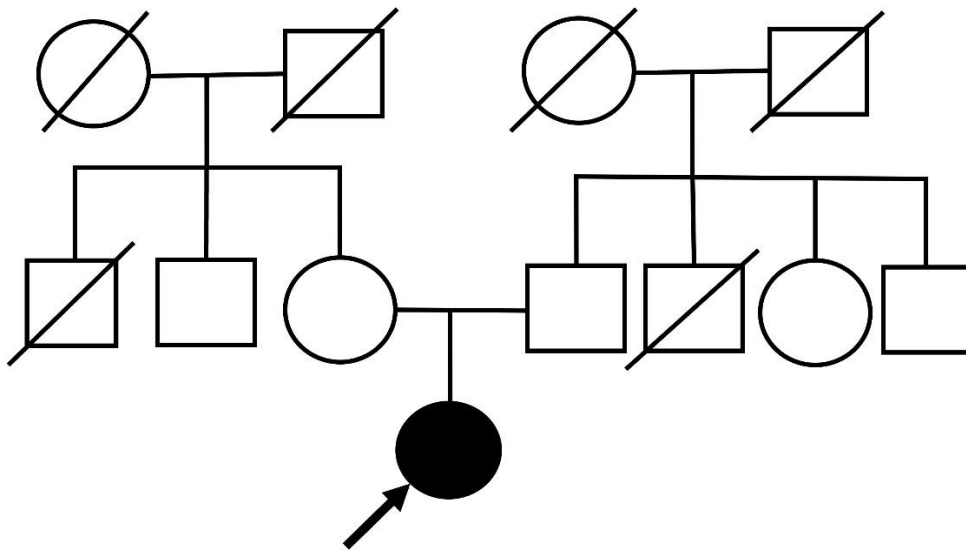
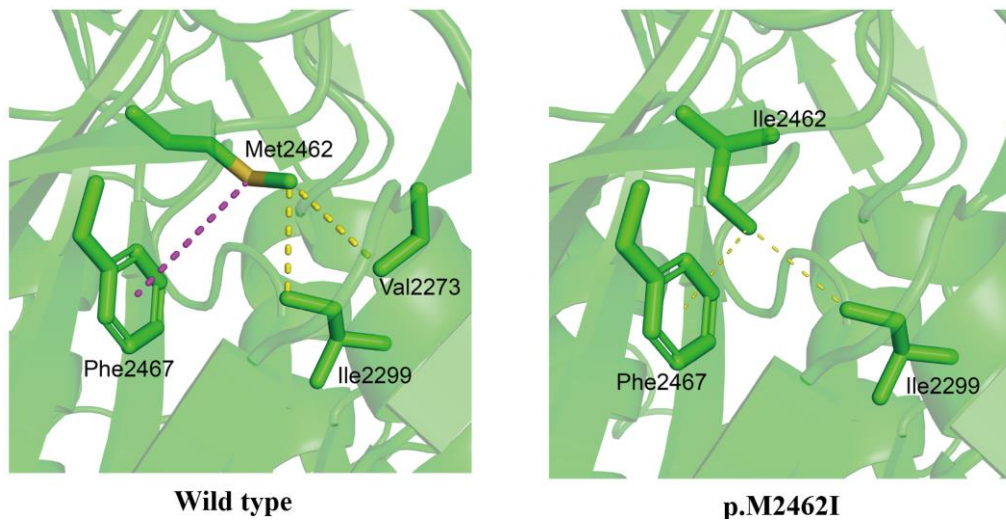


Supplementary Figure 1



Supplementary Figure 2



Supplementary Figure 3

| | |
|-----------------|---|
| H.sapiens | S MEECEALCTRLAI M VNGKFQ C IGSLQHIKSRFGRGFTVKVHLKNNKVTMETLTKFMQLH |
| P.troglodytes | S MEECEALCTRLAI M VNGKFQ C IGSLQHIKSRFGRGFTVKVHLKNNKVTMETLTKFMQLH |
| C.L. familiaris | S MEECEALCTRLAI M VNGRFQ C IGSLQHIKSRFGRGFTVKVHLKNTKVSMDTLTRFMQLH |
| E. caballus | S MEECEALCTRLAI M VNGRFQ C IGSLQHIKSRFGRGFTVKVHLKNTRVSM E ALTRFMQLH |
| M.musculus | S MEECEALCTRLAI M VNGRFQ C IGSLQHIKSRFGRGFTVKVHLKNNKVS M ETLTKFMQLH |
| G.gallus | S MEECEALCTRLAI M VNGSFQ C IGSLQHIKSRFGRGFTVKMHLNSSTVDTETL T EFMKSH |

Supplementary Figure Legend

Figure 1

Family pedigree of this patient.

Figure 2

In *silico* modeling of the wild-type and p.M2462I mutated *ABCA12* proteins was performed, which revealed the configuration change of *ABCA12* mutant

Figure 3

The c.7386G>C (p.M2462I) mutation could affect highly conserved amino-acid residues.