

Rett Syndrome (English)

Information on the disease pattern, its background, diagnosis and causes, with useful contacts.

WHAT IS RETT SYNDROME?

Rett Syndrome is a developmental disorder caused by genetic mutation which leads to severe mental and physical disablement. The disease almost exclusively occurs in girls. It was first described by the Viennese paediatrician Professor Andreas Rett (†) in 1966.

All affected children and adults show the same clinical symptoms, the most characteristic of which is involuntary hand movements such as wringing, washing and kneading.

After a normal pregnancy there are at first no apparent abnormalities. It is only later that seclusion, loss of acquired skills and social interaction, loss of speech or slowed head growth are noticeable. Many children do not learn to walk or walk only clumsily. Typical concomitant features include scoliosis, epilepsy and breathing issues.

WHAT ARE THE STAGES OF RETT SYNDROME?

Rett syndrome develops in stages. Predominantly normal early growth and development is followed by stagnation. Acquired skills are lost and interest in the environment disappears.

Stage I begins between 6 and 18 months of age and is characterized by a slowing of development, which may be subtle at first. The infant may show less eye contact and reduced interest in toys. There may be delays in gross motor skills such as sitting and crawling. This stage may last for a few months or continue for more than a year.

Stage II usually begins between ages 1 and 4. A decline of general development becomes evident. The girls rapidly lose spoken language and acquired purposeful hand movements. There is slowdown of head growth. The girls are irritated, they cry and begin stereotyped hand movements. Some girls show signs of autism such as reduced social interaction and communication.

Stage III usually begins between ages 2 and 10. It lasts for years and shows some stabilization. The children may regain certain skills, improve their communication and increase interest in their surroundings. However, apraxia, motor problems and seizures are also prominent during this stage.

Stage IV (late stage) begins at age 10. Prominent features include reduced mobility, curvature of the spine (scoliosis) and muscle weakness. Cognitive, communicative and manual skills are retained and do not decline. Life expectancy of Rett girls and women is not generally reduced, though there seems to be a slightly increased mortality.

HOW IS RETT SYNDROME DIAGNOSED?

Diagnosis of Rett syndrome is extremely difficult because the various symptoms combine in rather different forms in the individual case. In addition to clearly defined main diagnostic criteria specific conditions and supportive criteria and typical of Rett syndrome and affect almost all children.

CRITERIA OF CLINICAL DIAGNOSIS

- Relatively normal development during first 6 to 18 months of life
- Normal head circumference at birth
- commonly reduced growth of head circumference between ages 1 and 4
- Temporary loss of social skills
- Disruption of spoken language and communicative skills, varying degrees of mental retardation
- Loss of acquired purposive hand movement between ages 1 und 4
- Repetitive hand movements (hand wringing, squeezing, kneading, clapping)
- Gait abnormalities

COMMON ATTENDANT SYMPTOMES

- Seclusion
- Seeming lack of interest in surroundings and fellow human beings
- Teeth-grinding
- Inappropriate laughing or screaming spells
- stereotype movements and related apraxia
- different types of Epilepsy
- Scoliosis
- Ataxia
- Digestive disturbances
- Breathing disturbances (such as hyperventilation, apnea (breath holding), and air swallowing)
- Salivation
- Sleep disturbances
- Short stature, small feet
- Poor blood circulation of extremities, cold and blue feet and lower legs

GENETIC DIAGNOSIS

The clinical diagnosis of Rett syndrome can be substantiated by a genetic test since 1999. This is 80-85 per cent positive in children with a ‚classic‘ course of Rett syndrome. Thus, atypical courses and asymptomatic carriers can be detected in girls (and very few boys) who do not display all typical symptoms.

WHAT CAUSES RETT SYNDROME?

Rett syndrome is caused by mutations in the *MECP2* gene (pronounced ‚meck-pea-two‘). In most cases mutations are located in this gene. The *MECP2* gene controls the functions of many other genes. It’s important to know that the diagnosis of Rett syndrome is initially based on clinical criteria. In children fulfilling the diagnosis a genetic test on *MECP2* gene can be made. If a mutation is detected, the diagnosis is confirmed. If no mutation is found, Rett syndrome may still be given.

IS RETT SYNDROME CURABLE?

So far, there is no therapy or medication that can cure Rett syndrome. Various therapeutic measures such as physiotherapy, music therapy, hippotherapy, ergotherapy and speech therapy may be applied to improve the quality of life of affected girls and women. In particular, the method of "Augmentative and Alternative Communication" (AAC) can facilitate exchange with people with Rett syndrome.

Not every therapy is appropriate for everybody. In order to avoid overload, a specific selection of therapeutic measures should be made with respect to the requirements of the individual case.

WHAT ARE THE CHANCES OF HAVING ANOTHER CHILD WITH RETT SYNDROME?

The chance of having more than one child with Rett syndrome is very small, much less than 0.5 percent. The few genetic tests confirming Rett syndrome in siblings suggest a minimum probability which is not clearly defined. In such a case, prenatal diagnosis may be an option. But this should be discussed individually with a skilled genetic counsellor.

FURTHER INFORMATION AND GUIDANCE

RETT.DE – THE WEBSITE OF RETT DEUTSCHLAND e.V. -

Elternhilfe für Kinder mit Rett-Syndrom (Rett parents support group, Germany)

Our website www.rett.de features up-to-date information on the causes, diagnosis and therapeutic measures of Rett syndrome; news from the Rett parents support group; reports on individual cases as well as on new scientific research and findings.

THE RETT SYNDROME HANDBOOK

The Rett Syndrome Handbook by Kathy Hunter assembles more than 700 pages of scientific contributions on the clinical picture from professional Rett syndrome experts, but also emotional personal reports of the daily life from families with a Rett child. The German version *Das Rett-Syndrom Handbuch* is available through *Rett Deutschland e.V.* (info@rett.de).

RETTLAND MEMBERS' MAGAZINE

The magazine is published by Rett Deutschland e.V. Each issue features individual case reports; an overview of new medical and therapeutical developments, of current legal and social issues; and information on upcoming events, news and activities of *Rett Deutschland e.V.* .

BECOMING A MEMBER

Parents of a child with Rett syndrome are cordially invited to become a member of *Rett Deutschland e.V. - Elternhilfe für Kinder mit Rett-Syndrom*. We share concerns and needs, listen and give advice on all matters. As a member, you are invited to all meetings within your region (or beyond) and to the annual general meeting. Your active engagement is also highly welcome. You will receive our member magazine *RettLand* twice a year.

The annual membership fee amounts to 45 euros (or more) for families and 30 euros (or more) for single parents.

BECOMING A SPONSORING MEMBER

If you wish to support us without having a child with Rett syndrome yourself, you may become a sponsoring member. As a sponsoring member you are, upon request, invited to all meetings within your region and to the annual general meeting. Your active engagement is also highly welcome

Sponsoring membership amounts to 20 euros (or more) per person and year. For an additional fee of 12 euros per year you will receive our members' magazine *RettLand*.

DONATIONS AND PROMOTION

We see it as our foremost task to make Rett syndrome more widely known, to support scientific research and to help affected children and their parents as quickly as possible.

To achieve this, we organize regular information campaigns for paediatricians and advanced training courses. We attend to families of members and give comprehensive advice with questions of contacts, therapeutic options and practical help with everyday life.

More detailed information, applications forms and our privacy statement may be found on our website www.rett.de.

MISSION OF RETT DEUTSCHLAND E.V.

On the initiative of Professor Folker Hanefeld, the parents support group was founded in Göttingen in 1987. His aim was to bring together parents whose children had the same disability. Today, *Rett Deutschland e.V.* has more than 1700 members, nearly 700 of whom are children and adults with Rett syndrome. *Rett Deutschland e.V.* provides information on Rett syndrome for those affected or interested and encourages the exchange of experience amongst parents – an important means to counteract the isolation of families which is often imminent after the diagnosis of Rett syndrome.

Birgit Lork, mother of a daughter with Rett syndrome, is available as a first contact in our office in Witten.

AKTIVITIES

- Guidance to parents, doctors, teachers, therapists and all those concerned with Rett syndrome in their daily life
- Establishing contacts between parents
- Annual general meeting with specialist lectures
- Information weekends for families with fresh diagnosis
- Family weekends and meetings of regional groups und provincial associations
- Training programs
- Holiday camps
- Work group »Adults with Rett syndrome«
- Presence at congresses, symposia and specialized fairs
- Spread of information on Rett syndrome

- Sponsoring research project of Professor Stuart Cobb, Edinburgh

Your contact on the federal board

1. Chairperson - Gabriele Keßler, g.kessler@rett.de
 2. Chairperson - Christine Reiter-Metzler, c.reiter-metzler@rett.de
- Treasurer - Stilianos Brusenbach, s.brusenbach@rett.de

Contact for parents

Birgit Lork, Stockumer Straße, 3 58453 Witten

T 02302 – 962 56 60 F 02302 - 962 56 69 info@rett.de

Contact regarding commercial issues

Katja Mischarin Gaswerkstraße 13 52525 Heinsberg

T 02452 – 180 99 90 - F 02452 - 180 99 99 k.mischarin@rett.de

There are regional groups of *Rett Deutschland e.V.*. With closer proximity to the members, they arrange for more intensive support on the spot such as regular information and families' meetings. Groups and contacts in your vicinity may be found under <https://www.rett.de/de/Anlaufstellen>

Medical advisor

Prof. Dr. med. Bernd Wilken wilken@klinikum-kassel.de

Ambassador: Leslie Malton

Patron: Erdal Keser

Association account

IBAN DE56 5209 0000 0042 2178 08

BIC GENODE51 KS1

Research account

IBAN DE34 5209 0000 0042 2178 16

BIC GENODE51 KS1

Should you have further questions, do not hesitate to contact us. We will try to find a native-speaking contact as soon as possible.