



CoMPaSS-NMD



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Computational models for new patients stratification strategies of neuromuscular disorders

Issue 2 - April 2025

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

Your health, our CoMPaSS

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.
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The consortium



For more information visit our website

Project progresses

Did you know that CoMPaSS-NMD is present in the Orphanet platform?

Orphanet is a comprehensive online resource dedicated to gathering and providing information on rare diseases and orphan drugs. Established in France in 1997 at the advent of the internet, its primary goal is to improve the diagnosis, care, and treatment of individuals living with rare diseases. It maintains a database of rare diseases, including their classifications and related genes and provides directories of:

- Orphan drugs
- Patient organizations
- Healthcare professionals and institutions
- Expert centers
- Diagnostic laboratories

- Ongoing research and clinical trials

From 2000, this initiative became a European endeavour supported by grants from the : Orphanet has gradually grown to a network of 41 countries, within Europe and across the globe. Over the past 20 years, Orphanet has become the reference source of information on rare diseases. As such, Orphanet is committed to meeting new challenges arise from a rapidly evolving political, scientific, and informatics landscape. To empower everyone – healthcare professionals, patients, and caregivers – with readily accessible, high-quality online information, facilitating the identification of rare disease patients and contributing to the creation of extensive, computable, and reusable scientific data.

[Visit the CoMPaSS-NMD Orphanet page!](#)

Nice to Meet You, We Are CoMPaSS-NMD! 🤝

On **World Rare Disease Day, February 28th, 2025**, we weto kick off our awareness campaign: "**Nice to meet you, we are CoMPaSS-NMD!**".

Have you ever wondered how cutting-edge technology can revolutionise the lives of those affected by rare diseases? We're here to show you.

Enjoy **the video interview of our coordinator Rossella Tupler, [Università degli Studi di Modena e Reggio Emilia](#)**, by clicking on the image below.

CoMPaSS-NMD

Modelli computazionali per nuove strategie di stratificazione dei pazienti affetti da malattie neuromuscolari ereditarie



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In the coming months, the project partners will:

- *Recruit patients to collect data for the validation of the AI algorithms. If interested, read more about [the clinical study](#).*
- *Optimisation of the [ATLAS architecture](#).*
- *Define patient superclusters and their multi-omics profiles.*

Feature topic



Genes, muscles, and answers: how genetic counseling can help with neuromuscular diseases

Neuromuscular diseases (NMDs) present significant diagnostic and management challenges due to their diverse genetic origins and clinical manifestations. Understanding the intricate relationship between an individual's genetic makeup (genotype) and observable characteristics (phenotype) is crucial for effective diagnosis and personalized care. This article explores the complexities of genotype-phenotype correlations in NMDs, highlighting how factors beyond the primary mutation, such as modifier genes, sex, age, and environment, contribute to the variability in disease presentation. Furthermore, it emphasizes the indispensable role of genetic counseling in navigating these complexities, aiding in diagnosis, treatment planning, and family guidance.

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



Standardization and automation for re-analysis of unsolved rare muscle disease patient exomes

Diagnosing rare neuromuscular diseases (NMDs) is challenging due to the complexity of analyzing vast genetic data from whole exome sequencing (WES), often leaving patients without answers. Bioinformatics tools like Exomiser streamline this process by prioritizing disease-causing variants using patient phenotypes and diverse databases, achieving high accuracy in studies. These tools offer more efficient analysis and re-analysis, a consistent approach, and potential for new gene discoveries. However, data quality and unknown genetic causes remain limitations, requiring expert interpretation. Future advancements in sequencing (WGS) and tool development promise faster, more accurate diagnoses, with a need to address non-Mendelian inheritance patterns for complex cases.

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

Past events

The CoMPaSS-NMD seminar, "Omics Analysis of Skeletal Muscle Homeostasis under Normal and Pathological Conditions,"

took place on March 11, 2025, at the I.G.B.M.C. in Strasbourg, France, as a hybrid event (both in-person and online). Aimed primarily at healthcare professionals and researchers in neuromuscular diseases (NMD), the seminar featured three speakers: **Gisèle Bonne** discussed European-level omics strategies focusing on the genetics and pathophysiology of NMDs, particularly laminopathies and collagen VI-related myopathies, with an emphasis on translational research. **Fabrice Bertile** presented muscle proteomics research in hibernating brown bears, exploring links to physiology, aging, and environmental contamination. **Alain Mayer** focused on transcriptomic analysis of inflammatory myopathies with the goal of improving early diagnosis and patient monitoring. Registration was required to attend, with online access provided to remote participants and recordings available for those unable to attend live.

[You can find the slides and the recording of the seminar here on the website.](#)



Next events - SAVE THE DATE

- **Mid May 2025 - CoMPaSS-NMD event in Modena for Italian patients, family members and caregivers**, in collaboration with UILDM (Unione Italiana Lotta alla Distrofia Muscolare).
- **Young Investigator Training initiative:**
 - **June 2025: YIT 2 – Part 1 Electronic Histological data Clinical Report Form (eHCRF)**
 - **July 2025: YIT 2 – Part 2 Electronic MRI data Clinical Report Form (eMRICRF)**
 - **September 2025: YIT 3 – Electronic Genetic data Clinical Report Form (eGCRF)**

[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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