Appendix Posters

NEUROGENETICA *(chapter pag.59)*

Late-onset Huntington's disease : description of a large family.

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Huntington's disease (HD) is hinerited as an autosomal dominant trait; begin usually between 35 and 50y; patients die 10 to 20 y after onset. The genetic mutation has been identified as an expansion of CAG repeat sequence in IT15 gene on 4p 16.3.
We describe a large family (123 persons) with 4 members affected by late-onset HD (positive DNA analysis) and, respectively: 58, 64, 72, 73 years; other 4 cases of probable late-onset HD died aged over 75y old or later. In this family were also presents yuvenile cases and typical onset cases. The present observation indicates that senile chorea diagnosis might be done only after an accurate genealocic investigation and the systematic use of DNA analysis, considering the HD phenotypic variability.

EPILESSIA *(chapter pag.123)*

Ipsilateral cortico-cortical inhibition in patients with cryptogenic partial epilepsy.

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An altered balance between excitation and inhibition has been postulated as the neural basis of epileptic phenomena. Transcranial magnetic stimulation (TMS) can give information on such balance in the human motor cortex. We studied with TMS a group of eight patients affected by cryptogenic partial epilepsy (CPE) (mean age 30, S.D. ± 11.8), whose epileptic focus was located in the right hemisphere, and nine normal controls (mean age 28.7 S.D ± 5.7). TMS was delivered through a round coil centered at the vertex; motor evoked potentials (MEPs) were recorded from the right and left first dorsal interosseous muscle (FDI). We determined the following variables: (i) the threshold for evoking MEPs in the resting and active FDI; (ii) the cortical silent period; (iii) the ipsilateral cortico-cortical inhibition and facilitation curve (conditioning shock = 0.8x and test shock= 1.2x resting threshold). Either hemisphere was stimulated separately, and the results were subject to inter-side comparison. There was also a comparison between homologous hemispheres of patients and controls. In the patient group, the variables measured showed no difference between the right hemisphere, containing the epileptic focus, and the contralateral one. Comparing CPE patients with controls, we found that: (i) the resting motor threshold was increased in both the patient hemispheres; (ii) the active motor threshold was increased only in the hemisphere containing the focus; (iii) the cortical silent period was prolonged in the hemisphere contralateral to the focus; (iv) the ipsilateral cortico-cortical inhibition (for intervals 1 and 3 msec) was reduced in the patients, more evidently so for the hemisphere site of the focus.

NEUROFISIOLOGIA *(chapter pag.97)*

NEUROPHYSIOLOGICAL STUDY OF A EREDOATAXIA PATIENT WITH INCREASED REFLEXES.

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A patient, 64 years old, came to our observation for a progressive lower limb weakness, cerebellar ataxia, upper limb paresthesias and deep sensory loss.
His father and one of his brothers had gait disorders but they died when they were young. When he was 55 years old, had a ischemic heart disease so an aorta-coronaries by pass was made. The neurologic examination showed incoordination for all the limbs, increased of reflexes with heel bilateral clonus, right Babinski sign, deep sensory loss and a pyramidal ataxic gait. Contrast-enhanced CT demonstrated a small ischemic area in left external capsule but not cerebellar atrophy. MRI could not be made for the presence of metallic clips. Neurophysiologycal studies became very important in the diagnosis. Central and peripheral conduction times were increased during magnetic stimulation, as well as all sensory evoked potentials latencies. A sensory and motor neuropathy was demonstrated with electromyography. Genetic tests for SCA 1 and 2 were negative. The patient was affected by eredoataxia with increased tendon reflexes.