

DGAP170: 46,XX,t(1;19)(p34.1;p13.1)

This individual suffers from generalized epilepsy, developmental delay, behavioral problems, and fluctuating ataxia. The case was ascertained through a poster presentation at ASHG, in which British neurologist, Dr. Tolmie, described an individual with a *de novo* translocation and epilepsy of unexplained etiology. Our analysis of the breakpoint on chromosome 1 showed that the break does not disrupt any known genes, but occurs about 50 kb from gene *SLC2A1* (Fig. DGAP170-1). This gene encodes GLUT1, a major glucose transporter in blood-brain barrier, and its mutations cause an autosomal dominant GLUT1 deficiency syndrome. This syndrome could have variable severity with the most constant features being seizures, delayed mental and motor development, deceleration of head growth, and ataxia. The proximity of the breakpoint to the *SLC2A1* gene and the overlap between clinical presentation of GLUT1 deficiency syndrome and the clinical phenotype of DGAP170 suggest a translocation-related position effect on the expression of *SLC2A1*. Dr. Tolmie has been contacted with our recommendation to test the patient for GLUT1 deficiency. If the diagnosis is confirmed, it would lead to an important change in the patient's management as a ketogenic diet is proven to be beneficial in controlling epilepsy in GLUT1 deficient patients (most of these patients are refractory to anticonvulsants). If the patient is indeed GLUT1 deficient, our next step would be confirmation of position effect and a search for an upstream regulatory element. This case in particular illustrates the benefit of DGAP research to a participant's diagnosis and management.

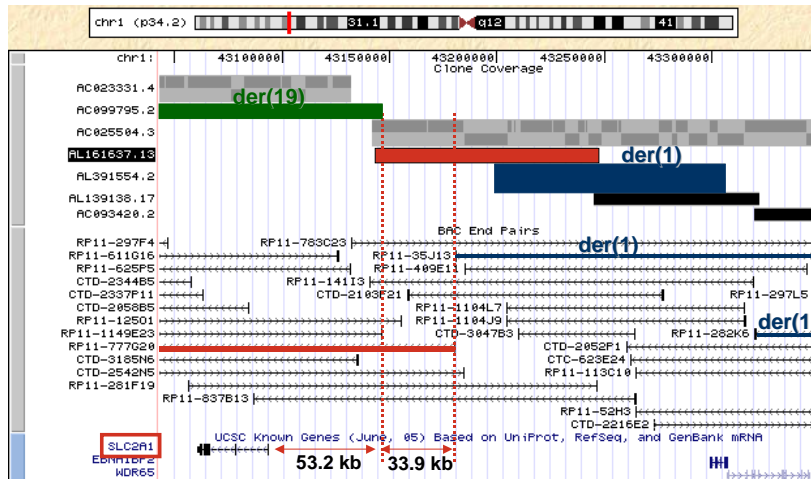


Figure DGAP170-1. Map of the 1p34.1 Breakpoint Region of DGAP170. This figure was obtained from the genome browser at <http://genome.ucsc.edu>. BAC clone hybridizing to der(19) is colored in green, BAC clones hybridizing to der(1) are colored in blue, and clones that split between both derivatives are colored in red. The vertical dashed lines enclose the breakpoint region (narrowed to 33.9 kb). A red square marks gene *SLC2A1* that is located 53.2 kb from the breakpoint region.