

Treatments for FSHD: Living a healthy life

DOMENICO MURATORI
FHSI ITALIA, ITALY
NICOL VOERMANS
Radboud University Nijmegen, The Netherlands



Why this document matters: a note on our strategy

This document provides recommendations for living a healthy life with FSHD. Importantly, it is more than a piece of health information. It sets out, in practical terms, one of the strategic aims that FSHD Europe has adopted: improving quality of life and access to effective treatments.

Whether and when new targeted therapies become available does not lie within our control. How well people with FSHD live today, and how ready they are to benefit from those therapies when they arrive, largely does. Making full use of the treatments and care that already exist – multidisciplinary, personalised, and guided by the renewed international standards of care – is therefore not simply good clinical practice: it is the first, concrete step in our community's preparation for the era of disease-modifying treatments.

This is what we call **treatment readiness**, and it has two dimensions. At the **individual level**, it means being in the best possible physical, mental and social condition to benefit from a therapy when it becomes available. At the **level of health systems**, it means being organised to deliver that therapy in practice: able to identify and inform eligible patients in good time – including the many who received a diagnosis years ago, sometimes without genetic confirmation, and are no longer followed by a specialist centre – to verify eligibility without creating bottlenecks, and to administer treatment regularly wherever people live, not only where specialist centres exist.

Alongside treatment readiness stands a distinct challenge: **treatment access**. Whether an approved treatment actually reaches the people who need it will also depend on national reimbursement and access procedures, and preparing patient communities to understand and navigate them is a strategic aim of FSHD Europe in its own right. Work is under way on both fronts – through the international Project Mercury on system preparedness and through the PaLaDIn project on access – because a treatment can only change a life if the person is in a condition to benefit from it, the system is able to provide it, and it can actually be obtained.

This document focusses on treatment readiness at the individual level. It is the work of a clinician; a patient advocate contributed to framing this introduction – a small example of the way **FSHD Europe co-creates**, with clinical expertise and lived experience side by side. We hope it will be read, translated, adapted and used by our member organisations across Europe – not only as information for individuals and families, but as a shared agenda for what our community can do now, without waiting.

Treatments for FSHD: Living a healthy life

Treatment and cure

People often hear that there is currently “no cure for FSHD.” While this is true, it does not mean that there are no treatments available. A cure and a treatment are not the same thing. A cure would completely remove the disease, whereas a treatment aims to improve health, reduce symptoms, or slow disease progression. In fact, most of the new therapies currently being developed for people with FSHD are targeted treatments rather than cures. Healthcare professionals have long used a range of approaches—including exercise, physiotherapy, assistive devices, surgery, and medication—to help people manage symptoms and maintain their quality of life. These symptomatic treatments can make a meaningful difference, even though they do not eliminate the underlying cause of FSHD.

Figure 1: The spectrum of prevention, treatment and cure. Current treatments are symptomatic, addressing the complications of FSHD.



While we wait for new targeted therapies to become available, it is important to make full use of the symptomatic treatments that already exist. These can help manage symptoms, maintain function, and improve quality of life. They are important not only for your health today, but also for the future. Being in the best possible condition may increase your ability to participate in ongoing and upcoming clinical trials, and to benefit mostly from future therapies. This is sometimes referred to as being ‘trial-fit’ and ‘treatment-fit’.

At the same time, researchers and clinicians are working to improve the design and conduct of clinical trials—a process known as “trial readiness.” Hence, patients can play

an important role in the good execution of clinical trials by staying as healthy and active as possible and making use of the treatments and support that are available.

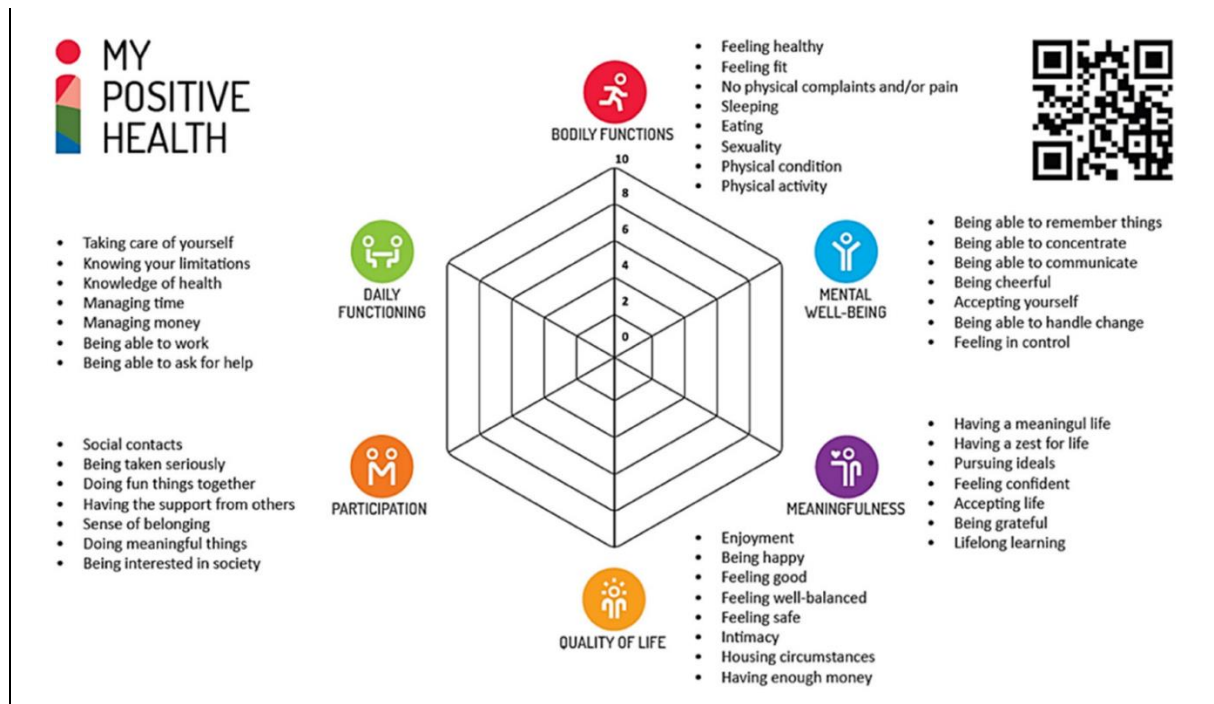
Multidisciplinary, personalized care

FSHD is more than a medical condition that affects muscles. It can influence many aspects of a person's life, including how they feel physically, emotionally, and socially. Hence, the impact of FSHD is not limited to symptoms alone. Living with FSHD can affect daily activities, work, education, relationships, and participation in society. In this way, the experience of illness is shaped not only by the condition itself, but also by its wider social consequences and the environment in which a person lives.

Positive health

The concept of Positive Health, developed by Machteld Huber ([Positieve gezondheid - Stichting Institute for Positive Health \(iPH\)](#)) takes the broad impact of FSHD into account. It encourages us to look beyond disease and focus on a person's ability to adapt and manage the physical, emotional, and social challenges of life. For people with FSHD, this means that health is not defined solely by muscle strength or physical limitations. It also includes aspects such as mental well-being, meaningful relationships, participation in daily activities, and a sense of purpose. Even when living with a chronic condition like FSHD, there are many ways to strengthen health and quality of life by focusing on what matters most to the individual. This underscores that improving health is not solely the responsibility of healthcare professionals and can occur in many aspects of daily life.

Figure 2: The Positive Health framework views health as more than the absence of disease. It helps people with FSHD reflect on six dimensions of health and well-being, including physical, emotional, and social functioning. By assessing each domain, individuals can identify areas that are going well and areas where additional support or lifestyle changes may help improve their quality of life, and discuss this at their visit to the clinic.



This broader perspective on health also highlights the importance of multidisciplinary care in FSHD. Managing symptoms and maintaining quality of life often requires the expertise of several healthcare professionals working together. In my centre in the Netherlands, people with FSHD are seen in a specialized clinic led by a rehabilitation physician, who collaborates closely with physical therapists, occupational therapists, speech and language therapists, psychologists, dietitians, and social workers. Together, they address the many physical, emotional, and social challenges that can accompany FSHD.

Expert consensus recommendations on the diagnosis and management of FSHD

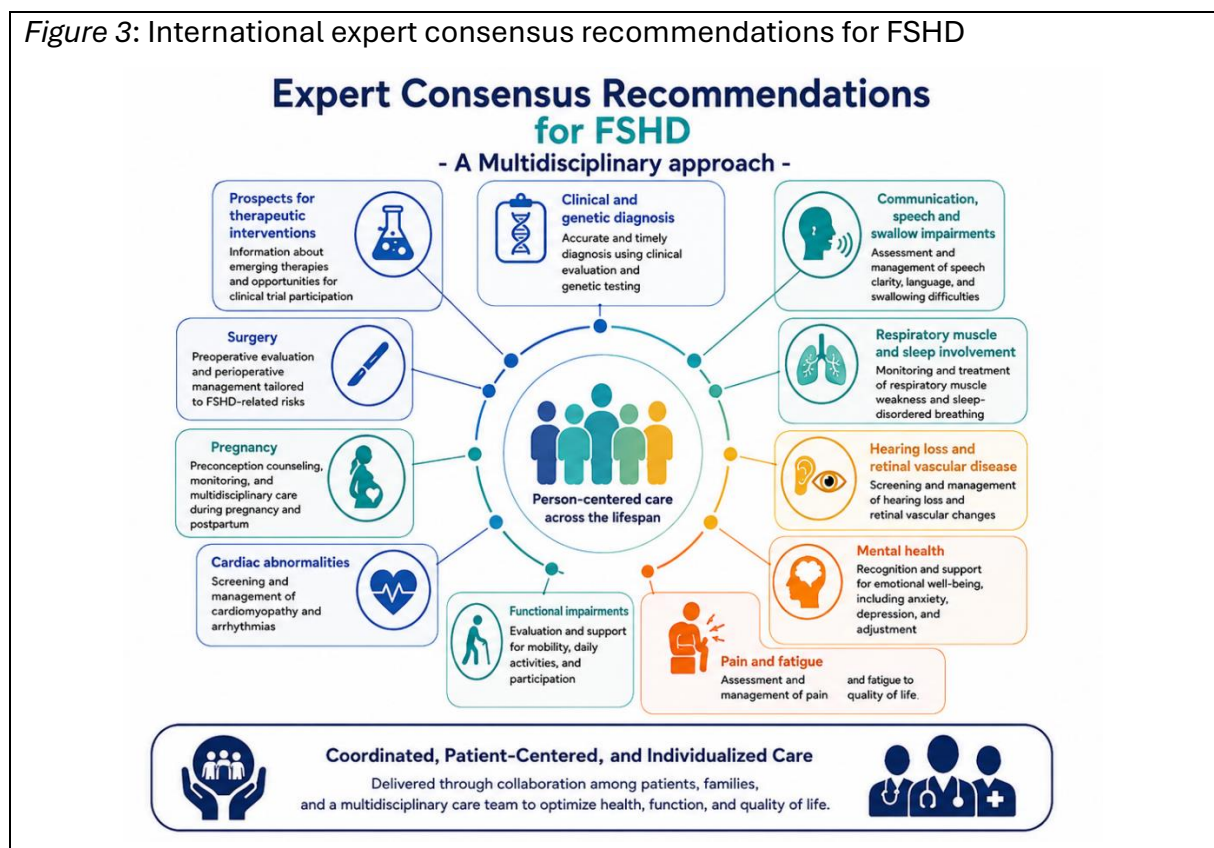
This wide range of topics is included in the renewed international expert consensus recommendations on FSHD, which are expected to become available by the end of 2026 (Figure 3). The last decade, the most widely used evidence-based guideline has been the 2015 guideline from the American Academy of Neurology. This guideline, led by Rabi Tawil from Rochester focused on diagnosis, genetic testing, pain, scapular fixation surgery, retinal screening, hearing assessment, respiratory function, and exercise recommendations. Since then, an international multidisciplinary working group coordinated by the FSHD Society, EURO-NMD and FSHD Europe has developed an updated international expert consensus recommendations. This effort involved approximately 40 experts from multiple countries, together with patient representatives. The updated guideline expands substantially on the 2015 version and reflects current knowledge and the emerging era of disease-modifying therapies.

The updated international expert consensus recommendations covers 13 major domains, including:

- Clinical and genetic diagnosis
- Pain and fatigue management
- Functional limitations and rehabilitation
- Exercise and physical activity
- Pulmonary and sleep disorders
- Cardiac assessment
- Orthopaedic and surgical interventions
- Hearing loss
- Retinal vascular disease
- Communication, speech, and swallowing
- Mental health
- Pregnancy, birth, and parenting
- Emerging pharmacological therapies and treatment readiness

A central theme of the new expert consensus recommendations is that FSHD care should be multidisciplinary, personalized, proactive, and focused on participation and quality of life, rather than solely on muscle weakness. The guidelines also emphasize optimizing health before disease-modifying therapies become widely available. We will discuss some of the topics below.

Figure 3: International expert consensus recommendations for FSHD



Symptomatic treatment for FSHD should be personalized. Although people with FSHD share the same diagnosis, the challenges they face can differ considerably. For one person, shoulder weakness may be the main concern; for another, fatigue, pain, difficulties with mobility, or problems at work may have a greater impact on daily life. Therefore, treatment should focus on the issues that matter most to you. When discussing treatment options, it can be helpful to ask yourself: What are the biggest challenges I am facing? Which activities have become more difficult because of FSHD? What would I most like to improve? And what goals are realistic and meaningful for me at this stage of my life? By identifying your personal priorities, you and your healthcare team can work together to develop a treatment plan that best supports your health, independence, and quality of life.

Breathing and cardiac care

The simplest way to assess breathing muscle strength in people with FSHD is through spirometry, a test that measures how much air you can breathe out and how quickly you can do so. In neuromuscular disorders, weakness of the respiratory muscles can reduce the *forced vital capacity* (FVC), which is the maximum amount of air that can be exhaled after taking a deep breath. Ideally, spirometry should be performed both while sitting and while lying down. When lying flat, the diaphragm has to work harder because the abdominal contents shift upwards, and this position can reveal weakness that may not be apparent when sitting.

Weakness of the breathing muscles may lead to inadequate ventilation during sleep, causing carbon dioxide to build up in the blood. This can result in symptoms such as shortness of breath when lying flat or bending forward, restless or unrefreshing sleep, morning headaches, daytime fatigue, and excessive sleepiness. These symptoms occur because elevated carbon dioxide levels can repeatedly disrupt sleep throughout the night. Although these interruptions are often too brief to be noticed, they can prevent the deep, restorative stages of sleep from being reached.

For this reason, regular assessment of respiratory function is important, even in people who do not experience breathlessness during the day. In FSHD, respiratory weakness is particularly common in individuals who use a wheelchair or have significant scoliosis. Studies suggest that up to 40% of these individuals may develop respiratory muscle weakness severe enough to require additional monitoring or respiratory support.

Cardiac evaluation is also often performed when a person with FSHD first establishes care with a specialist. Reassuringly, FSHD does not usually affect the heart muscle itself, and cardiac imaging, such as an echocardiogram (ultrasound of the heart), is typically

normal. However, some people with FSHD may have minor abnormalities in the electrical conduction system of the heart that can be detected on an electrocardiogram (ECG). The most common finding is a right bundle branch block, which means that the electrical signal travels slightly more slowly through part of the heart. In most cases, this does not cause symptoms, does not affect heart function, and does not worsen over time. If such an abnormality is detected, a follow-up ECG may be recommended after one or two years. Patients are also informed of the finding so that they can mention it if they undergo cardiac testing in the future.

Chronic pain and fatigue

Chronic pain and fatigue are common symptoms in FSHD and can have a major impact on daily life. These issues are important to discuss with your physical therapist, occupational therapist, or rehabilitation physician, as a range of treatment strategies may be available. In addition, educational resources on pain management, such as those developed by Dr. Katherine Mathews and colleagues (available on FSHD Society website), can provide practical advice and support.

Research has shown that non-pharmacological interventions can be effective in reducing fatigue and improving quality of life. For example, a study led by Nicole Voet evaluated a home-based exercise programme combined with cognitive behavioural therapy aimed at increasing physical activity in daily life. Compared with usual care, participants became more physically active, slept better, experienced less fatigue, and were able to participate more fully in social activities.

Another rehabilitation programme, known as the Energy Self-Management Programme, was developed for people with neuromuscular disorders experiencing severe fatigue. Participants learned practical strategies to manage their energy levels and balance activities throughout the day. The study found improvements in walking ability, physical endurance, and social participation, and these benefits were still present nearly a year later. An additional advantage reported by many participants was the opportunity to connect with others facing similar challenges. The support and shared experiences within the group were often described as valuable and lasting benefits of the programme.

Promotion of a healthy lifestyle

Lifestyle interventions are an important part of managing FSHD and maintaining overall health. In the general population, muscle mass and strength increase during childhood and early adulthood, remain relatively stable for some years, and then gradually decline from around the age of 30 to 35. The amount of muscle strength a person has throughout

life is influenced not only by genetics and disease, but also by lifestyle factors such as physical activity, nutrition, sleep, and smoking.

There is considerable variation in muscle strength and physical fitness between individuals. Although FSHD causes muscle weakness, lifestyle factors can still make an important difference. A person with FSHD who maintains a healthy lifestyle may be fitter and stronger than someone without FSHD who is physically inactive or has other health conditions such as diabetes or chronic lung disease. Lifestyle interventions cannot cure FSHD, but they can help preserve health, improve resilience, and provide a meaningful advantage over time.

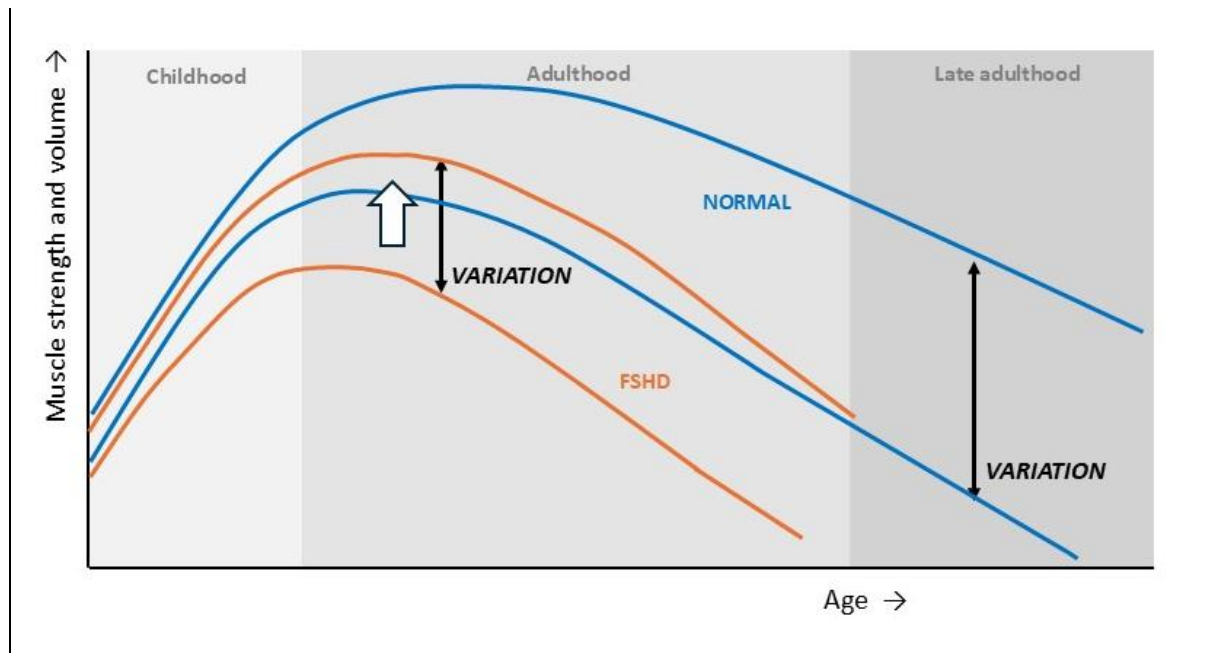
Improving your lifestyle is not about taking extraordinary food supplements or engaging in high-intensity sports. It is about making sensible, everyday choices that support a healthier life. Eat a varied and balanced diet, avoid overeating, maintain a healthy balance between activity and rest, and incorporate physical activity into your daily routine. This can include walking, cycling, taking the stairs, gardening, standing, household activities, or participating in adapted sports. It is also important to avoid (illicit) drugs. This includes avoiding nicotine and alcohol. The WHO has recently emphasized an important message regarding alcohol consumption: the healthiest choice is not to drink alcohol at all. Alcohol is toxic to the body, and even relatively small amounts can have adverse effects on health. Regular alcohol consumption is associated with a wide range of health risks. Therefore, when it comes to alcohol, less is better—and none is best.

For this reason, I often ask my patients: What aspects of your lifestyle could you improve over the next few years? Which changes are realistic and sustainable for you? What support would help you achieve them? No one has a perfect lifestyle, but there is almost always an opportunity to make small changes that can benefit health and well-being in the long term.

Figure 4: Muscle strength and volume over the lifespan in people with and without FSHD.

Muscle strength and volume normally increase during childhood and early adulthood, reach a peak in early adulthood, and gradually decline with ageing. In people with FSHD, muscle strength and volume are generally reduced compared with the general population, and the decline may start earlier or progress differently. However, there is considerable variation between individuals with FSHD, with some people maintaining relatively high levels of function for many years while others experience more rapid decline.

This variation is influenced by multiple factors, including genetic background, disease severity, lifestyle, physical activity, and supportive care. The goal of current management is to preserve muscle function, maximize health and participation, and maintain the highest possible level of independence throughout life (white arrow).



Mental and social well-being.

Health and well-being involve more than physical health alone. Mental and social well-being are equally important, particularly when living with a chronic condition such as FSHD. Some people, especially adolescents and young adults, may hesitate to seek psychological support because they do not feel they have a mental health problem. However, psychological care is not only for people in distress. Just as physical therapy helps strengthen muscles and improve physical function, psychological support can help people develop skills to cope with challenges, build resilience, and improve their quality of life.

One approach used by many psychologists is Acceptance and Commitment Therapy (ACT), which focuses on helping people live meaningful and fulfilling lives while adapting to the realities of a chronic condition. These skills can be valuable at any stage of life and can help people manage uncertainty, changing abilities, and the social impact of FSHD. Another important aspect of well-being in FSHD is facial weakness and its effect on communication and social participation. Research has shown that facial weakness can influence how people express emotions and how they are perceived by others. In a study of people with FSHD, many older participants reflected that facial weakness had been particularly challenging during adolescence and young adulthood, when self-image and social acceptance are especially important. Although many reported becoming more confident over time, they also felt that additional support would have been valuable when they were younger.

Today, with the growing importance of photographs, video calls, and social media, facial weakness may be an even greater concern for some young people with FSHD. For this

reason, facial expression and communication should be considered as part of multidisciplinary care. Speech and language therapists can help people develop strategies to compensate for facial weakness and improve communication in daily life. As our understanding of these challenges grows, new approaches, including group-based support programs, may help people share experiences, develop practical skills, and build confidence.

Care for children with FSHD

More recently, childhood-onset FSHD has come to be viewed as part of the broader FSHD disease spectrum rather than as a separate condition. This recognition is important because it supports the inclusion of children and adolescents in clinical research and may help ensure that they have access to emerging therapies as these become available in the future.

Children who develop symptoms of FSHD before the age of 10 often have a more severe form of the condition. They are at increased risk of hearing loss and abnormalities of the blood vessels in the retina, the light-sensitive tissue at the back of the eye. Regular hearing and eye examinations are therefore recommended. Early detection is important because timely treatment can help prevent vision loss and reduce the impact of hearing impairment on speech and language development.

There are also children who are diagnosed early because of a family history of FSHD and who have only mild symptoms, such as difficulty smiling, closing their eyes completely, running, or keeping up with physical activities. Although these challenges may not always be visible to others, they can have a significant impact on daily life. Many children with FSHD work hard to keep pace with their peers and may experience fatigue, reduced physical endurance, or difficulties at school. Because these challenges are often invisible, they may be overlooked. Providing appropriate support at home, at school, and within the healthcare system is essential to help children reach their full potential.

FSHD also affects families, not just individuals. For this reason, genetic counselling is an important component of care. Genetic counselling can help individuals and families understand the inheritance of FSHD, discuss the implications for relatives, and explore reproductive options when relevant.

Prepared for medical emergencies

Finally, it is important to be prepared for medical emergencies. If you are involved in an accident or require urgent medical care, healthcare professionals may not be familiar with FSHD. Carrying a medical alert card can provide important information about your

condition and help guide medical decision-making, particularly if you are unable to communicate. Many patient organisations provide medical alert cards that can be printed and carried in a wallet. It may also be helpful to store relevant medical information in an “In Case of Emergency” (ICE) application on your phone so that it can be accessed quickly when needed.

Conclusion

To conclude, concurrent management of FSHD extends far beyond treating muscle weakness and requires a multidisciplinary, personalized approach focused on health, participation, and quality of life. While disease-modifying therapies are emerging, optimizing symptomatic treatment, lifestyle, physical and mental well-being, and treatment readiness can help individuals maintain function, independence, and resilience throughout life.