THE CURRENT VIEW ON RETT SYNDROME

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Rett syndrome is a rare genetic neurodevelopmental disorder which affects brain development and leads to motor skills and speech. This disorder usually affects girls. Children with Rett syndrome develop normally during the first 6-18 months, but then the levels of their previously developed skills decrease (e.g. crawling, walking, communication, and using hands). As time passes, children experience difficulties related to using muscles to control their movements, coordination and communication as well. And as a result, it can lead to seizures, abnormal hand movements (e.g. repetitive rubbing or clapping), and intellectual disabilities in the future.

Children who are diagnosed with Rett syndrome typically are born after normal pregnancy and delivery without difficulties. As it was already mentioned, they develop normally till the age of 6 months, and after that the first signs and symptoms of Rett syndrome start to occur. These changes in behavior and development usually occur at the age of 12-18 months during the period of several weeks or months.

The symptom list is presented below (the severity of the expression of symptoms is indiusaly):

1) slowed growth. Microcephaly is one of the first sign which is common among children diagnosed with Rett syndrome;
2) decreased levels of normal movement and proper coordination (e.g. problems with crawling and working normally and decreased ability to control hand movements);
3) low communicative skills. Children diagnosed with Rett syndrome lose their communicative skills, make less eye contact, and are less interested in other people and playing with toys;
4) abnormal hand movements;
5) abnormal eye movements;
6) difficulties with breathing (e.g. hyperventilation, forceful exhalation of saliva or air);
7) irritability and/or crying. In addition, some children with Rett syndrome experience fear and/or anxiety;
8) cognitive impairment;
9) seizures;
10) abnormal curvature of the spine - scoliosis, which is usually diagnosed at the age of 8-11 years, and progress with age;
11) problems with heartbeat;
12) sleep problem (e.g. walking at night with screaming or crying, irregular sleep time);
13) other abnormal behavior and other symptoms (e.g. strange facial expression, laughing for a long time, hand licking, grasping of hair, bones prone to fractures, bowel problems, teeth grinding, small hands and feet).

There are four stages of development of Rett syndrome:

1) Stage 1: early onset. Babies at this stage (approximately at the age of 6-18 months) show less eye contact, and their interest in toys decreases. Additionally, they can have delays in crawling and/or sitting;
2) Stage 2: rapid deterioration. At the age of 1-4 years old, children start to lose rapidly or gradually skills which they have already developed. Symptoms which are present on this stage are: slowed head growth, abnormal hand movements, increased ventilation, screaming and/or crying with no reasons, problems related to movements and/or coordination, low communicative skills and the level of social interaction;
3) Stage 3: plateau. This stage usually begins at the ages of 2-10 years, and can be present for a long period of time. Problems with movements, and coordination are still present, but at the same time, behaviors, such as crying, irritability, hand use and communication, can slightly improve. Additionally, there can be seizures which are not present before the age of 2;
4) Stage 4: late motor deterioration. This stage starts at the age of 10 years, and lasts for years or decades. Symptoms which are linked to this stage are: decreased mobility, muscle weakness, joint contractures, scoliosis. Unfortunately, communicative and hand skills remain on the same low level of can improve, but nonsignificantly. As to seizures, they are still present, but less often.

As it was already mentioned, Rett syndrome is a genetic neurodevelopmental disorder. The genetic mutation related to this syndrome occurs randomly, and usually is present in the MECP2 gene. This mutation is caused by problems related to protein production which is critical for brain development. There are few cases in which it was reported that Rett syndrome was inherited.

All in all, the exact causes of Rett syndrome are not very well understood, and now there are a lot of studies which focus on their studying.

Due to different chromosome combinations, boys who have genetic mutation related to Rett syndrome are affected in a significantly bad way. Usually, they die before the birth or during the period of early infancy.

The number of boys, who can have other mutations with less destructive influence from Rett syndrome, is low. Like girls with Rett syndrome, boys also have intellectual and developmental problems.

Based on studies, it can be concluded that there are no risk factors which can lead to development of the genetic mutation which in terms influence occurrence of Rett syndrome.

The only possible risk factor which was identified in some studies was inherited factors (e.g. having a close relative who is diagnosed with Rett syndrome), but this factor was present only in a few studies.

The list of possible complication which can be present in children with Rett syndrome include:
1) sleep problems which are caused by sleep disruption;
2) eating problems which result in poor nutrition and delays in growth;
3) problems with bowel and/or bladder, including constipation, gastroesophageal reflux disease, gallbladder disease, bowel or urinary incontinence;
4) problems with bones, joints and muscles;
5) pain which is caused by gastrointestinal problems and/or bone fractures;
6) Behavioral problems and/or anxiety which influence negatively social functioning;
7) need for help and assistance in everyday life;
8) shortened life span: due to heart problems children with Rett syndrome may not live as long as the average person.

Diagnostic interventions for Rett syndrome include observation of a child's growth and development, collection of information from parents related to their medical history. The diagnosis is usually established if the growth of head is slow and simultaneously there is loss of previously acquired skills.

In addition, therapist should eliminate disorders and illnesses which have similar to Rett syndrome symptoms:
1) Autism spectrum disorder;
2) Cerebral palsy;
3) Metabolic disorders (e.g. phenylketonuria);
4) Disorders linked to hearing or vision problem;
5) Other genetic disorders;
6) Degenerative disorders;
7) Brain disorders caused by trauma or infection or prenatal brain damage

In order to eliminate these disorders and illnesses, the following tests should be prescribed:
1) blood and urine tests;
2) magnetic resonance imaging or computerized tomography scans;
3) hearing tests and vision examinations

A therapist gives diagnosis of Rett syndrome if a child meet the following criteria:
1) lose of hand movements;
2) loss of communicative skills;
3) problems with walking;
4) repetitive hand movements which do not have any purpose (e.g. hand-wringing, clapping, tapping, rubbing movements)

These criteria can vary slightly depending on a child, and the expressiveness of these symptoms can be different. Additionally, therapists can ask present to do DNA analysis to test if there is a mutation of MECP2 gene mutation to confirm the diagnosis.

Despite the fact that there is no particular treatment for Rett syndrome, interventions focus on dealing with symptoms and providing support for improving movement, communicative and social skills. These interventions include:
1) regular medical care, along with monitoring physical changes (e.g. heart problems and scoliosis);
2) medications which are used for dealing with seizures, muscle stiffness, breathing, heart, gastrointestinal and sleep problems;
3) physical therapy (e.g. cats and braces) which deals with scoliosis, and helps to support joints and hands. Additionally, physical therapy is used to improve movements, walking skills, and balancing.
4) occupational therapy which is used to improve hand movements
5) speech-language therapy
6) nutritional support;
7) behavioral interventions which focus on sleep problems

There are several physical and behavioral signs which parents should pay attention to:
1) decreased growth of child’s head and/or other body part;
2) low mobility and coordination;
3) repetitive hand movements;
4) decreased level of eye contact and/or low interest in normal play;
5) problem with speech development and/or loss of previously acquired communicative skills;
6) various losses of previous acquired motor skills.

To sum up, it can be said that there should be more studies to explore possible causes of Rett syndrome and focus on development of new treatment methods which can improve the quality of life of patients diagnosed with this syndrome.

References: