

Role of Physiotherapy in Rett Syndrome


Jackie Harris



Development

- Normal early development? (up to 6 – 18 months of age)
- Evidence to show that early development is not always normal
Burford, Kerr & Macleod 2002 & Einspieler et al 2005

Development

- Retrospective video analysis has shown the Rett baby is usually placid, can have unusual muscle tone, posture and movement, and displays repetitive movements of the upper limb
- Implications for earlier detection/diagnosis 

Development

- Development arrests, onset between 6 – 18 months of age
- Development then regresses, between 1 – 4 years of age (if they have achieved walking, they may stop doing this)
Charman et al 2002

Development

- Condition stabilizes at about 4 – 5 years of age, some kids don't walk till about 4 years of age
Kerr & Ravine 2003
- Many girls become wheelchair bound with age

Gait

- Ataxic
- Wide BOS
- Small step length
- Truncal ataxia
- Side to side rocking
- Generalised slow movements

Gait

- Gait disturbance may not be due to ataxia but a failure of inter-limb co-ordination (as seen in crawling too)
- Rocking of the trunk from side to side may not be due to truncal ataxia but a spontaneous or voluntary movement to induce a step forward

Gait

- Improvement of locomotive movement of the leg by tip-toe-walking explained as provocation of the intraspinal circuit in the spinal cord.
- Lack of tonic innervation from the supraspinal segment to the lumbar-sacral stepping generator is the cause of failure of locomotion.

Segawa 2001

Clinical Features

- Decline in gross motor function & standing ambulation
- Loss of transitional movements
- Poor postural control
- Dystonia
- Reduced purposeful hand use/ jerky uncoordinated movement
- Generalised slowness of movements

Other Complications

- Joint/muscle contractures
- Muscle wasting
- Scoliosis
 - Associated with immobility & inability to climb stairs
 - Can affect posture, mobility, digestion & respiratory function

Kerr et al 2003

Other Complications

- Osteoporosis
- Sedentary lifestyle diseases
- Constipation (can be positively affected by physical activity)

Assessment

- Joint ROM & muscle length
- Muscle tone
- Functional abilities
- Transitional movements
- GAIT/mobility
- Posture
- Seating

Treatment

- Prevent deformity such as joint & muscle contractures, foot deformities & scoliosis:
 - Maintain a good foot position
 - AFO's & splints
 - Serial casting
 - Positioning – prone lying, long sitting
 - Standing frame
 - Upright position, seating

Treatment

- Prevent deformity such as joint & muscle contractures, foot deformities & scoliosis:
 - Care with handling – lifting from shoulders with hip flexion contractures increases lumbar lordosis
Kerr et al 2003
 - Brace treatment of scoliosis can buy time, but becomes less affective & more restrictive as scoliosis worsens
Kerr et al 2003

Treatment

➤ Maintain/improve walking ability

- Daily walking program
- Use of standing and walking frames
- Use of AFO's, wrap-arounds
- Walking can be retrained after it has been lost
Larsson & Engerstrom 2001

Treatment

- Maintain/improve transitional movements
 - STS
 - Floor to stand / stand to floor
 - In/out of bed
 - Sitting on floor to crawl
 - Can be retrained if lost
- Larsson & Engerstrom 2001*

Treatment

➤ Improve fitness

- Daily physical fitness (i.e. – treadmill) improves fitness & functional ability (knee-stand, get up, walk, stairs/slopes) (for girls with independent mobility)
Lotan, Isakov & Merrick 2004
- Increasing fitness can prevent the sedentary lifestyle diseases
- Walking, swimming, riding, stairs

Treatment

➤ Hydrotherapy

- Gait training
- Fitness
- Recreation
- Stretches
- Improve muscle power
- Core stability



Treatment

- Good post-operative follow-up:
 - Plan early
 - Early mobilisation
 - Casting/splinting
 - Retraining

Treatment

- Find out what motivates them
 - People
 - Toys
 - Activities, places to visit
 - Food
 - Etc.

Summary

- Keep them as active and mobile as possible!

References

- Malhotra, Kumar, Gupta. Rett's syndrome – a neurodevelopmental disorder: report of two cases. *Neurol India* 2002; 50:330-3
- Lotan, Isakov, Merrick. Improving functional skills and physical fitness in children with rett syndrome. *J Intellect Disab Research* 2004; 48: 730-735
- Larsson, Engerstrom. Gross motor ability in Rett syndrome – the power of expectation, motivation and planning. *Brain & Devel* 2001; S77 – S81
- Einspieler, Christa, Kerr, Alison, Prechtl, Heinz. Is the early development of girls with rett disorder really normal? *Ped Research* 2005; 57:696-700
- Burford, Kerr, Macleod. Nurse recognition of early deviation in development in home videos of infants with Rett disorder. *J Intellect Disab Research* 2003; 47:588-596
- Haas, Dixon, Sartoris, Hennessy. Osteopenia in Rett syndrome. *The J Pediatrics* 1997; 131: 771-774

References

- Charman, Cass, Owen, Wigram, Slonims, Weeks, Wisbeach, Reilly. Regression in individuals with Rett syndrome. *Brain & Devel* 2002; 24:281-283
- Kerr, Webb, Prescott, Milne. Results of surgery for scoliosis in Rett syndrome. *J Child Neurol* 2003; 18:703-708
- Segawa. Pathophysiology of Rett syndrome from the stand point of clinical characteristics. *Brain & Devel* 2001; 23:S94-S98
- Pozzi, Rosemberg. Rett syndrome: clinical and epidemiological aspects in a Brazilian institution. *Arq. Neuro-Psiquiatr* 2003; 61
- Kerr, Ravine. Review article: Breaking new ground with Rett syndrome. *J Intellect Disab Research* 2003; 47:580-587
- Sarojini, Budden, Gunness. Possible mechanisms of osteopenia in Rett syndrome: Bone histomorphometric studies. *J Child Neurol* 2003; 18:698-702