

**DGAP106: 46,XX,t(3;5)(q27;q31.1),t(11;13)(p15.3;q14.1)**

This is a 2 year-old female with developmental delay, behavioral issues including self-injurious actions and agitation, failure to thrive and growth retardation. Weight, length and head circumference were all in the lowest 5% for the age range. Strabismus and ptosis were present. Prenatal ultrasound showed a "short" fetus with an apparently normal weight. FISH mapping of the 3q breakpoint narrowed the break to 5.5 Mb, between RP11-114M1 and RP11-259I19, but also identified a deletion of at least 1.5 Mb, between the BACs RP11-355N16 and RP11-510K16, inclusive. This deletion encompasses several genes, including *WIG1*, *USP13* and *PEX5L*. Further FISH is underway to delineate the breakpoint and the deletion boundaries. Subsequently, the 5q breakpoint was narrowed to ~800 kb between RP11-4E3 and RP11-233E5, with deletion of at least 500 kb, including clones RP11-21C10 and RP11-21J3. The 11p breakpoint was mapped to a 1.5 Mb interval bound by RP11-651M4 and RP5-859D17, with *METT5D1* and *KIF18A* as the only known genes within the region. A split clone, RP11-307O13, has been identified at the 13q breakpoint, and no known genes map within this breakpoint interval. Due to the karyotypic complexity involved, further experiments on this case have been discontinued.

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