

**Figure S2. Results of MLPA of the 9p region in patient A, TS and parents.** Results obtained with MLPA arrays for the 9p21 region were ordered according to the genomic location of the probes. Gene dosage quotients for the 41 probes and relative ID numbers are shown, for patient A and TS in full bars, and for parents in empty bars. The deletion detected in the two sisters extends from *CDKN2B* to *MLLT3* genes, and includes *CDKN2A*, *MTAP*, *IFNA1*, *KLH9*, *IFNW1*, and *IFNB1* genes; in contrast, *ELAVL2* and *TEK* centromeric to *CDKN2B* and *GLDC* and *DOCK8* telomeric to *MLLT3* showed normal gene dosage quotients indicating retention of both gene copies.

