# Summary

Neurodegenerative disorders of childhood encompass a large number of heterogeneous diseases that result from specific genetic and biochemical defects, and a significant group of conditions of unknown cause. Most are inherited in an autosomal recessive fashion and show involvement of different parts of the central and sometimes peripheral nervous system with a variety of progressive deficits, especially dementia, epilepsy, blindness, ataxia and disorders of tone and reflexes ***(Brett and Lake, 1997).***

Although the outcome is invariable fatal, and current therapeutic attempts have been unsuccessful, it is important to make the correct diagnosis so that genetic counseling may be offered and prevention strategies can be implemented

Recent advances in the recording of somatosensory, brain stem auditory and visual evoked potentials provide relatively non-invasive techniques for the evaluation of the different afferent pathways from the periphery to the cerebral cortex ***(Chiappa et al., 1997).*** In addition, CT and MRI are of value in the differential diagnosis of neurodegenerative brain diseases ***(Vanhanen et al., 1994).***

The aim of this study is a trial to evaluate the importance of different modalities of evoked potential in the detection of degenerative brain disorders and to assess the benefits obtained when adding neurophysiological modalities to neuroimaging procedures in order to refine our knowledge about the pathophysiology of these diseases.

To reach the goal 45 patients were the subject of this study and 20 normal volunteers acted as a control matched with the same age and gender for the patients. The patients were 25 males and 20 females and their ages ranging from 6 months to 13 years.

The patients were selected from neuropediatric clinic at New Children Pediatric Hospital, Faculty of Medicine Cairo University.

All patients were chosen from those showing progressive loss of previously acquired motor and mental milestones or failure of achievement of normal milestone of development associated with positive family history. All patients were subjected to thorough clinical examination and neuroimaging studies [CT or/and MRI]. Some patients were subjected to specific investigations as detected by their clinical pictures in order to reach specific etiological diagnosis.

The patients were categorized into demyelinating, neuronal storage and miscellaneous groups based on clinical data, neuroimaging studies as well as specific laboratory diagnosis.

Demyelinating group comprising 30 patients (14 patients with metachromatic leukodystrophy (MLD), one with adrenoleukodystrophy (ALD), two with Canavan's disease, two with Alexander's disease, one with pelizaeus-merzbacher disease (PMD), three with glutathione synthetase deficiency, two with possible diagnosis of krabbe disease and five with undefined etiology).

Neuronal storage group comprising 9 patients [2 paitents with GM2 gangliosidoses, four with Niemann-Pick's disease and three with mucopolysaccharidoses].

Miscellaneous group comprising 6 patients [1 patient with Wilson's disease, one with mitochondial encephalomyopathy, one with Friedreich's ataxia, one with hereditary spastic paraplegia and two with unknown etiology but the clinical course was highly suggestive of neurodegenerative disease].

Multiple comparison study of multimodality evoked potentials [SEP, VEP, BAEP] was carried out in the 3 studied patients groups and the normal control group.

## *The results can be summarized as follow:*

### *SEPs*

- The patients suffering from demyelinating diseases were found to have a significantly delayed cortical responses in addition to prolonged central conduction time between cervical and cortical potentials. This suggests a conduction defect in the large fiber sensory system above the lower medulla and below the thalamus.

- Also we found a significant delay in Erb's potential. This can be simply a reflection of peripheral nerve involvement in the patients with metachromatic leucodystrophy (MLD) and Krabbe disease.

- SEP study in the patients with neuronal storage disorders showed the same abnormalities as in the patients suffering from demyelinating diseases. Delayed Erb's potential could be explained by the presence of peripheral neuropathy in some patients with Niemann-Pick disease.

- The patients with miscellaneous neurodegenerative disorders were found to have a significantly delayed cervical potential pointing to

involvement of the large fiber sensory system central to the brachial plexsus and below the lower medulla.

***VEP:***

The 3 studied patients groups were found to have a significantly abnormalities in the visual evoked responses. Either the response was significantly attenuated or had marked prolongation of the latency of P100. There were statistically significant increase of P100 latency in miscellaneous group as compared with either demyelinating or neuronal storage group.

***BAEP:***

Wave I which is generated by the activity essentially of the extra-medullary portion of the 8$^{th}$ neve was significantly delayed in the 3 studied patient groups.

Also there were statistically significant delay in absolute latency of wave III & V in patients with demyelinating and neuronal storage diseases. However in the present study we did not found any significant delay in the interpeak latencies.