Rett Syndrome

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Rett syndrome is a condition of the nervous system that affects all body movement and is primarily found (almost exclusively) amongst females.

It is a genetic neurological disorder that is caused by a mutation of a gene (MECP2 gene) found on the X chromosome.

There are 4 stages of Rett syndrome.

Stage 1 (early onset)

Infants in the first stage of this disorder may experience normal (or close to normal) development until the ages of 6 to 18 months.

Their development then begins to slow and they start to lose (slowly or suddenly) skills that they have previously acquired.

Stage 2 (rapid destructive stage)

During the rapid destructive stage, the ability to vocalise words or sounds and intentionally perform hand movements may be lost. The slowing of head growth usually becomes noticeable at this stage and symptoms similar to autism may show.

Also, breathing irregularities (e.g. hyperventilation) may become apparent and characteristic hand movements, such as repeated clapping or wringing, can also emerge.

This stage of Rett syndrome can last for weeks or months and normally occurs between the ages of 1 to 4.

Stage 3 (plateau stage)

Children in this stage of the disorder (aged between 2 and 10) will usually experience an improvement in behavior. However, the emergence and frequency of seizure activity and movement difficulties (motor skills) may become more apparent. It is at this stage that some females may remain for the rest of their lives.

Stage 4 (late motor deterioration)

The late motor deterioration stage, which can last for years, includes the progressive decline of muscle strength and movement.

Jerky movements and sustained muscle contractions can occur, causing pain in the affected muscles. However, repetitive hand movements may decrease and attention span may increase during this stage.

Frequency

Affects between 1 in 9,000 live female births in Australia.

Diagnosis

Diagnosis of Rett syndrome is conducted clinically by medical professionals, who observe the signs and symptoms during the child's early stages of growth and then continue to perform regular evaluations. There is also a genetic test that searches for the MECP2 gene on the X chromosome which can be done to accompany the diagnosis.

Signs and Symptoms

- Development regression
- Loss of conscious hand movement

- Loss of vocal skills (partial or complete)
- Walking irregularities (e.g. unsteady walking or inability to walk)
- Repetitive hand movements

Treatment

There is no cure for Rett syndrome at this point in time. However, therapy can aid in slowing down the progressive movement loss.

Some therapies include:

- Physiotherapy (helps prevent joint deformities and improve movement ability)
- Occupational therapy (aids in improving hand movement)
- Music therapy
- Hydrotherapy
- Horseback riding

Contact IDEAS

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Disclaimer

This fact sheet provides general information about the disability and is for informational purposes only. It is not a guarantee that you will meet the disability requirements in section 24 of the NDIS Act.

References

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https://www.ninds.nih.gov/disorders/patient-caregiver-education/fact-sheets/rett-syndrome-fact-sheet#3277_6