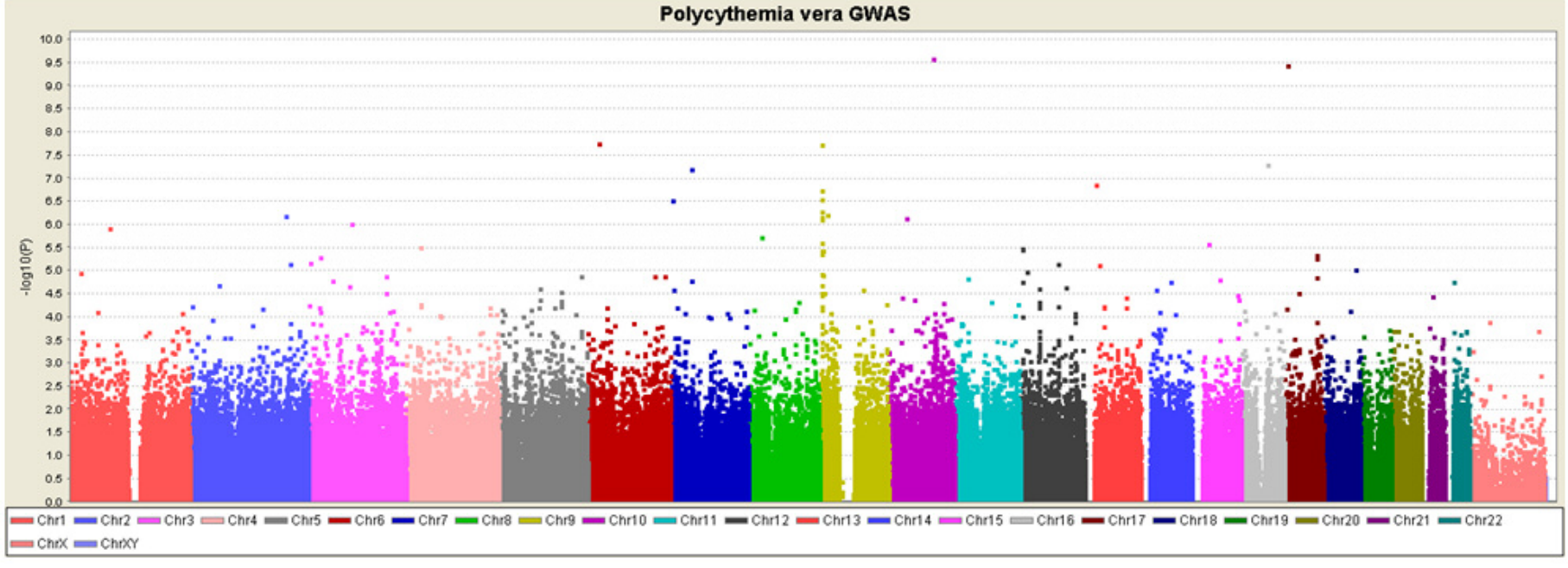
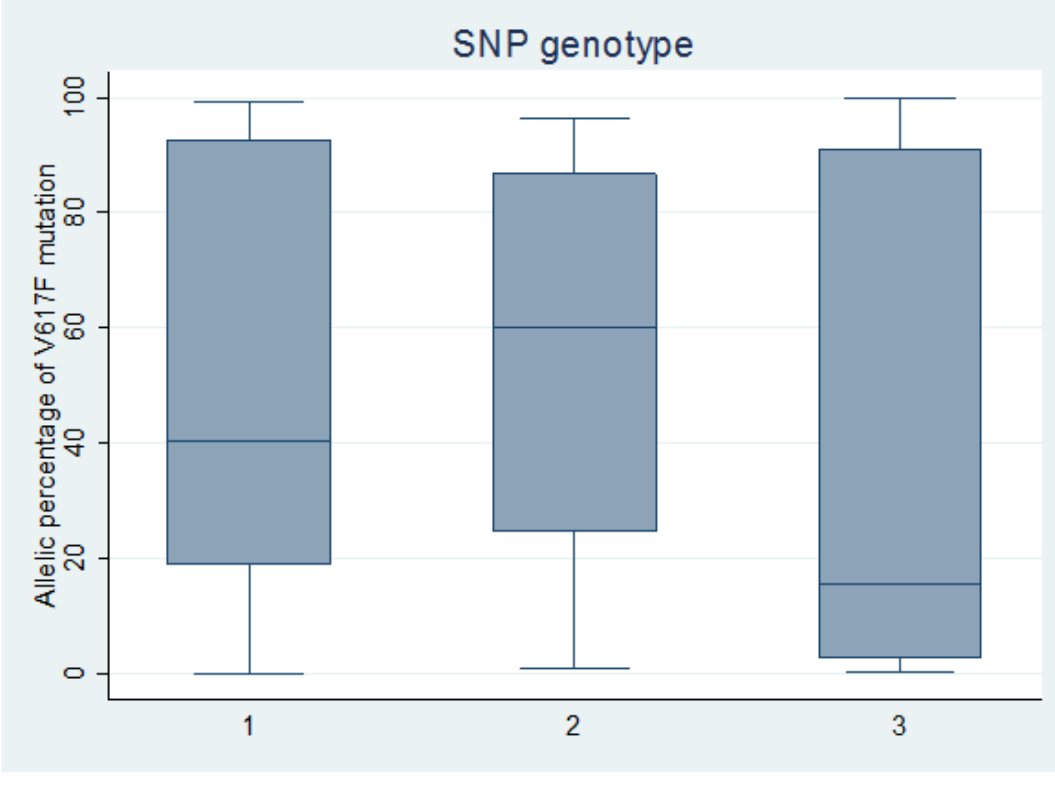
## Supplementary Figures

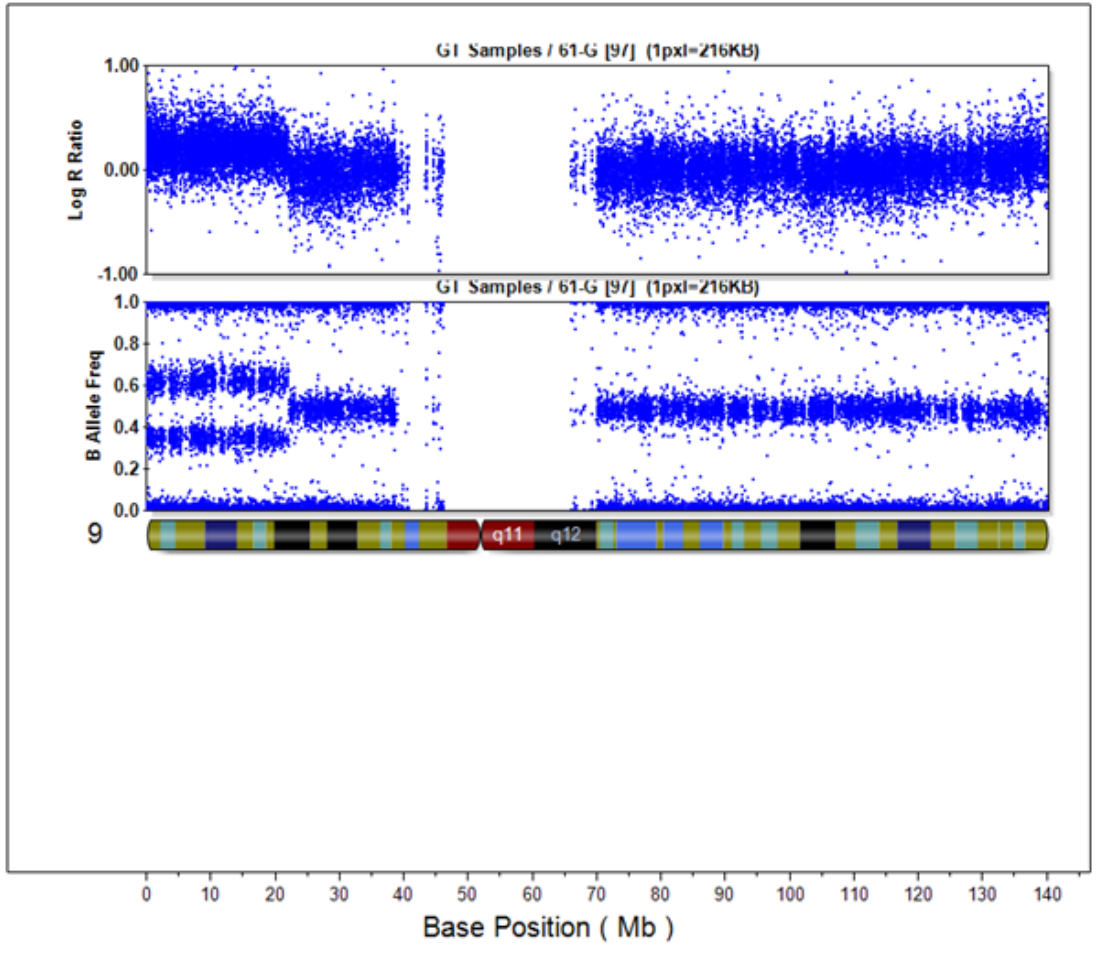
**Supplementary Figure 1**. Manhattan plot of the case/control association test on polycythemia vera. Multiple correlated SNPs surrounding JAK2 support association at the9p24.1 locus. Several isolated SNPs show strong association signals, which are due to random noise caused by small sample size.



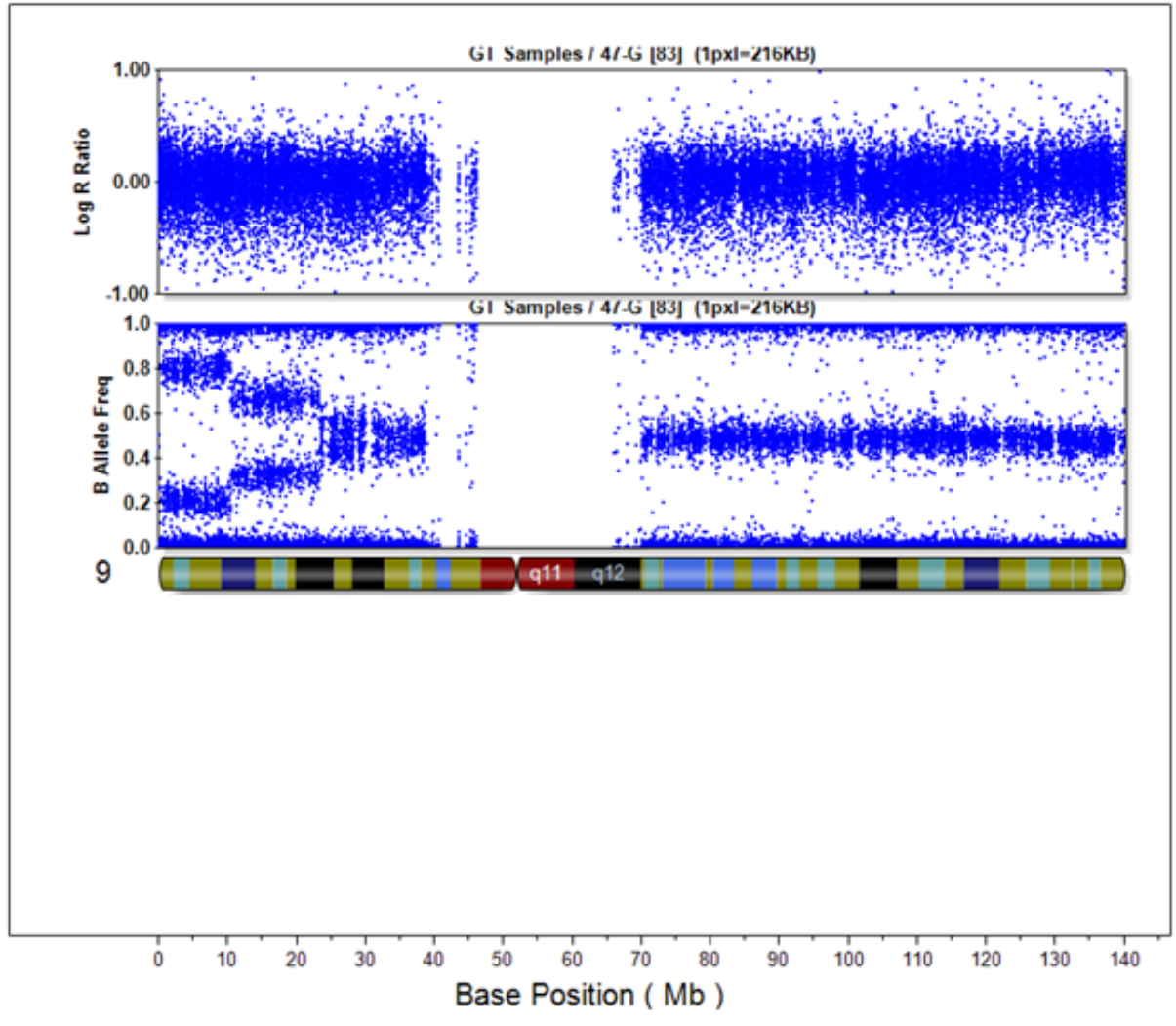
**Supplementary Figure 2**. Box plot of the percentage of V617F mutation in JAK2 over each of the three germline genotypes for rs11999802 in T-cells from 32 PV patients. No significant association was observed (P=0.75), possibly due to the small sample size. The SNP genotypes 1, 2, and 3 refer to homozygous non-risk alleles, heterozygotes, and homozygous risk alleles, respectively. Given the very wide range (large variance) of the allelic fraction and the relatively small sample size, the lack of statistically significant association is likely to be due to lack of power.



**Supplementary Figure 3**. Illustration of chromosomal duplications on chromosome 9p in granulocytes from one PV patient. Elevated Log R Ratio and splitting of B Allele Freq are hallmarks of chromosomal duplications.



**Supplementary Figure 4**. Illustration of stepwise chromosomal copy-neutral loss-of-heterozygosity on chromosome 9p in granulocytes from one PV patient. The Log R Ratio measures total signal intensity for two alleles, and it is centered around zero, indicating lack of copy number changes. The B Allele Freq measures allelic intensity ratios, with three distinct breakpoints, suggesting the presence of three distinct subclonal expansion events in this subject.



**Supplmeentary Figure 5**. A model for the stepwise events that occur during PV pathogenesis. Two homologous chromosomes with A and B germline alleles are depicted, and the red circle represents somatic V617F mutation.