

Reference

Boban S, Wong K, Epstein A, Anderson B, Murphy N, Downs J, Leonard H. Determinants of sleep disturbances in Rett syndrome: Novel findings in relation to genotype. American Journal of Medical Genetics Part A. 9999A:1–9.

Background

Sleep problems are known to occur in Rett syndrome and can have a significant impact on the affected child and the family. We investigated how common they were and whether or not some girls and women were more at risk than others in a large international sample of individuals with Rett syndrome.

What We Did

Families with a child with Rett syndrome registered in the International Rett Syndrome Phenotype Database (InterRett) were invited to participate. A high proportion of families filled out the questionnaire, mainly online, and provided information on the presence, nature, and frequency of their child's sleep problems.

What We Found

Sleep problems in Rett syndrome occurred much more frequently than in the general population. Night waking was the most common sleep problem with nearly half of the girls and women currently waking often at night. Getting to sleep and staying asleep was most disrupted for younger children and those with a p.Arg294* mutation. Severe seizure activity and being unable to walk were associated with a greater likelihood of daytime napping.

What It Means

This was the first study to explore sleep problems in Rett syndrome using a large international sample. Our findings highlight the different effects caused by abnormalities in the MECP2 gene and explain some of the variation in sleep disturbances. We were surprised to find more sleep difficulties associated with the p.Arg294* mutation given this mutation is usually associated with milder symptoms in Rett syndrome.