IMPACT SUCCESS STORIES

HNN 3.0
HEALTH-NCP-NET

CoMPaSS-NMD overall purpose is to create a new generation of methods for "precision diagnosis" that allow researchers and/or healthcare professionals to successfully classify patients affected by Hereditary Neuromuscular Diseases.

Through the application of methods based on the use of Artificial Intelligence (AI) and Machine Learning (ML), the study aims to provide new perspectives in the knowledge of the pathogenetic and transmission mechanisms of HNMDs, leading to a more accurate diagnosis, thus improving the prognosis, which is often difficult to predict in the clinical practice.

CoMPaSS-NMD

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1. Describe your project through key words/key phrases that identify it.

CoMPaSS-NMD is a project dedicated to transforming how hereditary neuromuscular diseases (HNMDs) are diagnosed, interpreted and understood using AI. The project brings together experts in genomics, clinical medicine, data science, and ethics to create a unified, AI-enhanced pathway that shortens the "diagnostic odyssey" faced by many rare disease patients. By combining advanced machine learning with harmonised clinical and genetic data, the project aims to deliver earlier and more accurate data disease interpretation across Europe.
CoMPaSS-NMD is a collaborative effort to modernise precision medicine, improve healthcare efficiency, and ensure that patients can access cutting-edge tools. Through its work, the project hopes to build a model that can be adopted widely, helping health services deliver faster answers and better care to individuals living with rare neuromuscular disorders.

2. In terms of impact, what will be the most tangible your project will achieve?

By integrating genomics, structured clinical data, and AI-driven decision support, the project will produce novel and universal tools for the diagnostic stratification of patients suffering from HNMDs aiming at precise personalised treatments to overcome high-impacting dependency and early death. This is a clinically validated pathway that can be directly adopted by hospitals and national health systems. CoMPaSS-NMD will provide tools that clinicians can use immediately, including SOPs (standard operation procedures), machine-learning models, able to support differential diagnosis, harmonised data standards, and digital workflows to reduce waiting times in patients. The project, by standardising data and enabling AI support, will provide patients with the same diagnostic quality all around Europe. Finally, CoMPaSS-NMD will generate significant health-system savings by reducing misdiagnosis, ineffective treatments, and avoidable procedures. The cumulative effect is a measurable improvement in precision medicine in Europe, patient treatment and efficiency for clinicians.

3. Please describe your project's overall impact, if applicable, at the European level.

CoMPaSS-NMD is setting a new standard for how HNMDs are diagnosed through data interpretation, managed, and understood. By creating a unified, AI-enhanced pathway, the project directly supports the EU's ambition to reduce health inequalities, strengthen precision medicine, and accelerate the digital transformation of healthcare systems.
CoMPaSS-NMD is creating a framework of harmonised data and clinical practices: CoMPaSS-NMD, together with its partners, align phenotypic, genetic, and clinical datasets, creating interoperable resources that can be shared, reused, and scaled. This directly contributes to the European Health Data Space by demonstrating how secure, high-quality, cross-border health data can be used to improve diagnosis and patient care.
Through its validated AI-driven tools, the project will enable more equitable access to better care, ensuring that patients can benefit from a high level

of accuracy in investigating the disease across Europe.
Moreover, CoMPaSS-NMD strengthens Europe's leadership in ethical and trustworthy AI in healthcare, providing a concrete example of how AI can be integrated into clinical workflows safely, transparently, and in compliance with EU regulations such as the AI Act and the GDPR.
Finally, the project will reduce the long-term cost in health associated with diagnostic delays and misdiagnoses, supporting more sustainable European health systems. By improving early detection and reducing unnecessary procedures, CoMPaSS-NMD advances the EU's goals of efficiency, resilience, and patient-centred care.

4. As an applicant, what advice would you have wanted in the Horizon project design process? What support did you receive from National Contact point (NCP) and your organisation, and what improvement of support would you benefit from?

Horizon proposals are not simply technical documents, they are strategic narratives that require alignment across scientific, clinical, societal, and policy dimensions. UNIMORE, as the lead partner, after a project codesign with all partners involved, early contacted the National Contact Point (NCP) to receive instruction on the fulfillment of the application requirements better shaping the project idea accordingly to the application form. The NCP provided useful clarifications on eligibility, call requirements, and strategic alignment, and offered targeted feedback on the structure of the proposal. Their input was particularly helpful ensuring that the project responded clearly to the expected outcomes and impacts.

5. Please highlight aspects of your Horizon project's strengths that you consider important and that may constitute good practice for other applicants.

One of the strongest aspects of our Horizon project was the early and sustained codesign among all partners, including clinicians, data scientists and industry, focussing on building a balanced and complementary consortium. We did not wait until the final drafting stage; instead, we built the concept collaboratively from the beginning. This created a proposal that was coherent, well-integrated, and clearly aligned with real clinical and patients' needs.
Moreover, the project's scientific ambition is concrete, featuring measurable impacts based on clearly defined activities and outcomes.
CoMPaSS-NMD invested in robust data management and ethics compliance planning throughout both its co-creation and implementation phases. This planning specifically focused on advancing science through AI while simultaneously protecting patients and their data.

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