Rett Syndrome

What is Rett Syndrome?

Rett Syndrome is a rare regressive neurological disorder that usually affects only girls. Girls with Rett Syndrome appear to develop normally until six to eighteen months of age when they start to lose acquired skills. This begins with the loss of communication skills and purposeful use of their hands and is followed by progression to unusual movements such as hand washing, disturbances in gait and slowing of head growth. Seizures and breathing problems may also appear.

Birth Defect Research For Children, Inc

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How many children have Rett Syndrome?

Rett Syndrome affects one in every 10,000-15,000 live-born female babies in all racial and ethnic groups worldwide.

What causes Rett Syndrome?

Rett Syndrome is caused by a MECP2 gene mutation. Since the disorder usually occurs spontaneously, there is less than a 1% risk of another child in the same family being born with Rett Syndrome.

How is Rett Syndrome Diagnosed?

Rett Syndrome is diagnosed by a combination of clinical observation for typical signs and symptoms and a genetic test to search for the MECP2 mutation on the child's X chromosome.

Stages of Rett Syndrome

Stage 1 - Early onset. Around 6 -18 months of age the baby may show less eye contact and lose interest in their toys. There may also be delays in sitting and crawling. Hand-wringing and slowed head growth may begin.
Stage 2: - Rapid decline. Between ages 1-4, the child may lose purposeful hand skills and spoken language. More disturbing hand movements (wringing, clapping, tapping and frequent hands in the mouth) may accelerate. Breathing problems including sleep apnea and hyperventilation may begin. Many girls with Rett Syndrome may have autistic-like symptoms in the areas of social interaction and communication.
Walking and balance may be unsteady and decreased head growth becomes noticeable.
Stage 3- Plateau stage: Between 2-10 years of age, apraxia, motor problems and seizures may be prominent, but alertness, attention span and communication skills may improve. Some girls with Rett Syndrome stay in Stay 3 for the rest of their lives.
Stage 4 - Late motor deterioration stage: Girls in this stage may have reduced mobility, scoliosis, muscle weakness, spasticity and increased muscle tone leading to abnormal positioning of their limbs or the top of their body.

Treatment for Rett Syndrome

Although there is no cure for Rett Syndrome, many girls with this disorder live into middle age. Treatment is supportive and focuses on managing symptoms like seizures and breathing problems. Occupational therapy may be helpful in life skills development. Physical therapy and hydrotherapy may improve mobility. Federal research is underway to develop better methods of diagnosis and new therapies to manage symptoms of Rett Syndrome.

Fact Sheet by:

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