



CoMPaSS-NMD



Funded by
the European Union

Computational models for new patients stratification strategies of neuromuscular disorders

Issue 3 - October 2025

Welcome to the third issue of the periodic project newsletter that will keep you updated on the CoMPaSS-NMD latest achievements and upcoming events.

In this issue:

1. The launch of our first **webinar series** dedicated to patient, family members and caragivers.
 2. Feature topic: **Why One Gene Can Cause Different Diseases**
 3. Past and next **events**
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Nice to Meet You, We Are CoMPaSS-NMD! 🤝

The project aims to create a new generation of methods for “precision diagnosis” that allow researchers and healthcare professionals to successfully classify patients affected by **Hereditary Neuromuscular Diseases (HNMDs)**, by adopting a new multidimensional approach based on Artificial Intelligence (AI) for **better and faster diagnoses and personalised treatments**.

HNMDs often strike early in life, causing severe disabilities and significantly reducing life expectancy. These conditions hinder daily functioning, leading to social isolation and the need for constant care. Many patients require long-term institutionalisation, putting strain on both families and healthcare systems.

What does CoMPaSS-NMD do?

In CoMPaSS-NMD, artificial intelligence guides the physicians towards a more accurate diagnosis of HNMDs.

CoMPaSS-NMD will analyse a large amount of diverse data sources, among which genetic and clinical information, collected from project partner clinics. Thanks to these data, it will create one of the most advanced diagnostic computational tools.

Artificial intelligence will give the chance to identify connections between data that are not recognisable with standard methods, **extending the diagnostic proficiencies of the physicians**.

Impact

The main expected outcomes of the project are:

- **An AI-based method that enables a faster and more accurate diagnosis of HNMDs able to promote** effective actions by European national health systems, ameliorating the

quality of life of patients and caregivers and reducing needless expenses.

- **The CoMPaSS-NMD Neuromuscular Genome Atlas: a repository of genetic, imaging, and histopathological data of HNMD patients from six clinical centres in five countries**, which will incentivise the development of strategies that integrate health data to support patients, healthcare professionals and citizens.

The consortium



For more information visit our website

Navigating NMDs: Your CoMPaSS to Care



From September to December 2025 the CoMPaSS-NMD project is happy to invite you to attend a **free webinar series on the future of neuromuscular disease diagnosis and treatment.**

Discover how our project is harnessing the power of a unique 'socio-technology' to create a better future for the nearly 80,000 patients affected by these conditions. We'll show you how we're using Artificial Intelligence to speed up the diagnostic journey, enhance quality of life, and empower patients and their families.

Why You Should Attend

These webinars are for anyone with a stake in the future of NMD care. Whether you are a **patient, a family member, or a caregiver**, this is your opportunity to gain clear, trustworthy information and find answers to your most pressing questions.

What You'll Learn

- ▲ **AI for NMD diagnosis, Simplified:** We'll demystify how AI is being used as an ally for doctors and patients.
- ▲ **Find Answers and Clarity:** Learn how we're tackling the lack of awareness and understanding around NMDs.
- ▲ **Data Security and Privacy:** Understand the robust security and privacy measures adopted to protect your sensitive health data.
- ▲ **A New Resource for NMD Care:** Get a firsthand look at the

ATLAS, an innovative tool designed to improve the diagnostic journey.





!! Important! The official language of the webinar is English, but simultaneous translation into German and Italian is available.

Registration is free and grants you access to all four live webinars. If they Can't make it live? Register anyway to receive the recordings.

[Register here!](#)

Navigating NMDs: Your CoMPaSS to Care

The free webinar series on the future of [neuromuscular disease diagnosis and treatment](#)

- **NMD care: bridging today and tomorrow for you**
22 September 2025 at 5pm
Gain Clarity and Understanding
- **AI in NMDs diagnosis, an ally for doctors and patients**
20 October 2025 at 5pm
Demystify Artificial Intelligence
- **Your sensitive data: secure and handled with care**
25 November 2025 at 5pm
Understand Your Data Privacy
- **Beyond the symptoms: how the CoMPaSS-NMD Atlas is improving patient care**
15 December 2025 at 5pm
Discover Powerful New Resources

[Patients](#) | [Family members](#) | [Caregivers](#) | [Healthcare professionals](#) | [Researchers](#)

REGISTER HERE

Feature topic



Why One Gene Can Cause Different Diseases

For years, the foundation of medical genetics rested on a simple, elegant idea: **one gene, one disease**. This model successfully explained many rare disorders, including numerous **neuromuscular diseases (NMDs)**.

But as research advances, reality is proving far more complex. We now know that the same gene can be implicated in completely different diseases, depending on the specific mutation and the tissues affected. This "split personality" of genes is reshaping our understanding of NMDs and is key to developing **personalized treatments**.

Beyond the Simple Map

Our latest blog article, "**Beyond the 'One Gene – One Disease' Paradigm,**" dives deep into this fascinating challenge.

You'll discover:

- **Striking Examples:** See how mutations in a single gene like **LMNA** (lamin A/C) can lead to distinctly different conditions, including various forms of muscular dystrophy, heart failure, or

neuropathy, based on whether the nuclear envelope is destabilized in the muscle, heart, or nerve.

- **A "Split Personality" Gene:** We highlight the gene *DNM2* as a perfect illustration. Depending on the mutation, it can cause either **Centronuclear Myopathy (CNM)**—a muscle disease— or **Charcot–Marie–Tooth (CMT) neuropathy**—a peripheral nerve disease.
- **Opposite Therapies for the Same Gene:** The most remarkable finding? The CNM mutation is a **gain of function** (too much protein activity), while the CMT mutation is a **loss of function** (too little activity). This means effective treatments must move in **opposite directions**, underscoring the critical need for precision.

The Path to Precision

Understanding how one genetic change can "speak different languages" in the body is more than just a scientific curiosity—it is the direct path toward more accurate diagnoses and more effective, targeted therapies.

This complexity is precisely why the **CoMPaSS-NMD consortium** exists. By bringing together experts to integrate genetic, cellular, and clinical data, we are working to decode these intricate mechanisms and move rare neuromuscular disease treatment into a new era of **precision medicine**.

[Read our new article on the CoMPaSS-NMD website](#)

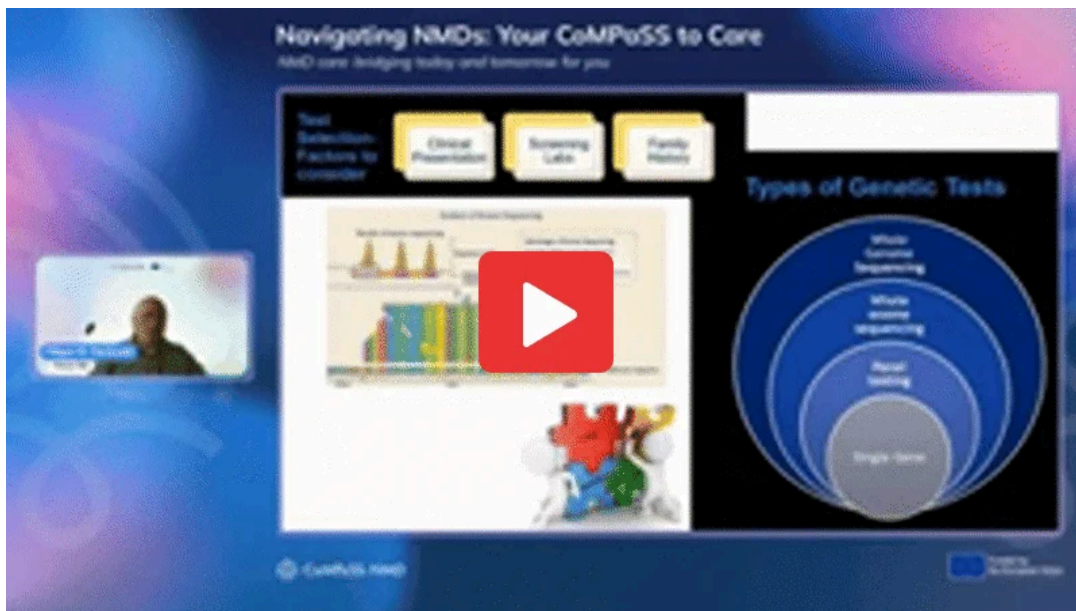
Past webinars

Did you miss our first webinar of the series "Navigating NMDs: Your CoMPaSS to Care"?

You can watch the recording of the first webinar: **NMD care: bridging today and tomorrow for you.**

Host: **Rossella Tupler**, Geneticist at University of Modena and Reggio Emilia, Italy

Speaker: **Filippo Maria Santorelli**, MD at IRCCS Fondazione Stella Maris, Italy



Next events - SAVE THE DATE

- **Synchronous Distance Learning Course on Rare Genetic Neuropathies in Pediatric Age** - Organised by [ACMT-Reteper la malattia di Charcot-Marie-Tooth](#). **Official language: Italian.** Read the course details and register [here](#)

- **20 October 2025** at 17:00 CET join our second webinar: **AI in NMDs diagnosis, an ally for doctors and patients.**

!! Important! The official language of the webinar is English, but simultaneous translation into German and Italian is available.

[Register here!](#)

- **Young Investigator Training initiative 2026:**
 - **Spring 2026 - The CoMPaSS-NMD Atlas**
 - **YIT Summer School 2026**
 - [Visit the YIT page to stay updated on and register for the webinar.](#)

VISIT OUR WEBSITE FOR UPDATES ON THE EVENTS!

CoMPaSS-NMD

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