Rett syndrome

Template:Infobox - genetic disease **Rett syndrome** is a genetic neurodevelopmental disease caused by a mutation of the *MECP2* gene on the X chromosome. It occurs almost exclusively in **girls**; occurrence in boys is very rare. Boys with a mutation in this gene were thought to die before birth. New studies show that these boys may survive and then develop more severe symptoms earlier than girls. [1]

The phenotype of this classic form of Rett syndrome partially overlaps with other diagnoses (general name "Rett-like phenotype"). In the past, these were described directly as variants of Rett syndrome, but today they are already referred to as separate diagnoses. These are, for example, neurodevelopmental diseases caused by mutations in the *CDKL5* or *FOXG1* genes (**CDKL5 deficiency or FOXG1 syndrome**)[2][3].

The syndrome was first described in 1966 by Austrian pediatrician Andreas Rett.

Symptoms of Rett Syndrome

- **psychomotor development regression** - between 6 and 18 months of age,
- **dyspraxia** (inability to use the hands purposefully), stereotypical hand movements - washing, clapping, squeezing, mouthing, finger manipulation,
- **apraxia**,
- **unsteady walk** - clumsy, puppet-like; loss of ability to walk,
- **intelligence disorders**,
- **communication dysfunction**, regression of speech development, up to complete loss of speech,
- **respiratory dysfunction** - as a result of insufficient development of the respiratory center,
- **hypotonia**,
- **gastrointestinal problems** - problems with chewing, swallowing, reflux, constipation,
- **bruxism** (teeth grinding), strabismus,
- **total growth retardation**,
- **epileptic seizures**,
- **emotional symptoms** - restlessness, dissatisfaction, frequent mood swings, sudden crying,
- **sleep disorders**.

Stages of Rett Syndrome

The disease is very individual for each of the girls. The syndrome occurs in varying degrees of severity, which are determined by the type of mutation, phenotype, and other factors.

Stadium	Symptoms
1. stage: between 6th to 18th month	Psychomotor development slows down, eye contact decreases, no interest in toys, generally impaired contact with the environment.
2. stage: between 1st and 4th year	Faster and more pronounced deepening of disorders of psychomotor development. In a short period of time, loss of speech and the functional use of hands, and the growth of a head also slows down. Symptoms of dementia with autistic features appear. Interest in social contact is lost, they are irritable and dissatisfied.
3. stage: pre-school period	Ataxic walking dominates, doesn't even have to start walking. Deterioration of motor skills, very frequent epileptic seizures. Autistic features usually recede and there may be renewed interest in communication, striving for eye contact.
4. stage: after 10th year	Severe movement and orthopedic problems, (scoliosis, kyphosis). Most of the girls are already confined to a wheelchair by this time. There is an improvement in emotional contact.
5. stage: adolescence and adulthood	They mature mentally, improvement of social feeling. Using eye contact is their strength. They usually live around 40 to 50 years.

Diagnosing Rett Syndrome

First, a clinical diagnosis is established, followed by a genetic examination. Genetically, the clinical diagnosis is confirmed in less than 80% of girls with a clinically determined syndrome.

ICD-10 criteria for Rett syndrome
A. Prenatal and perinatal development is apparently normal, apparently normal psychomotor development for the first five months, normal head circumference at birth.
B. Retardation of head growth between five months and four years, loss of functional manual skills between five and thirty months, along with communication dysfunction, impaired social interaction, unsteady gait and trunk movements.
C. Severe speech impairment, severe psychomotor retardation.
D. Stereotypic movements of the hands around the central axis at the time when loss of functional manual skills occurs.

Rett syndrome treatment

There is currently no treatment that can stop or cure this syndrome. Supportive therapy is used to alleviate the symptoms of the syndrome. A multidisciplinary approach is important.

Symptomatic therapy includes:

- solving gastrointestinal problems – reflux, constipation, nutritional support,
- treatment of scoliosis,
- improving communication skills,
- treatment of sleep disorders,
- Selective serotonin reuptake inhibitors,
- antipsychotics,
- and more.

Links

Related Articles

- Transcription factors
- Pervasive Developmental Disorders

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