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 lumbarsacral 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 un-coordinated 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-around
  - 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

References