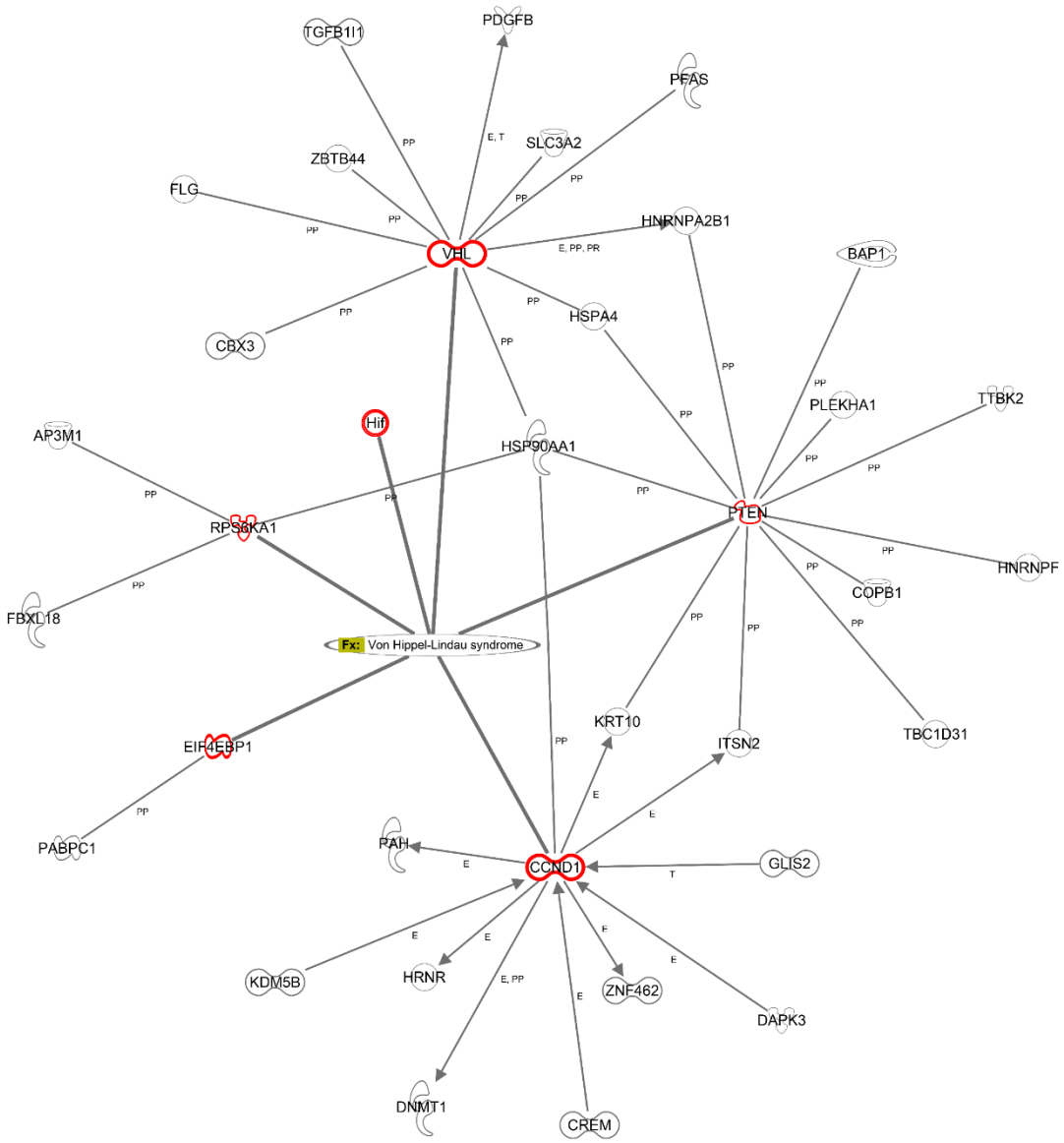
Supplementary Figure 1

GO analysis in VHL pathways for deleterious mutation detected in 22 unsolved NVI cases

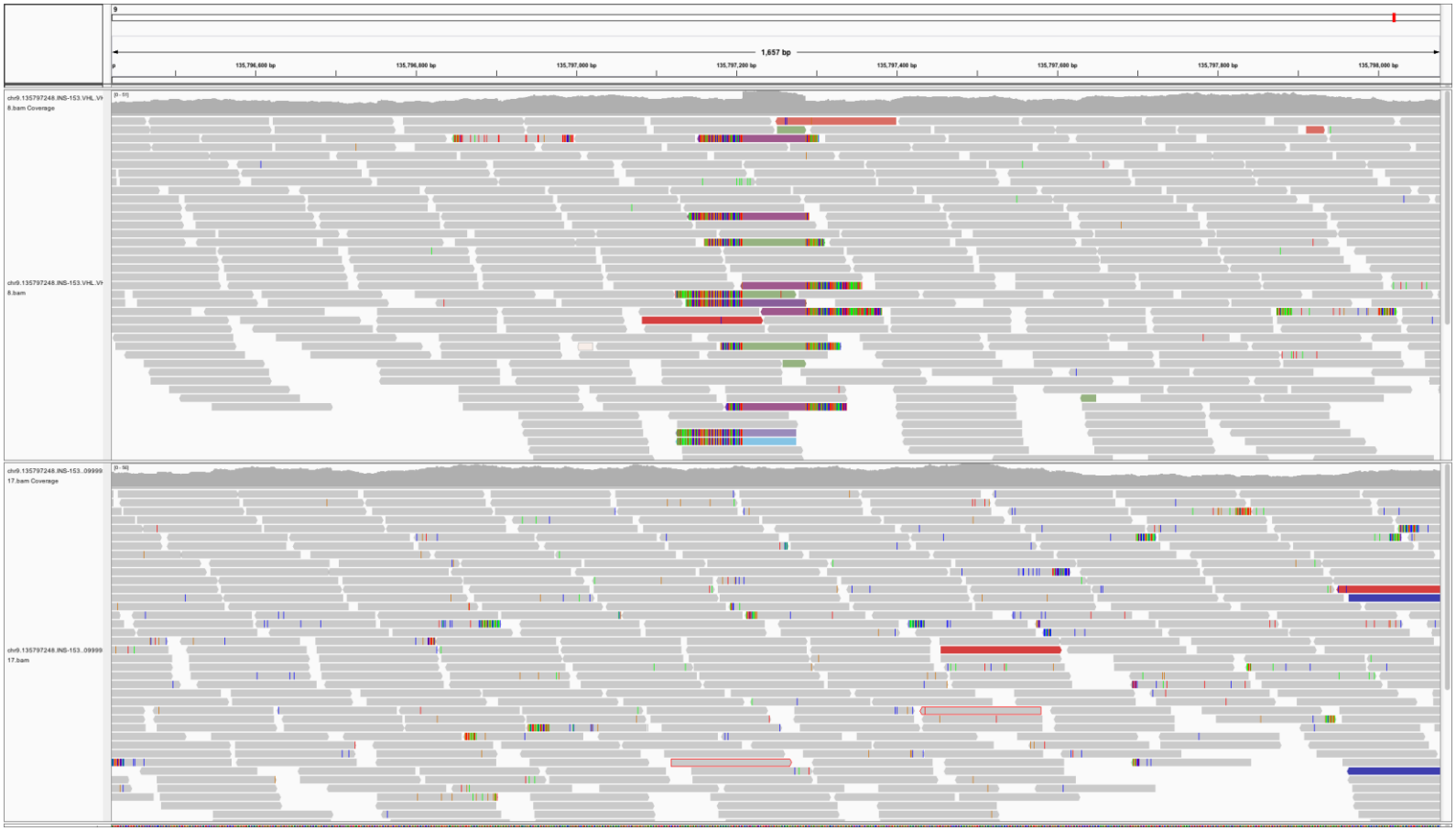


Supplementary Figure2. IGV shows a SV candidate of *TSC1* gene in VHL NVI case

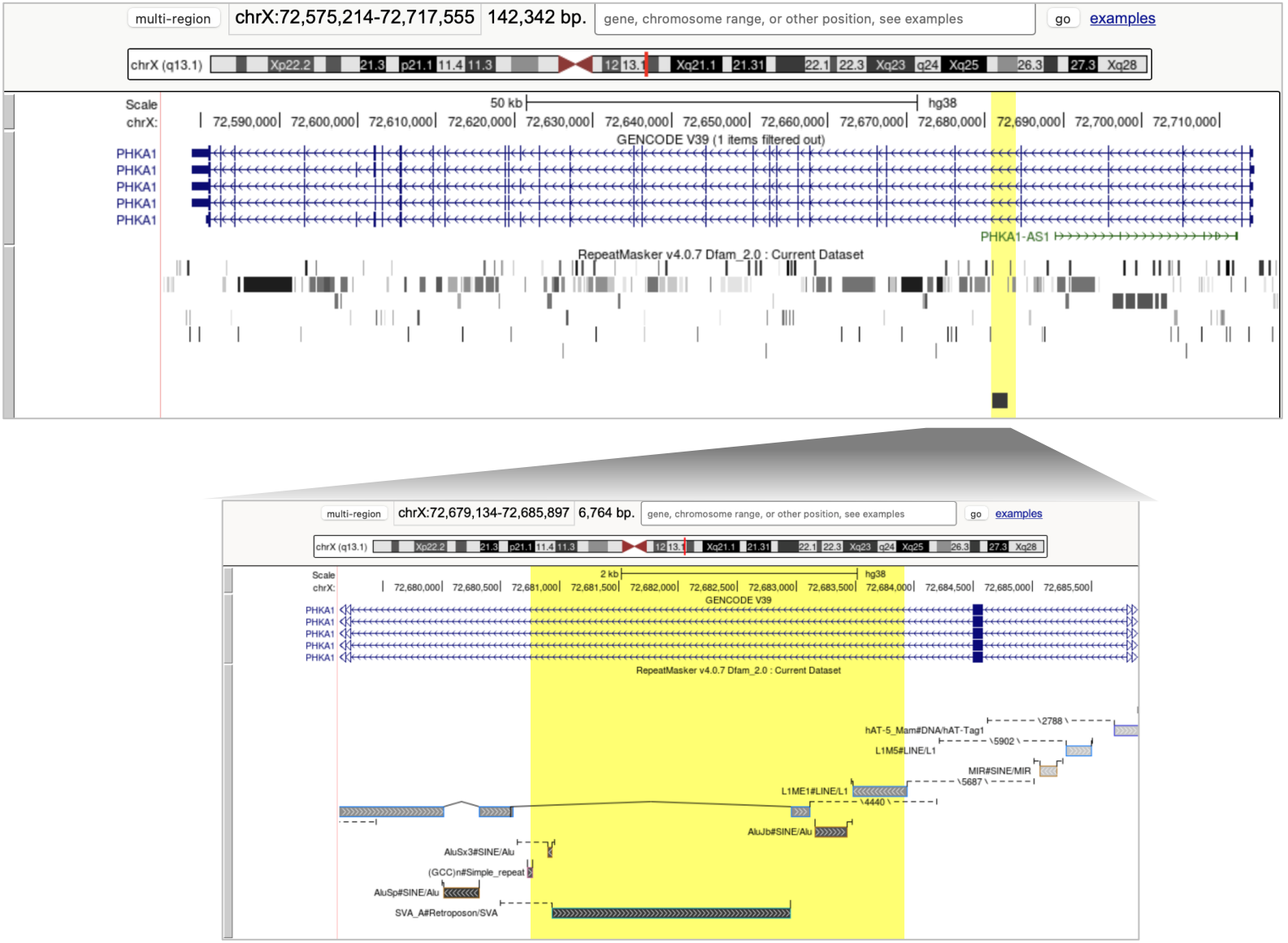
TSC1 exon7 (chr9:135797248 possible 153 bp insertion) in NIM Case08

VHL_08

BBJ: W18



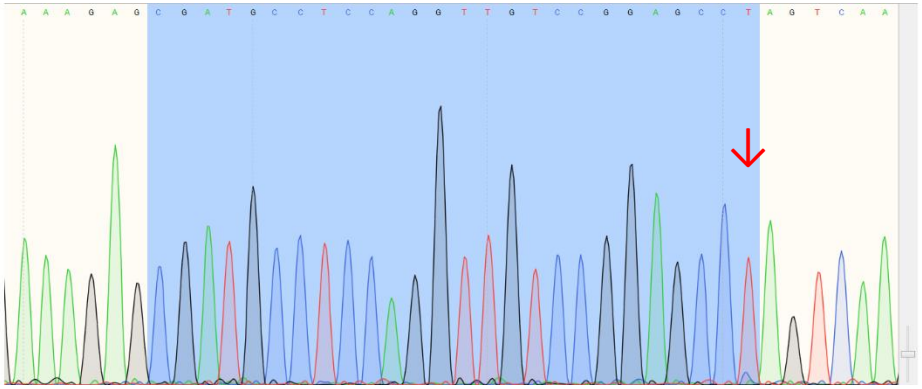
Supplementary Figure 3. ME insertion of *VHL* was derived from SVA in *PHKA1* intron at chromosome X.



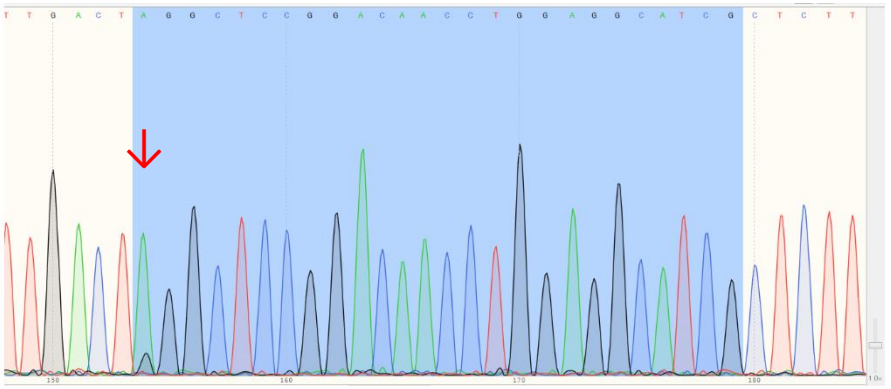
Supplementary Figure 4. Sanger sequencing validated mosaic mutations of *VHL* in NVI Case6 (c.T506C, AF 8.42%) and NVI Case15 (c.C481T, AF21.94%).

NVI Case6

Forward sequencing



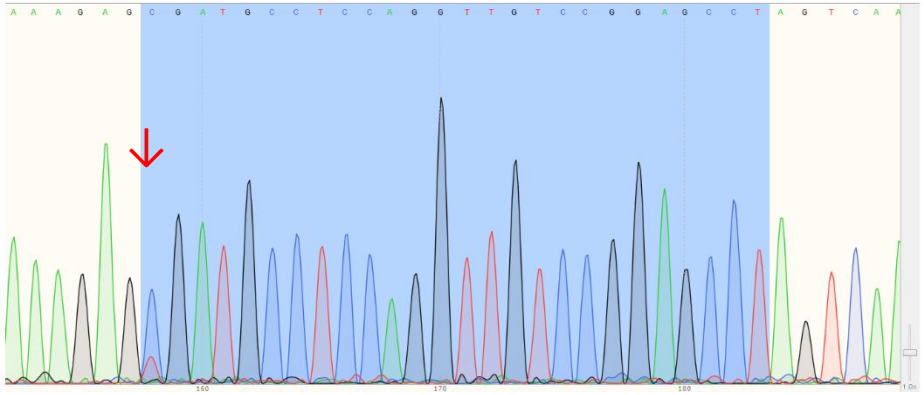
Reverse sequencing



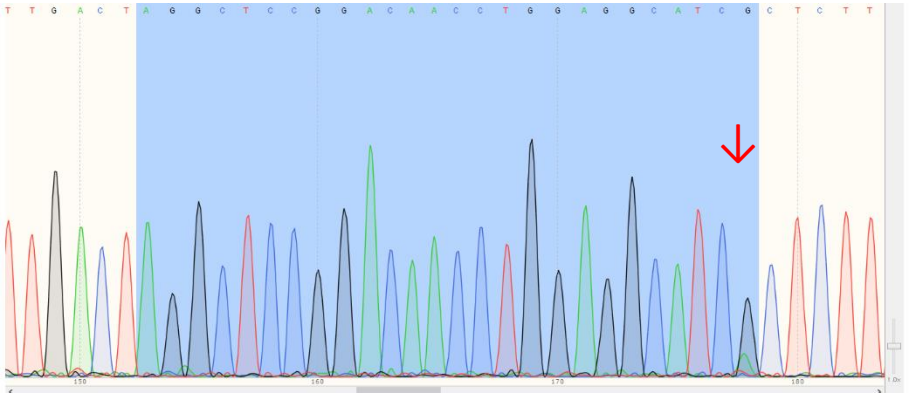
c.T506C

NVI Case15

Forward sequencing



Reverse sequencing



c.C481T