Fact Sheet 4

Diagnosis of 'atypical' Rett syndrome

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Diagnosis of 'atypical' Rett syndrome

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'. 'Atypical' Rett Syndrome is diagnosed when a child or adult has some, but not all, of the essential diagnostic criteria set down for 'typical' Rett syndrome, together with a certain number of symptoms that are associated with the disorder. As with 'typical' Rett syndrome, most of those diagnosed as having the 'atypical' form, are female. Detection of a mutation in the MECP2 gene simply confirms the diagnosis made according to the criteria. A diagnosis of 'atypical' Rett syndrome can also be made when there is **NO** mutation in the MECP2 gene.

'Atypical' Rett syndrome

The criteria that now must be met for an individual to receive a diagnosis of 'atypical' Rett syndrome are:

A period of regression followed by recovery or stability

At least **two** of the following four essential criteria for 'typical' Rett syndrome -

- Partial or complete loss of acquired purposeful hand use
- Partial or complete loss of acquired spoken language
- An impaired ability to coordinate walking or absence of the ability to walk
- Stereotypic hand movements such as wringing/squeezing, clapping/tapping, mouthing, washing/rubbing

At least **five** of the following 11 symptoms -

- Breathing disturbances while awake
- Disrupted sleep pattern
- Cold and/or small hands and feet
- Inappropriate laughing/screaming spells
- Intense use of eyes for communication
- Teeth grinding
- Curvature of the spine or neck
- Abnormal muscle tone
- Growth retardation
- Diminished response to pain.

Source: RSAA, 30 December 2020