

Rett Syndrome

Some of you may have heard about it but probably have not had too much exposure to this diagnosis. This is another one of those diagnoses that was usually based on symptoms although there is now a genetic test available to confirm the diagnosis.



(6 months)



(5 years)

One of the things that must be most frustrating and heart breaking for parents is that there is a period of apparent normal development until 6-18 months of age. Most of the patients are female. It is believed to affect 1 in 10,000 females.

Diagnostic Criteria include:



(10 months)

(7 years)

- **Period of normal development from 6-18 months.**
 - **Normal head circumference at birth that then slows.**
 - **Loss of verbal language**
 - **No purposeful hand movements**
 - **If able to walk, the gait is usually wide-based and stiff legged.**
- **Torso and/or limbs have shakiness, especially if upset**

Most also develop seizures and may also have irregular breathing and motor-control problems. Most live to adulthood, but never regain the use of their hands or verbal language skills.



The cause has been identified as a defect in the MECP2 gene on the X chromosome. It is often misdiagnosed as autism, cerebral palsy or a non-specific developmental delay. It was once thought to be a progressive, degenerative disorder, but most researchers now agree that it is a developmental disorder.

This information was compiled from several web sites.

www.rsrf.org (Rett Syndrome Research Foundation) (pictures used from this web site with permission)

www.isn.net/~jypsy/rett.htm and

www.rettsyndrome.org (International Rett Syndrome Association).

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