

**DGAP131: 46,XX,t(1;5)(p22;q13)**

This female is dysmorphic, developmentally delayed, and has cerebral palsy and a seizure disorder. She is hypotonic and has abnormal findings by MRI (increased white matter) and EEG studies, though she has had normal EMG and nerve conduction studies. Additionally, she was tested for Angelman and Prader-Willi syndromes, for which the results were negative. The 5q13 breakpoint in this seizure disorder case is very close to the 5q14-q15 locus linked to febrile and idiopathic generalized epilepsy {Nakayama, 2000 #104}, {Durner, 2001 #106}. Although a candidate gene, *MASS1*, in this region has been proposed, the mutation studies suggested that it is not likely to contribute to the cause of febrile seizures in the majority of the families with linkage to 5q14 {Nakayama, 2002 #105}. This finding strongly suggests the possibility of the presence of another gene in the region that could be responsible for the seizure phenotype. Interestingly, *MASS1* is disrupted in DGAP127, an individual in whom seizures have been reported. Our FISH mapping of the 5q13 breakpoint has narrowed the breakpoint region to ~100 kb. Several ESTs map within the breakpoint region, but no expression of these ESTs was identified in the DGAP131 cell line based on RT-PCR experiments using combinations of primers.

A potential double hit case, DGAP191, with the breakpoint in 5q14 and an overlapping phenotype (seizures) is being co-investigated. The breakpoint in DGAP191 has been narrowed to ~2 Mb.

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