Amplification of NOTCH1 and ABL1 gene loci is a frequent aberration in enteropathy-type T-cell lymphoma.

We have shown previously that amplification of chromosomal region 9q34 is the most frequent aberration in enteropathy-type T-cell lymphoma (ETL). To determine the minimum amplified 9q34 region and identify possible candidate gene(s), we performed a detailed microsatellite screening and quantitative real-time PCR (QPCR) on 26 ETL cases. Microsatellite analysis revealed allelic imbalance in both ABL1 and NOTCH1 gene loci (microsatellites D9S290-D9S1847 and D9S158 flanking the former and latter genes, respectively) localized in the band 9q34. The results were confirmed by TaqMan-based QPCR showing amplification of ABL1 and NOTCH1 exons in 50% and 65% of cases, respectively. Amplifications of the NOTCH1 gene were more frequent than of the ABL1 gene; moreover, the analyzed NOTCH1 exon consistently displayed higher levels of amplification than ABL1 coding sequences. From 9q34 known genes, NOTCH1 could thus be the primary target of genomic DNA amplification in ETL.