

Deafblindness in French Canadians from Quebec: A predominant founder mutation in the *USH1C* gene provides the first genetic link with the Acadian population

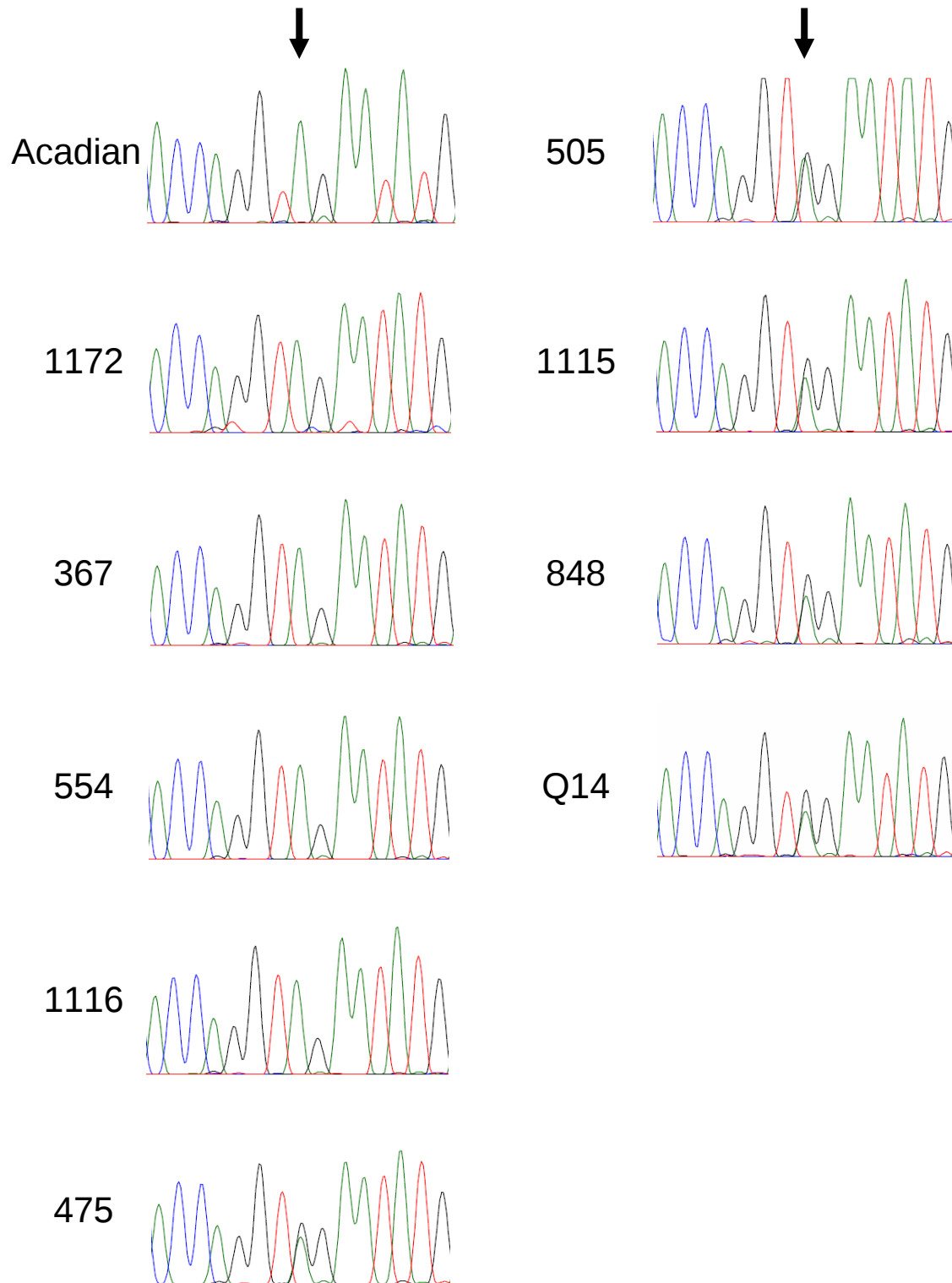
Ebermann et al.

RAW DATA

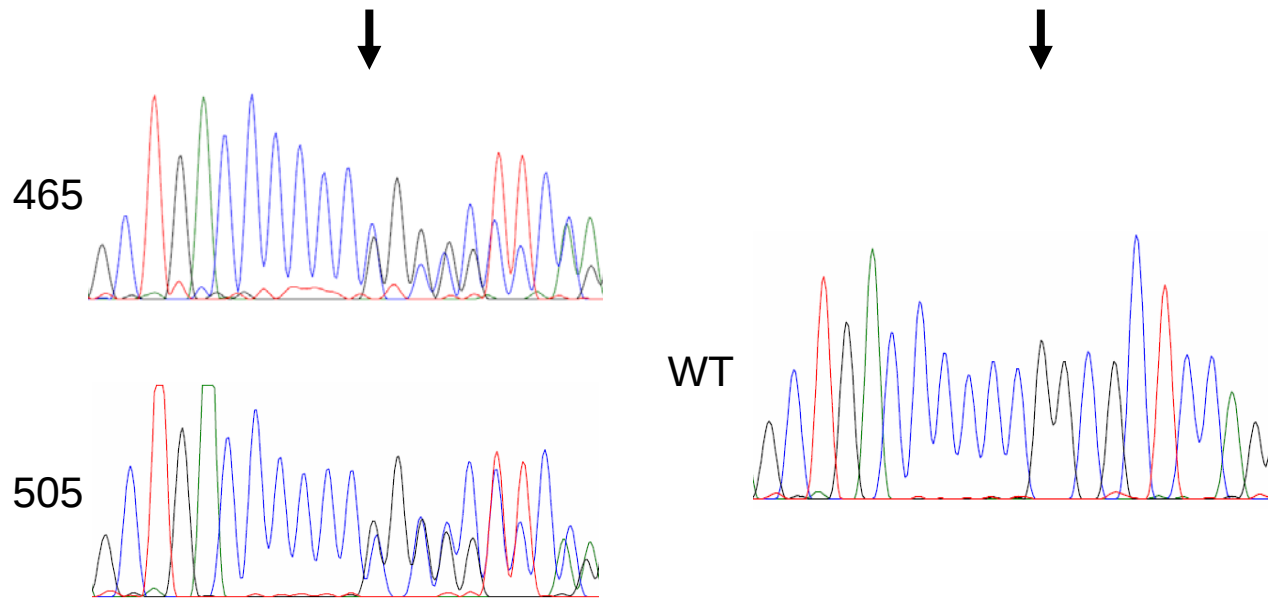
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USH1 gene mutations identified in this study

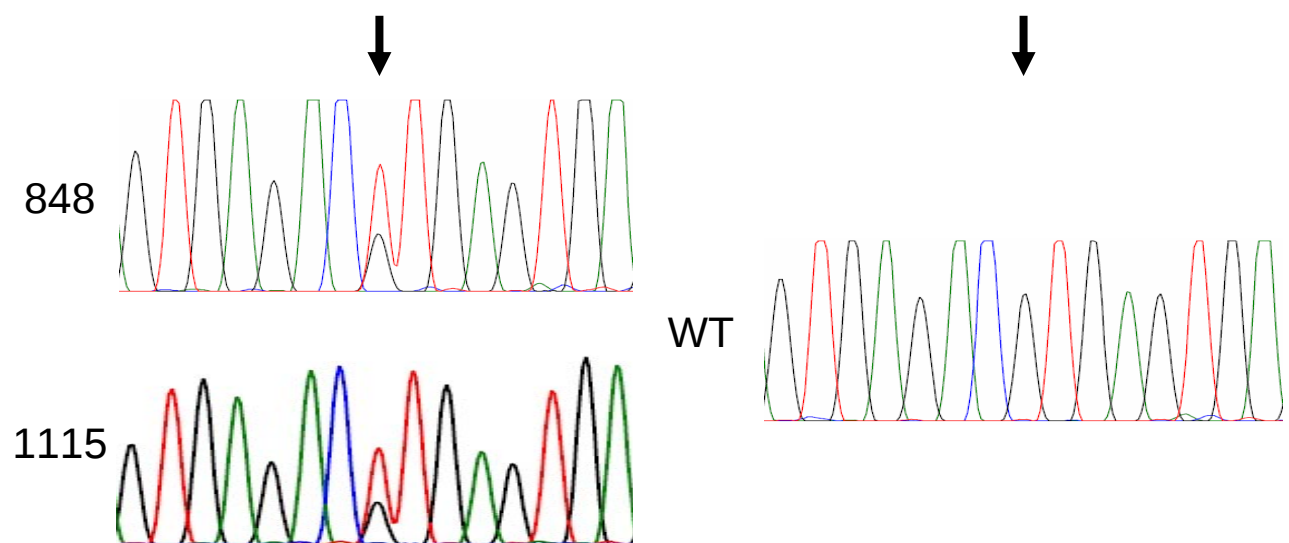
USH1C, c.216G>A



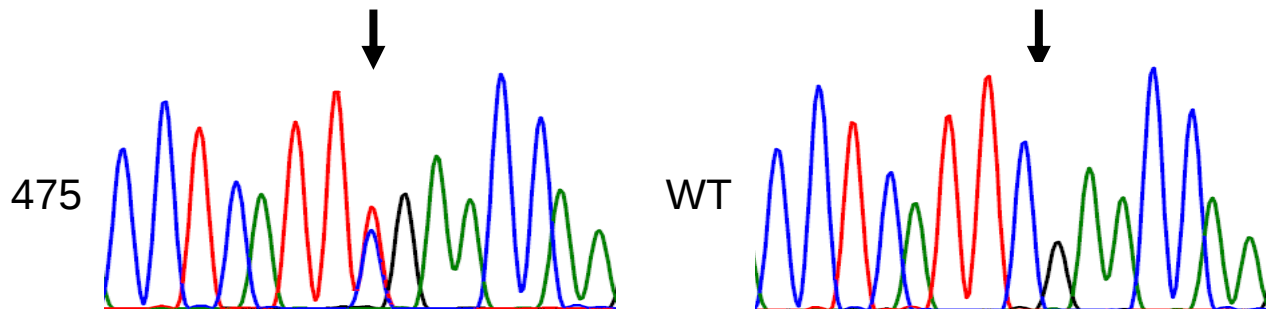
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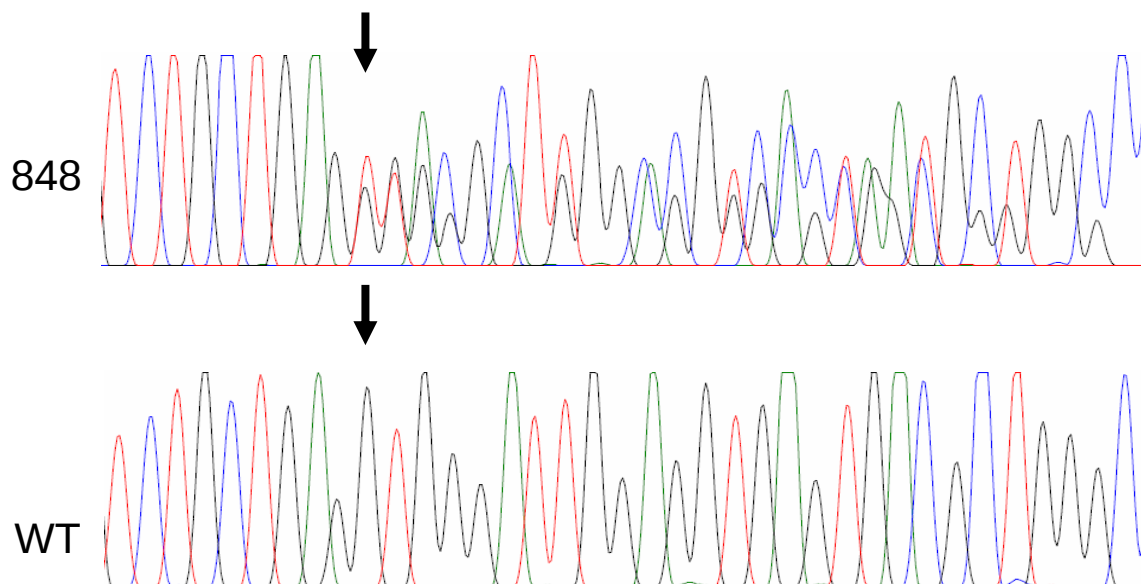
USH1C, c.496+1G>T



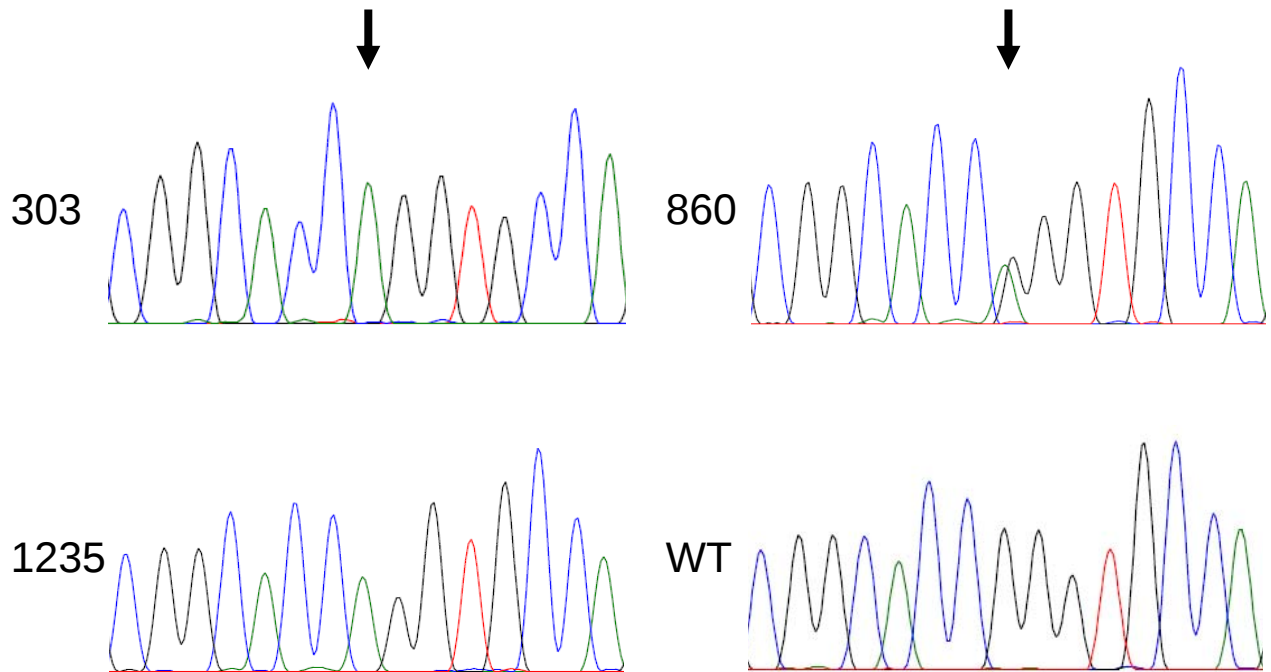
USH1C, c.463C>T (p.R155X)



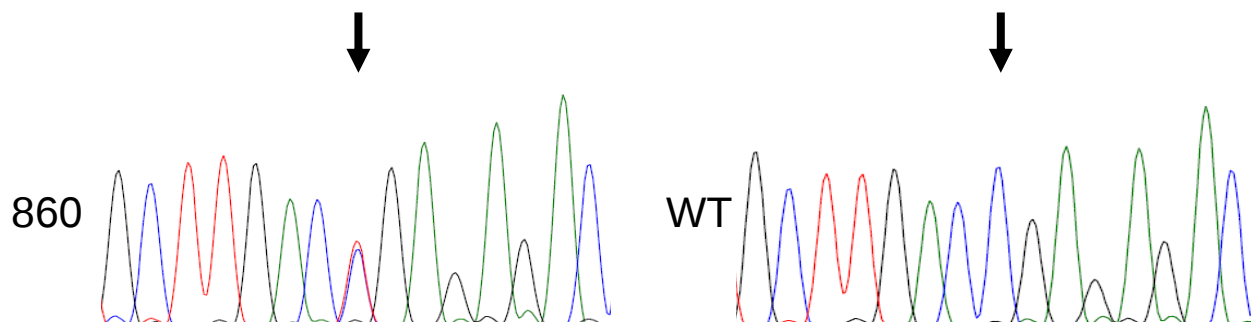
USH1C, c.748-759+5del



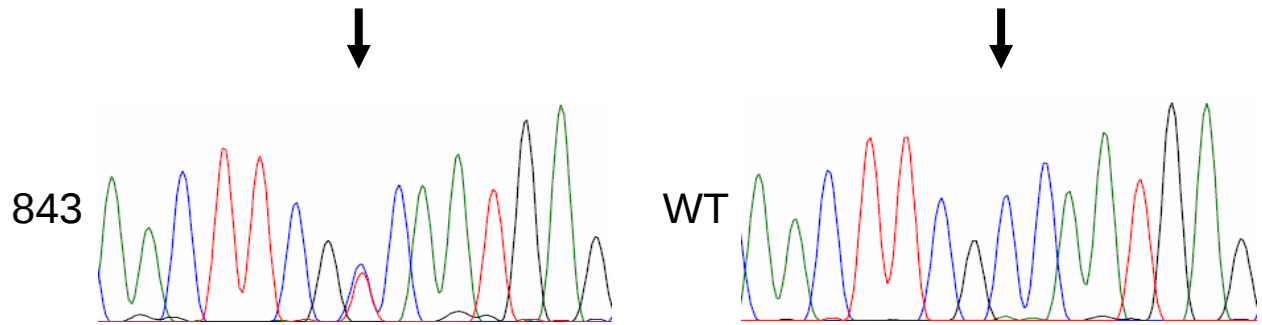
CDH23, IVS45-9G>A



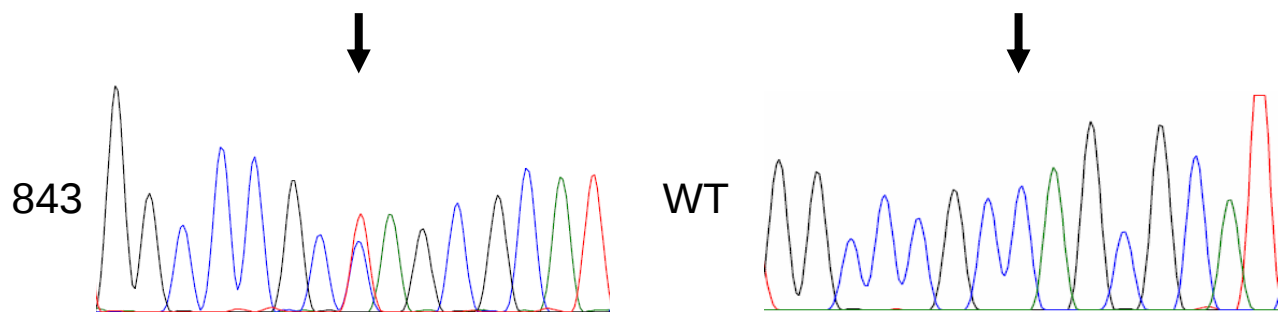
CDH23, c.2206C>T (p.R736X)



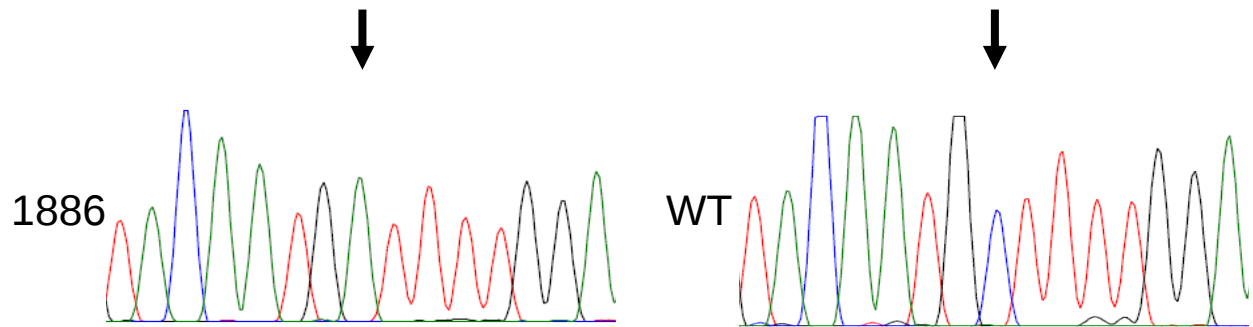
MYO7A, c.1370C>T (p.A457V)



MYO7A, c.2443C>T (p.Q815X)



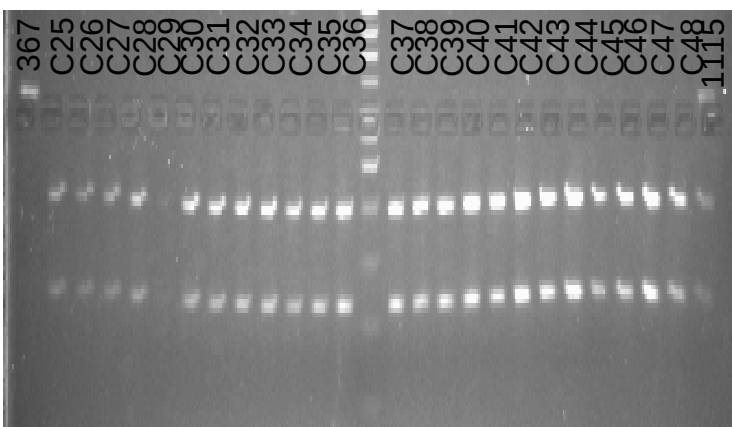
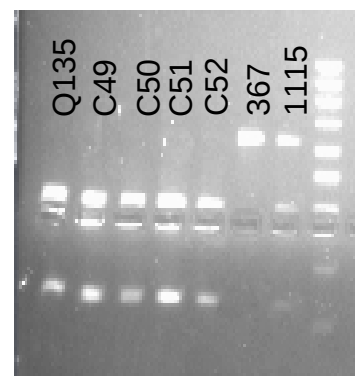
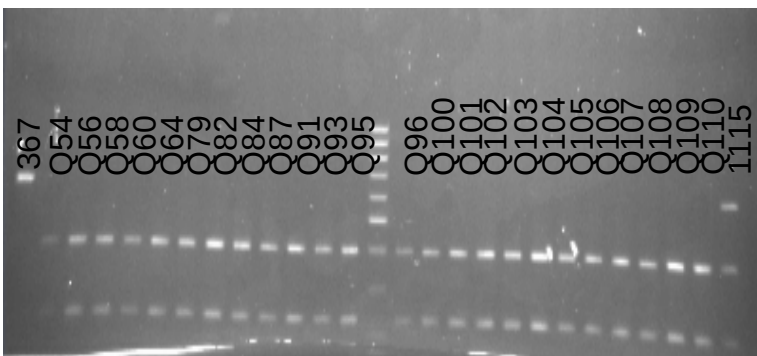
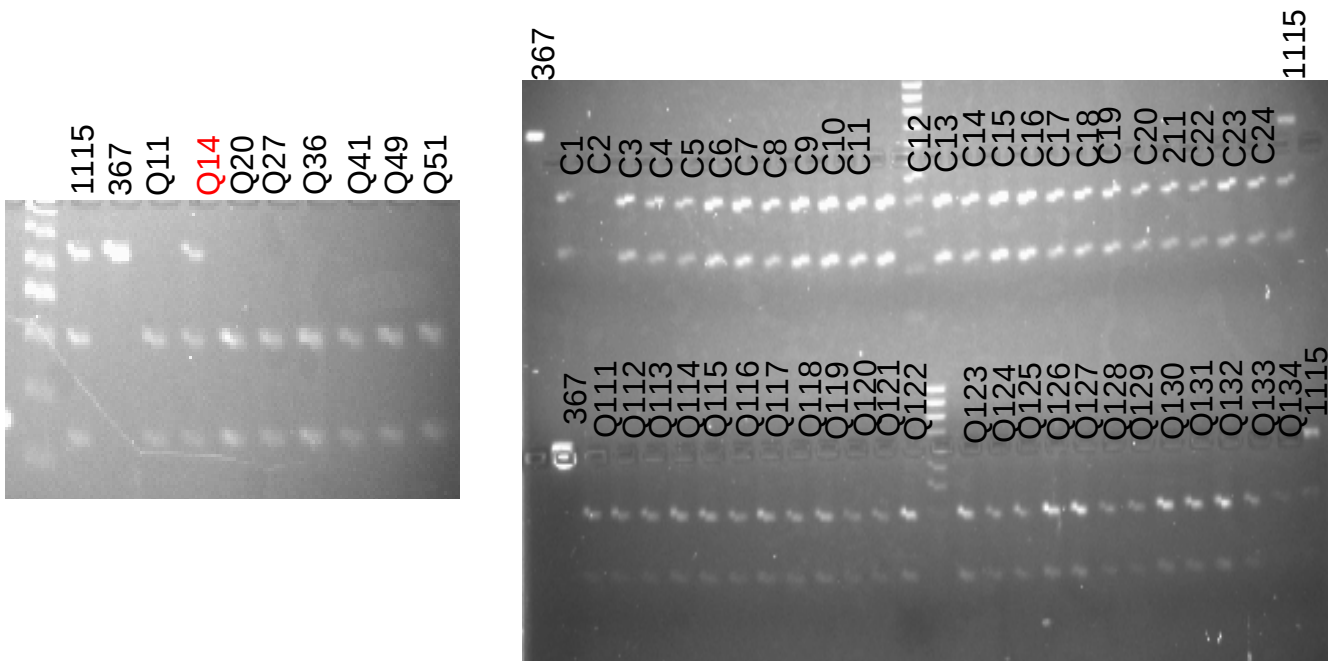
USH3A, c.368C>A (p.A123D)



c.216G>A (<i>USH1C</i>): Genotyping of 227 French Canadian healthy control individuals
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PCR products (625 bp) were digested with *Dra*III. One *Dra*III site is present in the wildtype, resulting in fragments of 230 and 395 bp. Presence of c.216G>A results in loss of the *Dra*III site.

Controls for c.216G>A (*USH1C*, exon 3) by restriction digest with *Dra*III



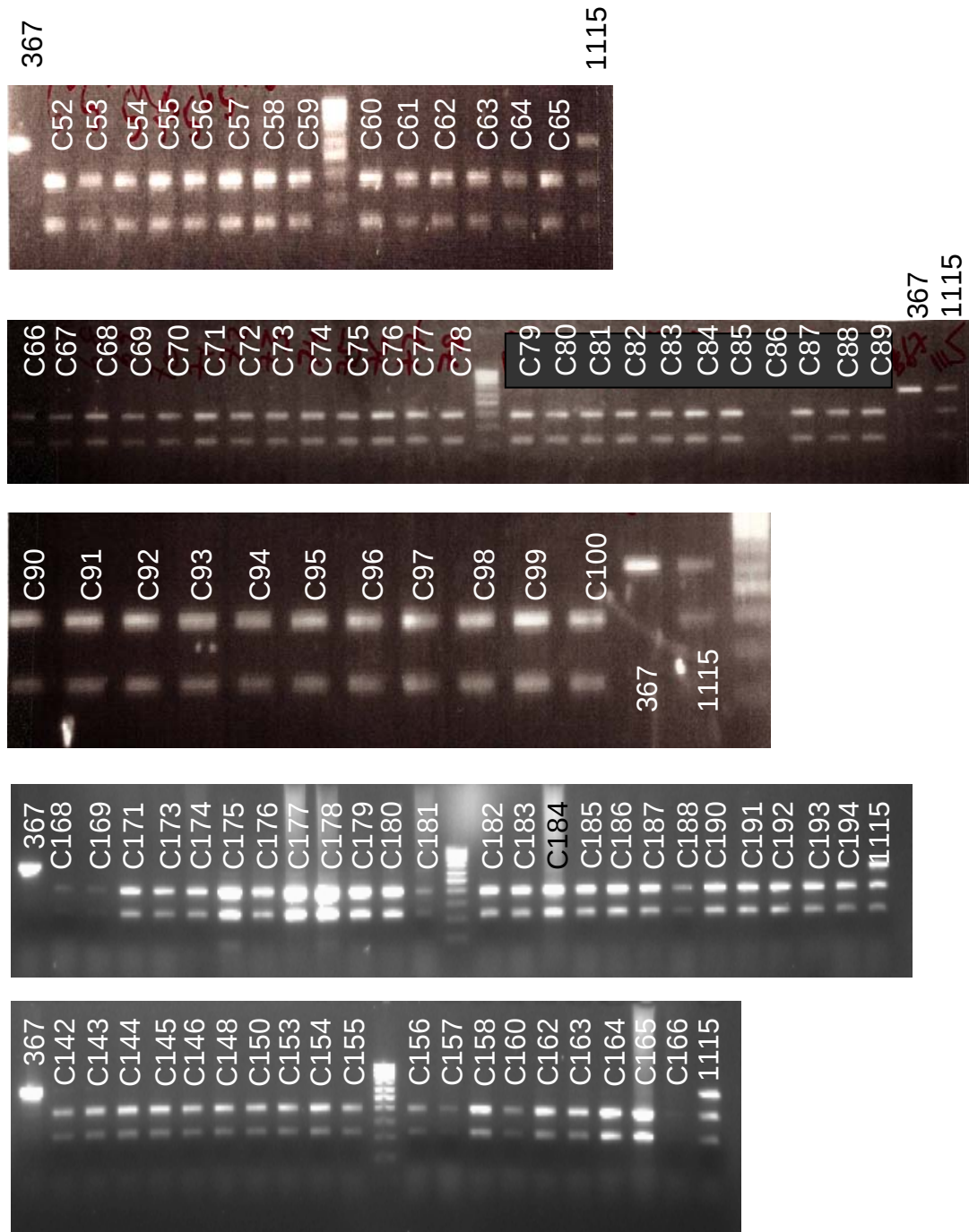
PCR product: 625 bp

367 = c.216G>A (homozygous)
1115 = c.216G>A (heterozygous)
Q14 = healthy carrier

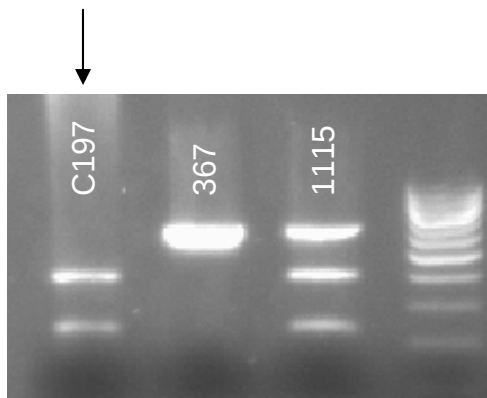
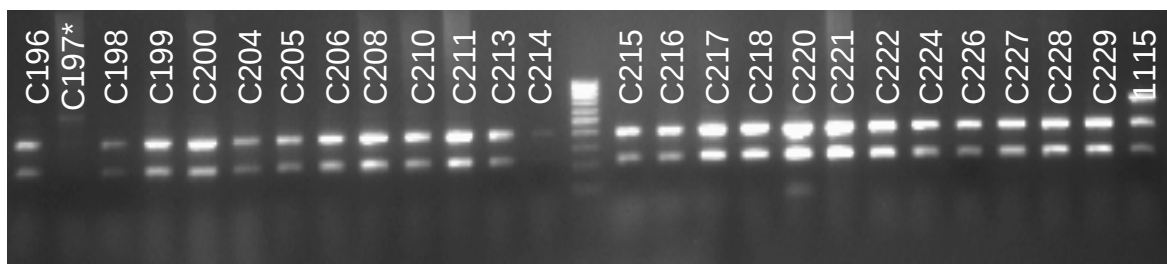
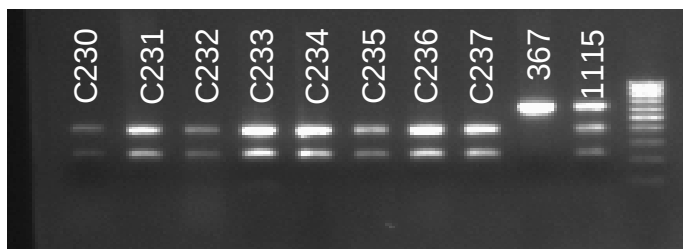


WT = 1x *Dra*III restriction site
230+395 bp
Mutant = 625 bp

Controls for c.216G>A (USH1C, exon 3)
by restriction digest with *Dra*III



Controls for c.216G>A (USH1C, exon 3)
by restriction digest with *Dra*III



*digestion of control C197 was repeated and shown to be homozygous for c.216G (wildtype)

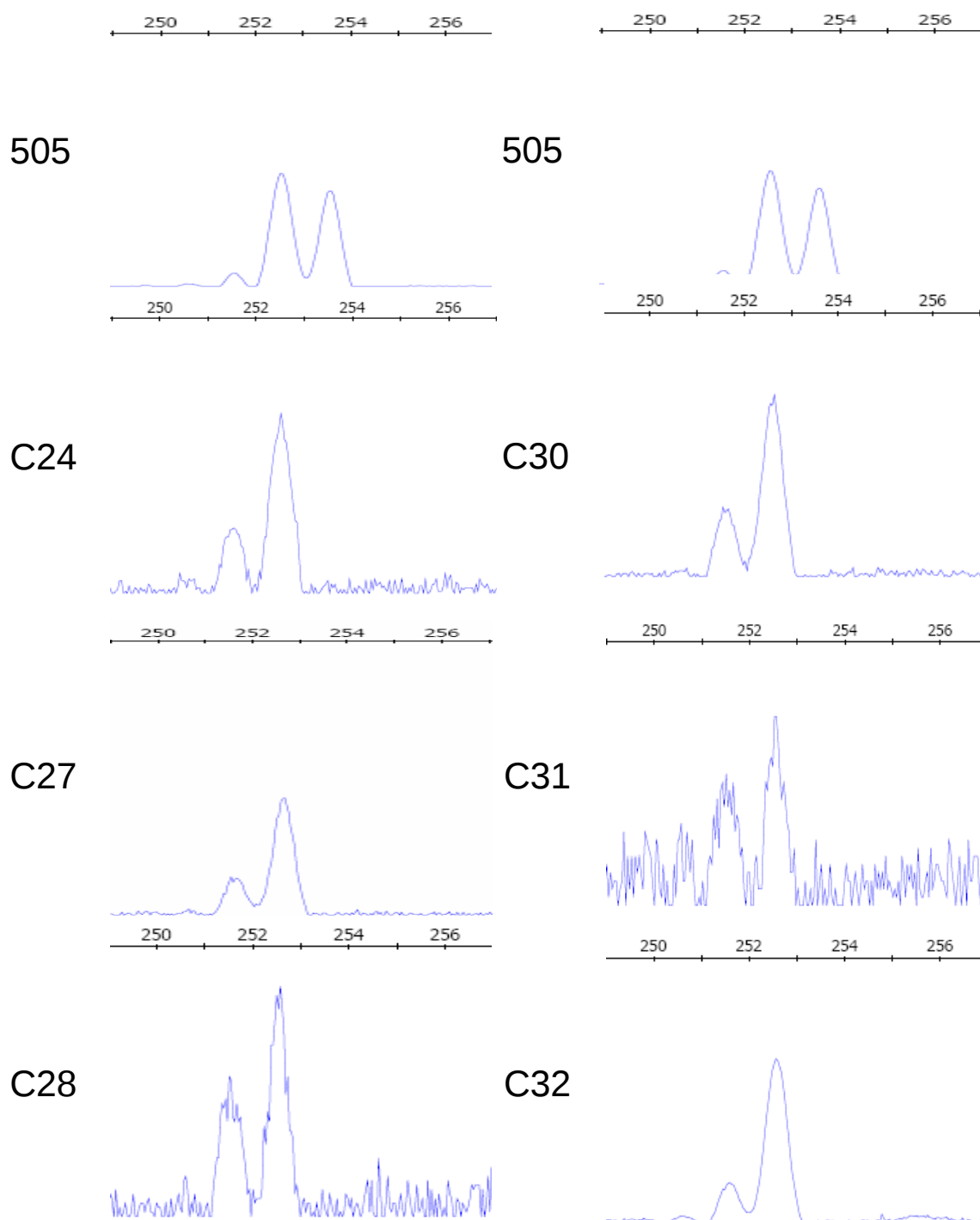
Σ227 controls

(Q2, Q133, Q134, C29, C66, C67, C86, C166 and C214 not counted (weak signal))

**c.238-239insC (*USH1C*):
Genotyping of 100 French Canadian healthy control individuals**

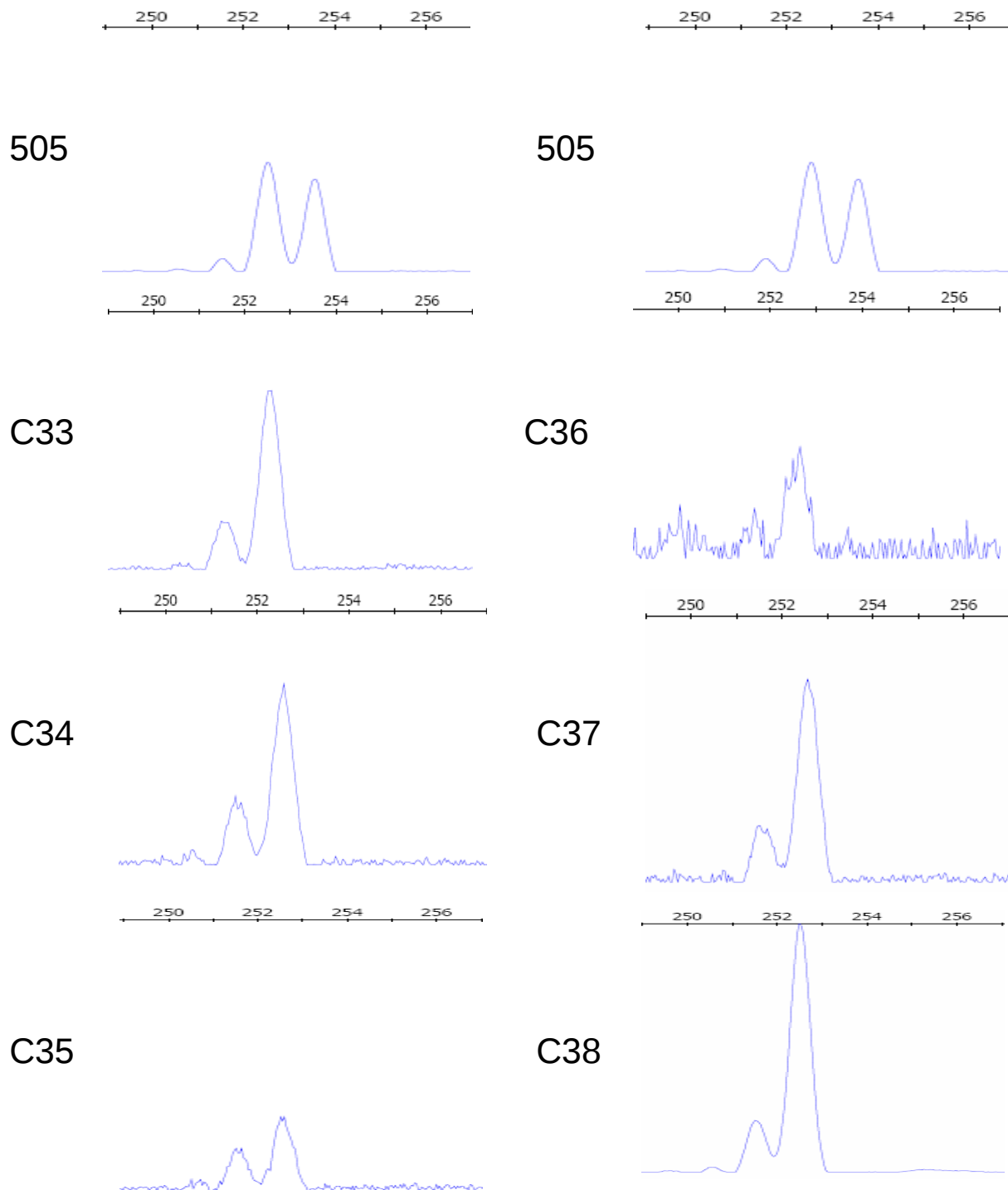
- fragment length analysis of exon 3 PCR products (GeneScan) -

USH1C, controls for c.238-239insC by GeneScan

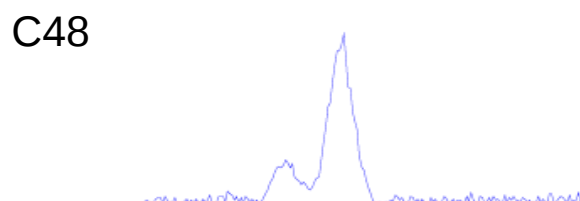
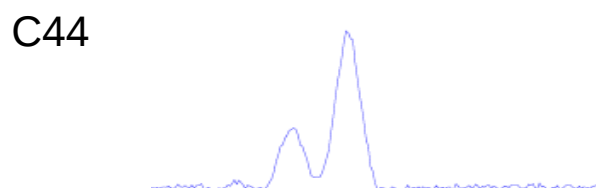
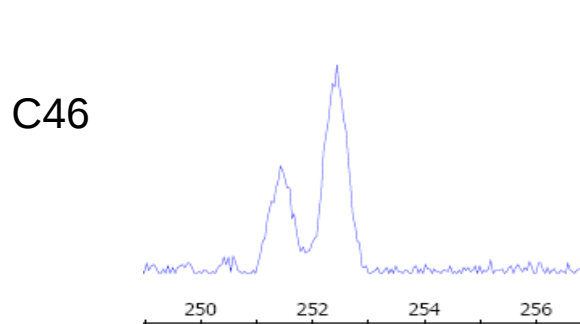
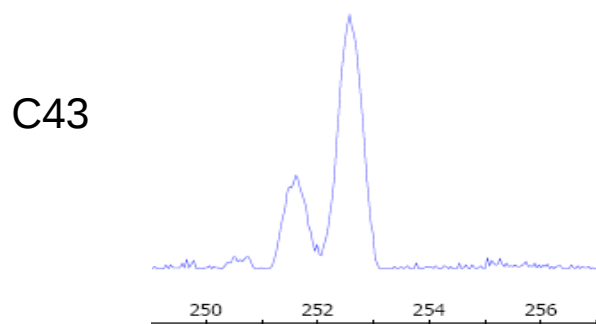
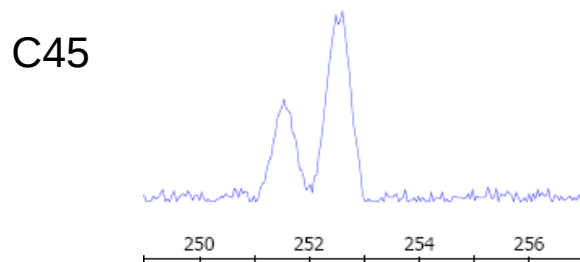
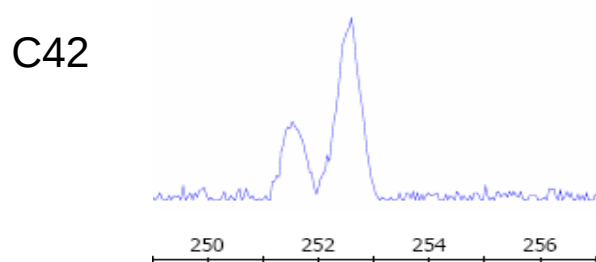
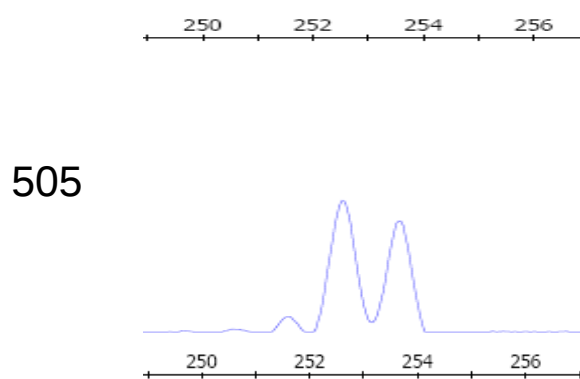
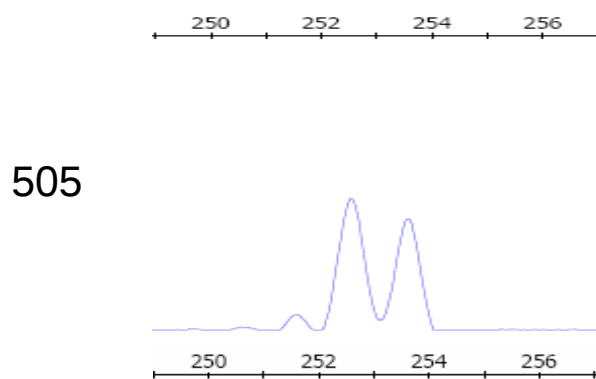


505: patient heterozygous for c.238-239insC (> two peaks)

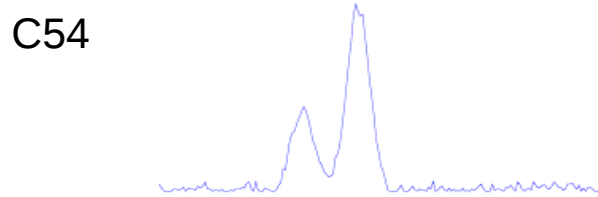
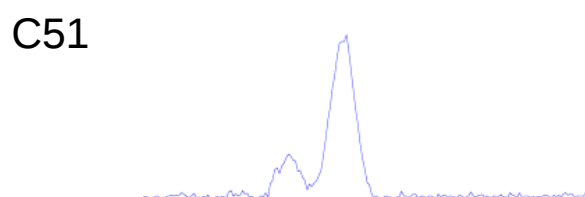
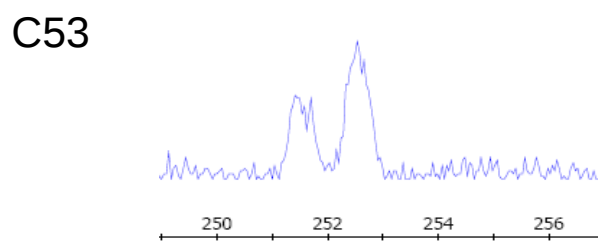
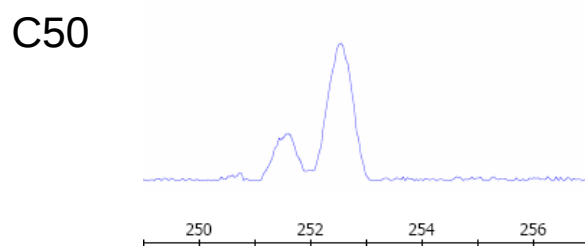
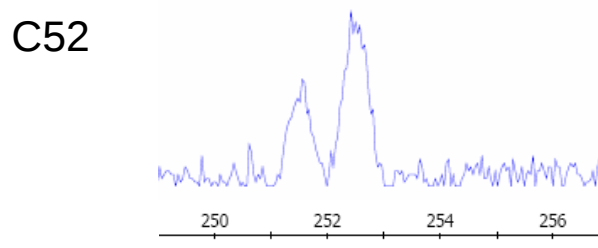
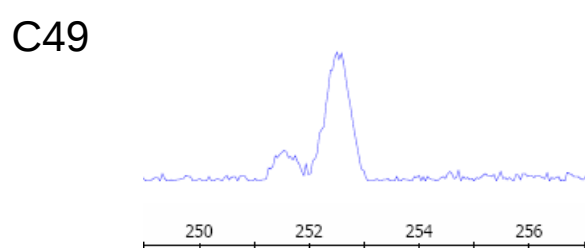
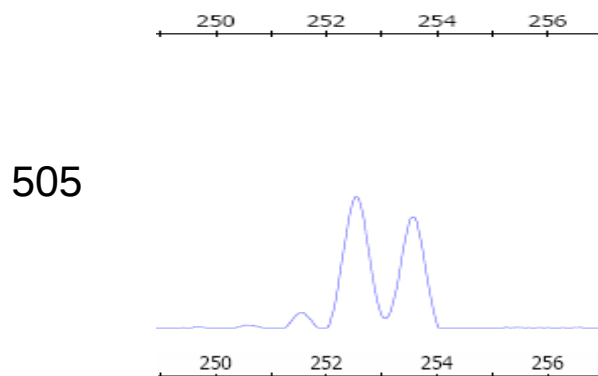
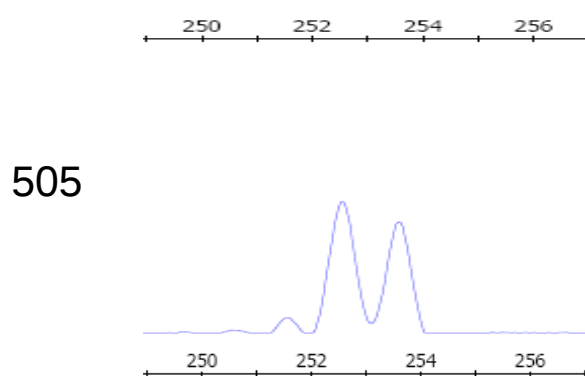
USH1C, controls for c.238-239insC by GeneScan



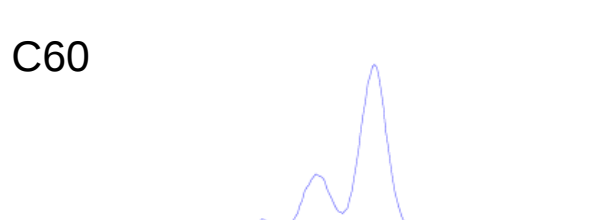
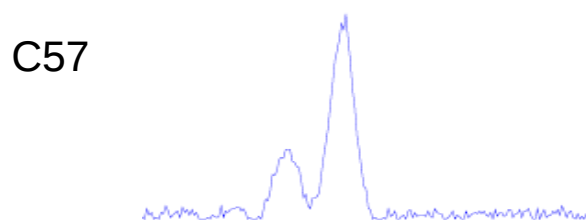
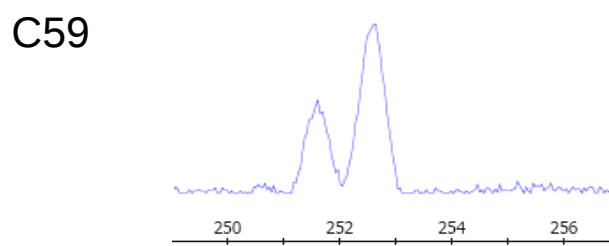
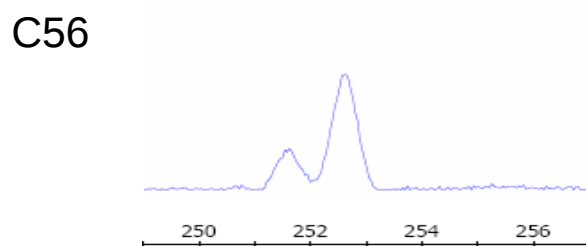
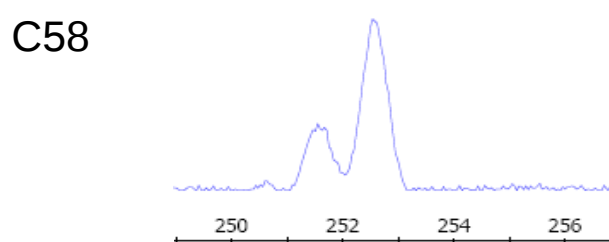
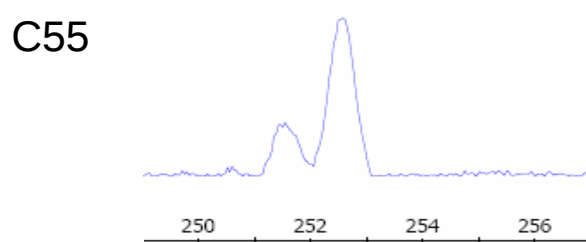
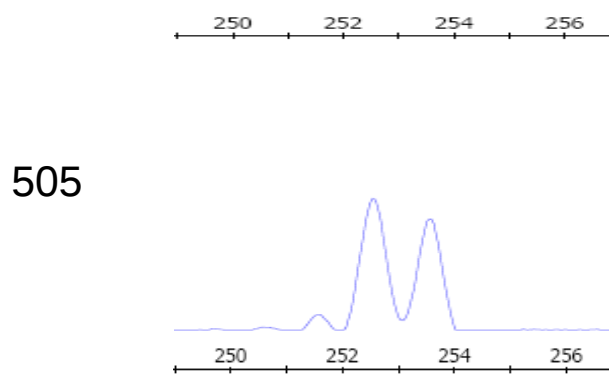
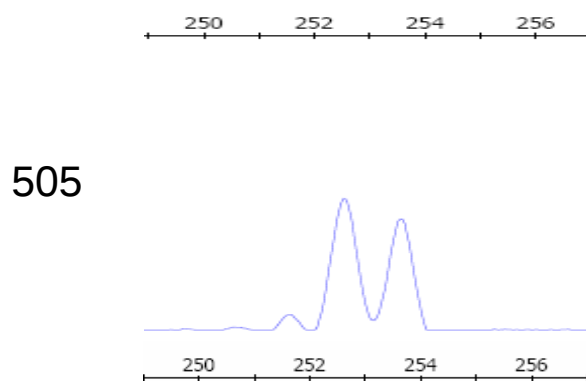
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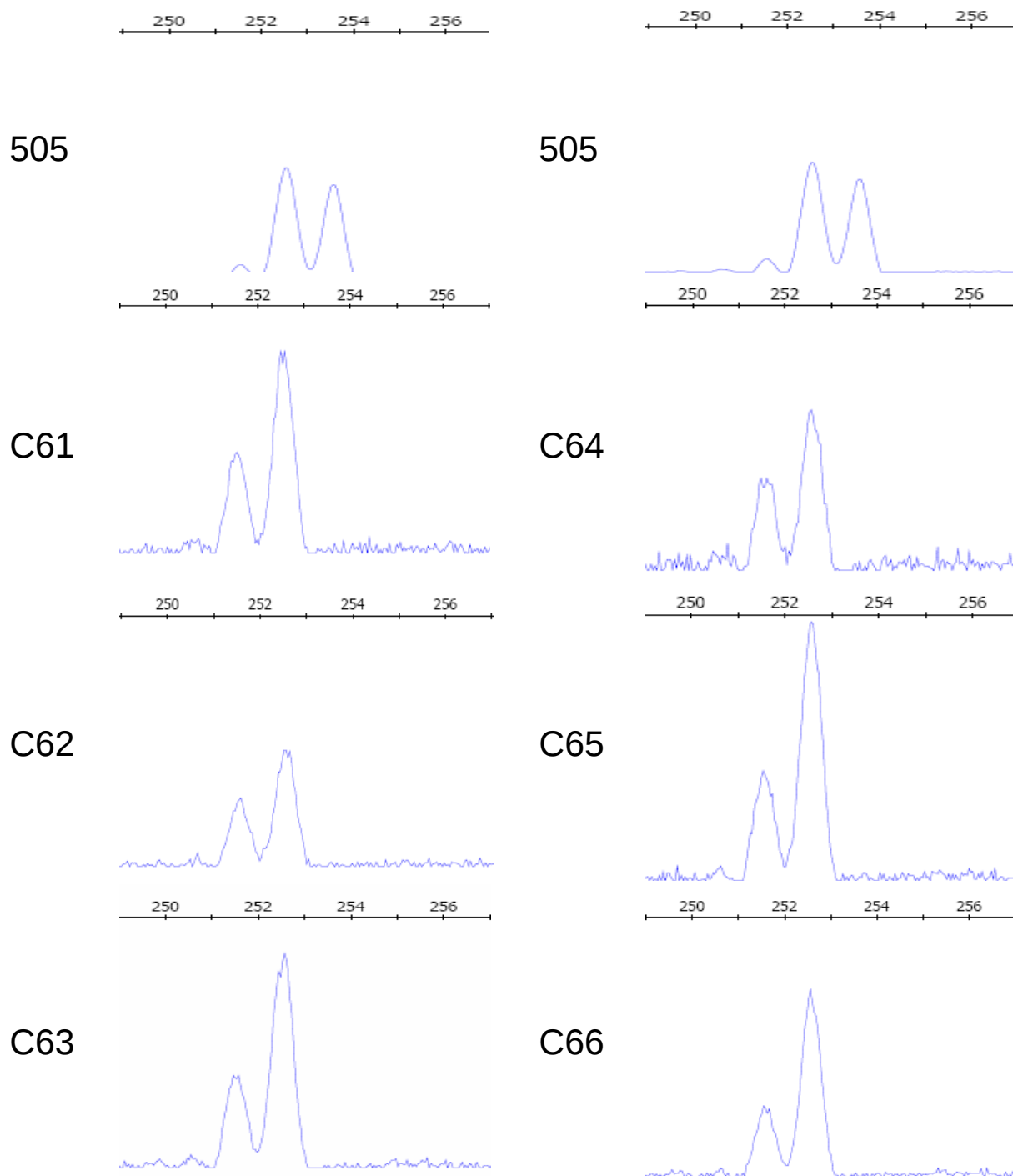
USH1C, controls for c.238-239insC by GeneScan



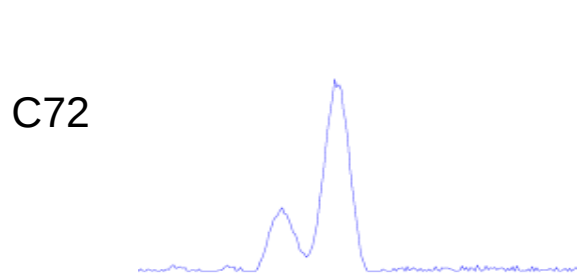
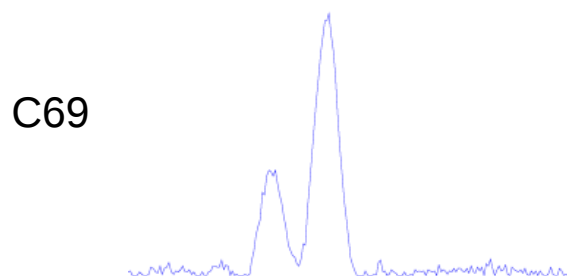
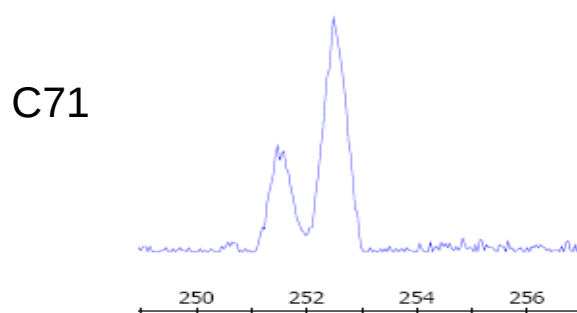
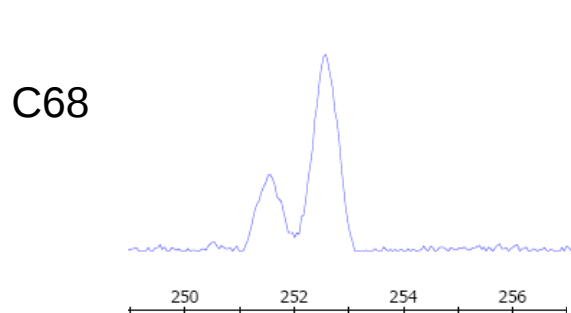
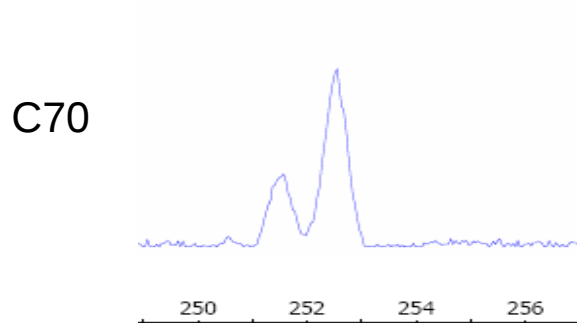
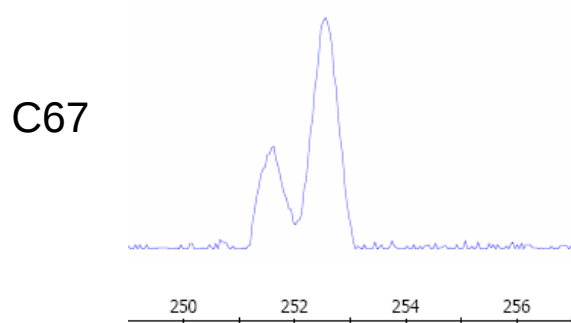
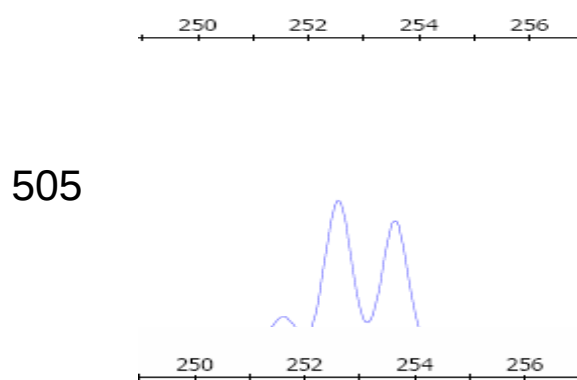
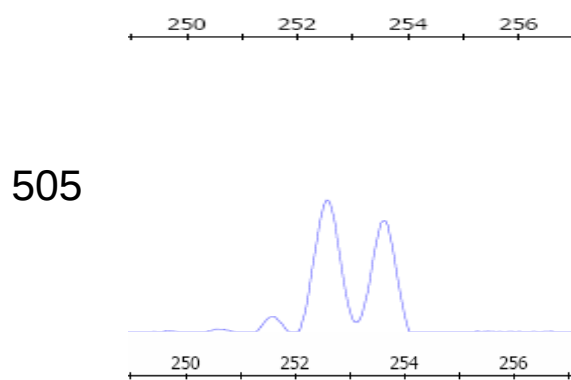
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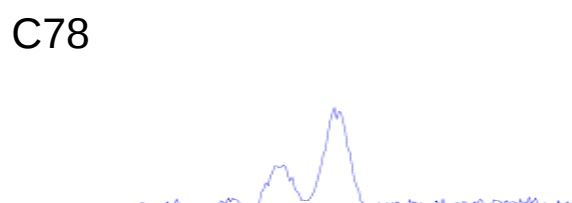
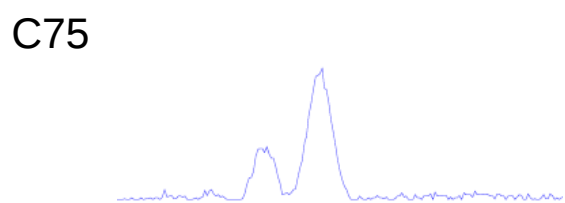
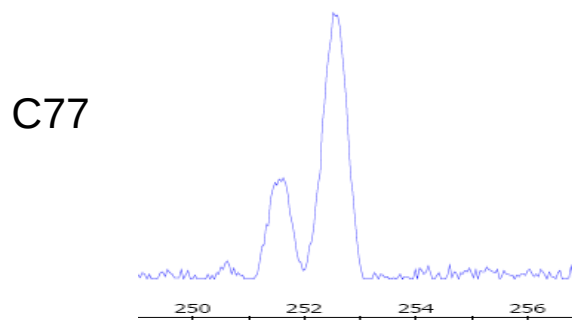
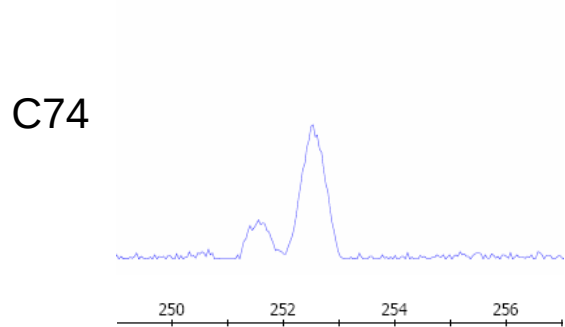
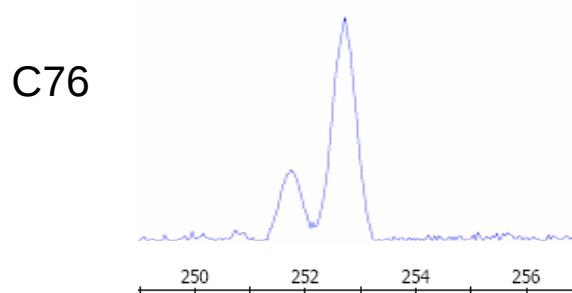
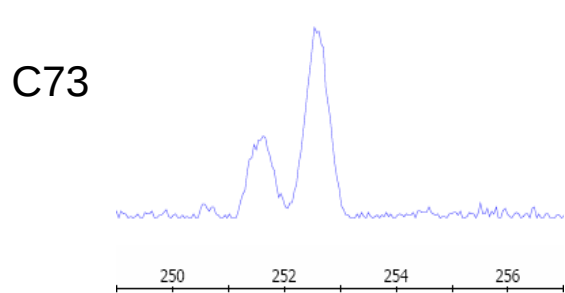
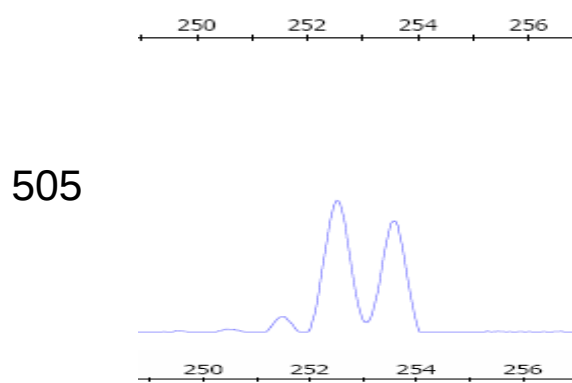
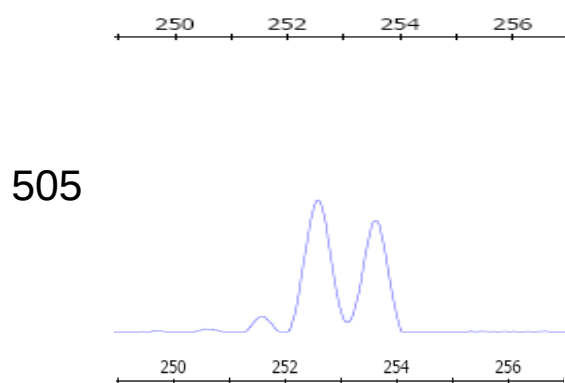
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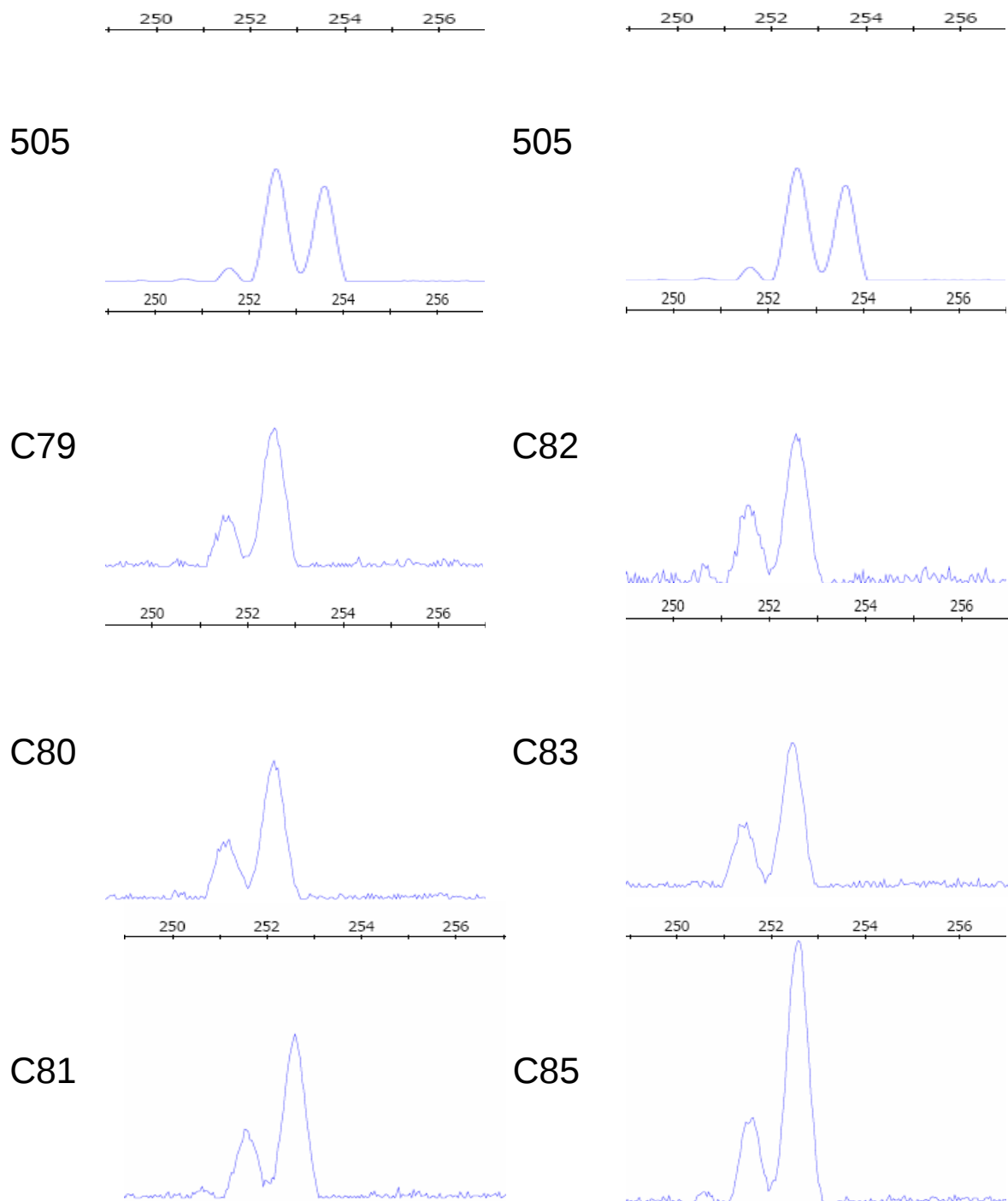
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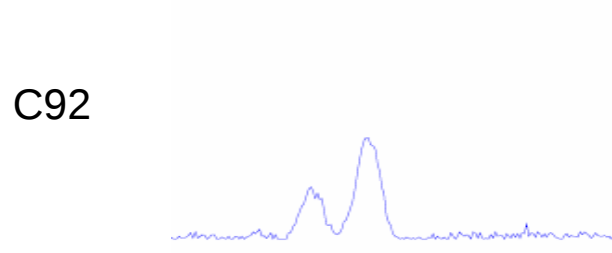
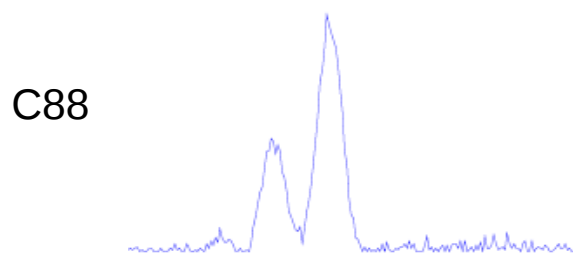
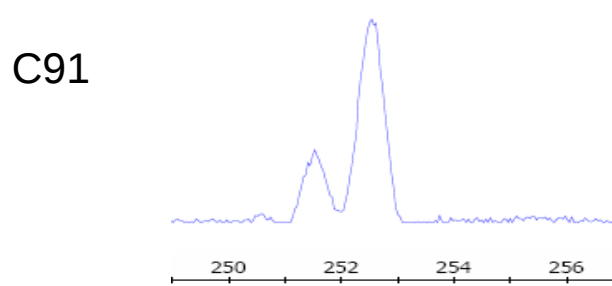
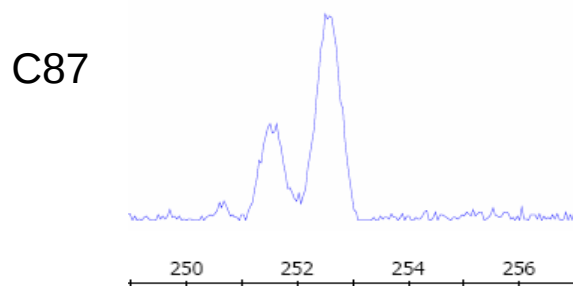
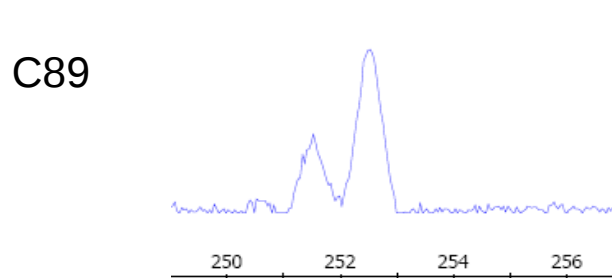
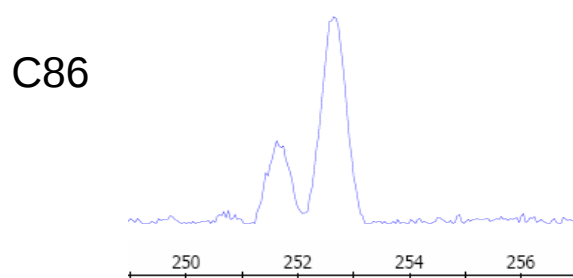
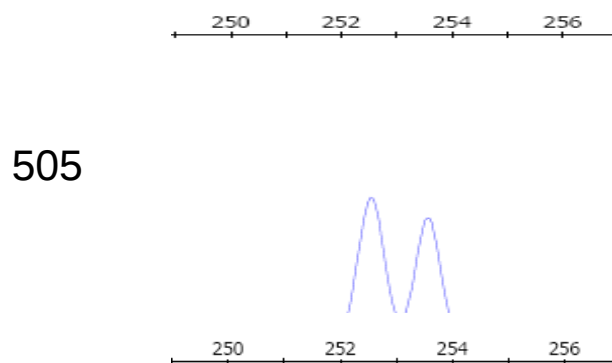
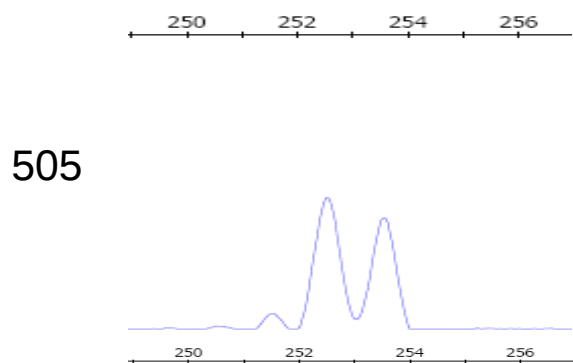
USH1C, controls for c.238-239insC by GeneScan



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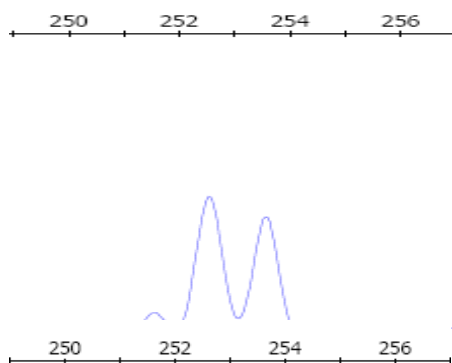


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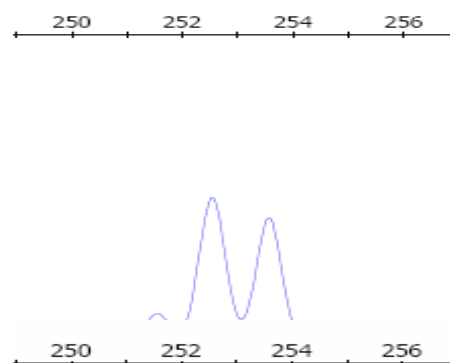


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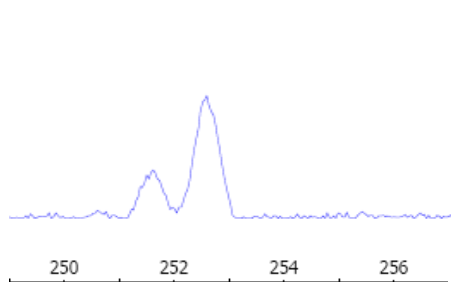
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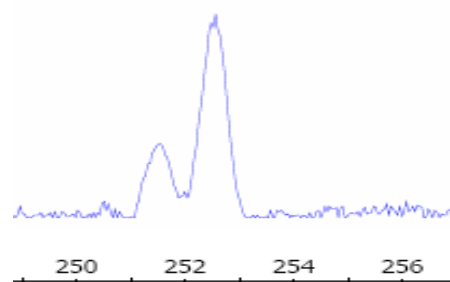
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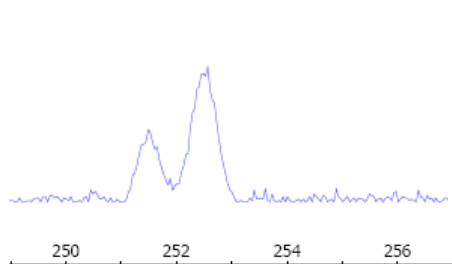
C93



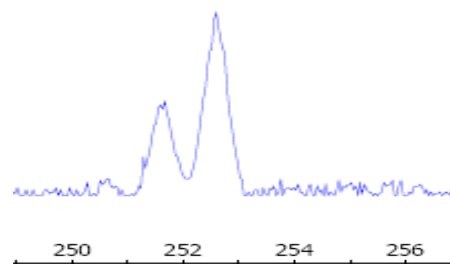
C96



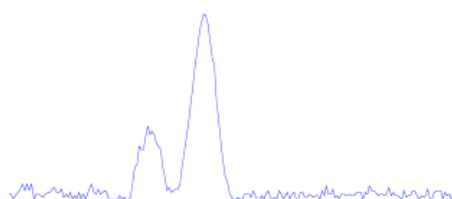
C94



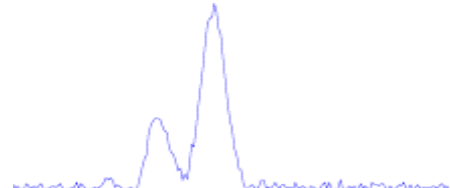
C97



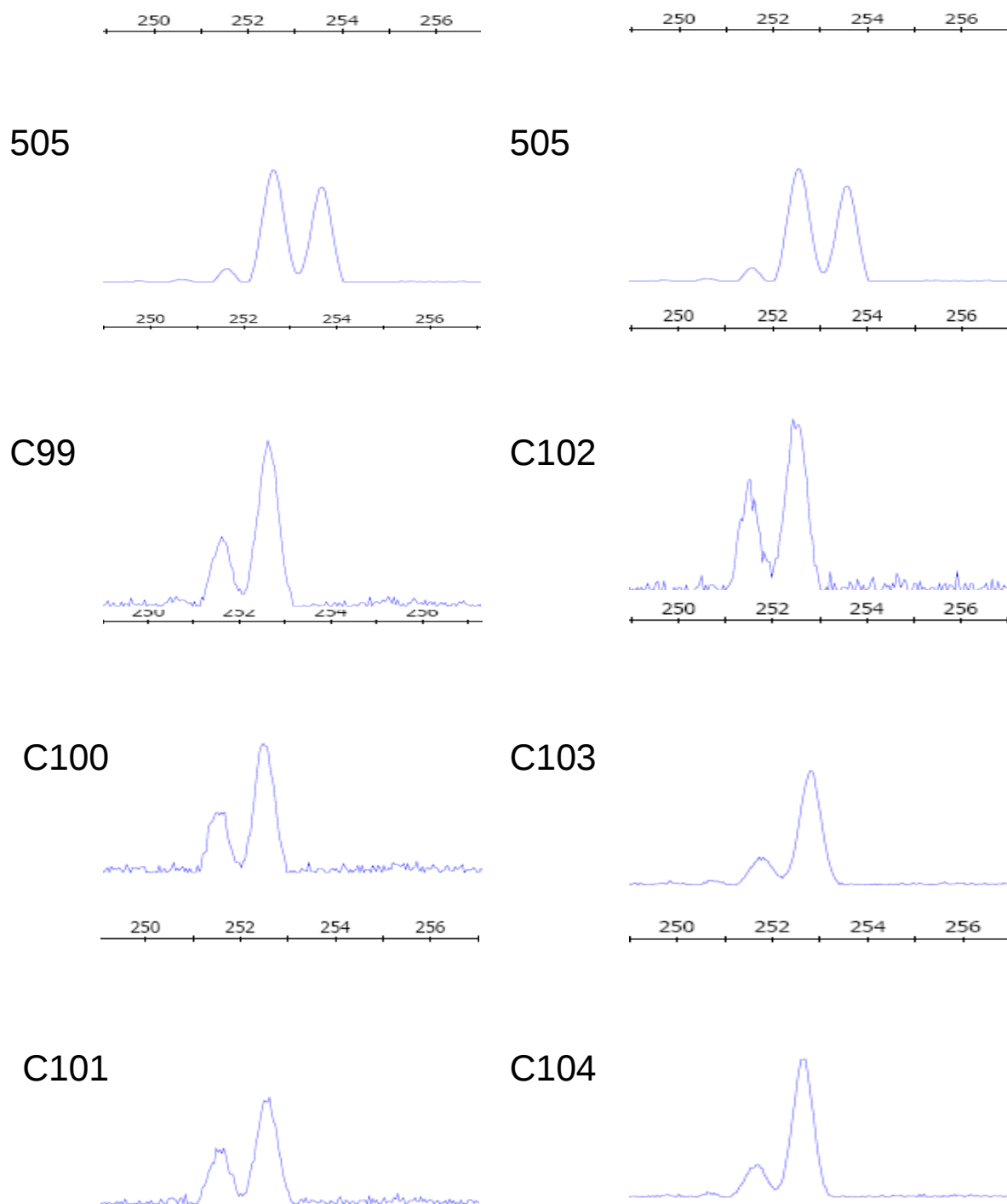
C95



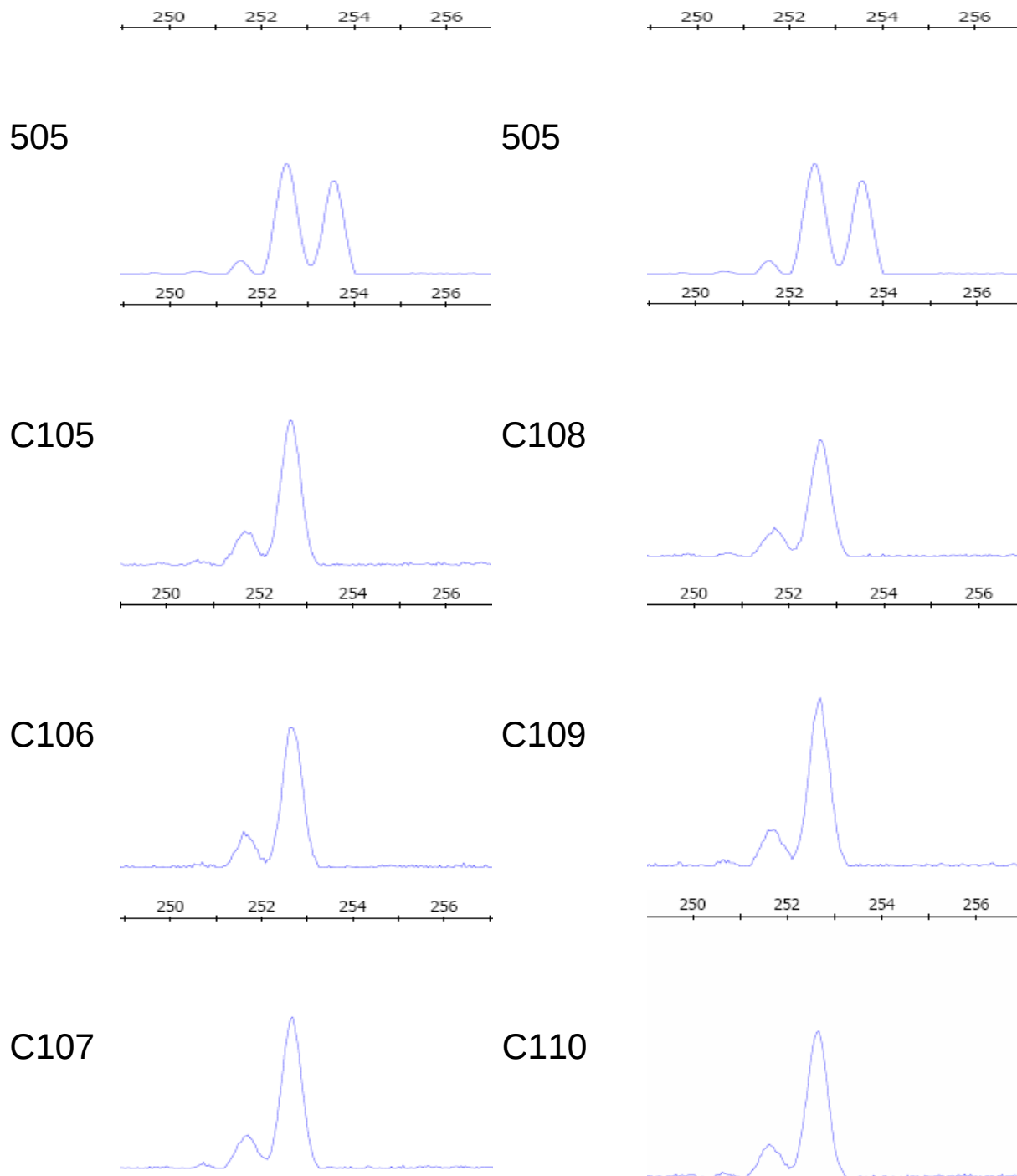
C98



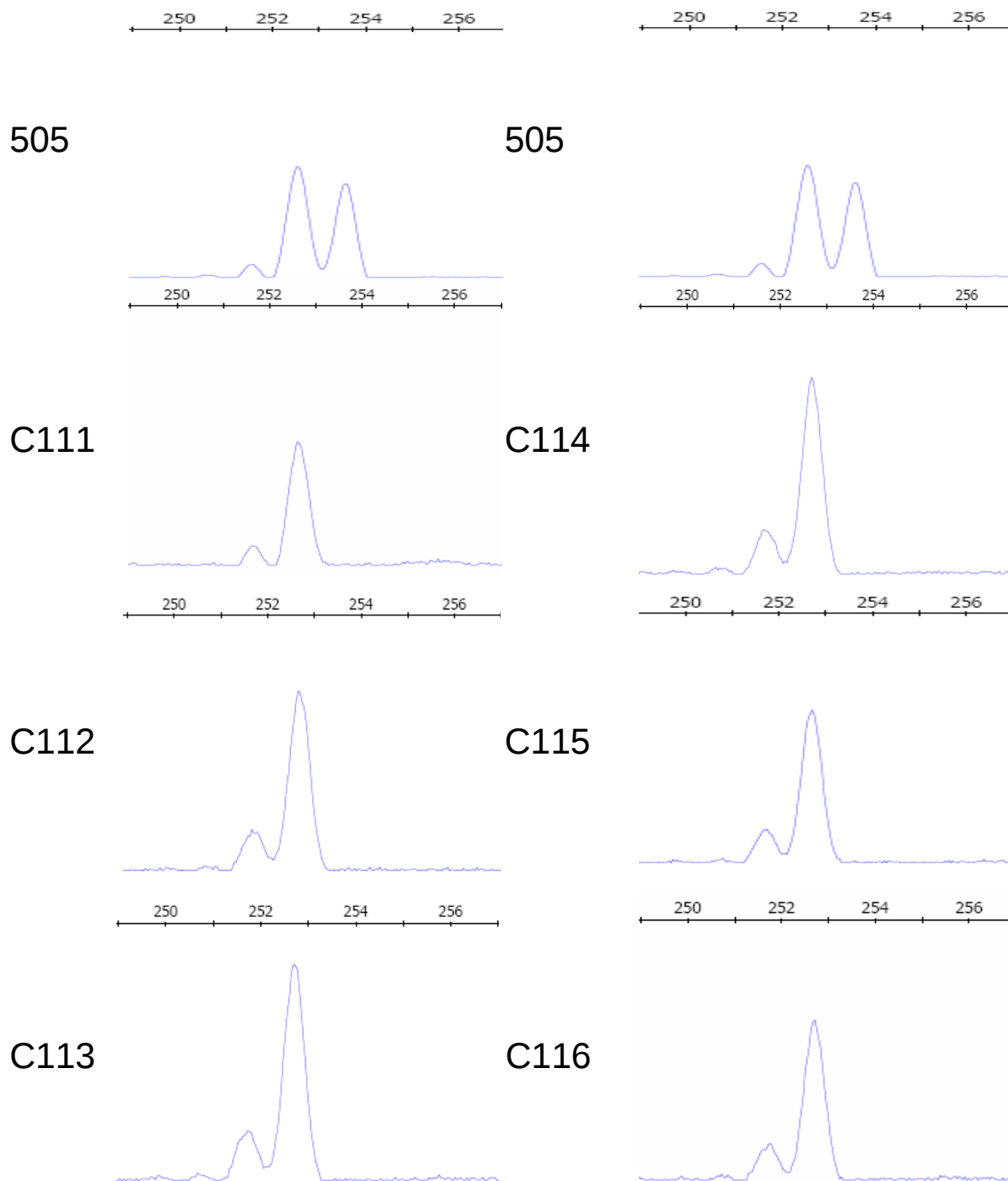
USH1C, controls for c.238-239insC by GeneScan



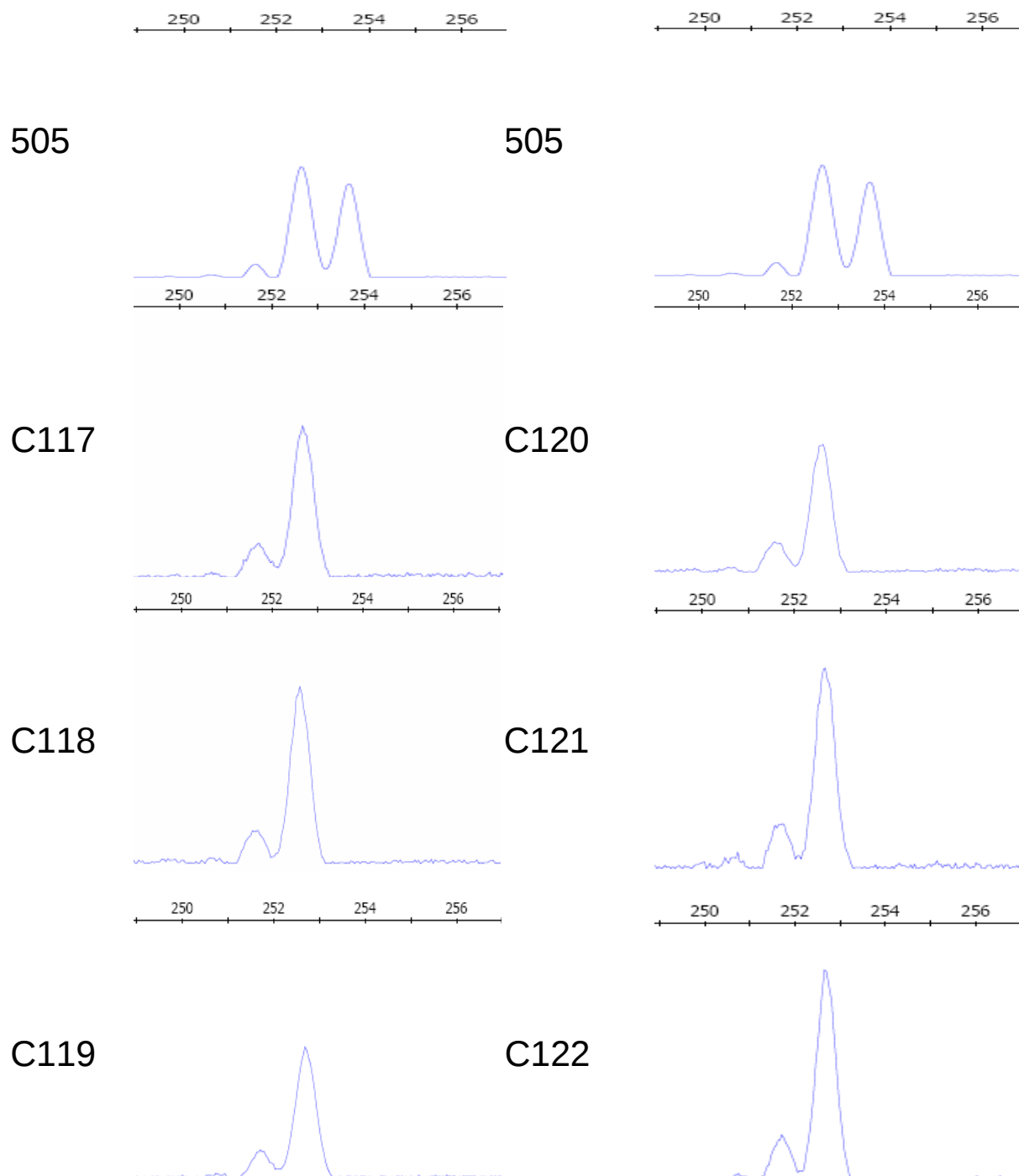
USH1C, controls for c.238-239insC by GeneScan



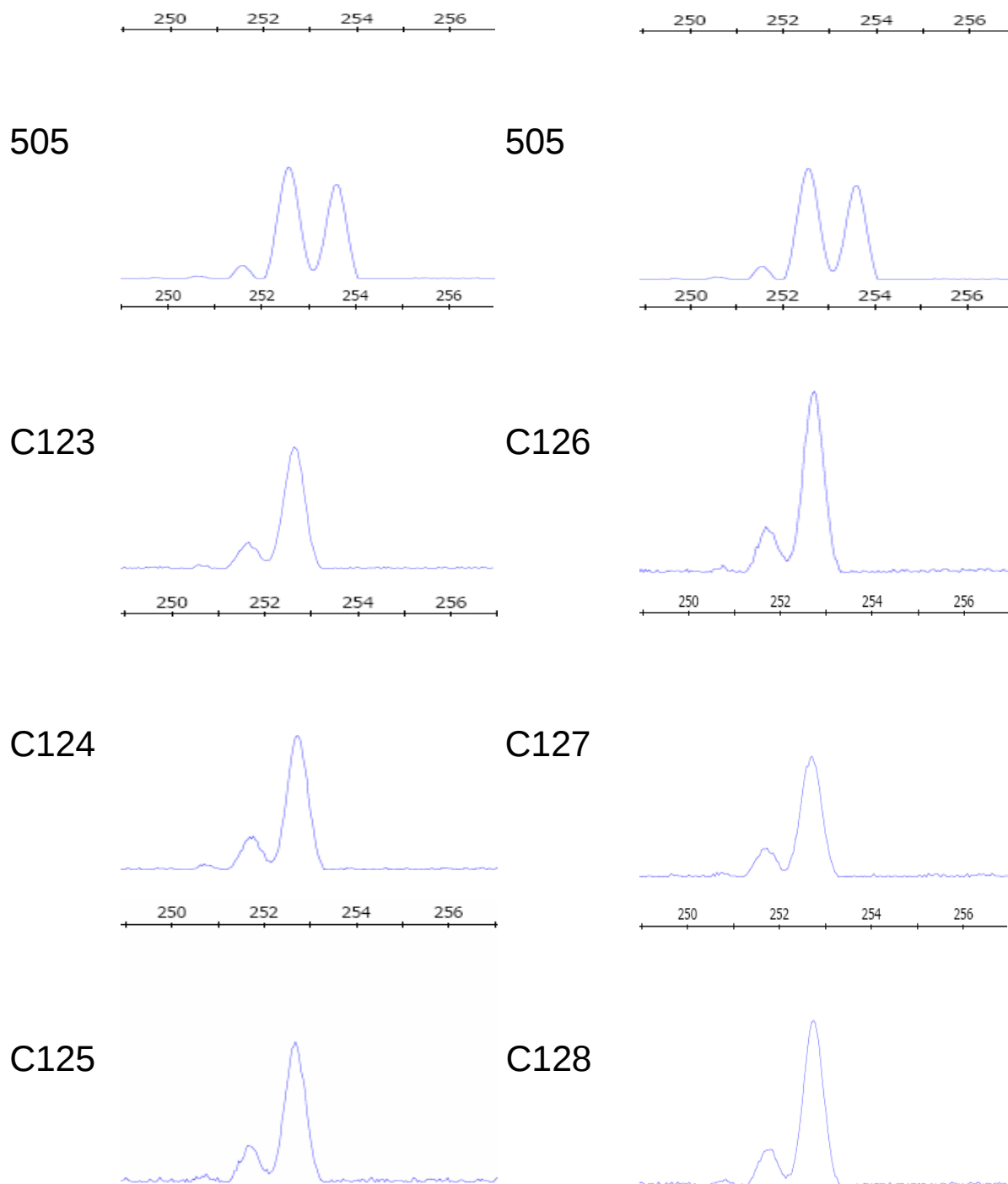
USH1C, controls for c.238-239insC by GeneScan



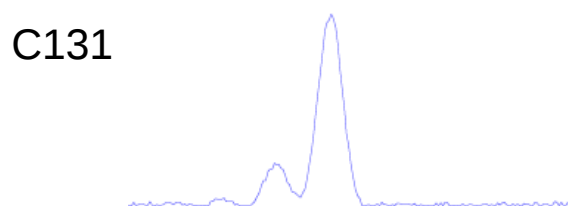
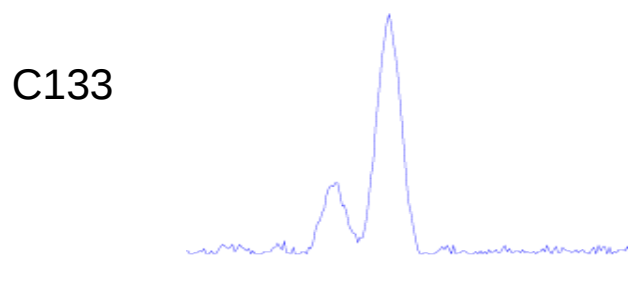
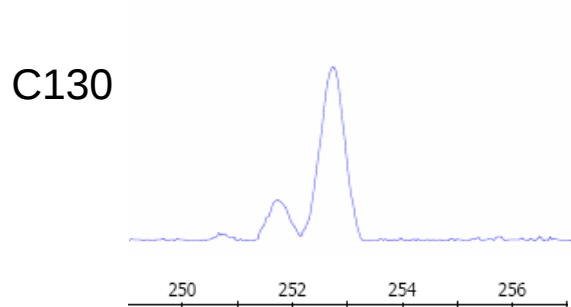
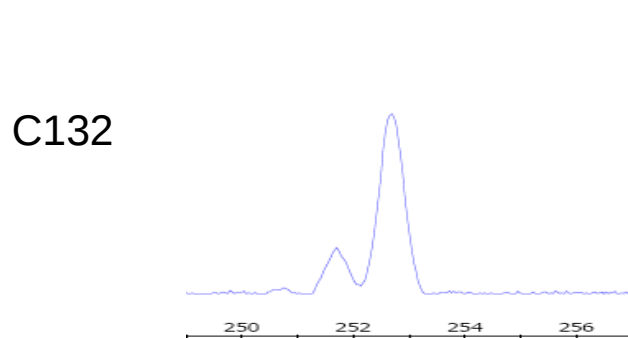
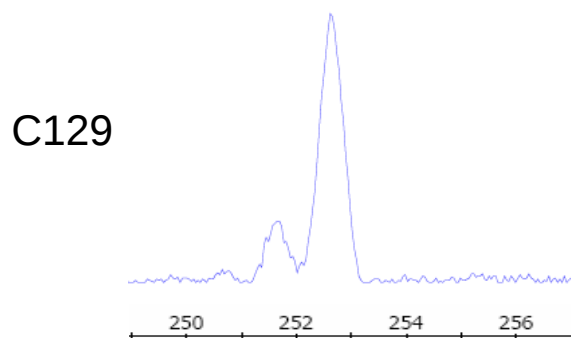
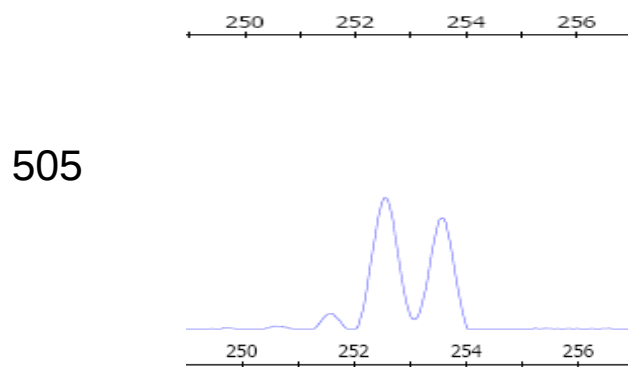
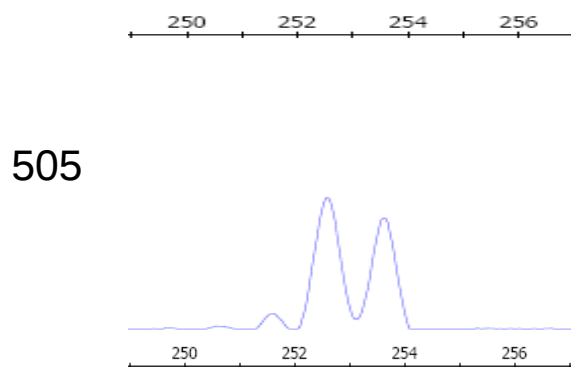
USH1C, controls for c.238-239insC by GeneScan



USH1C, controls for c.238-239insC by GeneScan



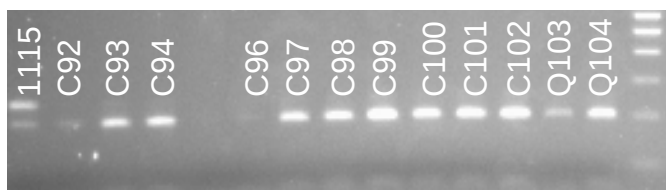
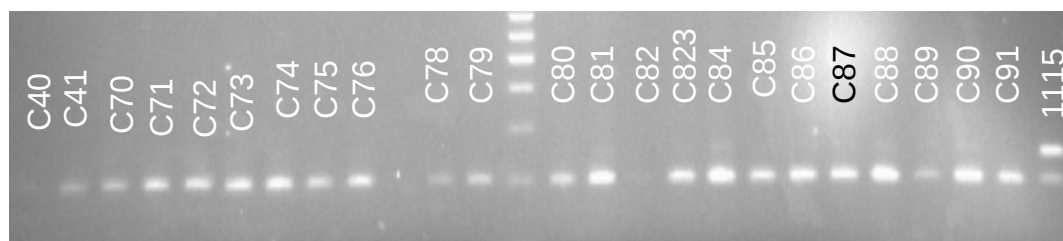
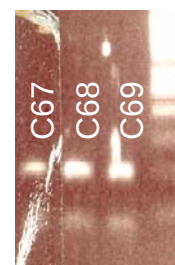
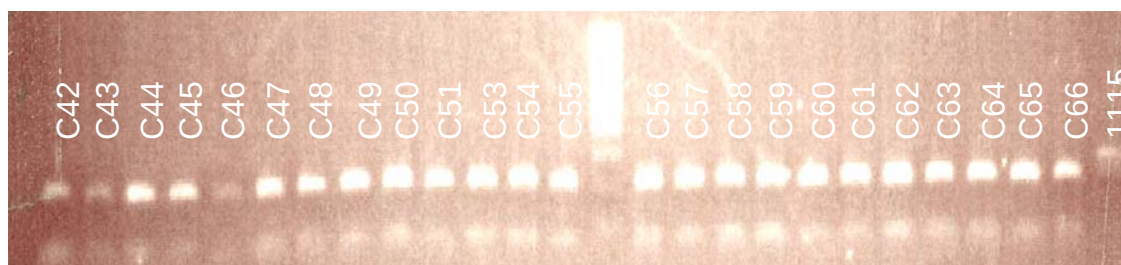
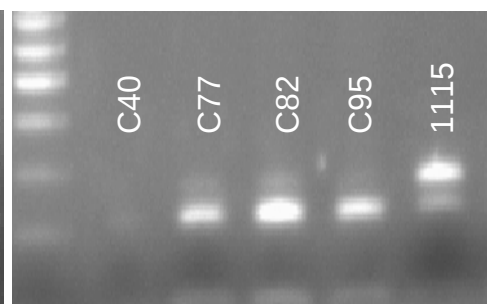
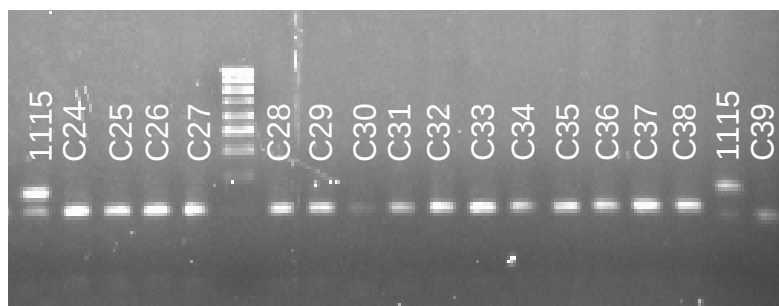
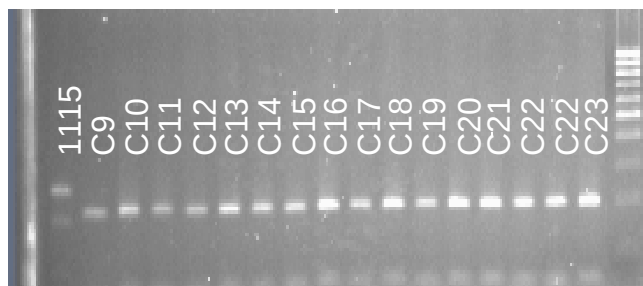
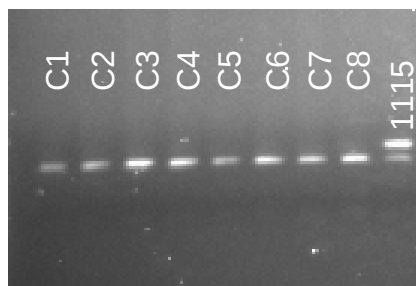
USH1C, controls for c.238-239insC by GeneScan



c.496+1G>T (<i>USH1C</i>): Genotyping of 100 French Canadian healthy control individuals
--

PCR products (297 bp) were digested with *Esp3I*. One *Esp3I* site is present in the wildtype, resulting in fragments of 208 and 88 bp. Presence of c.496+1G>T results in loss of the *Esp3I* site.

Controls for c.496+1G>T (*USH1C*, exon 5)
by restriction digestion with *Esp3I*

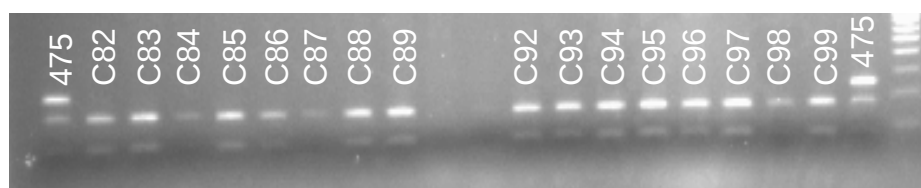
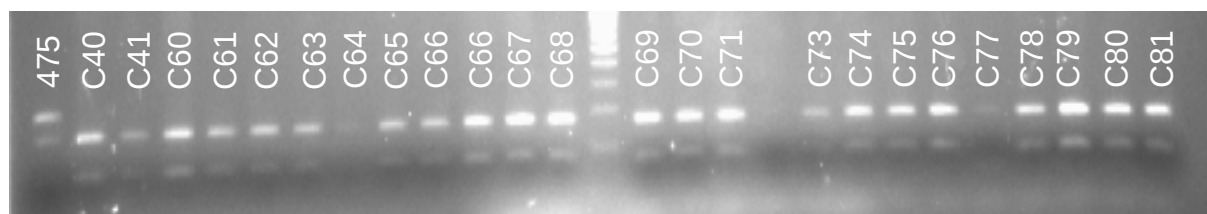
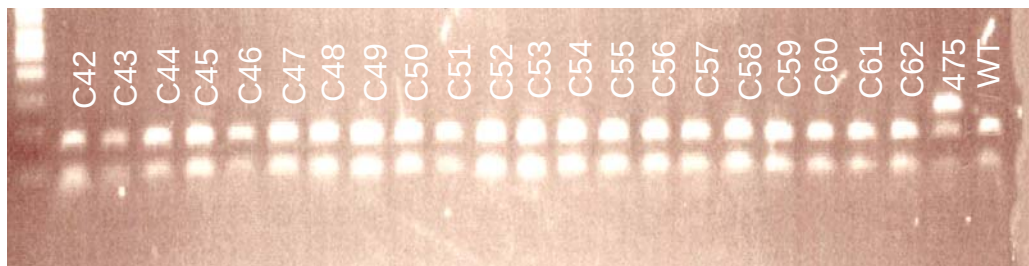
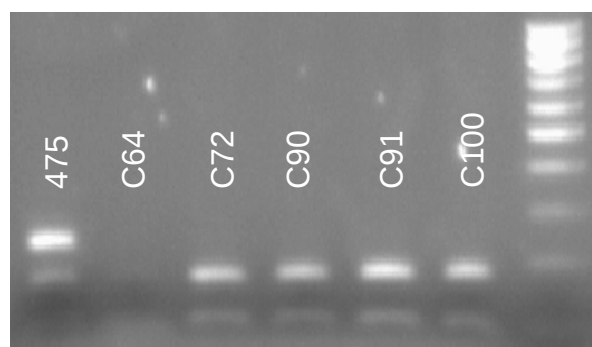
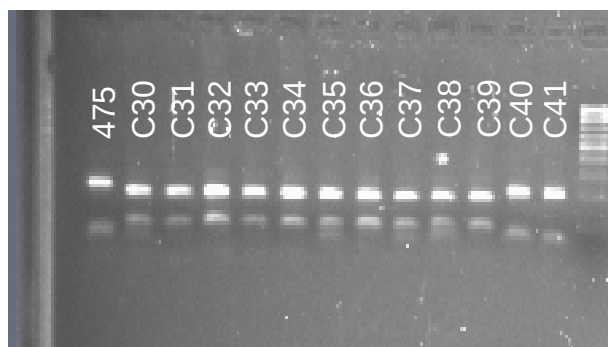
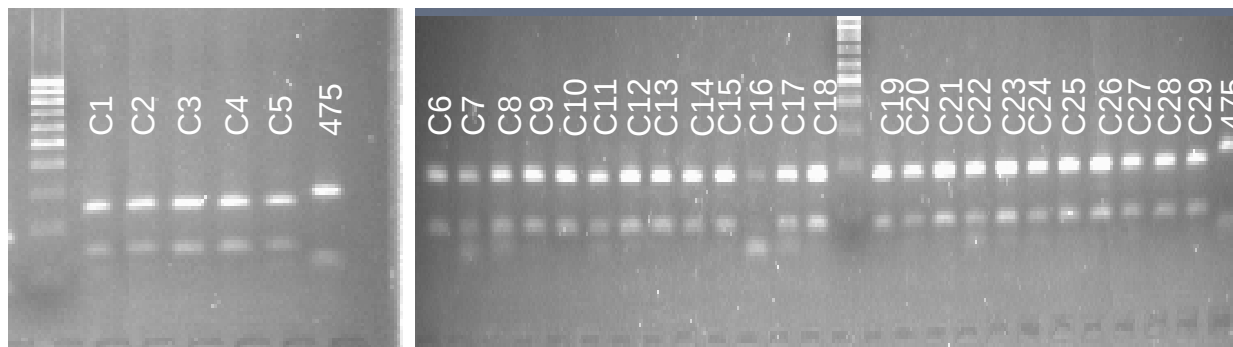


1115: patient heterozygous for c.496+1G>T

p.R155X (<i>USH1C</i>): Genotyping of 100 French Canadian healthy control individuals
--

PCR products (297 bp) were digested with *Bsp119I*. One *Bsp119I* site is present in the wildtype, resulting in fragments of 174 and 123 bp. Presence of c.463C>T results in loss of the *Bsp119I* site.

Controls for c.463C>T/p.R155X (*USH1C*, exon 5)
by restriction digest with *Bsp119I*

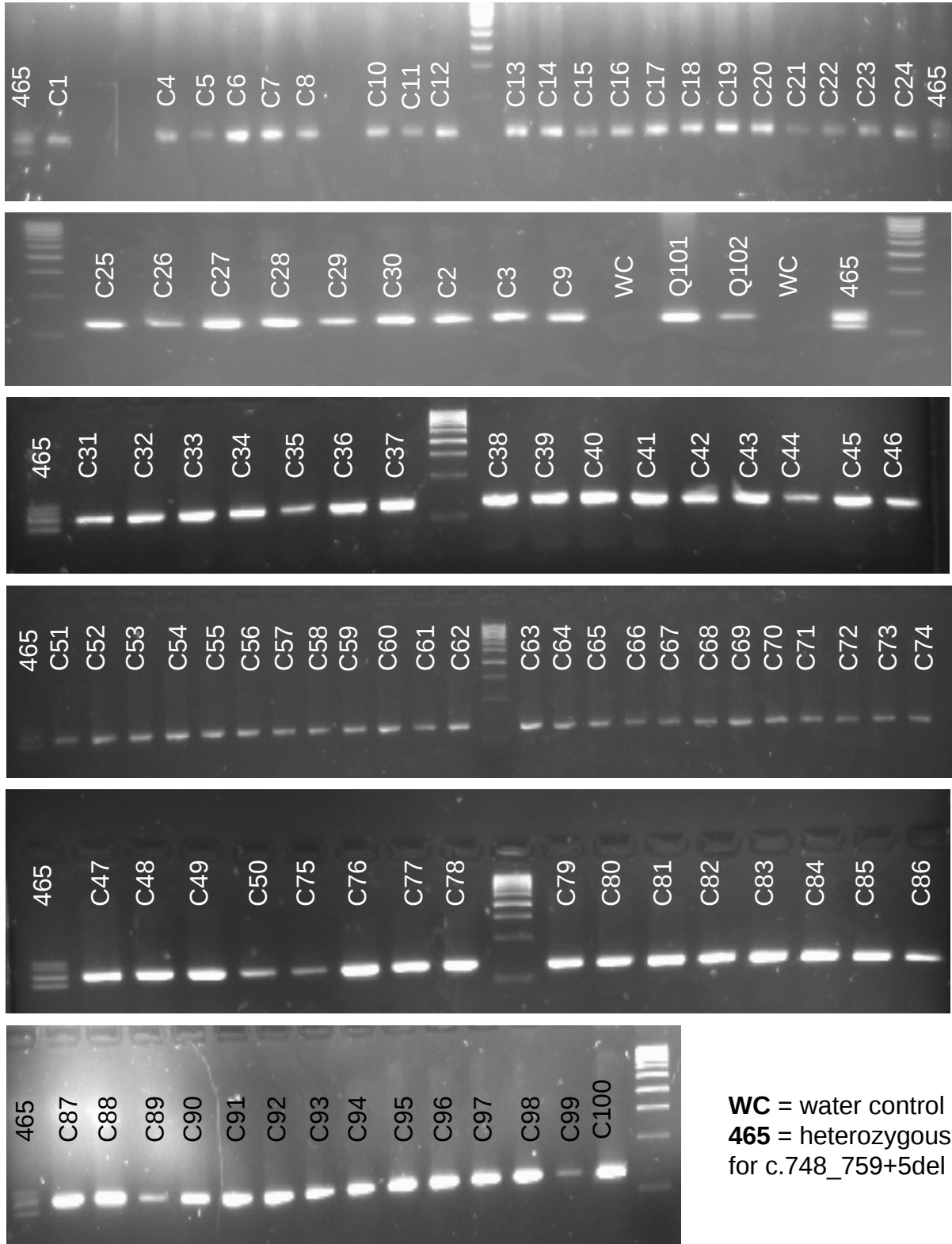


475: patient heterozygous for p.R155X

c.748-759+5del (<i>USH1C</i>): Genotyping of 100 French Canadian healthy control individuals

PCR products (225 bp) were run on a 3% agarose gel. The PCR product heterozygous for c.748-759+5del shows a second band of 208 bp.

Controls for c.748 759+5del (*USH1C*, exon 9)
by electrophoresis on a 3% agarose gel



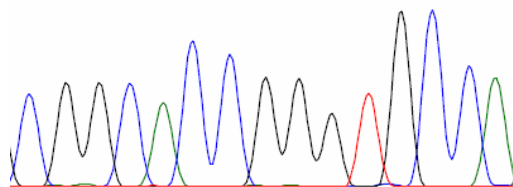
**IVS45-9G>A (*CDH23*):
Genotyping of 100 French Canadian healthy control individuals**

- by direct sequencing of PCR products -

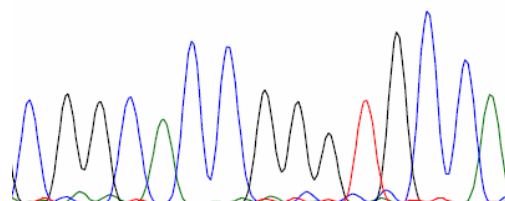
Controls for *CDH23*, IVS45-9G>A



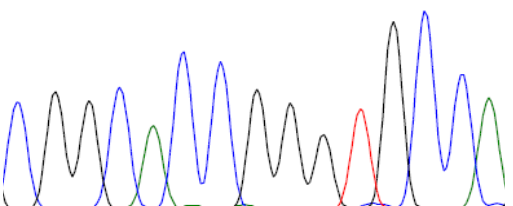
C1



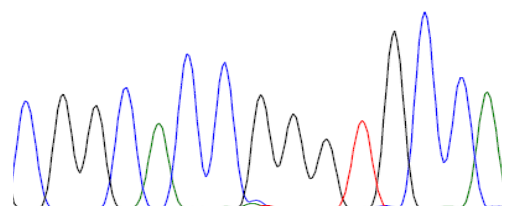
C2



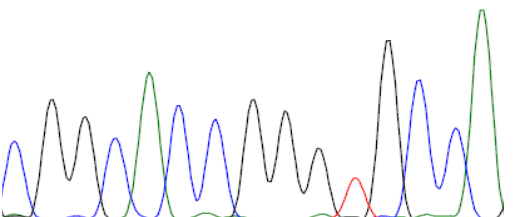
C3



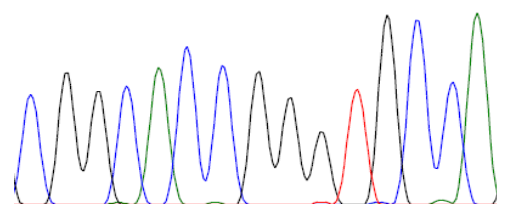
C4



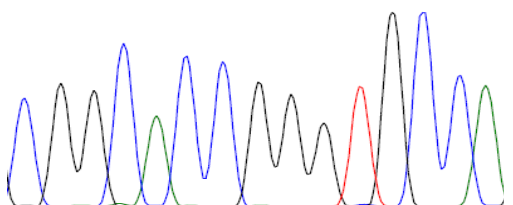
C5



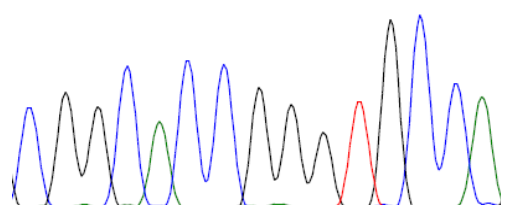
C6



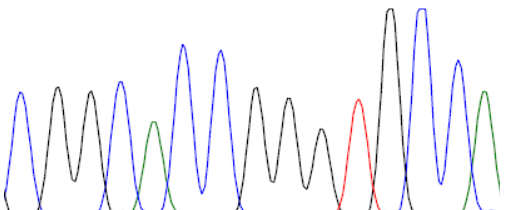
C7



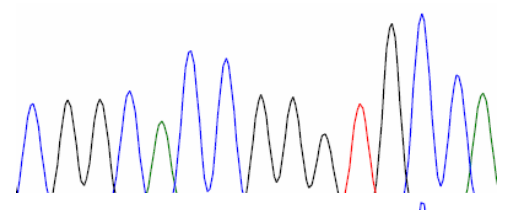
C8



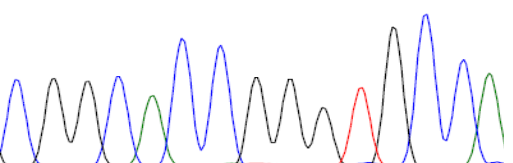
C9



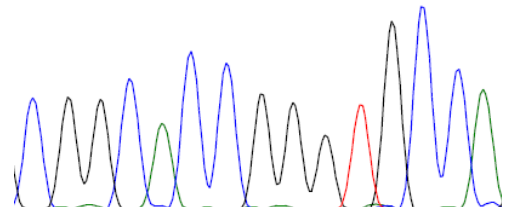
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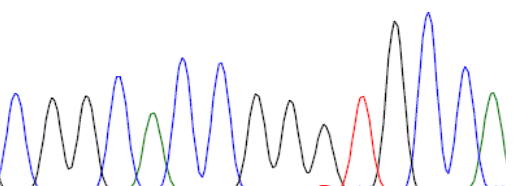
C11



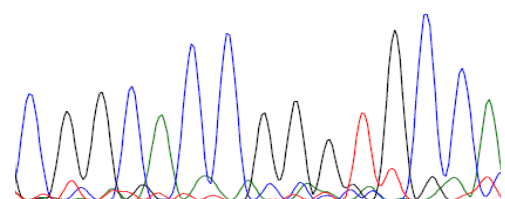
C12



C13



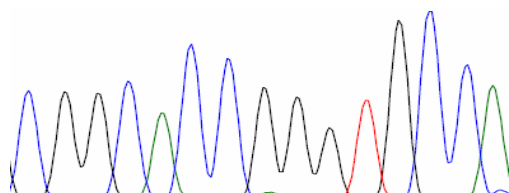
C14



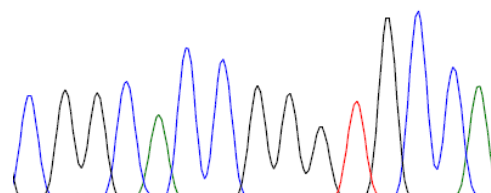
Controls for *CDH23*, IVS45-9G>A



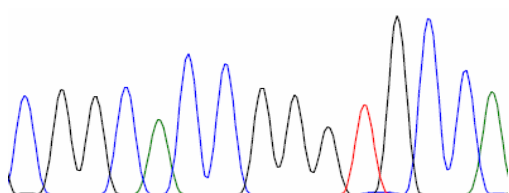
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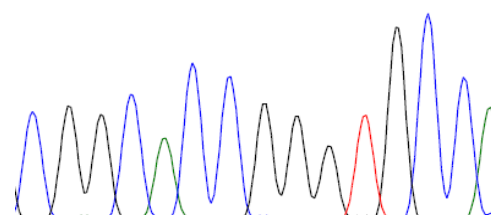
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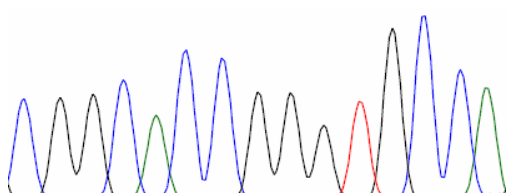
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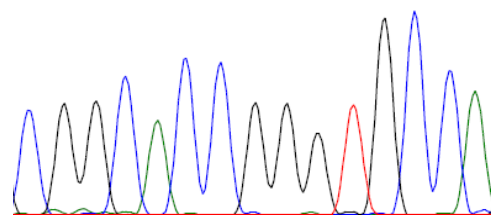
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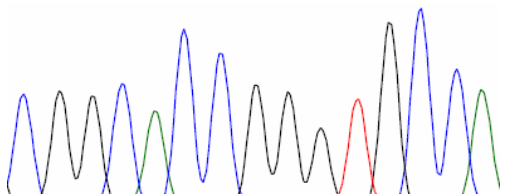
C19



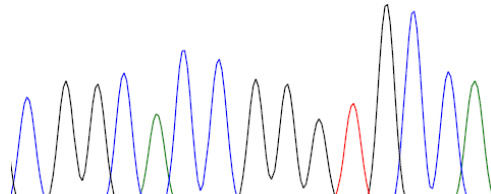
C20



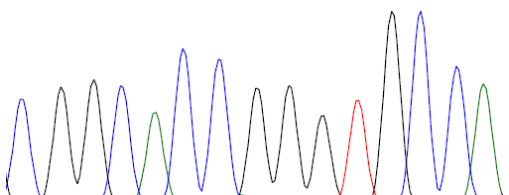
C21



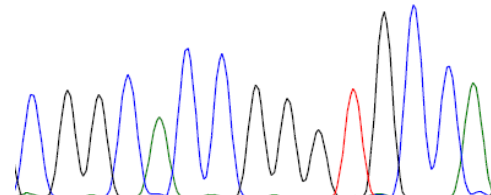
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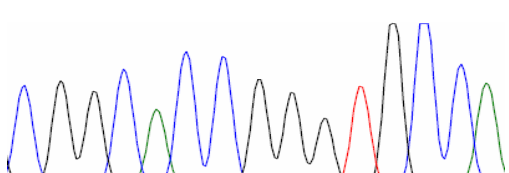
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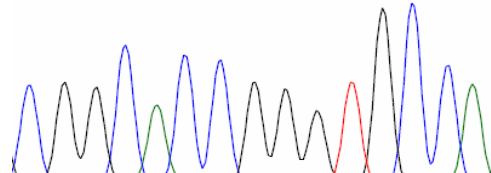
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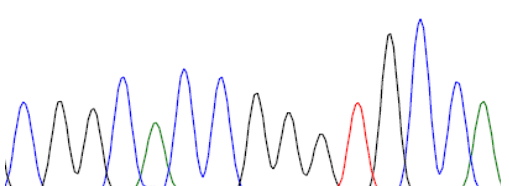
C26



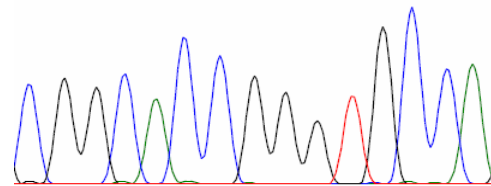
C27



C28



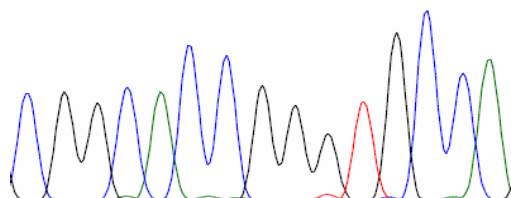
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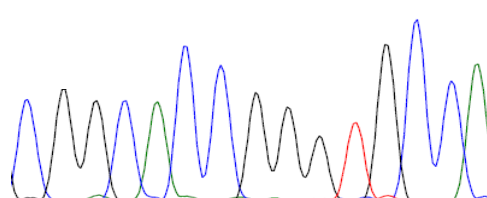
Controls for *CDH23*, IVS45-9G>A



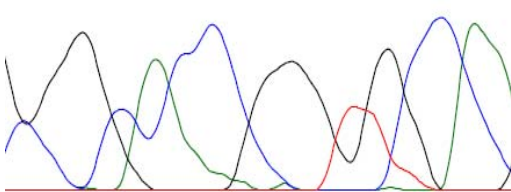
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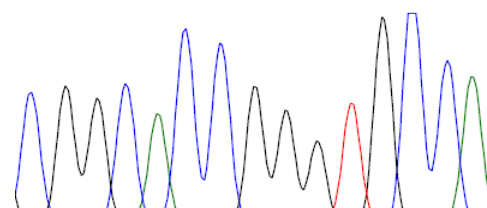
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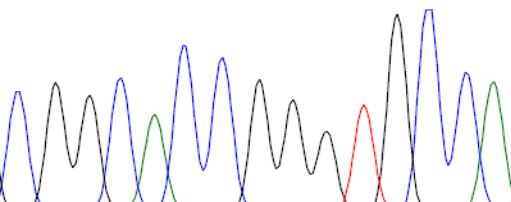
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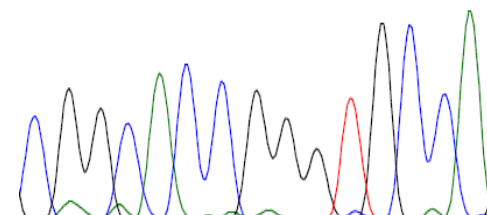
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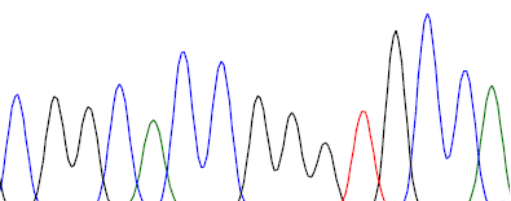
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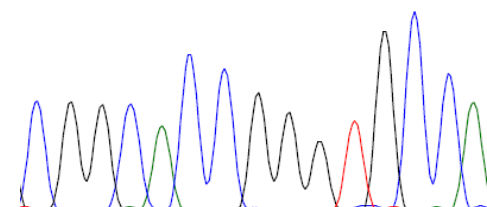
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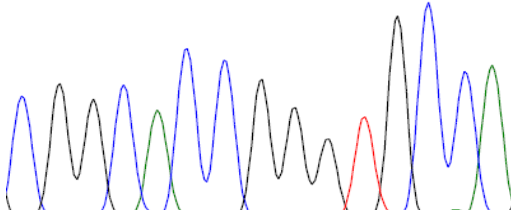
C36



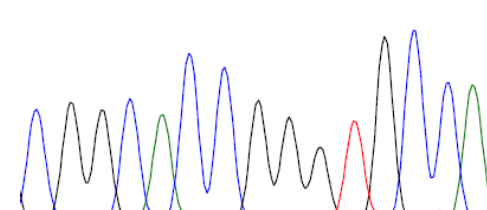
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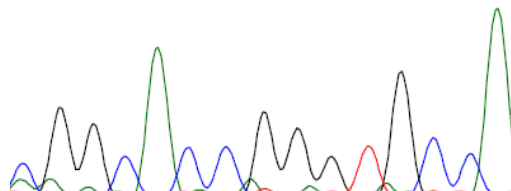
C38



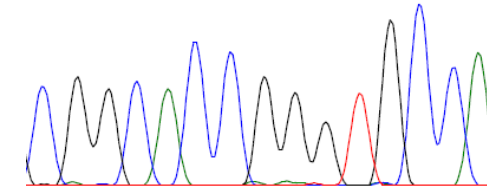
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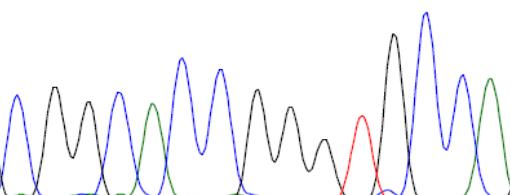
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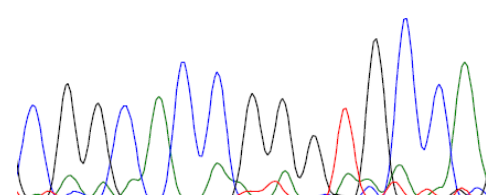
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C43



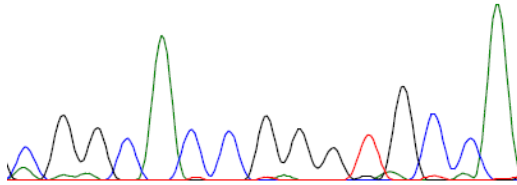
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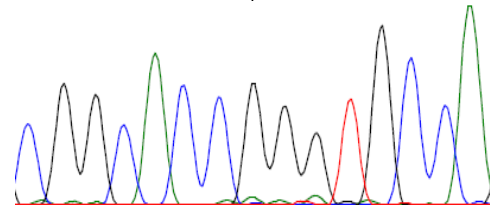
Controls for *CDH23*, IVS45-9G>A



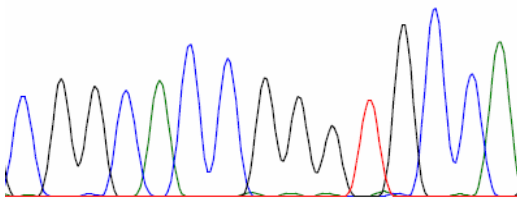
C45



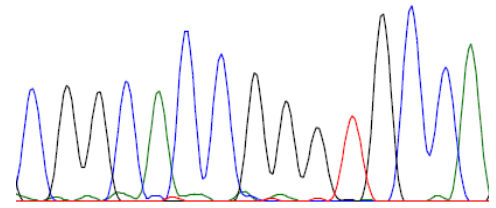
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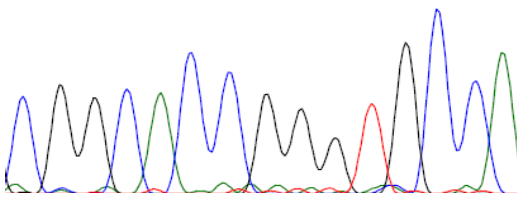
C47



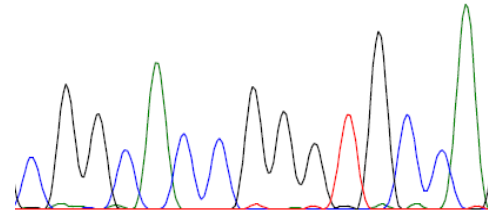
C48



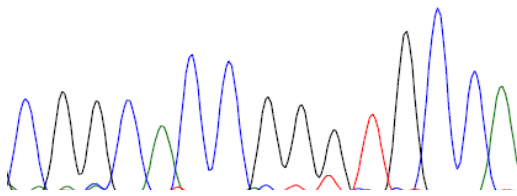
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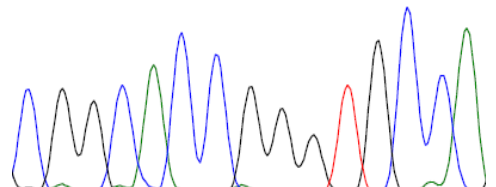
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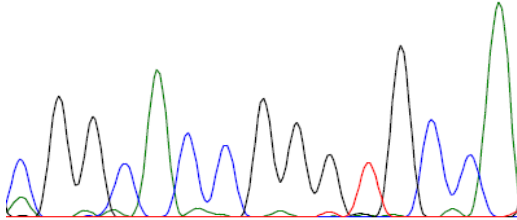
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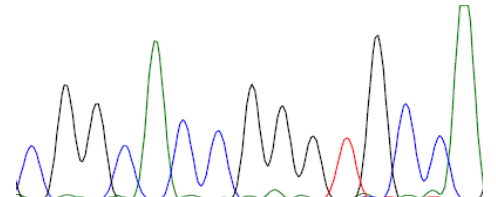
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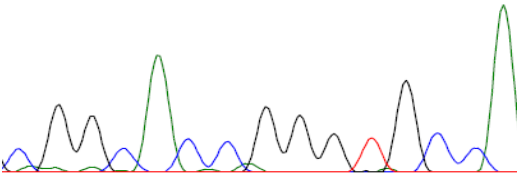
C53



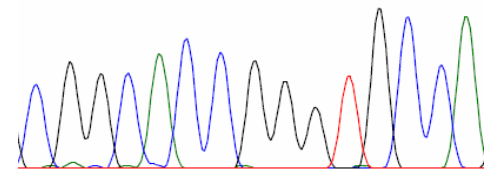
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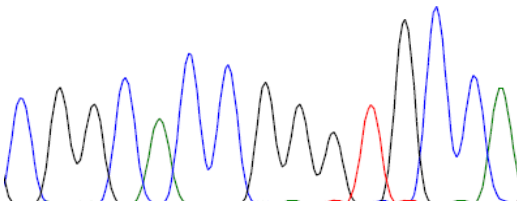
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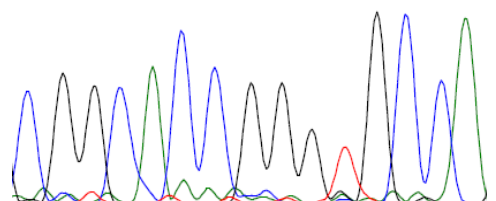
C56



C57



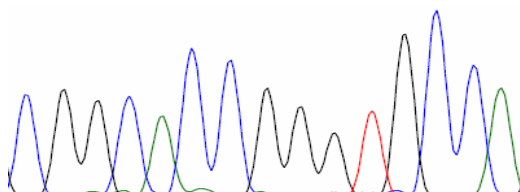
C58



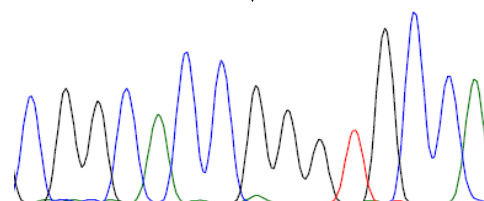
Controls for *CDH23*, IVS45-9G>A



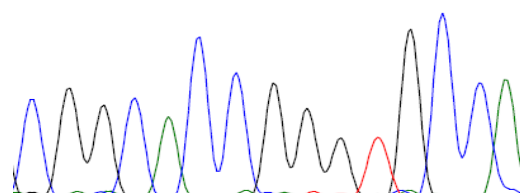
C59



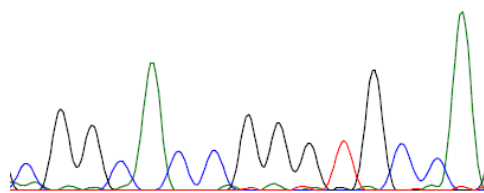
C60



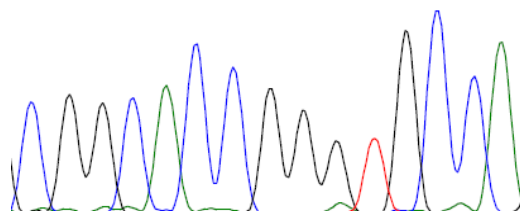
C61



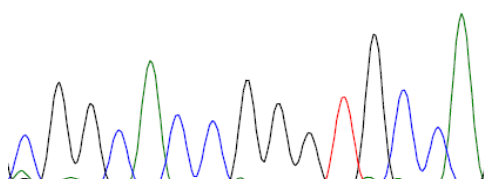
C62



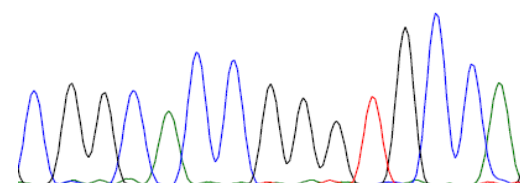
C63



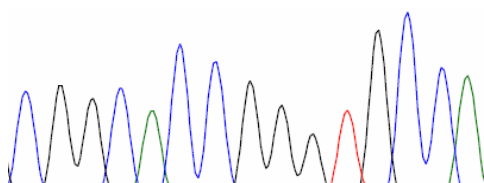
C64



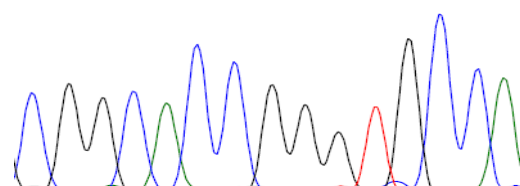
C65



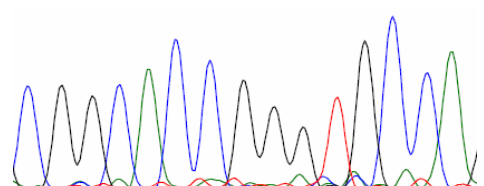
C66



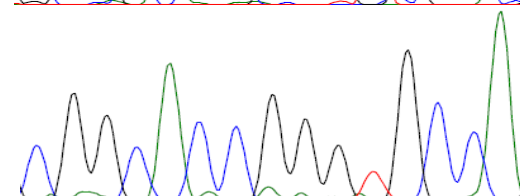
C67



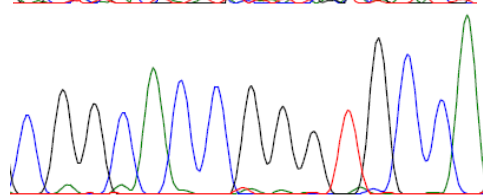
C68



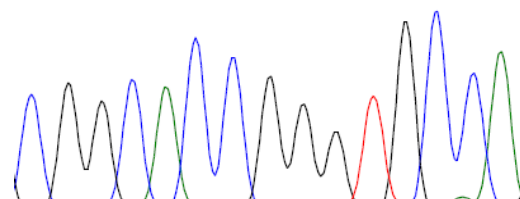
C69



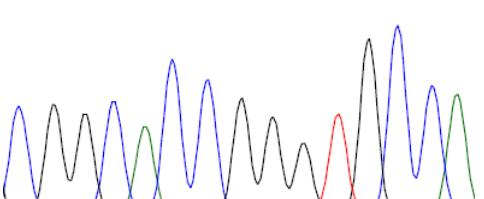
C70



C71



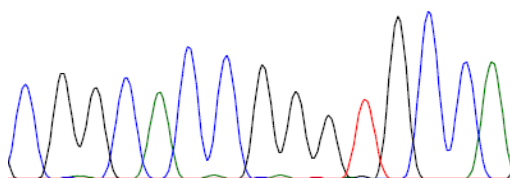
C72



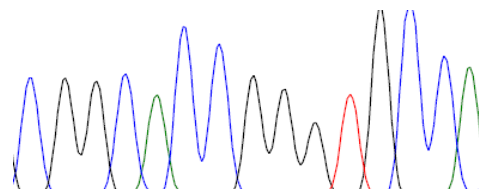
Controls for *CDH23*, IVS45-9G>A



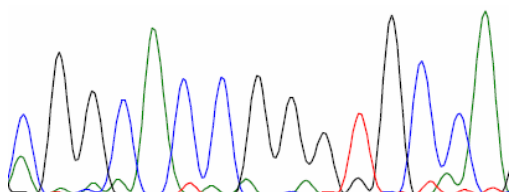
C73



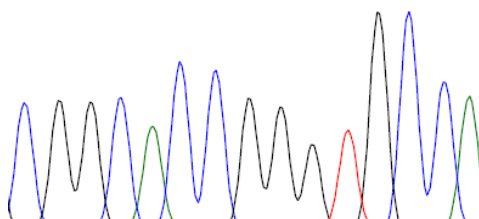
C74



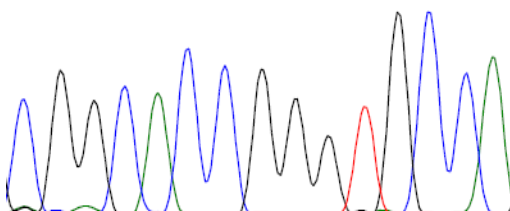
C75



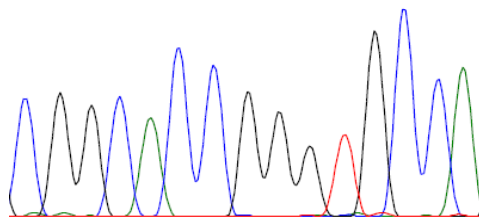
C76



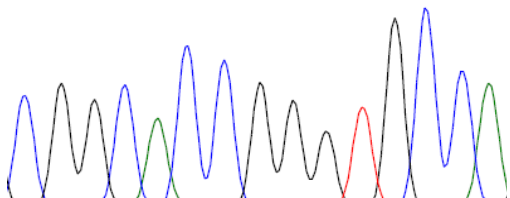
C77



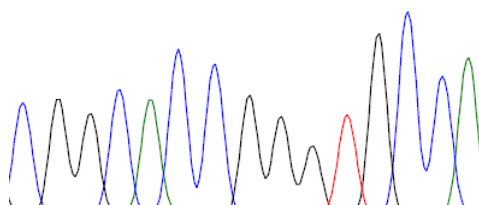
C78



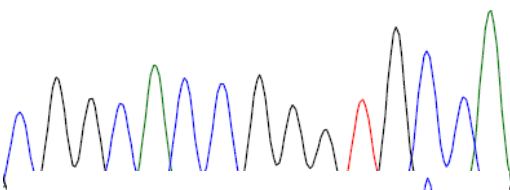
C79



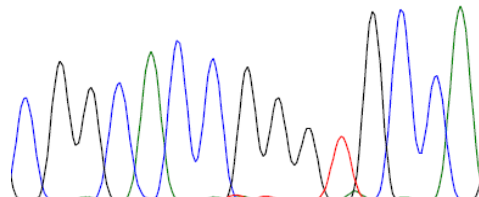
C80



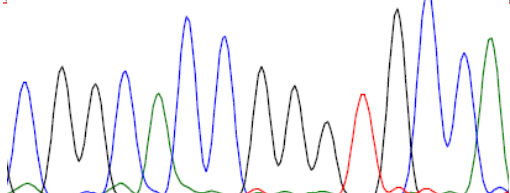
C82



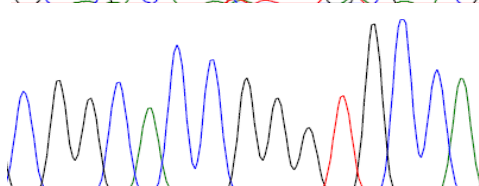
C83



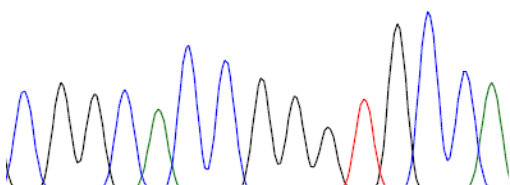
C84



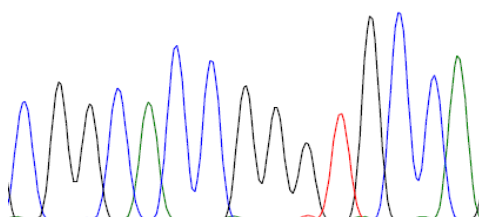
C85



C87



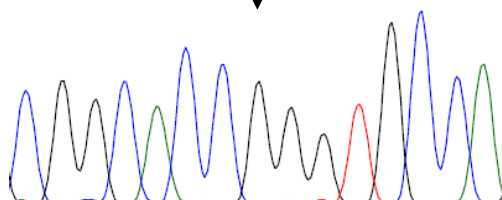
C88



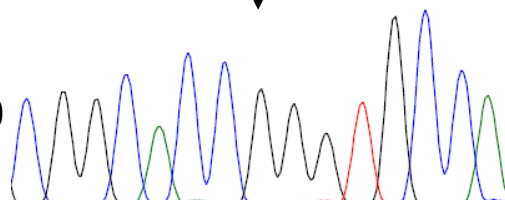
Controls for *CDH23*, IVS45-9G>A



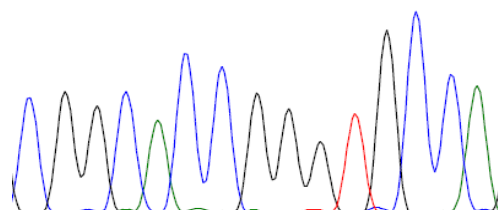
C89



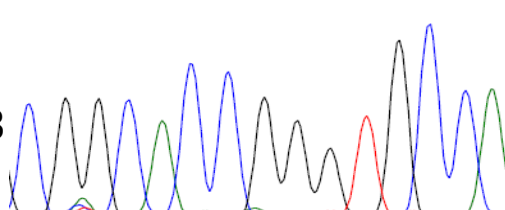
C90



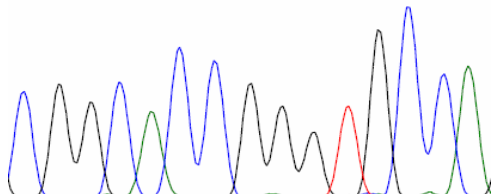
C91



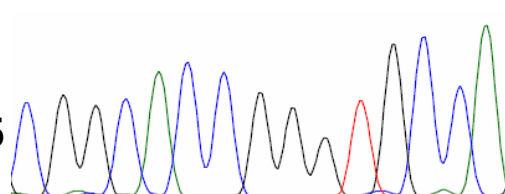
C93



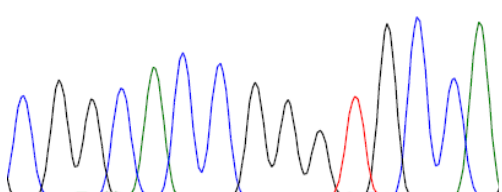
C94



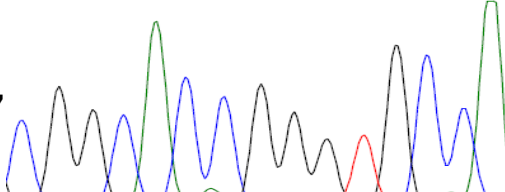
C95



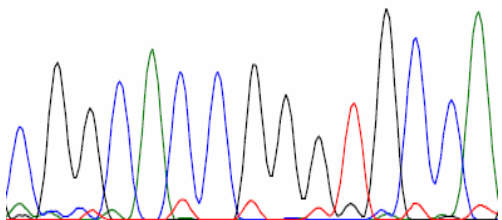
C96



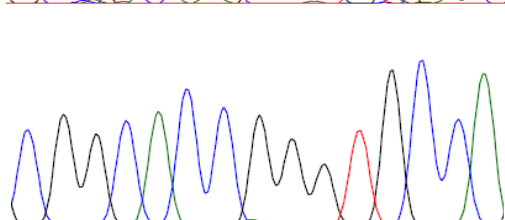
C97



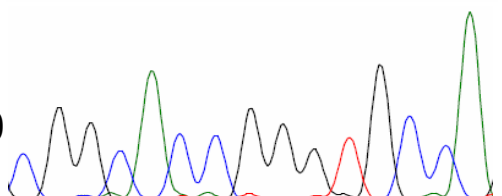
C98



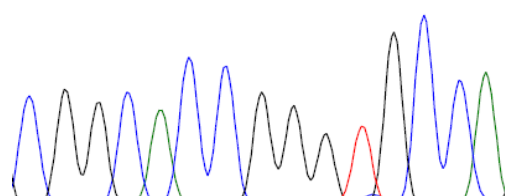
C99



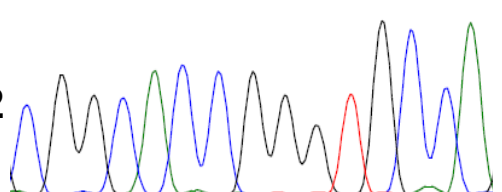
C100



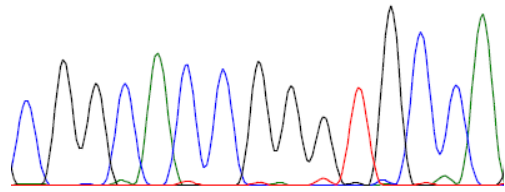
C101



C102



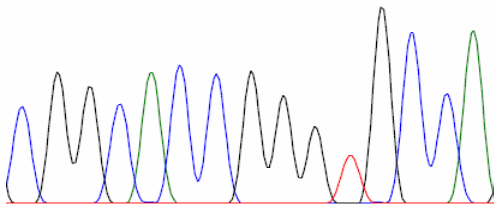
C103



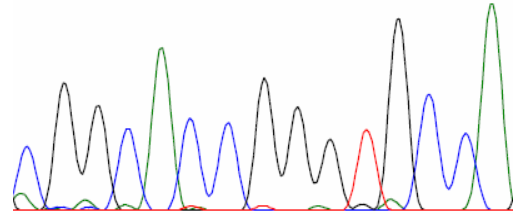
Controls for *CDH23*, IVS45-9G>A



C102



C110



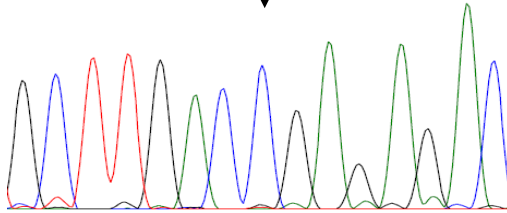
p.R736X (*CDH23*):
Genotyping of 100 French Canadian healthy control individuals

- by direct sequencing of PCR products -

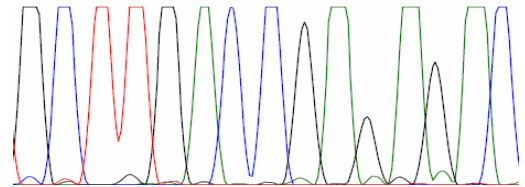
Controls for *CDH23*, c. 2206C>T (p.R736X)



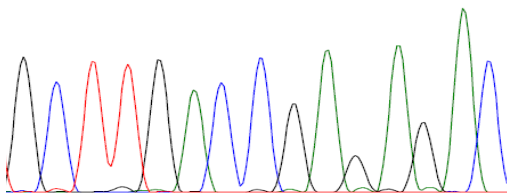
C2



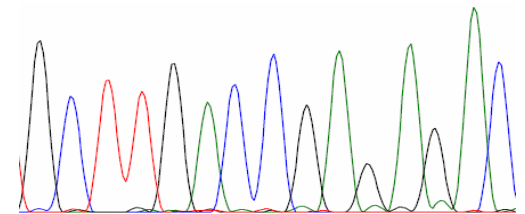
C3



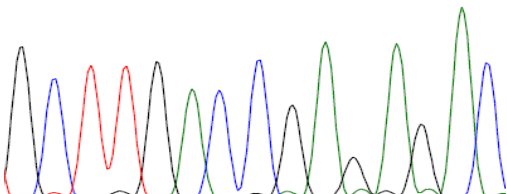
C4



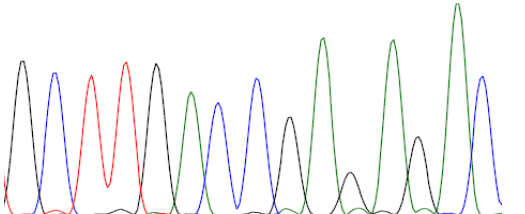
C5



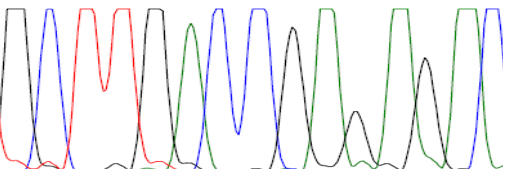
C6



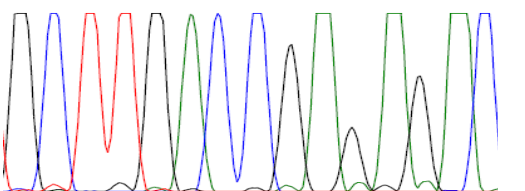
C7



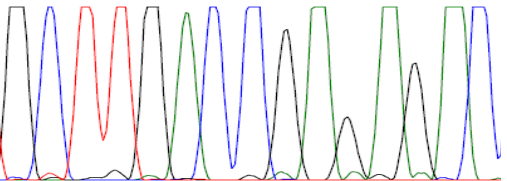
C8



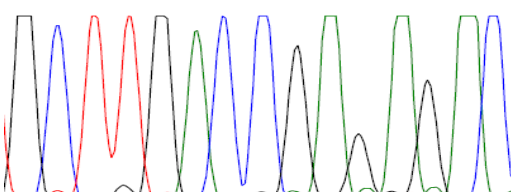
C9



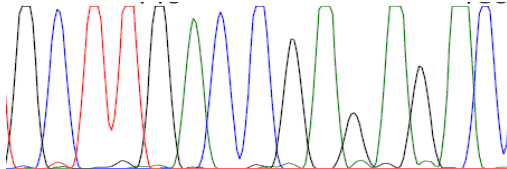
C10



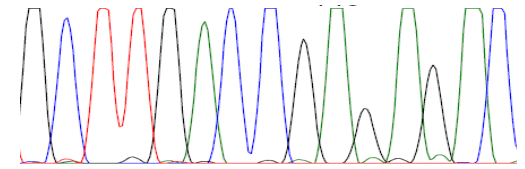
C11



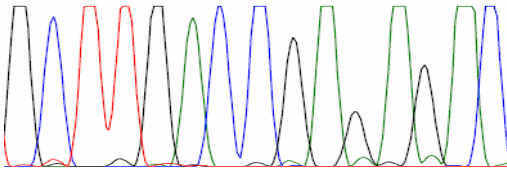
C12



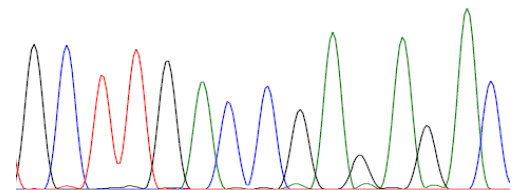
C13



C14



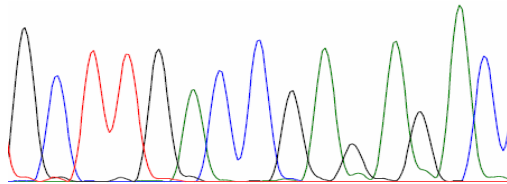
C15



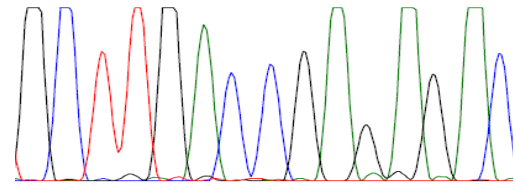
Controls for *CDH23*, c. 2206C>T (p.R736X)



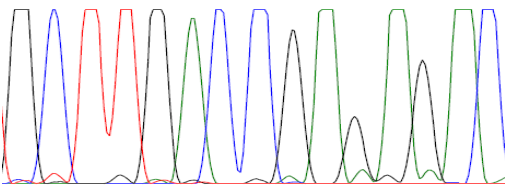
C16



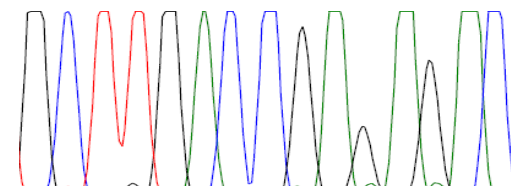
C17



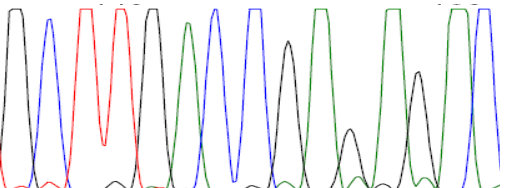
C18



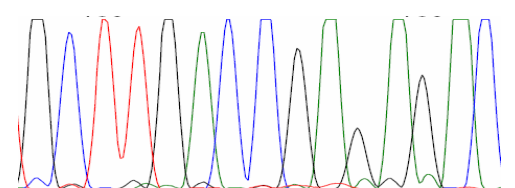
C19



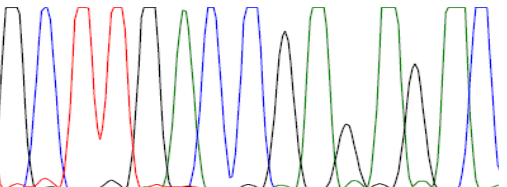
C20



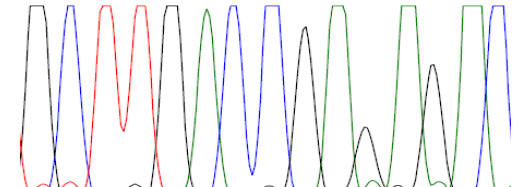
C21



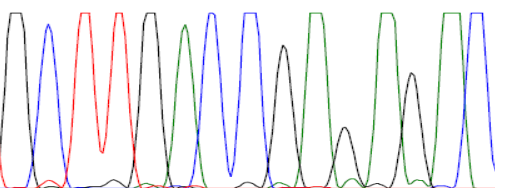
C22



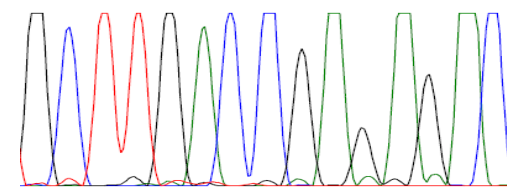
C23



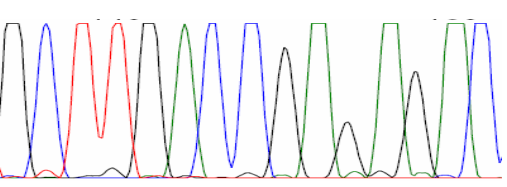
C24



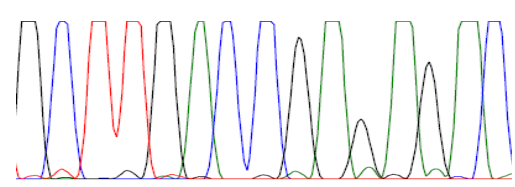
C25



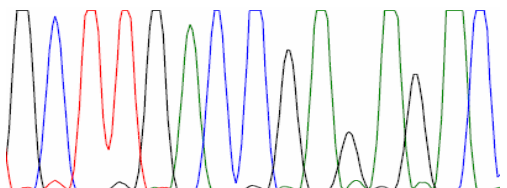
C26



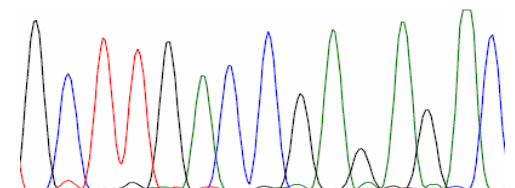
C27



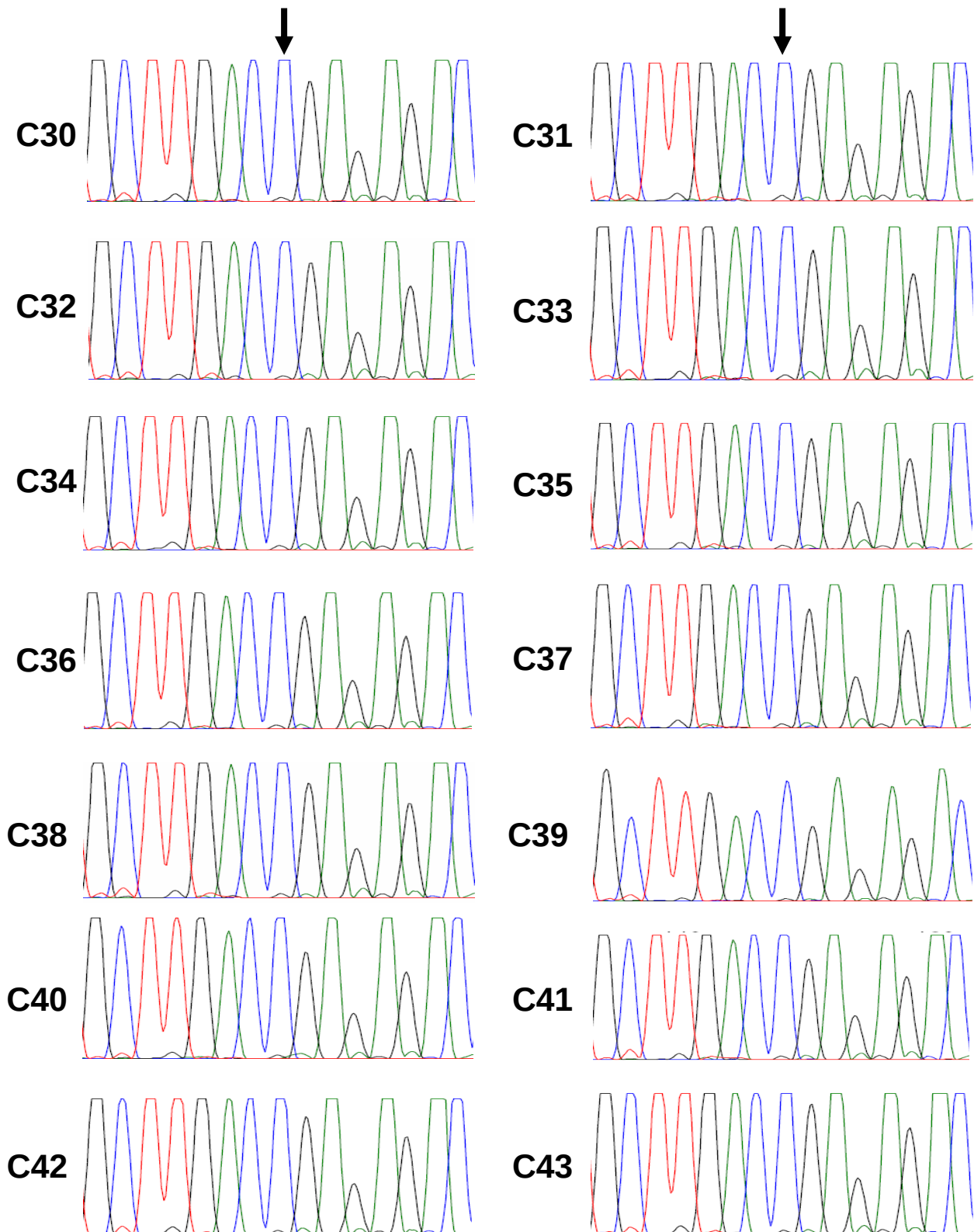
C28



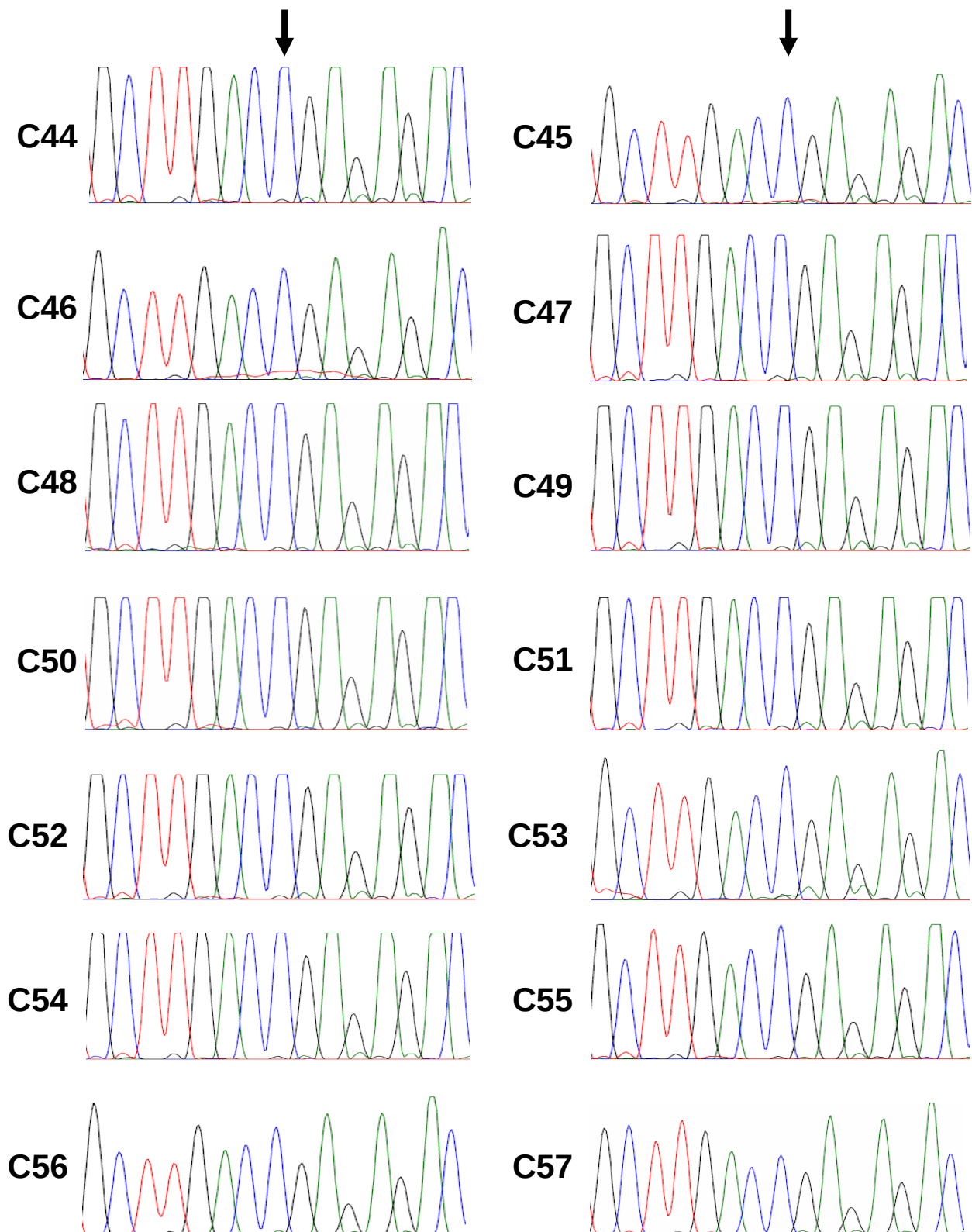
C29



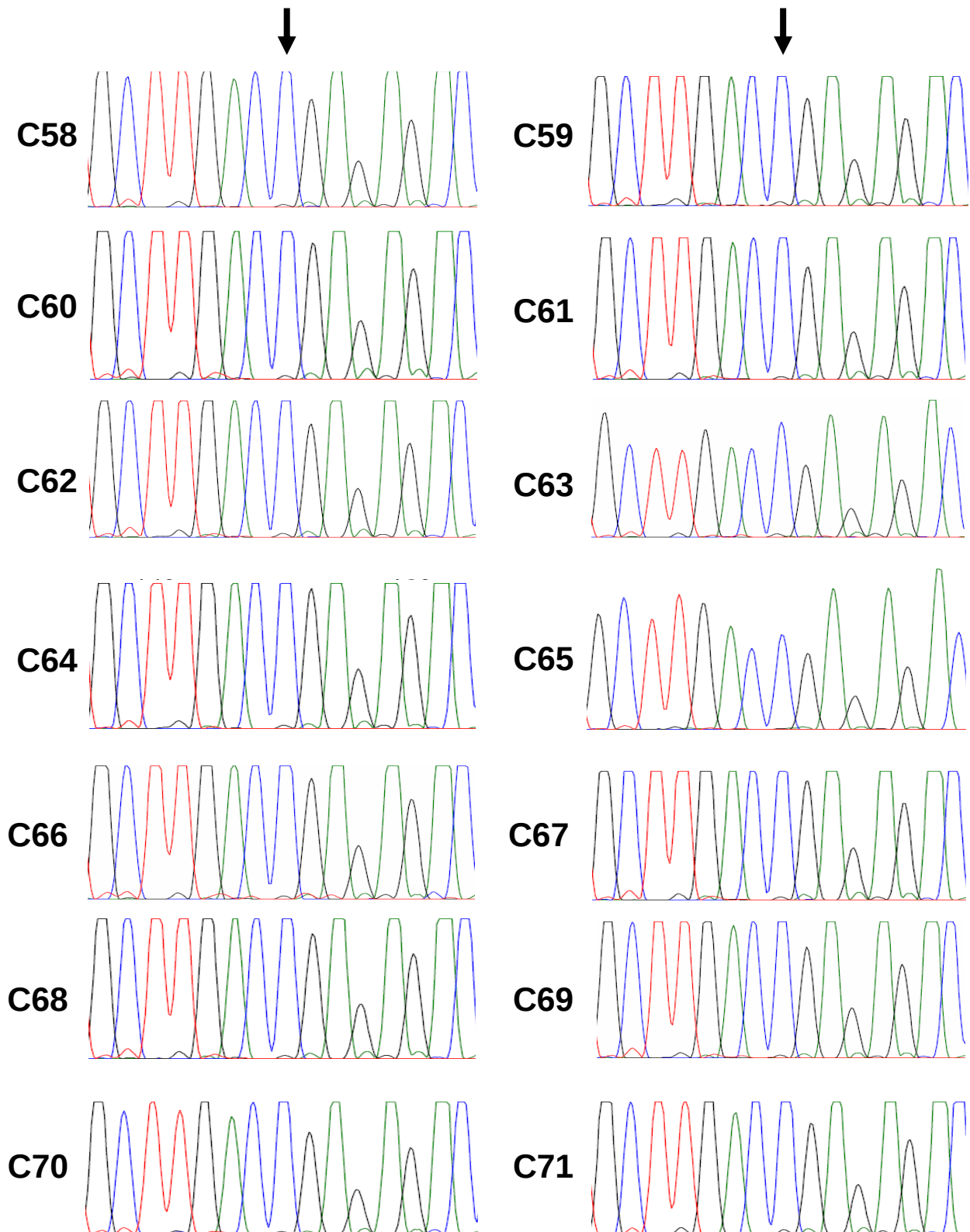
Controls for *CDH23*, c. 2206C>T (p.R736X)



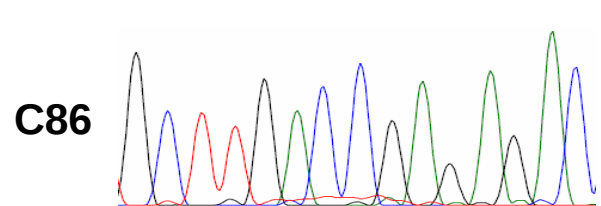
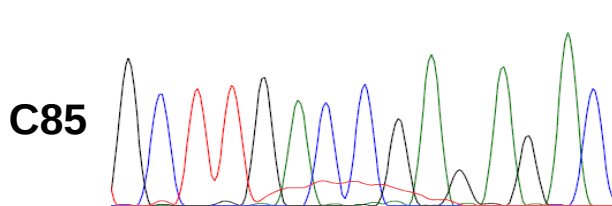
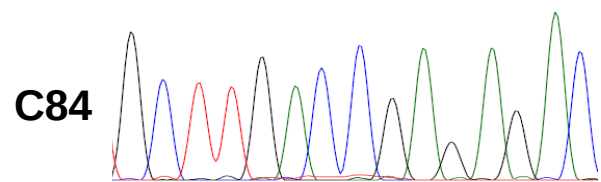
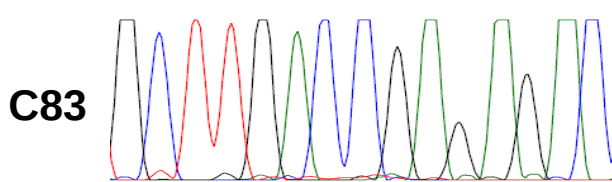
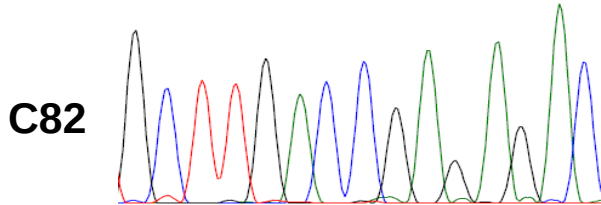
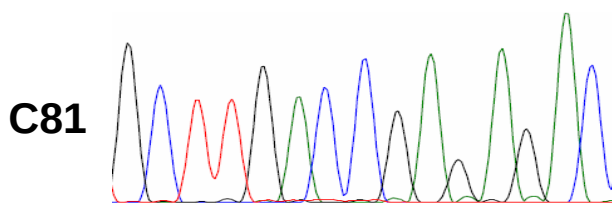
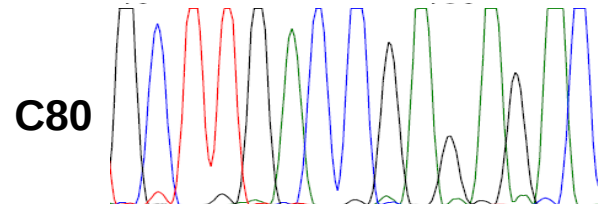
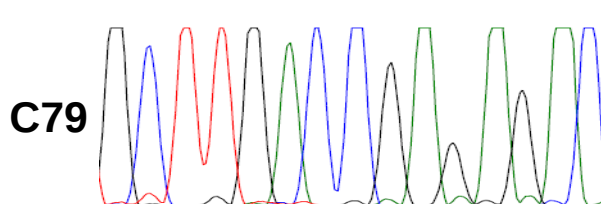
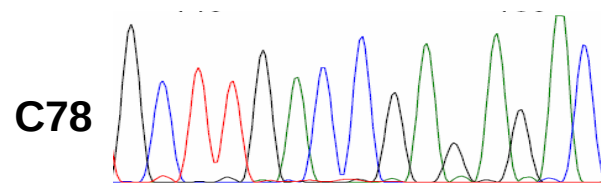
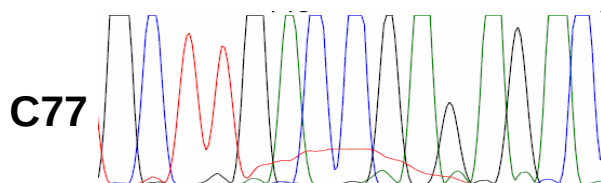
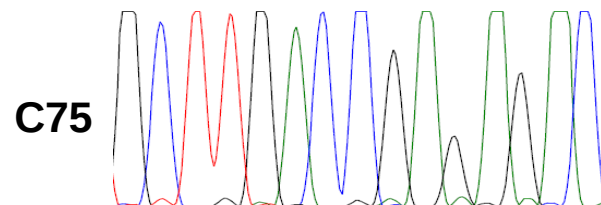
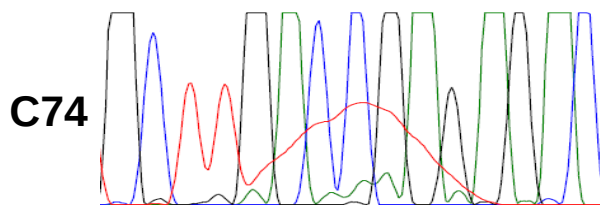
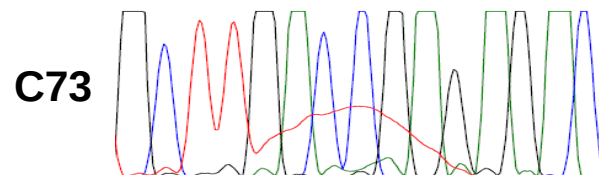
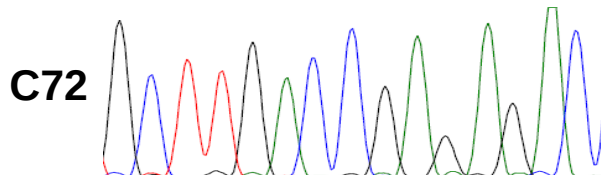
Controls for *CDH23*, c. 2206C>T (p.R736X)



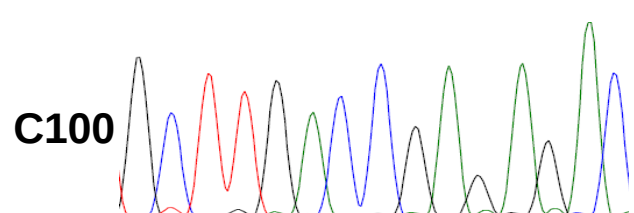
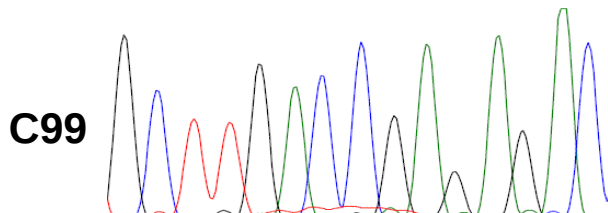
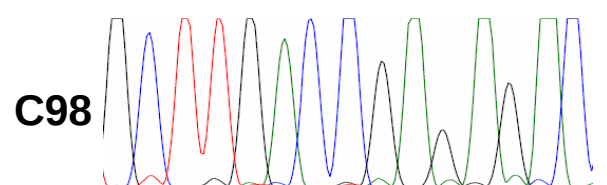
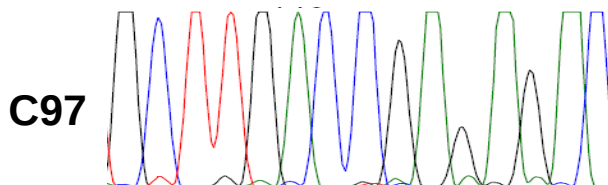
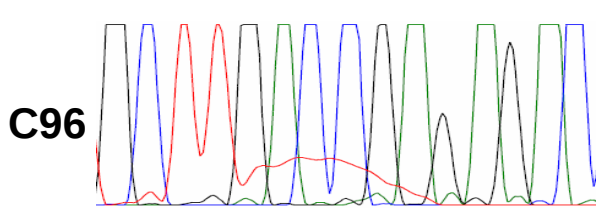
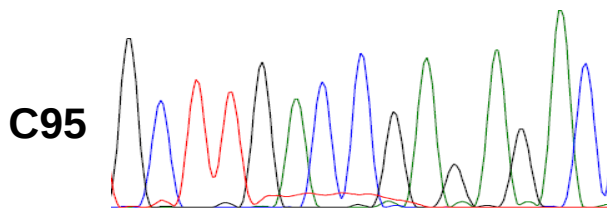
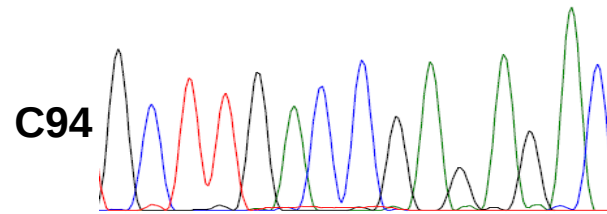
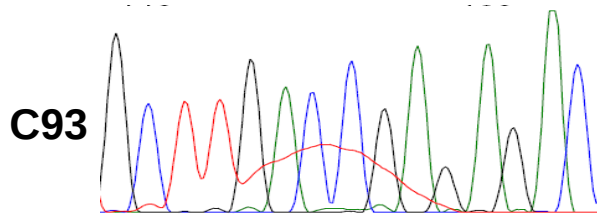
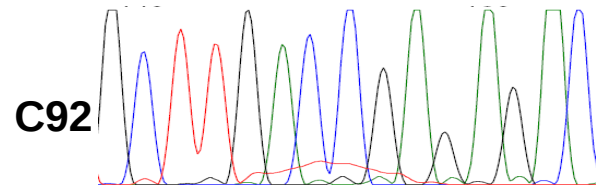
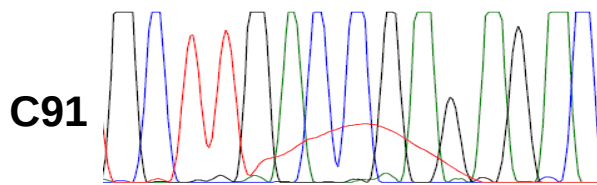
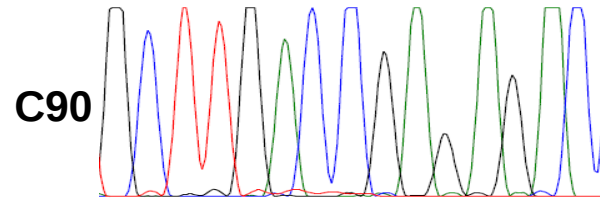
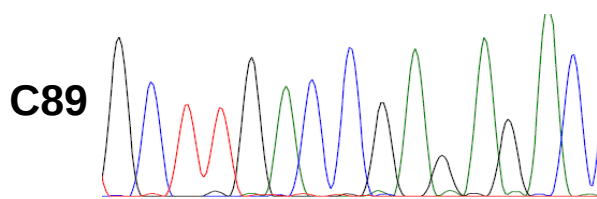
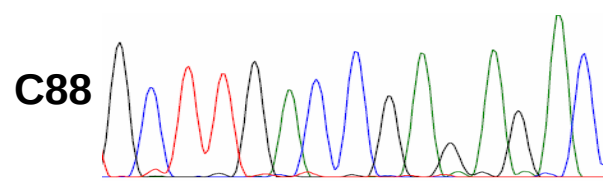
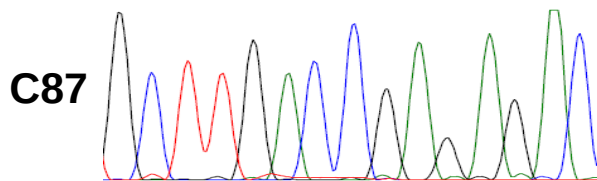
Controls for *CDH23*, c. 2206C>T (p.R736X)



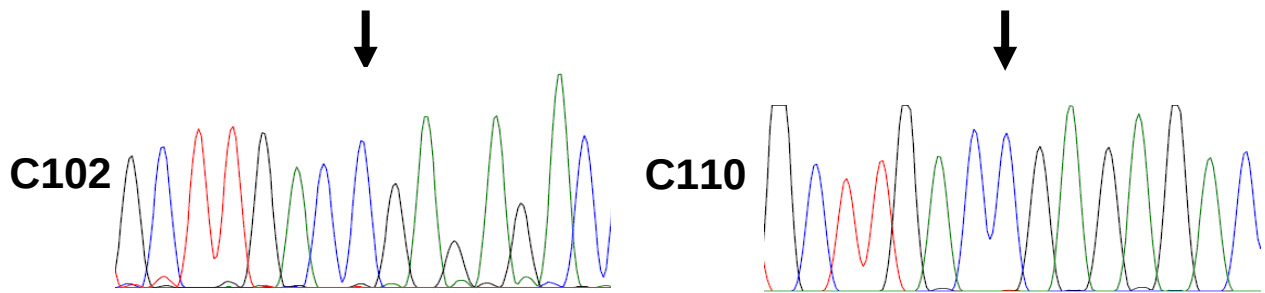
Controls for *CDH23*, c. 2206C>T (p.R736X)



Controls for *CDH23*, c. 2206C>T (p.R736X)



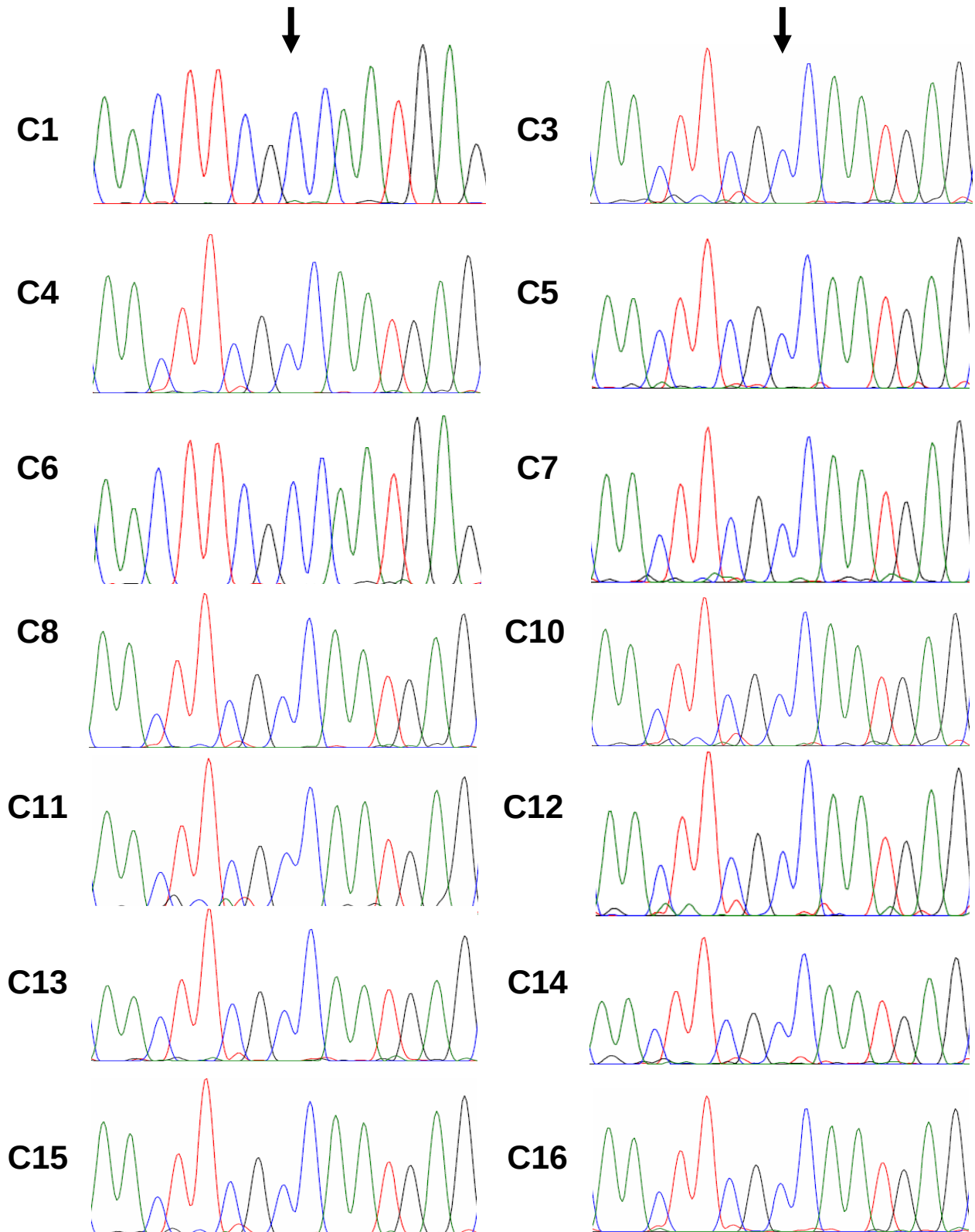
Controls for *CDH23*, c. 2206C>T (p.R736X)



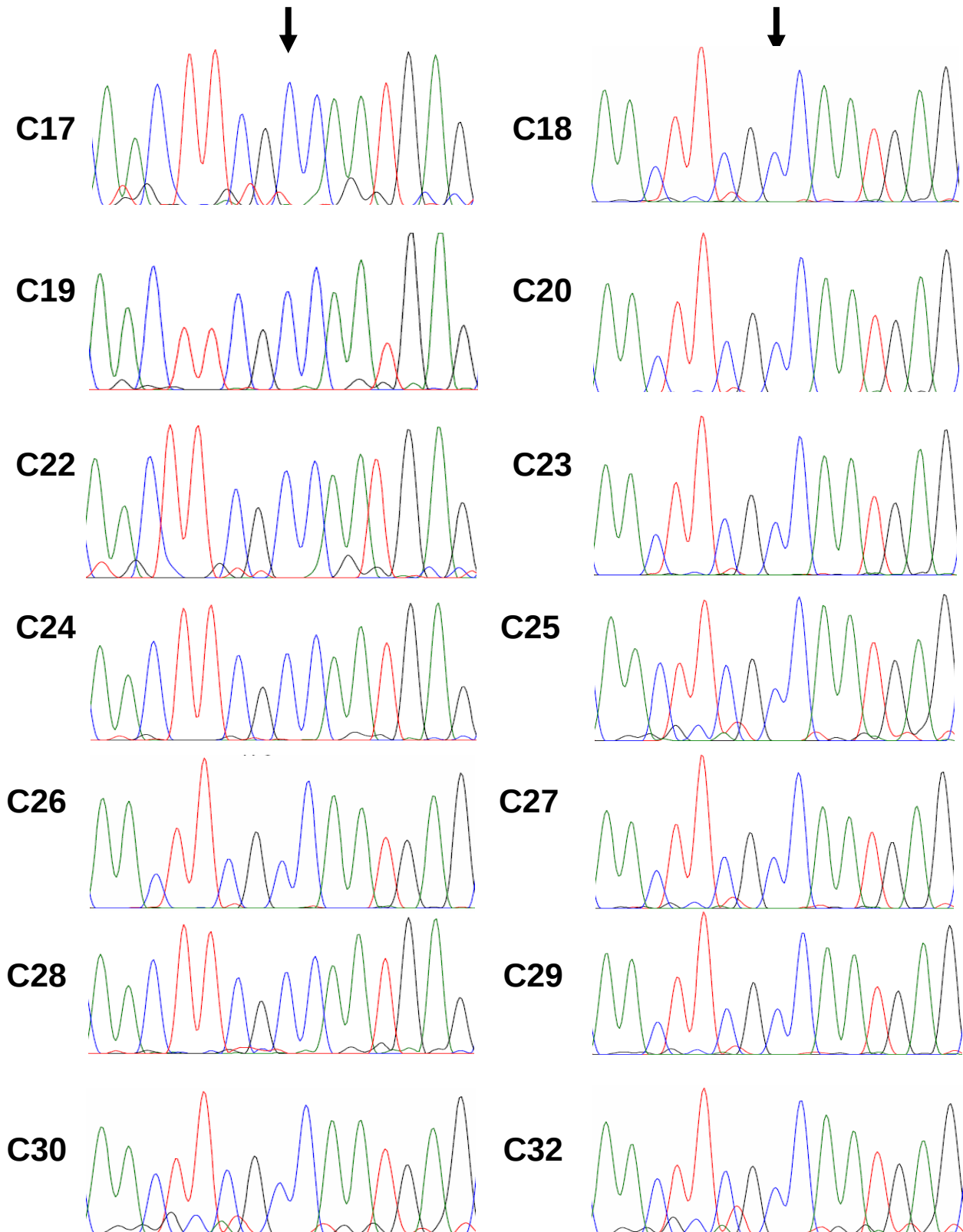
p.A457V (*MYO7A*):
Genotyping of 100 French Canadian healthy control individuals

- by direct sequencing of PCR products -

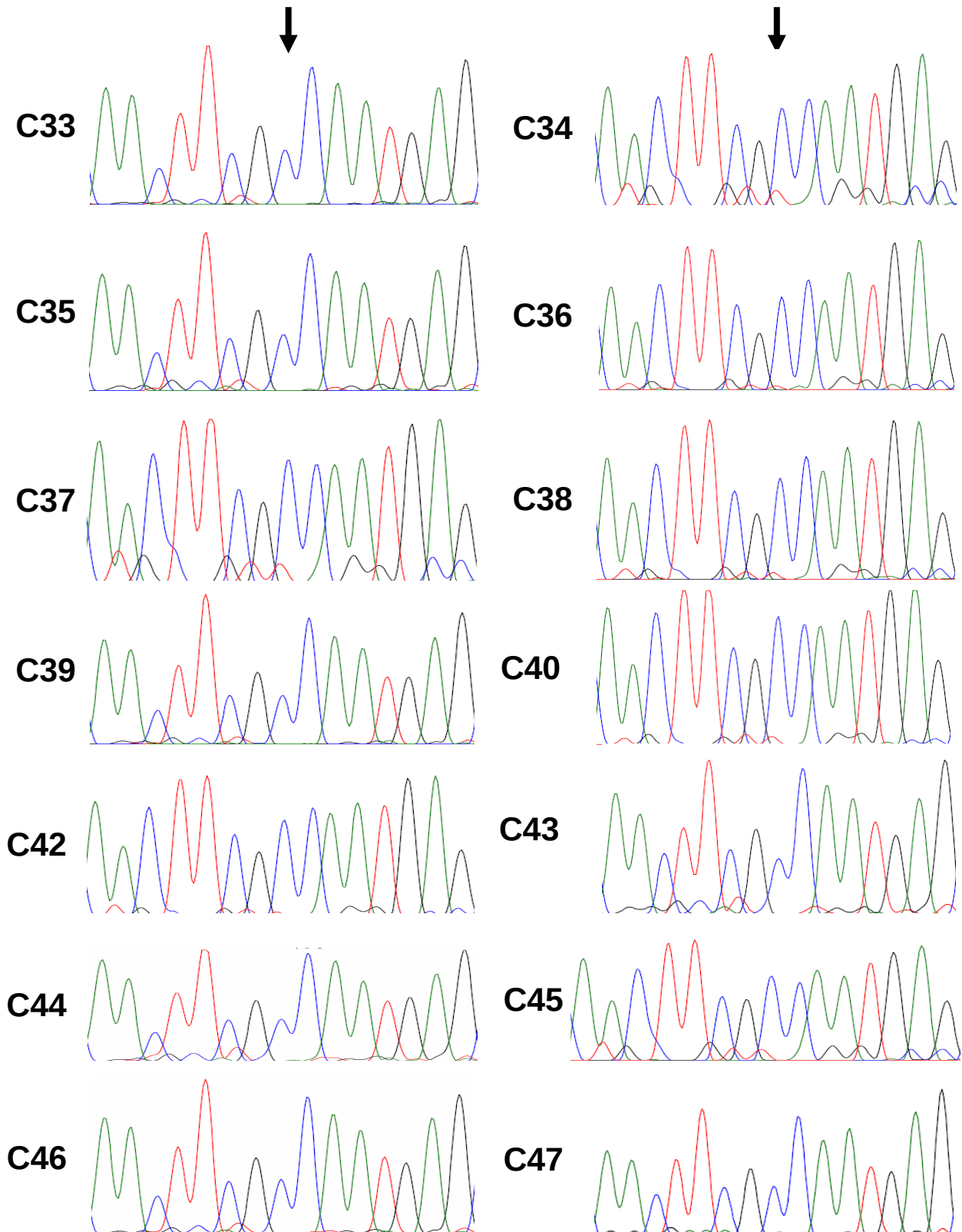
Controls for *MYO7A*, c. 1370C>T (p.A457V)



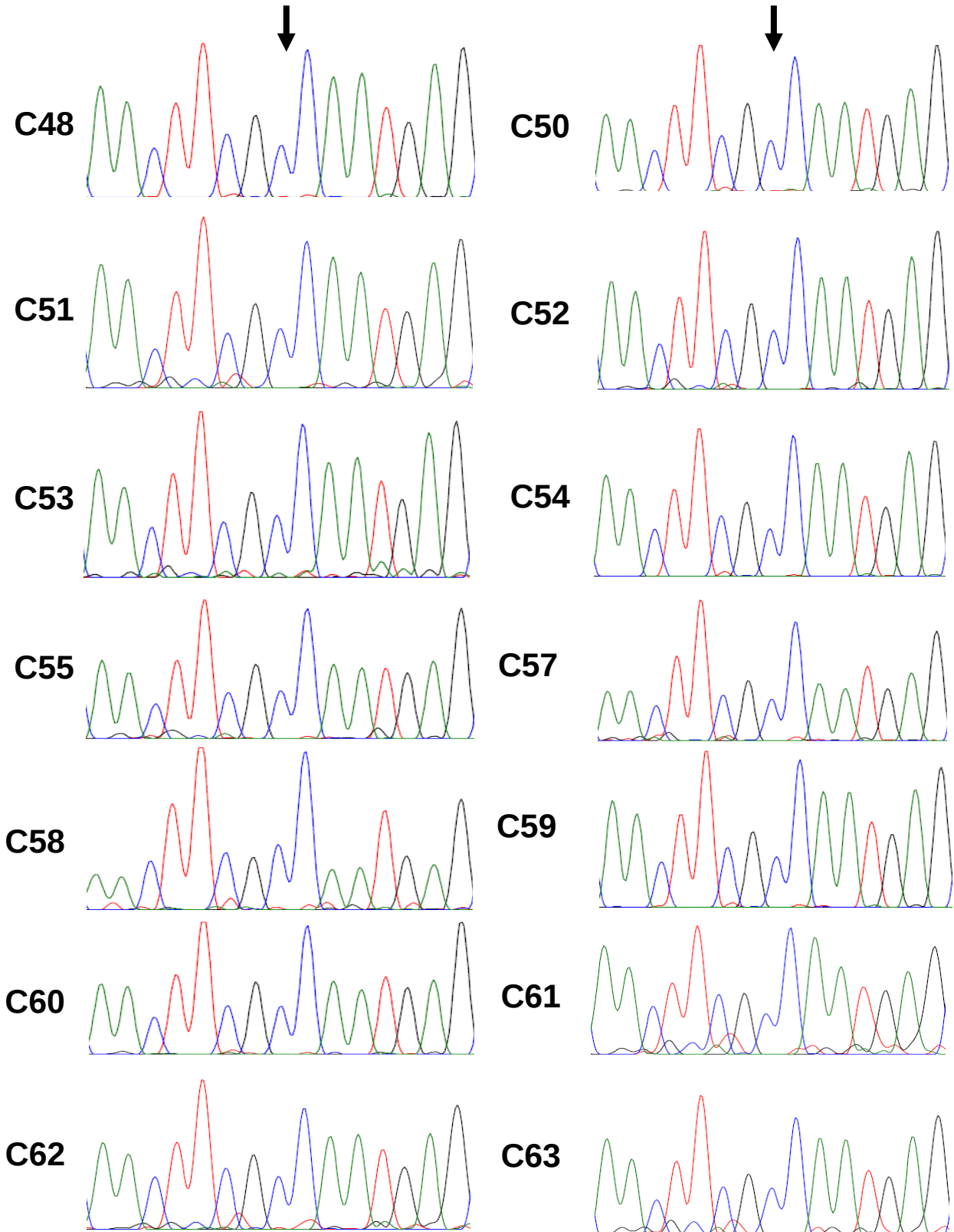
Controls for *MYO7A*, c. 1370C>T (p.A457V)



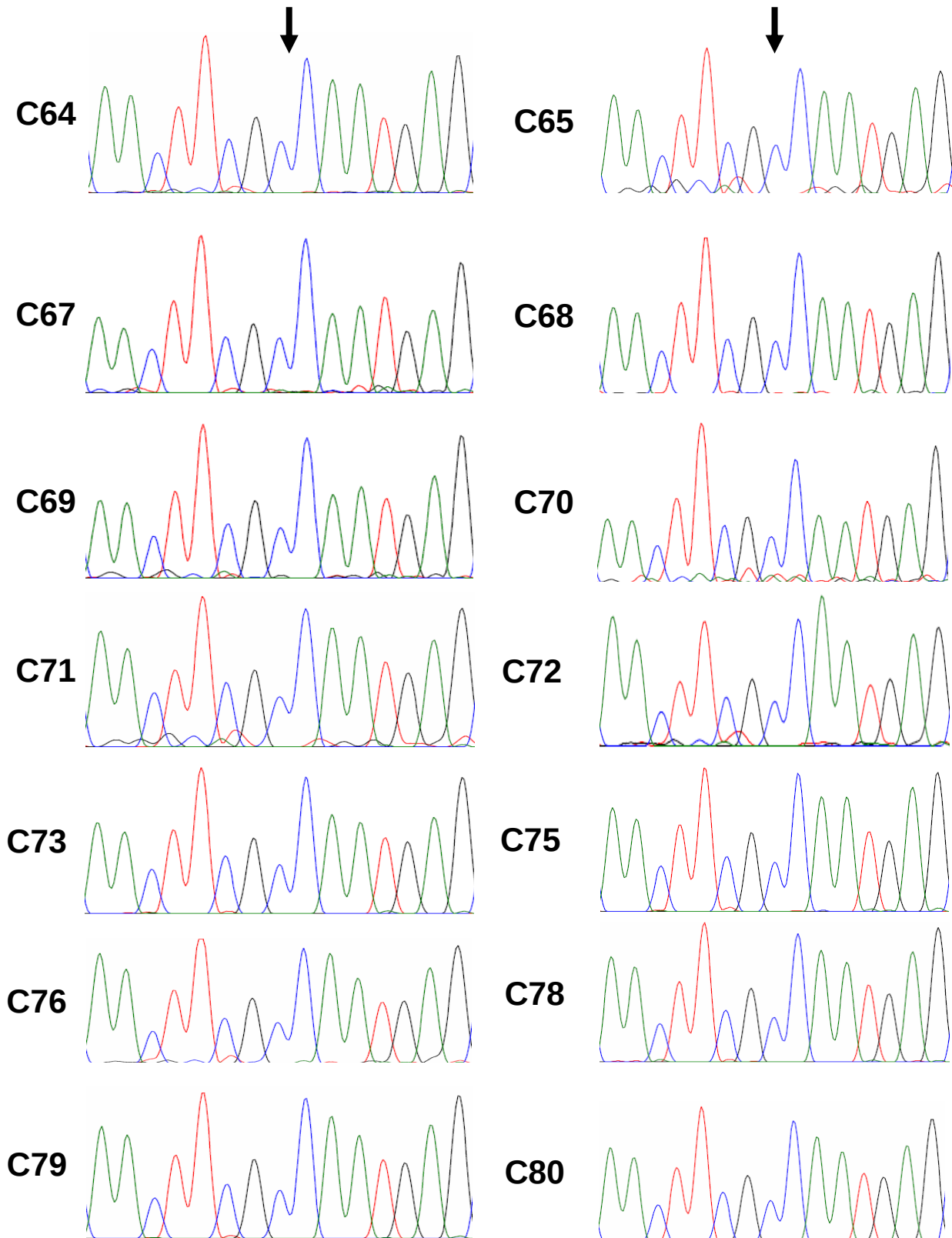
Controls for *MYO7A*, c. 1370C>T (p.A457V)



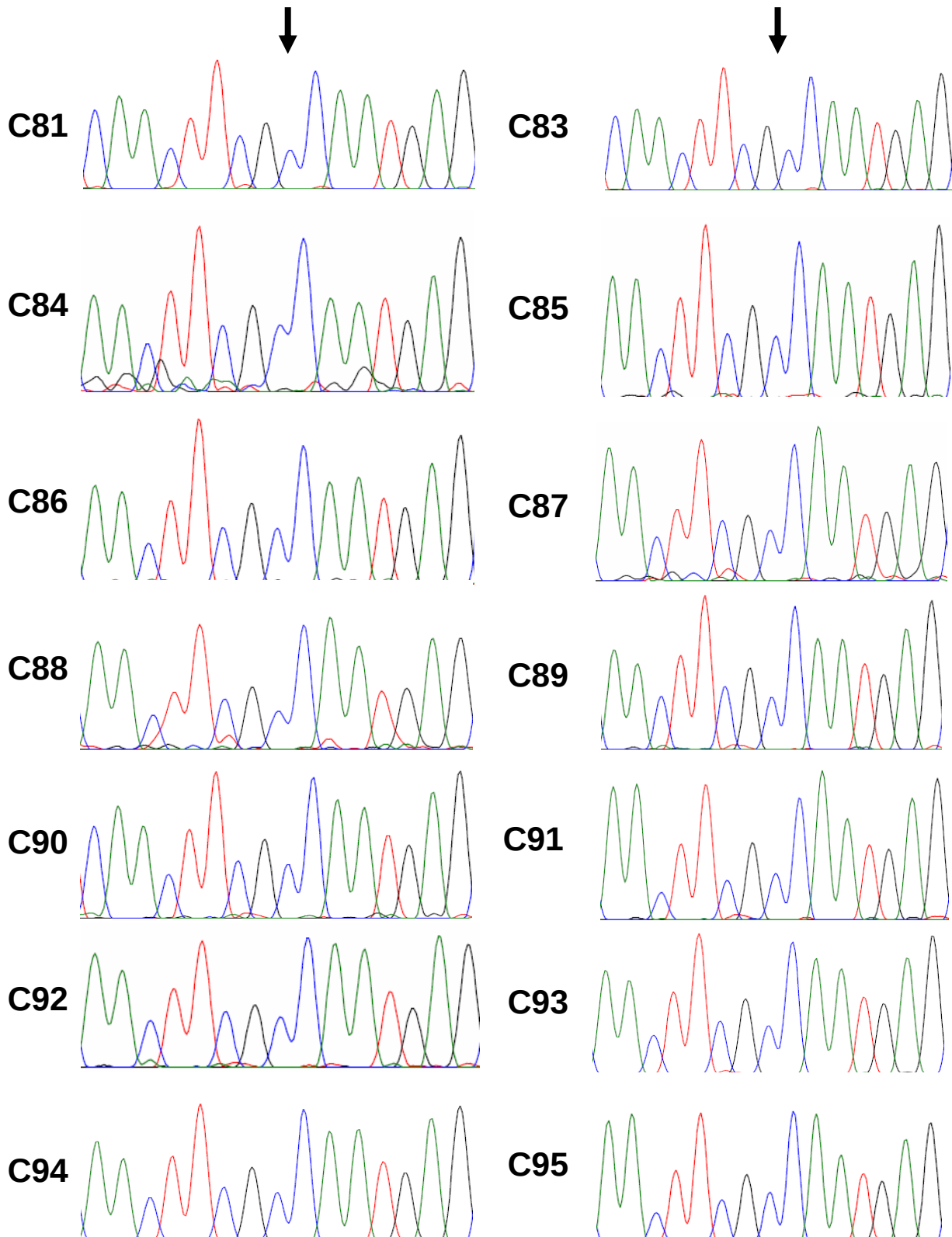
Controls for MYO7A, c. 1370C>T (p.A457V)



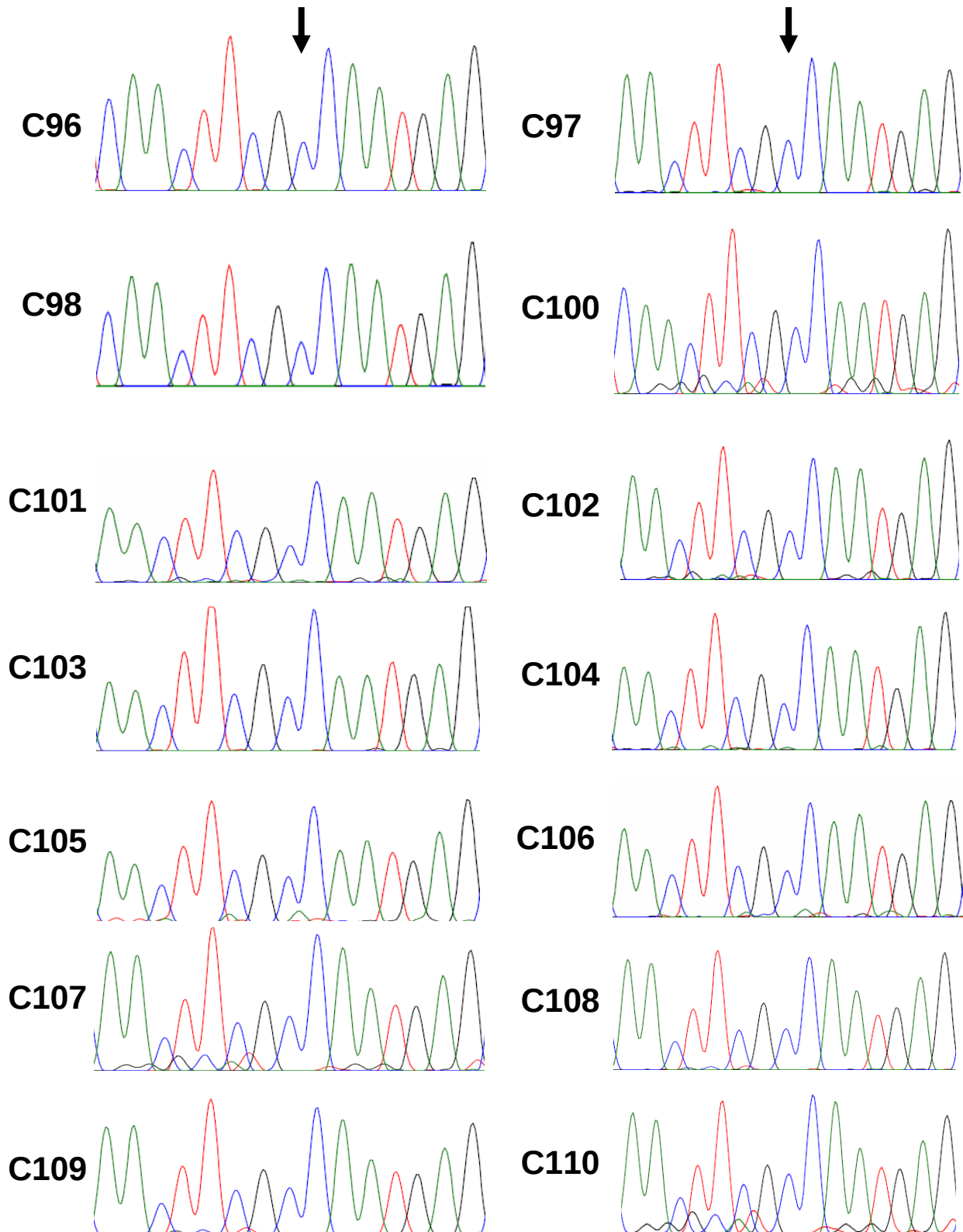
Controls for *MYO7A*, c. 1370C>T (p.A457V)



Controls for *MYO7A*, c. 1370C>T (p.A457V)



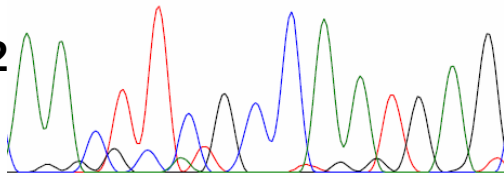
Controls for *MYO7A*, c. 1370C>T (p.A457V)



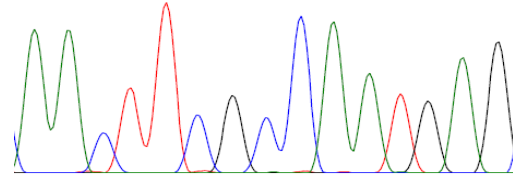
Controls for *MYO7A*, c.1370C>T (p.A457V)



C112



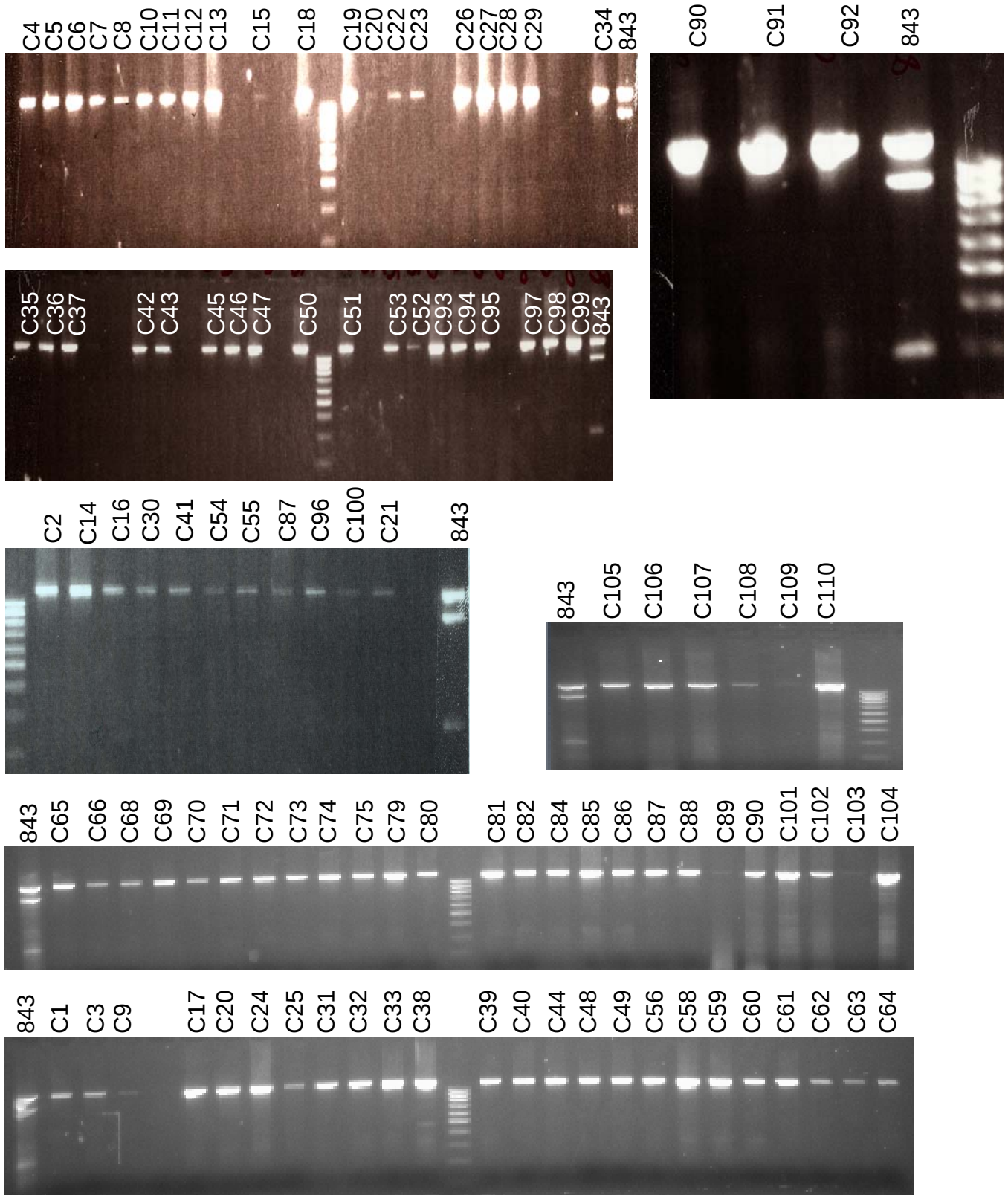
C110



p.Q815X (<i>MYO7A</i>): Genotyping of 100 French Canadian healthy control individuals
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PCR products (1199 bp) were digested with *NheI*. Presence of c.2443C>T creates a *NheI* site in the mutant, resulting in fragments of 295 and 904 bp.

Controls for c.2443 C>T/p.Q815X (MYO7A, exon 21)
by restriction digest with *Nhe*I



843: patient heterozygous for p.Q815X

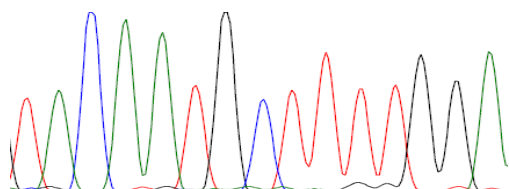
p.A123D (*USH3A*):
Genotyping of 100 French Canadian healthy control individuals

- by direct sequencing of PCR products -

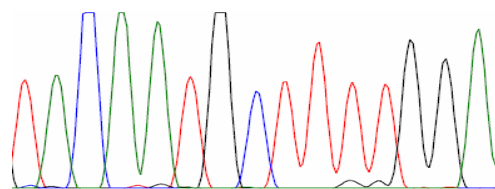
Controls for *USH3A*, c.368C>T (p.A123D)



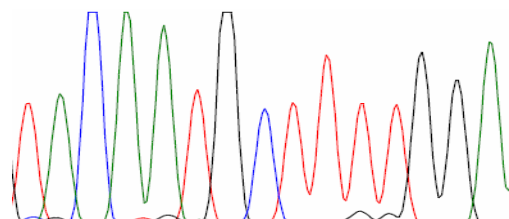
C1



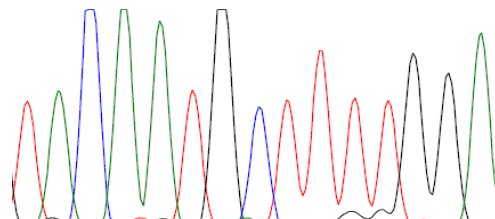
C2



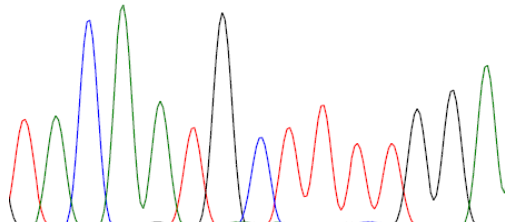
C3



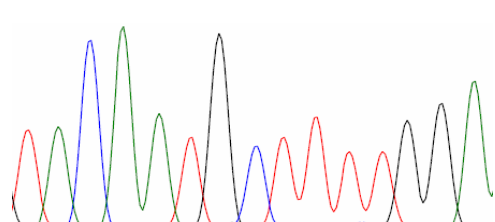
C5



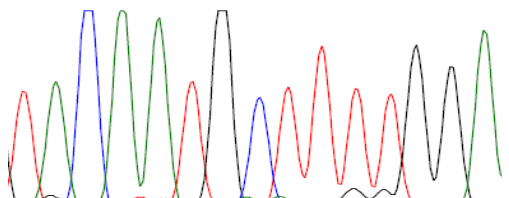
C6



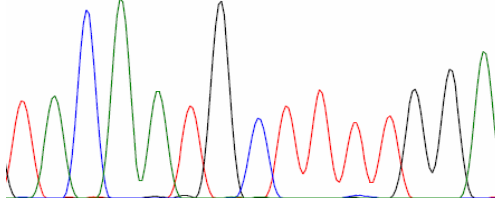
C6



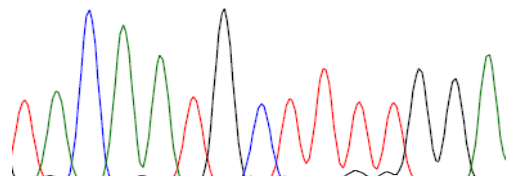
C7



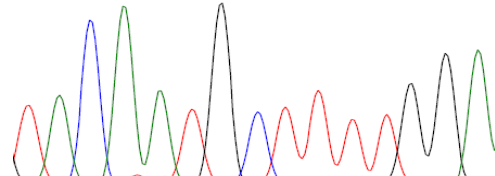
C8



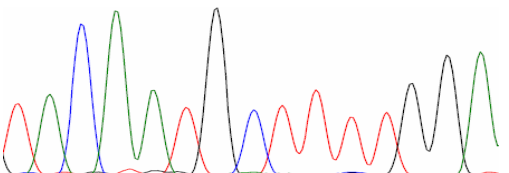
C9



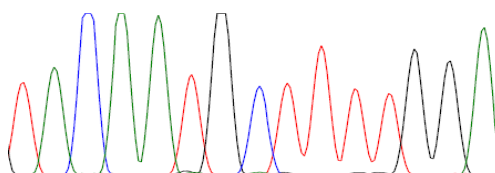
C10



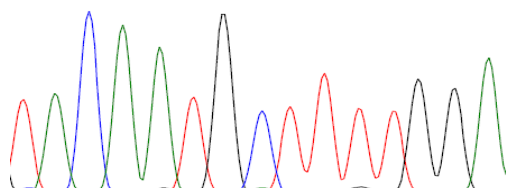
C11



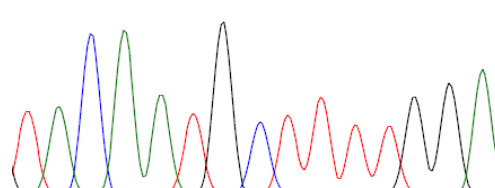
C13



C14



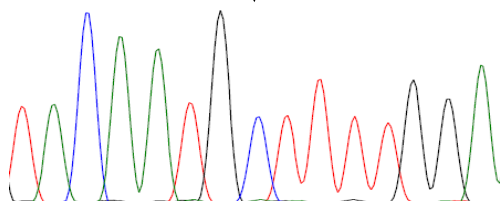
C15



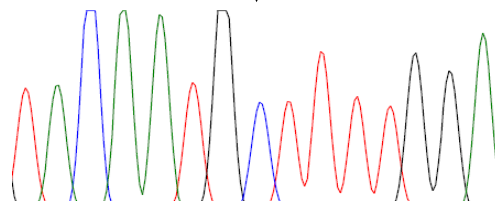
Controls for *USH3A*, c.368C>T (p.A123D)



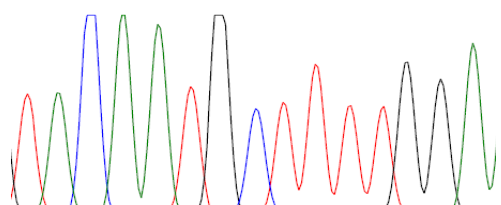
C16



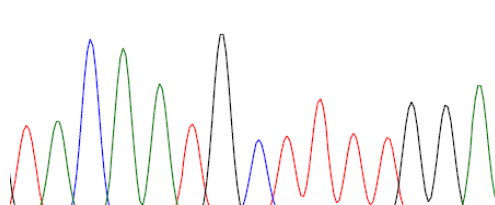
C17



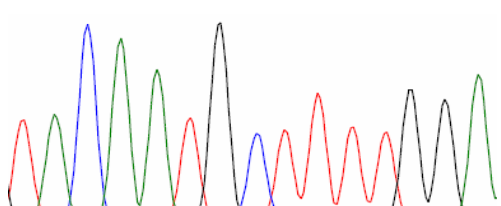
C18



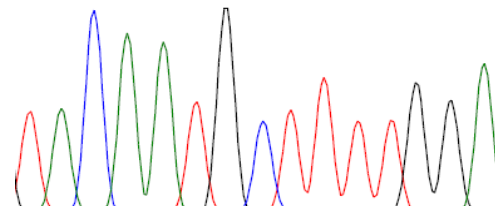
C19



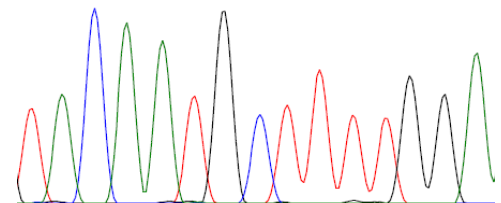
C20



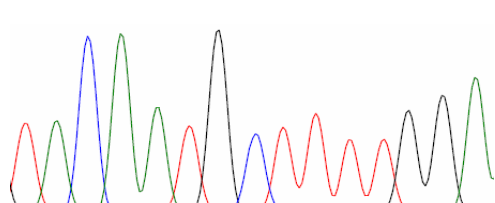
C21



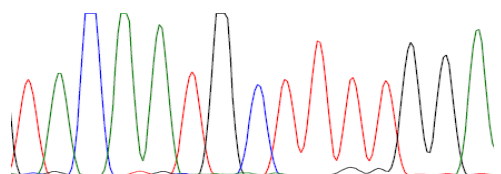
C23



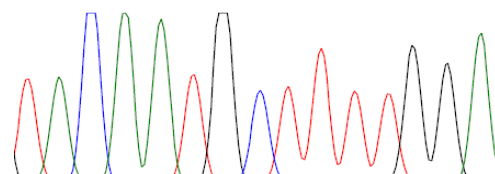
C24



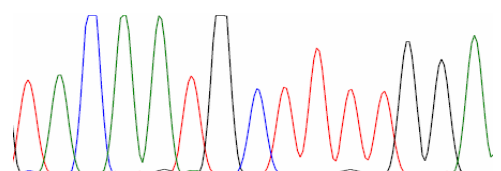
C25



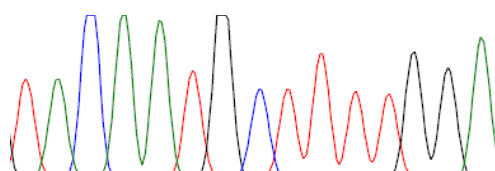
C26



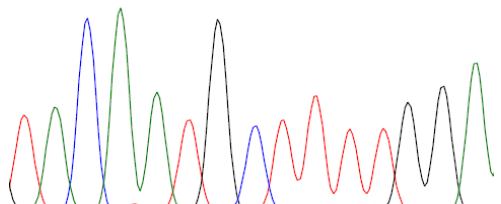
C27



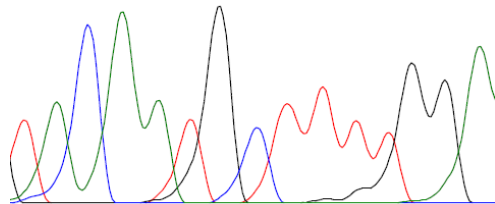
C28



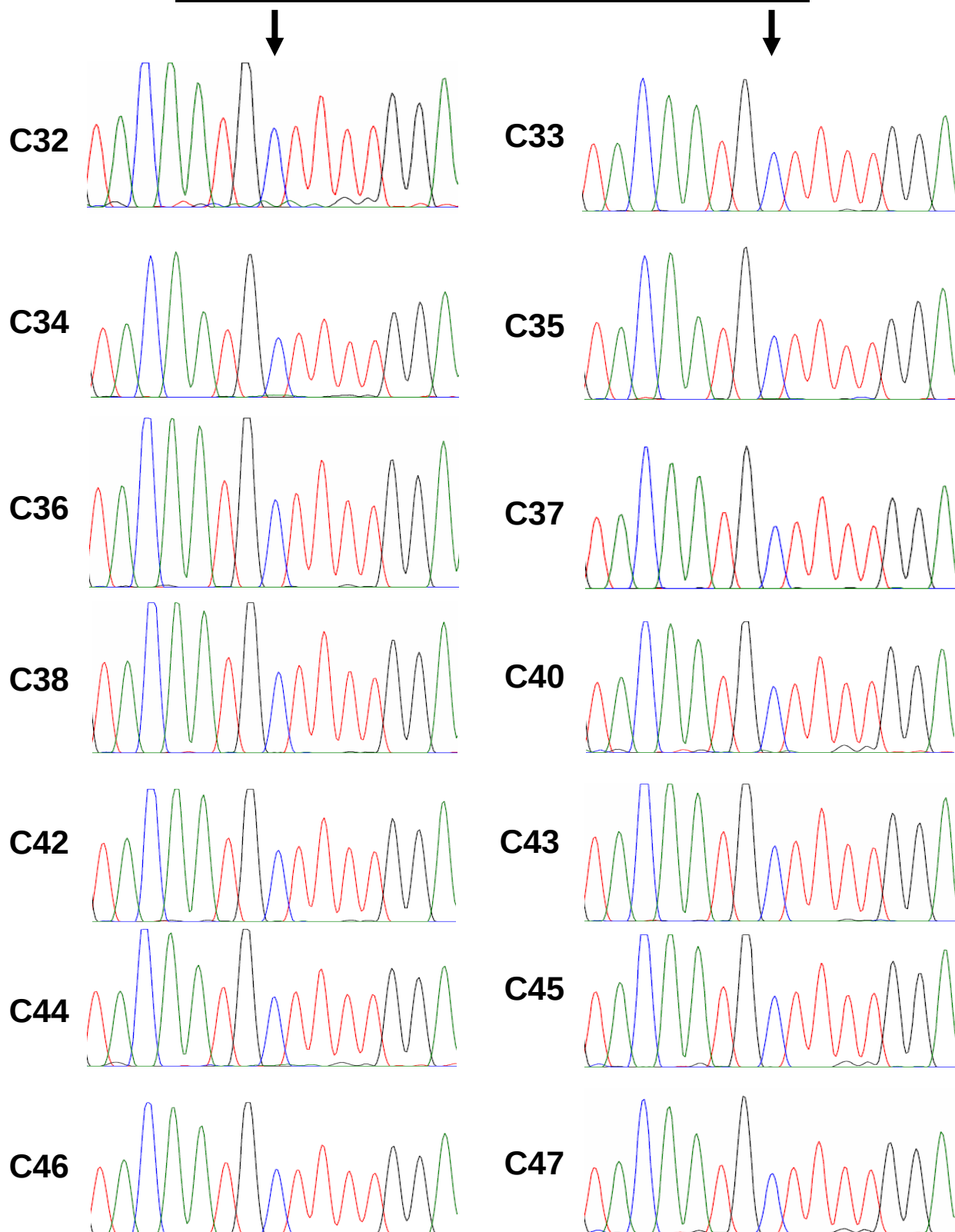
C29



C31



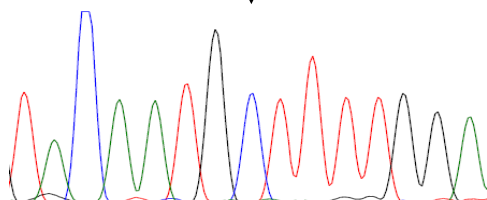
Controls for *USH3A*, c.368C>T (p.A123D)



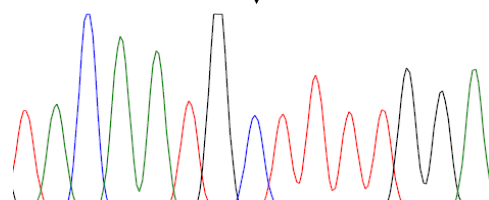
Controls for *USH3A*, c.368C>T (p.A123D)



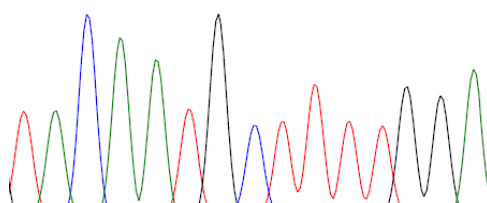
C48



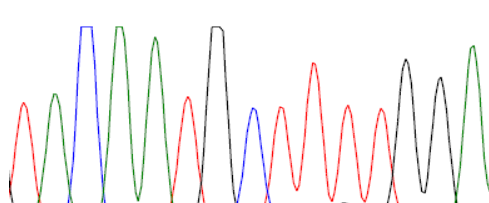
C49



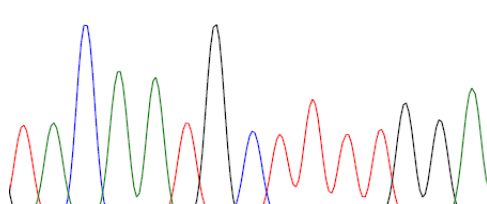
C50



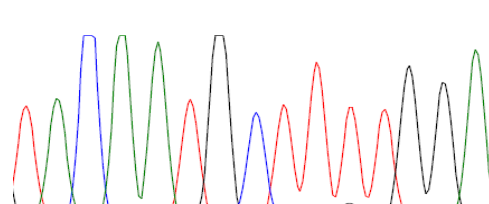
C51



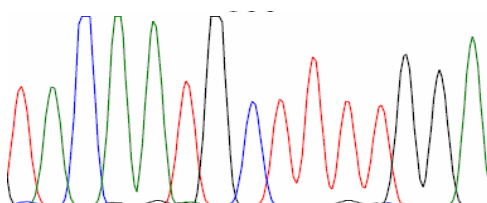
C53



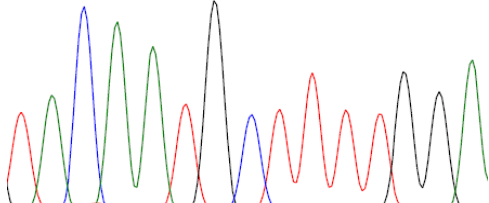
C54



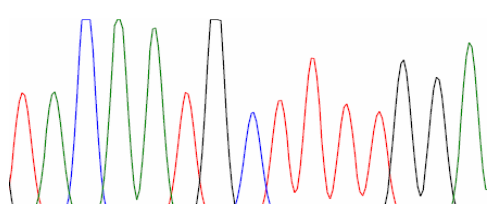
C55



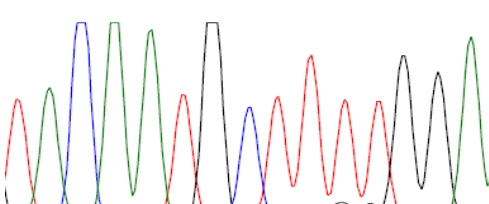
C56



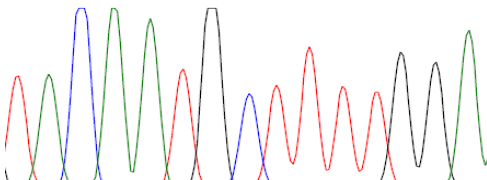
C57



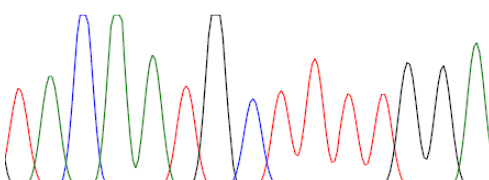
C58



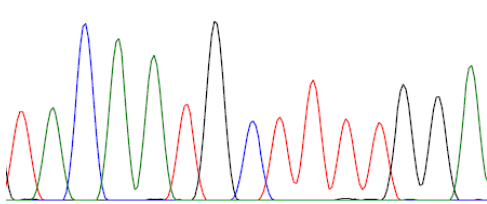
C59



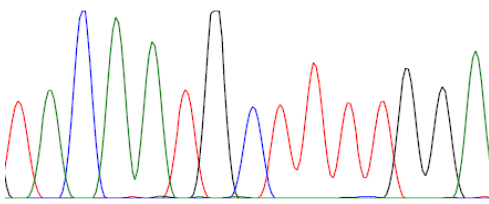
C60



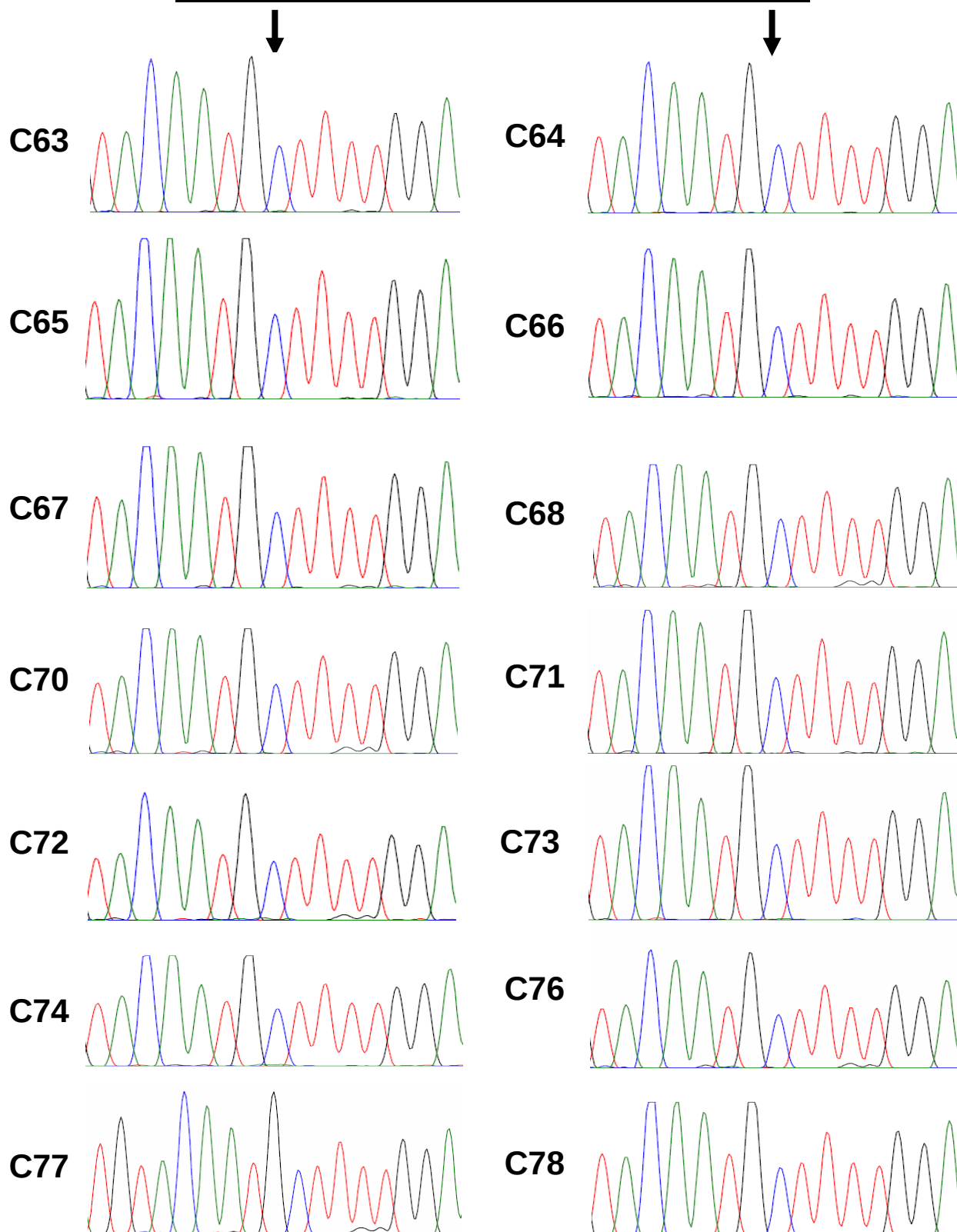
C61



C62



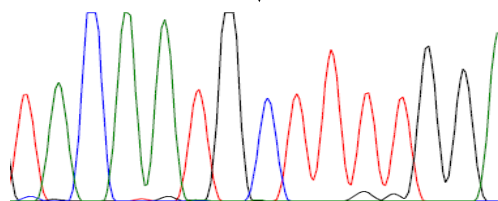
Controls for *USH3A*, c.368C>T (p.A123D)



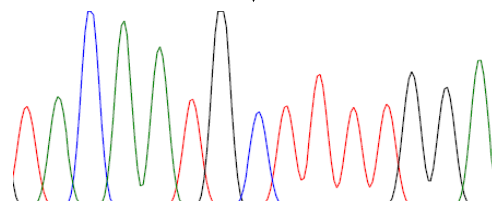
Controls for *USH3A*, c.368C>T (p.A123D)



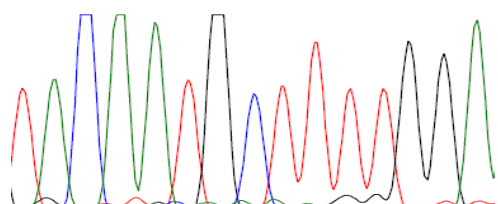
C79



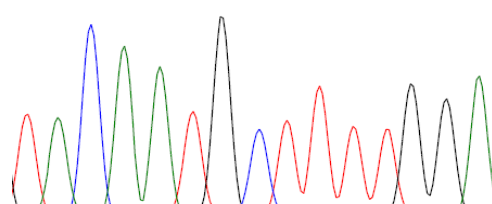
C80



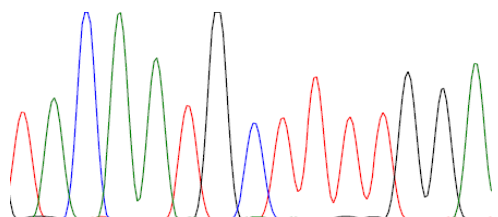
C81



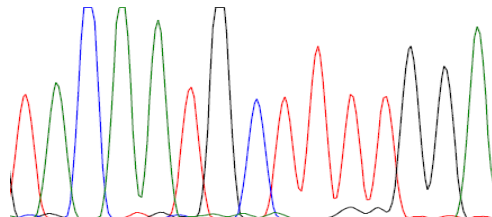
C82



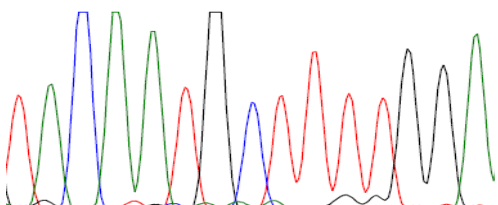
C83



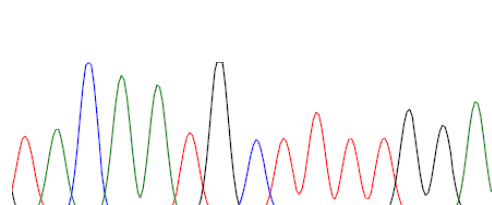
C84



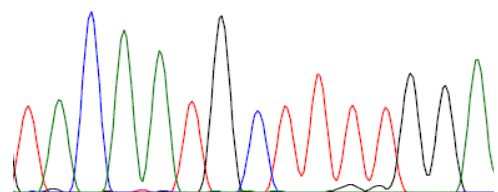
C85



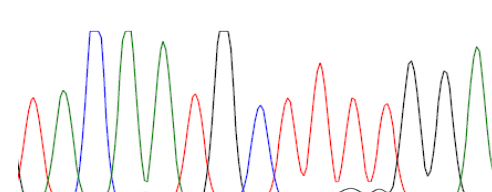
C86



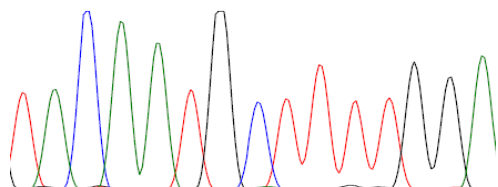
C87



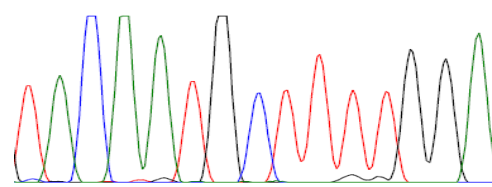
C88



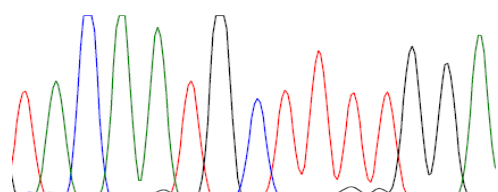
C89



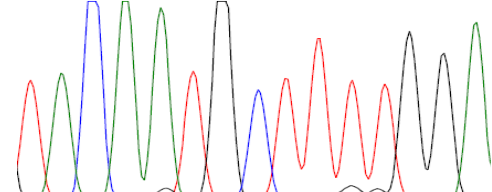
C90



C91



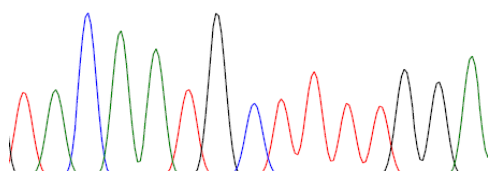
C92



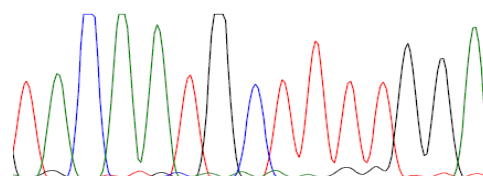
Controls for *USH3A*, c.368C>T (p.A123D)



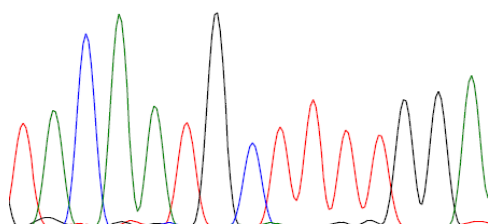
C93



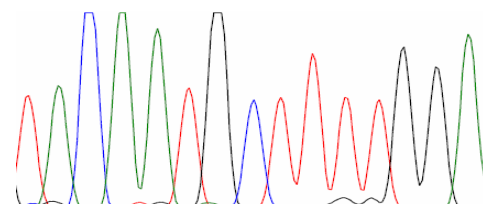
C94



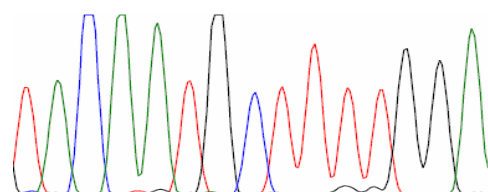
C95



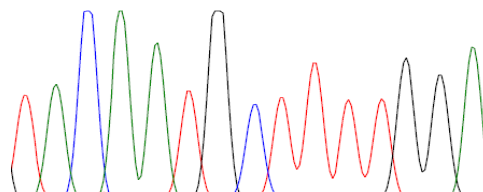
C97



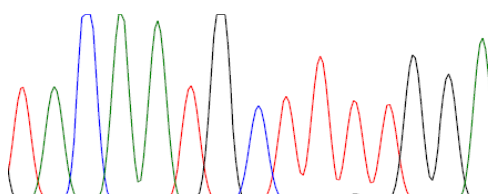
C98



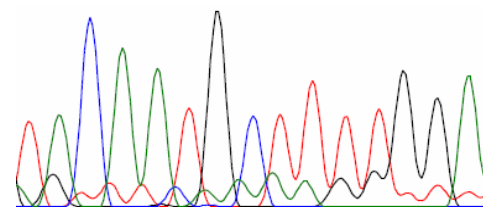
C99



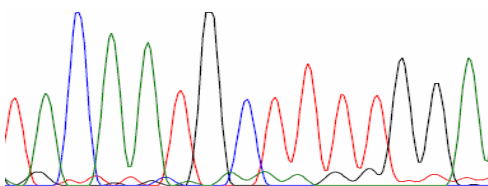
C100



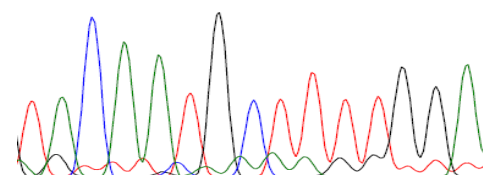
Q11



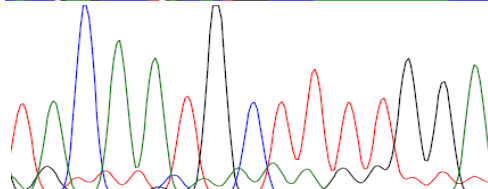
Q14



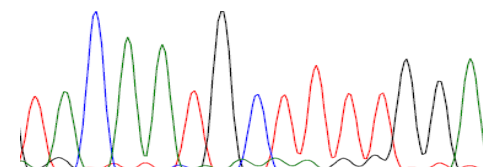
Q20



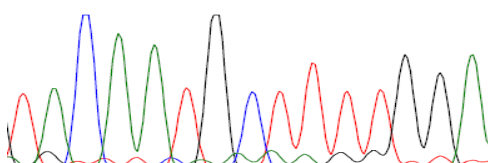
Q27



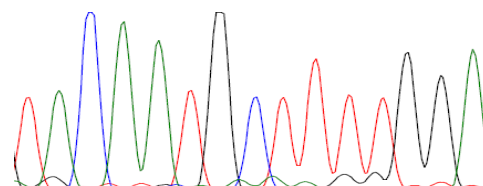
Q36



Q41



Q49



Controls for *USH3A*, c.368C>T (p.A123D)

