



DGAP180/181: **46,XY,t(2;11)(q31;q13.5)pat**

Revised: 46,XY,t(2;11)(q32;q14)pat

The individual has developmental delay, speech and language difficulties, sensory impairment and has been diagnosed with Tourette syndrome, OCD/ADHD/ODD, sensory integration disorder, and an arachnoid cyst. His father, who also carries the rearrangement, suffers from tics, attention issues, anxiety, learning difficulties and speech difficulties as a child. FISH experiments narrowed the 2q breakpoint to within the BAC AC112720. This breakpoint appears to disrupt the predicted gene *C2orf10*, a gene of unknown function and expression. Additionally, FISH on the 11q breakpoint has identified a 1.5 Mb breakpoint region, between RP11-89H11 and RP11-141H6. Several genes map within this region and one of them, *GRM5* is an attractive candidate for further study. This gene encodes a glutamate receptor precursor; L-glutamate is the major excitatory neurotransmitter in the central nervous system and activates both ionotropic and metabotropic glutamate receptors. Knock-out mice {Lu, 1997 #227} that lack this gene show impairment in the acquisition and use of spatial information. Electrophysiologic measurements indicated that long-term potentiation in the mutant mice was significantly reduced in the NMDA receptor-dependent pathways, whereas LTP remained intact in the mossy fiber synapses on the CA3 region, an NMDA receptor-independent pathway. As such, *GRM5* may play an important role in spatial learning and memory.

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