GENETICA Y BIOLOGIA MOLECULAR

A Peruvian Child with 18p-/18q+ Syndrome and Persistent Microscopic Hematuria.

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Abstract

Chromosome 18 pericentric inversion carriers could have offspring with recombinant chromosomes, leading to patients with clinical variable manifestations. Patients with 18p-/18q+ rearrangements share some clinical characteristics, while other characteristics differ. Factors for such divergence include the length of the inverted segment, among others. Here, we describe a Peruvian child with dysmorphic features, intellectual disability persistent microscopic hematuria, aortic pseudocoarctation, and descending aorta arteritis, among others. Karyotype analysis of family members determined the mother as the carrier of a pericentric inversion: 18[inv(18)(p11.2q21.3)]. This child carries a recombinant chromosome 18, with chromosomal microarray analysis detecting two genomic imbalances in patient's chromosome 18: one duplicated region and one deleted segment in the large and the short arms, respectively. Persistent microscopic hematuria has not been reported among 18p-/18q+ phenotypes. Our patient elucidates that other factors play significant and yet unknown roles for not fulfilling the proposed genotype-phenotype correlation associated with hemizygosity in this type of recombinant chromosome 18 or presenting these features as the patient ages.