Rett Syndrome

Rett syndrome (RTT) is a problem that shows up in the first 6 to 18 months of life. A syndrome is a collection of symptoms and medical problems. RTT is a life-long problem that has a variety of mental and physical issues. It is almost exclusively found in girls.

CAUSES

RTT is caused by mutation of genes found on the X chromosome. This leads to a decrease in the size and maturity of the brain. The actual cause of the mutation is not known. This is called a spontaneous mutation means RTT (meaning it is not inherited). The chance of having a second child with RTT is less than 1 %.

SYMPTOMS

Children with RTT usually appear normal in the first 6 to 18 months of life. Problems that develop can be mild to severe. Some children follow certain phases of symptoms. RTT can vary from one child to another. Common symptoms include:

- Slowing down in development.
- Reduced muscle tone (*hypotonia*) and coordination.
- Poor eye contact.
- Difficulty relating to people.
- Periods of irritability or prolonged crying.
- Difficulty in expressing feelings.
- Odd hand movements.
- Walking or crawling problems
- Seizures (*convulsions*).
- Poor weight and height gain.
- Small head size.
- Irregular breathing.
- Feeding problems.
- Difficulty swallowing.
- Constipation.

Some of the social problems often improve with time.

DIAGNOSIS

Diagnosis of RTT may be based on the history and physical exam. There are certain criteria that must be present to make the diagnosis. Genetic testing can be done if the diagnosis is not clear.

TREATMENT

There is no cure for RS. Treatment depends on the child's issues. Your child's caregiver may suggest:

- Medicine for seizures.
- Occupational, communication and physical therapy.
- Nutritional support for eating problems and poor weight gain.
- Referral to a genetics specialist if you have concerns about risks of RTT.

HOME CARE

Children with RTT usually need help for most daily activities. Special education approaches are needed. Some children can be toilet trained and can learn to feed themselves. Children and adults with RTT can continue to learn and enjoy interactions at home, school and other social settings. Support groups for parents of children with disabilities are helpful.

SEEK MEDICAL CARE IF:

- You are having problems coping with your child's behavior or disabilities.
- Your child has convulsions (*seizures*).
- Your child's seizures happen more often or are different.
- Your child has constipation.
- Your child has frequent choking, spitting up or swallowing problems.
- Your child has sleep problems.

SEEK IMMEDIATE MEDICAL CARE IF:

- A seizure lasts for more than 5 minutes.
- One seizure follows another without a return of consciousness.

WHAT WILL HAPPEN

In spite of the severe problems that occur with RTT, most children can be expected to reach adulthood. Most survive at least into their 40s. However, there is a risk of sudden unexplained death.

Research is continually being done to learn more about this problem.

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