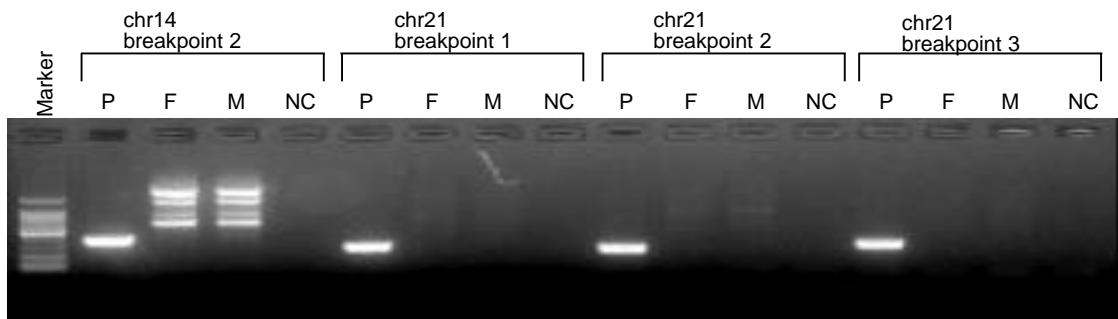


Additional file 2: Fig. S1 G-banding of the present patient. Arrows point chromosomes with aberrant structure.



Additional file 2: Fig. S2 Representative results of the PCR-amplification of DNA fragments containing the fusion junctions. The numbers of the breakpoints correspond to those of Fig. S3. P, F, M, and NC denote the patient, father, mother, and negative control, respectively.

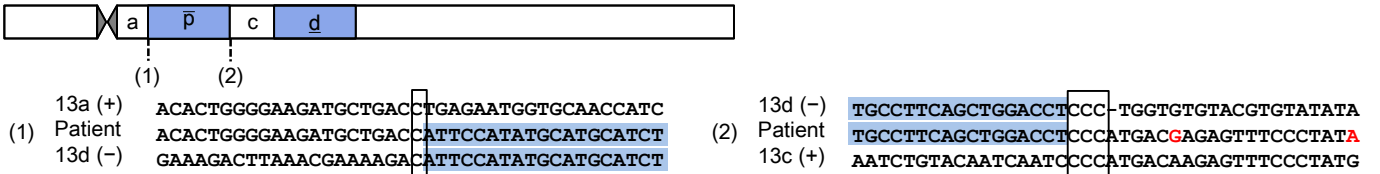
chromosome 2



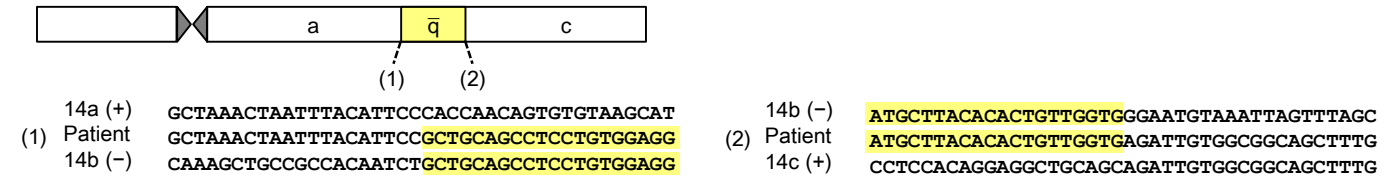
chromosome 6



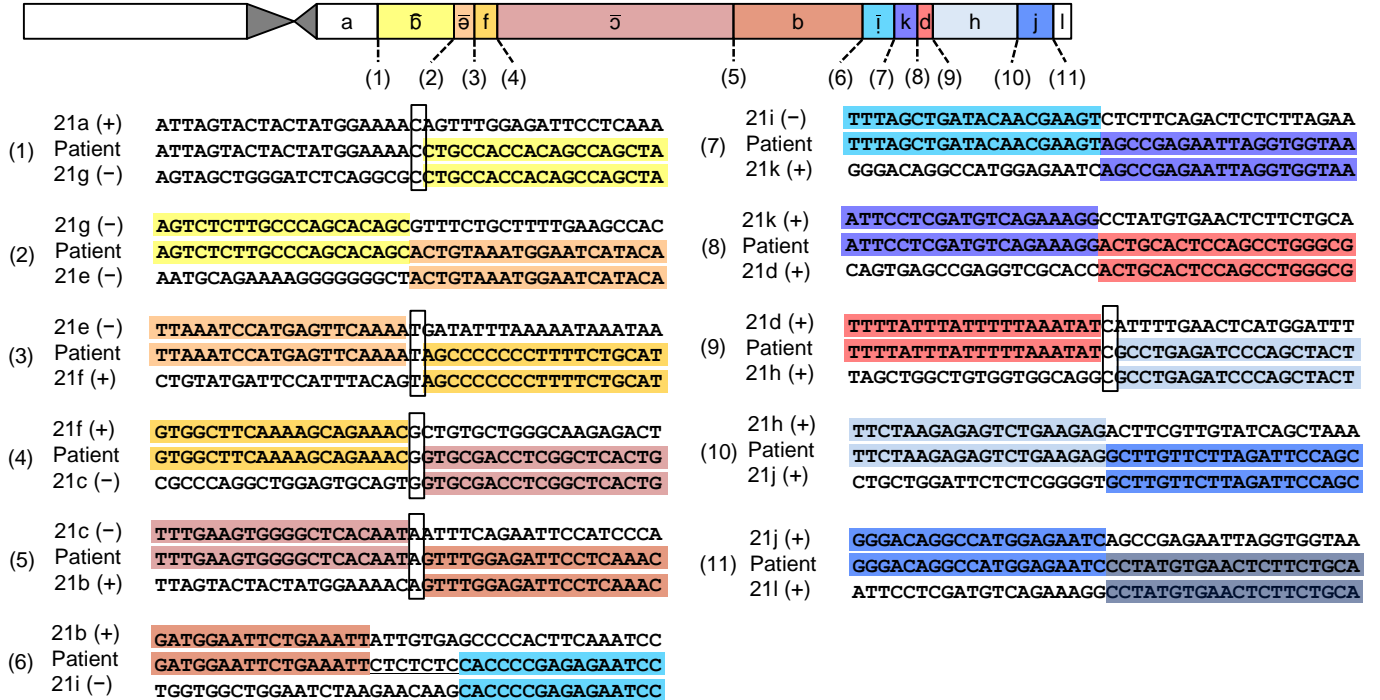
chromosome 13



chromosome 14



chromosome 21



Additional file 2: Fig. S3 Breakpoint structures of the rearrangements. Sequences of the patient are aligned with the human reference data (GRCh37 on Ensemble Genome Browser 91, http://grch37.ensembl.org/Homo_sapiens/Info/Index). Microhomologies at the fusion junctions are shown in boxes. An untemplated insertion is underlined. The (+) and (-) symbols indicate forward (CEN → q-ter) and reverse (q-ter → CEN) directions, respectively. Red letters on chromosome 13 denote single nucleotide variants shared by the patient and his father.