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How Modern Physiotherapy Can Transform the Lives of People Living With Rare Diseases

Before, during and after diagnosis – and how objective data can influence outcomes

For people living with rare and ultra-rare diseases, the path to diagnosis is often long and arduous. Across Europe, patients wait an average of **five years** to receive a correct diagnosis, with many waiting significantly longer (EURORDIS, 2023). During this time, they undergo repeated consultations, misdiagnoses and unnecessary investigations, often living for years with pain, functional decline and uncertainty.

Even after diagnosis, more than **90% of rare diseases have no curative treatment**. Those affected must live with chronic symptoms, fluctuating capacity and long-term functional limitations. Against this backdrop, **modern, data-driven physiotherapy** is increasingly establishing itself as a central, yet still under-recognised, pillar of care for people with rare diseases.

Rare diseases: individually rare, collectively significant
In Europe, a condition is considered rare when it affects fewer than **1 in 2,000 people** (Orphanet, 2024). Taken together, however, more than **30 million people in Europe** live with a rare disease (EURORDIS, 2023).

Conditions such as **Hypophosphatasia (HPP)** and **Ehlers-Danlos syndromes (EDS)** exemplify rare diseases with pronounced musculoskeletal involvement. HPP is a genetic metabolic bone disorder characterised by impaired bone mineralisation, chronic pain, muscle weakness and recurrent fractures (Whyte, 2017). EDS encompasses rare connective tissue disorders marked by joint hypermobility, instability and chronic pain, with diagnosis often delayed due to symptom overlap with more common conditions (Malfait et al., 2017).
Beyond HPP and EDS, specialised physiotherapy is increasingly relevant for rare conditions with complex pain syndromes or neurological and neuromuscular involvement. These include **Complex Regional Pain Syndrome (CRPS)**, which requires graded and sensorimotor retraining; **erythromelalgia**, where multidisciplinary rehabilitation can improve physical functioning; **trigemino-autonomic headache disorders and rare migraine variants**, where manual therapy targeting cervical dysfunction plays an important role; and selected **rare neuromuscular diseases**, in which physiotherapy can significantly enhance physical function.
Despite their clinical diversity, these conditions share common challenges: delayed diagnosis, fluctuating symptoms, limited treatment options and a lack of objective, long-term functional data. Structured physiotherapeutic assessment is therefore valuable not only for symptom management, but also for **diagnostic clarification, treatment evaluation and long-term care planning**.

Physiotherapy as an evidence generator

Modern physiotherapy in rare diseases goes far beyond rehabilitation alone. It represents a **long-term partnership**, supporting patients before diagnosis, during therapeutic decision-making and throughout the course of their condition.

A central element of this approach is **repeated objective measurement**. In rare diseases, clinically meaningful changes may be subtle, yet they can have a profound impact on daily functioning and independence. Comprehensive strength testing, pressure pain threshold assessments and validated activity measures allow physiotherapists to identify patterns that might otherwise remain undetected (World Health Organization, 2022).

At the heart of this work is the MWS Score, a baseline measure of wellbeing and physical performance that is currently undergoing validation. By integrating multiple dimensions of physical capacity into a single longitudinal metric, the MWS Score enables patients, clinicians and payers to better understand functional trajectories despite the presence of a rare disease. This represents a promising approach to capturing subtle functional changes that standardised instruments may overlook.

Supporting earlier diagnosis

Physiotherapy plays a critical role in supporting **earlier diagnostic clarification**. When patients present with persistent pain, weakness or fatigue that does not follow typical patterns, longitudinal physiotherapy data can reveal atypical responses to standard interventions. By documenting unexpected functional trajectories, physiotherapy can prompt earlier referral for specialised assessment, genetic testing or metabolic evaluation, thereby helping to shorten the diagnostic odyssey (EURORDIS, 2023). This is particularly relevant in conditions such as CRPS, where early recognition is crucial for prognosis, and erythromelalgia, where symptom recognition is often challenging.

A Swiss case example: Hypophosphatasia

A Swiss patient with Hypophosphatasia illustrates this impact clearly. She received her diagnosis in adulthood, at the age of **37**, after more than **ten years** of unexplained pain, recurrent fractures, declining strength and repeated misdiagnoses across two countries.
Following diagnosis, comprehensive strength testing, pressure pain threshold assessments and longitudinal monitoring using the **MWS Score** enabled both clinically meaningful functional improvement and objective documentation of change over time. These data served several essential purposes: they demonstrated the real-world effectiveness of the prescribed therapy, supported clinical decision-making through measurable outcomes beyond subjective symptom reporting, and provided objective functional evidence to support the physician's justification for continued health insurance reimbursement of a high-cost treatment.

The MWS Score also serves as a **practical patient support tool**. By making changes in physical capacity visible and understandable, it enables patients to manage their symptoms more dynamically, adjust activity levels safely and participate actively in therapeutic decision-making.

Managing fatigue and the invisible burden

Fatigue and cognitive impairment ("brain fog") are among the most burdensome symptoms experienced by people with rare diseases, yet they are frequently underestimated in clinical practice (Moss-Morris et al., 2020). Overexertion, even with low-intensity activity, poses a significant risk.

For this reason, **pacing strategies** are central to sustainable care. Activities are aligned with individual energy reserves to avoid "boom-and-bust" cycles (NICE, 2020). This approach is equally essential for people with CRPS, where fear-driven movement avoidance must be addressed through graded exposure, and for those with erythromelalgia, where trigger management plays a key role.

Key insights

- **Physiotherapy generates evidence, not just exercises**
  Objective, longitudinal data, including the MWS Score, support diagnosis, guide therapeutic decisions and demonstrate real-world impact.

- **Physiotherapy shortens the diagnostic journey**
  Documenting atypical functional trajectories accelerates referral to specialists and correct diagnosis.

- **Objective data secure access to high-cost therapies**In conditions such as Hypophosphatasia, physiotherapy-derived measurements provide essential evidence to support reimbursement decisions and long-term treatment access.

From evidence to action

What distinguishes specialised physiotherapy in rare diseases is **precision combined with personalisation**. It integrates clinical expertise with long-term, patient-centred data, enabling care that continuously adapts to the realities of living with a rare condition.
At practices such as **MWS Physio in Switzerland**, physiotherapy is delivered as an **evidence-generating discipline**. By integrating comprehensive functional assessments such as the MWS Score, this approach supports patients, clinicians and payers alike, translating complex symptom patterns into measurable insights that positively inform daily life, clinical decision-making and long-term care planning.

For patients and families who often live with uncertainty for years, and for clinicians who require objective data to support diagnosis, therapeutic decisions or reimbursement discussions, specialised physiotherapy offers meaningful support. Rare diseases demand an exceptional degree of precision, collaboration and persistence. Modern physiotherapy, when understood and practised as an evidence-generating discipline, can make a decisive contribution to all three.

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