Mutational analysis: DNA was extracted using a QIAamp DNA Blood Mini Kit (Qiagen, MD, USA). Genomic DNA samples were amplified using primers for each exon, as previously described (11). Amplified products were gel-purified using a QIAEX II Gel Extraction Kit (Qiagen). Sequencing reactions were performed using a GenomeLab DTCS Quick Start Kit (Beckman Coulter, CA, USA) with a cycle sequencing protocol, while sequencing reactions were separated on a CEQ 8000 Genetic Analysis System (Beckman Coulter). Splicing aberrations were tested using an immortalized cell line that was established from the patient by infecting with Epstein-Barr virus (EBV) (12) obtained from B95-8 cells. Total RNA was extracted from the EBV-immortalized lymphoblastoid cell line using a QIAamp RNA Blood Mini Kit (Qiagen) and subjected to reverse transcription (RT)-PCR using a Long-Range 2-Step RT-PCR Kit (Qiagen) with oligo dT. The forward and reverse primers for exons 2 and 6 were 5′-GCTGAGAGCGAAGTTTCAGA-3′ and 5′-CCAGGAATTCCAAAGGGTCGAAG-3′, respectively.