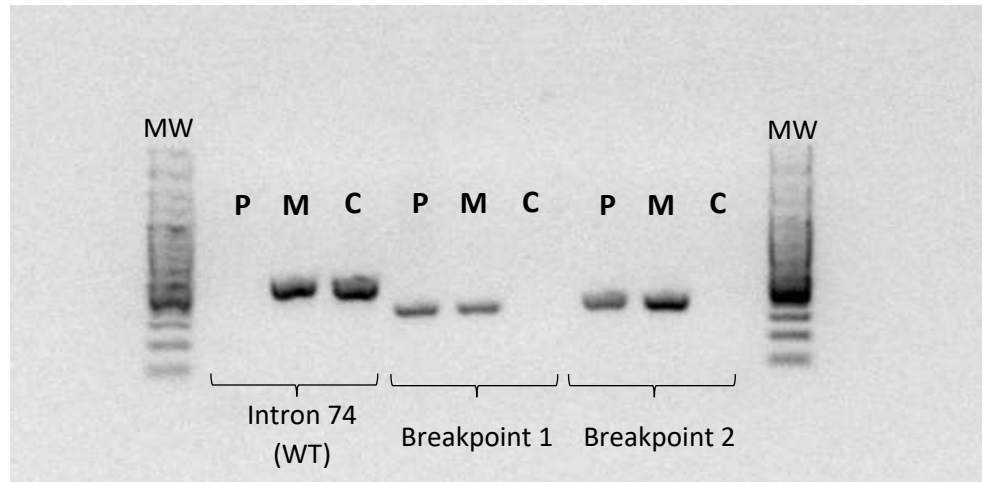


Supplementary Material IV



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).