Up to one-third of patients with myotonic dystrophy type 1 (DM1) die suddenly. Thus far, no intervention has effectively prevented sudden death. The objective was to determine whether an invasive strategy (IS) based on systematic electrophysiological studies and prophylactic pacing is associated with longer survival in patients presenting with DM1 and major infranodal conduction delays than a noninvasive strategy (NIS). A retrospective study, the DM1 Heart Registry, included 914 consecutive patients older than 18 years with genetically confirmed DM1 who were admitted to the Myology Institute of Pitié-Salpêtrière Hospital, between January 2000 and December 2009. Among 486 patients whose electrocardiogram showed a PR interval greater than 200 ms, a QRS duration greater than 100 ms, or both, we compared the outcome of 341 (70.2%) who underwent an IS with 145 (29.8%) who underwent a NIS. A propensity score risk adjustment and propensity-based matching analysis was used to account for selection biases. Over a median follow-up of 7.4 years (range 0–9.9 years), 50 patients died in the IS group and 30 died in the NIS group (hazard ratio [HR], 0.74 [95% CI, 0.47–1.16]; P = .19), corresponding to an overall 9-year survival of 74.4% (95% CI, 69.2%–79.9%). Regardless of the technique used to adjust for between-group differences in baseline characteristics, the IS was associated with a longer survival, with adjusted HRs ranging from 0.47 (95% CI, 0.26–0.84; P = .01) to 0.61 (95% CI, 0.38–0.99; P = .047). The survival difference was largely attributable to a lower incidence of sudden death, which occurred in 10 patients in the IS group and in 16 patients in the NIS group, with HRs ranging from 0.24 (95% CI, 0.10–0.56; P = .001) to 0.28 (95% CI, 0.13–0.61; P = .001). Among patients with DM1, an invasive strategy was associated with a higher rate of 9-year survival than a noninvasive strategy.

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S.P.27
Electrophysiological study with prophylactic pacing and survival in adults with myotonic dystrophy and conduction system disease
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S.P.28
Reported incidence of orthopedic and cardiopulmonary complications in patients with congenital muscle disease
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Congenital muscle disease, including congenital myopathies (CM) and congenital muscular dystrophies (CMD) may develop hip dysplasia, progressive joint contractures, and scoliosis, sometimes leading to surgical intervention. Both the incidence of orthopedic problems and cardiopulmonary abnormalities that may impact surgical management and patient outcome in certain CM/CMD subtypes are unknown. Defining the incidence and outcome of orthopedic and cardiopulmonary involvement per CM/CMD subtype will direct pre and post-surgical management to improve patient care. The objective was to determine the incidence of orthopedic, pulmonary and cardiac problems in CM/CMD subtypes. A PubMed search of peer-reviewed literature limited to human case reports, series, and multicenter studies was performed with the keywords “congenital myopathy,” “congenital muscular dystrophy” and CM/CMD specific genes. We excluded articles that did not report on orthopedic, pulmonary or cardiac involvement and excluded patients without genetic confirmation or if symptom onset was after 2 years of age. Twenty-eight articles met the review criteria, which included 365 patients who had CM/CMD with confirmed mutations in either ACTA1, COL6, DNM2, FKTN, LAMA2, LMNA, POMT1, POMT2, POMGnT1, LARGE, RYR1, or SEPN1. There were 504 orthopedic findings, including congenital hip dislocation (24), torticollis (15), joint contractures (201), joint laxity (42), and abnormal spinal curvatures (222). More than half of patients (210) had respiratory compromise with 114 patients requiring ventilatory support. 27 patients had documented cardiac abnormalities. Orthopedic manifestations, respiratory compromise, and cardiac abnormalities are common in patients with congenital muscle disease. Pre-surgical cardiac and respiratory evaluations are indicated.

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S.P.29
Respiratory dysfunction of dysferlinopathy
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Dysferlinopathy is an autosomal recessive muscular dystrophy caused by DYSF mutations. There are two major phenotypes of dysferlinopathy, proximal dominant limb-girdle muscular dystrophy (LGMD) 2B and distal dominant Miyoshi myopathy (MM). Respiratory dysfunction has been rarely reported in dysferlinopathy patients. Objective: To examine the respiratory function in patients with dysferlinopathy. We retrospectively reviewed the respiratory function (% forced vital capacity [FVC] for 27 dysferlinopathy patients confirmed by genetic analysis and/or immunohistochemical staining in our hospital. The mean age at the onset was 22.8 ± 8.5 (range 10–46 years). Of 27 patients, LGMD2B, MM, and unclassified were 13, 13, and 1, respectively. Serum CK level was 5171 ± 3005 (range 192–12,671 IU/l). Twenty-one patients were still ambulant and four were wheelchair-bound. The %FVC was 99.0 ± 29.1 (range 15.3–131.0). In three (11.1%) patients, %FVC was <80% (23.9–41.4%) and two patients used non-invasive positive pressure ventilation. The average %FVC of patients within 25 years from the onset was 108.8 ± 13.5% (n = 19), while 68.2 ± 43.6% (n = 8) in patients over 25 years and 109.5 ± 12.3% in ambulatory patients (n = 21), while 43.3 ± 28.9% in wheelchair-bound (n = 4). The Linear regression analysis to determine the relationship between %FVC and other clinical parameters revealed the age (p = 0.004), the duration from the onset to present (p = 0.003), and the CK (p = 0.009) as to be significantly correlated with %FVC. There was no significant difference in %FVC of LGMD2B and MM. Dysferlinopathy can cause severe respiratory failure. Respiratory dysfunction in patients with dysferlinopathy, which is characterized by long disease duration and advanced weakness and muscle atrophy, should be carefully monitored.

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S.P.42 Effects of a physical exercise programme in adults with myotonic dystrophy type 1 – A one-year follow-up study
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PHYSICAL THERAPY AND GENERAL ASPECTS – POSTER PRESENTATIONS

S.P.42 Effects of a physical exercise programme in adults with myotonic dystrophy type 1 – A one-year follow-up study
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Reduced physical activity can be a consequence of a progressive neuromuscular disease like myotonic dystrophy type 1 (DM1) and lead to an increased risk for secondary chronic conditions. Regular physical activity has a positive effect on health in able-bodied populations, but there is limited clinical research on people with DM1. The aim of this study was to evaluate functioning, in particular walking capacity, lower-extremity performance, and mobility and balance, one-year after the completion of a physical exercise programme. Thirty-five persons with DM1, stratified for functional level, were assigned to a control group (10 women and seven men, mean age 41 years) or a training group (10 women and eight men, mean age 44 years). The training group participated in an individually adapted group exercise programme incorporating aerobic activities, strength training and flexibility exercises, for 60 min twice a week during 14 weeks. Eleven persons completed the exercise programme with at least 75% adherence. No negative effects were reported. Primary outcome measure was the six-minute walk test (6MWT) and secondary were the timed-stands test and the timed up-and-go test. Assessments were made before, immediately after and 12 months after the intervention period by two independent physiotherapists, blinded to group allocation. A linear mixed-model design was used for evaluation. Intention-to-treat analyses did not reveal any significant differences between groups or interaction effects between time and group. There was, however, a significant time effect for the 6MWT (p = 0.027). Pair-wise comparisons showed that the mean 6MWT distance decreased with 13.3 m (95% CI: 1.2–25.5), p = 0.022, from before to 12 months after the intervention, and with 15.7 m (95% CI: 3.5–28.0), p = 0.013, from immediately after to 12 months after the intervention. In conclusion, there were no long-term effects of the physical exercise programme and walking capacity decreased over time.

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S.P.44 Evaluation of a balance exercise programme in DM 1: A single-subject experimental study with 11 participants

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The aim of this study was to evaluate the effects of an individualised balance exercise programme in patients with myotonic dystrophy type 1 (DM1). A single-subject experimental design was used: baseline, intervention and second baseline (ABA) with repeated assessments. Eleven DM1 patients with subjective or objective balance difficulties (outpatients) participated in a balance exercise programme with individualised exercises at the hospital-based physiotherapy department. The balance exercise programme was attended during 10 weeks (two sessions/week). Primary outcome measure was self-assessed balance confidence in three self-chosen activities, in which the balance was a problem. Secondary outcome measures were the Activities-specific Balance Confidence (ABC) scale, dynamic balance performance assessed with Step test and Timed Up and Go, together with Timed 10 m walk at maximum pace, isometric muscle force in leg muscles and number of falls. The most important findings were: 7/11 patients experienced a significant increase in balance confidence (+>2SD) in at least one self-selected activity, 9/11 scored a higher overall balance confidence in ABC scale (mean +13%, p = 0.014), and increased balance performance in Step test (mean +18%, p = 0.008). All patients deteriorated in the ankle dorsiflexor force (mean −17%, p = 0.003). The knee extensor force remained unchanged. The study showed that self-assessed balance confidence, as well as balance performance measured with Step test, is likely to increase following an individualised balance exercise programme in persons with DM1. It is important to note that total muscles in DM1 patients are at risk of exhaustion during exercise, these patients therefore require close supervision when participating in these training sessions.

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S.P.45 ENERGETIC, a multidisciplinary self-management group program for reconditioning and managing fatigue

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Persons with neurological disorders may experience fatigue and secondary physical de-conditioning. A multidisciplinary team developed a client-centred self-management group program including several components: aerobic training; education about training; education on managing fatigue and implementation of training and energy conservation...