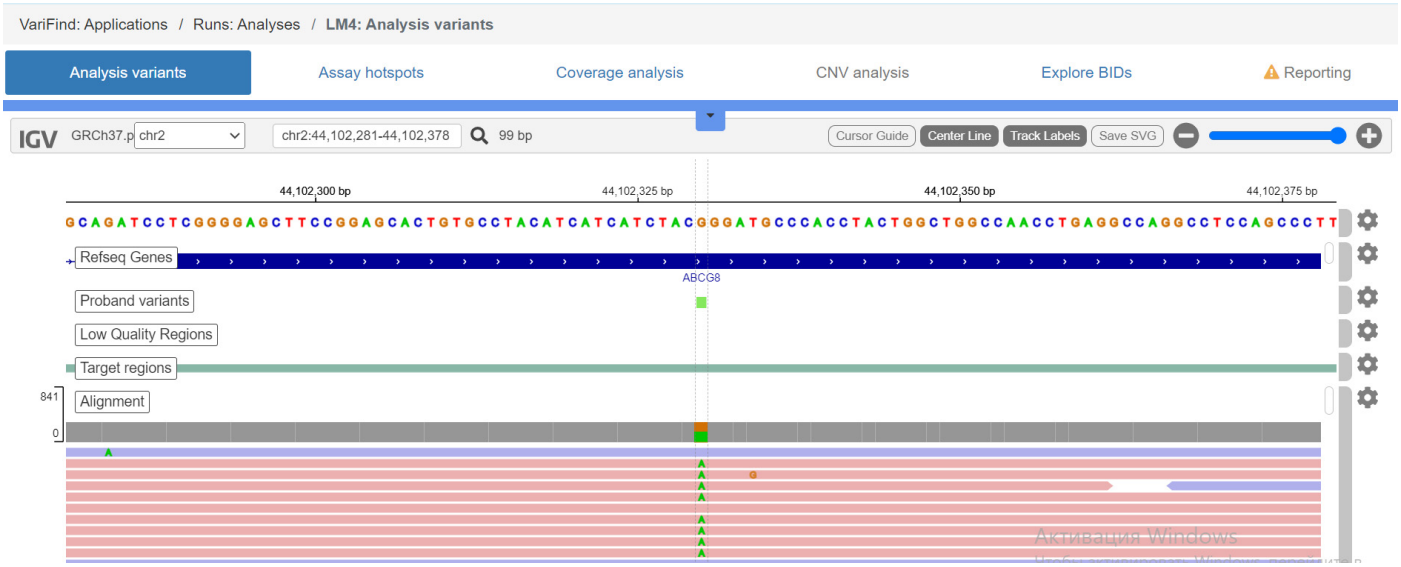
Case 2

Variant 1

chr2:44102330 (GRCh37) NM_022437.3:c.1534G>A p.(Gly512Arg) (rs376069170)

NGS sequencing:



Sanger sequencing results (reverse reads):

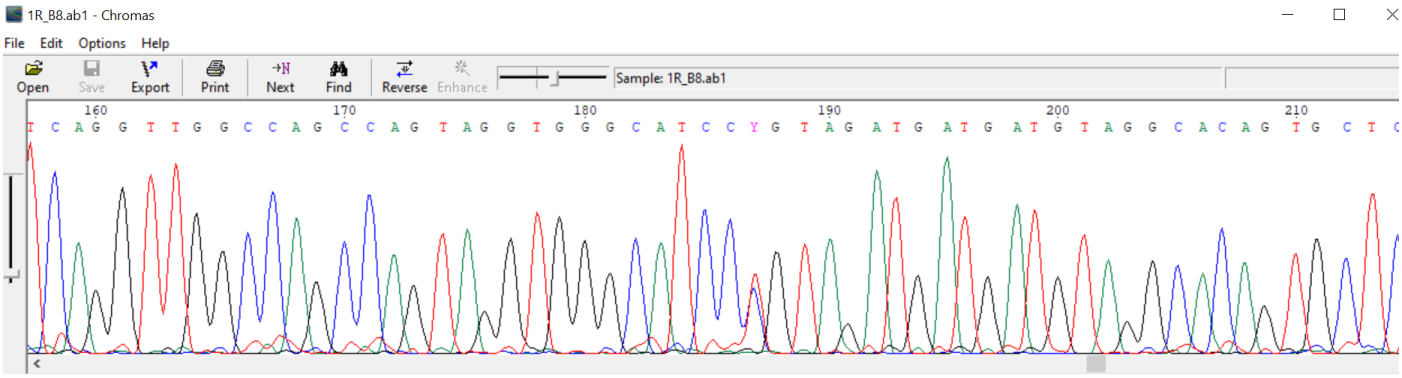
Probe identification

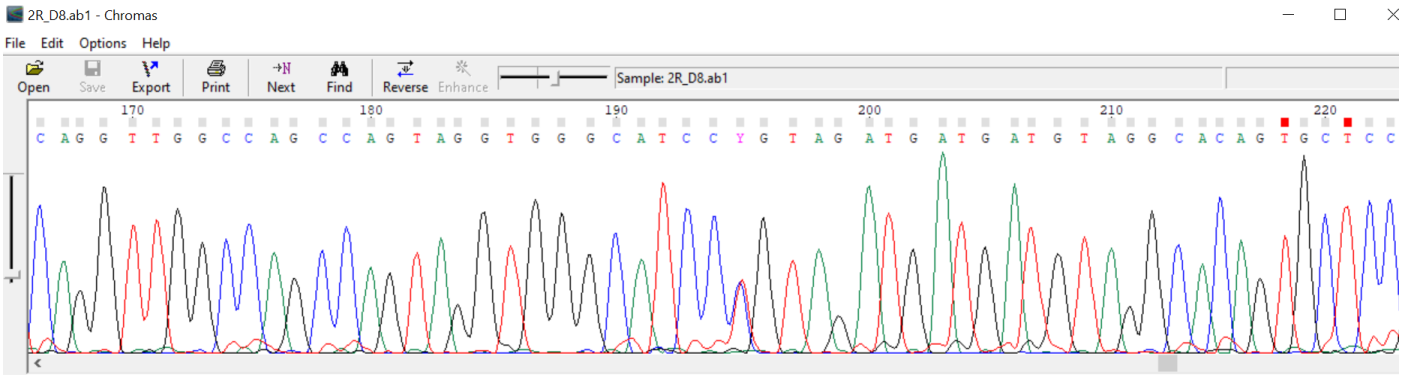
Proband - 1

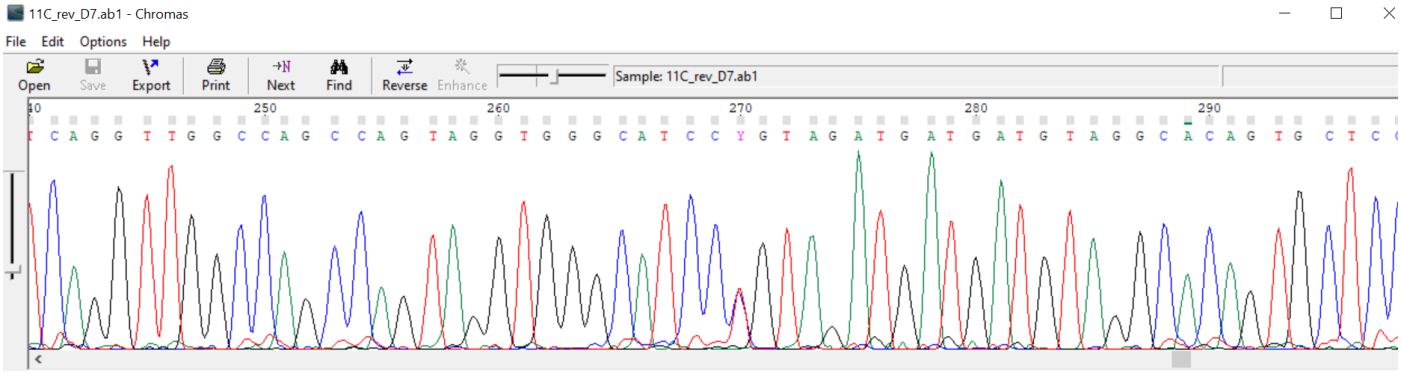
Proband's mother - 2

Proband's sister - 11C

Proband's father DNA – not available



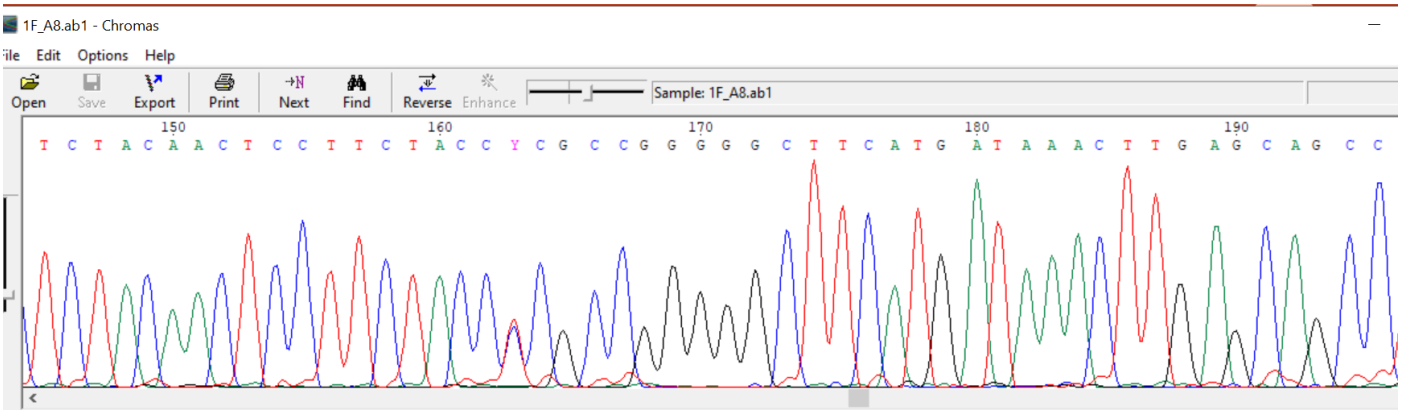


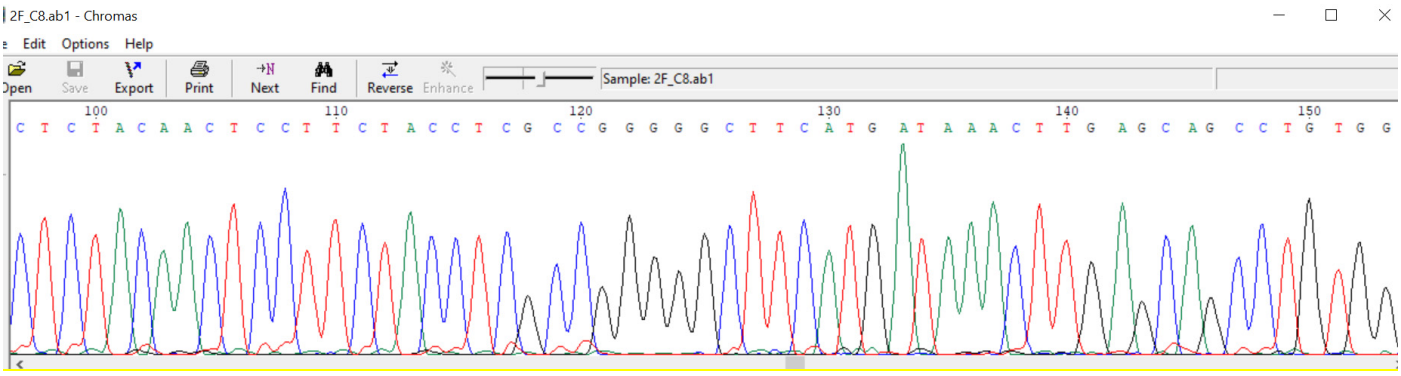


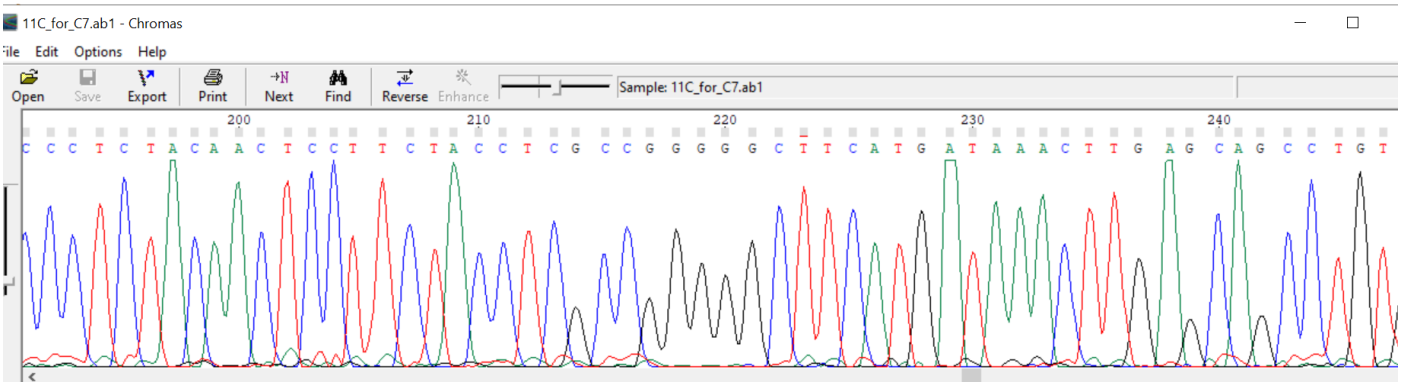
Variant 2
chr2:44102511 (GRCh37) NM_022437.3(ABCG8):c.1715T>C p.(Leu572Pro) (rs769576789)
NGS sequencing:



Sanger sequencing results (forward reads):
Probe identification
Proband - 1
Proband's mother - 2
Proband's sister - 11C
Proband's father DNA – not available





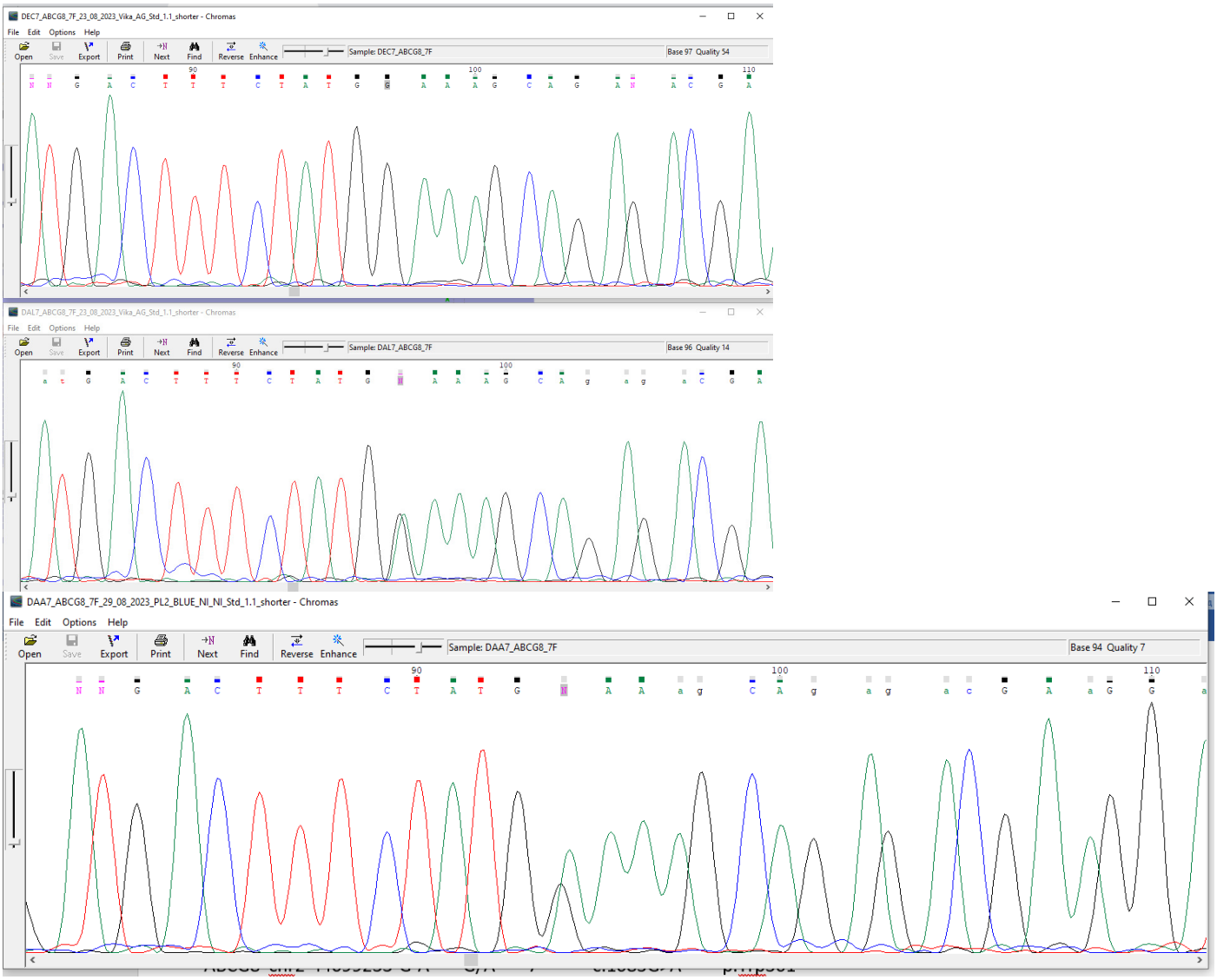


Case 3

ABCG8 chr2:44099233G>A NM_022437.3:c.1083G>A p.Trp361* (rs137852987)

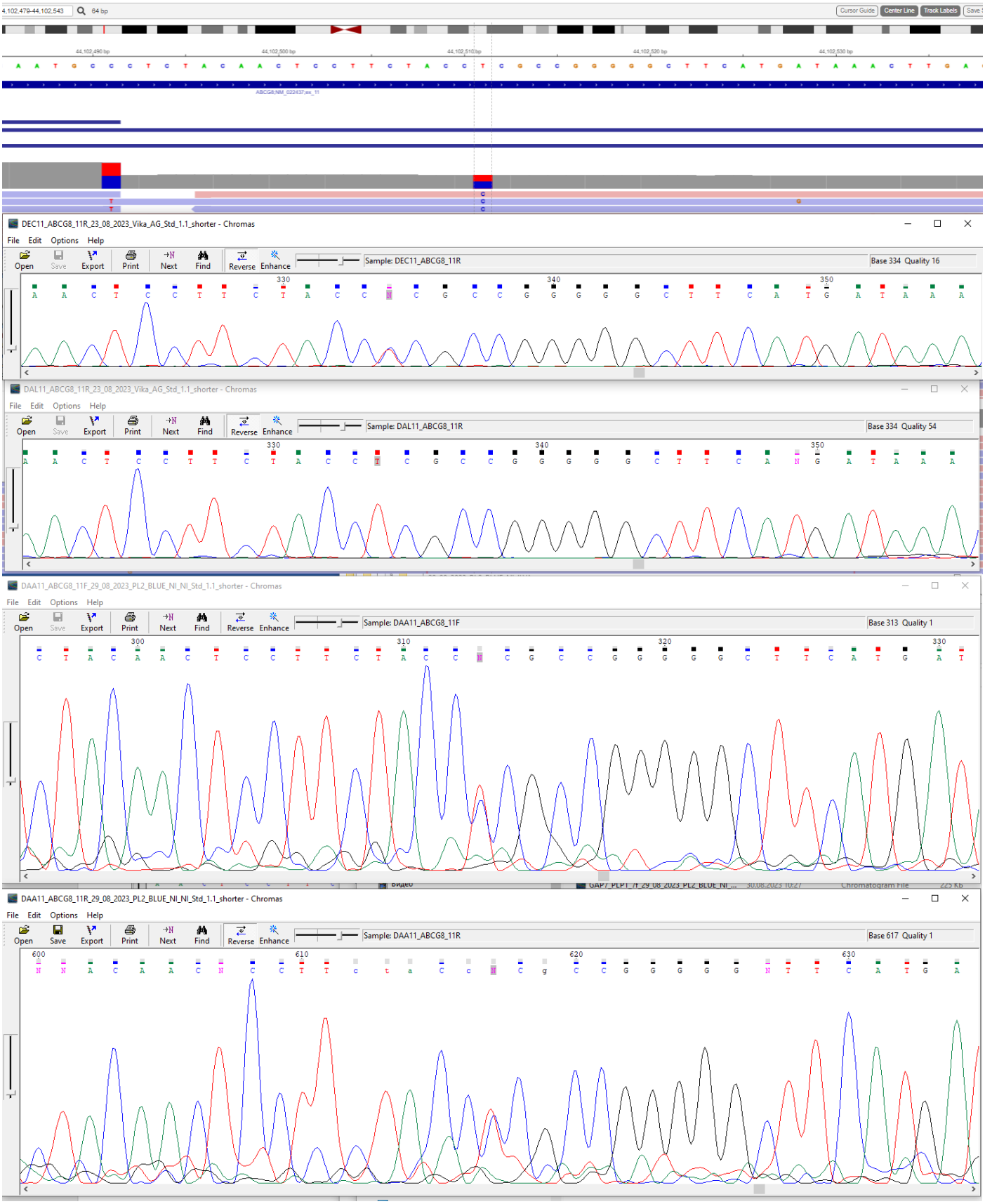


Sanger sequencing results (forward reads):
Probe identification
Proband's mother - DEC7
Proband's father - DAL7
Proband - DAA7



chr2:44102511 (GRCh37) NM_022437.3(ABCG8):c.1715T>C (p.Leu572Pro) (rs769576789)

Sanger sequencing results:
Probe identification
Proband's mother - DEC11
Proband's father - DAL11
Proband - DAA11


DEC11_ABCG8_11R_23_08_2023_Vika_AG_Std_1.1_shorter - Chromas
File Edit Options Help
Open Save Export Print Next Find Reverse Enhance
Sample: DEC11_ABCG8_11R
Base 334 Quality 16
DAL11_ABCG8_11R_23_08_2023_Vika_AG_Std_1.1_shorter - Chromas
Sample: DAL11_ABCG8_11R
Base 334 Quality 54
DAA11_ABCG8_11F_29_08_2023_PL2_BLUE_NI_NI_Std_1.1_shorter - Chromas
Sample: DAA11_ABCG8_11F
Base 313 Quality 1
DAA11_ABCG8_11R_29_08_2023_PL2_BLUE_NI_NI_Std_1.1_shorter - Chromas
Sample: DAA11_ABCG8_11R
Base 617 Quality 1