objective evaluation of human movement thanks to their portability, user-friendliness, and continuous monitoring. The aim of this preliminary study is to assess the usability of IMU sensors to quantify gait parameters in DMD patients during the 6MWT. Five ambulant DMD patients (age 7-13 years, height 120-130.5 cm, weight 24-46 kg) were enrolled in the experiment. 5 IMUs (APDM OPAL, 200 Hz sample frequency) were positioned on the pelvis, shanks and ankles. Gait events were identified from 3D accelerations and angular velocities. Time and frequency analyses were proposed to describe the gait in terms of spatio-temporal parameters (i.e. average speed, stance and swing duration), smoothness and symmetry (i.e. harmonic ratio, power spectral analysis). Results highlighted a reduction of gait symmetry and smoothness in comparison with physiological data available in the scientific literature. Moreover, in accordance with the clinical evaluation, the pelvis kinematics might be influenced by the upper body rotational compensations. The procedure was well tolerated by patients. Future research will concentrate on a larger population and a longitudinal monitoring to identify significant outcomes for the pathology progression and the correlation with clinical assessment.

InGene 2.0: How a technological platform supports the clinicians in the diagnosis and treatment of neuromuscular disorders

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Neuromuscular disorders (NMDs) are rare conditions if taken singularly, but as a whole they represent a huge problem for the community and the healthcare providers. One of the main challenges for the clinicians is represented by their heterogeneity, making their diagnosis and proper treatment particularly tricky. To this extent, recent technological advances in terms of genetic facilities (e.g., the introduction of NGS approaches), as well as in more efficient ways to perform specific examinations (i.e., neurological examination, functional assessment, etc.) and to collect and analyze data generated have been shedding a light on the genotype-phenotype characterization of a specific patient or group of individuals, in turn contributing to increasing the diagnostic accuracy and making the planned treatments tailored, therefore more efficient and effective.

With this in mind, the InGene 2.0 consortium, composed of Italian players within the research, academia and clinical universe, and coordinated by the CNR-IFC in Pisa, has developed a suite of technological tools whose core is represented by Health360, a modular, user-friendly, GDPR-compliant, small electronic health record capable of storing and displaying data coming from different sources and making them available at a glance for the clinician, to follow the clinical trajectories of the patient, and for the data scientist for a proper, yet offline, investigation. Actually including modules for personal data, anamnesis, muscular MR imaging, physiotherapy tests, neuromuscular examination, genetics, the tool will soon include facilities to allow agile data analysis and to finally attempting to unravel the complex relationships between genotype and phenotype in NMDs.

Usability of a mobile health application to support home-based aerobic exercise in neuromuscular diseases

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Barriers to home-based aerobic exercise in people with neuromuscular diseases (NMD) include reduced possibilities for monitoring by supervising physiotherapists and difficulties to train within target intensity zones. To overcome these barriers we developed the 'Keep on training with ReVi' app (ReVi-app). This study evaluated the usability of the ReVi-app.

Patients followed a 4-month polarized home-based aerobic exercise program, with 3 weekly sessions, on a cycle or rowing ergometer, supported by the ReVi-app. The app registered training data, including heart rate and ratings of perceived exertion, provided real-time feedback on reaching target intensity zones and enabled monitoring via an online dashboard. Patients and physiotherapists evaluated usability using self-developed questionnaires, covering different usability components; efficiency, effectiveness and satisfaction.

Amyotrophic Lateral Sclerosis Holistic Outpatient Exercise (ALS HOPE) Group

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Amyotrophic lateral sclerosis (ALS) is a progressive neurological condition affecting 30, 000 United States (US) Americans at any given time, 5,000 of which are US military Veterans. Veterans are twice as likely to be affected by the disease. Even though they are served by the largest healthcare system in the nation, access to specialized ALS clinics is limited.
The natural progression of the ALS disease presents unique challenges to a physical therapist (PT) treating persons living with ALS (pALS). Physical therapists strive to stay ahead of the patient's physical decline with therapeutic measures and issuance of durable medical equipment to assure safety and promote independence. Over the last decade, there has been increased interest in the role of exercise in slowing the functional decline in the ALS population. Veterans living with ALS have limited access to specialized physical therapists who can guide them through an appropriate and safe exercise program that promotes function and minimizes fatigue, pain, and other indications of over use. Traveling to specialized clinics is a significant burden for an individual living with disabilities and can lead to extended periods of fatigue. The Puget Sound Healthcare System, Seattle VA, has created an innovative way to provide a safe means for pALSs to exercise under the supervision of a specialized ALS physical therapist and physical therapy assistant without having to leave the comfort of their home. In this platform presentation, we will review the literature on ALS and exercise and provide details pertaining to this virtual exercise group.

Supporting Physical Exercise in patients with Neuromuscular Diseases: AI-assisted training

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In the context of Neuromuscular Diseases (NMDs), physiotherapy plays a crucial role in preserving patients' mobility and mitigating the adverse impacts of immobility and contractures, as seen in conditions such as Duchenne Muscular Dystrophy (DMD), Becker Muscular Dystrophy (BMD), and Charcot-Marie-Tooth Disease (CMT). For instance, in DMD, a regimen of regular stretching and, in select cases, low-impact exercises such as swimming or cycling is recommended to slow disease progression. A collaborative team of healthcare professionals including physical medicine and rehabilitation specialists, neuromuscular nurses, nurse practitioners, physical therapists, and social workers actively support individuals at various stages of their NMD journey. Recognizing the potential of digital technologies to alleviate the time burden on both patients and healthcare providers, these innovations can facilitate remote patient monitoring and enhance the frequency of care, particularly for those facing challenges with travel logistics.
Within this context, we introduce an experimental AI system tailored to assist patients and their caregivers in performing physical exercises effectively. This AI system, powered by a live video feed capturing the patient's exercise routine, can assess in real-time the accuracy of the patient's movement and offer guidance and corrections when errors are detected. Furthermore, the system can track and log eight key parameters (i.e., angles between the torso and limbs), which can aid in monitoring the progressive decline in muscle function over time. Developed at the School of Informatics at the University of Bari, this experimental system prioritizes user-friendliness and addresses primary concerns related to acceptability. Additionally, the system is GDPR-compliant and is poised for pre-clinical evaluation, marking a promising step towards more effective and accessible NMD care management.

An ICT suite of tools for for deep phenotyping and genotype correlation in neuromuscular diseases: the Phenotype module

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A detailed and standardized neurological evaluation in neuromuscular diseases (NMDs) that are often characterized by a wide and complex clinical picture, is recommended for an accurate recording of phenotypes, interpretation of genetic analysis and definition of natural history. In this scenario, the availability of ICT-based tools collecting data from various sources (clinical, instrumental, etc.), with the ultimate aim to

implement and train Artificial Intelligence (AI) - based algorithms helping the clinicians with a better characterization of patients is of utmost importance. In this regard, the possibility to perform a "morpho-functional digitization" of NMD phenotypes, perfectly characterizing the patient's clinical condition, overall, becomes pivotal for both the clinician and the data scientist. To this end, we developed a module specifically dedicated to the phenotype, to be integrated within a comprehensive, cloud-based suite, the Health360 platform. The module allows clinician to enter data related to neuromuscular examinations by selecting the single muscular district within a graphical user interface displaying the patient's silhouette and associated scores related to the functionality of the district (including the MRC Score), but also allows including the codes from the Human Phenotype Ontology (HPO), to reduce subjectivity of data inclusion and, later on, analysis and interpretation. Taken together with the other modules, this part could effectively support clinicians in a better clinical characterization of individuals with NMDs, at the same time providing the data scientist with a precious, usable tool for genotype-phenotype correlation in such clinical conditions.

Artificial-Intelligence fosters the muscular MRI interpretation by the clinician: a proof-of-concept study

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The present study aims at developing convolutional neural networks (CNNs) for automated segmentation of muscles in the thighs. Eighteen patients were retrospectively enrolled: 8 patients with neuromuscular alterations on MRI and 10 with unremarkable MRI. All patients performed MRI including dual echo, STIR sequences and DWI with b value set to 0 and 800.

A radiologist manually segmented thighs muscles with a different mask for each of the three muscle compartments (anterior, posterior and medial) on out-of-phase sequence. Manual segmentations were employed as groundtruth to train and validate two CNNs with different automatic segmentation UNet architectures, one for the thigh muscles together (UNet1) and one for the three muscular compartments, separately (UNet3). For each patient, the 2D slice at the middle third of the thighs was selected. Then, images of left and right thighs were divided into two separated datasets to double the sample size. Particularly, 15 thighs were used for training, 10 for validation, 11 for testing. The CNN performances were measured in terms of dice similarity coefficient (DSC).

Net1 and UNet3 achieved an accuracy of 96% and 95%, respectively. UNet1 showed a DSC of 0.94, while UNet3 showed a mean DSC of 0.92, 0.80, and 0.90 for the segmentation of the anterior, medial, and lateral compartment, respectively.

Overall, UNet1 and UNet3 achieved an excellent performance in thighs muscles segmentation. These results paved the way for further, potentially effective AI-based models for quantification of neuromuscular alterations or radiomics features extraction, leading to a more practical, less time-consuming workflow.

Application of digital tools in a management model for transition phase from paediatric to adult care in dystrophinopathies

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The transition phase from the pediatric care specialist center to the adult neurologist represents a complex and sensitive process involving patients in their late-childhood and adolescence age, caregivers, and health professionals. In this framework we developed the project of a dedicated program for case management of "transition" patients with Duchenne and Becker Muscular Dystrophy (DMD/BMD) in Tuscany region, involving the Neurology Unit from University of Pisa, responsible for adult patients, and the Metabolic and Hereditary Diseases Unit of Meyer Hospital and Neurology and Rare Diseases Unit of IRCCS Stella Maris for pediatric patients. We plan to include in a period of three years an estimated number of 25 DMD/BMD patients. Various health care professionals will be identified and included in the dedicated multidisciplinary team. Patients will be evaluated by the adult neurologist and the childhood neurologist in the pediatric Center on bimonthly-scheduled visits, and then the following exami-

nations will happen in the adult Center in Pisa. Starting from the present collaboration between the Neurology Unit and the CNR Center from Pisa on the development and use of the digital application Health360 in phenotyping neuromuscular patients, the whole process will take advantage of a dedicated common platform to collect and share relevant clinical data between the multidisciplinary team members, in order to evaluate the role of IT in supporting data collection and management in this delicate clinical moment. We believe that recording of diagnostic and functional parameters during this peculiar phase of our patients' life will provide a better knowledge on how to optimize global management but also on disease natural history.

Digital biomarkers

Wearable sensors to measure indirect fatigue in Myotonic Dystrophy (DM1A).

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Wearable sensors are becoming increasingly popular for complementing classical clinical assessments of gait deficits. They have been used to inform on additional aspects of gait, especially walking-related fatigue. Monitoring gait during the 6MWT offers a unique opportunity to investigate the dynamic changes that occur in prolonged walking. We try to determine which gait parameters worsen during sustained walking and to examine the clinical correlates of gait fatigability in patients with myotonic dystrophy.

Eleven patients with myotonic dystrophy were enrolled and recruitment is ongoing. Data were collected during a standard 6MWT wearing a sensor (GSensor, BTS) around the waist at L5 level. The sensor included a 3D accelerometer and a 3D gyroscope. Gait parameters were extracted from raw signals using validated algorithms, corresponding to early, middle and late segments of the 6MWT. Non-parametric test were performed to compare gait parameters between segments.

Preliminary results show an increase in gait acceleration and significant changes in smoothness and stability of gait in the DM1A group between the early and the late section of the 6MWT.

Our preliminary results suggest that gait parameters associated to fatigability can be measured during a standard 6MWT using a wearable device and that the method allows to highlight variations during sections of the 6MWT in patients with Myotonic Dystrophy. These findings can be used, in combination with patient-reported gait fatigue, as markers for fatigability in DM1A patients.

Wearable-devices assisted clinical protocol for assessment of fatiguability in SMA

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Spinal Muscular Atrophy (SMA) is an inherited neuromuscular disease affecting the lower motor neuron. With the new therapeutic options, sensitive clinical evaluation and outcome measures are needed, especially in adult patients, in which the expected clinical response is mostly stabilization of disease progression. With treatment, patients often subjectively report amelioration of fatigability in daily life activities. Based on this, we developed a dedicated evaluation scale, ENDUSMA, as a tool specifically designed for adult SMA patients by assessing the muscle endurance dimension for the upper and lower limbs. To enhance the sensitivity of the scale to capture changes in patients' endurance during motor tasks, a wearable devices protocol (AUTOMA) composed of one biaxial electro-goniometer and a surface electromyographer has been applied during the administration of the motor scales protocol. The system has been tested on two SMA patients so far. Electro-goniometer signals are acquired at 1000Hz by a 4-channels Bluetooth electronic module connected to a laptop; for the shoulder, EMG electrodes are placed on the deltoid and are acquired by another Bluetooth module connected to the same laptop. EMG signals are simultaneously processed with the same software that can synchronize events.

Endurance clinical evaluation supported by the AUTOMA acquisition system can integrate the standard clinical monitoring in the follow-up and in evaluating response to treatment in SMA adult patients.

Device Development

A novel technology-based tool for performing motor assessment in neuromuscular disorders

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Motor assessment is pivotal in evaluating the functional abilities of individuals with neuromuscular disorders (NMDs). Tests are usually conducted at clinics, and tests like the Six-Minute Walk Test (6MWT), the North Start Ambulatory Assessment (NSAA), the Performance of the Upper Limb (PUL) are used to monitor patients' functional conditions both at baseline and follow-up. Moreover, these tests are used in many clinical trials to understand the efficacy of new drugs. However, the lack of technological tools to help performing such examinations makes the work of the clinicians burdensome, mainly relying on pen-and-paper, with considerable use of dedicated staff and poor efficiency in retrieving old data from the archives for comparison of clinical trajectories. Within the InGene 2.0 project, aiming at building a technology-based suite of tools to support the clinicians and data scientists in NMDs, some solutions specifically devoted to motor assessment were developed. Health360, the software implemented for collecting multimodal data in NMDs is linked to various modules, including the PhysioTest, in turn allowing the clinicians to perform motor assessments through a dedicated interface operating on PC/MAC and tablets. The tool enables performing tests and storing data related to 6MWT, NSAA, PUL and others; feedbacks from clinicians highlighted significant improvement in timesaving and efficiency of their job thanks to the solution. More recently, an App was developped for both Android and iOS to allow performing the tests directly by the smartphone, with further significant time and resources reduction, not sacrificing the accuracy and usefulness of the assessment.