

Rett Syndrome Information Sheet

(Optional sheet to be included where the recipient of a PHR has Rett syndrome)

The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version 'Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

Introduction

Rett syndrome is a neurodevelopmental disorder seen almost exclusively in females. Originally described by Andreas Rett in 1966, this disorder has only been diagnosed in Australia since 1983. It involves severe intellectual and physical handicap, although IQ assessment by traditional methods is difficult.

Rett syndrome is a genetic disorder caused in most cases by *de novo* mutations in the MECP2 gene. There are eight common mutations and some are associated with milder or more severe clinical manifestations. Rarely the mother carries the mutation and there is the chance that other children will have the disorder.

Prevalence

There is a prevalence of 1 in 14,000 girls aged between five and 18 years in Australia and on average twelve girls are born each year who will be diagnosed with this disorder by the age of twelve. Life expectancy is unclear as people born before 1963 may have never been diagnosed.

Characteristics, Classification and Diagnosis

Diagnosis is made by observation and clinical assessment with genetic testing confirming about 80% of cases. As the clinical features develop over time, diagnosis is challenging and misdiagnosis as Prader-Willi syndrome, Angelman syndrome, autism or cerebral palsy can occur in the early stages.

Provisional classification is into three diagnostic subgroups:

Classical Rett Syndrome - all the necessary criteria are met. (Diagnosis is tentative until ages 2-5 years)

Atypical Rett Syndrome - clinical characteristics are present but not all the necessary criteria are met.

Provisional (Potential) Diagnosis –some clinical evidence of Rett syndrome in girls (aged 1-3 years old) who are too young to meet many of the criteria. (Genetic testing may confirm the diagnosis of these cases.)

Necessary criteria for diagnosis of Rett syndrome.

- Apparently normal pregnancy and delivery
- Apparently normal development until 6 months, although there may be developmental delay from birth
- Normal head circumference at birth, followed by slowing of head growth between 5 months to 4 years.
- Loss of acquired purposeful hand skills, associated with communication dysfunction and social withdrawal, between ages 6 and 30 months.
- Severely impaired expressive language (speech) which, combined with the loss of purposeful hand movements, makes the assessment of understanding and intelligence difficult
- Fixed patterns of repetitive hand movements such as wringing, squeezing, clapping, tapping, mouthing, washing and rubbing, appearing after purposeful hand skills are lost.
- Appearance of unsteady or jerky gait and shakiness of the torso (ataxia) between ages 1-4 years.

Supportive Criteria occur commonly but are not mandatory for the diagnosis. These include abnormal breathing patterns, EEG abnormalities, seizures (in up to 80% of cases), scoliosis (curvature of the spine), poor circulation with very cold feet, slow growth with small hands and feet, increasing spasticity of muscles and decreasing mobility with age, abdominal bloating, constipation, teeth grinding, chewing and swallowing difficulties, abnormal pain sensation, eye pointing, 'autistic' behaviours which decrease with age, and disturbed sleep with irritability and night laughing.

A short fourth metatarsal or a short ulna is often seen on X-ray. Osteopenia (thinning of bones) and fractures are common.

Key Recommendations for Management of Rett Syndrome

- Diagnosis, genetic counselling and referral to support services
- Management of seizures (video-EEG monitoring may be helpful)
- Referral to orthopaedic specialist for monitoring and management of scoliosis
- Nutritional management (involving use of gastrostomy if indicated)
- Physiotherapy and occupational therapy to maintain mobility and help prevent osteopenia
- Promotion of communication, exploration of alternatives to speech and communication aids
- Music and hydro therapy
- Active management of constipation
- General supportive and preventative care for patient and family

National Support Association

Rett Syndrome Association of Australia Inc
GPO Box 3497
Melbourne, 3001

President: Bill Callaghan
Phone: (03) 5243 3723
E-mail – rettaust@bigpond.com.au

The Association was established in response to the needs of Rett Syndrome children and their families, to develop understanding and awareness of the syndrome, and to further the advancement of study, research, therapy and care.