foster a holistic approach to living with rare diseases—addressing both the physical and emotional aspects of the journey and enable patient engagement in research. Individuals with FSHD were thus empowered to gain a comprehensive understanding of medical information, while also discovering that the narrative of FSHD extends beyond clinical aspects.

Declaration of competing interest

None.

Acknowledgements

We are grateful to FSHD Europe, in particular Mrs. Ingrid Molegraaf and the FSHD Europe board members for organizing the FSHD Connect Europe meeting in Amsterdam, 13–15 June 2025. We are thankful to all sponsors of that meeting.

The presentations of the meeting can be seen here: https://fshd-europe.info/fshd-connect-europe/

The movie of Marion Sellenet and Laëtitia Moreau is currently available on Vimeo: https://vimeo.com/ondemand/marionorthemetamorphosis

If no longer available on this platform, you can reach out to Marion Sellenet: https://www.marionsellenet.com/