Comparing Social Reciprocity in Preserved Speech Variant and Typical Rett Syndrome during the Early Years of Life

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Background: In recent years, understanding more precisely the relationships between, and defining characteristics of, a genotype-phenotype match for both mutation type and variants of Rett syndrome (RTT) has become an increasing focus for research. This study explored questions relating to early social awareness in RTT as a contribution to extending knowledge and understanding of the characteristics of classic or typical RTT and the milder ‘Zappella’ or preserved speech variant (PSV), both before and during the regression phase.

Aims: (a) to explore whether individuals with RTT demonstrate social awareness (social reciprocity) during the early years of life; (b) to determine whether there are changes over time which may span the pre- and post-regression period; (c) to ascertain whether there are differences in these social markers between typical RTT and PSV.

Method: Retrospective video analysis of 10 toddlers with typical RTT and five with PSV investigated participants’ orientation to their name being called between the ages of 5 and 24 months, prior to their diagnosis.

Results: From analysis of the recordings two distinct profiles were apparent. Although response rate was higher in girls with typical RTT than PSV at 5 to 8 months this noticeably reversed from 9 to 12 months onwards. By two years of age there was a markedly higher rate and range of responses from girls with PSV.

Conclusion: This study contributes to the delineation of different profiles for the variants of RTT.

Preserved Speech Variant and Typical Rett Syndrome during the Early Years of Life. Research in Developmental Disabilities. doi:10.1016/j.ridd.2015.06.008.