InterRett:
International Rett Syndrome database

IRSF

Bebbington, A., Anderson, A., Ravine, D., Fyfe, S., Pineda, M., de Klerk, N., Ben-Zeev, B., Yatawara, N., Percy, A., Kaufmann, W. E., & Leonard, H. (2008). Investigating genotype-phenotype relationships in Rett syndrome using an international data set. *Neurology, 70*, 868-875.

Background

Rett syndrome is a rare neurodevelopmental disorder caused by mutations in the *MECP2* gene. There is considerable variation in the clinical features likely to be related to the type of mutation affecting the individual person. This study used information from an international Rett syndrome database to investigate the relationship between clinical characteristics and different mutation types.

What we did

Information on 346 girls and women provided to the InterRett database by families and clinicians was used in this study. We examined overall severity and individual characteristics of girls and women with each of the common mutations.

What we found

The most common mutations identified were p.R106W, p.R133C, p.T158M, p.R168X, p.R255X, p.R270X, p.R294X, p.R306C and C-terminal deletions. Those with a p.R133C or p.R294X mutation tended to have milder symptoms overall and those with the p.R255X or p.R270X mutation were more severely affected. The type of mutation also appeared to influence the age at regression, age at development of hand stereotypies, feeding skills and abilities to walk, speak, and use hands.

What does it mean

This study provides information that helps to predict the likely clinical characteristics associated with some of the common *MECP2* mutations. This study also illustrates the need to collect information about large numbers of cases in order to detect differences between mutation types.

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