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ORIGINAL ARTICLE

Empowering clinicians with artificial intelligence in hereditary neuromuscular disorders

Training the Next Generation through the CoMPaSS-NMD Young Investigator Training program and the CoMPaSS-NMD Autumn School Workshop Report

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Artificial Intelligence (AI) is the ability of machines to perform tasks that typically require human intelligence, such as learning, problem-solving, and decision-making. Its integration into healthcare may revolutionize many areas of medicine, including the diagnosis and management of neuromuscular disorders (NMDs).
These disorders, characterized by their clinical and genetical complexity and heterogeneity, demand innovative approaches to improve patient outcomes. Among these approaches, AI-driven solutions hold immense potential. However, the success of these solutions depends on preparing a new generation of clinicians equipped to harness the multifaceted power of AI.
One remarkable initiative addressing this need is the CoMPaSS-NMD project, which pioneers an interdisciplinary framework for developing AI-driven strategies to stratify patients using multiple clinical, histopathological, MRI e genetic datasets. By fostering a shared working language and integrating diverse competencies, the project aims to advance knowledge dissemination and bridge gaps between traditional disciplines. This approach is vital for addressing the challenges posed by NMDs, where early diagnosis and personalized treatment plans are critical.
To support this mission, the Young Investigator Training (YIT) initiative within CoMPaSS-NMD fosters education and scientific exchange among early-career researchers. By promoting high-quality clinical assessments and multidisciplinary training, YIT prepares a new generation to meet the evolving challenges in NMD care and research.

Key words: neuromuscular disorders, artificial intelligence, genetics, MRI, histology

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Artificial Intelligence (AI) is the ability of machines to perform tasks that typically require human intelligence, such as learning, problem-solving, and decision-making. Its integration into healthcare may revolutionize many areas of medicine, including the diagnosis and management of neuromuscular disorders (NMDs)[1].

These disorders, characterized by their clinical and genetical complexity and heterogeneity[2], demand innovative approaches to improve patient outcomes[3]. Among these approaches, AI-driven solutions hold immense potential. However, the success of these solutions depends on preparing a new generation of clinicians equipped to harness the multifaceted power of AI.

One remarkable initiative addressing this need is the CoMPaSS-NMD project[4], funded by

the European Union within the Horizon project (ClinicalTrials.gov ID NCT06734949), which pioneers an interdisciplinary framework for developing AI-driven strategies to stratify patients using multiple clinical, histopathological, MRI[5] and genetic datasets[6]. By fostering a shared working language and integrating diverse competencies, the project aims to advance knowledge dissemination and bridge gaps between traditional disciplines. This approach is vital for addressing the challenges posed by NMDs, where early diagnosis and personalized treatment plans are critical.

Central to this vision is the engagement and education of young scientists. The CoMPaSS-NMD project prioritizes equipping future clinicians with a technological perspective and the ability to collaborate across disciplines. This requires a skill set that extends beyond medicine and informatics to include data analysis, machine learning, and systems biology. Mastery of these competencies will enable clinicians to navigate the complexities of modern healthcare, where AI applications are becoming indispensable. As AI continues to evolve, its potential in addressing the challenges of neuromuscular disorders will only expand. By investing in education, fostering interdisciplinary collaboration, and promoting standardized tools, we can ensure that this powerful technology is utilized to its fullest, ultimately improving the lives of patients and advancing the field of medicine.

CoMPaSS-NMD autumn school: building the future of NMD research

A key component of the CoMPaSS-NMD initiative is the Young Investigator Training (YIT) program, designed to foster scientific exchange among emerging researchers. The program ensures high-quality clinical assessments by qualified clinicians while uniting young researchers in the field of NMDs.

By promoting a multimodal and multidisciplinary training framework – integrating clinical practice, bioinformatics, imaging, histology, genetics, data analysis, and knowledge dissemination – the YIT program prepares clinicians to adopt a Gestalt approach to neuromuscular disease (NMD) care, emphasizing that the whole is greater than the sum of its parts. A key aspect of this training is the adoption of standardized tools like the Human Phenotype Ontology (HPO), which provides a consistent vocabulary for describing phenotypic abnormalities linked to human diseases, including rare NMDs[7]. Widely used in diagnostics, research, and genomic studies, HPO enables precise and unified communication across disciplines, facilitating the integration of artificial intelligence into clinical practice. Furthermore, the program encourages participants to move beyond the traditional 'one gene, one disease' paradigm by exploring the broader genetic architecture and its contribution to phenotypic variability in NMDs, thereby encouraging a deeper understanding of genetic complexity in patient care.

The CoMPaSS-NMD Autumn School, held from September 29th to October 1st, 2024, in Tirrenia, Italy, was a landmark event for advancing this mission[8]. Bringing together young researchers from diverse cultural and academic backgrounds, the program exemplified international collaboration, multidisciplinary learning, and translational research. Of the twenty-seven participants, sixteen (59%) were females, twenty-three Italians (85%), three (11%) Polish, one (4%) French. Eight participants (30%) were neurology residents, three participants (11%) were medical genetics residents, two (7%) were child neurology residents, one of the participants (4%) was a neurology resident and PhD student. Two participants (7%) were child neurologists and PhD students, three (11%) were attending neurologists. Three participants (11%) were biotechnologists and PhD students in genetics, two participants (7%) were post-doc biologists, and three (11%) were PhD students in computer sciences. The immersive environment dismantled disciplinary silos, promoting a culture of knowledge transversality to improve patient care.

Day 1 agenda - 29 September

Time	Session	Speaker
13:00	Registration	
14:00	Welcome lunch	
14:45	Introduction to the key topics of the School	Filippo Santorelli (Fondazione Stella Maris)
15:00	COMPASS-NMD: what is important for the next phase and beyond	Rossella Tupler, University of Modena and Reggio Emilia, CoMPaSS-NMD coordinator
15:30	The complex genetics of inherited muscle diseases	Marco Savarese (Folkhalsan)
16:30	Modeling muscle diseases in zebrafish	Maria Marchese (Fondazione Stella Maris)
17:30	What their muscle have taught us: a long and exciting journey	Corrado Angelini (University of Padova)
18:30	Q&A Section - General discussion	All
19:30	End of the first day (*dinner on your own*)	

Day 2 agenda - 30 September

Time	Session	Speaker
9:30	Multiple Omic studies and how to apply to NMD	Jocelyn Laporte (IGBMC institute)
10:30	Artificial intelligence systems to support the diagnosis and monitoring of NMD diseases	Joanna Polanska (Silesian University of Technology)
11:30	Muscle biopsy in 2024: What? Why? The experience of LGMD	Lucia Ruggiero (Federico II University Hospital)
12:30	Discussion, what you have learned, Clinical challenge & Goals of Coffee House (coffee served during this slot)	Filippo Santorelli (Fondazione Stella Maris)
13:00	Lunch	
14:00	Molecular logic of cerebral cortex development, evolution, and disease	Marco Onorati (University of Pisa)
15:00	The skeletal muscle in mouse models of neuromuscular diseases	Elisabetta Ferraro (San Filippo Neri Hospital, Rome)
16:00 - 18:00	**Coffee House** - Project idea developments (four classes on four topics) (coffee station 17:00-18:00)	
18:00	**Case quiz**: preparing 3-4 case studies in small groups	
18:30	End of the day 2	
19:30	Getting together and dinner	

Day 3 agenda - 1 October

Time	Session	Speaker
9:00	What we learned from InGene2.0	Raffaele Conte (National Research Center)
10:30	Sharing results of Coffee House	
11:30	**Coffee break**	
11:45	Case quiz presentations	
12:30	Wrap up of the meeting- The next training in COMPASS NMD	Rossella Tupler (University of Modena and Reggio Emilia), project coordinator Filippo Santorelli (Fondazione Stella Maris)
13:00	**Farewell light lunch**	

Figure 1. Overall program of the first CoMPaSS-NMD Autumn School.

The program seamlessly blended cutting-edge science with real-world applications, highlighting the synergy between genetics, molecular biology, bioinformatics, and clinical neurology (Fig. 1). Sessions delved into intricate topics such as the complex genetics of inherited muscle diseases emphasizing the importance of detailed phenotypic characterization and a structured functional analysis of genetic mutations for accurate interpretation of Next-Generation Sequencing (NGS) results. At the same time, the discussion underscored the ongoing need for robust preclinical models to investigate pathophysiological mechanisms and develop potential therapeutic strategies, highlighting their pivotal role in translating laboratory discoveries into improved patient care. Equally compelling were discussions on the long and exciting journey of muscle biopsy, tracing its past, present, and future, and underscoring how foundational research continues to shape modern medical practice. The workshop focused on Limb-Girdle Muscular Dystrophies (LGMDs), a heterogeneous group of disorders characterized by progressive muscle weakness and atrophy, predominantly affecting the shoulder and pelvic girdles. While NGS has transformed the diagnostic landscape of LGMDs, muscle biopsy remains an essential complementary tool for resolving uncertain genetic findings, confirming protein deficiencies, and differentiating LGMDs from other myopathies. Furthermore, it provides critical insights into disease mechanisms, biomarker discovery, and therapy evaluation, particularly in cases with variants of uncertain significance (VUS) or undetected mutations. This transdisciplinary approach struck a powerful chord with participants, motivating them to embrace holistic perspectives in their work. Adding further depth, the program explored the rapid evolution of artificial intelligence since the mid-20th century, illustrating how this transformative technology is poised to increasingly integrate into both our professional and personal lives. AI is increasingly being integrated into the diagnosis and management of HNMDs[9]. AI-driven tools leverage the concept that medical images are not merely visual representations but quantifiable data sources from which valuable metrics – such as muscle volume, fat fraction, fat distribution, and elevated short tau inversion recovery (STIR) signal – can be extracted. AI algorithms are increasingly being utilized to analyse medical images with greater speed and accuracy than traditional methods, as well as to assess genetic sequences, identify mutations, and predict their pathogenic potential, thus paving the way for more precise and personalized approaches in NMDs management.

Breaking barriers: a model for collaborative learning

One of the most remarkable aspects of the Autumn School was its commitment to breaking down barriers between disciplines. By bringing together participants from various countries and academic fields, the program created a vibrant environment where ideas flowed freely. This diversity enriched discussions and highlighted the interconnectedness of scientific fields. The design of the program blurred the lines between basic science, translational research, and clinical practice, providing a comprehensive understanding of NMDs and their management.

The "Coffee House" session stood out as a hub of creativity and collaboration, designed to spark innovative thinking and teamwork. Participants were grouped to tackle specific topics, including NMD clinical aspects, genetics, preclinical models, imaging, and computer sciences. Through lively discussions and collective brainstorming, these teams worked to develop fresh ideas and propose forward-looking solutions aimed at advancing the future of CoMPaSS-NMD (Tab. I).

Table I. Learning from Different Perspectives: Insights Gained During the Autumn School.

Clinicians	The importance of establishing a diagnosis based on the clinical phenotype, moving beyond the *one gene, one disease* paradigm. Additionally, the need for accumulating and standardizing data (genetics, MRI, histopathology) to improve diagnostic accuracy.
Medical Geneticists	Gaining insights from clinical practice on the importance of standardized phenotype evaluation to complement genetic data and improve diagnostic precision
Biotechnologists	Learning that the clinical phenotype and its heterogeneity in humans are complex and not fully explained by preclinical models. Understanding the systemic nature of muscle disorders in human beings.
Computational scientists	Understanding what constitutes a clinical disorder and the significance of the investigations behind it. Developing algorithms to enable faster and more efficient diagnoses, bridging science and clinical care.
All participants	The whole is greater than the sum of its parts: achieving accurate diagnoses by unifying efforts, fostering collaboration across disciplines, and harnessing the potential of artificial intelligence.

This dynamic activity not only encouraged participants to combine their diverse expertise but also exemplified the program's commitment to fostering interdisciplinary dialogue and problem-solving.

In addition to structured activities, informal interactions during the Autumn School played a pivotal role in strengthening bonds among participants. These exchanges often led to spontaneous collaborations and discussions, enabling young researchers to share insights



Figure 2. Photo of the participants to the first CoMPaSS-NMD Autumn School.

and approaches from their respective fields. Such interactions are instrumental in creating a cohesive community of investigators dedicated to advancing NMD research (Fig. 2).

Despite the success of the CoMPaSS-NMD Autumn School, some limitations highlight areas for future improvement. One key challenge is the relatively small number of participants, which, while fostering an intimate and engaging learning environment, also restricts the broader impact of the initiative. To maximize the reach and influence of this interdisciplinary approach, a more extensive and widespread dissemination of the project among young researchers is essential, particularly among those outside the CoMPaSS-NMD Consortium. Expanding participation through additional training sessions, online modules, and stronger outreach efforts could ensure that more early-career clinicians and scientists benefit from the program's insights, ultimately accelerating progress in the field of HNMDs.

Empowering young investigators: the road ahead

The CoMPaSS-NMD Autumn School was more than a training program; it was a call to action for the next generation of neuromuscular researchers. The enthusiasm and engagement of the participants reflected their dedication to advancing the field (Fig. 3). By providing

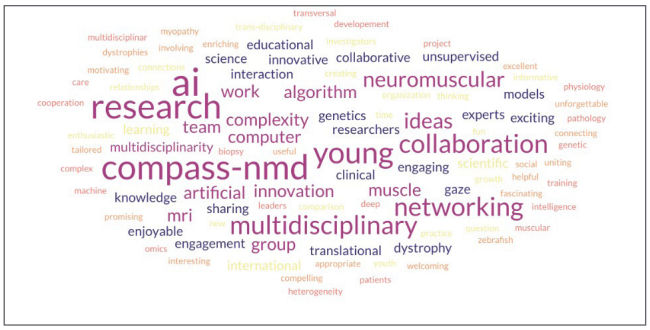


Figure 3. Word cloud with the most reported words from the CoMPaSS-NMD Autumn School participants.

opportunities for collaboration, mentorship, and skill development, the CoMPaSS-NMD project is accelerating progress in understanding and treating hereditary NMDs.

Looking forward, the success of this initiative serves as a model for future endeavors. The friendships formed, knowledge gained, and ideas exchanged during the Autumn School will undoubtedly ripple across the global NMD research community, paving the way for breakthroughs that transcend borders and disciplines. The YIT series is offering a program of webinars scheduled between July 2024 and April 2026. In 2025, the YIT series will continue with a spring webinar focused on utilizing the electronic Genetic CRF. Summer will bring another installment, this time delving into the electronic Histological Case Report Form. To round out the year, there will be a webinar on the electronic Magnetic Resonance Imaging CRF. The series concludes in Spring 2026 with the grand finale – an exclusive webinar unveiling the CoMPaSS-NMD ATLAS. Participants will gain hands-on knowledge to navigate the ATLAS and harness its transformative power to enhance their research.

The vision for a collaborative future

The broader implications of the CoMPaSS-NMD initiative extend beyond the field of neuromuscular disorders. The program's emphasis on interdisciplinary collaboration and innovation serves as a blueprint for tackling other complex medical challenges. By uniting experts from diverse fields, fostering a culture of open communication, and leveraging cutting-edge technologies, the initiative demonstrates how collective efforts can lead to meaningful advancements in healthcare. Moreover, the CoMPaSS-NMD Autumn School underscores the importance of supporting young investigators in their journeys. Empowering the next generation of scientists and clinicians is not only an investment in the future of research but also a commitment to improving patient outcomes worldwide. As participants carry forward the lessons learned and the connections made during the Autumn School, they become ambassadors for a more integrated and innovative approach to medical science.

A brighter future for NMD research

The CoMPaSS-NMD Autumn School exemplifies the power of collaboration and innovation in addressing complex medical challenges. By uniting young researchers from diverse backgrounds and breaking down disciplinary barriers, the initiative has created fertile ground for scientific discovery and improved patient care.

As we reflect on the success of this event, we are reminded of the words of Sir Isaac Newton: "If I have seen further, it is by standing on the shoulders of giants." The Autumn School has provided young investigators with those very shoulders – a foundation upon which they can build a brighter future for patients affected by hereditary neuromuscular disorders. The ongoing efforts of initiatives like CoMPaSS-NMD inspire hope for a future where collaboration between basic science, clinical practice and technology converge to overcome the challenges of rare diseases, ensuring that no patient is left behind.

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Conflict of interest statement

The authors declare no conflict of interest.

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Authors contribution

Conceptualization AN, MS, FMS, RT
Writing-original draft: AN Writing-review and editing: RT, MS, FMS.

Ethical consideration

No personally identifiable information is disclosed without explicit permission. Group photos are used in accordance with event policies. The content of lectures, workshops, and presentations is attributed to the original speakers.

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