

Efficacy and tolerability of antiepileptic drugs in 104 girls with Rett syndrome

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Approximately 30-80% of girls with Rett Syndrome suffer from epilepsy which represents one of the most severe problems clinicians have to deal with, especially when patients are 7-12 years old.

We included in this study 104 girls, aged 2-42 years: 89 had a mutation in MECP2, 5 in CDKL5, 2 in FOXP1, 8 unknown.

Epilepsy was present in 83 patients (79.8%). Mean age at onset was 4.1 years. 19.6% of patients presented with daily seizures, 21.6% weekly, 31.3% monthly, 27.4% sporadic.

Girls were divided into 5 groups according to age: 1-5, 5-10, 10-15, 15-20, >20 years.

Valproic acid (VPA) resulted as the most prescribed single therapy in young patients (<15 years), whereas carbamazepine (CBZ) was preferred by clinicians in older patients. The more frequently adopted association in the patients under 10 years and older than 15 was VPA with lamotrigine (LTG).

The group aged 10-15 years resulted the most difficult to treat, with a mean of 3 different antiepileptic drugs (AEDs) used. In this group clinicians adopted associations of AEDs, mostly including VPA.

12 patients (14.5%) were considered drug resistant.

VPA was reported as the most effective AED in younger girls (in 40% of patients aged 1-5 years, in 19% of girls aged 5-10 years), whereas CBZ reported the major efficacy in patients aged 15 years or older.

Adverse reactions did not differ from the expected; agitation and weight loss were the most frequently reported. In our sample, LTG was the less tolerated AED.

We did not find correlations with MECP2 mutations in term of efficacy or adverse reactions.