



**DGAP006: 46,XX,t(1;2)(p32;q11)**

**Revised: 46,XX,t(1;2)(p32.3;q13)**

This case was obtained from the NIGMS Human Genetic Cell Repository (NIGMS [GM04300](#)). Features include mental retardation, developmental delay, aggressiveness, no speech, frontal bossing, epicanthal folds, unilateral ptosis, low-set ears, and no binocular fixation. We have cloned and sequenced the breakpoint on the der(2) and have delineated the breakpoint on the der(1). The latter disrupts the single strand DNA binding protein (*SSBP3*) between exons 4 and 5, while the former disrupts *FLJ14681* between exons 6 and 7. No fusion gene is expected because the directions of transcription are oppositely oriented.

Interestingly, *C. elegans* fed with a *SSBP3* homolog in an RNAi experiment are reported to have poor movement and frequent lysis, the latter of which has been hypothesized to indicate a defect in vulva formation. Because one allele of each locus is disrupted, haploinsufficiency is a possible pathogenic mechanism. Members of the *SSBP* gene family are evolutionarily conserved modulators of differentiation. Despite an unknown function, *FLJ14681* also is a good candidate because it is expressed in fetal and adult brain. The 2q13 region has been linked with familial autistic disorders.

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