

Diagnosis

Even though mutations in the MECP2 gene have been found to cause Rett syndrome, diagnosis of the condition is based on an individual meeting certain criteria. There are two broad diagnostic groups in Rett syndrome, 'typical' and 'atypical'. Detection of a mutation in the MECP2 gene simply confirms the diagnosis that was made according to the criteria. A diagnosis of Rett syndrome can be made when there is NO mutation in the MECP2 gene.

Typical Rett syndrome

The five criteria that now must all be met for an individual to receive a diagnosis of 'typical' or 'classical' Rett syndrome are:

- A period of regression during the first 5 years of life followed by recovery or stability. Children start off normally then go 'backwards' in their development
- Partial or complete loss of acquired purposeful hand skills
- Partial or complete loss of acquired spoken language
 - Persons with Rett syndrome may have developed to a babbling stage with their speech or may have been speaking in words or sentences, before the loss of such
- Impaired ability to coordinate walking or an absence of the ability to walk
- Stereotypic repetitive hand movements such as wringing/squeezing, clapping/tapping, mouthing, washing/rubbing.

One or more symptoms such as irregular breathing, sleep disturbances, abnormal muscle tone, curvature of the spine, bouts of screaming, teeth grinding, cold and small hands and/or feet, growth retardation and an intense eye gaze, may also be evident.

'Atypical' Rett syndrome

Information about a diagnosis of 'atypical' Rett syndrome is contained in Fact Sheet 4.

Stages of Rett syndrome

Four stages have been suggested to characterise Rett syndrome from infancy to adolescence and are applicable to both 'typical' and 'atypical' Rett syndrome.

- Stage 1 (onset 6 to 18 months) - General slowing down of development
- Stage 2 (onset 1 to 3 years) - Rapid disintegration/regression period
- Stage 3 (onset 2 to 10 years) - Stationary or 'wake up' period
- Stage 4 (onset 11 years+) - Late motor deterioration.

The range of disability can vary considerably among persons with Rett syndrome. For instance, not all will necessarily experience stage 4 of the disorder. While not a common occurrence, two individuals of the same age can present totally different pictures of the disorder.

In summary, Rett children appear to be developing normally and usually reach early milestones of development. Then things start going backwards. They may lose speech. If walking, it may stop. Purposeful hand use may be lacking. And for their families, this is only the start of their Diagnosis of Rett s Rett journey.

Source: RSAA, 30 December 2020



FACT SHEET 3

Diagnosis of typical Rett Syndrome

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