Cognitive-behavioral characteristics and developmental trajectories in children with deletion 11qter (Jacobsen syndrome), and their relation to deletion size

Abstract

Subtelomeric deletions represent an important class of abnormalities to be considered when investigating genetic links to intellectual disability (ID). One subtelomeric deletion found on the long arm of chromosome 11q produces a characteristic phenotype that includes ID and is often referred to as Jacobsen syndrome (JBS). Previously, researchers found an inverse relationship between IQ and deletion size. While useful, IQ does not provide a comprehensive picture of the cognitive-behavioral strengths and weaknesses in JBS, nor does it reveal how the profiles evolve as these individuals age. One purpose of this study was to confirm the relationship between IQ or adaptive behavior (DQ) and deletion size. We also examined cognitive-behavioral profiles of children with JBS and the extent to which they changed over time. Initially, at T1, we examined 10 children, ages 5-20 years, diagnosed with JBS. Cognitive ability was assessed with the Stanford-Binet (4th Edition). Adaptive behavoir was evaluated with the Vineland Adaptive Behavior Scales (VABS). Eight children were reassessed 2 years later (T2). Results show a negative but non-significant correlation between IQ and deletion size. There was no statistically significant relationship between DQ and deletion size. As for our second aim, IQ and DQ scores were stable from T1 to T2.

Cognitive profiles were not significantly different from T1 to T2. However, there were significant changes in adaptive behavior domain scores from T1 to T2. Lack of a significant relationship between cognitive-behavioral measures and deletion size, as well as changes in cognitive-behavioral profiles are discussed.

Fisch GS. Cognitive-behavioral characteristics and developmental trajectories in children with deletion 11qter (Jacobsen syndrome), and their relation to deletion size. Am J Med Genet A. 2015 Jan;167A(1):45-53. doi: 10.1002/ajmg.a.36837. Epub 2014 Nov 25. PMID: 25425441.