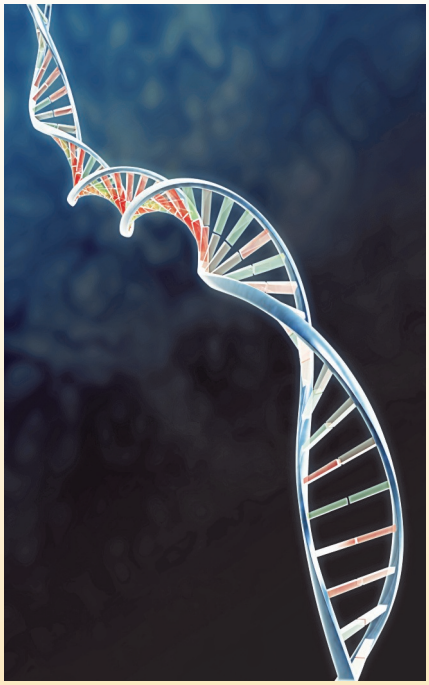
GENETICS CENTER

Rett Syndrome

Classic Rett syndrome is an X-linked condition known to be associated with a mutation in the *MECP2* gene. Associated features include progressive neurodevelopmental disorder that primarily affects girls. There is usually normal development during the first 6 to 18 months of life followed by a short period of stagnation. Then there is usually regression in language and motor skills with additional features of stereotypic hand movements, lack of purposeful use of the hands, teeth grinding, periodic breathing, seizures and acquired microcephaly.

Atypical Rett syndrome is similar to classic Rett syndrome; however, patients tend to have either milder or more severe findings compared to classic Rett syndrome. Atypical Rett syndrome is associated with mutations in either *FOXG1* or *CDKL5* genes. Specifically, mutations in the *CDKL5* gene can be associated with severe and early onset seizure activity. A small percentage of male patients have also been described with atypical Rett syndrome.



Genetics Center's Comprehensive Rett Syndrome Panel includes the following genes:

- ***MECP2***
- ***FOXG1***
- ***CDKL5***

Our laboratory accepts both peripheral blood, as well as buccal swab samples for molecular analysis. Rett syndrome testing is also available on a single gene basis.

As we are a comprehensive center, our medical geneticists and genetic counselors are available to provide clinical evaluations, as well as genetic counseling for patients and their families.

For further information about testing, CPT codes, and genetic patient referrals, please visit www.geneticscenter.com or contact us at:

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