P-765 - MANAGEMENT OF CHILDREN WITH ANGELMAN SYNDROME

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Purpose: Angelman syndrome (AS) is a neurogenetic disorder caused by various 15q11-q13 abnormalities, characterized by severe mental retardation, speech delay, ataxia, happy disposition. In this paper we present our experience regarding the management of children with AS.
Material and methods: Our study included 9 children (5 boys and 4 girls) with AS, all with interstitial deletion of 15q11-13, with aged ranged between 6 months and 12 years. In all children we noted: clinical phenotype, epilepsy history (seizures onset, type of seizures, response to antiepileptic drugs), psychomotor development. All children have been followed up regarding epileptic seizures control and psychomotor development.
Results: All patients presented the typical clinical pictures. The most frequent seizures were partial seizures, followed by atonic seizures and atypical absences. The EEG exhibited the characteristic AS pattern. Valproate was preferentially used anticonvulsant, in some cases in association with clonazepam, lamotrigine or levetiracetam . Corticotherapy was used in four cases, with good results; also, improvement of EEG abnormalities and a slight psychomotor amelioration was noted. 8 children are seizures free in present. All children made physical therapy and cognitive stimulation.
Conclusions: In our patients, the treatment of epileptic seizures was effective. Valproate was most effective, but association of other antiepileptic drugs was necessary in many cases. Corticotherapy had good effect, not only in controlling seizures, but also in improvement of psychomotor delay and EEG abnormalities. Physical therapy and cognitive stimulation were useful tools for the improvement of psychomotor development.
Acknowledgments: CNCSIS 1203 project, PN II 42-130 project.

https://doi.org/10.1016/S0924-9338(12)74932-5 Published online by Cambridge University Press