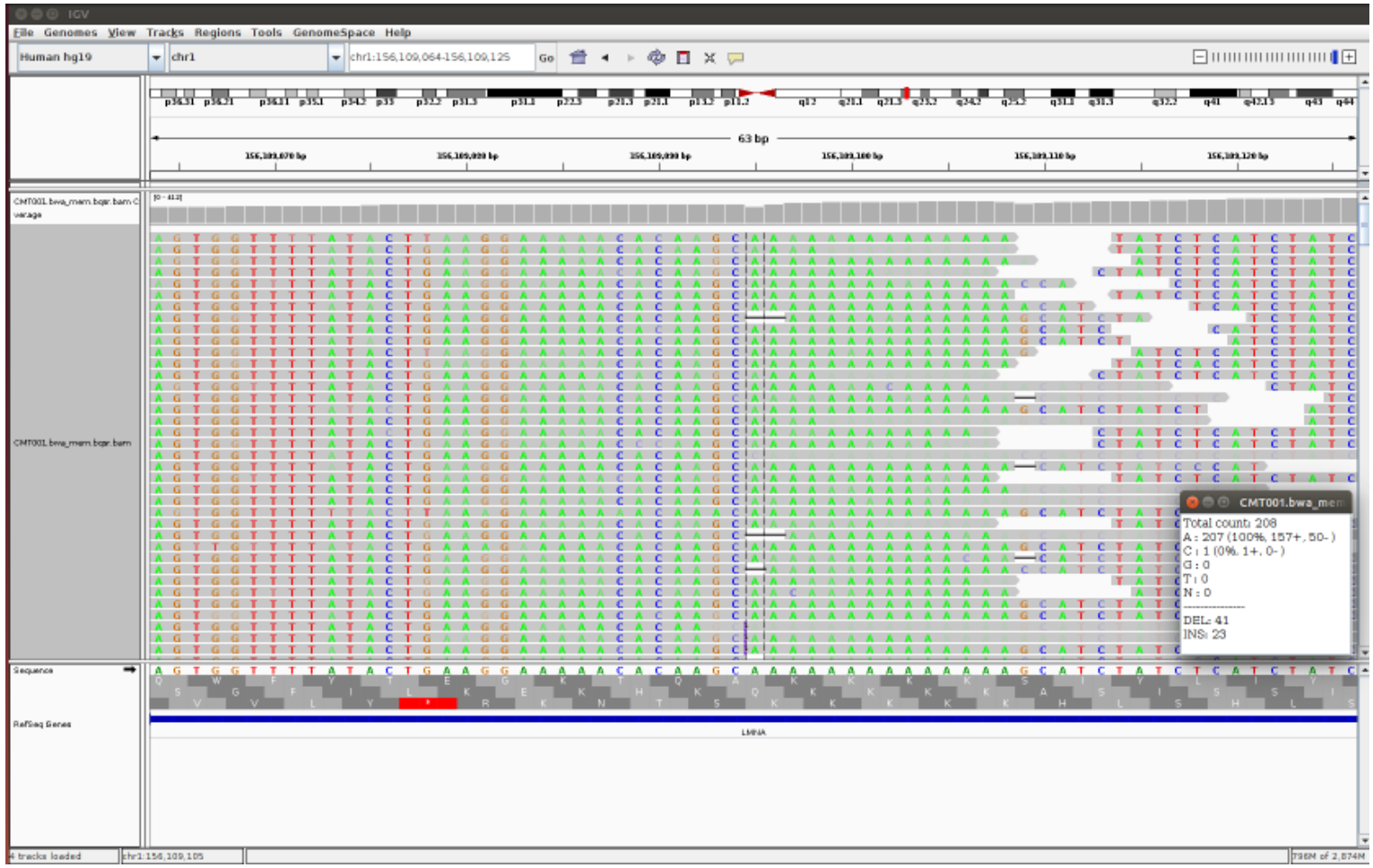
Supplementary material II

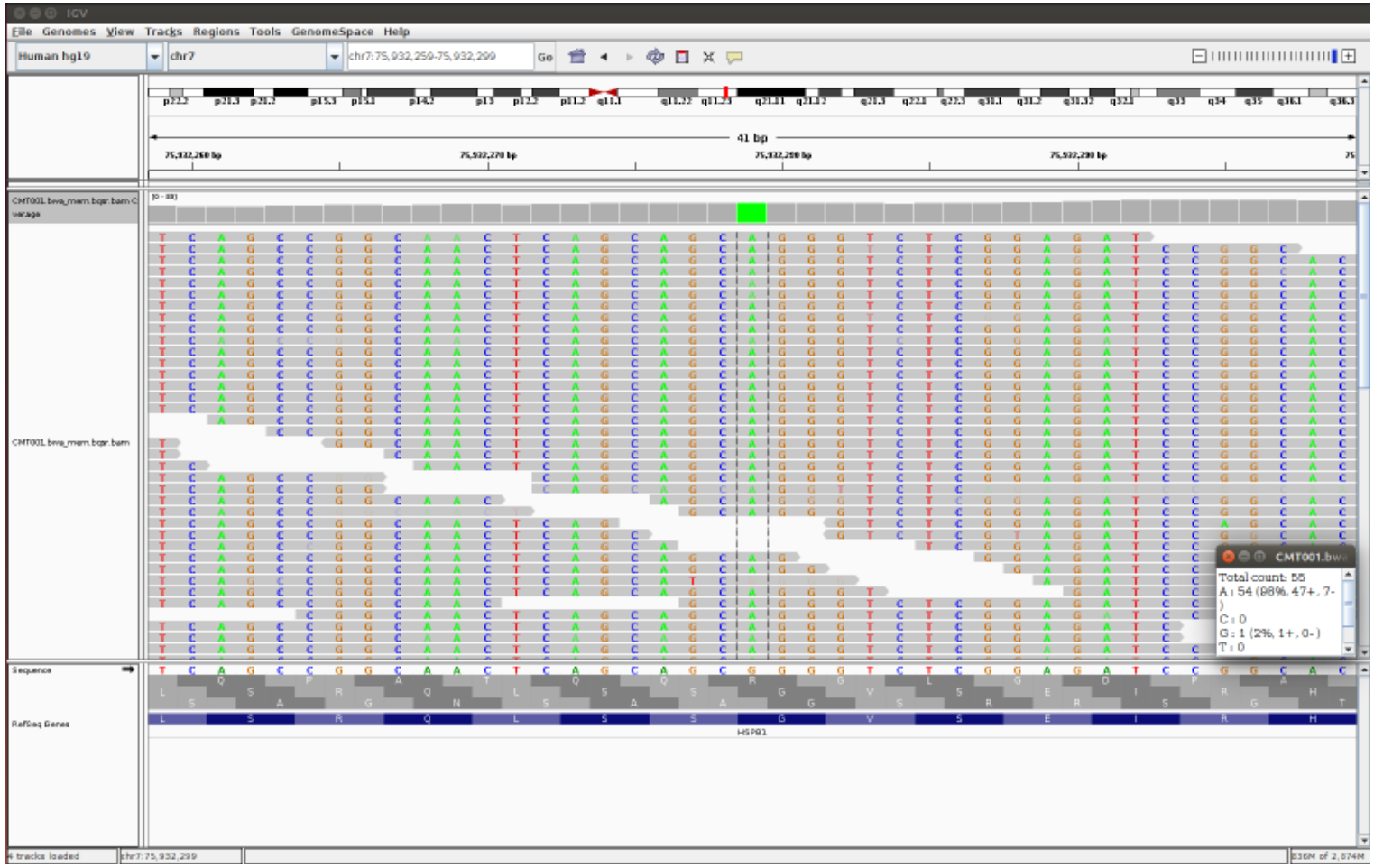
Manual verification of shortlisted variant calls

Case 1

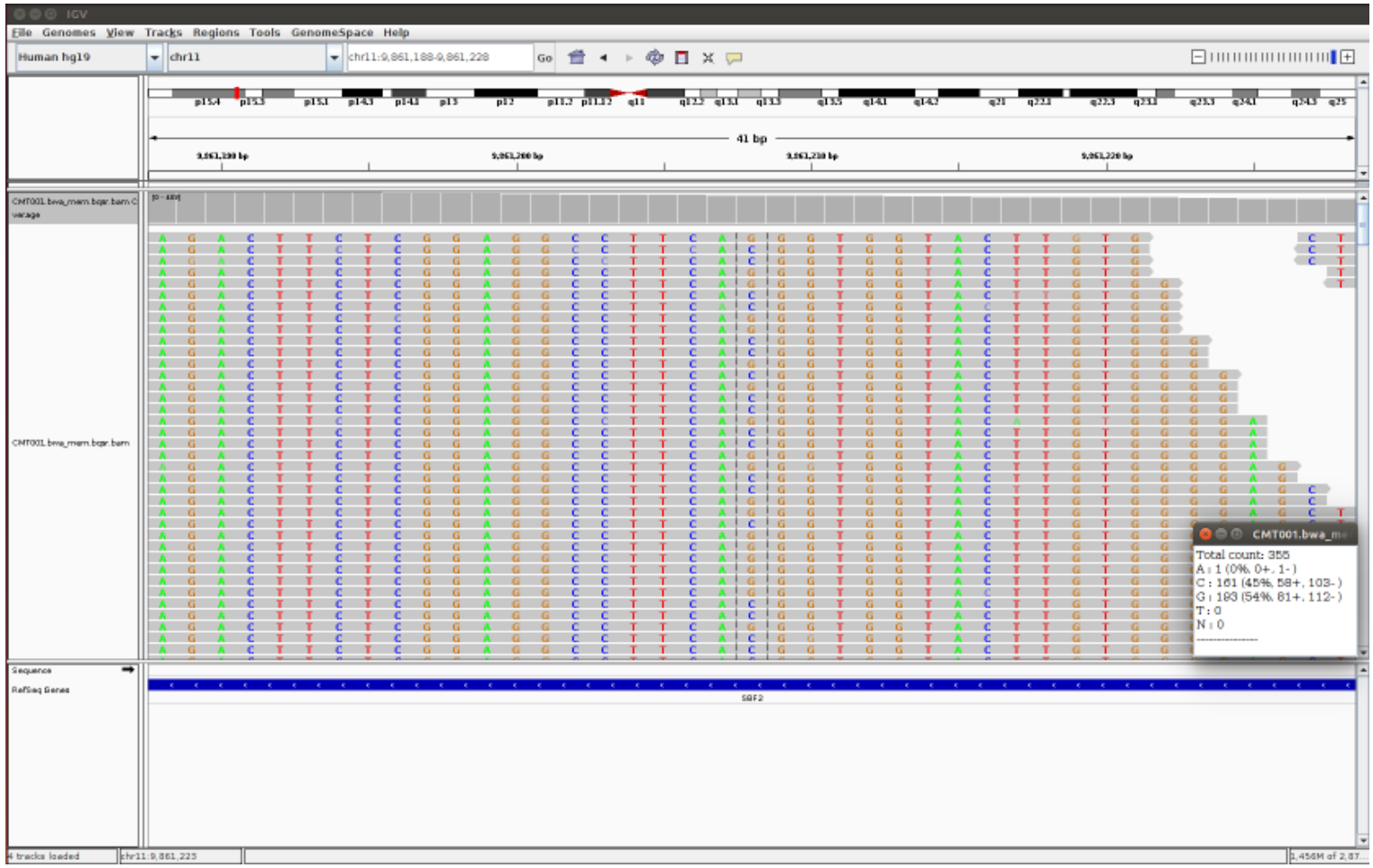
1. **chr1:156109095_156109095delA (likely sequencing error / miscalled variant)**
   LMNA:NM_170707:cDNA.2405_2405delA



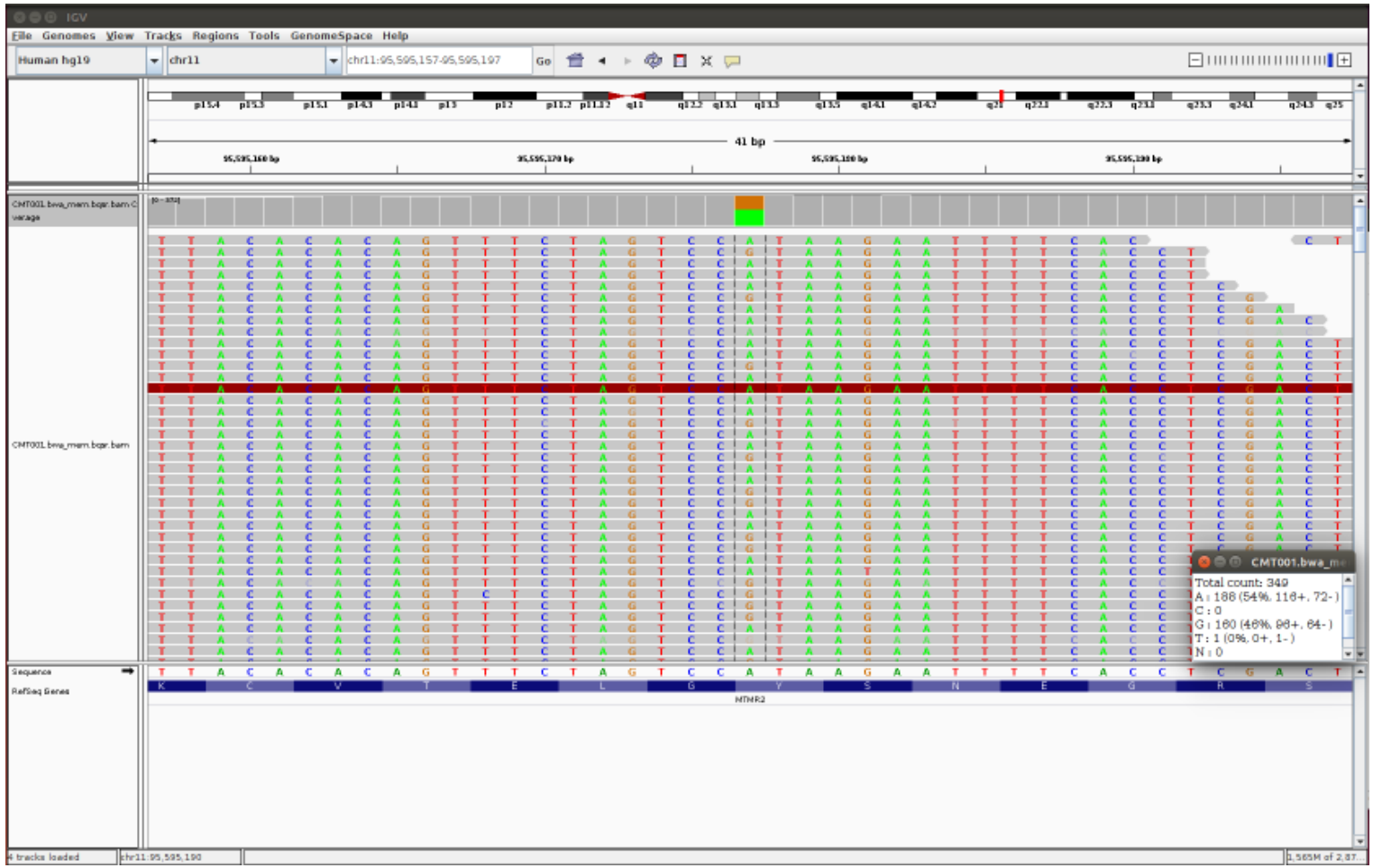
2. chr7:75932279G>A [Homozygous]
HSPB1:NM_001540:c.G250A(p.G84R)



3. chr11:9861208G>C [Heterozygous]
SBF2:NM_030962:c.C3292G(p.L1098V)

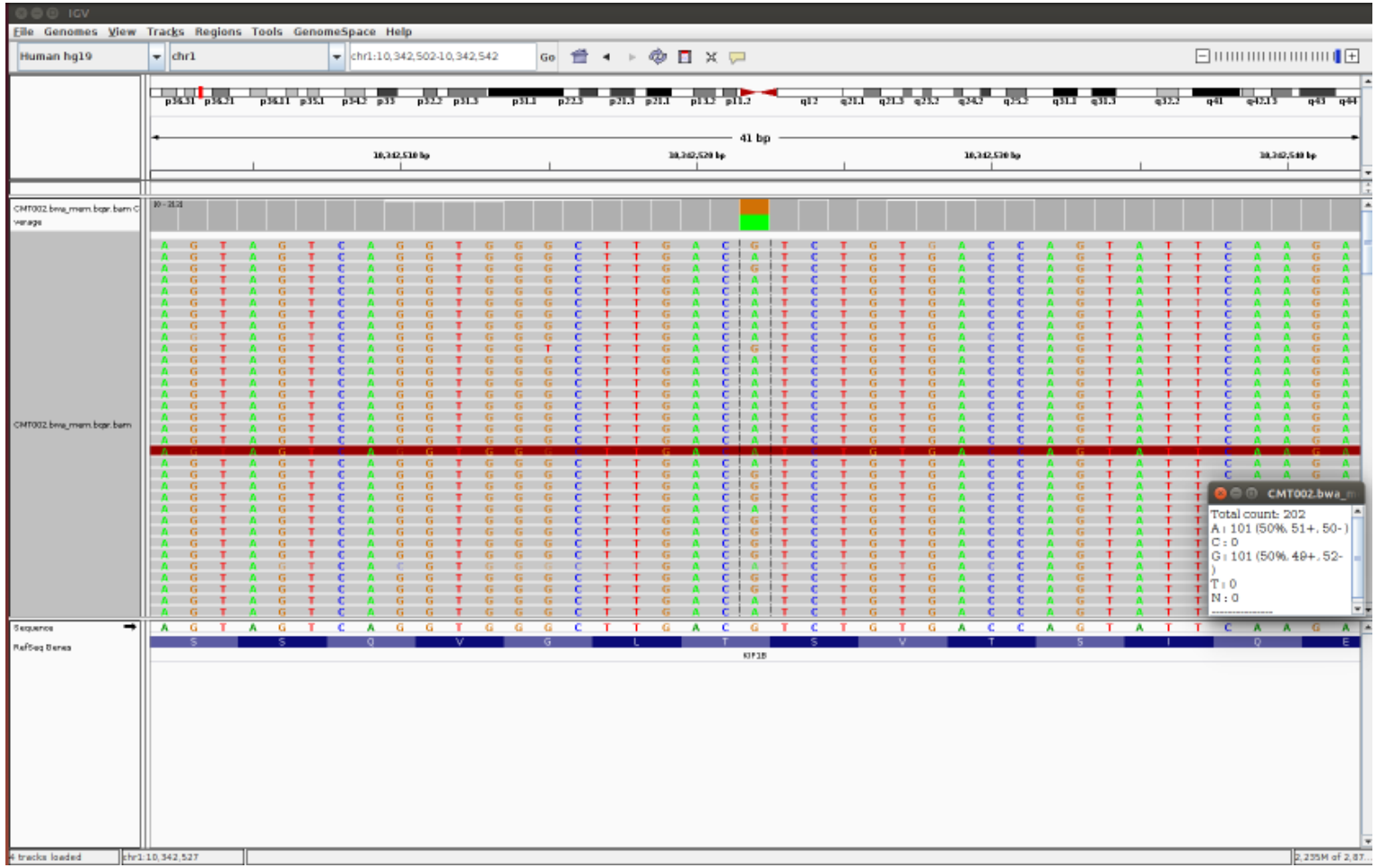


4. chr11:95595177A>G [Heterozygous]
MTMR2:NM_016156:c.T447C(p.Y149Y)

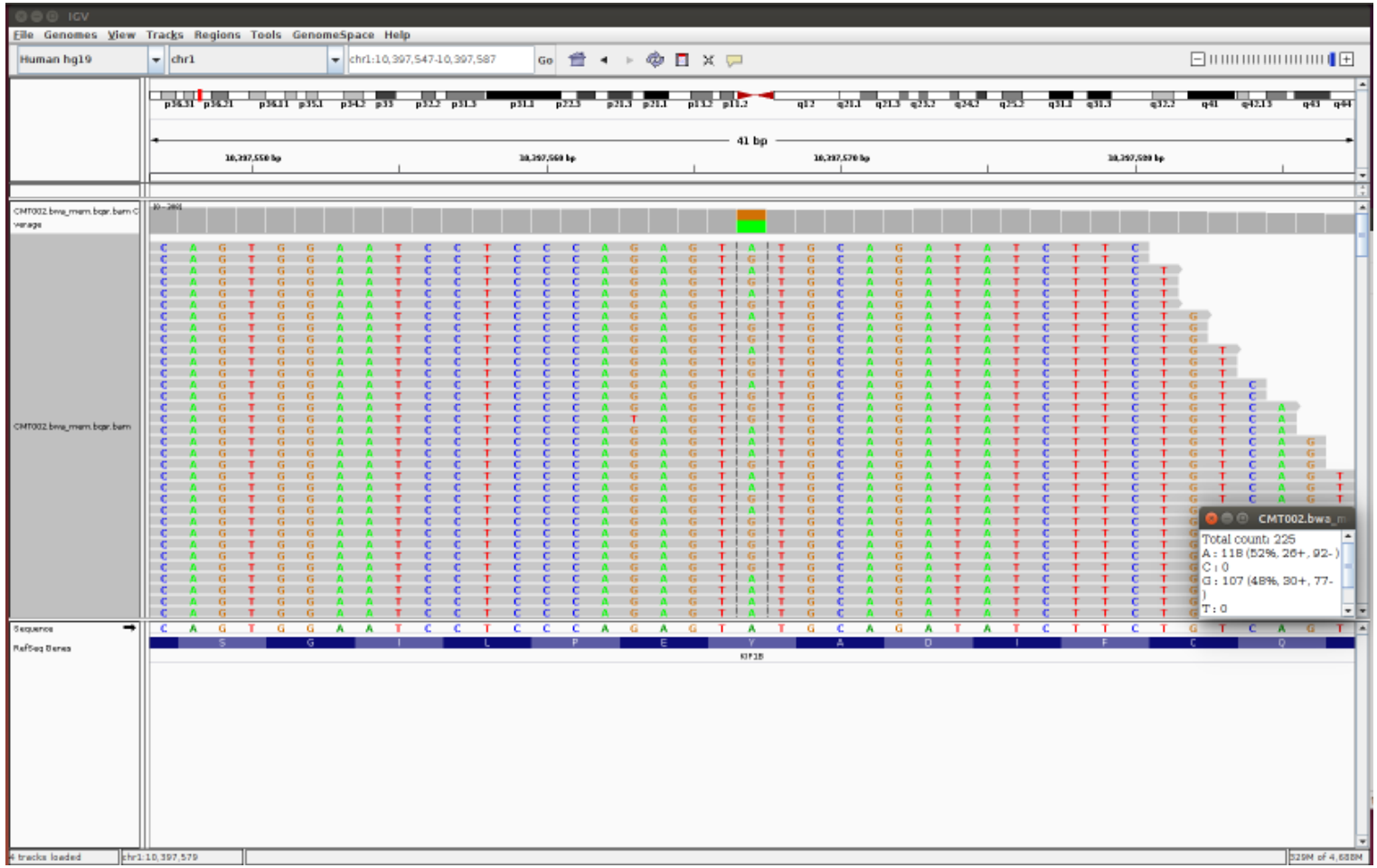


Case 2

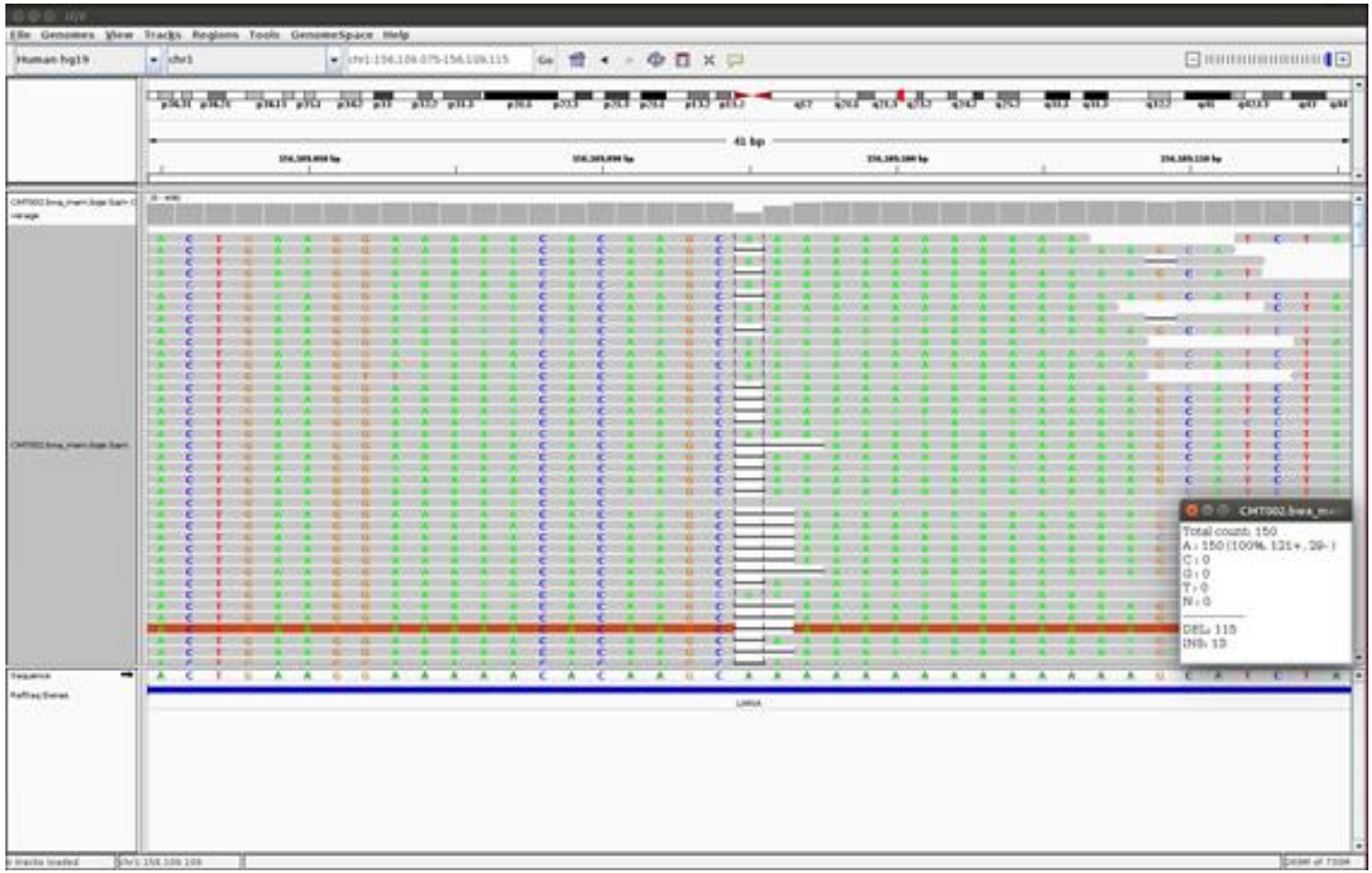
1. **chr1:10342522G>A [Heterozygous]**
   KIF1B:NM_015074:c.G1227A:p.T409T



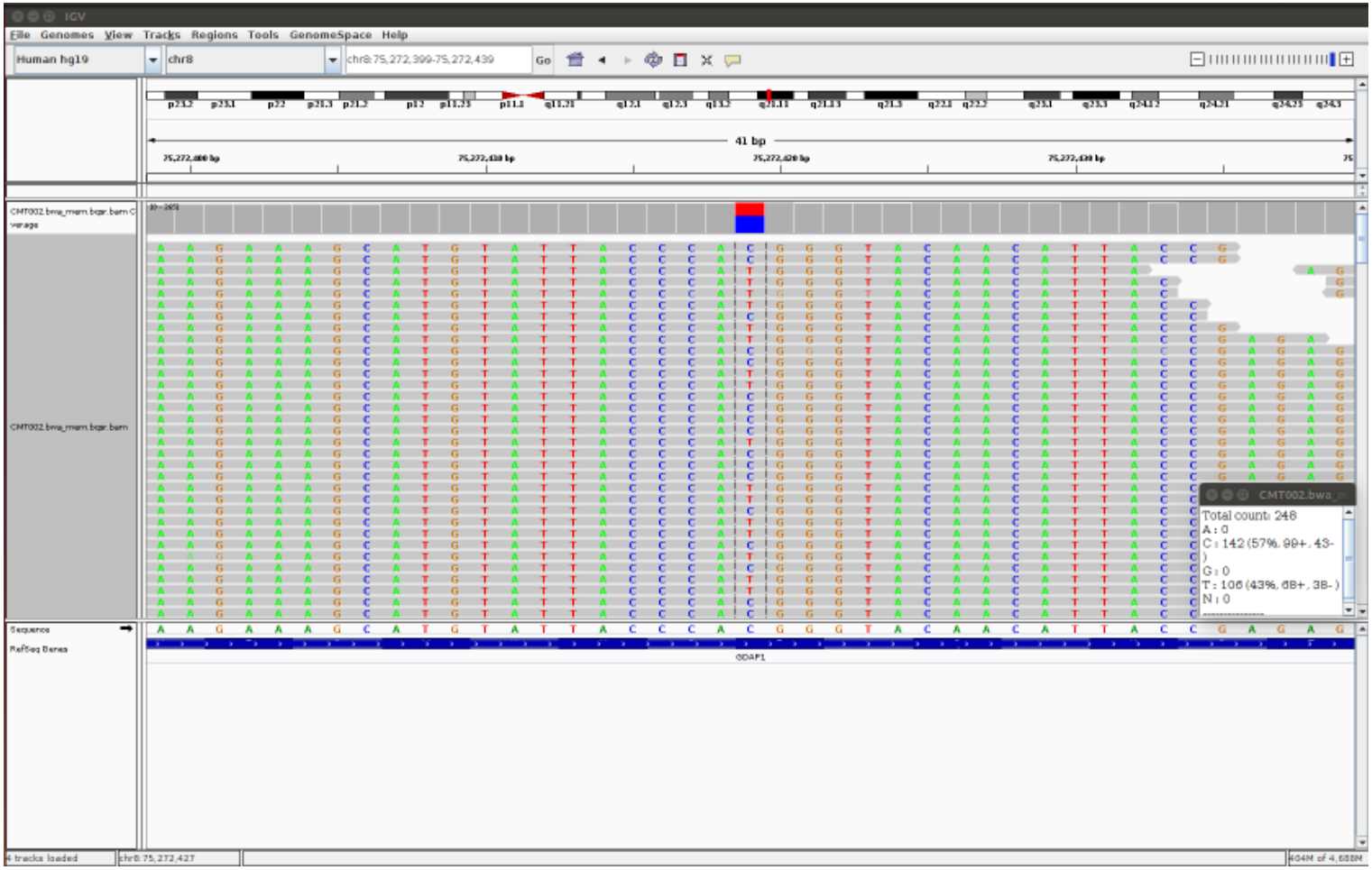
2. **chr1:10397567A>G [Heterozygous]**
   KIF1B:NM_015074:c.A3260G:p.Y1087C



3. **chr1:156109095_156109095delA (likely sequencing error / miscalled variant)**
   LMNA: NM_170707:cDNA.2405_2405delA



4. **chr8:75272419C>T [Heterozygous]**
   GDAP1:NM_018972:c.C358T:p.R120W



5. chr11:9990017G>A [Heterozygous]

SBF2:NM_030962:c.C1471T:p.L491F