

Rett Syndrome

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Rett Syndrome was first recognized by Andreas Rett in 1966 and is a neurological disorder affecting primarily females. Autopsies on the brains of these individuals indicate a pathology different than autism; however, children afflicted with Rett Syndrome often exhibit autistic-like behaviors, such as repetitive hand movements, prolonged toe walking, body rocking, and sleep problems.

The prevalence of Rett Syndrome is similar to the prevalence of autism; that is, estimates are between 1 in 10,000 births and 1 in 15,000 births.

Typical characteristics:

- Normal development until 1/2 to 1 1/2 years
- Shakiness of the torso, and possibly the limbs
- Unsteady, stiff-legged gait
- Breathing difficulties (hyperventilation, apnea, air swallowing)
- Seizures (approximately 80% have epilepsy)
- Teeth grinding and difficulty chewing
- Retarded growth and small head
- Functioning level is usually between severely and profoundly mentally retarded
- Hypoactivity

In most cases, there is a regression in cognition, behavior, social, and motor skills throughout their lifetime.

In 1999, Dr. Huda Zoghbi and her colleagues located the gene for Rett syndrome. The gene was located on one of the two X chromosomes that determine sex. Rett syndrome results from the mutation of the gene that makes methyl cytosine binding protein, resulting in excessive amounts of this protein.

For more information, contact:

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