In one family, previously linked to the CMT4B1 locus no mutations in the MTMR2 and GDAP1 genes have been identified. A follow up analysis of this family revealed a marked discrepancy between clinical course of disease between affected male and female (X dominant trait of inheritance). In two CMT families X-linked recessive trait of inheritance was detected. Surprisingly, we have identified numerous recessive CMT families in population of Poland which is characterized by a low ratio of the consanguineous marriages.

O-6

Markers of degeneration and regeneration in blood of Duchenne muscular dystrophy

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The microenvironment of dystrophic muscles is associated with variation in levels of markers of degeneration and regeneration. Markers of degeneration can be measured in terms of apoptosis and apoptosis gene expression, while markers of regeneration can be measured in terms of cytokine and growth factors. The present study is an attempt to demonstrate the extent of degeneration and regeneration in DMD pathogenesis. The levels of Fas and FasL and Bax/Bcl-2 and plasma of DNA fragmentation as markers of apoptosis were measured. Also Cytokine tumor necrosis factor alfa (TNF, TNF-α), as well as the growth factors basic fibroblast growth factor (bFGF) and vascular endothelial growth factor (VEGF) were measured as markers of regeneration. Results indicate that Fas/FasL and Bax/Bcl-2 are involved in muscle atrophy and degeneration in DMD patients, while the regeneration process does not cope with the degeneration.

O-7

Examining the lower orbicularis oculi muscle reduces the number of false negative results with respect to myasthenia gravis diagnosis: a Stimulated Single fiber EMG study

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Purpose. Stimulated Single-fiber EMG (SSFEMG) is a sensitive electrophysiological method for the determination of neuromuscular dysfunction. Normative values for SSFEMG have been reported for the orbicularis oculi (OOc) with no distinction made between the upper and lower OOc. The purpose of this study is to demonstrate that the upper and lower OOc differ in how they behave during dysfunction.

Methods. Patients in whom myasthenia gravis (MG) was suspected were included in this study. A single fiber potential was considered abnormal if it had a mean consecutive difference (MCD) value greater than 30 μsec, and the study was considered abnormal if more than 10% of the sampled potentials were abnormal and/or the mean MCD value in a sample of 20 single fiber potentials was greater than 20 μsec. The potentials had to have amplitude of at least 200 μV, with a rise time less than 200 μsec. MCD values greater than 100 μsec were discarded to avoid contamination with blink reflex.

Results. Seventy-six patients were entered prospectively into this study. Twenty-six patients completed the study and obtained a final diagnosis. Twenty-one patients had abnormal SSFEMG results. The lower OOc was found more likely to be abnormal based on the number of abnormal single fiber potentials (27.6% and 20.7% potentials from the lower and upper OOc respectively). Both upper and lower OOc were abnormal in 13 patients. The lower OOc was solely abnormal in 6 patients, of which 4 (66.7%) had a diagnosis of MG. The upper OOc was solely abnormal in 2 patients, neither of which had a diagnosis of MG.

Conclusions. The lower OOc is an important muscle to examine, not only to rule out MG, but also to prevent the occurrence of false negative results. Examination of the lower OOc increases the sensitivity of SSFEMG, without a change in specificity.

O-8

Investigating valproate as a treatment for McArdle’s disease

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McArdle’s disease is an inherited myopathy caused by the absence of the muscle isoform of phosphorylase. There is no treatment. In normal muscle the genes for the brain and liver isoforms are active in utero, in neonates and in regenerating mature muscle. This activity was found in mature muscle in McArdle’s disease sheep following the injection of vectors carrying cDNA for LAC Z or myophosphorylase or the injection of notexin or val-