Identification of a novel gene on chromosome 7q11.2 interrupted by a translocation breakpoint in a pair of autistic twins

Sultana, Razia

Yu, Chang En

Yu, Jun

Munson, Jeffery

Chen, Donghui

Hua, Wenhui

Estes, Annette

Cortes, Fanny

De La Barra, Flora

Yu, Dongmei

Haider, Syed T.

Trask, Barbara J.

Green, Eric D.

Raskind, Wendy H.

Disteche, Christine M.

Wijsman, Ellen

Dawson, Gera

We report here the identification and characterization of a novel gene (AUTS2) that spans the 7q11.2 breakpoint in a monozygotic twin pair concordant for autism and a t(7;20) (q11.2; p11.2) translocation. AUTS2 is 1.2 Mb and has 19 exons. The predicted protein is 1295 amino acids and does not correspond to any known protein. DNA sequence analysis of autism subjects and controls revealed 22 biallelic polymorphic sites. For all sites, both alleles were observed in both cases and controls. Thus no autism-specific mutation was observed. Association analysis with two exonic polymorphic sites and linkage analysis of four dinucleotide repeat markers, two within and two

flanking AUTS2, was negative. Thus, although it is unlikely that AUTS2 is an autism susceptibility gene for idiopathic autism, it may be the gene responsible for the disorder in the twins studied here.