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Neuromuscular Disorders 2

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Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) in a patient with Hashimoto's Disease (HD) and possible minimal renal lesion

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Background: The precise mechanism of CIDP is unknown, and it is believed to have an inflammatory background, suggesting an auto-immune physiopathology.
Objective: A case report of CIDP in a patient with kidney and thyroid damage, both caused by auto-immune mechanisms.
Patients and methods: A 23 year-old woman presented symmetrical, distal and progressive weakness and loss of vibratory sensibility in upper and lower members two months ago. She related dysuria and foamy urine, she had no other complaints. Neurological examination revealed peripheral facial paresis, proximal and distal weakness in upper and lower limbs, global arreflexia, absence of vibratory sensibility in distal limbs, and gait moderately ataxic. We have obtained Institutional Review Board approval, as necessary.
Results: The following additional tests were performed: (1) laboratory tests: TSH > 100 mUI/L, T4L < 0.4 ng/dL, IgG auto-antibodies positive, microalbuminuria and hematuria; (2) neurophysiological study: chronic sensorimotor, demyelinating polyneuropathy with secondary axonal features; (3) CSF had normal cellularity with increased protein (428 mg/dL); (4) thickening and contrast enhancement of motor and sensory roots on MRI; (5) renal biopsy: minimum injury due to moderate IgG positive diffuse, global and agranular deposits in capillaries and intra-glomerular mesangial cells.
Conclusion: CIDP and Hashimoto's Disease and nephropathy are not commonly found together; this association could reflect a dysregulation of the immune system and indicates that some antibodies may have affinity to bind to some particular antigen in different places, such as peripheral nervous system, thyroid and kidney.

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Neuromuscular Disorders 2
Autophagy dysregulation in amyotrophic lateral sclerosis

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Background: Amyotrophic lateral sclerosis (ALS) is a devastating neurodegenerative disease. Although the causes of the disease are not yet undefined, it is believed that a toxic gain of function resulting from abnormal protein aggregation is one of the mechanisms contributing to this disease.
Objective: The objective of this study is to investigate if ALS has autophagy alteration and if autophagy modifiers can be used to treat ALS.
Materials and methods: We used SOD1-G93A mutant ALS mouse model to examine the autophagy related molecules in the spinal cord and other central nervous system, and applied several autophagy modifiers to test if autophagy can be potential therapeutic target for ALS.
Results: We found that the macroautophagy is specifically activated in the motor neurons of the ALS mice at early stage, which included an accumulation of LC3-II and p62, an increase of autophagic vacuoles (AVs), aggregation of ubiquitin and SOD1 proteins associated with LC3-II deposition in motor neurons. In addition, we demonstrated that autophagy enhancer rapamycin causes accumulation of AVs, but fails to reduce SOD1 aggregates, indicating the abnormal autophagic flux in the ALS model. Rapamycin treatment results in severe mitochondrial impairment, higher Bax levels and greater caspase-3 activation. On the contrast, mTOR independent autophagic enhancer trehalose prolongs the motor neuron survival and ameliorates autophagic flux defect in the ALS model.
Conclusion: These studies indicate that defect in autophagic flux is a critical pathogenetic mechanism of ALS and mTOR independent autophagic enhancer may have a potential for the treatment of ALS.

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Amyotrophic lateral sclerosis in Moroccan population: environmental and clinical aspects

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Introduction: Amyotrophic lateral sclerosis (ALS) is the most common variety of motor neuron disease. Its etiology remains unknown, but several studies incriminate the environmental factors in its genesis.

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Aim of the study: is to describe the epidemiological, clinical and environmental aspects of ALS in Moroccan population.
Materials and methods: 60 ALS patients were recruited over a period of 5 years from January 2008 to September 2012 in the Neurology B and Neurogenetics Department and also Military Neurology Department of Rabat. Patients were evaluated by detailed record of exploitation. Statistical analysis was performed using SPSS 13.0.
Results: The average age of the population was 52 ± 11.2 years with a sex ratio of M/F = 1.5. The average age of onset was 50 ± 11.7 years. In the group of patients exposed to toxins, a significantly higher proportion of solvent exposure was found ($p = 0.02$).However there was no significant association with exposure to heavy metals, pesticides or with toxic and eating habits. ALS is frequent in the west region of Morocco ($p = 0.03$).
Conclusion: The positive association between exposure of solvents and ALS found in our population has been reported in the literature. The frequency of the ALS in the west region suggests may be environmental or genetic origin. These results are preliminary and require a multicenter study to have more data and better highlight the environmental characteristics of ALS in the Moroccan population.
Keywords: Amyotrophic lateral sclerosis, environmental factors, pathogenesis.

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Diagnostics of the heterotopic ossification in patients with spinal cord injury

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Aim: The aim of the study was to define the peculiarities of bone remodeling and identify specific parameters to development to heterotopic ossification.
Methods: Markers of bone formation (Osteocalcin, serum type 1 procollagen (N-terminal) (tP1NP)) and bone resorption (serum collagen type 1 cross-linked C-telopeptide (β-CTx)) were determined by the electrochemiluminescence immunoassay "ECLIA" for Elecsys user cobas immunoassay analyzer.

In the study were included 23 patients with spinal cord injury — first group (average age 26.8 ± 3.9, duration of spinal cord injury from 3 to 12 months) and 23 healthy people's appropriate age and gender (average age 30.6 ± 6.0, years). In the first group included 11 patients with spinal cord injury with the presence of heterotopic ossification — subgroup I and 12 patients with spinal cord injury without heterotopic ossification — subgroup II.
Results: The results of examination showed that patients of first group had significantly higher bone markers than control group: P1NP (256.7 ± 48.2 ng/ml vs 49.3 ± 5.1 ng/ml, $p < 0.001$), serum β-CTx (1.47 ± 0.23 ng/ml vs 0.45 ± 0.04 ng/ml, $p < 0.0001$), osteocalcin (52.2 ± 9.8 ng/ml vs 24.9 ± 2.08 ng/ml, $p < 0.001$). Levels of bone remodeling markers in patients with HO were significantly higher in comparison with patients without HO: P1NP (404.9 ± 84.9 ng/ml vs 133.2 ± 15.7 ng/ml, $p < 0.001$), serum β-CTx (1.75 ± 0.23 ng/ml vs 0.28 ± 0.14 ng/ml, $p < 0.0001$), osteocalcin (87.1 ± 18.9 ng/ml vs 29.4 ± 3.7 ng/ml, $p < 0.001$).
Conclusion: The bone formation and bone resorption markers in patient of first group were significantly higher than in healthy individuals of appropriate age. The rate of bone turnover markers in patient with HO was considerably higher than in patients without HO, and the process of formation dominated over the resorption in patient with HO.

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Bone mineral density in women with Parkinson's disease

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Aim: The aim of the research is to define the bone mineral density in patients, with Parkinson's disease.
Methods: We examined 12 women with Parkinson's disease and 12 healthy women of appropriate age (average age: -63.6 ± 6.25 vs 62.2 ± 6.83 years, $p = 0.5$). The duration of Parkinson's disease was at list 5 years. All patients received levodopa.
Results: BMD of women with Parkinson's disease was significantly lower compared with BMD of women of control group on the level of total body (T-score = −1.86 ± 1.32 vs. −0.71 ± 1.48, $p < 0.05$, Z-score = −0.35 ± 0.93 vs. 0.51 ± 1.05, $p < 0.05$), lumbar spine (T-score = −1.56 ± 1.22 vs. 0.10 ± 1.63, $p < 0.05$, Z-score = −0.66 ± 0.87 vs. 0.72 ± 1.53, $p < 0.05$) and at the distal forearm (T-score = −1.87 ± 1.32 vs. 0.71 ± 1.47, $p < 0.05$, Z-score = −0.51 ± 1.05 vs. 0.38 ± 1.22, $p < 0.05$). Hip BMD was not different from control group. It is important to note that one woman with Parkinson's disease had two hip endoprostheses after femoral neck fractures.
Conclusion. BMD in women with Parkinson's disease is significantly lower than in healthy women of the same age.

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Neuropathic pain component under musculoskeletal diseases

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Introduction: Neuropathic pain from the musculoskeletal diseases has recently been the highlight of numerous studies. The aim of this study was to estimate the structure of pain syndrome and reveal the presence of neuropathic pain component in patients suffering from the musculoskeletal diseases.
Material and methods: We examined 73 patients aged 45–85 years (average age 68.1 ± 1.2 years). Patients were divided into 3 groups: A — patients with osteoporosis ($n = 30$), B — patients with low back pain ($n = 23$), and C — patients with osteoarthritis of knee joints ($n = 22$). To assess the NP component, we used the painDETECT, LANSS, and DN4 questionnaires. For statistical analysis of results ANOVA, correlation and regression analysis were applied.
Results: Regression analysis showed correlation between the questionnaires: LANSS and painDETECT ($r = 0.76$, $p = 0.000001$), DN4 and painDETECT ($r = 0.8$, $p = 0.000001$). 66.7% of patients with osteoporosis examined by the painDETECT were unlikely to have the NP component, 18.5% might possibly, 14.8% — probably had one. By the LANSS scale: 23.3% probably had NP. By the DN4 scale: 36.7% probably had NP. 63.2% of patients with low back pain examined by the painDETECT were unlikely to have NP, 26.3% might possibly,

10.5% — probably c. By the LANSS scale: 29.4% probably had NP. By the DN4 scale: 43.5% probably had NP. 68.1% of patients with osteoarthritis of knee joints examined by the painDETECT were unlikely to have the NP component, 22.8% might possibly, 9.1% — probably had one. By the LANSS scale: 31.8% probably had NP. By the DN4 scale: 40.9% probably had NP.
Conclusion: In patients with musculoskeletal diseases the pain syndrome may have the NP features. Identification of these patients would promote a treatment strategy targeted at the NP.

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Beta sarcoglycanopathy caused by a novel mutation in the SGCB gene in a Palestinian family

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Background: The autosomal recessive limb girdle muscular dystrophies (LGMD2) are a genetically heterogeneous group of myopathies.
Objective: To reach final diagnosis in a child with Duchenne-like muscular dystrophy.
Patients and methods: An 8-year-old male presented with a history of motor delay and a highly elevated serum creatine kinase (50,000 U/l). A liver sonogram was normal. Parents are first cousins and of Palestinian descent.
Results: He showed a protruding abdomen and lumbar lordosis with mild calf hypertrophy, positive Gowers' sign and weak neck flexors. Deep tendon reflexes were hypoactive but symmetrical. His gait was limited and wobbly. A cardiovascular evaluation, including EKG and Echocardiogram, was normal. An MRI of the brain was normal. The muscle biopsy showed dystrophic changes and complete absence of the four sarcoglycans by immunohistochemistry. Molecular analysis (Prevention Genetics, Marshfield, WI) revealed a novel homozygous missense mutation in exon 5 of the beta sarcoglycan gene defined as c.630C > G resulting in a p.Ser210Arg amino acid substitution. Both parents were found to be heterozygous carriers of the same mutation. On follow-up examinations, his muscle strength reached a plateau at age 6 at which time treatment with Deflazacort was considered and started after cardiologic and pulmonary clearance. Side effects have been minimal. At last follow up he had become wheelchair dependent.
Conclusions: We report a child with LGMD2E caused by a novel missense mutation of the beta sarcoglycan gene.

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