# Clinical and molecular-cytogenetic evaluation of a family with partial Jacobsen syndrome without thrombocytopenia caused by an approximately 5 Mb deletion del(11)(q24.3)


## Abstract

Clinical manifestations of Jacobsen syndrome (JBS) depend on the size of the 11qter deletion, which usually varies between approximately 7 and 20 Mb. Typical JBS features include developmental delay/mental retardation, short stature, congenital heart defects, thrombocytopenia, and characteristic dysmorphic facial features. We report on a family in which a 4-year-old girl as well as her mother and maternal uncle present with subtle features of JBS. Notably, neither thrombocytopenia nor congenital anomalies were detected in this family. Cytogenetic analyses revealed normal karyotypes. Using fluorescence in situ hybridization (FISH) and whole-genome oligonucleotide array CGH analyses, we identified an approximately 5 Mb deletion of the terminal part of chromosome 11q in all the three affected family members. The deletion breakpoint was mapped between 129,511,419 and 129,519,794 bp. This is the smallest deletion reported in a JBS patient. Interestingly, the FLI1 (friend leukemia virus integration 1) hematopoiesis factor gene located approximately 6.5 Mb from 11qter and usually

deleted in patients with JBS, is intact. Our data support previous hypotheses that FLI1 haploinsufficiency is responsible for thrombocytopenia in patients with JBS.

Bernaciak J, Szczałuba K, Derwińska K, Wiśniowiecka-Kowalnik B, Bocian E, Sasiadek MM, Makowska I, Stankiewicz P, Smigiel R. Clinical and molecular-cytogenetic evaluation of a family with partial Jacobsen syndrome without thrombocytopenia caused by an approximately 5 Mb deletion del(11)(q24.3). Am J Med Genet A. 2008 Oct 1;146A(19):2449-54. doi: 10.1002/ajmg.a.32490. PMID: 18792974.