Nocturnal alterations in Rett syndrome: a polysomnographic study

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Purpose Rett syndrome is a severe neurodevelopmental disorder mainly affecting females and usually linked to mutations in the methyl-CpG-binding protein 2 gene, with an estimated prevalence of 1 in 10,000 live female births.
Clinical features which usually become more apparent over time include breathing dysfunction, seizures, spasticity, peripheral vasomotor disturbance, scoliosis, growth retardation, and hypotrophic feet, with a great variety of presentations. The clear immaturity in brainstem mechanisms is expressed by the presence of early sleep disorders such as nocturnal awakenings, bruxism, and difficulty falling asleep, and no conclusive findings were derived from the few polysomnographic studies about the sleep macrostructural aspects.

The aim of this study is to analyze the sleep macrostructural parameters, the nocturnal respiratory characteristic, and the
presence of periodic limb movements in a sample of children affected by Rett syndrome.

Materials and Methods Thirteen Rett subjects underwent a polysomnographic study, and the findings were compared with those obtained by a group of 40 healthy children.

Results The Rett group shows a great impairment in sleep macrostructural and respiratory parameters, with a higher percentage of pathological periodic limb movements than controls.

Conclusions This study may be considered a report about the ventilatory impairment during sleep in Rett syndrome and the first approach to the macrostructural aspects of sleep supported by the PSG data that could be considered mandatory
for a better comprehension of this very complex syndrome

Bibliografia

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