A Severity Comparison between Italian and Israeli Rett Syndrome Cohorts

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Abstract

Rett syndrome (RTT) is a neurodevelopmental disorder marked by profound cognitive, communication, and motor impairments. Despite identified genotype/phenotype connections, the extent of clinical severity varies even among individuals sharing the same genetic mutation. Diverse sociocultural environments, such as the level of inclusivity of the scholar system, the time spent with family, and the intensity of the rehabilitative intervention provided, might influence their development diversely. This study examines the severity of RTT in people in Italy and Israel, countries with distinct contradictory approaches to caring for those with intricate disabilities, across two age groups. Data from 136 Italian and 59 Israeli girls and women with RTT were assessed and divided into two age categories: above and below 12 years. The RARS, a standardized RTT-specific clinical severity tool, was administered. Despite no differences in age and genetic characteristics, the Italian group showed better scores in the RARS motor and disease-related characteristics areas in both age groups. Moreover, the young Italian participants gathered better total RARS scores and emotional and behavioral characteristics area scores. Furthermore, the young group showed significantly less scoliosis, foot problems, and epilepsy than the older group. These findings endorse the inclusion of girls with RTT in the regular schooling system for a limited daily period, investing in high activity levels within the home and community environments, and suggest continuously providing the person with daily occasions of physical activity and socialization.

see also:
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