SURGICAL FUSION OF EARLY ONSET SEVERE SCOLIOSIS INCREASES SURVIVAL IN THE CHILD WITH RETT SYNDROME: A POPULATION BASED STUDY

*Downs J[1,2], Torode I[3], Wong K[1], Ellaway C[4,5,6], Elliot EJ[5,6], Christodoulou J[4,5,6], Jacoby P[1], Thomson MR[7], Izatt MT[8], Askin GN[8], McPhee B[9], Bridge C[10], Cundy P[11,12], Leonard H[1]

1. Telethon Kids Institute, The University of Western Australia, Perth, WA, Australia

2. School of Physiotherapy & Exercise Science, Curtin University, Perth, WA, Australia

3. Department of Orthopaedics, Royal Children's Hospital, Melbourne, VIC, Australia

4. Discipline of Genetic Medicine, The University of Sydney, NSW, Australia

5. Discipline of Paediatrics and Child Health, The University of Sydney and The Children's Hospital at Westmead, Sydney, NSW, Australia

6. The Sydney Children's Hospitals Network (Westmead), Sydney, NSW, Australia

7. Department of Radiology, Princess Margaret Hospital for Children, Perth, WA, Australia

8. Paediatric Spine Research Group, Queensland University of Technology and Mater Health Services, Brisbane, QLD, Australia.

9. Department of Surgery, University of Queensland, Brisbane, QLD, Australia

10. Department of Orthopaedics, The Children's Hospital at Westmead, Sydney, NSW, Australia

11. Discipline of Orthopaedics and Trauma, University of Adelaide, SA, Australia

12. Department of Orthopaedic Surgery, Women's and Children's Hospital, Adelaide, SA, Australia

Introduction

Rett Syndrome is a rare genetic neurodevelopmental disorder usually affecting females. Scoliosis is a common comorbidity and spinal fusion may be recommended if severe. Little is known about long term outcomes. We examined the impact of spinal fusion on survival and risk of severe lower respiratory tract infection (LRTI) in Rett Syndrome.

Methods

Data were ascertained from hospital medical records, the Australian Rett Syndrome Database, a longitudinal and population-based registry of Rett Syndrome cases established in 1993, and the Australian Institute of Health and Welfare National Death Index database. An extended Cox regression model was used to estimate the effect of spinal surgery on survival in females who developed severe scoliosis (Cobb angle > 45 degrees). Generalized estimating equation modelling was used to estimate the effect of spinal surgery on the odds of developing severe LRTI.

Results

Severe scoliosis was identified in 140 cases (60.3%) of whom slightly fewer than half (48.6%) developed scoliosis prior to eight years of age. Scoliosis surgery was performed in 98 (69.0%) of those at a median age of 13 years 3 months (IQR 11 years 5 months – 14 years 10 months). After adjusting for mutation type and age of scoliosis onset, the rate of death was lower in the surgery group (HR 0.30, 95% CI 0.12, 0.74, P = 0.009) compared to those without surgery. Rate of death was particularly reduced for those with early onset scoliosis (HR 0.17, 95% CI 0.06, 0.52, P = 0.002). Spinal fusion was not associated with reduction in the occurrence of a severe LRTI overall (OR 0.60, 95%CI 0.27, 1.33, P=0.206) but was associated with a large reduction in odds of severe LRTI among those with early onset scoliosis (OR 0.32, 95%CI 0.11, 0.93, P=0.036).

Conclusion

With appropriate cautions, spinal fusion confers an advantage to life expectancy in Rett syndrome.