

Supplemental Figure S3

Supplemental Figure S3. Sequence analysis of RT-PCR products from *in vitro* splice assays. Sanger sequenced electropherograms for all the non-canonical splice sites variants and wild-types (WT) are given. All the electropherograms are numbered according to the fragments in Supplemental Fig. S2. Electropherograms obtained after gel purification are indicated by letter “G” encircled in green while letter “P” encircled in blue indicates the results of pGEM cloned bands. Black triangles indicates the substituted nucleotides in the mutant clone. The # symbol indicates that BA20 RT-PCR was performed using primers spanning *ABCA4* exons 29-31 instead of exons 28-31, yellow highlighted sequence for c.4538A>C_fr2 indicates the 29-nt elongation of exon 30 instead of expected 30-nt considered as a cloning artifact. Question mark (?) for BA23 fragment c.4773+5G>A indicates the 9-nt deletion of exon 33 due to 9-nt difference in size compared to wild-type fragment cannot be seen on gel but captured in pGEM cloning.

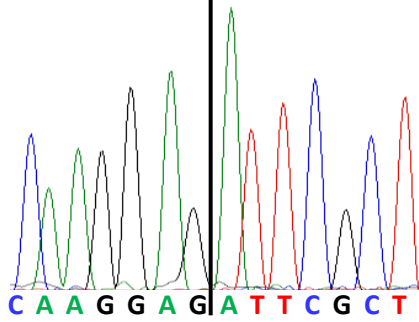
BA1

BA1_WT_fr1

G

RHO-e3

ABCA4-e2

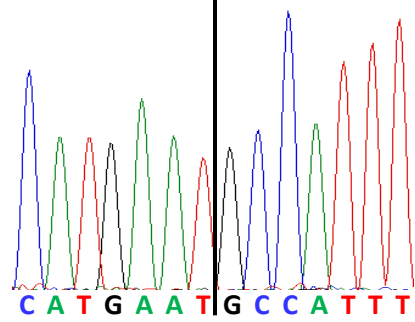


BA1_WT_fr1

G

e2

e3

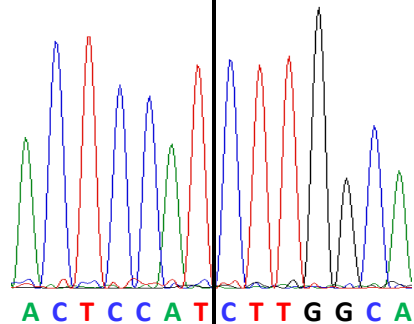


BA1_WT_fr1

G

e3

e4

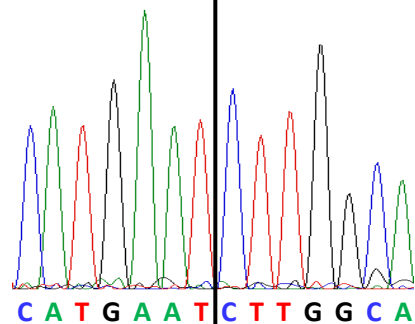


BA1_WT_fr5

G

e2

e4

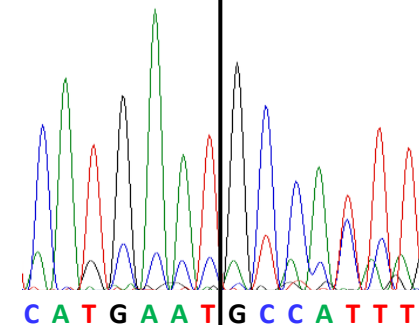


BA1_c.160+5G>C_fr1

G

e2

e3

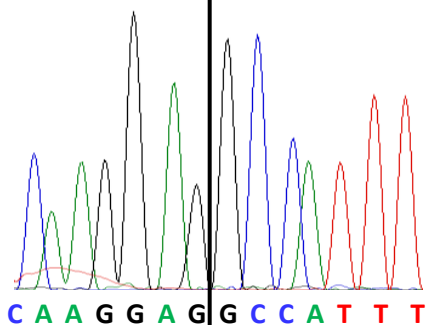


BA1_c.160+5G>C_fr3

P

RHO-e3

ABCA4-e3



BA1_c.160+5G>C_fr4

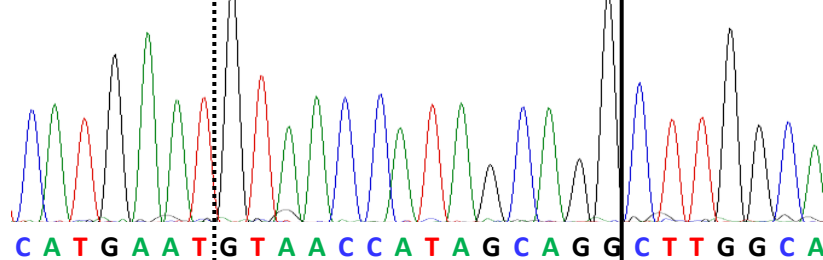
P

e2

i2

e4

← 14 nt →

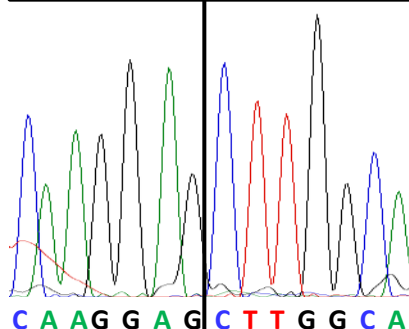


BA1

BA1_c.160+5G>C_fr6

G

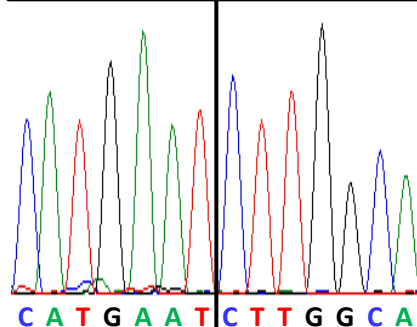
<i>RHO</i> -e3	<i>ABCA4</i> -e4
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BA1_c.161G>T_fr2

G

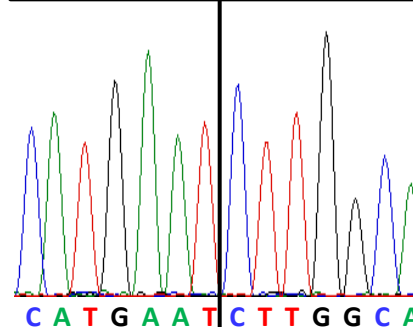
e2	e4
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BA1_c.302+4A>C_fr2

G

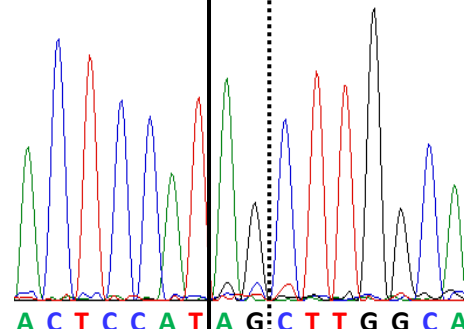
e2	e4
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BA1_c.303-3C>G_fr3

G

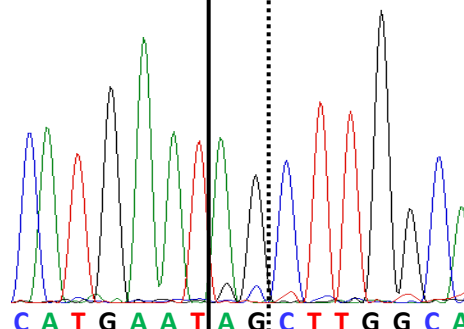
e3	i3	e4
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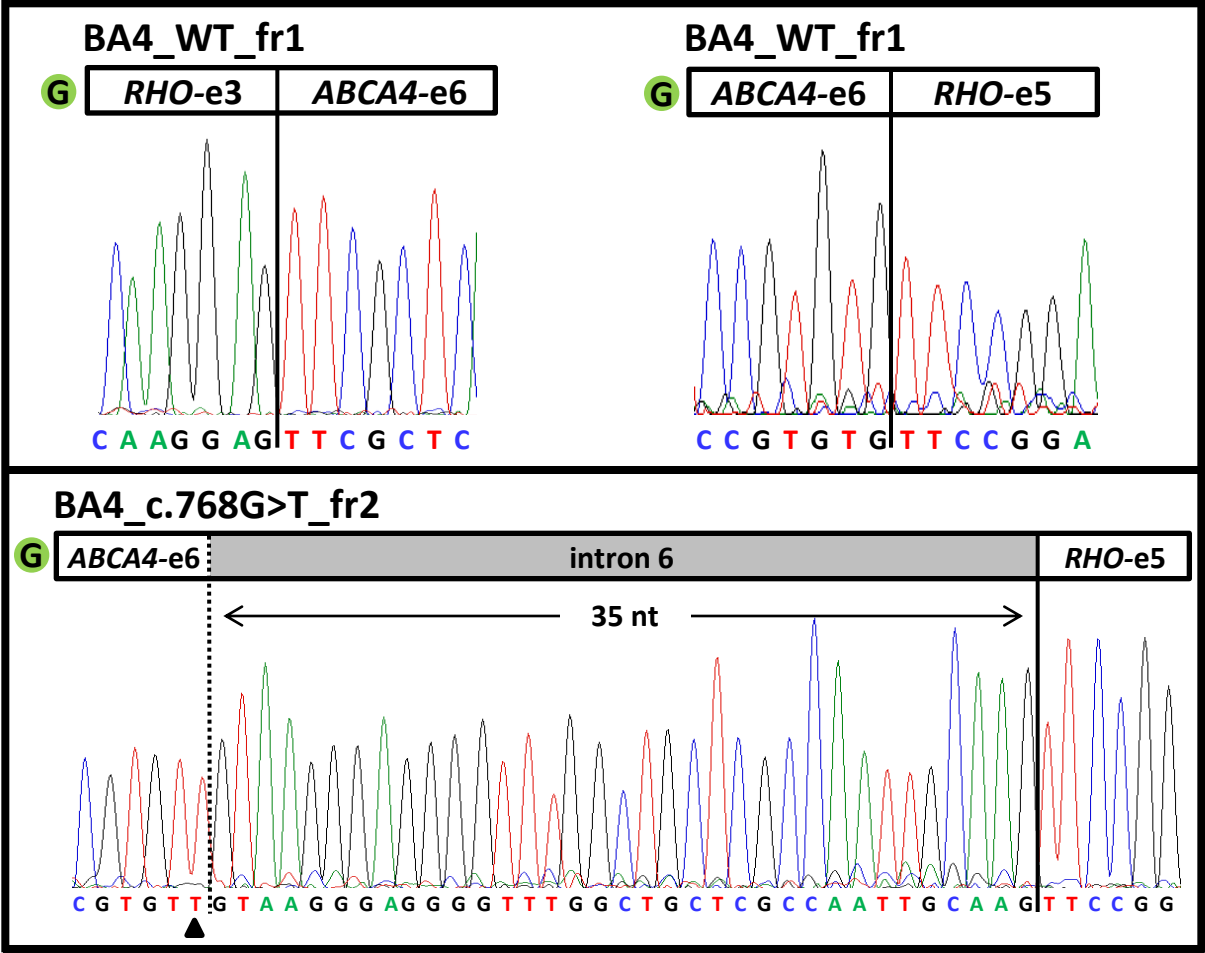
BA1_c.303-3C>G_fr4

G

e2	i3	e4
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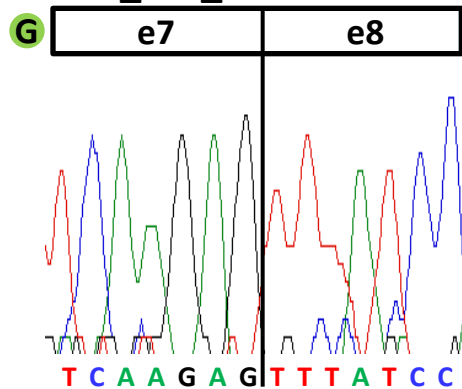


BA4

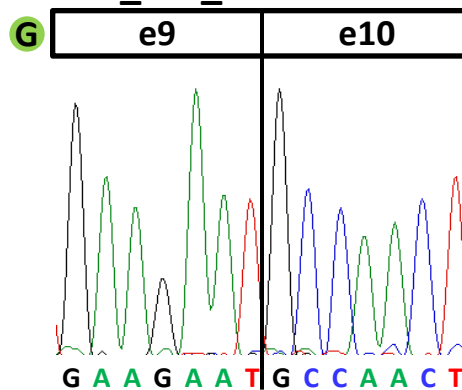


BA7

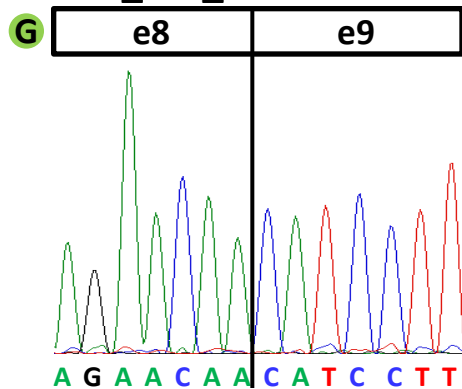
BA7_WT_fr1



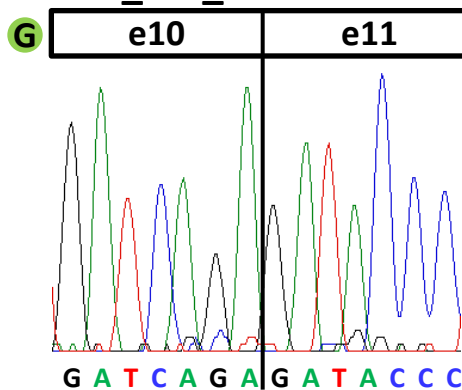
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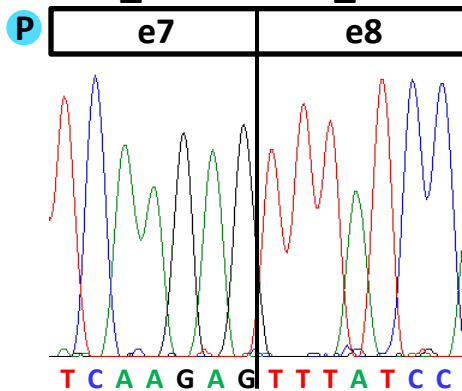
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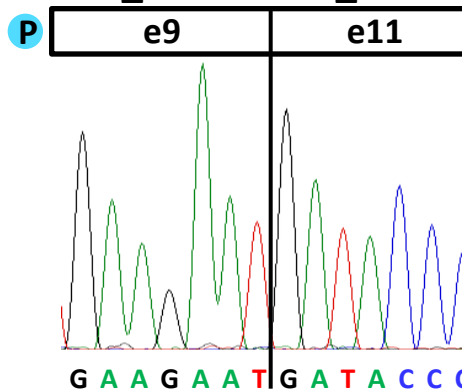
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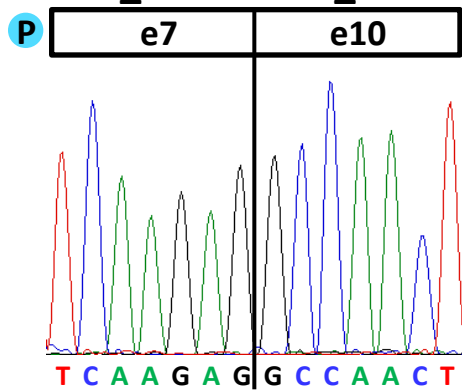
BA7_c.859-9T>C_fr1



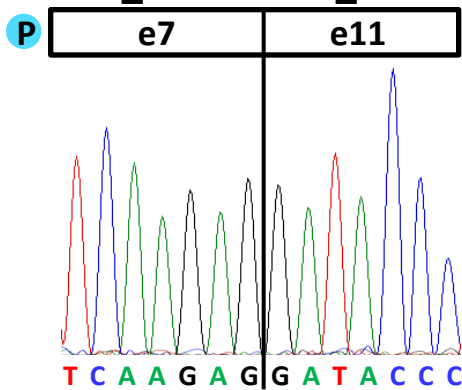
BA7_c.859-9T>C_fr2



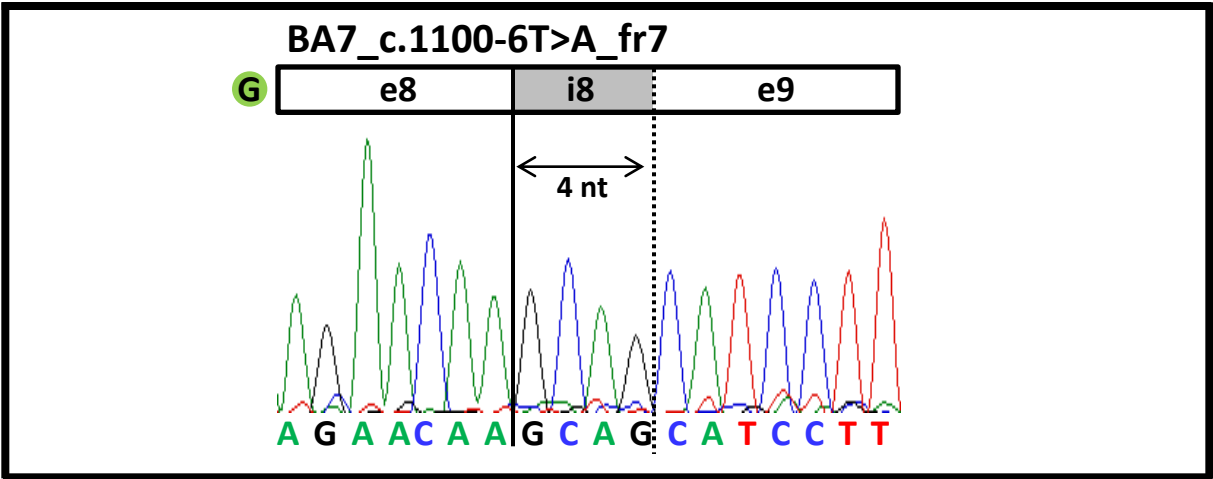
BA7_c.859-9T>C_fr5



BA7_c.859-9T>C_fr6

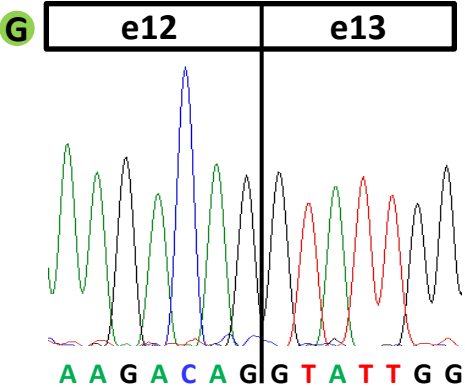


BA7

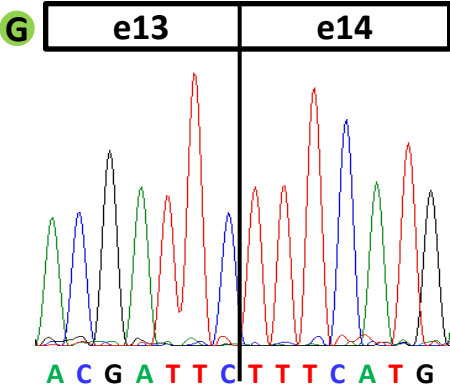


BA11

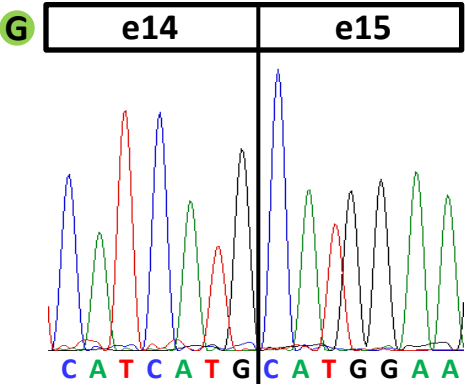
BA11_WT_fr1



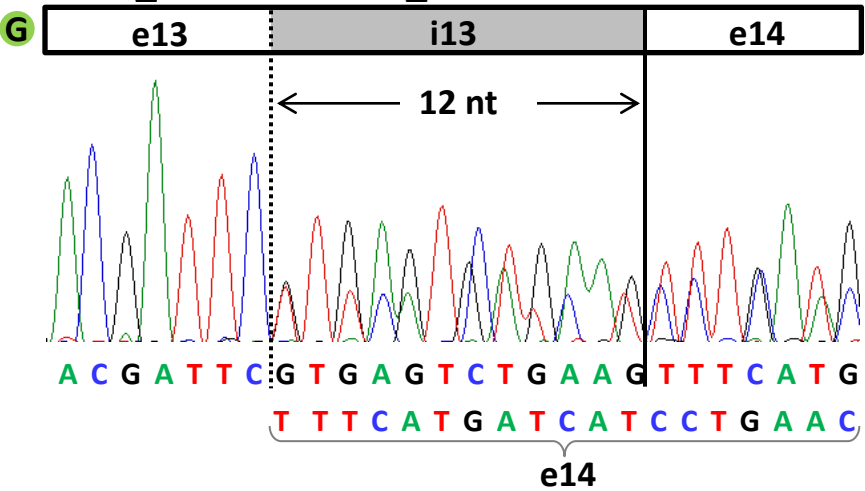
BA11_WT_fr1



BA11_WT_fr1

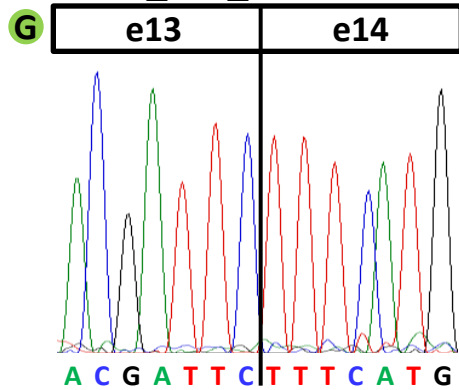


BA11_c.1937+13T>G_fr2

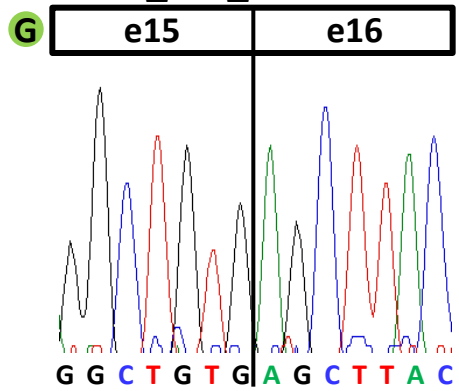


BA12

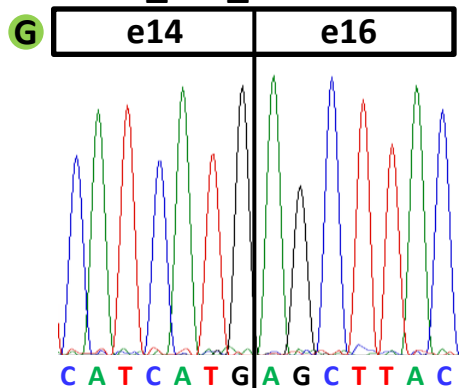
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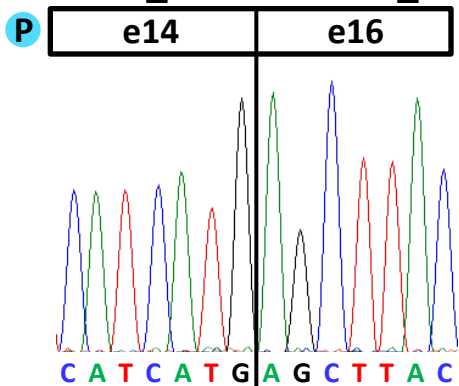
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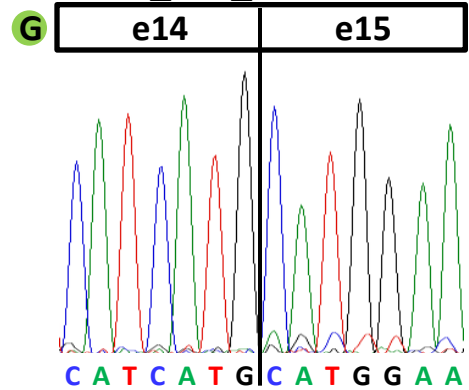
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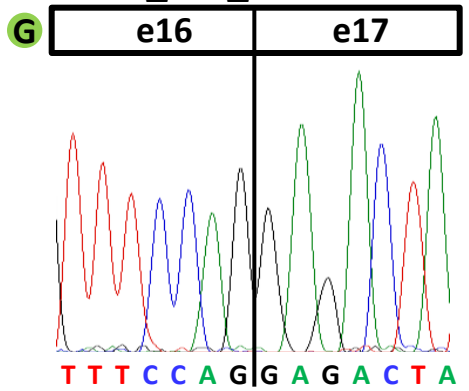
BA12_c.2382+5G>C_fr2



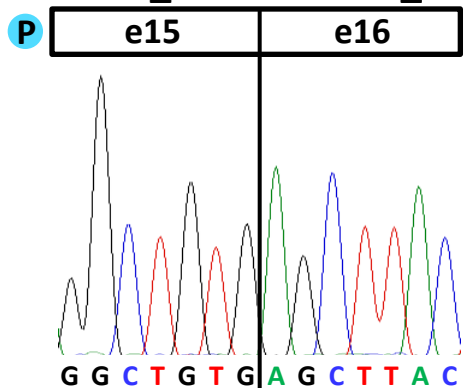
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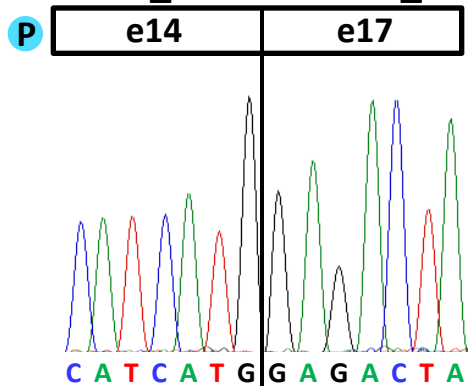
BA12_WT_fr1



BA12_c.2382+5G>C_fr1

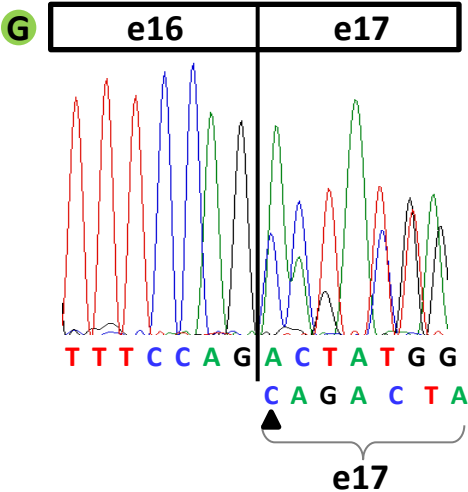


BA12_c.2382+5G>C_fr5

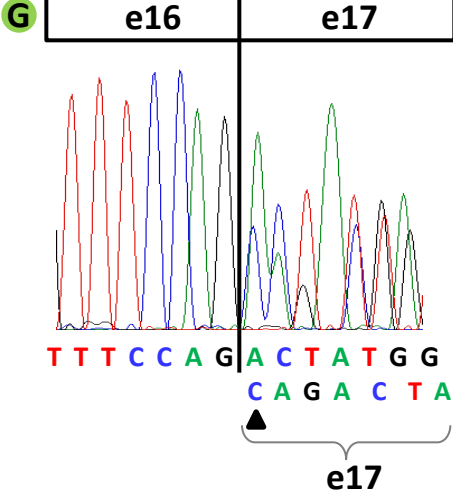


BA12

BA12_c.2588G>C_fr1/6

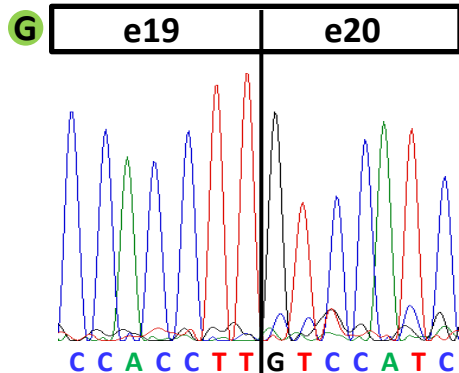


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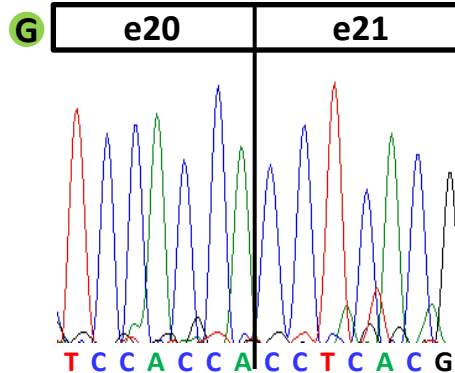


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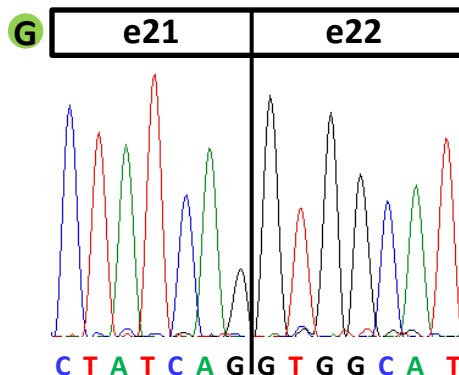
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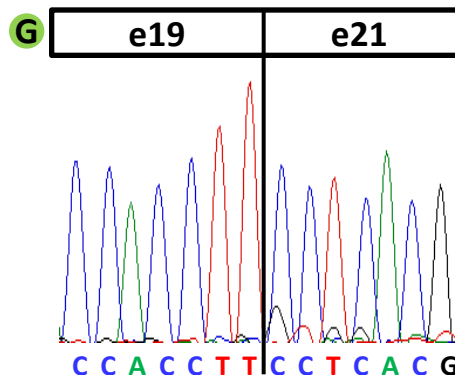
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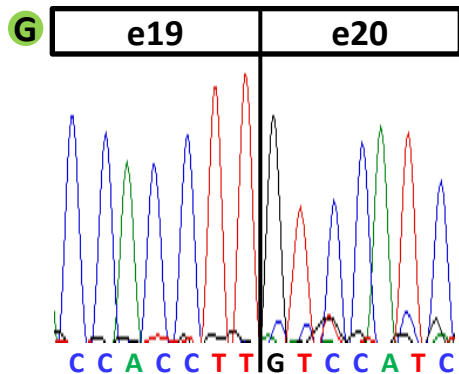
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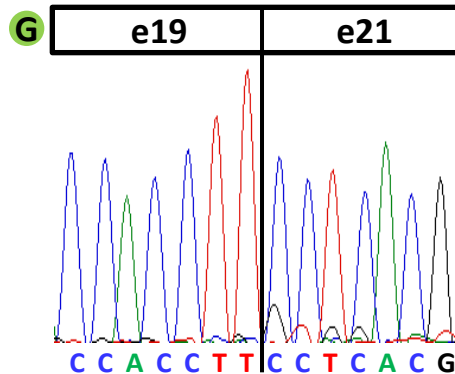
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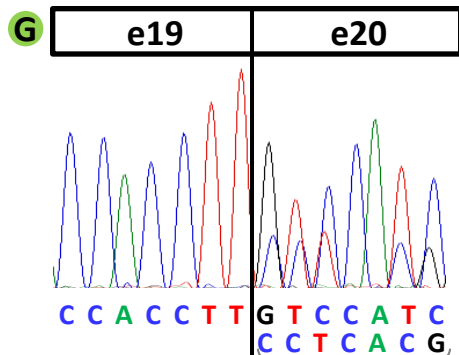
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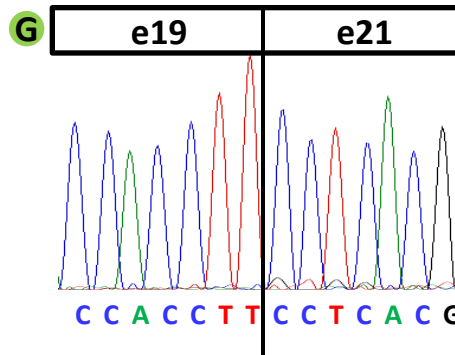
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BA15_c.2919-10T>C_fr1



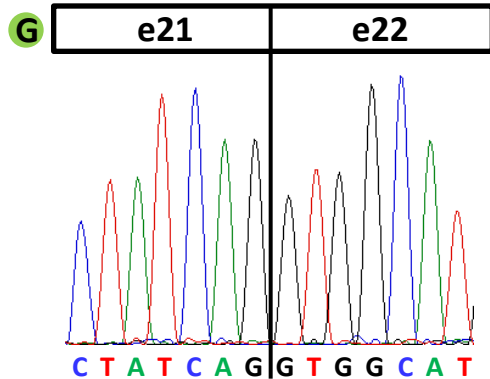
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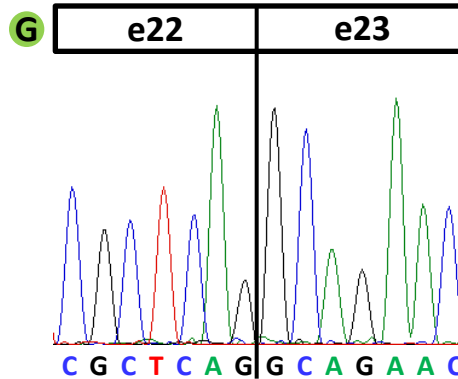
e21 (contamination fr. 2)

BA16

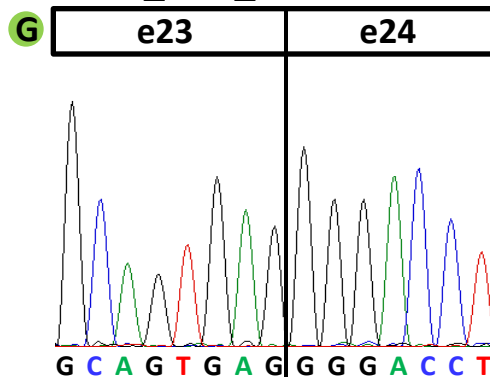
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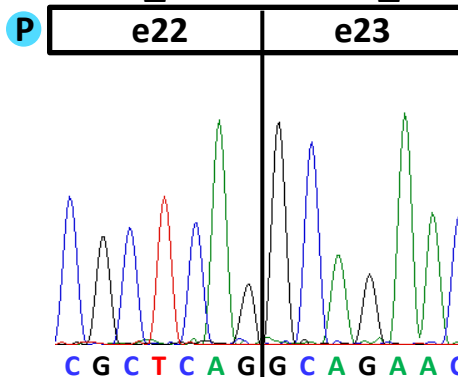
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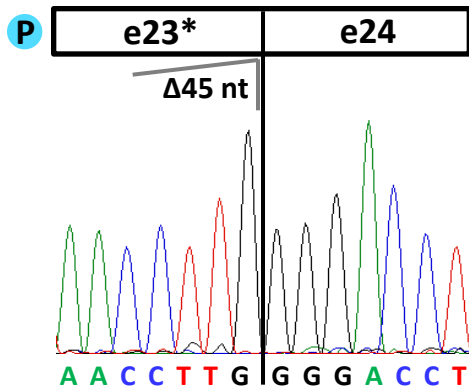
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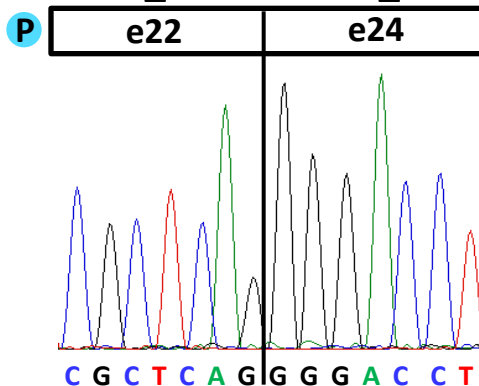
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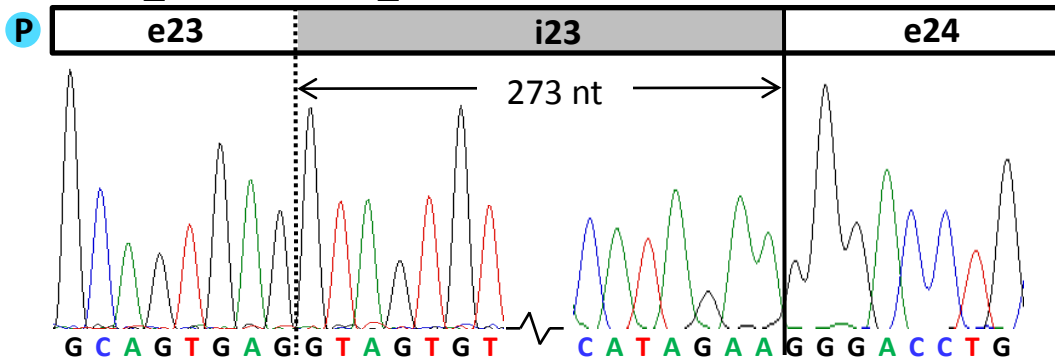
BA16_c.3522+5del_fr8



BA16_c.3522+5del_fr9



BA16_c.3522+5del_fr6

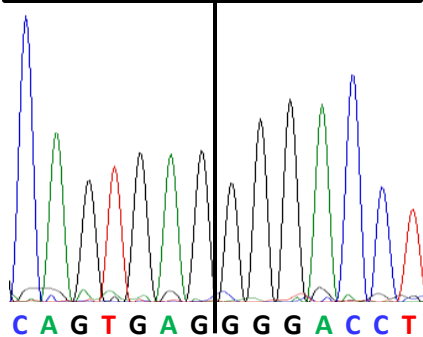


BA17

BA17_WT_fr1

G

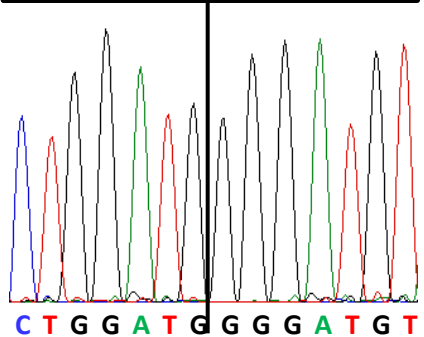
e23	e24
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BA17_WT_fr1

G

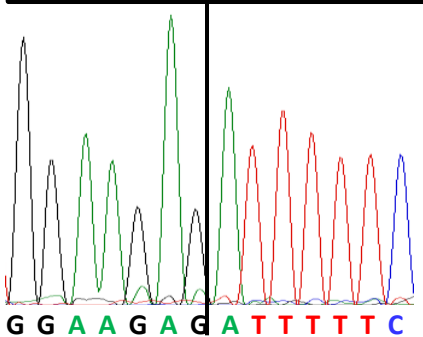
e24	e25
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BA17_WT_fr1

G

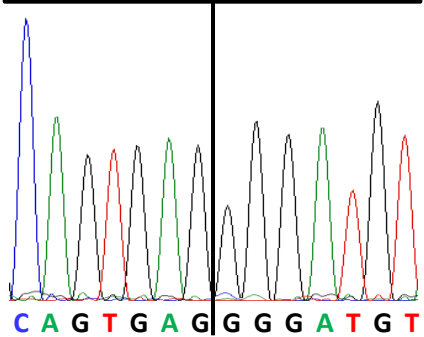
e25	e26
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BA17_c.3607+3A>T_fr3

G

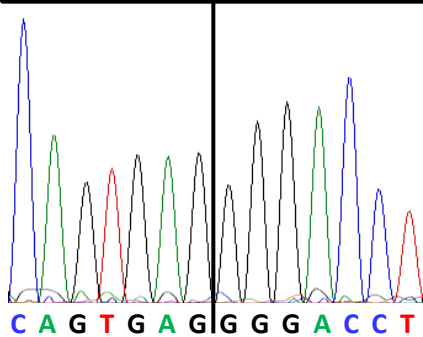
e23	e25
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BA17_c.3607G>A_fr1

G

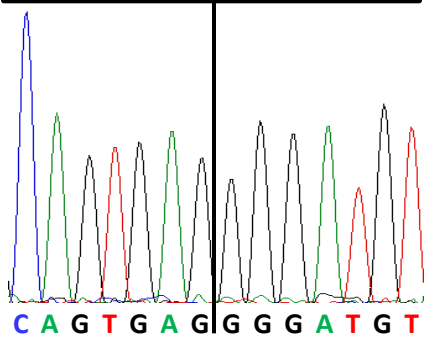
e23	e24
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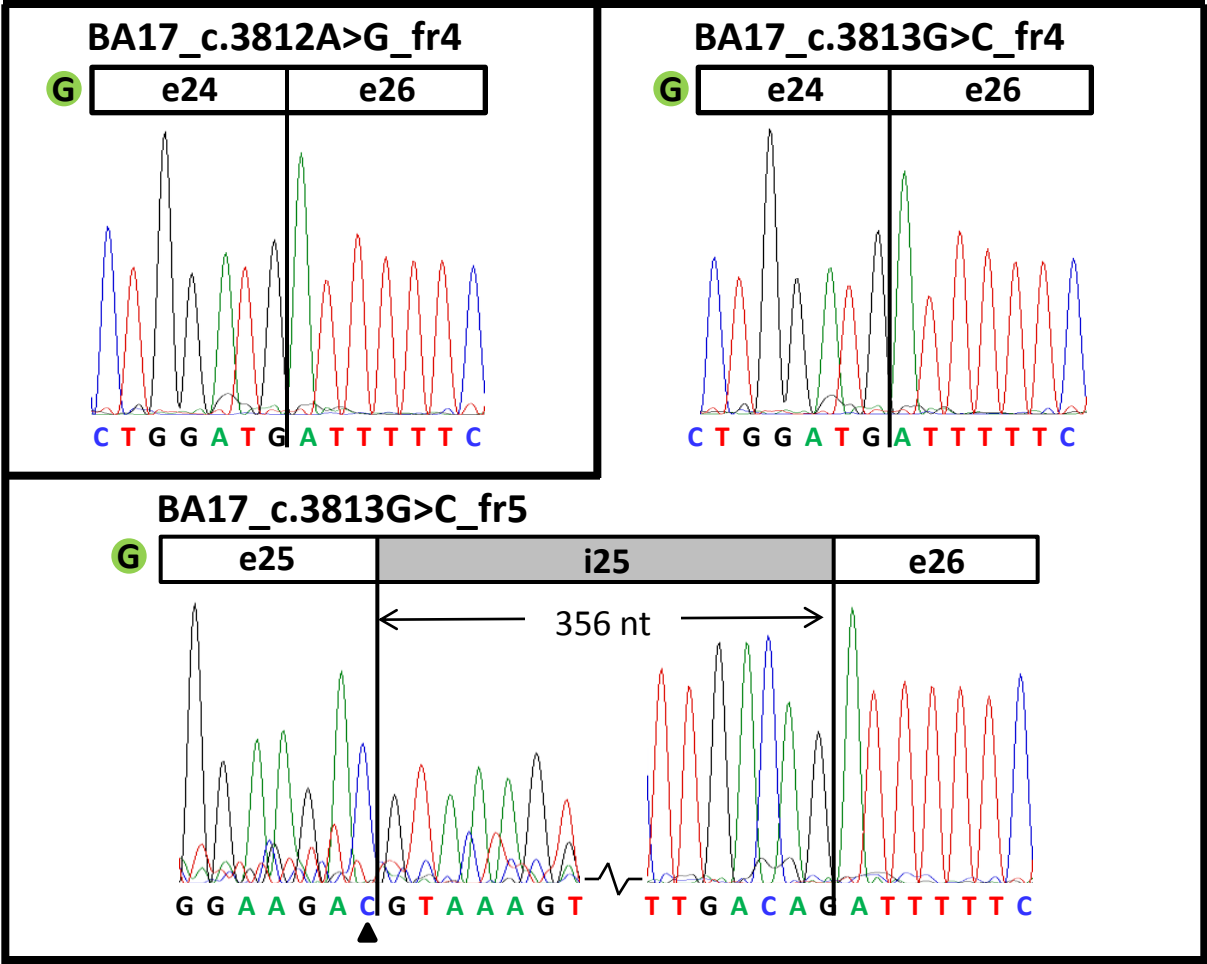
BA17_c.3607G>A_fr3

G

e23	e25
-----	-----

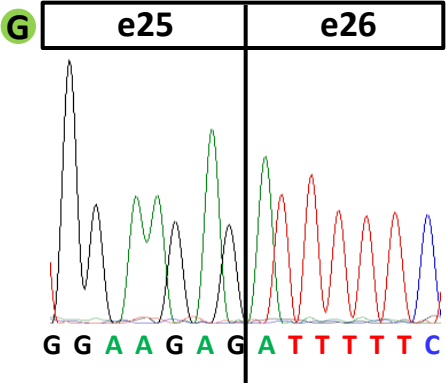


BA17

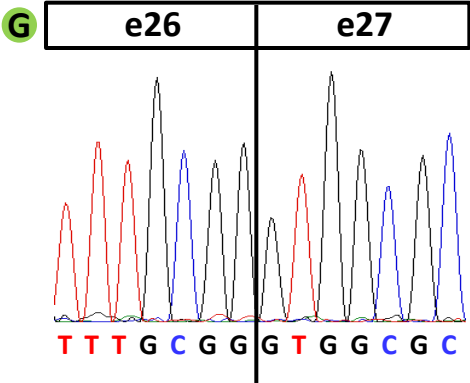


BA18

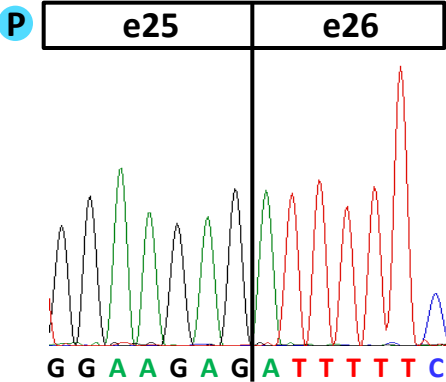
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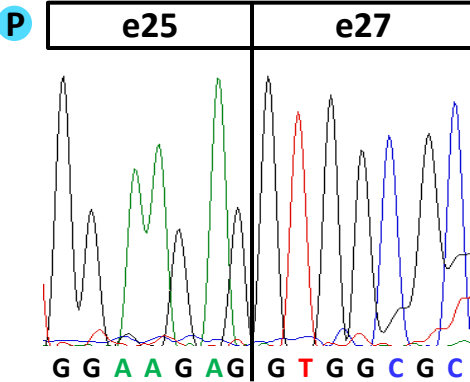
BA18_WT_fr1



BA18_c.3862+3A>G_fr1

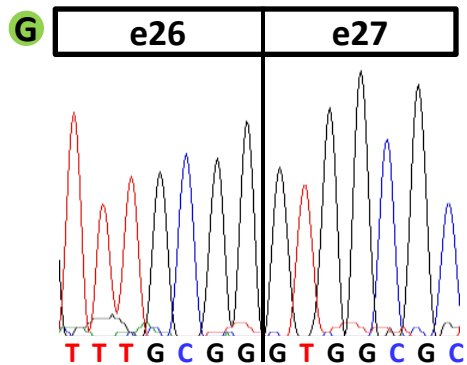


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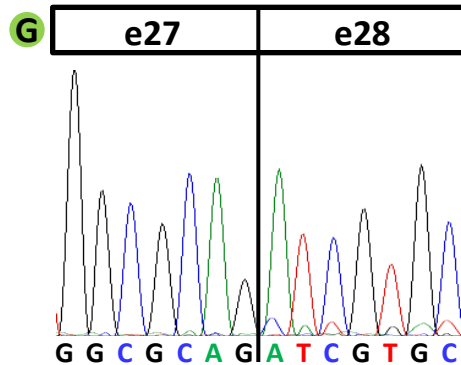


BA19

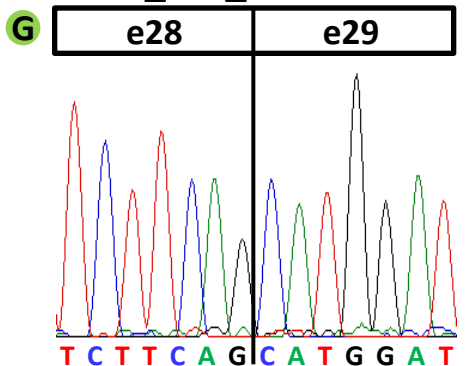
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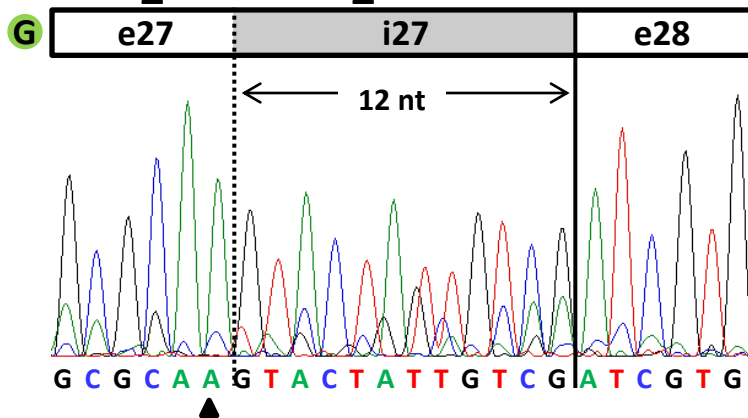
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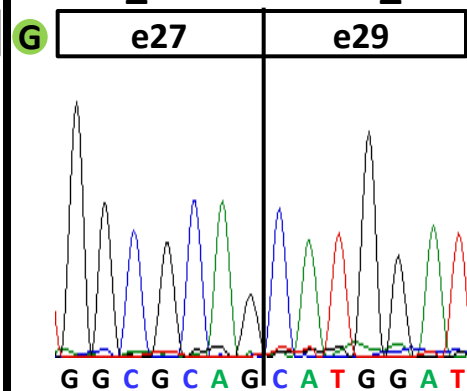
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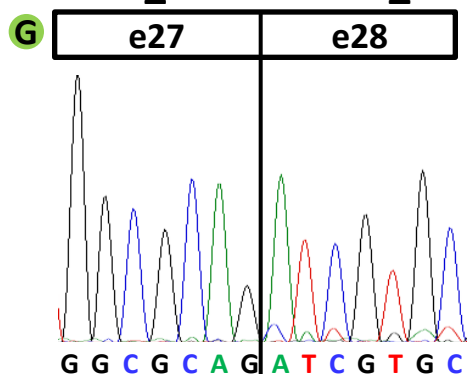
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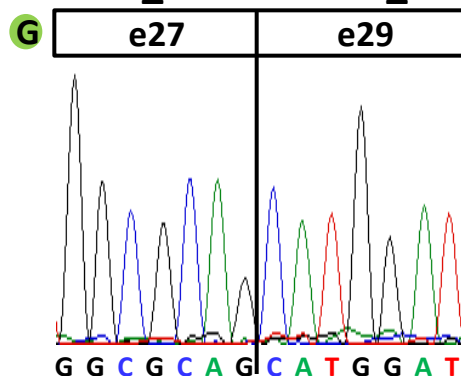
BA19_c.4253+5G>A_fr2



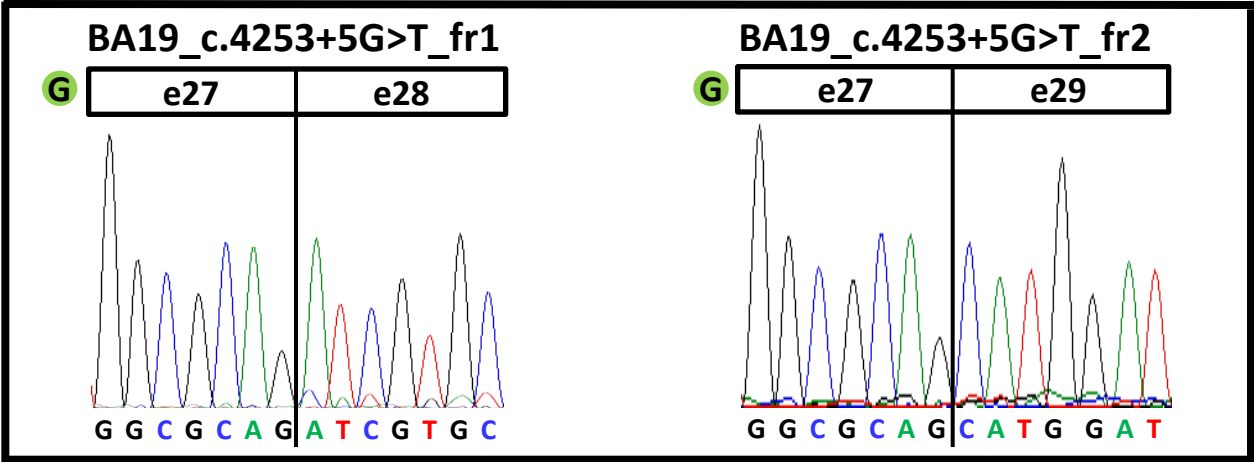
BA19_c.4253+4C>T_fr1



BA19_c.4253+4C>T_fr2



BA19



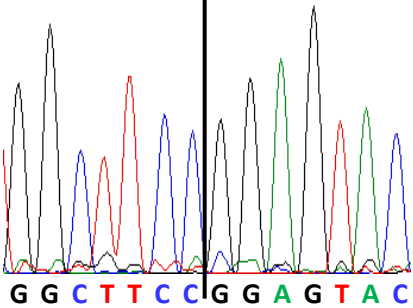
BA20#

BA20_WT_fr1

G

e29

e30

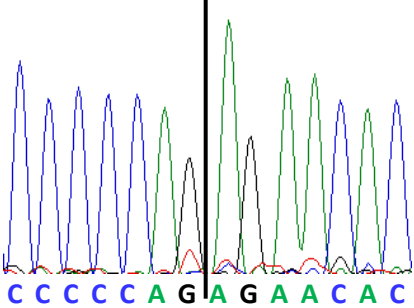


BA20_WT_fr1

G

e30

e31



BA20_c.4538A>G_fr2

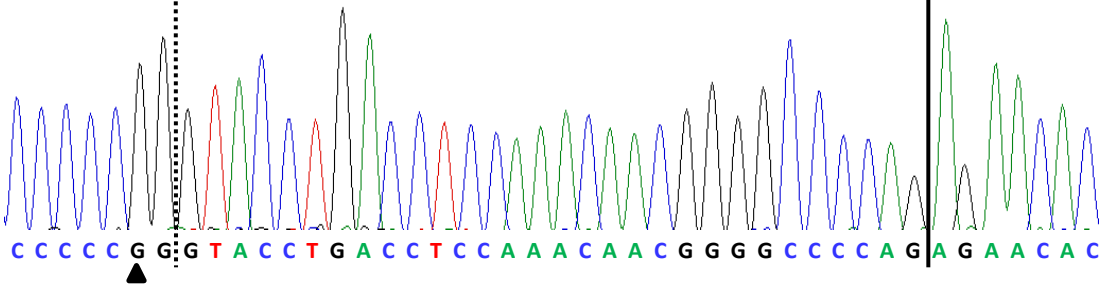
P

e30

i30

e31

30 nt



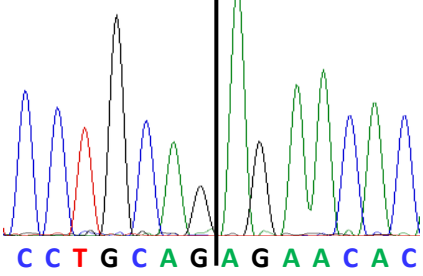
BA20_c.4538A>G_fr3

P

e30*

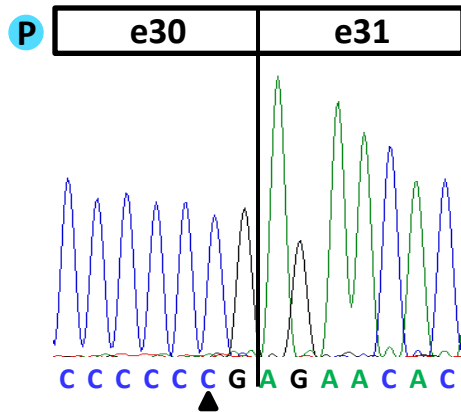
e31

Δ73 nt

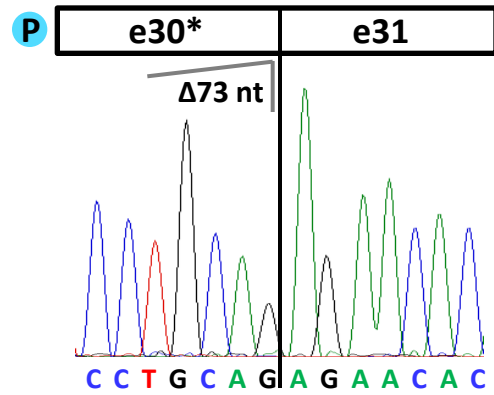


BA20

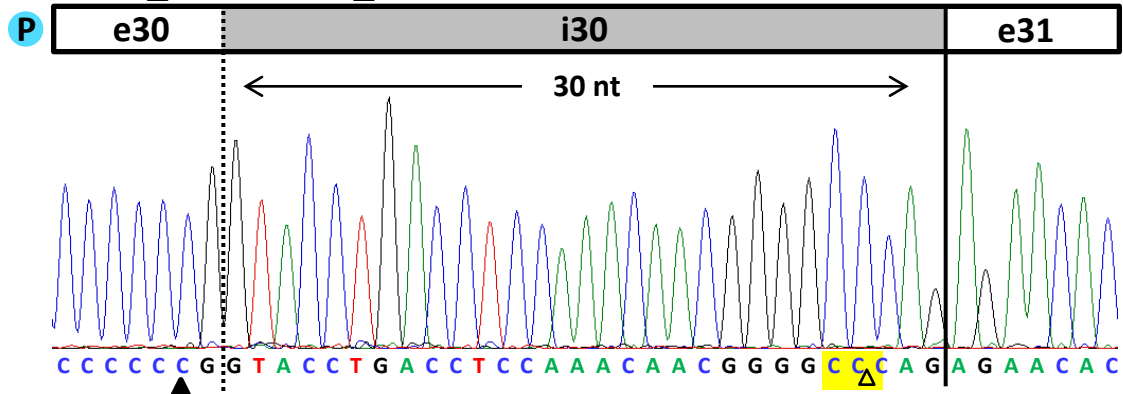
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BA20_c.4538A>C_fr3

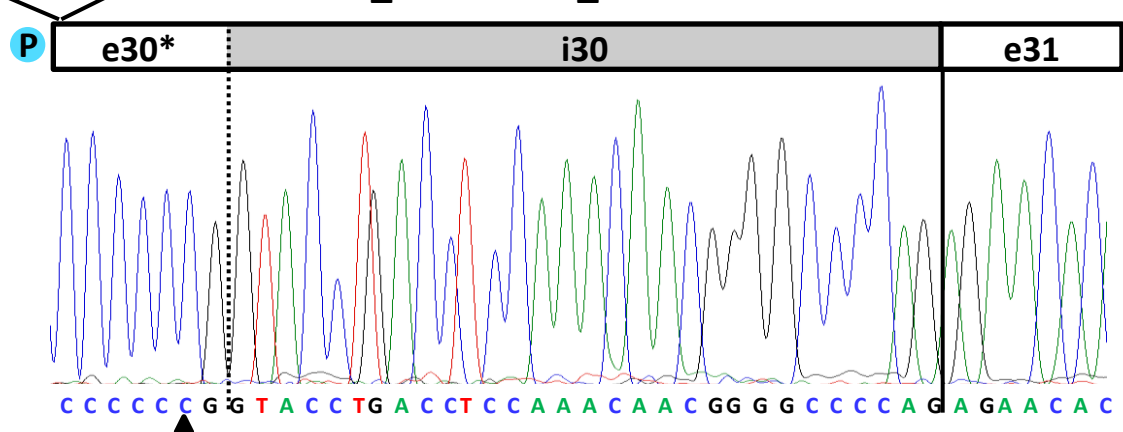


BA20_c.4538A>C_fr2



Δ 114nt

BA20_c.4538A>C_fr4



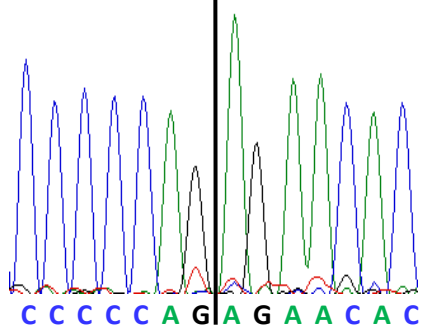
BA21

BA21_WT_fr1

G

e30

e31

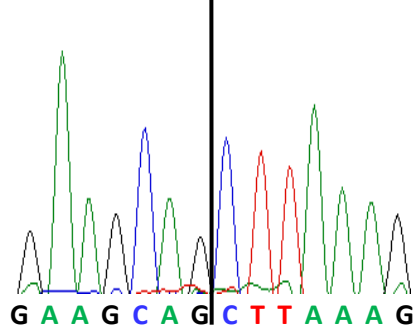


BA21_WT_fr1

G

e31

e32

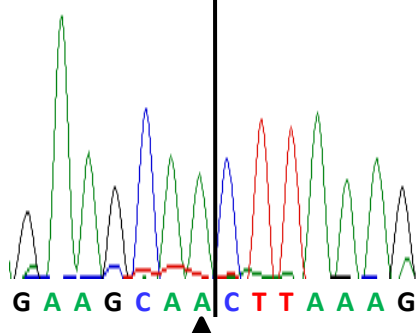


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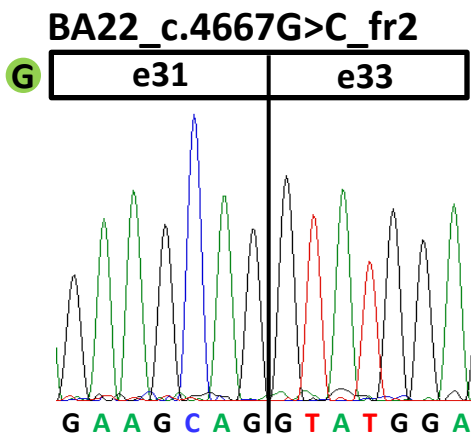
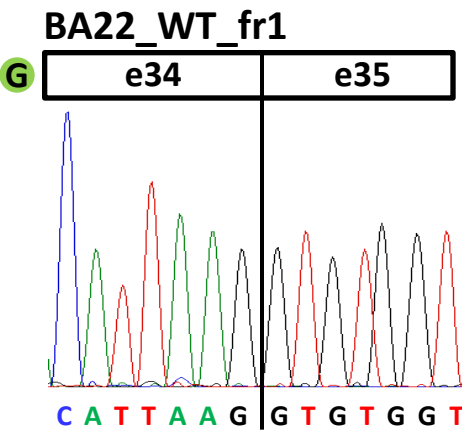
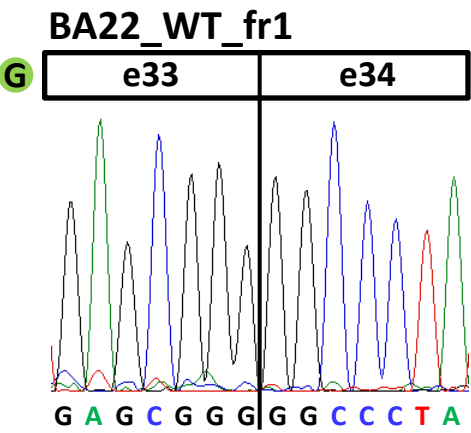
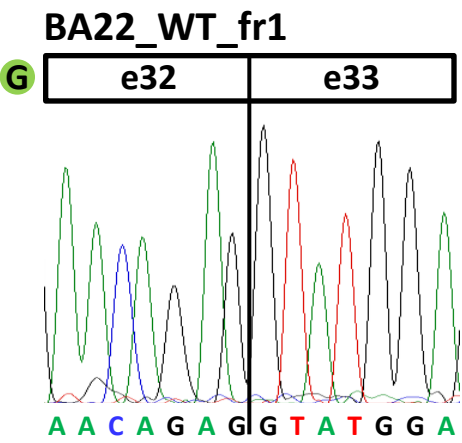
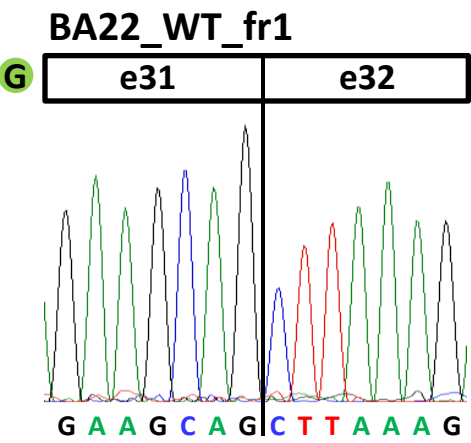
G

e31

e32

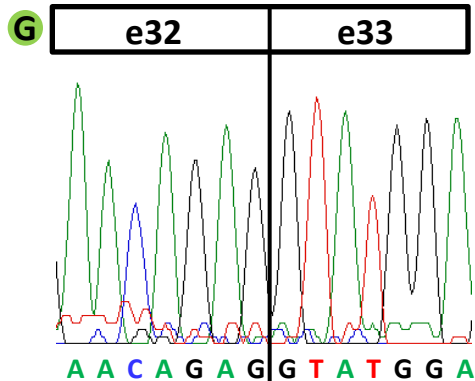


BA22

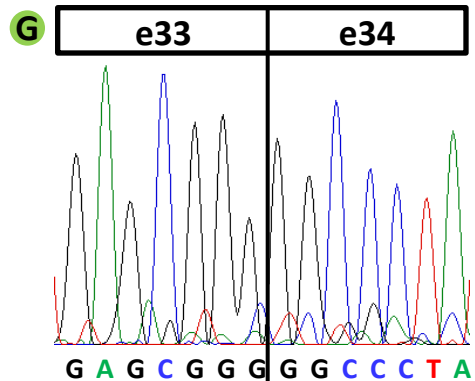


BA23

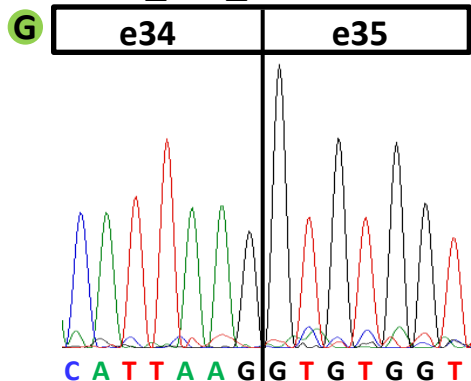
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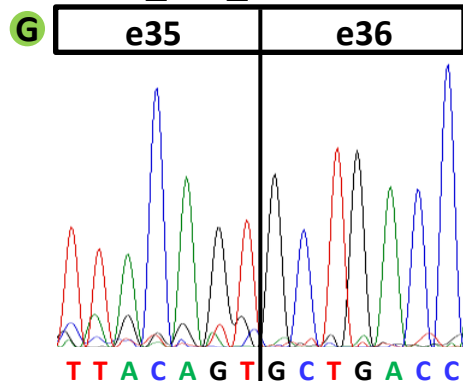
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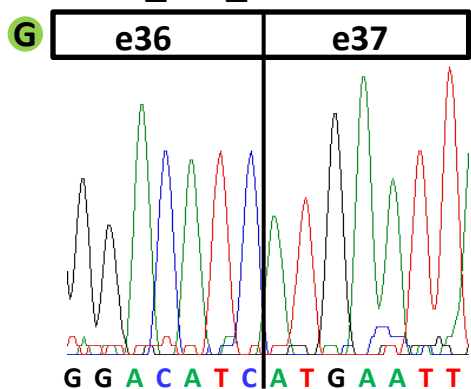
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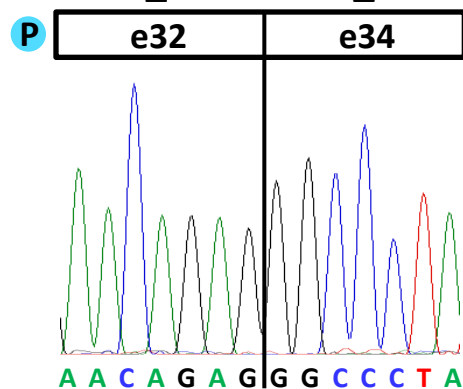
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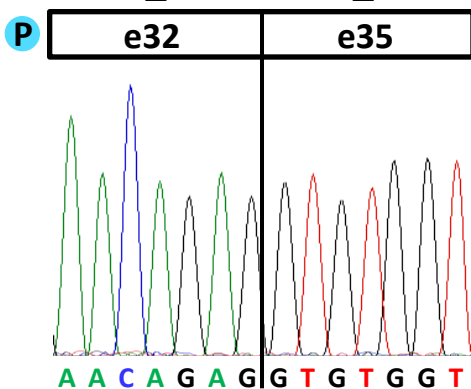
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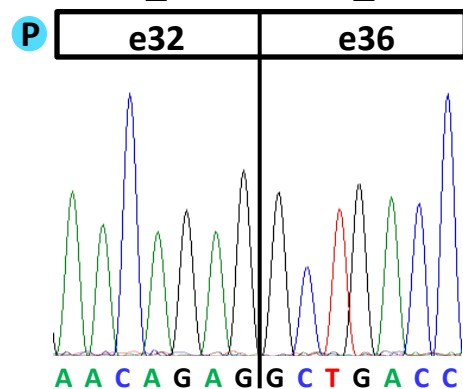
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BA23_c.4773G>C_fr4

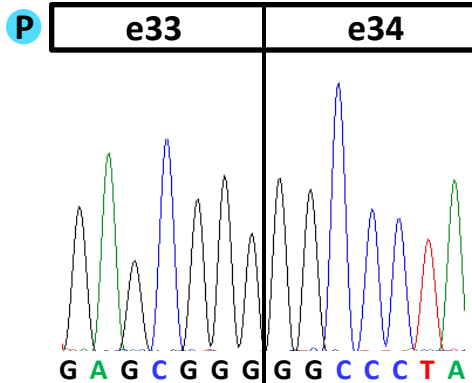


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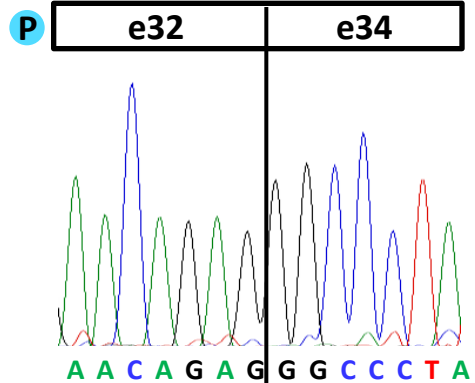


BA23

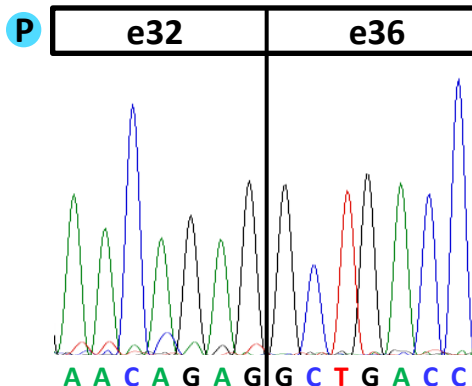
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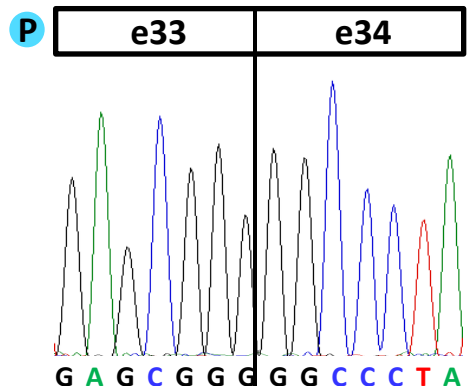
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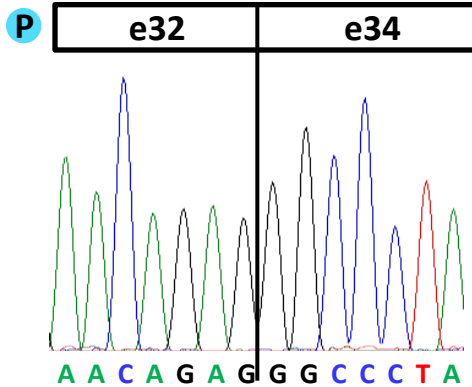
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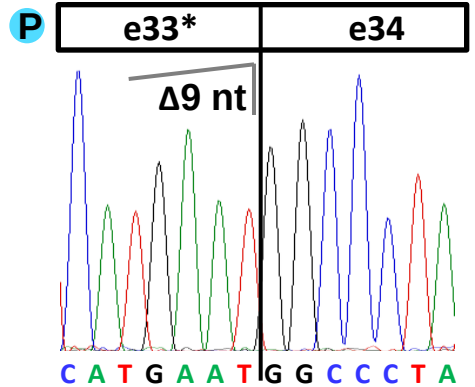
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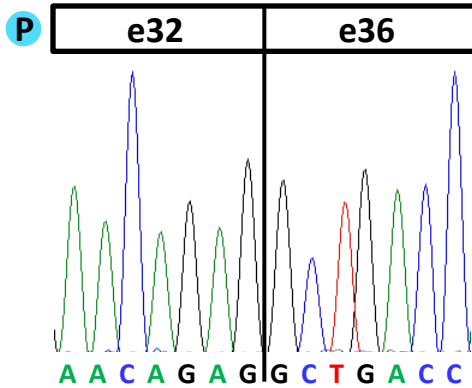
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BA23_c.4773+5G>A_fr?

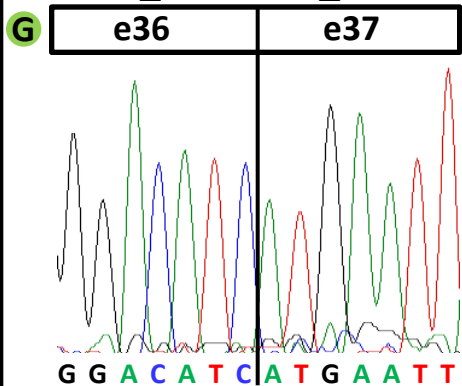


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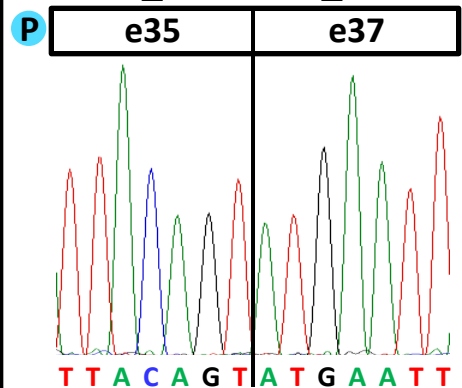


BA23

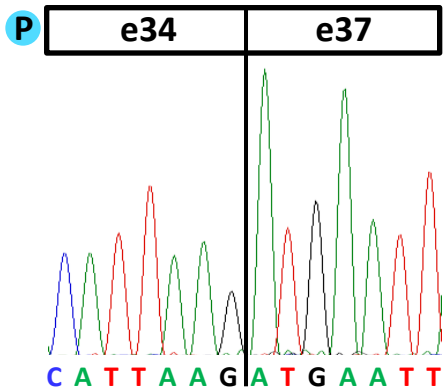
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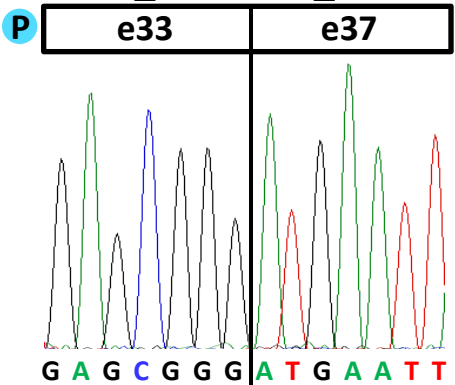
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BA23_c.5196+3_5196+6del_fr9

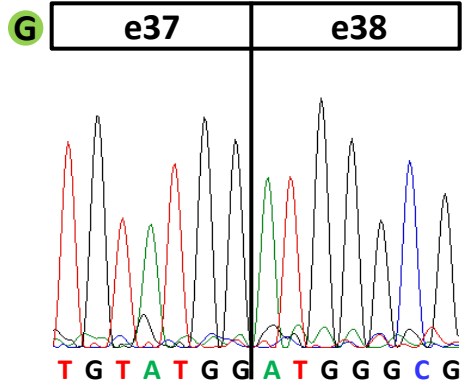


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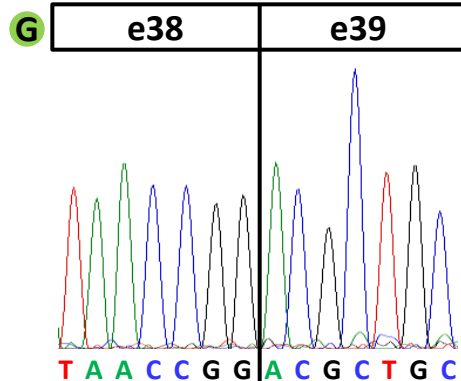


BA25

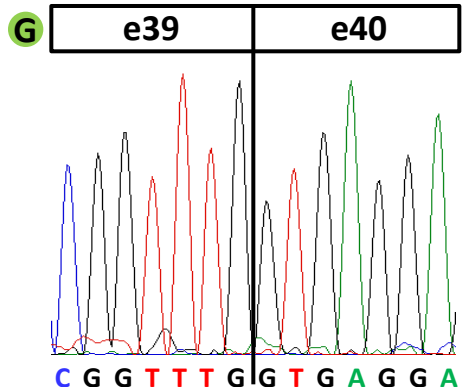
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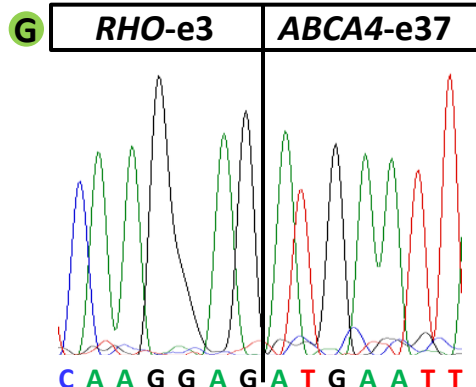
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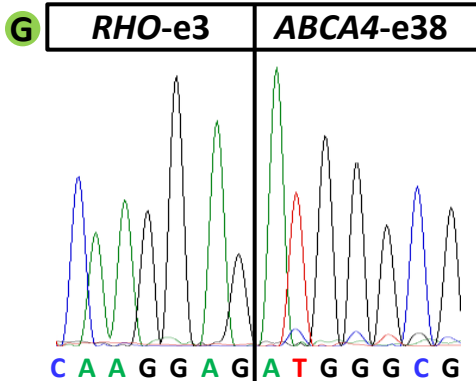
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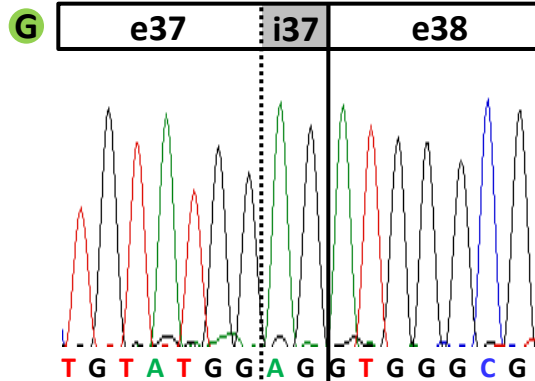
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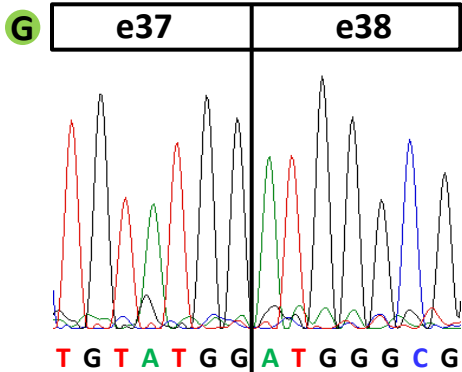
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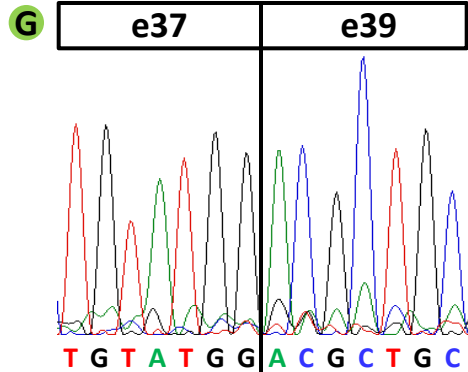
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BA25_c.5460+5G>A_fr5

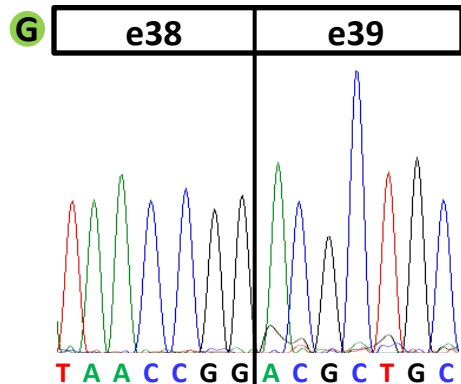


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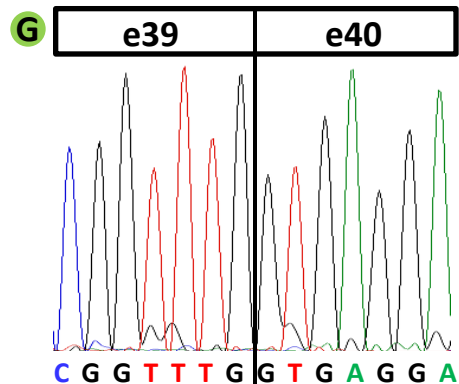


BA26

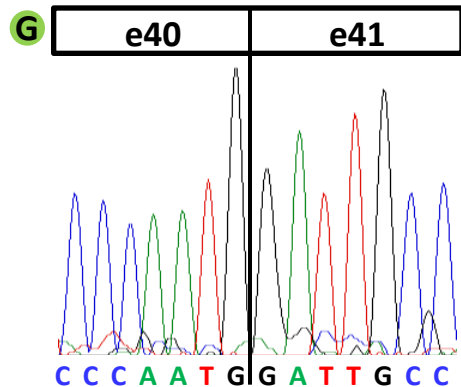
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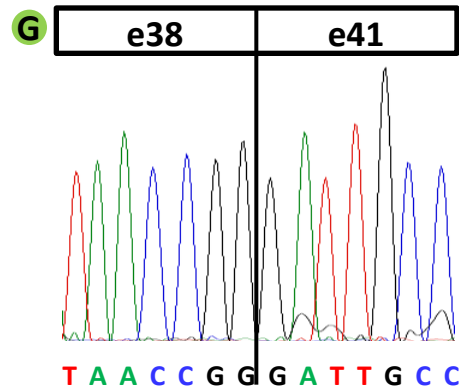
BA26_WT_fr1



BA26_WT_fr1



BA26_WT_fr5

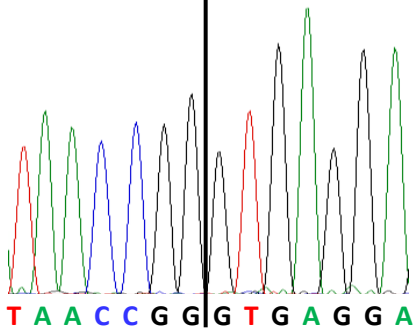


BA26

BA26_c.5461-10T>C_fr3

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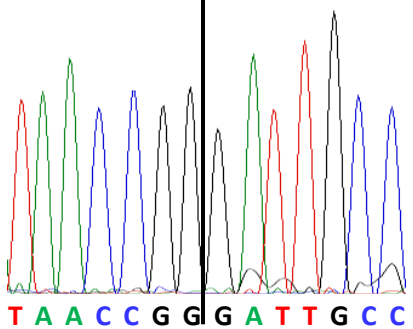
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G

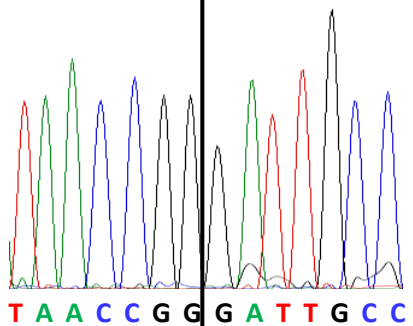
e38 e41



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G

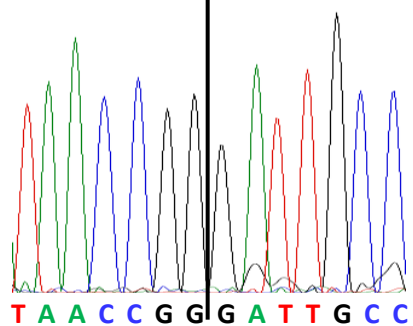
e38 e41



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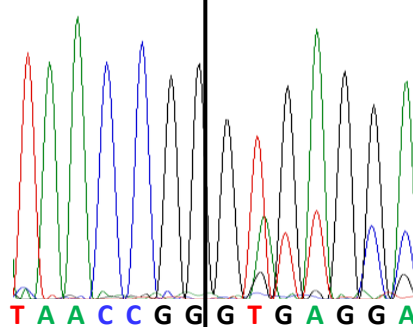
e38 e41



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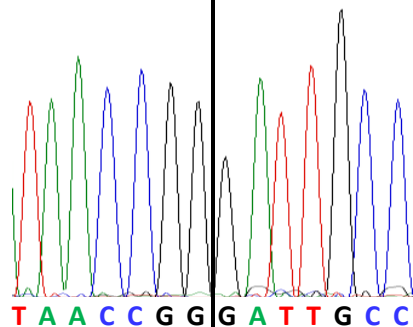
e38 e40



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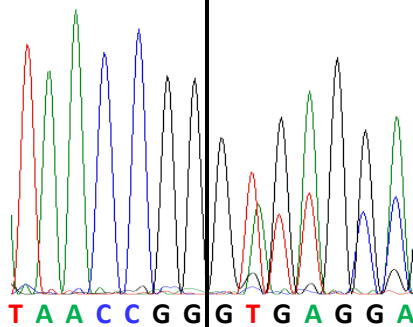
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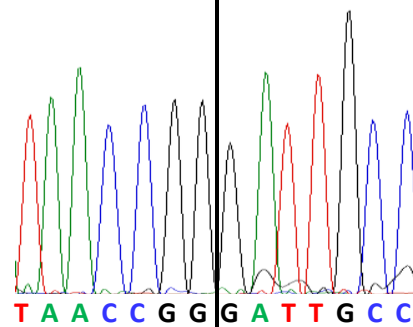
e38 e40



BA26_c.5584+6T>C_fr5

G

e38 e41

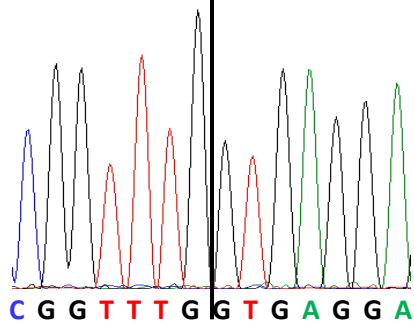


BA27

BA27_WT_fr1

G

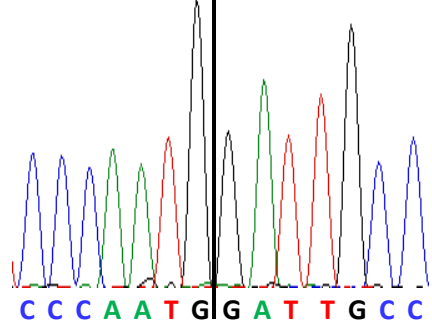
e39 e40



BA27_WT_fr1

G

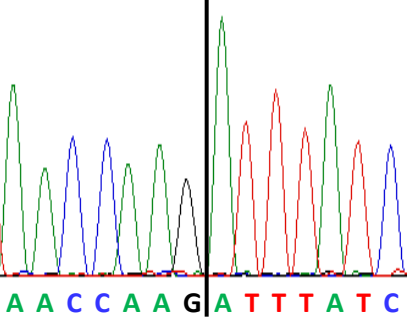
e40 e41



BA27_WT_fr1

G

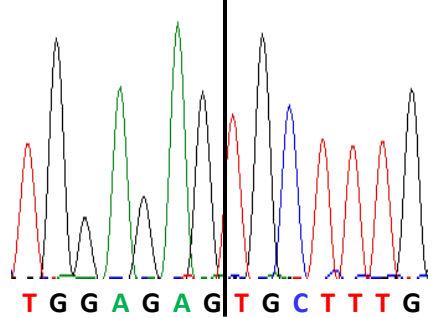
e41 e42



BA27_WT_fr1

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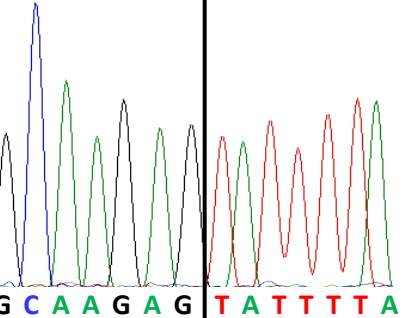
e42 e43



BA27_WT_fr1

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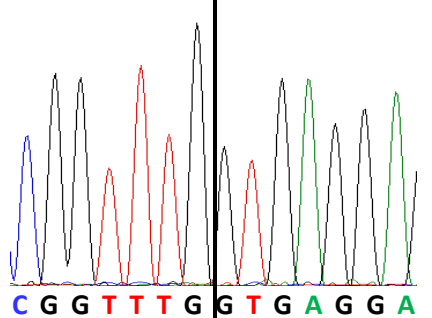
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G

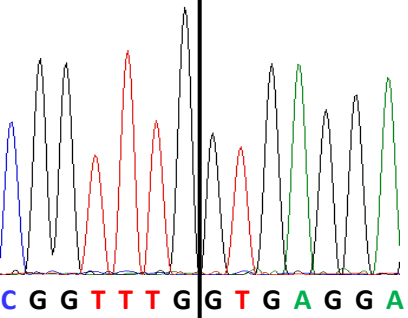
e39 e40



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P

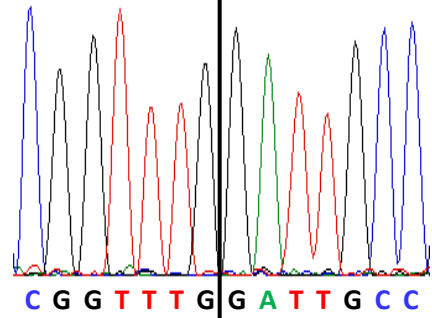
e39 e40



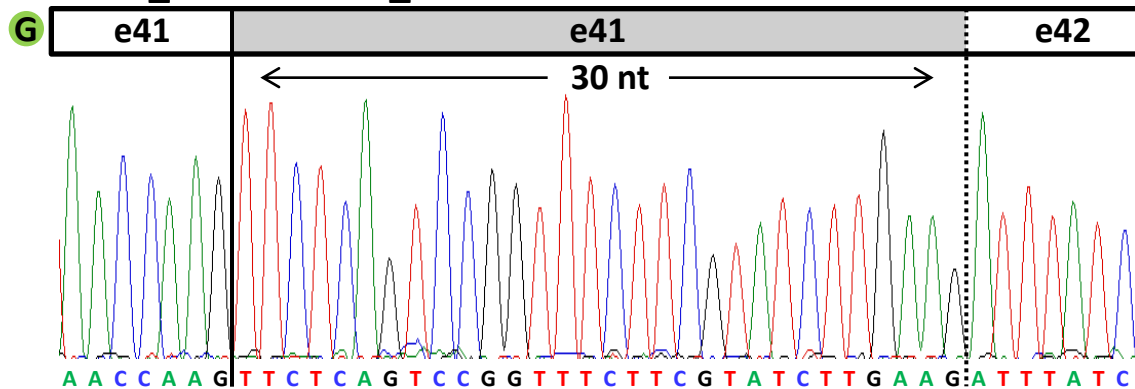
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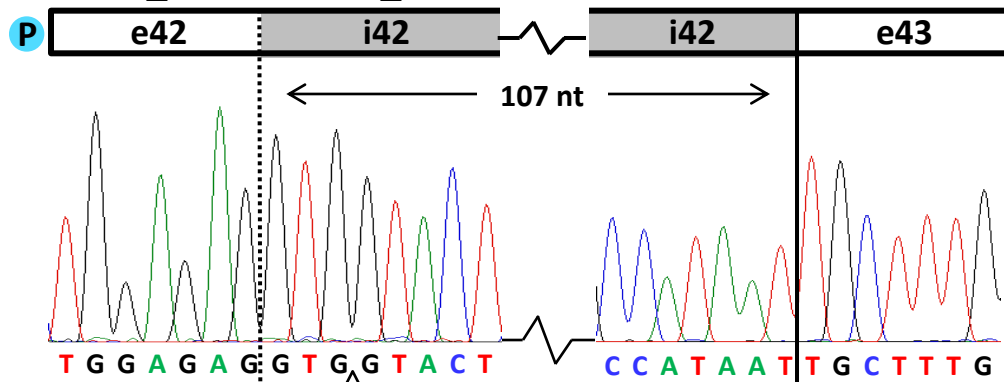
e39 e41



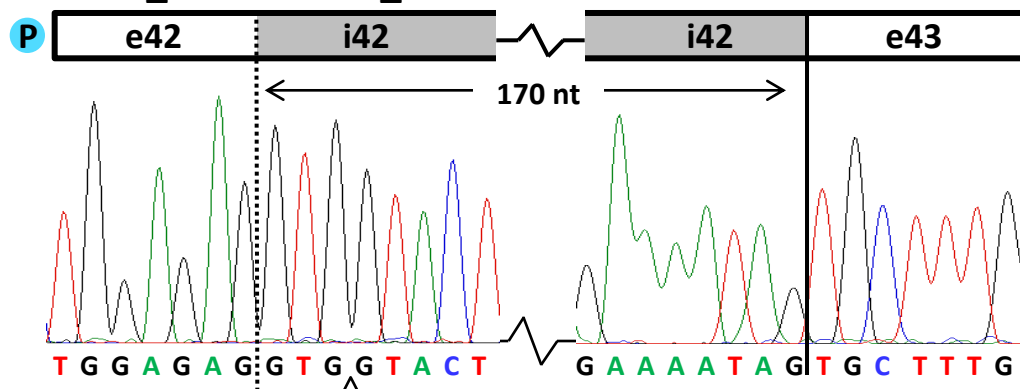
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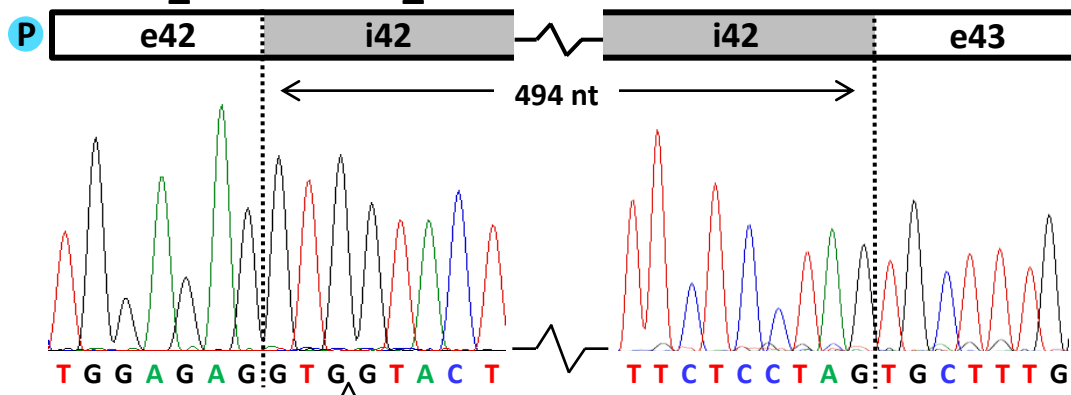
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BA27_c.5898+5del_fr5



BA27_c.5898+5del_fr3

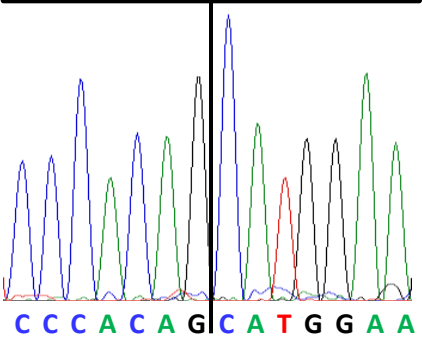


BA29

BA29_WT_fr1

G

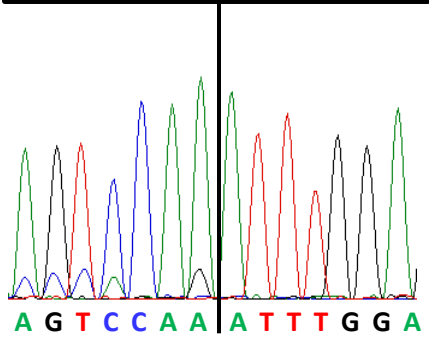
e46 e47



BA29_WT_fr1

G

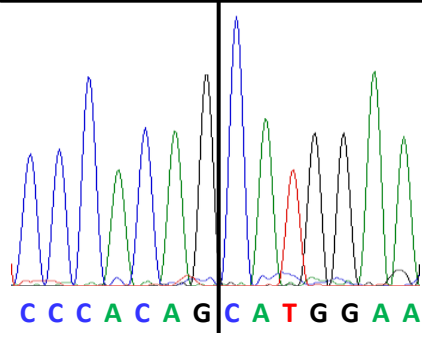
e47 e48



BA29_c.6478A>G_fr1

P

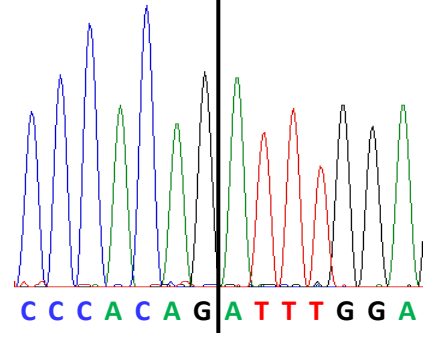
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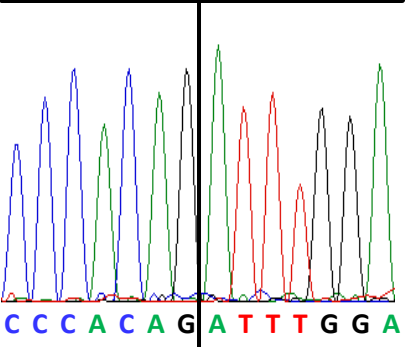
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G

e46 e48



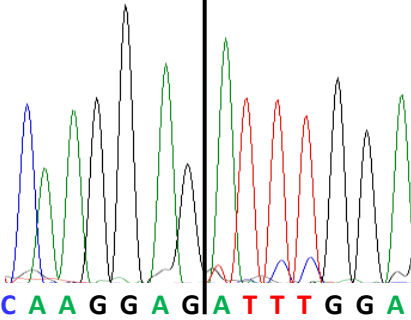
MG30

MG30_WT_fr1

G

RHO-e3

ABCA4-e48

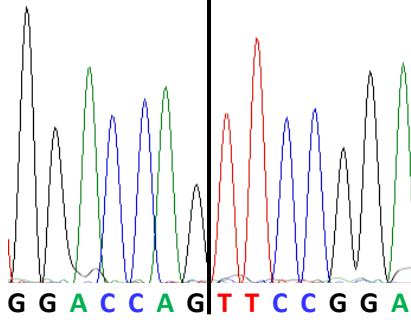


MG30_WT_fr1

G

ABCA4-e48

*RHO*e5



MG30_c.6729+5_19del_fr2

G

RHO-e3

RHO-e5

