

ANGELMAN SYNDROME: CHARACTERISTICS OF EPILEPSY, ELECTROENCEPHALOGRAPHIC ABNORMALITIES AND CORRELATION TO GENETIC MECHANISMS (ABSTRACT) *. **THESIS. SÃO PAULO, 2001.**

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Angelman syndrome is characterized by severe mental retardation, speech disorder, stereotyped jerky movements, peculiar behavioral profile and typical facial traits. Eighty to 90% of these patients present epilepsy and suggestive electroencephalographic patterns which are used as diagnostic criteria and become important when the phenotype is not suggestive enough, as in infants. The syndrome results from distinct genetic mechanisms [deletion (75-80%), paternal uniparental disomy (1-2%), imprinting center abnormality (2-3%) and UBE3A point mutation, found in ¼ of the negative genetic cases (20-25%)], which affect the maternal chromosome 15. The preservation of a larger genic contingent is probably related to a milder clinical phenotype as observed in patients without deletion making its recognition less evident.

The aim of this study was to analyze the correlation between distinct genetic mechanisms that determine Angelman syndrome and the severity of epilepsy and electroencephalographic abnormalities in a series of 26 consecutive patients with clinical and/or genetic diagnosis.

Patients were classified according to their genetic studies in cases with deletion (n=19), disomy (n=3) and negative genetic studies (n=4). Sixty-eight EEGs were studied for the characterization of the EEG patterns, background activity and the occurrence of electrographic seizures. Epilepsy was studied through clinical history, V-EEG and film revision and occurred in 84.6% (22/26) of these patients. Suggestive EEG patterns were found in 96.1% (25/26). Prevalence of epilepsy was sig-

nificantly higher in patients with deletion. Parameters indicating severity of epilepsy such as earlier onset age, higher occurrence of daily, disabling or multiple seizures, *status epilepticus* and febrile seizures presented a higher tendency of occurrence in patients with deletion. Patients with disomy did not present epilepsy and negative cases were less severe.

Suggestive electroencephalographic patterns were observed in the three groups. Delta pattern was the most frequent followed by posterior discharges. Theta pattern was observed only in patients with deletion. Prolonged, continuous or quasicontinuous bursts, as well as electrographic seizures and unspecific interictal epileptiform discharges, were predominantly observed in patients with deletion. Normal age-related background activity was more frequent in patients without chromosomal deletion.

We concluded that, although epilepsy has been observed in non-deletion patients, epilepsy was more severe in patients with deletion. This is probably due to a major involvement of genes, which decodify GABA receptors through this mechanism. In spite of the presence of epilepsy or of a less suggestive clinical phenotype and regardless the genetic mechanism involved, EEG could be used as a diagnostic criterion in patients in all groups.

KEY WORDS: Angelman syndrome, EEG, epilepsy, genetic mechanism, deletion, uniparental disomy.

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TEACHING INFANTILE NEUROLOGY IN BRAZIL: CURRENT ASPECTS, CHALLENGES AND PERSPECTIVES (ABSTRACT)*. **THESES. CAMPINAS, 2001.**

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This study intends to identify how many and which are the formation and training centers in Infantile Neurology in our country and its particularities. Besides, it tries to find out whether or not there are differences between the services which are *accredited* by the National Commission of Medical Residence and the services that are not accredited by this Commission.

Another purpose is to find out why the Institutions that have medical residence in Neurology and in Pediatrics do not have Infantile Neurology residence even though they have the necessary conditions to offer it.

It is also about the improvement course in Infantile Neurology for non doctor professional of the health area that is taught by the Universidade Estadual de