Partial monosomy of 11q22.2q22.3 including the SDHD gene in individuals with developmental delay

Abstract

Deletions in the middle portion of 11q are not as well described in the literature as terminal 11q deletions that result in Jacobsen syndrome. One confounding factor in the older literature is that the G-banding pattern of 11q13q21 is very similar to 11q21q23. The advent of fluorescence in situ hybridization and later microarray technologies have allowed for a better resolution of many of these deletions, but genotype-phenotype correlations are still difficult since these deletions are rare events. We present five individuals who presented with developmental delays with de novo 11q22.2q23.3 deletions. Deletions were observed by standard G-banded chromosome analysis with clarification of breakpoints and gene content by SNP microarray analysis. Of note, all individuals had identical distal breakpoints. All deletions include SDHD, which is implicated in hereditary paraganglioma/pheochromocytoma, for which the patients will need to be monitored in adulthood. In spite of the large deletions of 8.6 Mb (Patients 1 and 3), 13.98 Mb (Patient 2), and 12.6 Mb (Patients 4 and 5) all patients show somewhat mild intellectual disability and dysmorphism.

Yelavarthi K, Cabral H, Wilson GN, Rohena L, Risheg H, Penton A, Schleede J, Burnside RD. Partial monosomy of 11q22.2q22.3 including the SDHD gene in individuals with developmental delay. Am J Med Genet A. 2015 Apr;167A(4):695-700. doi: 10.1002/ajmg.a.36956. Epub 2015 Mar 3. PMID: 25735893.