

Disease

# Rett syndrome<sup>1, 2</sup>

Gene

## MECP2

|                                                 |                                                                                                                                                                                                                                           |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | XL                                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | Trofinetide                                                                                                                                                                                                                               |
| <b>Evidence for Treatment</b>                   | guideline <sup>3</sup>                                                                                                                                                                                                                    |
| <b>Non-Molecular Testing Availability</b>       | no                                                                                                                                                                                                                                        |
| <b>Disease Frequency (per 100,000)</b>          | 3.575 <sup>1</sup>                                                                                                                                                                                                                        |
| <b>Genes Included in Disease Frequency</b>      | MECP2                                                                                                                                                                                                                                     |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Rett+Syndrome&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Rett+Syndrome&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

<sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1497/>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/38318312/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/32984552/>