SNP array and phenotype correlation shows that FLI1 deletion per se is not responsible for thrombocytopenia development in Jacobsen syndrome

Abstract

Jacobsen syndrome (JBS) is a rare chromosomal disorder caused by terminal deletion of the long arm of chromosome 11. We report on four prenatally diagnosed patients with JBS with variable prenatal and postnatal phenotypes and 11q deletions of varying sizes. Precise characterization of the deleted region in three patients was performed by SNP arrays. The severity of both the prenatal and postnatal phenotypes did not correlate with the size of the haploinsufficient region. Despite the large difference in the deletion size (nearly 6 Mb), both of the live-born patients had similar phenotypes corresponding to JBS. However, one of the most prominent features of JBS, thrombocytopenia, was only present in the live-born boy. The girl, who had a significantly longer deletion spanning all four genes suspected of being causative of JBS-related thrombocytopenia (FLI1, ETS1, NFRKB, and JAM3), did not manifest a platelet phenotype. Therefore, our findings do not support the traditional view of deletion size correlation in JBS or the causative role of FLI1, ETS1, NFRKB, and JAM3 deletion per se for the development of disease-related thrombocytopenia.

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