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Neuromuscular Disorders

Myelopathic-tropical spastic paraparesis caused by HTLV-1: I

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MYELOPATHIC-TROPICAL SPASTIC PARAPARESIS CAUSED BY HTLV-1:
Introduction: Myelopathic-Tropical spastic paraparesis caused by HTLV-1 is a neurological disease that causes a chronic and progressive inflammatory process of the spinal cord, progressing to demyelination in patients with HTLV-1s The neurological condition is characterized by spastic paraparesis with extensive involvement of the pyramidal tracts, causing motor and sensory impairment, bladder and bowel sphincter disturbances, and erectile dysfunction in men.
Objective: Report a case of tropical spastic paraparesis caused by HTLV-I infection.
Report of case: Female patient of 39 years old, diagnosed with HTLV-I, infected by blood transfusion, having muscular weakness as initial clinical symptom 12 years ago, and worsened progressively in recent years, associated with frequent falls, and 05 years ago stopped walking, requiring a wheelchair. Presents today recurrent urinary infections as a result of urinary incontinence and use of diapers for 3 year. At the neurological exam, showed maximum force in all the segments of upper limbs, while in the lower limbs presented force level 2 in the left and right proximal segments and in the left distal segment; the right distal segment presented level 3. Regarding to reflexes, the bicipital, tricipital and estilorradial presented hyperactives, having positive Hoffmann. Patellar and aquileo, also showed to be hyperactives, with Babinski and positive clonus. The tactile and vibratory sensitivity were normal in the upper and lower limbs. Muscle spasticity using the Ashworth scale was grade 3 for the adductor and quadriceps femoris bilaterally, and grade 04for sural triceps bilaterally. Was made the relative quantification of gene expression of Th1 cytokines (IFN and TNF) that were greatly expressed, Th2 (IL-4) that showed reduced by antagonism with Th1 and a profile of regulatory T cells (IL-10 and TGF), where IL-10 was very low, but the TGF was high due to its function of neural repair.
Discussion: The motor disability of patients with HAM/TSP and low back pain significantly interferes with daily activities and can lead to functional limitations. Muscle strengthening is critical to clinical improvement. The inflammatory response triggered by IFN is probably involved in the progression and clinical outcome of the disease in infected individuals, but not in relation to IL-4. Studies show that IFN is closely related to the development of HAM/TSP.

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Neuromuscular Disorders

Clinical and genetic aspects of late-onset pompe disease in northeast Brazil (State of Ceará)

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Background: Late-onset Pompe disease (LOPD) is characterized by a slowly progressive myopathy due to deficiency of α-glucosidase.
Objective: To report the clinical, electrodiagnostic and genetic findings of 4 patients with LOPD from Northeast Brazil.
Material and Methods: We reviewed the medical, genetic and electrodiagnostic findings of 4 patients with LOPD seen at Hospital Universitário Walter Cantídio (2 were diagnosed at another institution and referred for evaluation of pulmonary status).
Results: Mean age of diagnosis was 40.3 ± 11.4 years (SEM), 3 men. All 4 had heterozygous mutations: 1.c.-32-13 T > G(5´UTR) & c.2501_2502delCAp.T834Rfs*49 (exon 18); 2. c.32-13 T > G & c.525delT p.E176RFs*45 (exon 2); 3 and 4. c-32-13 T > G (intron 1) & deletion on exon 18 (brother and sister). No patient was wheelchair-bound. Two patients (brother and sister) had classic limb-girdle phenotype and onset around 19-20 years. Two older patients had very atypical presentations. One had exercise-induced dyspnea since young-adulthood but was only diagnosed after respiratory failure and pneumonia. The other had a limb-girdle weakness pattern since age 40 and developed progressive dyspnea since age 50. Electrodiagnostic testing revealed neuropathy and myopathy in 2 and myopathy with paravertebral myotonic discharges in 2. One patient was initially diagnosed with critical care neuromyopathy with phrenic nerve paralysis. All 4 patients were treated with Myozyme (alglucosidade α) for at least 6 months, with significant clinical improvement.
Conclusion: The spectrum of LOPD is diverse and clinical course significantly improved by enzyme replacement. Recent availability of genetic testing has greatly improved our ability to establish diagnosis of LOPD in the state of Ceará, Brazil.

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Neuromuscular Disorders

Hypokalemic paralysis and crohn's disease (cd): report of two cases

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Background: Hypokalemic Paralysis (HP) is a rare condition that can be acquired or due to an inherited channelopathy.
Objective: To describe the clinical, electrodiagnostic and neurological findings of two patients from a cohort of Brazilian Inflammatory Bowel Disease (IBD) patients who presented with hypokalemic paralysis.
Methods: Patients underwent neurological evaluation, including detailed neurological exam and electrodiagnostic testing. The study was approved by the local IRB.
Results:

Patient 1: A 18 year-old man with a 2-year history of CD presented with acute generalized muscle weakness, dysarthria and limb paresthesias. Initial tests revealed hypokalemia (2.8 mEq/L), hypocalcaemia and hypomagnesaemia. Hypokalemic paralysis was diagnosed. Replacement of electrolytes was performed, with full recovery of muscle strength. Exams showed normal urinary electrolytes and preserved renal function. Patient had had major weight loss in the prior months and important generalized muscle atrophy. EMG revealed generalized axonal sensorimotor neuropathy.
Patient 2: A 30 year-old female with 10 years of CD presented with acute generalized paralysis and hyporreflexia. Initial evaluation showed hypokalemia (1.5 mEq/L), hypocalcaemia and hyponatremia. After

electrolytes replacement, the paralysis was reversed. Further investigation showed normal urinary electrolytes and preserved renal function. She subsequently had seizure episodes, several bouts of CD relapse, associated with fistulae as well as additional episodes of weakness also associated with lower limbs paresis and edema. Clinical improvement happened after the stabilization of the CD relapse. EMG revealed right peroneal neuropathy at the fibular head.

Conclusions: HP may be present in patients with CD, secondary to fecal/diarrheal potassium loss and bowel inflammation.

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Neuromuscular Disorders

Pompe disease in a brazilian city: the prevalence of a rare disease in a symptomatic population sample

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Background: Pompe disease, also known as glycogen type II deposit disease, happens because of a glycogen lysosomal deposit due to the acid-alfa-glucosidase enzyme deficit. It is a rare metabolic myopathy with a broad range of clinical presentations, which includes weakness and hypotonia. The diagnosis is made by the measurement of the enzyme activity in a dry drop of blood (DBS) and confirmed by the measurement in another tissue.
Objective: To evaluate the prevalence of Pompe disease in a population with suggestive symptoms in the city of Brasilia-DF, Brazil.
Patient and Methods: Patients with suggestive symptoms or electromyography pattern of myopathy were elected to perform the DBS test. If the result was positive, it was confirmed with the measurement of the enzyme activity in a fibroblast culture (FCM) to assure the diagnosis.
Results: Between 2011 and 2015, 87 patients were submitted to DBS test, 5 of them had a positive result. However, when submitted to the second test only 3 of them had a confirmed diagnosis, the other two, heterozygotes, did not had enough criteria for treatment. Considering that the FCM was the gold standard, in this symptomatic sample, 60% of the DBS positive test were true positive.
Conclusion: The prevalence of Pompe disease in our population was of 3, 45%. It was in accordance with two other studies realized in neuromuscular centers, with a prevalence of 4 and 5, 3%. This number is much bigger than the general prevalence of 1: 40.000, because it was calculated in a symptomatic population.

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Neuromuscular Disorders

The potential role of cell surface complement regulators and circulating cd4+ cd25+ t-cells in the development of autoimmune myasthenia gravis

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Background: Regulatory T cells (Treg cells) and regulators of complement activity (RCA) involving CD55 and CD59 play an important role in the prevention of autoimmune disease, however their role in the pathogenesis of autoimmune myasthenia gravis (MG) remains unclear.
Objective: Our study aimed to assess the frequency of peripheral blood $CD4^+CD25^+$ T-reg cells and $CD4^+$ T-helper cells and the RBCs level of expression of CD55 and CD59 in MG patients.
Patients and Methods: Fourteen patients with MG and ten age matched healthy controls participated in the study. We assessed the percentage of peripheral $CD4^+CD25^+$ T-reg cells and $CD4^+$ T-helper cells and the level of expression of CD55 and CD59 on RBCs in the peripheral blood of patients and controls.
Results: There was a statistically significant decrease in the percentage of peripheral blood $CD4^+CD25^+$ Treg cells and $CD4^+CD25^+$ T-reg/$CD4^+$ T-helper cell ratio in the MG patients group ($\boldsymbol{P}$ value < 0.05). Moreover, the level of expression of CD55, CD59 and dual expression of CD55/CD59 on RBCs were statistically significantly lower in MG patients than those of healthy controls ($\boldsymbol{P} < 0.001$). However; regression analysis revealed that there was no significant correlation between all the measured parameters and disease duration or staging.
Conclusion: We can conclude that functional defects in the Treg cells and RCA may play a role in the pathogenesis of autoimmune MG and their functional modulation may represent an alternative therapeutic strategy for MG treatment.

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Neuromuscular Disorders

Electrochemical skin conductance in patients with carpal tunnel syndrome

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Background: Studies suggest that C-fibers are affected in entrapment neuropathies. In carpal tunnel syndrome (CTS) there is reduction of intraepidermal nerve fiber density.
Objective: To assess small fiber function using electrochemical skin conductance (ESC) in patients with CTS.
Patients and Methods: 10 patients with electrophysiologically confirmed CTS and 20 control subjects. Sweat gland function in the palms was assessed non-invasively with SUDOSCAN (Impeto Medical, Paris France) device. Nerve conductions were performed according to AANEM. Stevens´s scale (three grades) was used for grading of severity of nerve conduction abnormalities. The Boston CTS questionnaire symptoms domain (BCTQ-symptoms) was used to assess subjective symptoms.
Results: CTS patients (4 with hypothyroidism, 1 with diabetes), BCTQ-T was 23.1 ± 10.3. Seven CTS were bilateral. Severity: mild = 4, moderate = 4, severe = 2. In CTS patients mean ESC in hands was $68.2 \pm 9.6\mu S$ and in controls was $68 \pm 15.8\ \mu S$. In bilateral CTS patients, mean ESC in hands was $69.6 \pm 2.1\ \mu S$. In unilateral CTS mean ESC in the affected hand was $62.3 \pm 12\ \mu S$ and the non affected hand was $69 \pm 11.3\ \mu S$. Asymmetry in unilateral CTS was 9 ± 2.8 % and in controls was $3.5 \pm 2.9\%$.
Conclusions: Assessment of electrochemical skin conductance could be useful to detect palm sweat gland dysfunction in a subgroup of CTS patients.

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