Published on *My46* (https://www.my46.org) 01/19/2022

My46 Trait Profile

Rett syndrome

Rett syndrome is a rare neurodevelopmental disorder primarily affecting females. It is caused by mutations in the MECP2 gene which makes a protein called methyl-CpG-binding protein 2.

Characteristics of Rett syndrome

Rett syndrome is a rare genetic condition that affects the development of the brain and nervous system. Girls with classic Rett syndrome usually have a period of normal development followed by a regression in development (loss of acquired skills) at 6 to 18 months of age. While some developmental skills may be gained back, there is typically a continued loss of previously acquired verbal skills and purposeful hand use. During this period of regression, repetitive hand movements usually begin. Many, though not all, have a normal head size at birth followed by a slowing of head growth after birth. Additional features may include the inability to walk or difficulty walking without assistance, breathing problems (breathing very fast and/or breath holding), teeth grinding (bruxism), seizures, poor growth, curving of the spine (scoliosis/kyphosis), disrupted sleeping patterns, autistic-like behaviors, and panic-like attacks.

Diagnosis/Testing

Rett syndrome can be diagnosed either by meeting certain clinical criteria or by genetic testing for a change or mutation in a gene called MECP2. This gene makes the methyl-CpG-binding protein 2. This protein is thought to help control the activity of certain genes, and is also thought to be important for the development of brain cells. Mutations in the MECP2 gene may interfere with the protein's normal function and the ability to interact with certain genes that are important for normal development and function.

Management/Surveillance

The management of Rett syndrome is based on the symptoms of the individual. Typically, physical, occupational, and speech therapies are required to improve developmental skills following the initial regression phase. Anti-seizure medications, nutrition supplementation, and other treatments are used as needed.

Individuals with Rett syndrome are also at an increased risk for certain changes in heart function that can cause the heart to beat irregularly. Periodic EKGs (tests that monitor the electrical activity of the heart) is often needed to watch for these changes.

Mode of inheritance

Rett syndrome is inherited in an X-linked dominant pattern. This means only one copy of the MECP2 gene whether in a female with two X chromosomes or in a male with one X chromosome must have a change or mutation for an individual to be affected with Rett syndrome. It is thought that in males, Rett syndrome is fatal before birth because a second normal X chromosome is needed for survival to birth. However, some males with Rett syndrome (or variants of Rett syndrome) have been reported. In most individuals, the mutation in the MECP2 gene occurs brand new (de novo). However in about 1% of cases, the mutation is inherited from the mother.

Risk to family members

The risk to family members depends on whether or not the individual with Rett syndrome inherited the mutation or if the mutation occurred de novo. Since the vast majority of individuals with Rett syndrome have a de novo mutation,

the risk of other siblings being affected is very low.

Special considerations

None

Resources

International Rett Syndrome Foundation
http://www.rettsyndrome.org/
Cure Rett
http://www.curerett.org.uk/
Genetics Home Reference: Rett syndrome
http://ghr.nlm.nih.gov/condition/rett-syndrome
Medical Home Portal: Rett Syndrome
http://www.medicalhomeportal.org/diagnoses-and-conditions/rett-syndrome

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Created: 09/2013 **Created by:** Lauren M. Baggett, MS, CGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC