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Gross Motor Abilities in Girls and Women with Rett Syndrome: A Longitudinal Video Study

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Background

Rett syndrome is a rare neurological condition associated with a MECP2 mutation. It results in severe physical and intellectual disability characterised by deceleration of head growth, cognitive impairment, loss of hand skills, severely impaired expressive and receptive language and gradual decrease in acquired gross motor abilities. The temporal changes which occur in gross motor abilities in this population is not known.

Purpose

This study identified changes in gross motor ability over three years in girls/women with Rett syndrome, recruited from a population-based data base. The relationships between this change and age, genotype and change in status of scoliosis were investigated.

Methods

Families participating in the Australian Rett Syndrome Database, were invited to participate in a video study illustrating activities of daily living. Ninety nine families provided videos of girls/women in 2004 and gross motor abilities were described using an assessment tool based on the Gross Motor Function Measure. Seventy five of these cases submitted a subsequent video in 2007, gross motor data were coded and compared with the 2004 data. Genetic data to describe the presence and type of MECP2 mutation was available. Scoliosis was described as absent, present and managed conservatively or surgically. Data were analysed using linear regression models.

Findings

Seventy five cases were categorised into four age groups, <8 years (n=19), 8≤13 years (n=13), 13≤19 years (n=24) and ≥19 (n=18). Preliminary observations suggest that there is some deterioration of gross motor functioning in girls/women with Rett syndrome.

Conclusion

Understanding the progression of Rett syndrome is central in providing information to guide therapeutic interventions, design clinical trials and assist in educating parents and caregivers.

Learning Outcomes

1. Increase understanding of the clinical progression of gross motor abilities in girls/women with Rett syndrome.
2. Gain knowledge for guiding therapeutic interventions when working with girls/women with Rett syndrome.