



DGAP019: 46,XX,t(2;13)(p13;q32)

Revised: 46,XX,t(2;13)(p12;q31)

This female has severe mental retardation and, while she is ambulatory, she lacks any interaction with her environment. Dysmorphic features include a hawk-like nose, small head, thin and dystrophic, long extremities and fingers. FISH mapping of the 2p12 breakpoint has narrowed the region to an approximately 1 Mb interval between RP11-502O9 and RP11-451C8 and includes one gene, *DHFRP1*. The 13q31 breakpoint has been narrowed to a 100 kb region containing no known genes. *SPRY2* and several of the *SLITRK* genes are near the breakpoint.

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