

Rett Syndrome

What is Rhett Syndrome?

Rhett Syndrome is a neuro-developmental disorder that affects females almost exclusively. It is a degenerative disorder caused by a mutation in gene MECP2. There are 4 stages to Rhett Syndrome which result in the slowing of physical and intellectual development.

What are the common features of Rhett Syndrome?

Stage 1- 'early onset':

- Occurs between the ages of 6-18 months.
- Usually lasts a month, but can continue for up to a year.
- Many of the symptoms are often overlooked as they are usually very subtle.
- May have reduced eye contact and interest in toys.
- May have delays in gross motor development in sitting and crawling.

Stage 2 - 'rapid destructive stage'

- Occurs between the ages of 1 and 4 years and can last for a week or months.
- Child loses purposeful use of hands and spoken language.
- Characteristic hand movements, such as wringing, tapping, clapping, washing and repeated movement of the hands to the mouth - these movements usually disappear during sleep.

- Apnea and hyperventilation may occur, but usually improves during sleep.
- Some Autistic type behaviours develop, such as a loss of social interaction and communication.
- Walking becomes unsteady and initiating motor movements becomes difficult.
- Slowed head growth

Stage 3 - 'plateau or psuedo-stationary stage'

- Begins between the ages of 2 and 10 years and can last for years.
- Apraxia
- Motor problems
- Seizures
- Behaviour improves with less irritability, crying and Autistic-type characteristics.
- Communication, interest in surroundings, alertness and attention-span can improve.

Stage 4 - 'late motor deterioration'

- Can last for years or decades.
- Reduced mobility
- Curvature of the spine (scoliosis).
- Muscle weakness
- Rigidity
- Spasticity
- Increased muscle tone with abnormal posturing of an arm, leg, or top part of the body.
- Children who were walking may lose this ability.
- Communication, cognition and hand skills do not generally decline further in this stage.

Common difficulties often (but not always) experienced by the child with Rhett Syndrome:

- Significantly impaired communication and cognitive abilities.
- May demonstrate an inability or impaired ability to perform tasks or

movements (a child with Rhett Syndrome may lose the ability to perform motor skills they had previously learned to do, such as walking or crawling).

- Difficulties accessing the school curriculum.
- Difficulty or the inability to perform self care tasks such as feeding, dressing and toileting.

Management strategies that support the child with Rhett Syndrome (at preschool, school and/or home):

- Medication may be needed for breathing irregularities and motor difficulties and anti-convulsant drugs may be used to control seizures.
- Implement routine/planned and structured events.
- Have a good knowledge of the child's strengths and weaknesses, interaction preferences and early signs of distress.
- Use of visual cues to describe the routine. Visual cues are especially useful when preparing for changes in routine.
- Extra time/support to transition to school.
- A physiotherapist can help improve muscle tone, coordination, fine and gross motor skills, prolong mobility through the use of hydrotherapy, development of braces to assist scoliosis and splints to modify hand movements.

Occupational Therapy approaches and activities that can support the child with Rhett Syndrome and/or their carers include:

- **Expanding abilities:** Developing a gradually broadening range of skill areas.
- **Social stories:** Providing ideas and education around social story development.
- **School transition:** Advocating and professionally supporting the transition to school and liaising with teachers, as required.
- **Visual cues** to support routine and to introduce new, or a change in, tasks.

- **Gross and fine motor abilities:** Determining the current age level of a child's gross and fine motor abilities.
- **Devise goals:** Setting functional and achievable goals in collaboration with the child, parents and teachers so that therapy has a common focus beneficial to everyone involved.
- **Educating** parents, carers, teachers and others involved in the child's care about Rhett Syndrome and the age appropriate skills a child should be demonstrating.
- **Management strategies:** Providing management strategies/ideas to assist the child in the home, at school and in the community.
- **Task engagement:** Providing alternative ways to encourage task engagement.
- **Developing Underlying skills** necessary to support whole body (gross motor) and hand dexterity (fine motor) skills, such as providing activities to support:
- **Equipment:** Providing recommendations in regards to equipment and/or home modifications to assist the child to participate in daily activities.
 - balance and coordination
 - strength and endurance
 - attention and alertness
 - body awareness
 - movement planning

Speech Therapy approaches and activities that can support the child with Rhett Syndrome and/or their carers include:

- **Speech and language assessment** to help the family to understand how the child is processing, understanding, learning and using language and communication.
- **Communication strategies:** Providing the family with strategies and techniques to increase and enhance communication with the child.
- **Daily activities:** Helping the child to understand the environment, routines and language.
- **Developing language:** Helping the child to understand and use richer language and to use language more spontaneously.
- **Conversation skills:** Developing conversation skills (e.g. back and forth

exchange, turn taking).

- **Visuals** can be used to help with understanding and the child's ability to express their needs, wants, thoughts and ideas.
- **Social skills:** Development of social skills (i.e. knowing when, how to use language in social situations).
- **Enhancing verbal and non-verbal communication** including natural gestures, speech, signs, pictures and written words.
- **Visual strategies:** Using visual information to help understand, organise and plan the routine for the day.
- **Liaising with educational staff** regarding the nature of the difficulties and ways to help the child to access the curriculum.

Why should I seek therapy for my child with Rhet Syndrome?

Diagnosis alone is NOT the solution. It simply opens the door to getting the help that is needed by arming all involved with the relevant information.

The 'help' still needs to be provided. The help that is provided (at least from a therapy perspective) will reflect:

- First and foremost what medical intervention is needed.
- What the parents/teachers/carers biggest concerns are for the child (i.e. what are the most significant functional challenges).
- The specific areas that are problematic to the child (which will vary even within children with the same diagnosis).
- The capacity of the child's environments to meet the child's needs.

If left untreated the child with Rhet Syndrome may have difficulties with:

- Following instructions within the home, kindergarten or school environment.
- Vocabulary whereby a child cannot clearly get their message across due to limited word knowledge.
- Understanding jokes and figurative language during interactions with others, and when watching TV shows and movies and reading books.
- Learning to talk, speech intelligibility and clarity.

- Managing a full school day due to poor strength and endurance.
- Participating in sporting activities leading to an inactive lifestyle, increasing the risks of other health related issues such as obesity, diabetes, cardiovascular disease or similar conditions.
- Self esteem and confidence when they realise their skills do not match their peers.
- Bullying when others become more aware of the child's difficulties.
- Fine motor skills (e.g. writing, drawing and cutting) due to poor core stability, meaning they do not have a strong base to support the use of their arms and hands.
- Completing self-care tasks (e.g. doing up shoelaces, buttons, zips, using cutlery).
- Self regulation and behaviour as the child is unable to regulate themselves appropriately to settle and attend to a task for extended periods of time.
- Accessing the curriculum because they are unable to attend to tasks long enough to complete assessment criteria.
- Sleep habits, impacting upon skill development due to fatigue.
- Social isolation because they are unable to cope in group situations or busy environments, impacting on their ability to form and maintain friendships.
- Anxiety and stress in a variety of situations leading to difficulty reaching their academic potential.
- Reading/understanding social situations and being perceived as 'rude' by others.
- Social communication, such as eye contact, appropriate distance when talking to someone, turn-taking within a conversation.
- Academic performance: Developing literacy skills such as reading and writing and coping in the academic environment.
- Academic assessment: Completing tests, exams and academic tasks in higher education.

More specific implications of not seeking treatment will be influenced by the common difficulties that are most influencing your individual child.

For more information see the relevant fact sheets under areas of concern or refer to the other relevant resources section below.

What does the diagnosis of Rhett Syndrome really mean for the child?

Diagnoses are used to label a specific set of symptoms that are being experienced by a child.

This label then helps to narrow down and specifically tailor what:

- Other issues commonly occur simultaneously.
- Medication might be appropriate.
- Therapies might help the child (e.g. Medical, Occupational Therapy, Speech Therapy, Psychology).
- Course of intervention (medical and/or allied health) might be and what outcome might be expected (prognosis).
- Can be done to help the child.

A diagnosis helps the child and their carers (parents, teachers, health professionals, carers) to:

- Access information about the relevant cluster of symptoms.
- Communicate the salient features of the child's challenges to all people involved in the child's care.
- Possibly interpret certain behaviours differently in light of the diagnosis.
- Obtain information about what can be done to help the child.
- Determine specifically where and how to help the child.
- Access funding or services that might not otherwise be accessible.

Contact us today to make an initial enquiry or book an assessment for your child
on 1800 KID SENSE (1800 543 736)

