Mitotic recombination and compound-heterozygous mutations are predominant NF1-inactivating mechanisms in children with juvenile myelomonocytic leukemia and neurofibromatosis type 1

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Online Supplementary Figure S1. (A) Short tandem repeat analysis of chromosome arm 17q in 10 leukemic samples from children with JMML and NF-1. The numbers in boxes indicate fragment lengths detected (base pairs). Gray indicates heterozygosity; black, homozygosity. The genomic position of each marker (Mbp, million base pairs) is indicated. The genomic position of the NF1 gene on chromosome 17 is at 26.66 Mbp (arrow). (B) Comparative genomic hybridization of chromosome arm 17q in JMML samples D341 and D566. A DNA array containing >236,000 probes was used. A proximal segment of 17q, spanning the genomic region from 25.2 million base pairs (Mbp) to 27.9 Mbp, is shown. The data indicate heterozygous deletions with the same proximal breakpoints at 26.023 Mbp in D341 (top) and D566 (bottom) and distal breakpoints at 27.351 Mbp and 27.386 Mbp. The vertical line indicates the position of the NF1 gene at 26.66 Mbp.
Online Supplementary Figure S2. Schematic representation of the NF1 gene and its protein domain structure. Mutations of patients are indicated above the protein structure. Exon numbers are based on GenBank accession number NM000267.1. Exon numbers in parentheses are according to Fahsold et al. **NF1 mutations described in the literature (Refs. 13-17 in the manuscript).