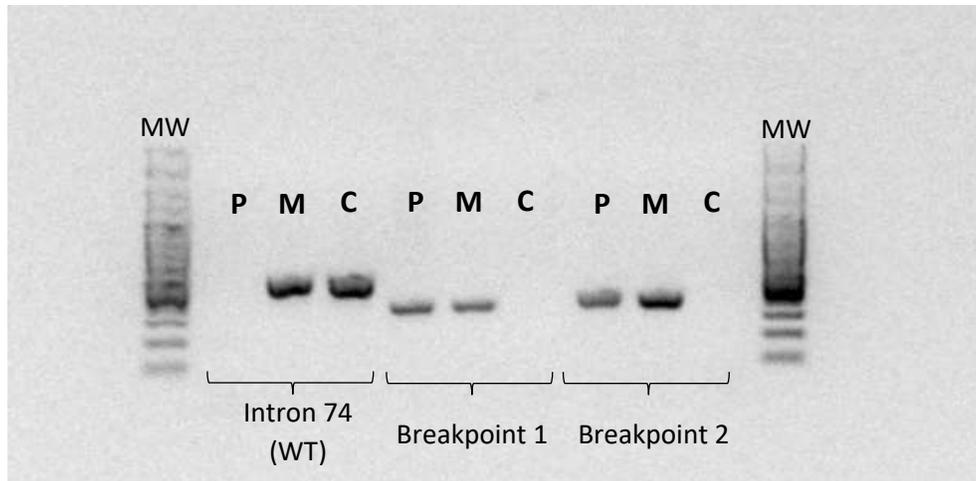


## Supplementary Material IV



**Family segregation studies.** PCR amplification of intron 74 reference sequence (WT) and both inversion breakpoints in patient (P), his mother (M) and a male control DNA (C), shows the presence of the inversion in the mother in a heterozygous state (MW- molecular weight marker).