Comprehensive neuro-rehabilitation of the children with mental problems improved their clinical and emotional background, memory and cognitive functions. EEG and NEC examination marked the trends in recovery of inter-hemispheric connections and in normalization of the bioelectrical activity in the brain.

O-3
Clinical features and outcome measures during 1 year enzyme replacement therapy in late onset GSD II patients

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The objective to identify appropriate outcome measures to use in clinical trials or observational studies. Natural history data of patients with the heterogeneous juvenile/adult form of glycogen storage disease type II (GSDII) are useful to evaluate enzyme replacement therapy (ERT).

We evaluated several outcome measures before and during ERT, such as a series of timed and graded functional tests i.e. Gait, Stairs, Gowers, Chair (GSGC) score, the Six-Minute Walk Test (6MWT), Forced Vital Capacity.

During an observational study we monitored a series of 32 patients using these outcome measures at 1 year after ERT and observed a partial but significant improvement. A significant decrease was observed in Gait time and time to raise from the chair and total GSGC score. A gain of function was observed in few cases.

Important, crucial topics seem to be: the use of different functional parameters in determining the efficacy of ERT, since not all juvenile/adult patients respond similarly, and, reliable outcomes of treatment.

O-4
The Italian Network for Laminopathies

G. Lattanzi and the Italian Network for Laminopathies[1]

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Laminopathies are a group of genetic diseases caused by mutations in the nuclear protein lamin A/C or in related proteins. A group of laminopathies targets specific tissues – tissue-specific laminopathies –, other laminopathies affect multiple tissues – systemic laminopathies.

The Italian Network for Laminopathies is a group of Clinical and Research Centers performing clinical and molecular diagnosis or biomedical research in the field of laminopathies. Aims of the Italian Network for laminopathies are: to connect Italian Centers involved in diagnosis and biomedical research of laminopathies; provide information on the clinical features of Laminopathies to family doctors, specialists and patients; provide the contact information of specialists involved in the diagnosis and research of laminopathies; provide updated information on biomedical research in the field of laminopathies; organize an Italian Registry for Laminopathies containing clinical and biological data; establish a bank of biological material; report news on relevant events and meetings; report on funding opportunities in the field of Laminopathies research.

The Italian Network for Laminopathies website provides updated information of the Network and of Laminopathies-related events.

O-5
Novel HLA II associations in myasthenia gravis

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Myasthenia gravis (MG) is a heterogeneous disorder encompassing several subtypes. In our patients population, the most numerous and homogeneous group is represented by patients with a late onset, seropositive, non-thymomatous MG (LO-Ab+noTh). We have analyzed the frequency of HLA-A, -B, - DRB1, -DQA1 and -DQB1 alleles in 81 unselected Italian MG patients and in 100 healthy controls. HLA allele frequencies were estimated by direct count and compared in a 2×2 contingency table analysis using the Fisher exact test; p values were corrected for multiple comparisons according to the Bonferroni method (pc). When the entire MG population was considered, no association was found between the occurrence of the disease and the presence of any of the alleles analyzed. However, when the LO-Ab+noTh patients were compared to controls, a positive association of the DRB1*16 and the DQB1*0502 alleles was observed (pc 0,0211 and 0,00768 respectively). On the contrary, the same association was not present in patients with either early-onset, seropositive, non-thymomatous MG, or with thymomatous MG. The association of DRB1*16 and DQB1*0502 with MG has been previously reported in Italian patients with different features, but not in LO-Ab+noTh MG. According to these results, DRB1*16 and DQB1*05:02 might be considered as genetic markers of LO-Ab+noTh MG, while their role as predisposing genetic factors for MG should be clarified by further investigations.