

Fig. S1. The p.Ala1457Thr mutation detected in the proband.

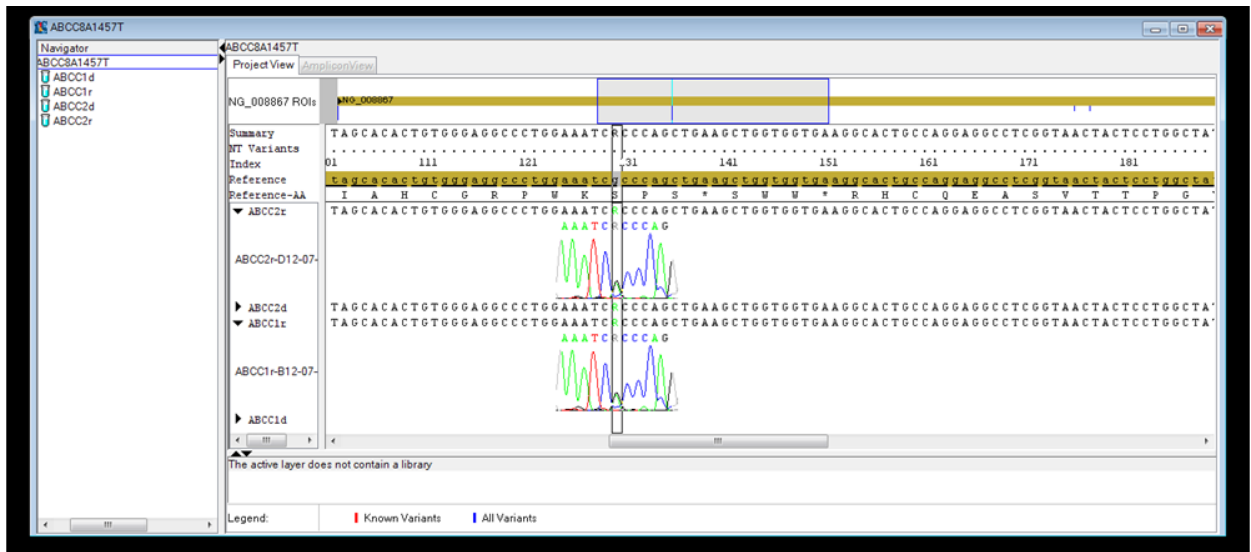


Fig. S2. p.Ala1457Thr mutation in the *PolyPhen-2* software.

