Through this case we are highlighting the importance of the **Next generation Sequencing (NGS)** as a part of routine diagnostic workup for clinical samples of myeloproliferative neoplasms (MPNs). As exemplified by our data, NGS facilitates:

- The identification of low frequency and rare variants, including rare combination of mutations
- Broad molecular profiling; this is important since JAK2 testing is more prevalent and generally sequential single gene testing has been excluded in the case of these cancers
- Definite reduction in turnaround time and cost for diagnostic workup

## HISTOLOGICAL EVALUATION:

Bone marrow biopsy showed diffuse marrow fibrosis (Grade 2), and further molecular.

## MOLECULAR EVALUATION:

The whole blood EDTA sample was used for molecular diagnosis. The DNA was extracted from the peripheral blood using qiagen DNA extraction kit. The sample was processed by two different molecular techniques - real-time Polymerase Chain Reaction (RT-PCR) and Next Generation Sequencing (NGS).

**RT-PCR Results:**

The whole blood EDTA sample was used for molecular diagnosis.

**NGS Results:**

Myeloid panel NGS test was performed on the sample. Myeloid panel NGS tests for various hotspot genetic alterations such as SNVs and fusions in genes-ABL1, BRAF, CBL, CSF3R, DNMT3A, FLT3, GATA2, HRAS, IDH1, IDH2, JAK2, KIT, KRAS, MPL, MYD88, NPM1, NRAS, PTPN11, SETBP1, SF3B1, SRSF2, U2AF1, WT1, ASXL1, BCOR, CALR, CEBPA, ETV6, EZH2, IKZF1, NF1, PHF6, PRPF8, RB1, RUNX1, SH2B3, STAG2, TET2, TP53 and ZRSR2.