
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

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.