IRF4-BLOC1S5: the first rearrangement gene identified in TEMPI syndrome

Authors

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Supplemental Figure 1

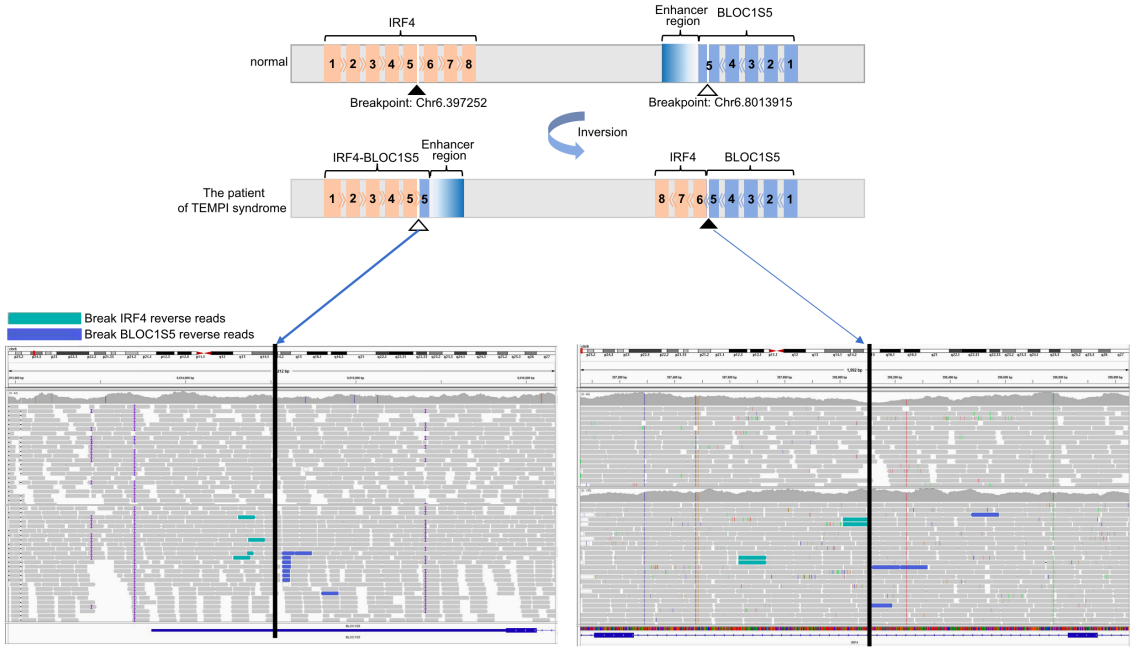

Figure S1. **Mate pair sequencing data of the patient**. Mate pair sequencing data of the patient was analyzed and the orientation of the mate pair reads is consistent with a chromosomal inversion on chromosome 6 between *IRF4* and *BLOC1S5*.

Supplemental Figure 2

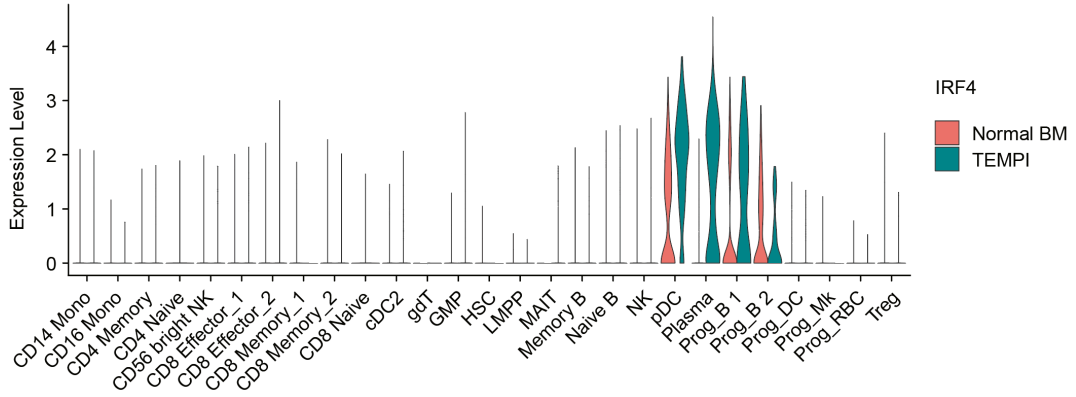

Figure S2. Expression level of IRF4 in different cell clusters. The bone marrow mononuclear cells (BMNCs) from normal donors and the patient were clustered by cell markers by single cell RNA sequencing. IRF4 was extremely highly expressed in plasma cell of the patient compared with that of normal donors.

Supplemental Figure 3

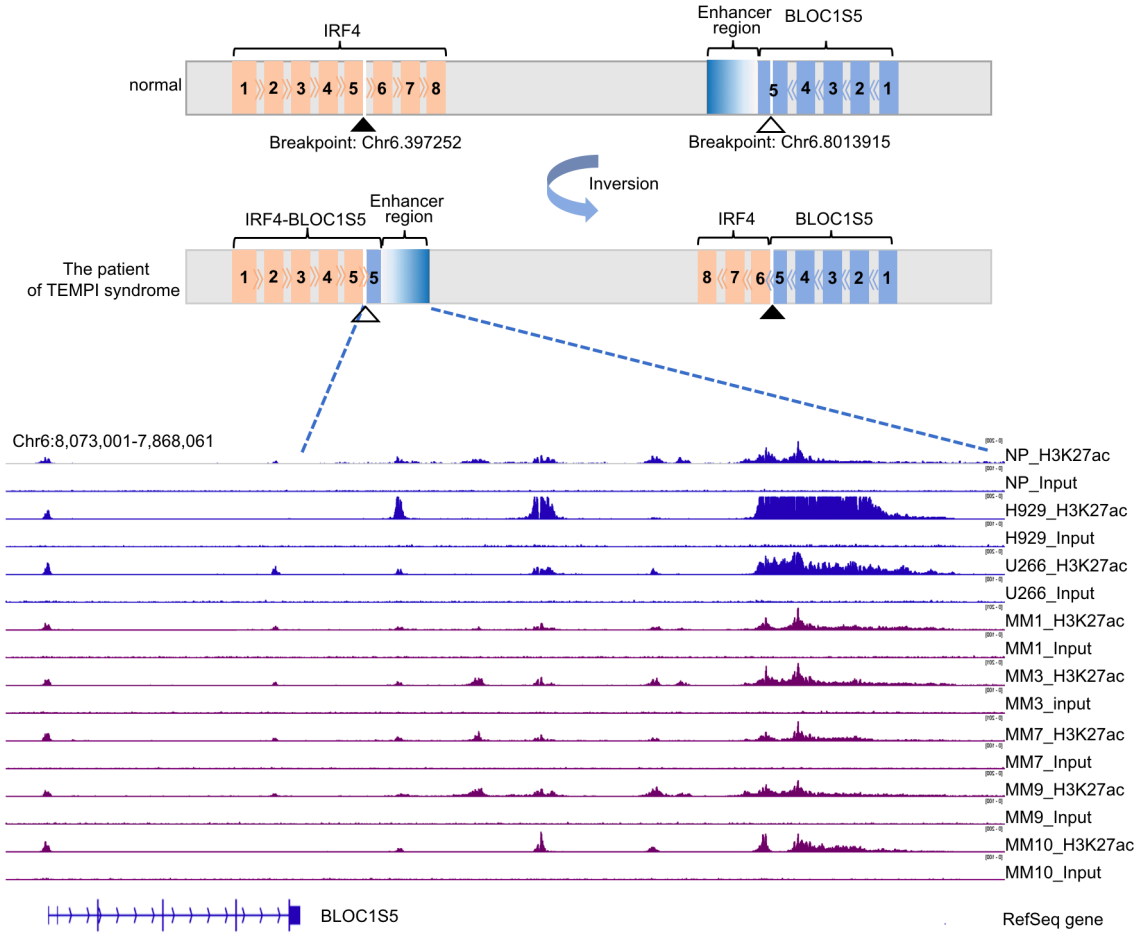

Figure S3. Integrative Genomics Viewer genome browser tracks show the level of H3K27 acetylation (H3K27ac) at the *BLOC1S5* transcription stop site in Chromatin immunoprecipitation (ChIP) samples over input. NP stands for normal plasma cell, H929 and U266 are multiple myeloma (MM) cell lines, MM1, MM3, MM9 and MM10 are primary MM cases. (Data download from GSE145839).