

Reference

Urbanowicz A, Downs J, Girdler S, Ciccone N, Leonard H. 2015. Aspects of speech-language abilities are influenced by MECP2 mutation type in girls with Rett syndrome. *Am J Med Genet Part A* 167A:354-362.

Background

Speech and language abilities are almost always severely affected in girls and women with Rett syndrome and often lost during the period of regression. This study explored relationships between speech and language abilities in girls with Rett syndrome and how they may be affected by the type of genetic mutation.

What We Did

We investigated the level of speech and language abilities before and after this period of regression, the age at which the regression occurred and whether this varied according to the girl's type of genetic mutation. To do this we used information families of over 700 girls had provided to the Australian Rett Syndrome Database and the International Rett Syndrome Phenotype Database (InterRett).

What We Found

Most of the girls either babbled or learned to use words during development. Of those, the majority but not all lost these abilities. Girls with a p.Arg133Cys mutation were the most likely to use one or more words prior to regression but to continue to be able to use words after regression. However some of those with a p.Arg168X mutation or a p.Arg306Cys mutation also continued to have the ability to use words after regression.

What It Means

This study examined speech and language in one of the largest samples of girls with Rett syndrome to date. Girls with Rett syndrome differ in their speech and language abilities and in their experience of regression, and these differences are partially explained by the type of mutation. Girls with mutations associated with milder features were most likely to use words before and after a regression, and also to lose language skills later than those with mutations associated with more severe traits.