Muscle and neuromuscular junction diseases

**EP2142**

**Isolated ocular distribution in double seronegative myasthenia with low density receptor-related protein 4 antibodies: a case series**


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**Introduction:** Antibodies targeting low density receptor-related protein 4 (LRP4), have been recently detected in patients with double seronegative myasthenia gravis (MG). The clinical phenotype of this particular subgroup of patients has not been clearly elucidated.

**Case series:** We present clinical and laboratory findings concerning 3 cases of double seronegative MG patients with LRP4 antibodies. A total of 2 females and 1 male patient of Caucasian origin were admitted to our Department with intermittent diplopia and eyelid ptosis. Symptoms had a fluctuating course and worsened during the day. Clinical examination did not reveal weakness in limb, axial, facial and tongue muscles. Deep tendon reflexes were elicited normal without signs of pyramidal tract involvement. Pharyngeal reflexes were preserved. Thorough blood tests and screening for systemic autoimmune diseases were unrevealing. Brain MRI and CT of the mediastinum were unremarkable. Serological tests for acetylcholine receptor antibodies and muscle specific kinase receptor antibodies were negative. Patients serum was also tested for antibodies against LRP4 using a cell-based assay. The results were positive. Repetitive nerve stimulation and single fiber electromyography confirmed the diagnosis of myasthenia gravis. All patients responded adequately to pyridostigmine.

After six months two of them remained free of symptoms. The remaining patient had substantial resolution of her symptoms after the addition of prednisolone.

**Conclusions:** Our double seronegative patients with LRP4 antibodies share a common phenotype, characterized by ocular distribution, mild or moderate severity and favorable response to pyridostigmine.

**Disclosure:** Nothing to disclose

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**EP2143**

**Distal myosin heavy chain-7 (thumb) myopathy due to the novel transition c.5566G>A with heterogeneous cardiac involvement**

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**Introduction:** Myosin-heavy-chain (MYH7)-myopathy manifests clinically with a distal, scapuloperoneal, limb-girdle (proximal), or axial distribution and may involve the respiratory muscles. In the majority of the cases the heart is affected, ranging from relaxation impairment to dilative cardiomyopathy with ventricular arrhythmias. Progression of cardiac involvement and earlier onset in successive generations has not been reported in MYH7-myopathy.

**Methods:** Case study.

**Results:** In a five-generation family MYH7-myopathy manifested with late-onset, distal > proximal myopathy and variable degree of cardiac involvement. The index patient developed myopathy from age 49y with anginal chest pain. Her mother presented with a similar phenotype but had only developed myocardial relaxation impairment. The daughter of the index patient had only mild distal myopathy but presented with left ventricular hypertrobrubeculation / noncompaction and required an implantable cardioverter defibrillator (ICD) because of ventricular arrhythmias since age 37y. Her daughter was diagnosed with dilated cardiomyopathy at infancy, without overt skeletal muscle disease. MYH7-myopathy in the presented family was due to the novel mutation c.1566G>A in the MYH7 gene.

**Conclusions:** There is cardiac involvement in MYH7-myopathy, and cardiac affection in MYH7-myopathy is highly variable between the generations ranging from relaxation abnormality to noncompaction, ventricular arrhythmias, and dilated cardiomyopathy. While manifestations and progression of MYH7-myopathy may be mild, cardiac disease in MYH7-myopathy may be highly variable and progress with successive generations.

**Disclosure:** Nothing to disclose
EP2144
Carnitine deficiency, a spectrum disorder: case series histopathological and ultrastructural characterization
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Introduction: The brain must first oxidize ketone derivatives of acetyl CoA and acetoacetyl CoA produced by hepatic β-oxidation of fatty acids before they can be used as an energy source. To achieve this, the “carnitine shuttle” facilitates the transfer of long-chain fatty acids from the cytoplasm across the inner mitochondrial membrane into the mitochondrial matrix for β-oxidation in muscle, liver and cardiac tissues. The shuttle consists of three enzymes (carnitine palmitoyl-transferase 1, carnitine acylcarnitine translocase, carnitine palmitoyl-transferase 2), and organic cation transporter type 2 (OCTN2). A deficiency in any of these components leads to carnitine deficiency, with a wide spectrum of presentations, and creates a diagnostic challenge for clinicians.

Methods: Case series of 6 patients with carnitine deficiency (age range 1.5 to 53 years).

Results: Neurological manifestations included hypotonia, burning pain, decreased endurance, sensory deficits, developmental delay, stiffness, poor coordination and muscle weakness. All cases had low serum carnitine levels. In one case, a new variant of the gene responsible for primary carnitine deficiency (SLC22A) was found. Two cases had abnormal muscle biopsies. Two cases had suspected primary carnitine deficiency, one had secondary carnitine deficiency and the other three were not clear. In all the six cases, neurological symptoms improved after initiation of carnitine supplementation.

Conclusions: Our case series highlights the wide spectrum of neurological complaints in patients with carnitine deficiency. Our report also underscores the importance of including carnitine deficiency in the differential diagnosis for many neurological signs and symptoms so that treatment can be initiated if appropriate.

Disclosure: Nothing to disclose

EP2145
Late onset myasthenia gravis – what is specific about it?
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Introduction: Myasthenia gravis (MG) is heterogenous regarding clinical features, age at onset, thymus pathology and different autoantibodies. Recognition of clinical subtypes is essential in management and prognosis of the disease.

Aim: To analyze if late onset MG (LOMG, onset >50 years) has specific features compared to early onset form (EOMG, onset <50 years).

Methods: 338 LOMG patients were treated in our Clinic from 2002 until 2010. This group was compared with 189 patients with EOMG from the MG Belgrade registry.

Results: Male predominance (1.7:1) was observed in LOMG, and female predominance in EOMG (2.8:1). Anti-AChR antibodies were more common in LOMG (90.3% vs. 79.7%; p<0.05). The presence of MuSK antibodies was similar in both groups of seronegative patients (18.5% in EOMG vs. 20% in LOMG; p>0.05). Severity of the disease was similar in EOMG and LOMG group (p>0.05). Pure ocular form was more common in LOMG group (21.0 vs. 10.6%; p<0.01). Thymoma was equally present in LOMG and EOMG patients (13.2% vs. 14.5%; p>0.05), while hyperplasia was more common in EOMG group (63.0% vs. 5.6%; p<0.01). Other autoimmune disorders were found in 22.2% of EOMG and in 8.9% of LOMG patients (p<0.01), while malignancies were present with similar frequencies in both groups (8.0% in LOMG vs. 5.3% in EOMG; p>0.05).

Conclusions: Male predominance, low incidence of thymus hyperplasia, higher frequency of AChR antibodies, pure ocular form and lower frequency of autoimmune disorders suggest that LOMG is a different entity of this disease.

Disclosure: Nothing to disclose
Design of a confirmatory phase 3, multicentre, randomized, double-blind, placebo-controlled study of ataluren in patients with nonsense mutation Duchenne muscular dystrophy

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Introduction: In ~13% of patients, Duchenne muscular dystrophy (DMD) is caused by a nonsense mutation (nm) in the dystrophin gene. Ataluren is an investigational oral drug designed to promote ribosomal read-through of premature stop codons in mRNA, leading to production of full-length, functional protein. We describe an ongoing confirmatory phase 3 placebo-controlled study designed to assess the efficacy and safety of ataluren 40 mg/kg/day in boys with nmDMD. The design of this study reflects lessons learned from prior studies and targets a study population to best show a treatment effect over 48 weeks.

Methods: All patients have a confirmed nonsense mutation in the dystrophin gene, are 7-16 years of age, are receiving a stable dose of corticosteroids, and have a screening 6-minute walk distance (6MWD) ≥150 metres but below the protocol-specified %-predicted threshold. Overall, 220 patients were randomized in a 1:1 ratio to placebo or ataluren. The primary endpoint is 6MWD after 48 weeks. Secondary efficacy measures include timed function tests, quality of life, North Star Ambulatory Assessment, and patient/parent-reported disease-related symptoms and activities of daily living.

Results: In a retrospective subgroup analysis of patients in the phase 2b trial of ataluren in nmDMD who met the current study criteria, the difference between ataluren 40 mg/kg/day (administered as 10, 10, 20 mg/kg tid; n=30) vs placebo (n=31) in mean change in 6MWD over 48 weeks was approximately 50 metres.

Conclusions: This study is designed to confirm the treatment effect of ataluren seen in the phase 2b trial.

Disclosure: JB, AR, RS, GLE, MH and SWP are all employees of PTC Therapeutics, Inc., which has developed ataluren.

EUROMAC: disease registry for McArdle disease and other pure muscle glycogenolytic disorders presenting with exercise intolerance

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Introduction: The European Union has funded the development of a new disease registry for McArdle disease and other rare glycogenolytic disorders presenting with exercise intolerance. The scope of the project is to identify as many patients as possible across all European countries and to collect important natural history and epidemiological data.

Methods: EUROMAC is a new European network which currently has 20 partners from 7 European countries and includes collaborators from Turkey and the USA. The registry will be accessed directly by patients via the EUROMAC website and aims to recruit as many patients as possible from all European countries. A database will be developed of diagnostic laboratories and specialist clinics in Europe which will be made freely available via the website. Patient support groups will also be involved.

Results: The EUROMAC consortium aims to improve genetic diagnosis by signposting relevant diagnostic laboratories. Standards of care will be developed, together with a plan to develop outcome measures for large multicentre clinical trials. The project will incorporate public participation and aims to improve access to patient support bodies across Europe. Data on natural history and epidemiology of patients living in Europe will be analysed.

Conclusion: the EUROMAC is an European registry for McArdle disease and other rare glycogenolytic disorders. We seek to recruit as many European partners as possible and welcome collaborators and volunteers both from health services and patient support organisations.

Apply online: http://euromacregistry.eu/info@euromacregistry.eu +34 934 894 054

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EP2148

Muscle MRI of scapular girdle in facioscapulohumeral muscular dystrophy (FSHD)

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Introduction: In Facioscapulohumeral muscular dystrophy (FSHD), the upper girdle is early involved and often difficult to assess only relying on physical examination. Our aim was to evaluate the pattern and degree of involvement of upper girdle muscles in FSHD compared with other muscle diseases with scapular girdle impairment.

Methods: We propose an MRI protocol evaluating neck and upper girdle muscles. A large cohort of consecutive symptomatic FSHD patients and patients affected by muscular dystrophies and myopathies with prominent upper girdle involvement underwent this protocol.

Results: The trapezius and serratus anterior were the most and earliest affected muscles in FSHD, whilst spinati and subscapularis were consistently spared even in late disease stages. Asymmetry and hyperintensities on short-tau inversion recovery (STIR) sequences were common features. The overall involvement appears to be disease-specific in FSHD as it significantly differed from that encountered in the other myopathies.

Conclusions: The detailed knowledge of single muscle involvement provides useful information for correctly evaluating patients’ motor function and to set a baseline for natural history studies. Upper girdle imaging can also be used as an additional tool helpful in supporting the diagnosis of FSHD in unclear situations, and may contribute with hints on the currently largely unknown molecular pathogenesis of this disease.

Disclosure: Nothing to disclose

EP2149

Heterogeneity of muscle and CNS involvement in Steinert’s disease (DM1): what links behaviour to brain imaging?

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Introduction: Myotonic dystrophy is a genetic and multisystemic disorder due to polynucleotide expansions being only partially reliable to predict phenotypic expression. Beyond muscular involvement DM1 phenotype can be characterized by functional/morphological brain abnormalities to different extents. From a neuropsychological point of view executive and visuo-spatial dysfunctions, mood and personality impairments are reported.

Methods: 40 subjects with established clinical-genetic diagnosis, underwent a complete neurological assessment, including psychological interview and neuropsychological evaluation. Main caregiver underwent patient’s Quality-of-life interview. A subgroup of 15 patients underwent brain MRI investigation.

Results: We found reduced scores in neuropsychological tests for frontal functions (61%) and visuo-spatial impairments (66%); interestingly verbal abilities were rather preserved (80%). Behaviour was characterized by mixed mood conditions (anxiety, depression, apathy) and by variable sets of pathological personality traits, even though without fulfilling diagnostic criteria for major psychiatric disorder according to DSM-IV. Patient’s and main caregiver’s reports showed internal discrepancies (63%), with patients tending to denial some aspects of their condition. Brain imaging revealed involvement of the white matter in frontal (53%), parietal (27%) and temporal (73%) lobes. Statistical analysis showed significant relationships between reduced spatial memory performances and temporal lobe white matter changes (Fisher-Exact-Test p<0.05).

Conclusions: Our study indicates that CNS involvement in DM1 is an heterogeneous condition characterized by cognitive/psychopathological dysfunctions; this could be a prominent feature in DM1, leading to an increased burden in management. White matter lesions are common in DM1 patients independently from CTG-repeat-expansion and disease-duration. CNS disorders could have significant relationships with white matter lesions and should be investigated since the early phases of illness.

Disclosure: Nothing to disclose
EP2150

Increased prevalence of malignancy in adult mitochondrial disorders

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Introduction: There are indications that patients with a mitochondrial disorder (MID) develop more frequently malignomas or benign tumours than the general population. Aims of the study were to find out if the prevalence of tumours is actually increased in MID-patients and which of the malignomas or benign tumours are the most frequent.

Methods: Retrospectively evaluated were the charts of MID-patients for the presence of malign or benign tumours. MID was diagnosed according to the modified Walker-criteria.

Results: Among 475 MID-patients screened for tumours, at least a single malignoma was found in 65 patients (13.7%), and at least a single benign tumour in 35 patients (7.4%). Among those with malignancy, 22 were men and 43 female. Among those with a malignancy 1 had definite MID, 9 probable MID, and 55 possible MID. The most common of the malignancies was breast cancer, followed by dermatological, gynecological, and gastrointestinal malignancies. The most frequent of the benign tumours was lipoma, followed by pituitary adenoma, meningeomas, carcinoids, and suprarenal adenomas. Compared to the general population, the prevalence of malignancies and of benign tumours was markedly increased. The female preponderance was explained by the frequent maternal inheritance of MIDs.

Conclusions: Adult patients with a MID, particularly females, carry an increased risk to develop a malignancy or a benign tumour. Since malignancy is an important determinant for their outcome, these patients should be more accurately screened for neoplasms, not to overlook the point, at which an effective treatment can no longer be provided.

Disclosure: Nothing to disclose

EP2151

Quantitative grip force assessment of muscular weakness in myasthenia gravis

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Introduction: Muscular weakness in myasthenia gravis (MG) is commonly assessed using Quantitative Myasthenia Gravis Score (QMG). More objective and quantitative measures of muscular weakness may complement the use of clinical scales and might detect subclinical affection of muscles. We hypothesized that muscular weakness in patients with MG can be objectively quantified with non-invasive grip force tasks and that pathological findings correlate with disease severity as measured by the QMG.

Methods: This was a prospective study investigating patients with confirmed diagnosis of MG. All data was compared to healthy controls (HC). Subjects and HC were asked to lift a device (250 and 500g) equipped with an electromagnetic sensor that measured three-dimensional changes in position and orientation. These were used to calculate position index coefficient (PI-C) and orientation index (OI-C) as measures for involuntary movements due to muscular weakness.

Results: 40 patients with MG (55.7 years, 42.5% female, mean QMG 7.2) were included. PI-C and OI-C were significantly increased in MG patients for the 500g device in the non-dominant hand. Subgroup analysis showed that patients with ocular myasthenia gravis (OMG) showed significantly higher values for the PI-C and OI-C in the non-dominant hand compared to HC. No correlation between QMG and grip force performance was found.

Conclusion: Quantitative Grip Force Assessment may be a useful objective tool for measuring muscular weakness in MG and seems to detect subclinical generalized muscle weakness in patients with OMG. Used as endpoint, it might increase the sensitivity and power of future clinical trials.

Disclosure: Nothing to disclose
Early diagnosis and early treatment in LOPD: when asymptomatic patients should be treated


Pompe disease is a lysosomal disorder caused by GAA deficiency. Late Onset Pompe Disease (LOPD) is characterized by progressive muscle weakness and/or respiratory failure but, sometimes, only by an asymptomatic hyperCKemia. It has been suggested that an early diagnosis is fundamental for a timely ERT start to maximize its efficacy. According to the current guidelines, ERT is recommended for patients clinically defined or in presymptomatic patients with detectable muscle weakness or reduction in respiratory parameters on clinical examination.

Objective: To discuss about current treatment guidelines for LOPD, focusing on early diagnosis.

In a recent high risk population study (LOPED study), involving 17 Neuromuscular Italian Centers, we were able to diagnose 17 new LOPD patients out of 1051 patient with suspected neuromuscular disorders. Among those patients, 35% manifested with asymptomatic hyperCKemia, 59% with hyperCKemia and limb girdle muscle weakness (LGMW) and 6% only with LGMW. The median time from onset of symptoms to diagnosis was 7.7 years. ERT has been initiated in 11 patients: 8 out of the 11 showed LGMW with hyperckemia whereas other 3 evidenced hyperCKemia without clinical symptoms but muscle morphology showed severe muscle damage and muscle MRI in proximal muscles revealed an sclero-adipose substitution.

Our study demonstrated that 35% of patients apparently with asymptomatic hyperCKemia showed a combination of clinical, morphological and neuroradiological data that suggesting to start ERT early. This study suggests that current treatment guidelines should be carefully updated.

Disclosure: Nothing to disclose
EP2154

Evaluation of the trail mediated apoptotic pathway in myasthenia gravis patients with thymic abnormalities

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Introduction: Myasthenia Gravis (MG) is accompanied by follicular thymic hyperplasia in 40-70% of cases, and by thymoma in 10-15%. TRAIL is a TNF family member, with a claimed role in negative selection of thymocytes in thymus. Yet the results are controversial. Alterations in the expression levels of TRAIL ligand and receptors and their inhibitors is significant in terms of evaluating different roles exerted by TRAIL, including apoptosis, in various tissues.

Methods: We investigated expression levels of TRAIL apoptotic ligand and receptors, and the antiapoptotic NFkB molecule immunohistochemically, in 22 MG patients with thymoma, 5 MG patients with thymic hyperplasia, and in 10 normal thymic tissue samples. Apoptotic cell counts were detected by TUNEL.

Results: Expression levels of DR4 and DR5 death receptors, and DcR2 decoy receptor were significantly higher in thymoma, while DR5 was increased in thymic hyperplasia compared to normal thymus. Furthermore, no detectable levels of active NFkB was evident in normal or abnormal thymus. Apoptotic cell count in normal thymus correlated with TRAIL expression.

Conclusions: Our results are compatible with an active apoptotic pathway for TRAIL in normal thymus and thymic hyperplasia, in terms of low decoy receptor levels, and absence of NFkB activation. Decoy receptors may be increased in thymoma as a protection from infiltration. Correlation of apoptotic cell count in normal thymus with TRAIL levels may suggest a possible role for TRAIL in thymic atrophy. These results, when combined with functional and in vivo tests, will be informative on applicability of a possible “TRAIL-mediated medical thymectomy”.

Disclosure: Nothing to disclose

EP2155

Myotonic dystrophy type 1 with myasthenia gravis: is this a by chance association?

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Introduction: Myotonic dystrophy type1(DM1) is an inherited muscle disease characterized by muscle weakness, myotonia and, within the multisystem involvement, the occurrence of immunological disorders. We describe three unrelated DM1 cases in which DM1 was associated with myasthenia gravis.

Case descriptions:
First case: 60-years-old male with diagnosis of DM1 since the age of 30 years. At the age of 56 he presented a rapid decline of clinical conditions, in particular in worsening of respiratory insufficiency, severe fatiguability in daily activities and appearance of left eyelid ptosis. Acetylcholine receptor antidody serum titre resulted positive and thoracic CT detected the presence of thymoma, confirming the diagnosis of thymomatous myasthenia.

Second case: 71-years-old male, affected by a mild form of DM1, complaining for some months of a sub-acute onset of dropped head and respiratory difficulties. Further investigations confirmed the diagnosis of myasthenia gravis associated with thymoma. In both cases the thymoma was removed surgically, with subsequent improvement of clinical conditions.

Third case: 33-years-old male coming to our attention for two-year history of handgrip myotonia and progressive nasal voice and presence, in his family pedigree, of an affected sister with occurrence of an otherwise typical ocular myasthenia with increased levels of acetylcholine receptor antibodies.

Conclusions: The rare but not exceptional occurrence of myasthenia gravis in DM1 is to be considered to correctly manage these at some extent clinically overlapping disorders. It remains to be elucidated which pathogenic mechanisms underlie possible autoimmune comorbidities in DM1 in the frame of a syndromic appearance or just a coincidence.

Disclosure: Nothing to disclose