Research Assistant:
Francesca Fadda

Research topic:
Management and monitoring of Pompe disease through the use of new wearable technologies

Abstract:
Pompe disease is a rare and progressive neuromuscular disorder that compromises both muscular and respiratory function, requiring continuous and multidisciplinary monitoring. The aim of this project is to develop an integrated model for the clinical management and monitoring of Pompe disease through the use of wearable technologies (G-Sensor and actigraphs), in combination with standard clinical assessments. The G-SENSOR, an inertial sensor worn on the lower back, will be used to quantitatively evaluate gait, static and dynamic balance, and performance in functional tests such as the Timed Up and Go (TUG). This device enables the segmentation and kinematic analysis of different movement phases, increasing sensitivity in detecting subtle motor changes that are often clinically imperceptible. The G-Sensor will be employed at baseline and during follow-up evaluations every six months. The ActiGraph devices will be used for continuous recording, over a period of seven consecutive days, of motor activity, rest, and sleep-related parameters. These wrist-worn devices allow for the ecological monitoring of sleep-wake cycles, as well as the quantity and quality of movement in real-life settings, including the influence of environmental factors such as light, noise, and temperature. The protocol includes the integration of these data with standard clinical assessment tools, including the 6-Minute Walk Test (6MWT), Walton and Gardner-Medwin (WGM) score, Gait Stairs Gower Chair (GSGC) score, Medical Research Council (MRC) scale, Forced Vital Capacity (FVC), and patient-reported outcome measures. The expected outcomes include greater sensitivity in detecting functional changes over time and objective support for clinical decision-making. This project aims to provide an innovative contribution to precision medicine in the field of rare diseases by promoting personalized, continuous, and remote monitoring, thereby improving both care quality and therapeutic planning.