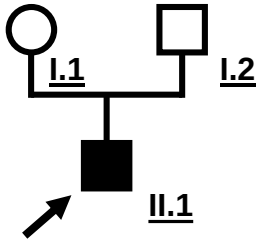
Supplementary Materials

Figure S1. Sanger sequencing and WES results of the patients in this study. f: forward; r: reverse.

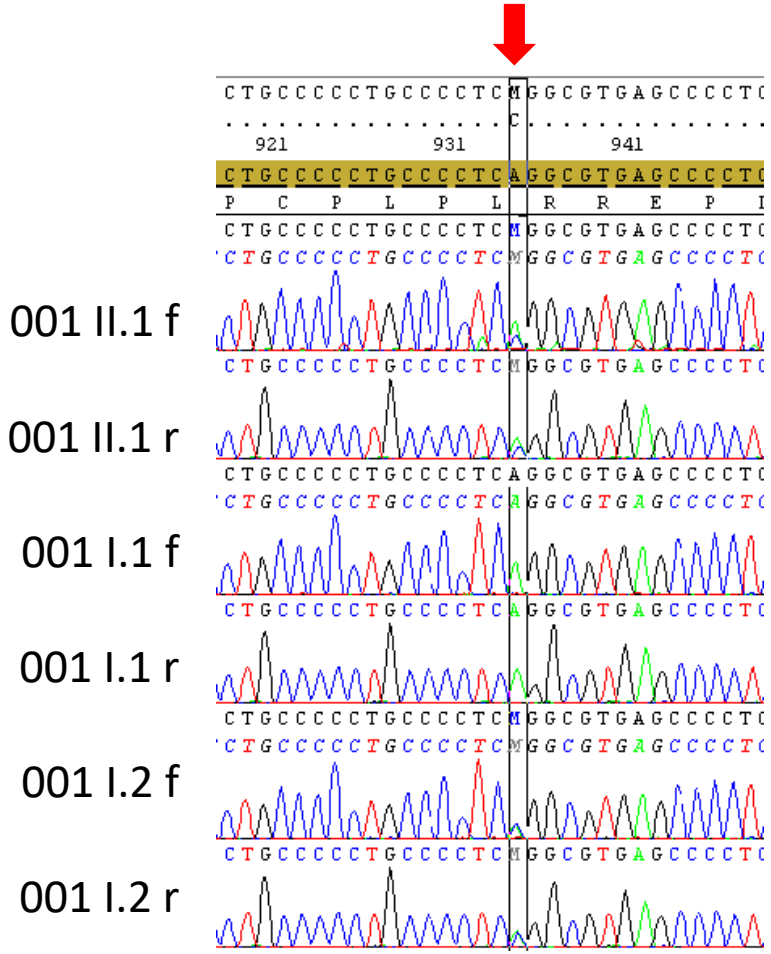
Patient 001

# Disease-associated gene: NR2E3

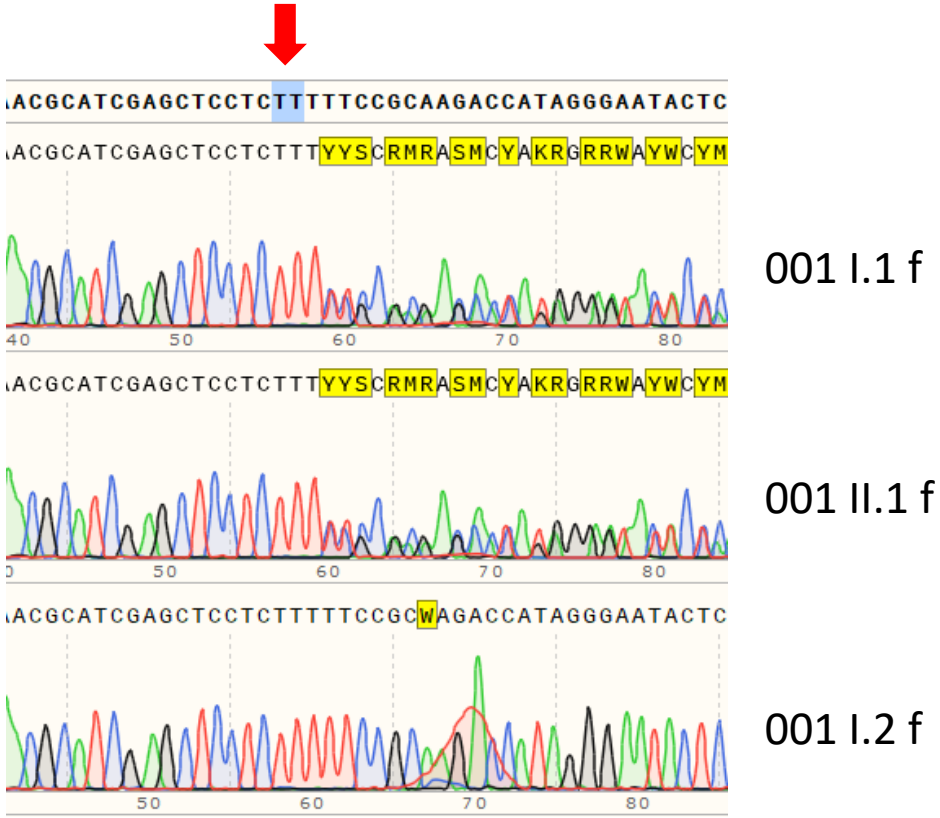
**001**




**c.119-2A>C**



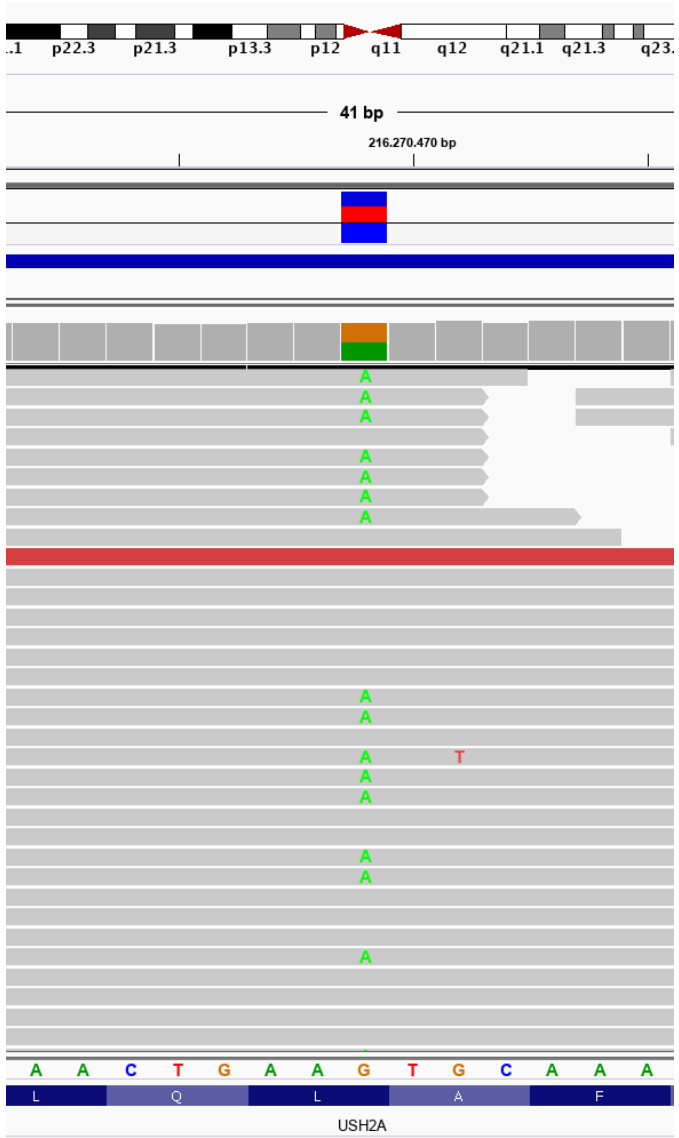

**c.1171_1172delTT**



Patient 002

# Disease-associated gene: USH2A

## c.4714C>T (173x Total Count)

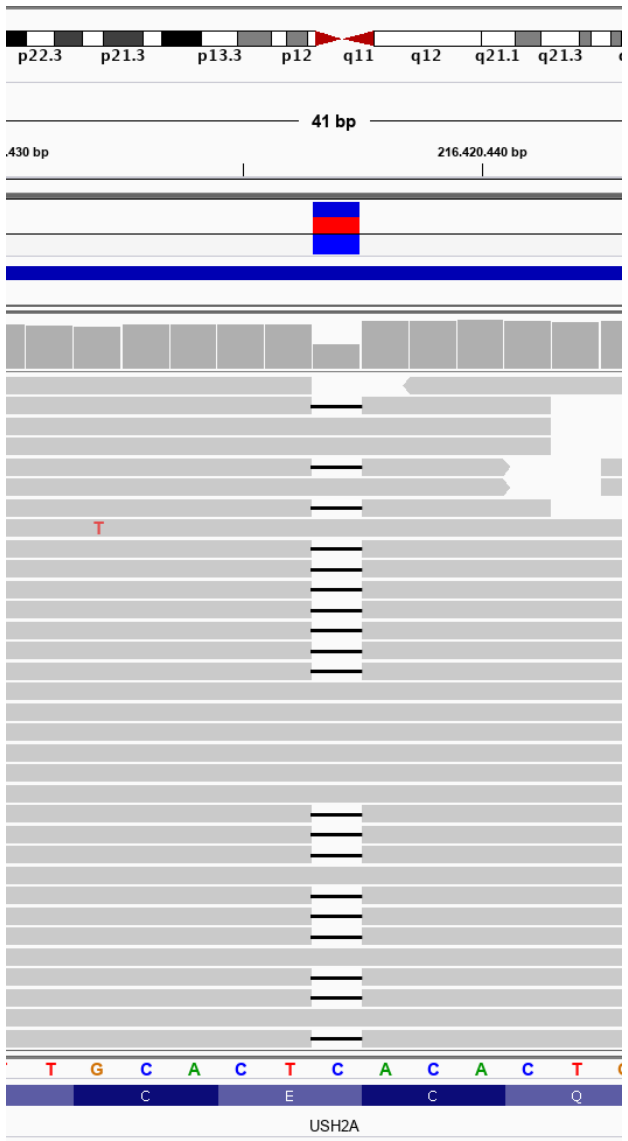



Chromosome 1

Chromosomal Position

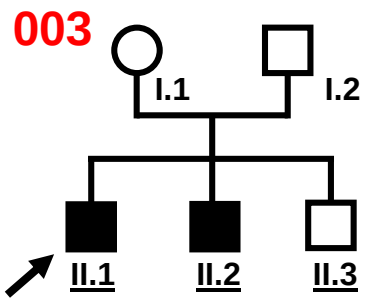
NGS Reads

## c.2299delG (63x Total Count)



Patient 003

# Disease-associated gene: USH2A




## c.4714C>T


003 II.1 f

003 II.1 r

003 II.3 f

003 II.3 r

003 II.2 f

003 II.2 r

## c.2299delG


003 II.1 f

003 II.1 r

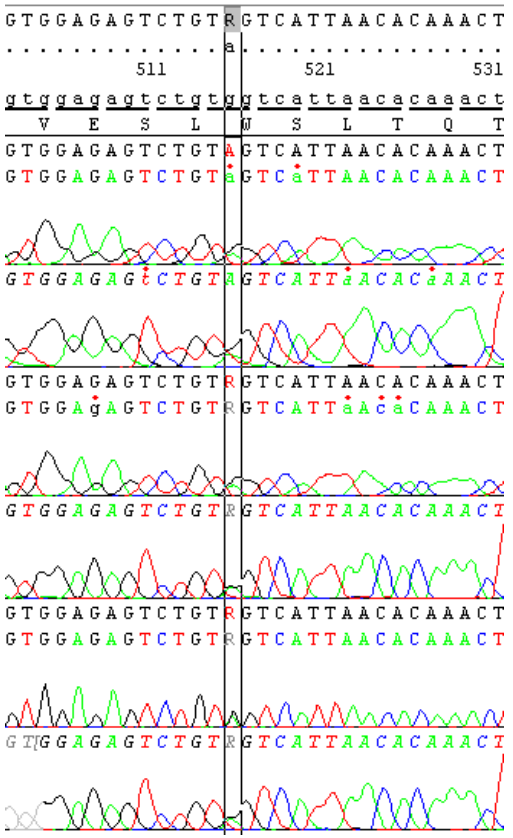
003 II.3 f

003 II.3 r

003 II.2 f

003 II.2 r

## c.11864G>A




003 II.1 f

003 II.1 r

003 II.3 f

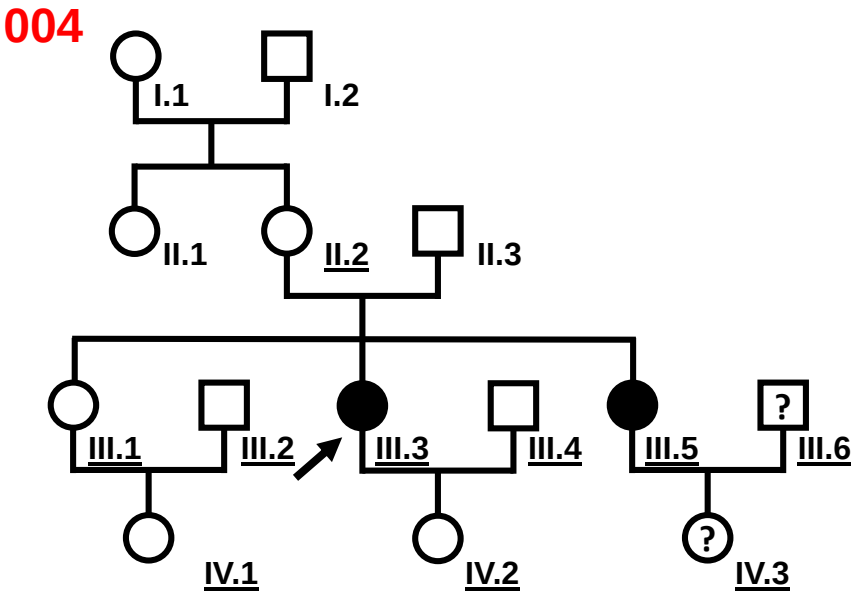
003 II.3 r

003 II.2 f

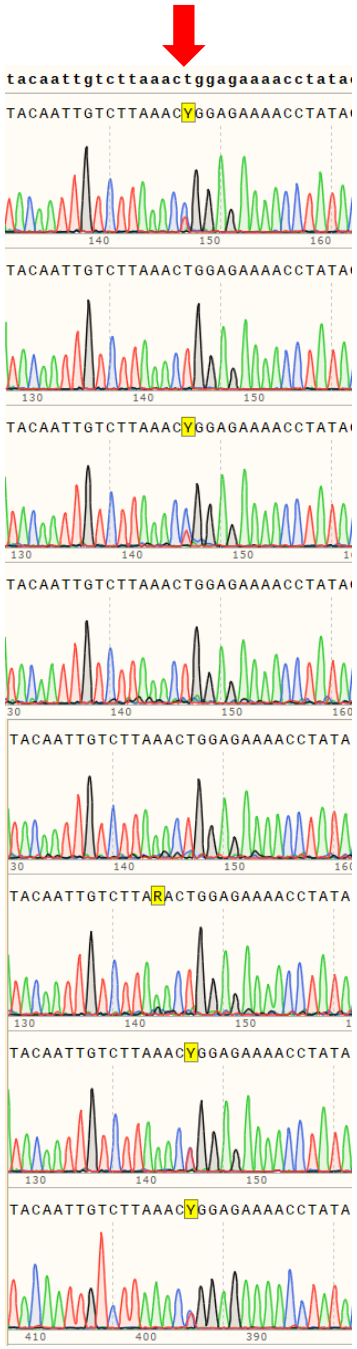
003 II.2 r

Patient 004

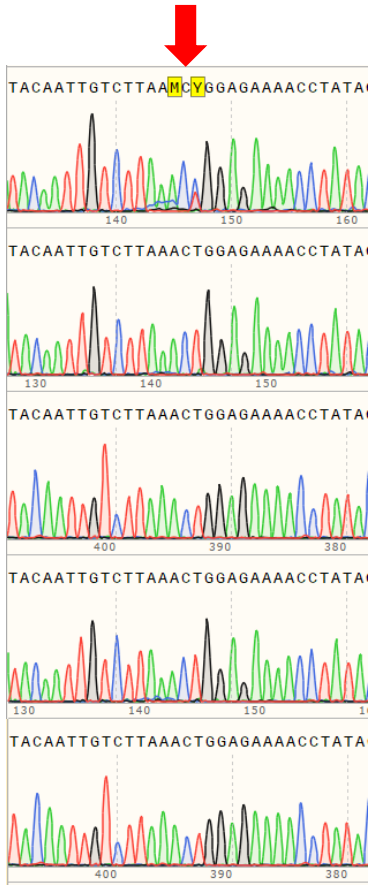
# Disease-associated gene: USH2A




**c.10561T>C**

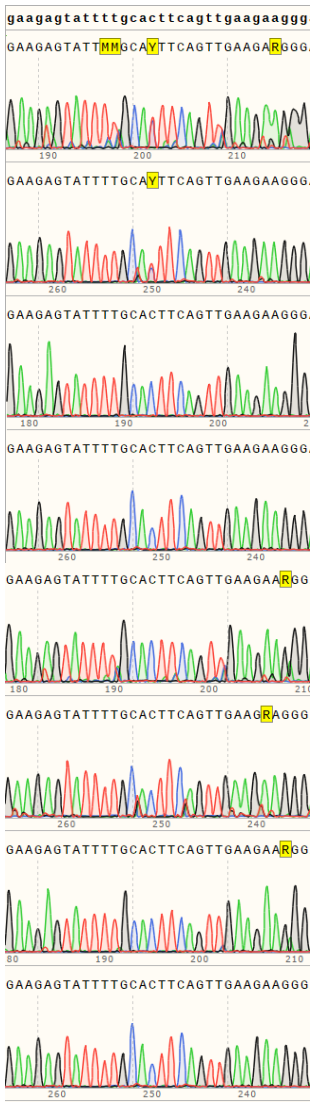



**c.10561T>C**



Patient 004

**c.4714C>T**



004 III.3 f

004 III.3 r

004 III.4 f

004 III.4 r

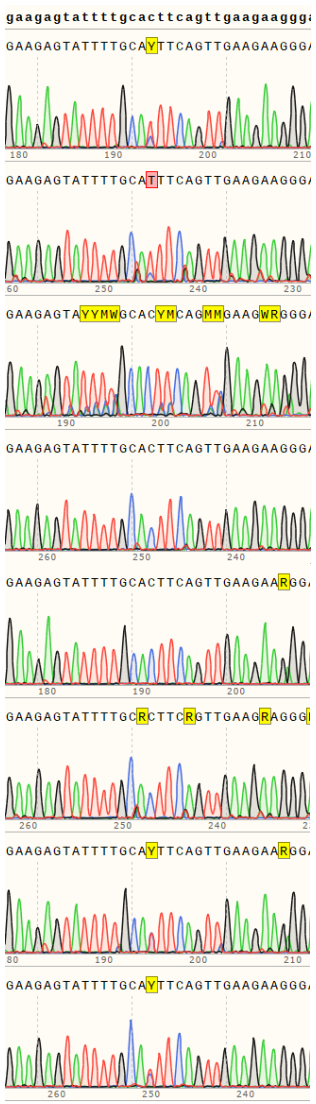
004 IV.2 f

004 IV.2 r

004 IV.1 f

004 IV.1 r

**c.4714C>T**



004 III.2 f

004 III.2 r

004 III.1 f

004 III.1 r

004 II.2 f

004 II.2 r

004 III.5 f

004 III.5 r

**c.4714C>T**

004 IV.3 f

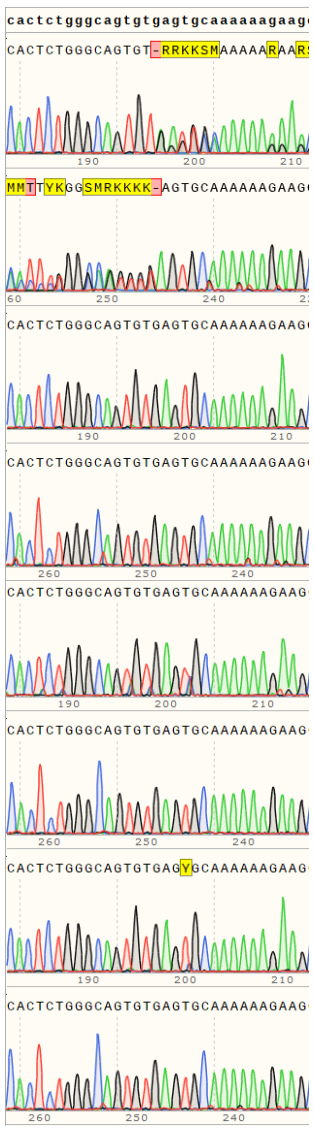
004 IV.3 r

004 III.6 f

004 III.6 r

Patient 004

**c.2299del**



004 III.3 f

004 III.3 r

004 III.4 f

004 III.4 r

004 IV.2 f

004 IV.2 r

004 IV.1 f

004 IV.1 r

**c.2299del**

004 III.2 f

004 III.2 r

004 III.1 f

004 III.1 r

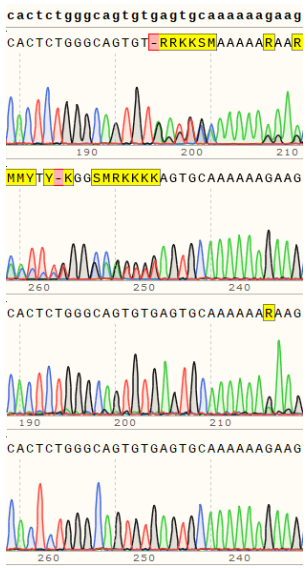
004 II.2 f

004 II.2 r

004 III.5 f

004 III.5 r

**c.2299del**



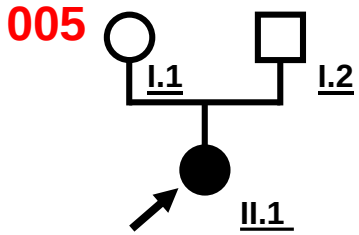
004 IV.3 f

004 IV.3 r

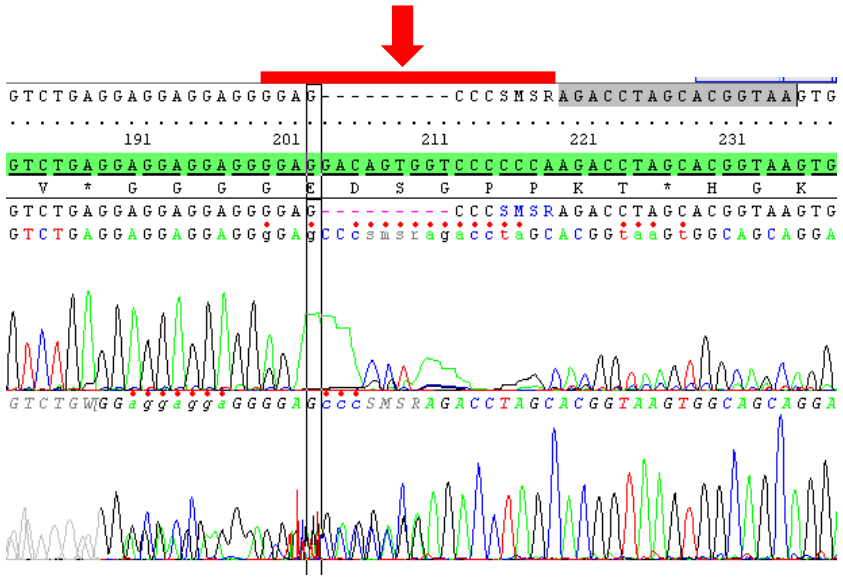
004 III.6 f

004 III.6 r

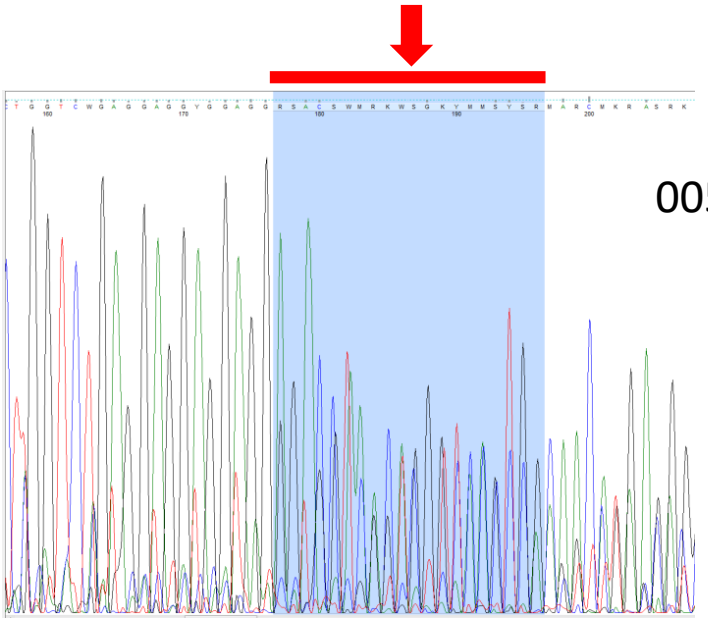
Patient 005

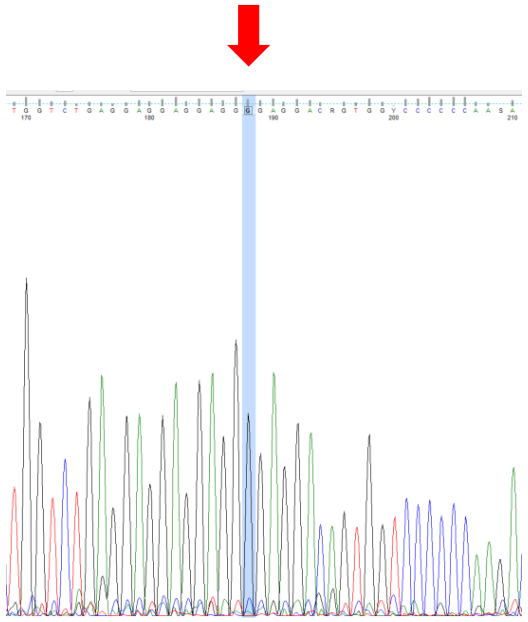



## c.1709_1728del



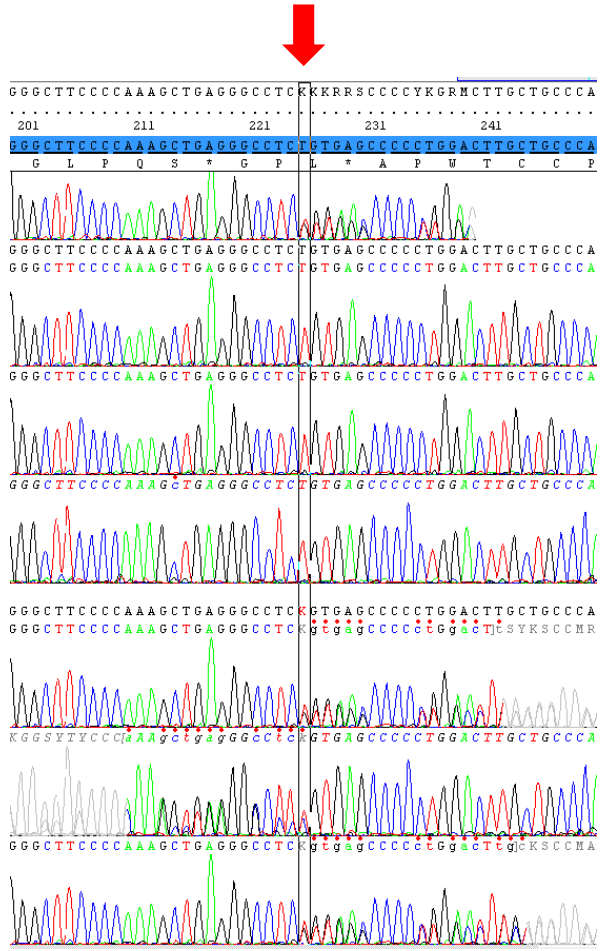

## c.1709_1728del




# Disease-associated gene: c2orf71

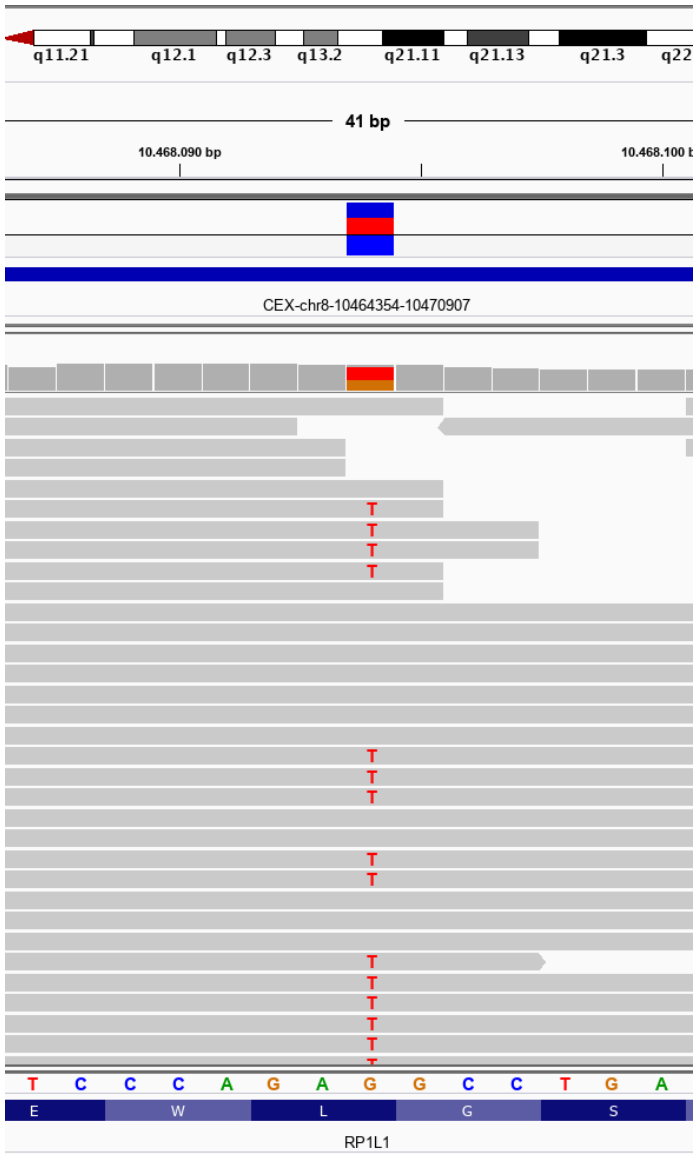
## c.2655delT



Patient 006

# Disease-associated gene: RP1L1

## c.3514C>A (85x Total Count)

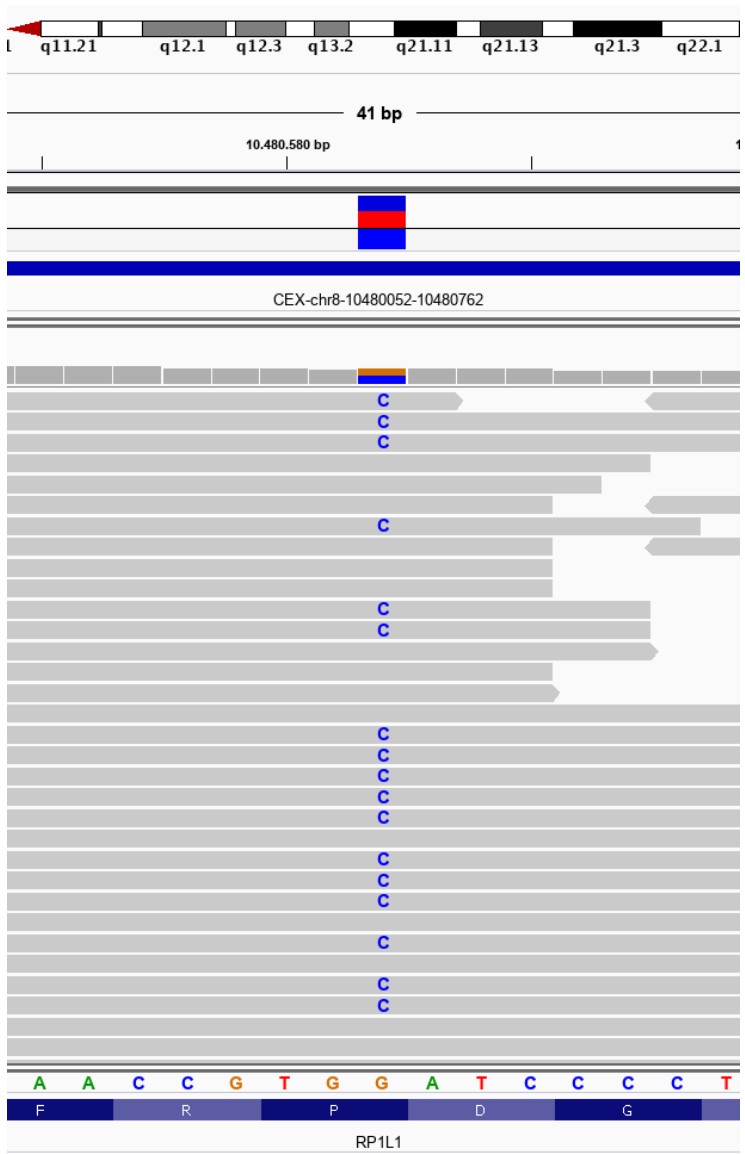



Chromosome 8

Chromosomal Position

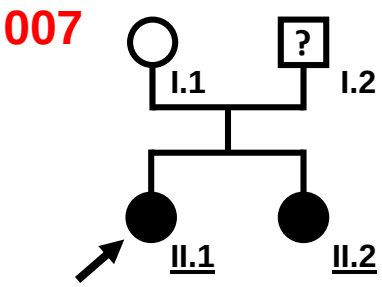
NGS Reads

## c.130C>G (53x Total Count)

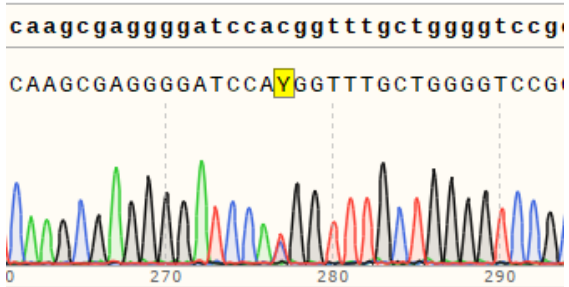


Patient 007

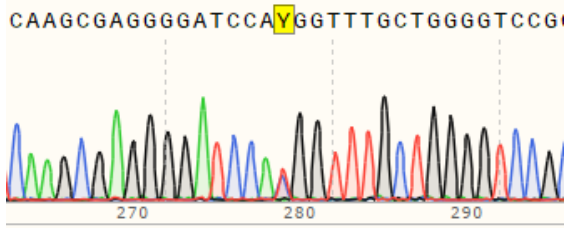
# Disease-associated gene: RP1L1



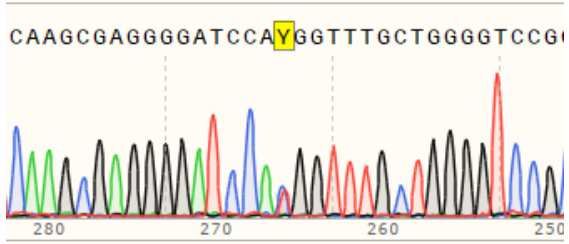

c.133C>T



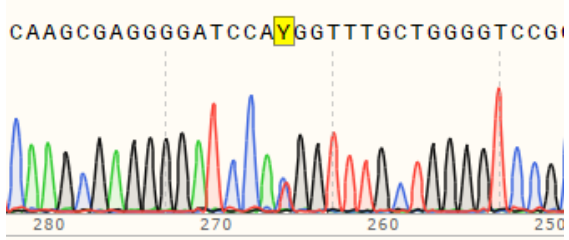

007 II.1 f




007 II.1 f

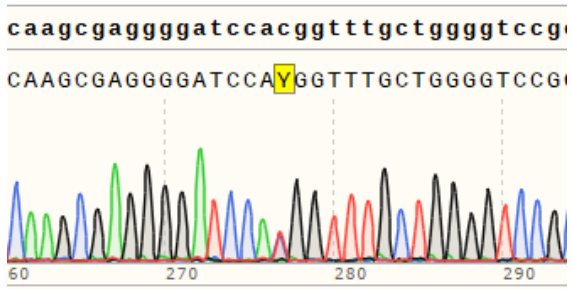



007 II.1 r

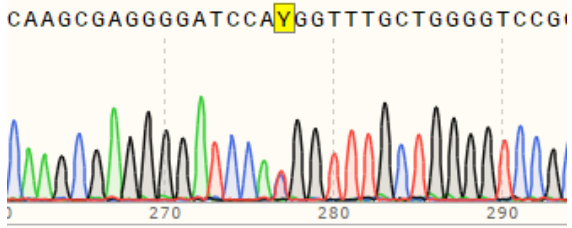



007 II.1 r

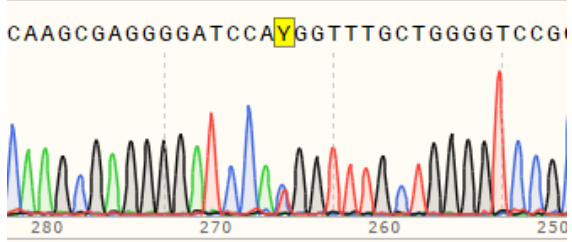
c.133C>T



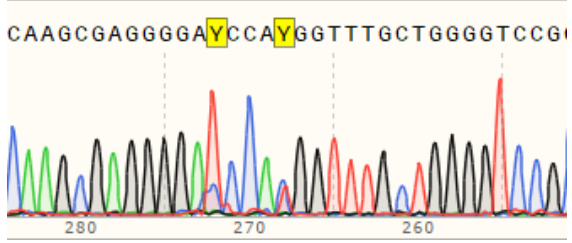

007 II.2 f




007 II.2 f




007 II.2 r

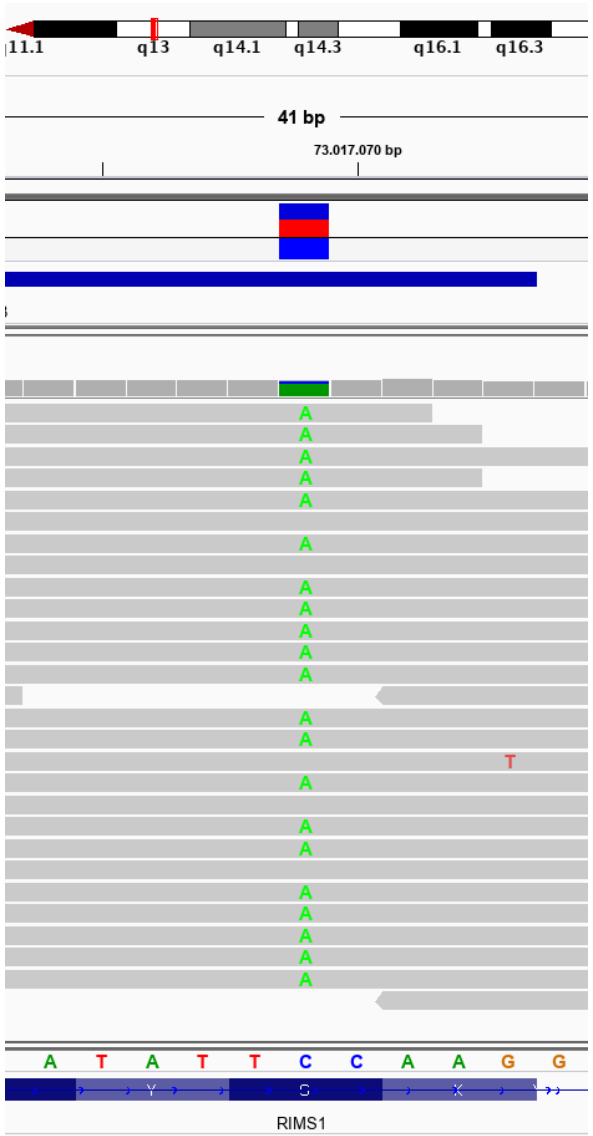



007 II.2 r

Patient 008

# Disease-associated gene: RIMS1

## c.1919C>A (26x Total Count)

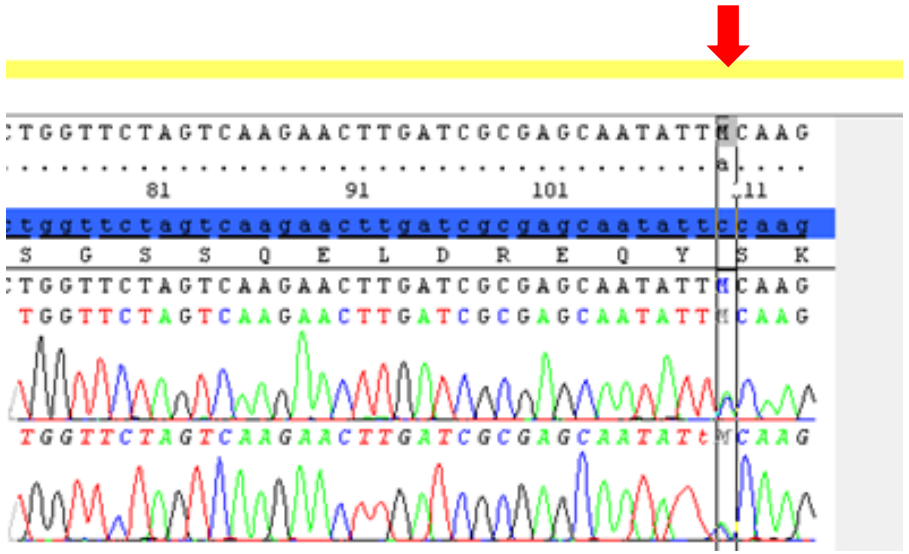



Chromosome 6

Chromosomal Position

NGS Reads

## c.1919C>A



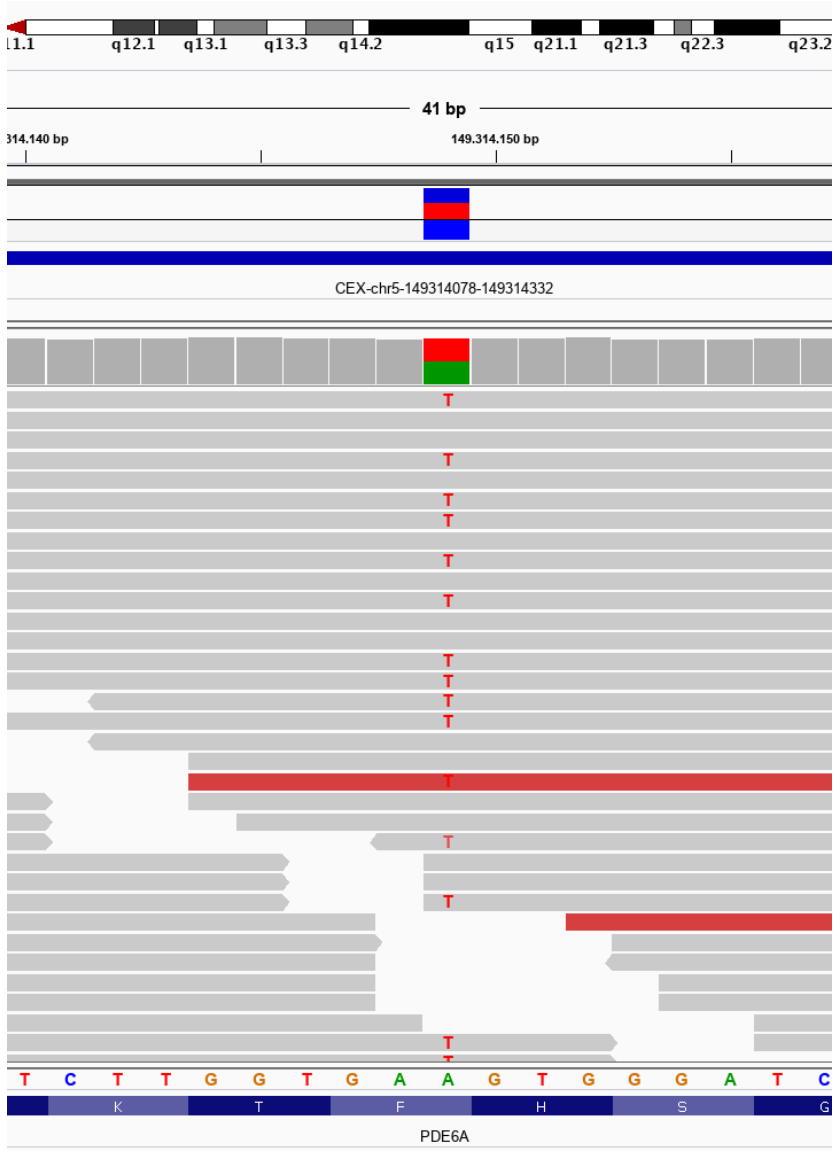

008 f

008 r

Patient 009

# Disease-associated gene: PDE6A

## c.607T>A (111x Total Count)




Chromosome 5

Chromosomal Position

NGS Reads

## c.607T>A


009 f

009 f

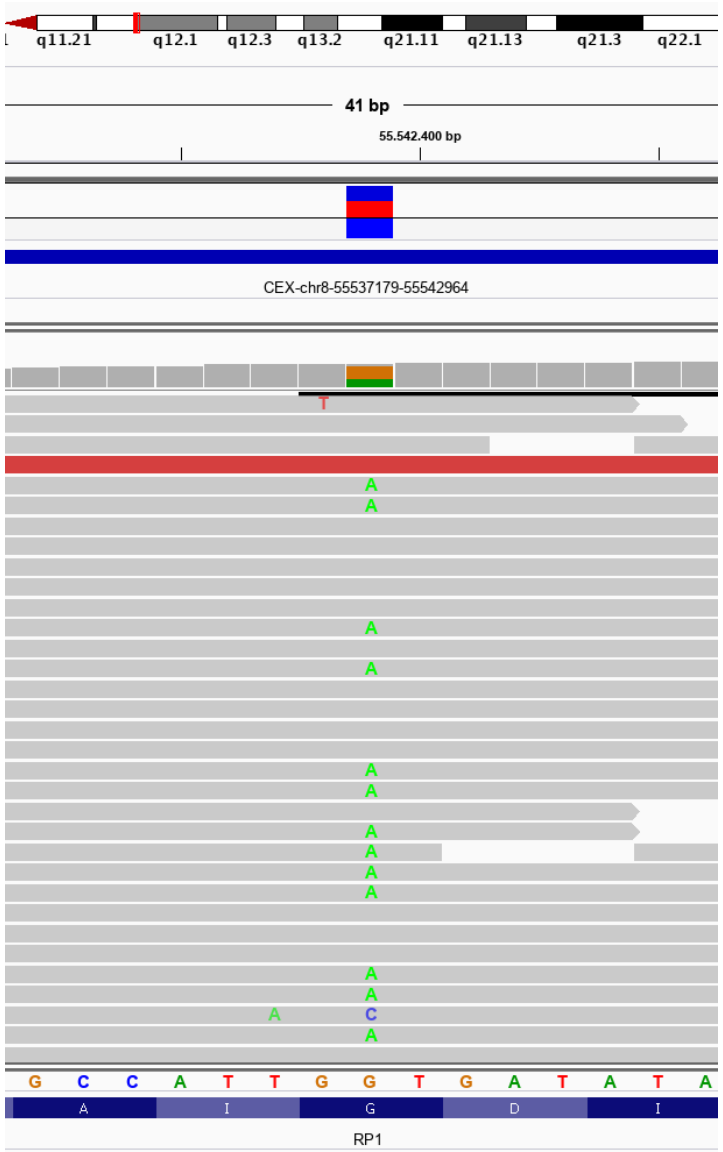
009 r

009 r

Patient 010

# Disease-associated gene: RP1

## c.5957G>A (96x Total Count)

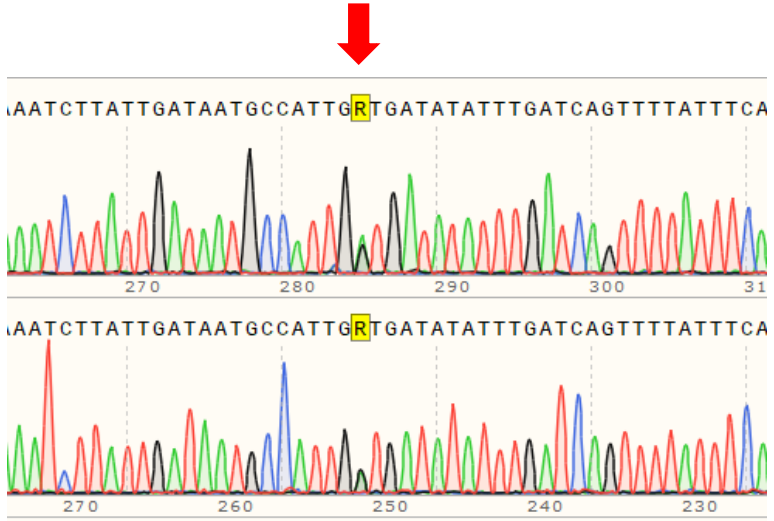



Chromosome 8

Chromosomal Position

NGS Reads

## c.5957G>A



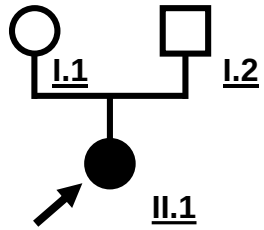

010 f

010 r

Patient 014

# Disease-associated gene: PROM1

014

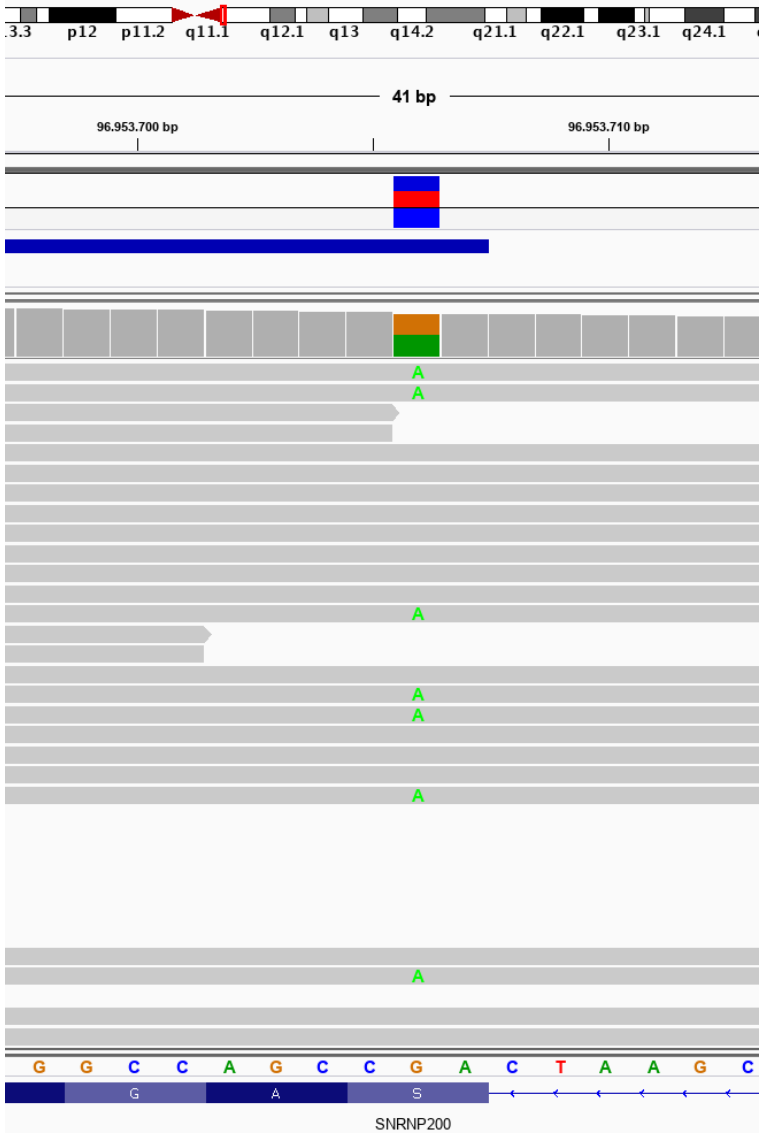



c.1069G>C

Patient 016

# Disease-associated gene: SNRNP200

## c.3260C>T (96x Total Count)




Chromosome 2

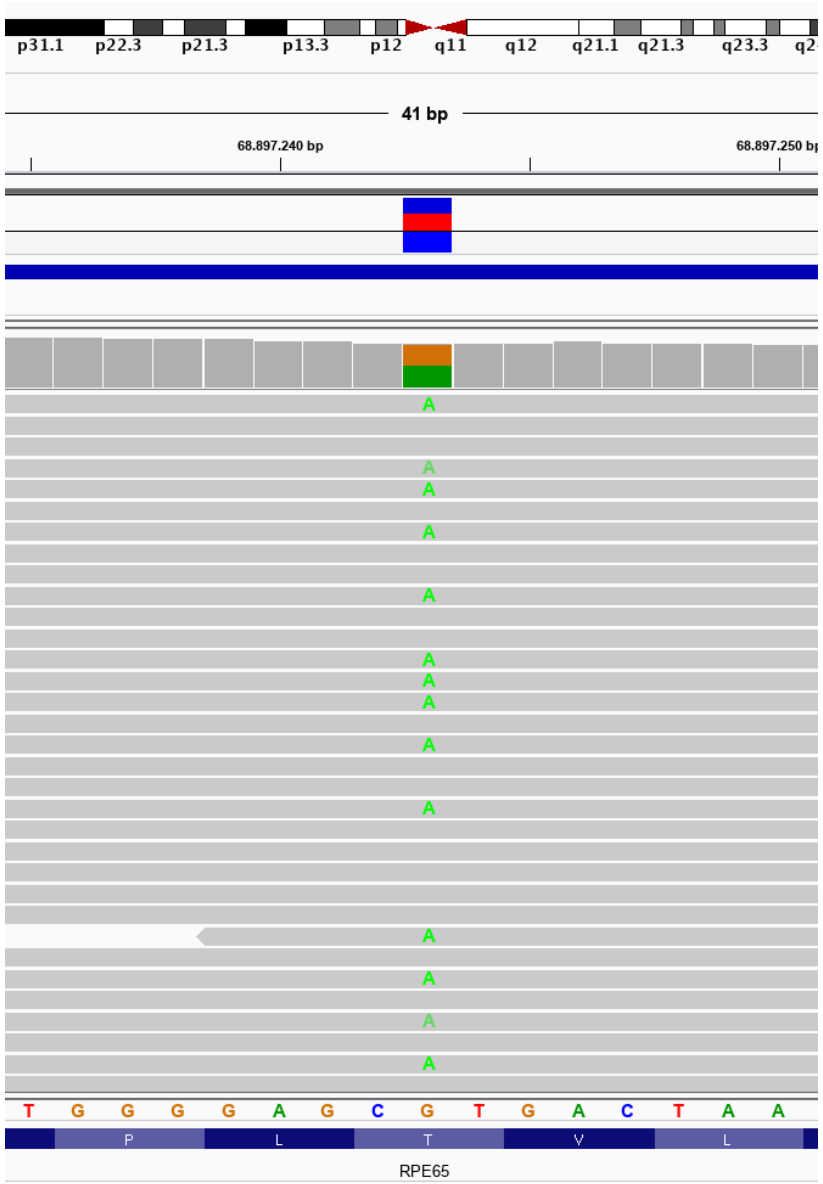
Chromosomal Position

NGS Reads

Patient 017

# Disease-associated gene: RPE65

## c.1154C>T (152x Total Count)




Chromosome 1

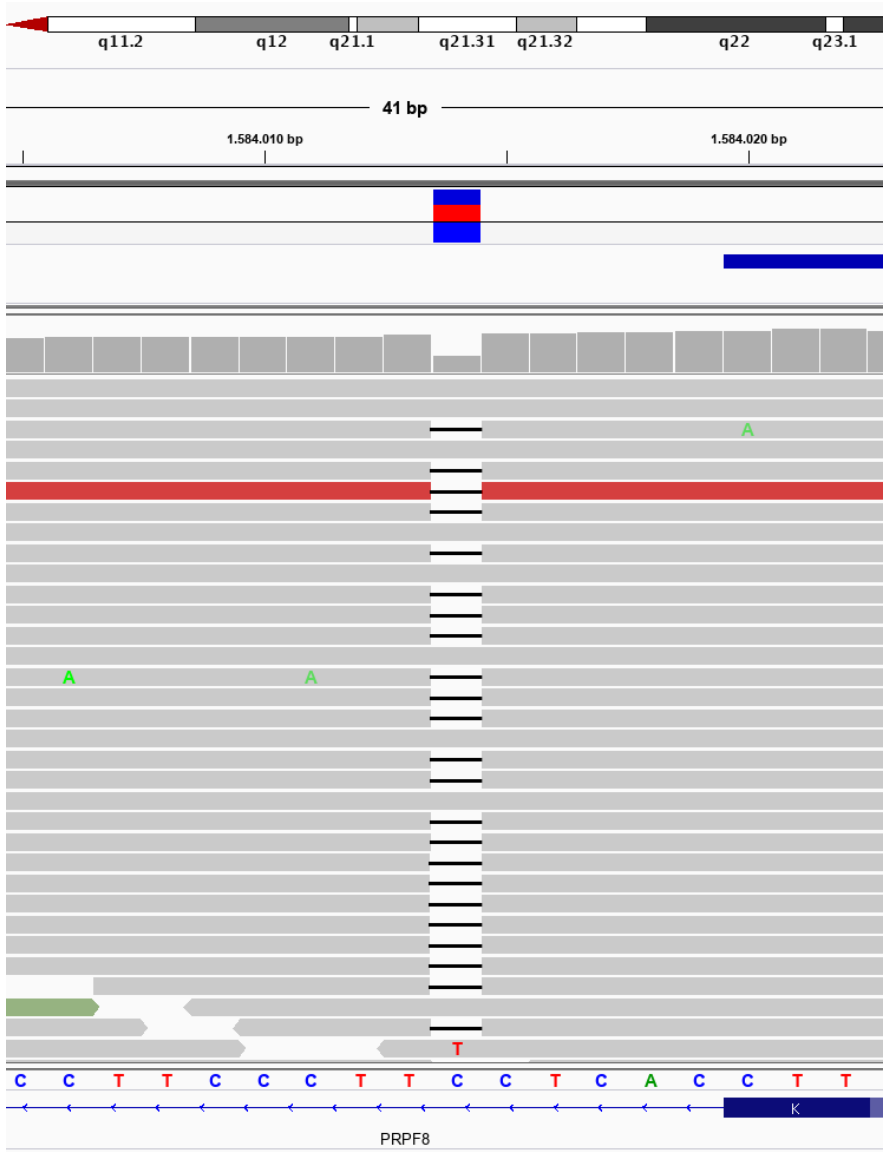
Chromosomal Position

NGS Reads

Patient 018

# Disease-associated gene:PRPF8

## c.1098+6del (48x Total Count)




Chromosome 17

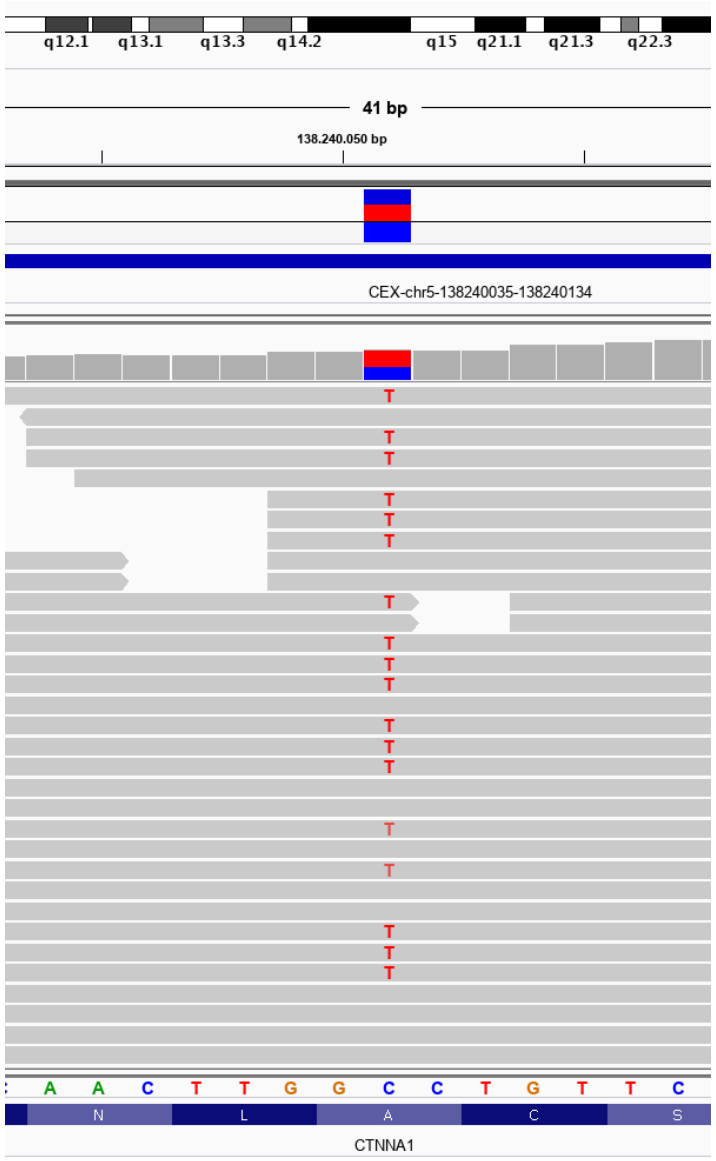
Chromosomal Position

NGS Reads

Patient 019

# Disease-associated gene: CTNNA1

## c.1310C>T (50x Total Count)




Chromosome 5

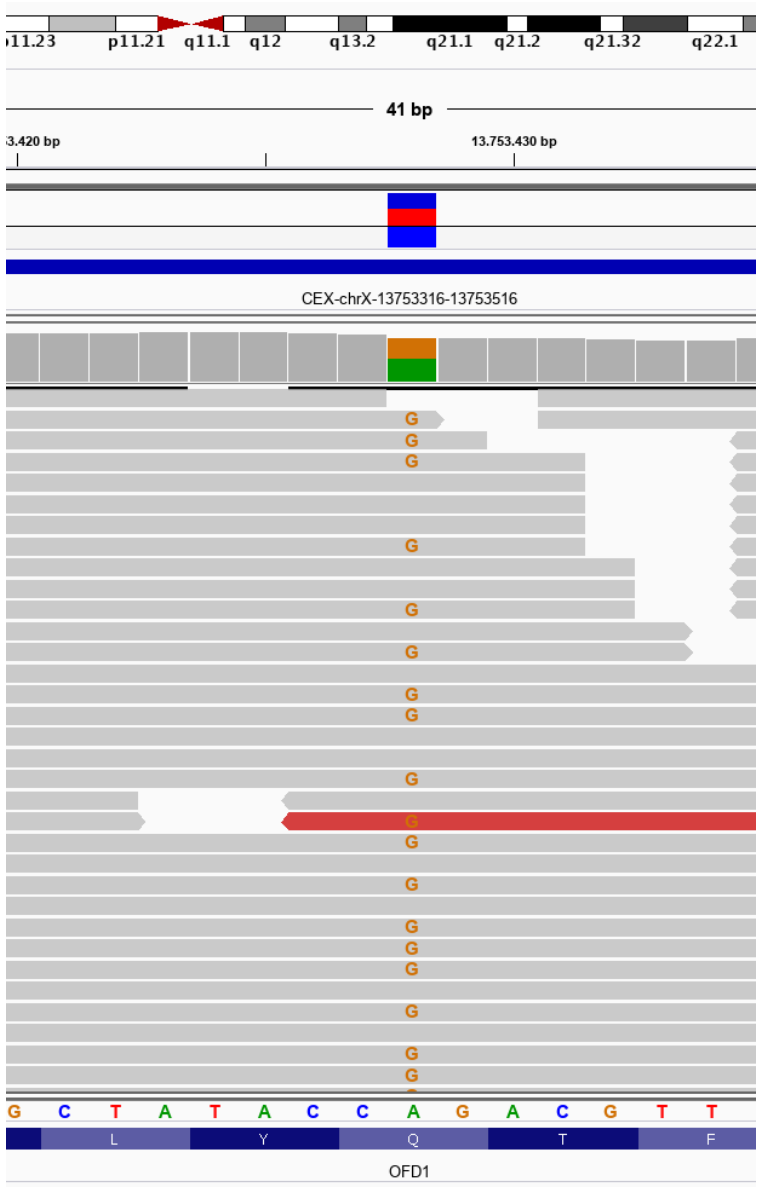
Chromosomal Position

NGS Reads

Patient 020

# Disease-associated gene: OFD1

## c.74A>G (256x Total Count)



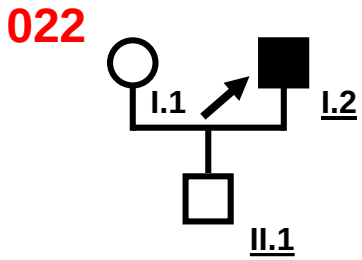

Chromosome X

Chromosomal Position

NGS Reads

Patient 022

# Disease-associated gene: ABCA4

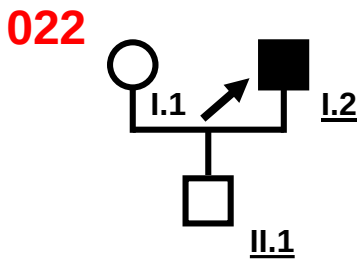



## c.5882G>A


## c.3113C>T

Patient 022

# Disease-associated gene: ABCA4



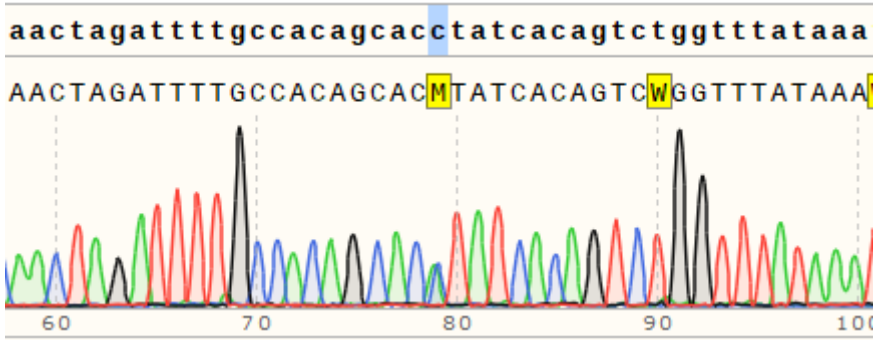

## c.1622T>C


## c.6006-5T>G

Patient 023

# Disease-associated gene: ABCA4

## c.4539+1770C>A




023 f

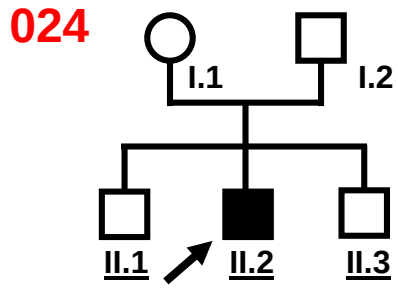
## c.5196+1015A>G


023 f

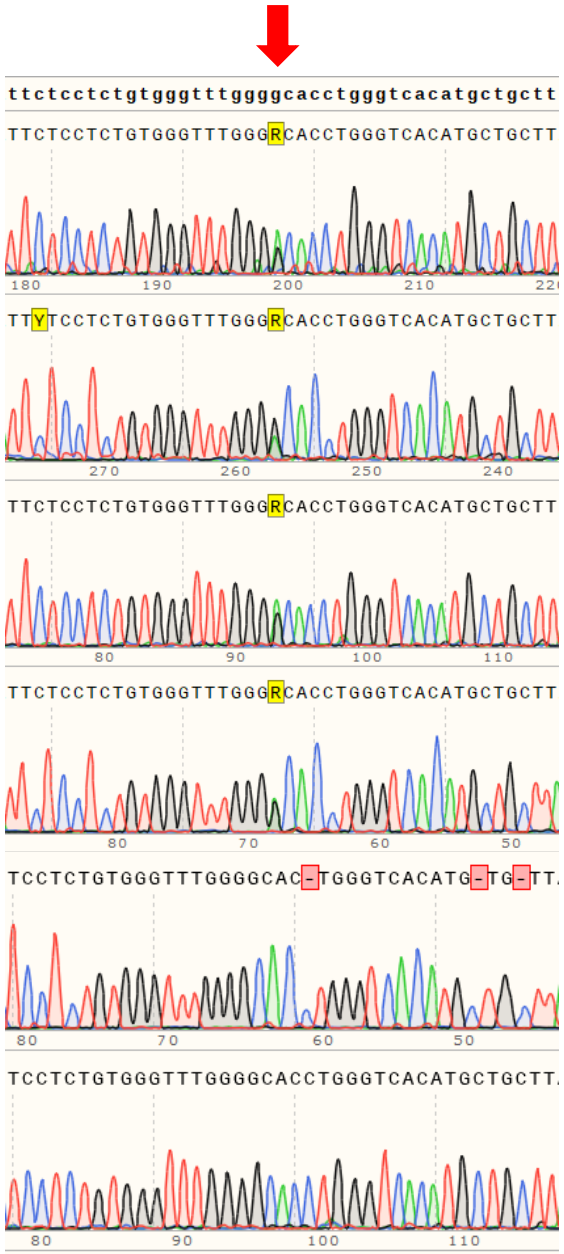
023 r

Patient 024

# Disease-associated gene: ABCA4




## c.4253+43G>A




024 II.2 f

024 II.2 r

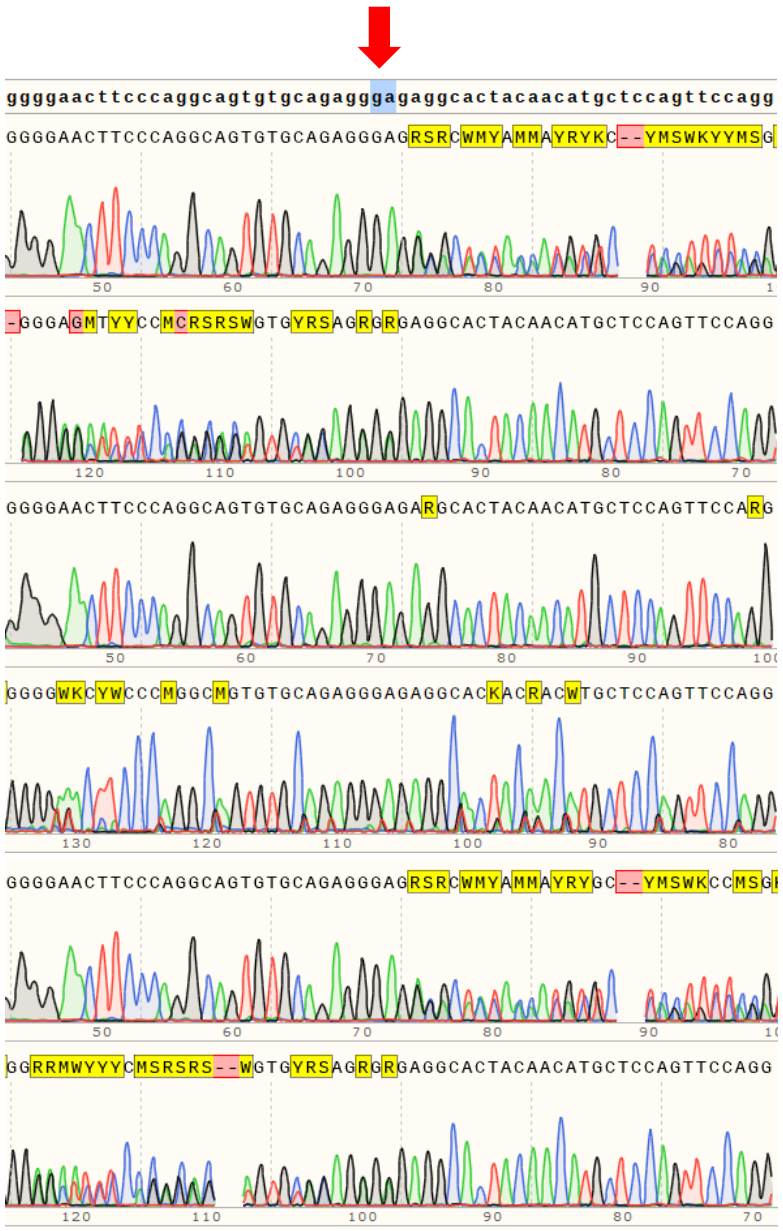
024 II.1 f

024 II.1 r

024 II.3 r

024 II.3 f

## c.6601_6602delAG




024 II.2 f

024 II.2 r

024 II.1 f

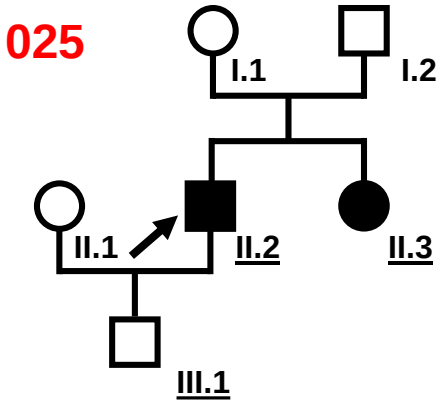
024 II.1 r

024 II.3 f

024 II.3 r

Patient 025

# Disease-associated gene: ABCA4




## c.3113C>T

025 II.2 r

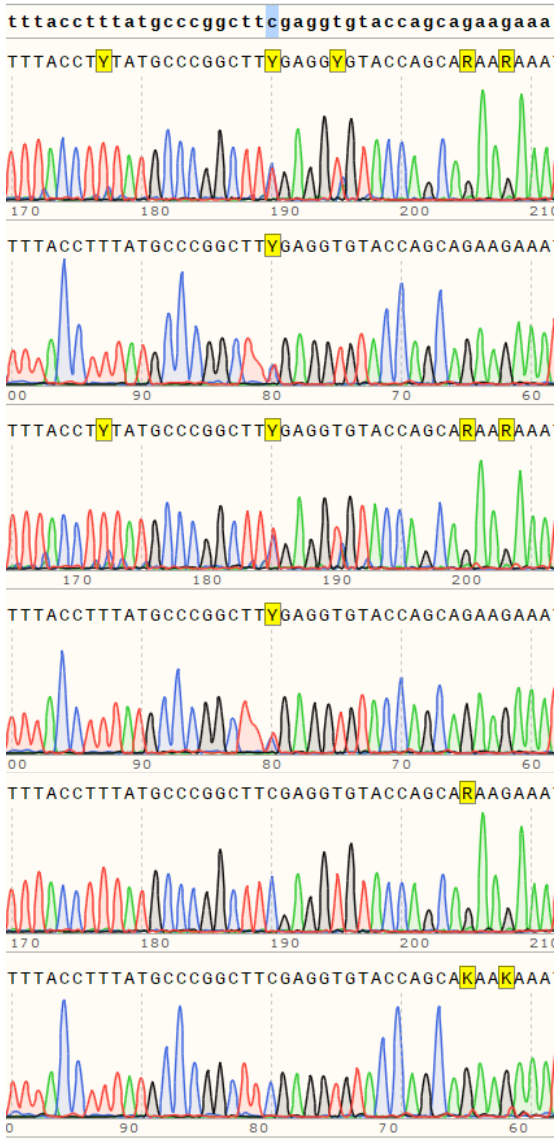
025 II.3 f

025 II.3 r

025 III.1 f

025 III.1 r

## c.6118C>T



025 II.2 f

025 II.2 r

025 II.3 f

025 II.3 r

025 III.1 f

025 III.1 r