



DGAP162: 46,XX,t(2;11)(p25.1;p15.1)

This ten year-old female has severe developmental delay, occasional self-injurious behavior, seizures, partial agenesis of the corpus callosum and delayed myelination, per MRI. EEG identified a biparietal disturbance and an epileptic discharge during sleep. FISH experiments refined the chromosome 2 breakpoint to less than 150 kb flanked by clones RP11-93D24 and AC092177. One predicted protein (LOC339789) lies within this region. The 11p breakpoint is localized to BAC RP11-764H14, and no known genes lie within this region, although the 3' terminus of SOX6 maps ~250 kb downstream.

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