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Developmental Trajectories for a Sibling Pair with Chromosome 15q11.2-13.1 Duplication Syndrome and Angelman Syndrome

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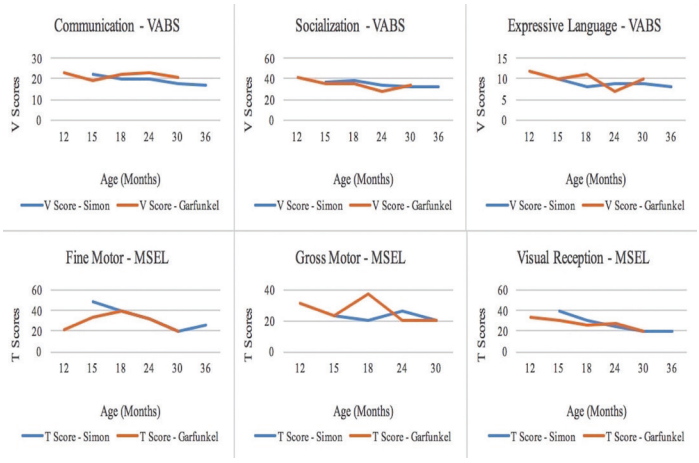
Several types of copy number variants, or a surplus or absence of genetic information encoded by DNA, appear in the 11.2-13.1 region of chromosome 15. Chromosome 15q11.2-13.1 duplication syndrome (dup15q) is caused by duplications of chromosome 15q11.2-13.1. Angelman syndrome derives from a deletion of the mother's genetic contribution from the 11.2-13.1 region of chromosome 15. To date, no studies have documented early childhood development for individuals with dup15q and Angelman syndrome. Here we present two unique case studies of brothers Simon and Garfunkel, who have dup15q (Simon) and dup15q *and* Angelman syndrome (Garfunkel). Garfunkel, therefore, has both a duplication and a deletion of parts of his genome in the specified chromosomal region.

Each child's developmental progress was tracked using the Vineland Adaptive Behavior Scales (VABS) and the Mullen Scales of Early Learning (MSEL) when the boys were 12 to 36 months of age. The VABS assessment seeks to measure the social and daily living skills of individuals, through the perspective of someone who is familiar with their typical day-to-day behavior, such as a parent. In contrast, the MSEL is administered by a trained examiner and indexes their language (expressive and receptive), motor (gross and fine), and visual reception skills.

We completed a thorough lateral analysis of and comparison between the VABS and MSEL results obtained for each of the boys. Because little is currently known about how children with these syndromes develop, a research question evolved out of the privilege of having two boys with these genomic copy number variants participating in an ongoing prospective study. Through the study, we discovered that by 30 or 36 months of age, both Simon and Garfunkel had global developmental delays across the domains of language (receptive and expressive), motor (gross and fine), visual reception, socialization, and daily living. However, as illustrated in the figure, the progression of these varied by developmental domain and did not differ significantly across Simon and Garfunkel.

Thus, while Garfunkel has an additional deletion on his maternal chromosome 15, it is clear, as indexed by the results of his VABS and MSEL assessments, that this additional genetic variant did not pose greater developmental risk. Specifically, it did not cause less developmental progress for Garfunkel during his first three years of life. At the examined ages, Simon and Garfunkel were developing fairly similarly and experiencing comparable deficits, despite having distinct genetic makeups.

Research advisor A. J. Schwichtenberg writes: "Ms. Sheehy and Ms. Nanninga delineated the developmental trajectories of two boys with copy number variants from a larger dataset of children developing at risk. Their question was savvy and unique—does multiple copy number variants on chromosome 15 confer greater developmental risk compared to only one copy number variant?"



Developmental trajectories for Simon and Garfunkel for select subscales on the VABS and the MSEL.

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