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6.4 Spondylo-epiphyseal dysplasia – a physiotherapy approach H Mato*, N Hasson, C Pilkington and S Maillard

Address: Great Ormond Street Hospital, London, UK

* Corresponding author

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Background

Spondylo-epiphyseal Dysplasia (SED) is a rare condition which is difficult to diagnose and can be mistaken for juvenile arthritis or Ehlers-Danlos Syndrome (EDS). The symptoms experienced by the young people are often due to a mixture of joint restrictions and hypermobility.

Materials and methods

A retrospective review was completed on 6 cases of SED. All physiotherapy assessments of joint range of movement, muscle strength, balance and gait were included. The physiotherapy programme included stretches to restricted joints and a progressive resisted strengthening programme (high repetitions and low resistance weights) completed into the full range of movement of most joints. Gait re-education and balance training were also included.

Results

Six children (5 M, 1 F) were included, 2 sibling pairs (2 M and 1 M, 1 F). 5/6 had initially been diagnosed with arthritis by their local hospital and one diagnosed with EDS. All had significant difficulties with walking and 3 required crutches. All had severe fixed flexion deformities (FFD) at the hips, 4 had FFD at knees and 2 hyperextension. All demonstrated extremely weak muscles especially hip abductors, hip extensors, inner range quadriceps and plantar flexors. All had very poor balance. Following treatment over 3 months (combination of IP and OP treatment) all had significantly improved the FFD in hips and knees and increased the muscle strength in all groups. Balance had increased in all and gait improved; though 1 still remained on crutches at school.

Conclusion

Physiotherapy can significantly improve the muscle function and quality of life for children with SED.