

Supplemental Information

Parent of Origin Effect Analysis

We analyzed possible parent of origin effect (POE) for the SNP rs2710102 using PLINK (<http://pngu.mgh.harvard.edu/~purcell/plink/>). This is a family-based TDT analysis. Transmissions from heterozygous fathers and mothers to affected offspring were estimated separately.

	"A" allele Transmitted:Nontransmitted	CHISQ	<i>p</i> -value	POE	POE
				Z-score	<i>p</i> -value
Paternal	20.5:13.5	1.441	0.2299		
Maternal	24.5:12.5	3.892	0.04852		
				0.517	0.6052

Although the maternal side finding is significant taken by itself, the difference between maternal and paternal is not significant in this small sample of 37 heterozygote parents.

Note: When both father and mother are heterozygous, the transmissions are ambiguous and counted as 0.5 for both mother and father. This led to non integer Transmitted:Nontransmitted counts.

Sex Effect Analysis

The sample was separated by retaining only boys or girls within each family. We then conducted sex effect analysis using FBAT by analyzing the boys and girls separately. rs2710102 was nominally significant (*p*-value = 0.041) for the boy group, but all other tests, including single SNP analysis and haplotype analysis for both boy and girl groups were clearly nonsignificant.