higher-level developmental skills that included walking, jumping, and running. GRO provides a standardized and consistent measure of motor function especially for clinical trials inclusive of participants 5 years of age or younger, as this group has become the predominant target for developing new therapies.

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External responsiveness of the Duchenne video assessment, a novel fit-for-purpose remotely collected outcome measure for Duchenne muscular dystrophy

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Determining treatment benefit in Duchenne Muscular Dystrophy (DMD) is challenging due to factors such as disease heterogeneity and lack of robust outcome measures. The Duchenne Video Assessment (DVA), accepted into FDA's Clinical Outcome Assessment Qualification Program, addresses these challenges and aligns with the decentralized clinical trial paradigm. The measure's performance was explored in a longitudinal study. The Duchenne Video Project (DVP) is an observational study of male participants with DMD and healthy controls. DMD study cohorts included early/late ambulatory and early/late non-ambulatory. The early ambulatory cohort included two age groups: 4-6 and 7+. The DVA was collected at baseline and 30 weeks (baseline and 12 weeks for ages 4-6), and Clinician Global Impression of Change (CGIC) was scored as a comparison between the two timepoints. Physical therapists scored the DVA's 17 tasks evaluating ease of movement, and a DMD clinician provided CGIC rating by task. Within-task DVA severity percentages were calculated and several composite algorithms were considered. Aggregated CGIC scores were calculated using median across tasks. CGIC and DVA change score association was evaluated using the Jonckheere-Terpstra test. Sixty-three participants were enrolled; 53 with DMD and 10 healthy controls. Across the study population, declines over time in DVA Total Composite Task Average and Compensation Sum scores were significantly associated with aggregate CGIC worsening (DVA change score median -0.5 (CGIC=No Change), 3.0 (Minimally worse), 8.0 (Much Worse), p=0.0018; 0.0 (No Change), 1.0 (Minimally worse), 3.0 (Much Worse), p=0.0013, respectively). Similar trends were observed within study cohorts. External responsiveness was exhibited via significant associations between DVA change over time and CGIC, further demonstrating the validity of the DVA and highlighting its utility for DMD clinical trials.

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Longitudinal multi-centric study to assess the digital outcomes issued from wearable magneto-inertial devices in ambulant DMD

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Duchenne muscular dystrophy is a muscle disease characterized by severe and rapidly progressive muscle weakness. One of the key challenges in treating this condition is identifying objective, reliable, and sensitive outcome measures to measure the effects of drug. For this purpose, the ActiMyo® (Sysnav, France), a magneto-inertial wearable device, was developed. Among the assessments with performed by this wearable device, the 95th centile stride velocity (SV95C) was qualified as the 2nd endpoint in 2019 by the European Medical Agency, representing the most rapid 5% of strides during real-life activities. The ActiLiège study is a multicentre clinical study with the aim of gathering data issued from a magneto-inertial wearable. Ambulant patients with DMD and healthy controls were included and will be followed up for three years. The patients wore the ActiMyo® on their ankle and wrist during the first 3 months after the inclusion and afterwards for one month every 3 months. Controls wear the device for a period of one month every 12 months. Digital outputs from the wearable sensors are compared to gold standard assessments. We enrolled seventy-six ambulant DMD patients aged between 4 and 20 years in 7 centres (Belgium, Poland, Hungary, Romania, Czech Republic, Slovenia, and Egypt). Thirty-five of them completed at least 1 year of follow-up. All ambulant patients were either on a 6-month stable course of steroids or started steroids at baseline. The baseline data of the patients and controls will be presented, along with longitudinal data that may provide an indication of sensitivity to change in comparison with other commonly performed outcomes.

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The international clinical outcome study for dysferlinopathy - ten years of natural history data

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Individuals with dysferlinopathies have highly variable clinical presentation presenting significant challenges for trial readiness. The International Clinical Outcome Study for Dysferlinopathy (COS) was the first multi-country natural history study in dysferlinopathy. 193 participants from 15 sites in eight countries for three years, with some sites collecting data for up to six years (NCT01676077). In the absence of any disease specific OMs, participants completed a variety of standardised assessments of strength and function, PROM, qualitative and quantitative MRI and biomarker studies. The North Star Assessment for limb girdle type muscular dystrophies (NSAD), was developed, validated and quantified dysferlinopathy presentation and progression. The extension study, COSII, recruited 203 participants (119 new) from 16 sites in nine countries. COSII participants were aged 14-78, (mean 40), 70% ambulant and 56% were female. Across COS1 and COS2 cohorts, mean follow up time was 43 months (0-117 months). 20 participants have now been followed for 10 years. We analysed annual progression for all participants over a 10-year period using the NSAD, by gender and ambulation status. Mean NSAD total score at first visit was 26.4 (±15.8), at last visit 18.5 (±15.6). There was statistically significant decrease in mean NSAD total score per year of age (95% CI -1.489 to -1.296, p <.001). Qualitative MRI defined a characteristic pattern of muscle involvement. Quantitative MRI methods including MRS and fat fraction captured change over three years. Utilising NSAD and MRI, COS confirmed Miyoshi and LGMD2B/R2 are not two distinct phenotypes, a critical finding for clinical management, clinical trial population definition and access to disease modifying treatments. COS/COSII, conducted with patient advocacy partners, the Jain Foundation, has developed and determined robust OMs, defined phenotype and progression, all critical components for clinical trial readiness. This study is supported by the Jain Foundation and is submitted on behalf of the Jain COS Consortium.

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Intra and inter-rater reliability of the MFM32 in myotonic dystrophy type 1

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Myotonic dystrophy type 1 (DM1) is an autosomal dominant neuromuscular disease. The disease is slowly progressive, and motor signs include muscle weakness and myotonia. Early motor signs are handgrip myotonia and distal weakness, and weakness of neck and abdominal flexors. Monitoring of motor function is important to track disease progression and to assess effects of prescribed exercise or medical treatment. The 32-item Motor Function Measure (MFM32) is a tool to assess motor function, developed for patients with neuromuscular disorders. MFM32 is validated in people between 6 and 60, and a range of different neuromuscular disorders including DM1. MFM32 consists of 32 items, scored on a scale from 0: Cannot perform the task to 3: Performs the task fully and normally. The items are classified in 3 domains: D1; Standing and transfers. D2; Axial and proximal motor function and D3; Distal motor function. Item scores in each domain are added up. The sum is divided by the maximum score of the domain and multiplied by 100 to give a percentage score. The sum of the 3 domains divided by 96 and multiplied by 100 constitutes the total percentage score. In this study, we use data from a large