Muscle and neuromuscular junction diseases

PP2165
Sustained remission in a case of MuSK (+) myasthenia gravis treated with i.v. rituximab as primary therapy
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Introduction: Muscle specific tyrosine kinase (MuSK) antibody positive myasthenia gravis accounts for 50-70% of seronegative cases. MuSK (+) patients are predominantly women with early, prominent bulbar and respiratory symptoms and poor response to standard therapies. Several reports suggest a role for rituximab, an anti-CD20 monoclonal antibody, in patients who fail conventional immunotherapies. We report our experience with rituximab as first line therapy for a MuSK(+) MG patient.

Methods: The patient is a 29-year-old woman who developed symptoms of bulbar weakness and respiratory insufficiency at age 22. She required recurrent intensive care unit admissions over the next 10 months due to type 2 respiratory failure that resulted in tracheostomy placement. She had minimal limb weakness and remained undiagnosed for the subsequent 4 years. She underwent re-evaluation at age 26.

Results: The clinical examination revealed bilateral ptosis, facial weakness, tongue weakness and atrophy. A tracheostomy was in place and mild proximal weakness in the upper and lower extremities. The repetitive nerve stimulation revealed a 27% decrement at 3 Hz. EMG and NCS were normal. The acetylcholine receptor antibodies and anti-striated muscle antibodies were negative. Anti-MUSK antibodies were elevated. The patient was treated with intravenous rituximab 375 mg/m2 weekly for 4 weeks. The patient clinically improved. The tracheostomy was discontinued. She remained asymptomatic off all medications for over 3 years.

Conclusions: Intravenous rituximab may induce sustained remission in MuSK(+) myasthenia gravis even after a single course and may be considered for primary therapy. Ideally a randomized controlled study would interrogate this hypothesis.

Disclosure: Nothing to disclose

PP2166
Clinical and genetic features of patients with MNGIE: cohort at the department of neurology, Istanbul Faculty of Medicine
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Introduction: Mitochondrial neurogastrointestinal encephalopathy (MNGIE) is a rare genetic disorder which is characterized by gastrointestinal symptoms, cachexia, ptosis, ophthalmoparesis, neuropathy and leukoencephalopathy. Mutation in the thymidin phosphorilase encoding TYMP gene, leads to dysfunction of the catalytic activity of the enzyme causing systemic accumulation of its substrates, that causes mitochondrial DNA instability and dysfunction in oxidative phosphorylation chain. In recent years treatment options urges early diagnosis of this disorder. Here, we present the genetic and clinical features of seven Turkish patients, diagnosed as MNGIE in our clinic.

Methods: Clinical and laboratory findings of seven Turkish patients from four unrelated families diagnosed with MNGIE at the Department of Neurology, Istanbul Faculty of Medicine between 2009 and 2013 were retrospectively evaluated.

Results: Six patients were male, only one patient was female. Mean age of onset was 16.2±8.35 years. Intra- and inter-familial variability in the age of onset and rate of disease progression were striking. Nausea and vomiting were the most common presenting symptoms which were usually followed by bilateral ptosis and ophthalmoparesis within two years. Ptosis was absent in one patient with rapid progression, whereas all other characteristic symptoms were evident. All patients had gastrointestinal symptoms, cachexia, neuropathy and distal muscle weakness. Three patients died due to disease related causes during the follow up. Notably, we described four novel mutations in the TYMP in our patients.

Conclusions: Our data indicates the marked intra- and inter-familial phenotypic variability in MNGIE as previously described and four novel pathological mutations in TYMP.

Disclosure: Nothing to disclose
PP2167

Angiogenic factors dynamics during skeletal muscle regeneration

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Introduction: The establishment of a well-organized, stable vasculature is a key process during skeletal muscle regeneration, and a key goal in regenerative medicine. These processes are orchestrated by a large panel of systemic and local signals. However, detailed mechanisms are still unclear.

Methods: Multiplex assay was used to measure level progression of an array of pro-angiogenic growth factors and cytokines during muscle regeneration in animal models. Crush injuries were performed with a forceps on the left gastrocnemius muscle. Muscle and blood samples were collected at specific time-points. Selected factors were further detected in situ, by immunolabelling. The contralateral muscle was used as control.

Results: Significant local and systemic levels were detected for pro-angiogenic, inflammatory cytokines from 24h up to 2 weeks post-injury. Likewise, the local levels of notorious angiogenic factors like vascular endothelial growth factors A and C and hepatocyte growth factor increased in the 2nd/3rd week post-injury, but were accompanied by high systemic levels compared to fibroblast growth factor 2. Surprisingly, we detected significant tissue levels, peaking at the end of the 1st week post-injury, for a set of angiogenic factors - amphiregulin, betacellulin, endoglin, follistatin and PLGF-2 - that were never taken into account and tested in such an experimental system. The distribution of the interstitial cells involved in their synthesis during muscle regeneration was pointed out by immunofluorescence.

Conclusions: Identification of new angiogenic factors secreted locally during normal regeneration opens a promising perspective for improving skeletal muscle healing after injury or muscle diseases.

Disclosure: Nothing to disclose

PP2168

Myopathy with muscle hypertrophy as a rare presenting feature of primary amyloidosis

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In systemic amyloidosis, the major sites of clinically important amyloid deposition are in the kidneys, heart, and liver. We report a rare case of primary amyloidosis associated with plasma cell dyscrasia initially presenting with musculoskeletal manifestations. 65-year-old woman was admitted with a 6-month history of difficulties in walking, progressive muscle weakness, stiffness, myalgia and spontaneous muscle overgrowth. Examination revealed visible increase in muscle bulk in the pelvic girdle muscle group and also marked macroglossi. Severe weakness of deltoid, triceps, biceps, iliopsoas and gluteus maximus muscles was confirmed in the absence of reflex or sensory changes. Electromyographic findings and mildly elevated CK levels were compatible with myopathy. Magnetic resonance imaging (MRI) of muscle demonstrated generalized muscle enlargement and patchy edema prominent in pectoralis major, serratus anterior, paravertebral and pelvic girdle muscles. Muscle biopsy taken from deltoid muscle revealed mild variation in fiber size, accumulation of amyloid in thickened blood vessel walls as well as in muscle fibers with Congo red staining. In further investigations, the diagnosis of systemic AL amyloidosis in association with plasma cell dyscrasia was established. Over 3 months, cardiac, gastrointestinal and visceral involvement was evident. She did not respond to systemic chemotherapy and died of systemic complications of amyloidosis. This case highlights the importance of investigations for amyloid myopathy in middle-aged patients with progressive myopathy or muscle hypertrophy of unclear cause.

Disclosure: Nothing to disclose
PP2169

A patient with CASPR2 myasthenic-myotonic syndrome

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48-year-old female patient was first admitted in September 2011 when she experienced gradually progressive symptoms of clumsiness in the left hand, periodically diplopia, dysarthria and partial ptosis on the left. In the department of neurology all clinical symptoms resolved by the next day. Blood tests and brain MRI were normal. In January 2012 patient was referred to the neurology department in West-Tallinn Central Hospital. Neurological exam demonstrated ptosis on the left, dysarthria, weakness and atrophies of small hand muscles with brisk reflexes and fasciculations in arm muscles, but no pyramidal signs. ENMG revealed subacute neurogenic changes in C8 and Th1 innervated muscles. Extensive clinical workup was unremarkable. In February 2012 patient was re-admitted as dysarthria, dyspnoe and muscle weakness had progressed. Also, weakness and atrophies of small hand muscles, fasciculations in the tongue and shoulder-girdle area were more marked, however no plantar extensor signs were demonstrated. Repeated ENMG confirmed progression with fasciculations and fibrillations in 3 segments and right diplets in frontal muscle. CSF was normal and oligoclonal bands negative. Anti CASPR2 IgM was positive in low titre (+1:10). Amyotrophic syndrome was diagnosed. In May 2012 patient’s symptoms had stabilised with less fasciculations and weakness, small hand muscle atrophies had subsided. ENMG demonstrated positive changes with no spontaneous activity in limb muscles, large motor units were present diffusely. Repeated analysis CASPR2 IgM demonstrated higher titer (IgM++ 1:32 ) than 4 months ago. The final diagnosis of CASPR2 positive neuromyotonia was made.

Disclosure: Nothing to disclose

PP2170

Anxiety and depression symptoms in patients with generalized myasthenia gravis

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Introduction: We investigated the symptoms of anxiety and depression in Myasthenia Gravis (MG) patients using practical psychiatric scales and aimed to emphasize the need for clinical awareness.

Methods: Thirty patients (21 women), between 48-59 years of age, who were registered in the Neuromuscular Unit in our Neurology Clinic were included in the study. The depression and anxiety symptoms were evaluated using the Beck Anxiety Scale (BAS) and the Beck Depression Scale (BDS). The correlation between these scores and age, gender, disease duration, intensive care unit experience, medications being used and the frequency of admission to a medical center were analyzed using the SPSS statistical program.

Results: The BAS and BDS scores were found to be higher than the normal ranges in >50 of the whole patient group (56% and 60%, respectively). One third of all the patients required medical psychiatric treatment. The disease duration (RS:0.68 and 0.56, p=0.016 for BAS and BDS, respectively), admission rate (RS:0.66 and 0.46, p<0.001, for BAS and BDS, respectively) and hospitalization (RS:0.64 and 0.48, p<0.001, for BAS and BDS, respectively) were statistically significantly related with the BAS and BDS scores.

Conclusions: Although there is a predictable relationship between psychiatric symptoms and MG, surprisingly few studies have evaluated this correlation. The symptoms of anxiety and depression may easily mask the myasthenic symptoms and leading to delayed diagnosis or misdiagnosis of MG. Furthermore, these symptoms may mimic the myasthenic symptoms and leading to over-treatments for MG. Thus, the use of practical psychiatric scales to evaluate the psychiatric status in routine neurological visits would help in deciding the specific treatment strategies.

Disclosure: Nothing to disclose
PP2171

Stiff limb syndrome: a case report

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Introduction: Stiff-person syndrome (SPS) is a rare disorder characterized by rigidity of the truncal muscles, painful spasms, and an exquisite sensitivity to external stimuli. Stiff-limb syndrome (SLS) is a rare variant of SPS. We report a SLS in a young woman.

Case report: A 27-year-old healthy woman presented with a sudden onset of spasms of right arm muscles, preceded by sudden movement, loud noise or emotional stress. Her physical examination was remarkable in that her right upper extremity was rigid on passive and active ranges of motion. Movements were severely limited and painful. No paraspinal or axial contractions were palpated. Sensory examination was normal, and there was no extrapyramidal rigidity. EMG testing showed continuous motor unit activity on the upper right arm muscles. Anti-GAD antibody examination was negative. Immunological data and the paraneoplastic antibodies were negative. The patient improved at high doses of clonazepam. She was given also IV immunoglobulin as an adjunctive therapy.

Discussion: SLS is a newly emerging entity presenting focally with rigidity and spasms involving one or more limbs. The distal leg is the most affected. EMG show continuous motor unit activity at rest, but the distribution is very different from SPS. GAD auto-antibody titers are raised in a smaller proportion. SLS can also be paraneoplastic. Response to treatment is partial in SLS in marked contrast with those with SPS. Our patient remains ambulatory.

Conclusion: Only two cases of SLS exhibiting upper limb signs were reported. Our case report illustrates this exceptional variant of SPS.

Disclosure: Nothing to disclose

PP2172

Val71Ala: a rare transthyretin variant in an American patient and response to liver transplantation

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Introduction: Mutations in TTR cause an autosomal dominant neuropathy, characterized by deposition of insoluble amyloid fibrils in the extracellular space in the nerves and muscles. Liver transplantation is the only treatment option in the USA. Although over 50 TTR mutations have been reported, the Val30Met mutation remains the most common. The Val30Met patients respond well to liver transplantation. A rare Val71Ala mutation was previously described in Europe; however, how these patients respond to liver transplantation is not known. Here, we report the first American patient with the Val71Ala mutation, the clinical characteristics and the response to liver transplantation.

Methods: Case report.

Results: A 42-year-old man of Swedish/Irish descent, presented with a 2-year history of progressive weight loss, weakness/numbness/pain in arms and legs. Examination revealed weakness of distal arms and legs and decreased pin prick sensation up to mid-thighs, and up to the elbows. Vibration sense was absent at ankles and knees, and decreased at wrists. Patellar and ankle reflexes were absent. EMG/NCS revealed a CIDP-like picture. Muscle and nerve biopsies showed amyloid deposits. DNA testing revealed Val71Ala mutation in the Transthyretin gene. 8 months later, the patient underwent a domino liver transplant, following which mild betterment of neurological weakness was reported.

Conclusions: Here we provide the first report of a rare Transthyretin mutation (Val71Ala) as a cause of Transthyretin amyloid neuropathy in an American patient. Our study also provides data to suggest that liver transplantation may be beneficial in the Val71Ala patients, similar to the Val30Met patients.

Disclosure: Nothing to disclose
PP2173

Both binding and blocking antibodies correlate with disease severity in myasthenia gravis
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**Introduction:** The purpose of this study was to compare assays of acetylcholine receptor (AChR) binding and blocking antibodies for their ability to the diagnosis of myasthenia gravis (MG) and to estimate the clinical severity of MG patients.

**Methods:** The patients enrolled in the study had been tested for both binding and blocking antibodies and had disease duration exceeding 2 years since diagnosis. The patients were divided into five main classes by the Myasthenia Gravis Foundation of America (MGFA) clinical classification. Again, the enrolled patients were divided into ocular and generalized group according to MGFA classification. We compared the type and titer of antibodies and the thymus status between the ocular and generalized group.

**Results:** Thirty-five patients met the inclusion criteria. Of these, 16 patients (47%) had both blocking and binding AChR antibodies, 11 patients (31%) had only binding antibodies, and 8 patients (22%) had only blocking antibodies. By defined clinical classification, the ocular and generalized groups included 10 and 25 patients, respectively. Sixteen patients in the generalized group possessed both AChR antibodies, with the remaining patients displaying only the binding antibody. All the patients with only blocking antibody were classified into ocular group.

**Conclusions:** Our study demonstrates that MG patients with both binding and blocking antibodies show more severe generalized MG or myasthenic crisis. Patients in the generalized group tend to have higher titers of binding and blocking antibodies. We suggest that both antibodies tests are useful in determining whether the disease will generalize.

**Disclosure:** Nothing to disclose

PP2174

Cholinergic transmission of outer hair cell impaired in myasthenia gravis
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**Introduction:** Nicotinic acetylcholine receptors (nACHRs) are located on outer hair cell (OHC) which is source of otoacoustic emission (OAE), and can be inhibited by alpha-bungarotoxin like muscular nACHR in myasthenia gravis (MG). The classical techniques such as repetitive stimulation, autoantibody, and response to ACh esterase inhibitor (AChEI) are sometimes not helpful for diagnosis of MG. The purpose of this study is to evaluate OAE for the possible role in diagnosis of MG.

**Methods:** We performed transient evoked otoacoustic emissions (TEOAEs) and distortion product otoacoustic emissions (DPOAEs) on 30 ears of 15 MG with normal hearing, and on 20 ears of 10 controls.

**Results:** Mean age was not different. TEOAE were significantly lower in MG (3.41 dB SPL) than in controls (8.69 dB SPL) (p<0.01). DPOAE were lower in MG at higher frequencies between 2,026 and 4,053 Hz (p<0.01). TEOAE and DPOAE were significantly lower in repetitive stimulation positive group and in AChR antibody positive group. TEOAE and DPOAE were correlated with titers of antibody.

**Conclusions:** The decrease of OAE in MG is probably related to the reduced cholinergic transmission at OHC level. This study supports the role of ACh in the efferent function of OHC, as well as the impaired AChRs on OHC in MG. Furthermore, more reduced OAE in repetitive stimulation positive or AChR antibody positive groups with the correlation between OAE and antibody titer suggests impaired OHC function by AChR antibodies in MG. Consequently, measuring TEOAEs and DPOAEs may be useful in the diagnosis of MG.

**Disclosure:** Nothing to disclose
**PP2175**

**Thymoma with or without paraneoplastic myasthenia gravis – is minimally invasive tumour resection really safe?**

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**Introduction:** Thymomas are malignant tumours, often associated with Myasthenia gravis and treated in neurological context. Complete operative resection is along with radiation and chemotherapy the mainstay of therapy and the most important prognostic factor. Regarding the surgical procedure no clear guidelines exist. Whilst there is agreement that an en bloc resection of the tumour, the complete thymus and the surrounding mediastinal fat is necessary, the technical approach differs considerably. Most treatment centres choose an open approach through a median sternotomy, but recently many minimally invasive techniques arise. These methods do have advantages, but it remains uncertain, whether they are as safe as the conservative approaches, especially in long term observation. Here we want to report five cases of tumour recurrence after minimal invasive thymoma resection and discuss the potential mechanism.

**Methods:** retrospective data evaluation.

**Results:** We can present five cases of patients with thymoma ± myasthenia gravis, with recurrence after a minimally invasive operation. In two cases thymoma and thymus gland were not completely resected but classified as a complete resection. In another case pleural carcinosis emerged years after the specimen pouch ruptured during operation.

**Conclusions:** We can show that recurrences of thymomas do emerge using minimally invasive techniques and that the occurrence can be related to the operation technique in some cases. Therefore we think that the use of these methods should be carefully considered, having in mind that recurrences of thymomas can occur many years later.

**Disclosure:** Nothing to disclose

**PP2176**

**Characteristic features of long-time survivors patients with Duchenne muscular dystrophy**

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**Introduction:** The prognosis of Duchenne muscular dystrophy (DMD) has been remarkably improved according to the progress of the respiratory care and cardiac protective therapy. At present, it is possible to prolong over 40 years in Japan. The aim of this study was to clear the clinical characteristics of long-time survivors patients with DMD.

**Methods:** The mean age of death among DMD inpatients was 32.1-years-old for the last ten years in our hospital. We investigated clinical features of DMD inpatients survived over 35 years.

**Results:** Eight of 14 inpatients clinically diagnosed as DMD were over 35 years old. Four of 8 patients showed deleted exons of the dystrophin gene by MLPA analysis and were picked up. The mean age of 4 patients was 44 years old. They were bedridden because of severe weakness of the skeletal muscles. They had non-invasive positive pressure ventilation all day due to severe respiratory dysfunction. On the other hand, their cardiac involvements were not so damaged. Their ejection fractions measured by ultrasonic echocardiography were 0.18-0.50. They had some medicines for cardiomyopathy, such as angiotensin converting enzyme inhibitor, diuretic, digitalis, and so on. Two patients received oral nutrition and 2 patients required tube feeding. The mean of body mass index was 13.6. Two patients were diagnosed as depressive state or adjustment disorder in spite of mild cognitive dysfunction by psychologist.

**Conclusions:** We experienced long-time survived patients with DMD. All of them had artificial ventilatory support but their cardiomyopathies were not so severe.

**Disclosure:** Nothing to disclose
PP2177

Correctness of referral to the repetitive nerve stimulation test and diagnostic outcome

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Introduction: Repetitive nerve stimulation test (RNST) is a useful method in the diagnosis of diseases of the neuromuscular synapse. The aim of this study was to verify the correctness of indications when referring to RNST and sensitivity of the method in the diagnosis of diseases of the neuromuscular junction (NMJ).

Methods: RNST was performed on 91 referred patients under the suspicion of NMJ disorders. Synergy PIU NCS EMG System was used and test performed on distal n. medianus - m. abductor pollicis brevis and proximal system n. accessorius - m. trapezius at low stimulation frequencies (1-3Hz). Any decrement in CMAP amplitude of more than 10% was defined as abnormal. Referral diagnosis was verified after the completion of planned examinations.

Results: After all tests were conducted, of 91 patients sent for RNST (mean age 15.34, SD 9.38), diagnosis of NMJ disorder was confirmed in 61 (67%), out of which 20 with congenital myasthenic syndrome (KMS), and 41 with autoimmune myasthenia gravis (aMG). 55% of KMS patients and 70.7% of aMG patients had abnormal RNST, without reaching statistically significant difference (Pearson Chi-Square 1.474, p=0.225). Eight (27.6%) had abnormal RNST on distal system, 16 (55.2%) on proximal, and 5 on both systems in the group with aMG. One patient with KMS had abnormal RNS findings on the distal, and the remaining 10 (90.9%) on the proximal system.

Conclusions: Results suggest high accuracy of the presumed diagnosis before referral to RNST, good sensitivity of the method and continuing need for its use in the diagnosis of NMJ disorders.

Disclosure: Nothing to disclose

PP2178

Unusual clinical manifestations of hereditary inclusion myopathy

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Introduction: Hereditary inclusion body myopathy (HIBM) is a group of progressive myopathy disorders, which are uncommon in the general world population. Case 1, 2 and 3: 41-year-old daughter, 70-year-old father and 60-year-old uncle presented bilaterally ptosis, progressive proximal muscle weakness and atrophy in the upper and lower extremities with diminished distal muscle. Blood tests for serum Creatine Kinase (CK) were in normal ranges. EMGs showed myopathic and neurogenic potentials especially in the proximal muscles. Muscle biopsy showed non-enflmatuar myopathic findings with inclusion bodies and rimmed vacuoles. Case 3 and 4: 36-year-old and 41-year-old sisters presented progressive weakness of the distal leg muscles of the upper and lower extremities with sparing quadriceps muscles. Distal leg muscles of the lower extremities were symmetrically atrophic and deep tendon reflexes were diminished. CK levels were normal. EMGs showed myopathic and neurogenic potentials especially in the proximal muscles. Muscle biopsy showed non-enflamatuar myopathic findings with inclusion bodies and rimmed vacuoles.

Conclusion: HIBM constitutes a unique group of neuromuscular disorders characterized by adult-onset and a typical muscle pathology including rimmed vacuoles and filamentous inclusions. Clinical presentations of HIBM are variable:
(1) An autosomal dominant form where the quadriceps are one of the first muscles to become weak.
(2) An autosomal recessive form so-called quadriceps-sparing myopathy.
(3) Inclusion body myopathy associated with Paget’s disease of bone and frontotemporal dementia.

We report 5 cases from 2 families in this study. To our knowledge, ptosis in HIBM is not described earlier.

Disclosure: Nothing to disclose
PP2179

Breathing pattern and central ventilatory drive in late-onset Pompe’s disease (LOPD)

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Background: Pompe’s disease is an autosomal recessive disorder characterized by GAA deficiency that results in intra-lysosomal glycogen accumulation mainly affecting skeletal muscles. Respiratory symptoms may represent LOPD onset and have been attributed to diaphragm involvement. Glycogen accumulation has been demonstrated also in peripheral nerves and in the CNS, either in patients or in animal models. Consequently, it has been suggested that there could be a potential neurogenic influence on LOPD patients respiratory dysfunction.

Aim: This study is to determine the breathing pattern and the central ventilatory drive in LOPD patients to better define the pathophysiology of respiratory impairment.

Methods: 15 LOPD patients were studied and compared with a control group of 15 age- and sex-matched individuals. The rate, time, depth of breathing and inspiratory occlusion pressure in mouth in the first 0.1 second (P0.1) were measured under basal conditions and compared with spirometric and respiratory pressure measurements.

Results: In LOPD patients, respiratory pattern including shallow breathing index and respiratory rate were increased, whereas P0.1 value was decreased, (p<0.001).

Conclusions: The respiratory pattern found in LOPD patients has also been documented in other neuromuscular disorders due to respiratory muscle weakness. Conversely, P0.1, that is an expression of the respiratory center output, in LOPD patients resulted decreased whereas in other muscle disorders resulted increased as compensatory event to the muscle weakness. Our data suggest that LOPD patients have a low activity of the respiratory drive, demonstrating that an involvement of CNS contributes to the pathophysiology of the respiratory failure.

Disclosure: Nothing to disclose

PP2180

The interaction between tropomyosin-related kinase-B receptor-S and serine kinases modulates acetylcholine release in adult neuromuscular junctions

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We conducted an electrophysiological study of the functional link between the tropomyosin-related kinase-B (trkB) receptor signaling mechanism and serine-threonine kinases, both protein kinase-C (PKC) and protein kinase-A (PKA). We describe their coordinated role in transmitter release at the neuromuscular junction (NMJ) of the Levator auris longus muscle of the adult mouse. The inhibition of the trkB receptor with K-252a results in a significant reduction in the size of EPPs indicating that this receptor may be coupled to ACh release stimulation. We found that the intracellular PKC pathway can potentiate ACh release without the involvement of the trkB receptor function. However the trkB pathway needs an operative PKC pathway to be coupled to the release mechanism and potentiate it. The operativity of trkB is a necessary condition and one effect of trkB may be PKA stimulation.

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Disclosure: Nothing to disclose
PP2181

Phenotypic variability of myotonic dystrophy type 2

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Introduction: The aim was to assess manifestations of myotonic dystrophy type 2 (DM2).

Methods: Study comprised 34 DM2 patients and 34 matched DM1 patients (67.6% females, 53±10 years).

Results: Proximal muscles were similarly affected in both groups, while distal were less involved in DM2 (p<0.01). Following symptoms were less common in DM2 (p<0.01) - ptosis (3% vs. 62%), mastication weakness (21% vs. 82%), impaired speech (21% vs. 94%), swallowing difficulties (15% vs. 38%), significant sternocleidomastoid and trapezius weakness (56% vs. 100% and 15% vs. 36%), handgrip and jaw myotonia (71% vs. 100% and 38% vs. 97%). Forced vital capacity <90% was found in 3% of DM2 patients and 52% of DM1 (p<0.01). Differences in EMG findings were not significant - myopathy was present in 93% of DM2 and 100% of DM1 patients and myotonia in 90% and 100%, respectively. Calf hypertrophy was found in 29% and hand tremor in 38% of DM2 patients, while were absent in DM1. Severe ECG abnormality was found in 9% of DM2 and 22% of DM1 patients (p<0.05) with shorter PQ in DM2 (0.16±0.03 vs. 0.21±0.02, p<0.01). Diabetes was more frequent in DM2 (32% vs. 7%, p<0.01). Frequency of eye cataract was similar in DM2 and DM1 (82% vs. 97%, p=0.05).

Conclusions: DM2, compared to DM1, is manifested with older age at onset, less involvement of distal, cranial and respiratory muscles, less pronounced myotonia and cardiac abnormalities. Presence of calf hypertrophy, hand tremor and diabetes is suggestive of DM2.

Disclosure: Nothing to disclose

PP2182

Enzymatic replacement therapy in patients with late-onset Pompe’s disease – a 5-year follow up

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Introduction: Late-onset Pompe disease (LOPD) is progressive metabolic myopathy, affecting skeletal muscles, which, if untreated, leads to serious disability and/or respiratory failure. Enzyme replacement therapy (ERT) improves muscle strength, respiratory function and prevents disease progression. We present a 5-year follow-up of 4 LOPD patients treated with ERT.

Methods: Four patients with LOPD received ERT: two started treatment in 2008, other two in 2010. Patients received recombinant human alpha-glucosidase in dose 20 mg/kg intravenously every two weeks. Physical efficiency was assessed in 6-minute walk test (6MWT). Spirometry was performed to examine FVC and FEV1. Liver enzymes, LDH, CK and CK-MB levels were assessed.

Results: Walking distance in 6MWT increased by average 9% in the first two years and 21% in the first three years of treatment in two patients with the longest treatment compared to the baseline. Similar changes were detected in spirometry: the biggest FVC increase was observed in two patients with the highest FVC values before treatment, which increased to normal values adjusted for age and sex in three years of treatment, that is by 28% and 34%. In two other patients FVC reached 88% and 76% of predicted values. ERT didn’t affect liver or muscle enzymes levels.

Conclusion: The improvements of exercise tolerance and FVC were observed in all patients in the first three years of treatment and were the biggest in patients treated the longest and with the least severe neurological and respiratory symptoms. Early ERT introduction results in higher improvement of respiratory and ambulation functions.

Disclosure: Nothing to disclose
PP2183

Metabolic syndrome in patients with myotonic dystrophy type 1

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Introduction: Aim was to investigate frequency and
features of metabolic syndrome (MetSy) in patients with
myotonic dystrophy type 1 (DM1).

Methods: Study comprised 66 DM1 patients (50% males,
41.9±10.5-years-old, with disease duration of 41.9±10.5
years and CTG repeat length of 751.9±280.6). New
worldwide consensus criteria for MetSy from 2009 were
used.

Results: Components of MetSy were present with following
frequencies: hypertriglyceridaemia 67%, low HDL
cholesterol 35%, hypertension 18%, central obesity 14%,
hyperglycemia 9%. MetSy was present in 11 (17%) of
patients, among them 7 (11%) had three components and 4
(6%) had four components. Presence of MetSy was not in
association with patients gender and age, severity and
duration of disease, neither with CTG repeats length
(p>0.05). Patients with MetSy had significantly lower total
SF-36 score as a measure of quality of life in comparison to
patients without MetSy (34.8±21.6 vs. 53.8±23.2, p<0.05).

Conclusions: Although certain components of MetSy are
very frequent in patients with DM1, only 17% of them met
the criteria for MetSy. Patients with MetSy had significantly
lower quality of life.

Disclosure: Nothing to disclose

PP2184

Subclinical cardiac involvement in thymomatous myasthenia gravis

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Introduction: Myasthenia Gravis primarily affects skeletal
muscles, however cardiac involvement has been reported in
thymomatous MG. We present a case of an MG patient
whom cardiac Magnetic Resonance Imaging (cMRI) in
accordance to premature ventricular beats (PVC) confirmed
the presence of cardiomyopathy and the application of
cortisone treatment reversed it.

Methods: A 58- year-old male presented with a two- month
history of eyelid ptosis after thymectomy and fatigue
induced deterioration of clinical condition which improved
at rest. The electrophysiological study showed disorder of
the neuromuscular junction and the serum acetylcholine
receptor antibodies had a titer of 46 nM (positive titer>0.6
nM). Pyridostigmine resulted in mild clinical improveme-
tment. A thorough cardiac evaluation was performed,
including electrocardiogram, cardiac ultrasound, 24 hour
ambulatory ECG (Holter) and cMRI. ECG and echo didn’t
show abnormalities related to MG while Holter showed
moderate number of PVC (>30 beats/hour) and one couplet
of PVCs. Exercise test was negative for coronary artery
disease while enhancement areas in the interventricular
septum and in the lateral wall of the LV were seen during
the cardiac MRI in T1 sequences indicative of fibrotic
process. The patient after the cardiac exams received
prednisolone 60mg/d with clear clinical improvement while
the repetitive cMRI was clear with no evidence of
gadolinium enhancement.

Conclusions: Although myocardial damage is usually
detected long after the diagnosis of MG, in our case, there
was concurrent presentation of cardiac involvement and
myasthenia process. The combination of cMRI and Holter
successfully detected cardiac abnormalities for which the
patient received cortisone.

Disclosure: Nothing to disclose
PP2185

Two cases of myotonic dystrophy type 1 associated with white matter abnormalities of the brain
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PP2186

Duchenne muscular dystrophy in Tunisian children
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PP2187

Prevalence and risk factors of cancer in a cohort of patients with myotonic dystrophy type 1
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PP2188

Miyoshi myopathy: a case report
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PP2189

Course of myasthenia gravis during pregnancy – experience in the Clinical Centre Nis, Serbia
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PP2190

Periodic paralysis: a thyrotoxic periodic paralysis case
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PP2191

Correlations between creatine kinase and blood pressure in repeated measurements
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PP2192

Immunological background of myasthenia gravis
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PP2193

Abstract withdrawn

PP2194

A case of late onset Pompe's disease
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PP2195

Antibodies to acetylcholine receptors in case of myasthenia gravis associated with concomitant autoimmune diseases
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Paper Poster Sessions

PP2196
Myasthenia gravis as a paraneoplastic syndrome associated with colon adenocarcinoma
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PP2197
Treatment of a seronegative myasthenia gravis patient with complications
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PP2198
Abstract withdrawn

PP2199
Ischemic infarction, sepsis and trombocytopenia after using intravenous immunoglobuline in a patient with chronical demyelinating inflammatory neuropathy
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PP2200
Myotonic dystrophy type 1 as a multisystemic disease – lessons from the Serbian Registry
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PP2201
Electromyographic correlation with clinical severity, antibody levels and treatment response in myasthenia gravis
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PP2202
Idiopathic inflammatory myopathies – clinical features, complementary investigation and treatment response
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