

**Supplementary Table 1.** Family Haplotype.

Microsatellite analysis of two STS markers flanking the amplified region, located at 5' and 3' of *SOX9*, shows that the probands share the same paternal haplotype for the region.

<b>Locus</b>	<b>Position (Kb)</b>	<b>P1</b>	<b>P2</b>	<b>S</b>	<b>M</b>	<b>(F)</b>
D17S807	62289	124/124	124/124	120/122	120/124	122/124
D17S939	72973	205/207	205/207	205/209	205/209	205 or 209/207

**Supplementary Table 2.** *SOX9* expression in lymphoblasts from patient and age matched controls. Quantitative real time PCR was performed using TaqMan assay on Demand (Applied Biosystems) following manufacturer's instruction and using Mx3000P apparatus.

	<b><i>SOX9</i>/β actin*§</b>	<b><i>SOX9</i>/GAPDH*§</b>
Patient	0.443±0.116	0.464±0.046
Control (n=6)	2.066±1.313	1.787±1.141
P value‡	0.08	0.14

\* Plus-minus values are means ± SD.

§ The relative expression of each mRNA was normalized to β actin and glyceraldehyde-3-phosphate dehydrogenase expression respectively, and then compared with the relative mRNA expression of the controls. This test was performed in triplicate on three separate patient samples.

‡ Ttest was used to evaluate P values.

**Supplementary Table 3.** Amplicon positions on chromosome 17 (bp)

RT1	chr17:67004087-67004176
RT2	chr17:67021968-67022040
RT3	chr17:67114802-67114877
RT4	chr17:67016545-67016601