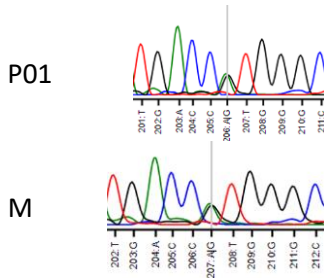
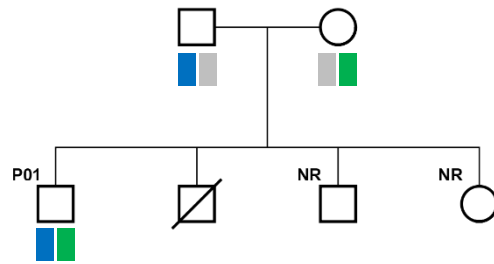


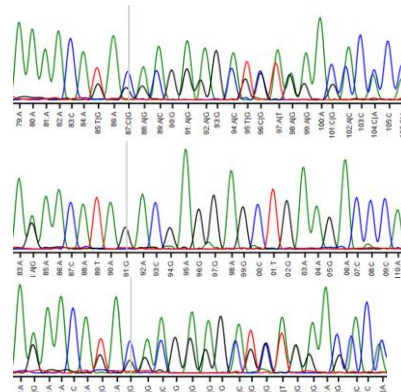
### RYR1-KDS Genotypes P01 and Relatives



Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Proband, Mother and Father).  
 Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Father)  
*RYR1:c.14524G>A*

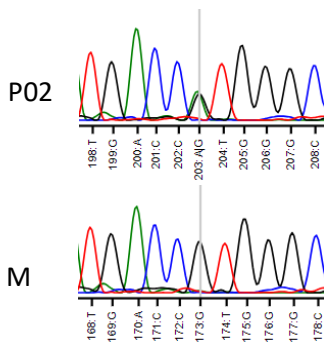
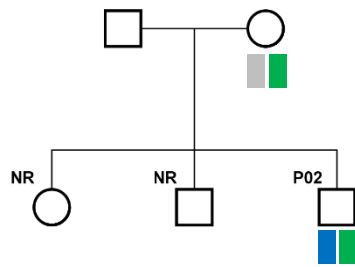
Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Proband, Mother and Father).

*RYR1:c.10348-6C>G*

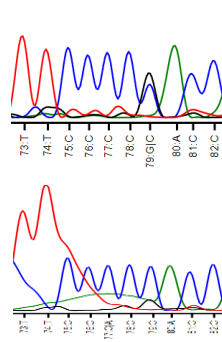


*RYR1:c.8342\_8343delTA*

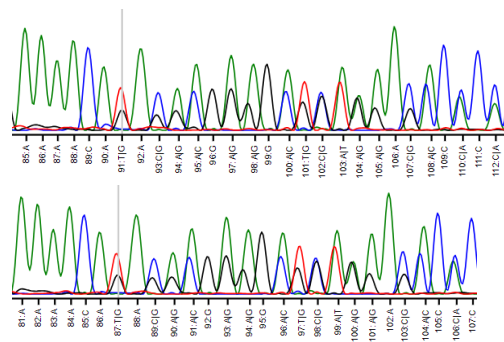
### P02 and Relative



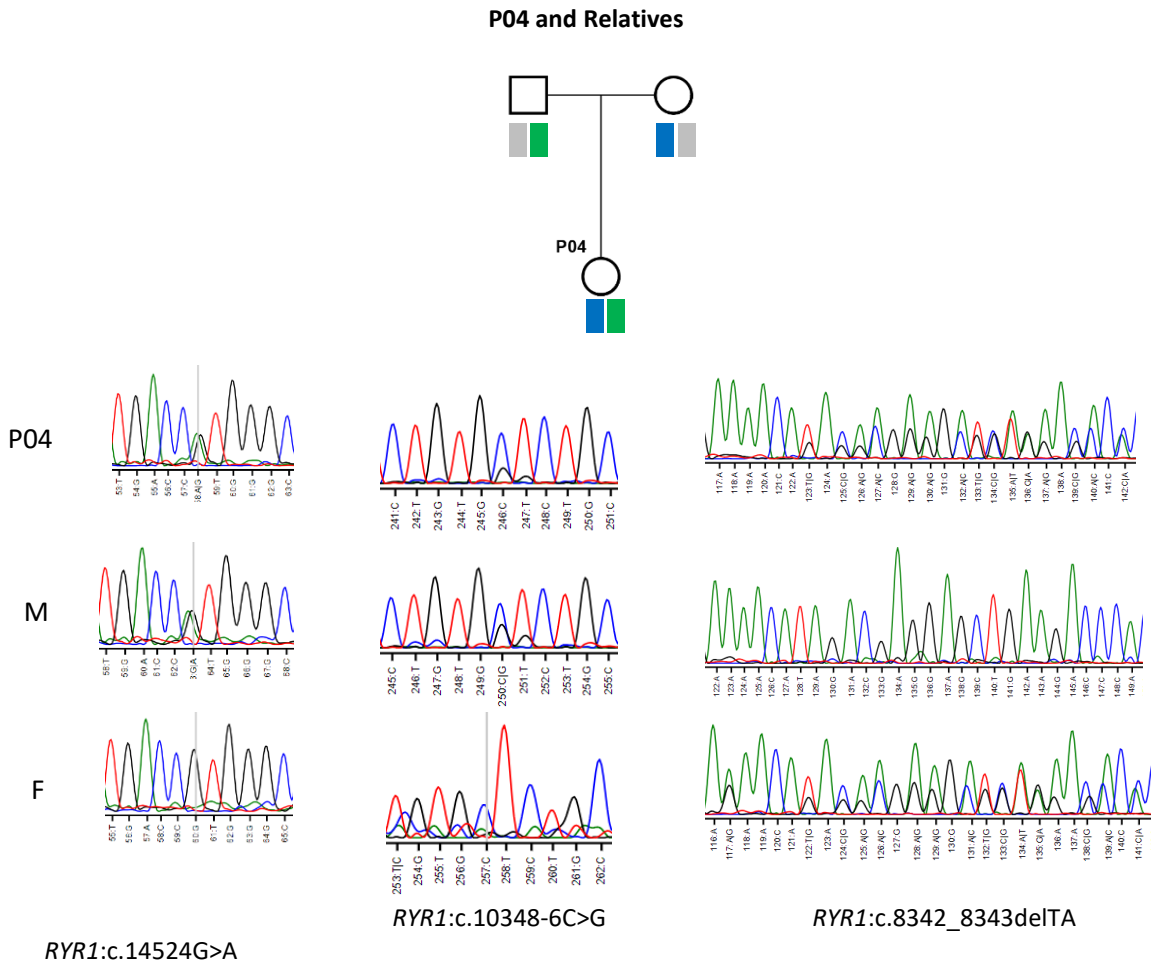
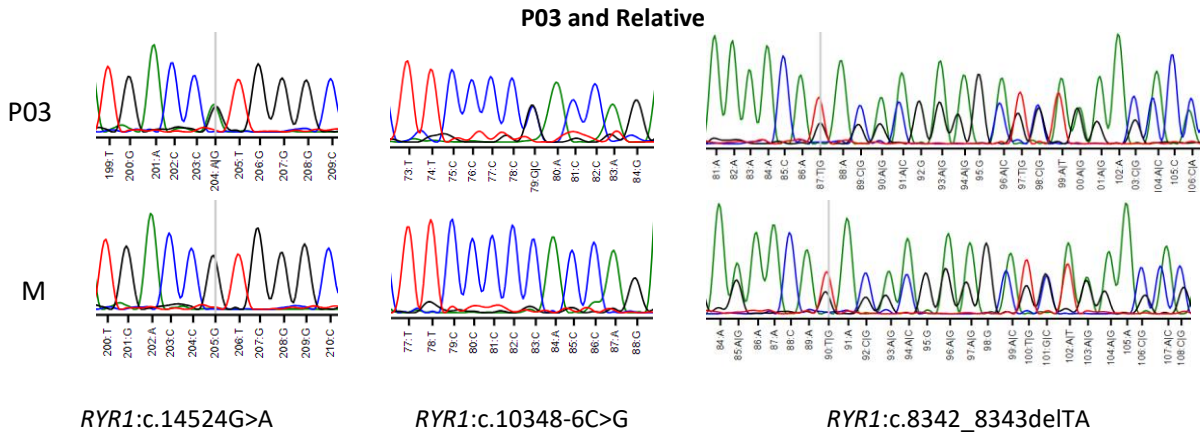
*RYR1:c.14524G>A*



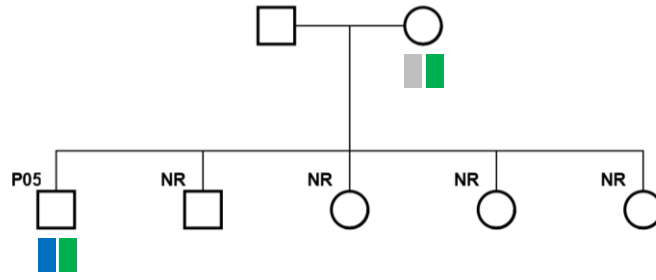
*RYR1:c.10348-6C>G*



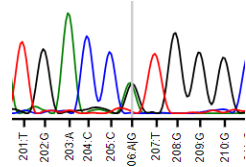
*RYR1:c.8342\_8343delTA*



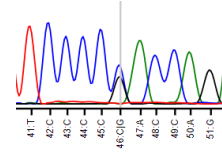
**P05 and Relative**



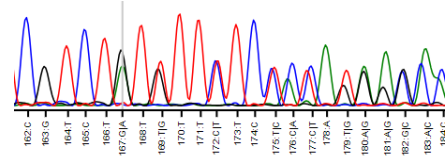
P05



*RYR1*:c.14524G>A

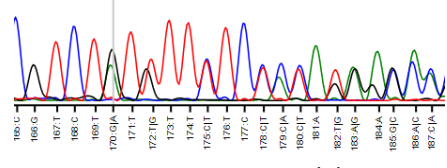
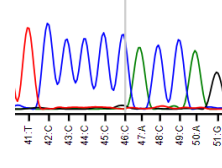
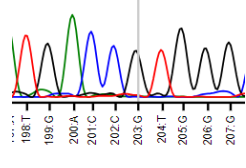


*RYR1*:c.10348-6C>G

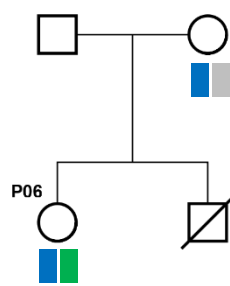


*RYR1*:c.8342\_8343delTA

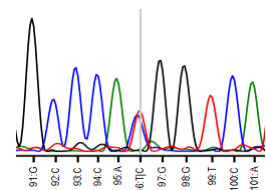
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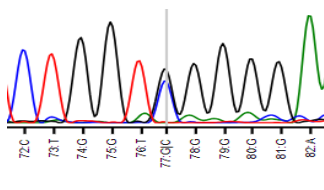
**P06 and Relative**



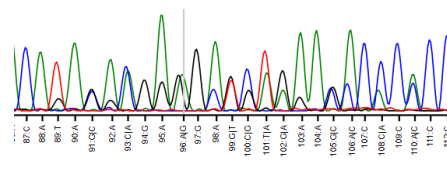
P06



*RYR1*:c.14524G>A

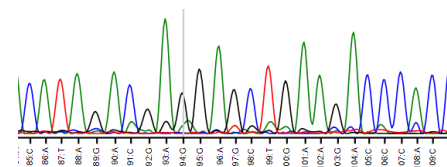
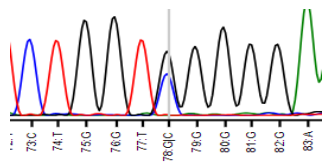
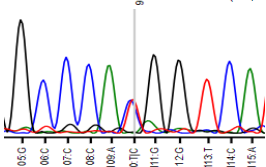


*RYR1*:c.10348-6C>G

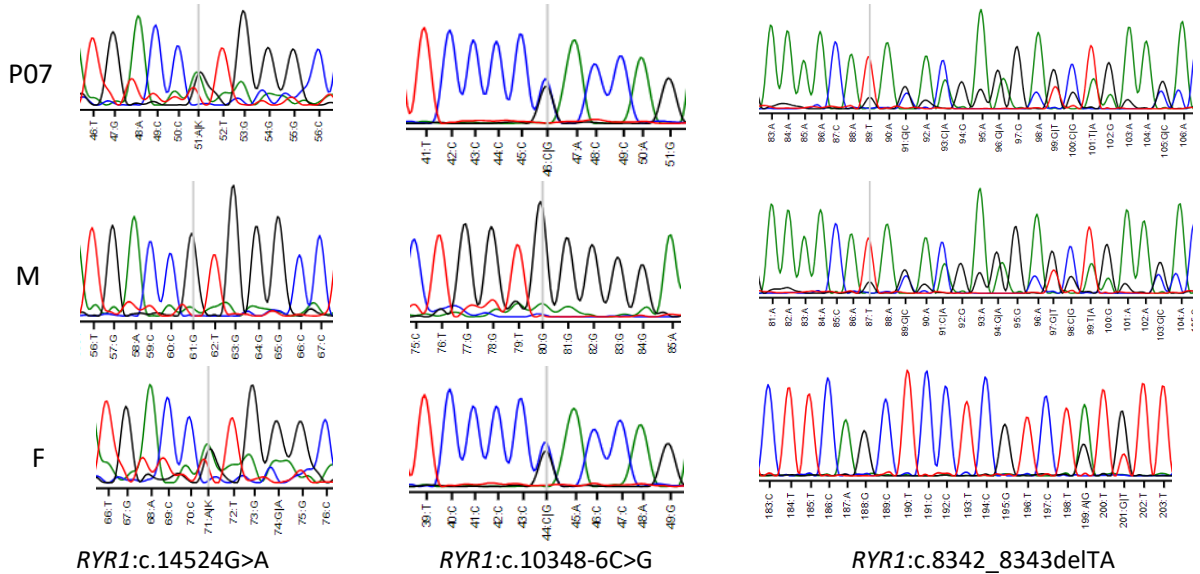
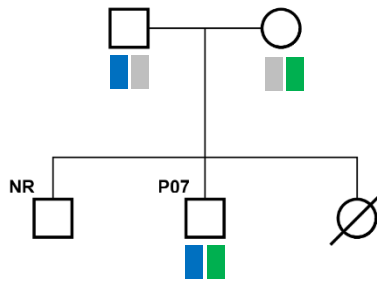


*RYR1*:c.8342\_8343delTA

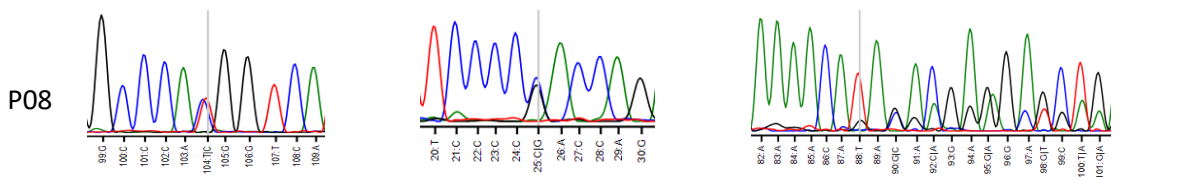
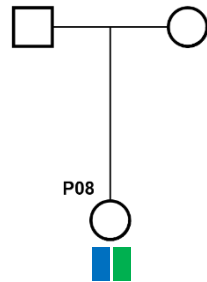
M



**P07 and Relatives**



**P08 and Relative**

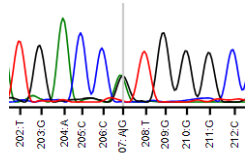
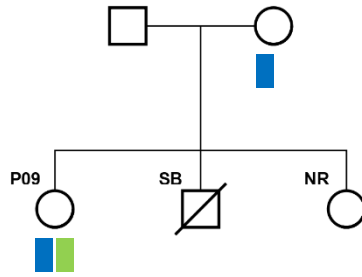


**RYR1:c.14524G>A**  
 Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Mother and Father)

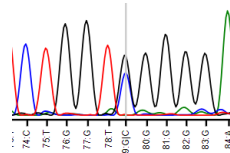
**RYR1:c.10348-6C>G**  
 Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Mother and Father)

**RYR1:c.8342\_8343delTA**  
 Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Mother and Father)

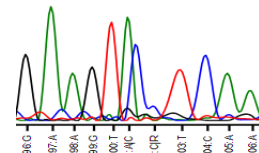
**P09 and Relative**



*RYR1:c.14524G>A*



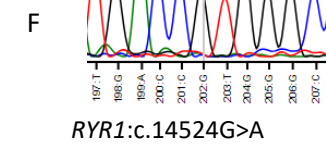
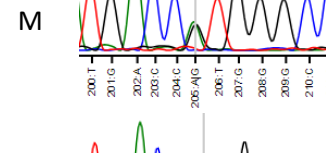
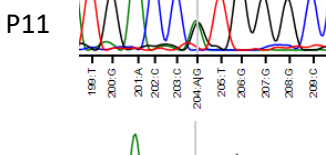
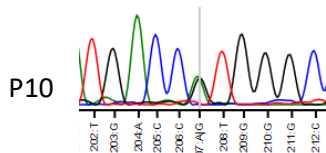
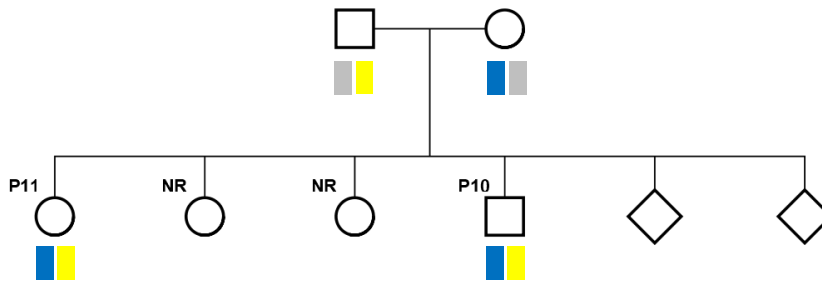
*RYR1:c.10348-6C>G*



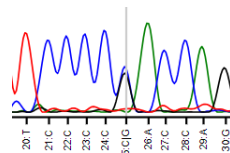
*RYR1:12625-1G>A*

Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Mother)

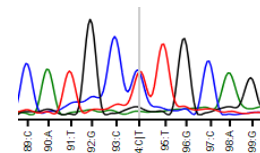
**P10 and P11 and Relatives**



*RYR1:c.14524G>A*



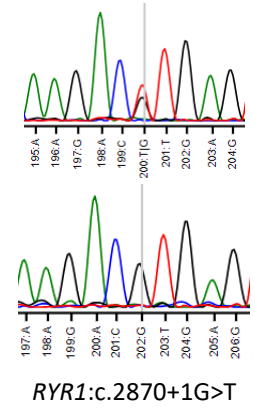
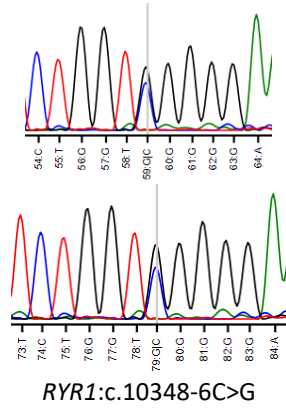
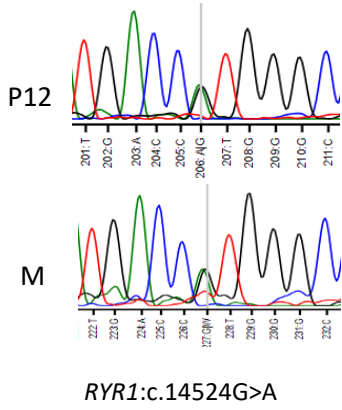
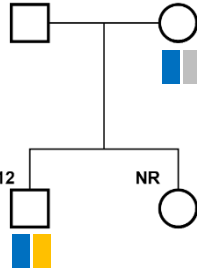
*RYR1:c.10348-6C>G*



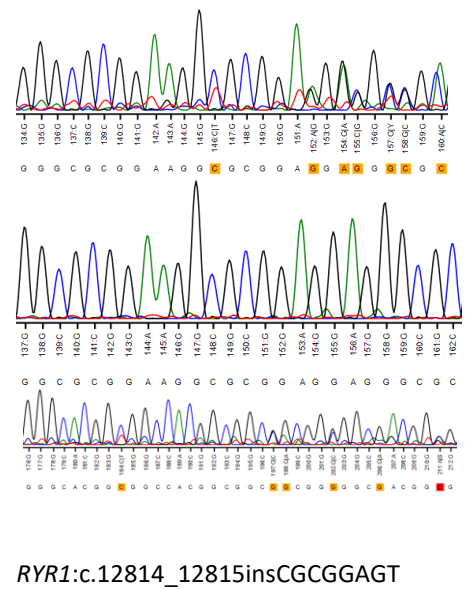
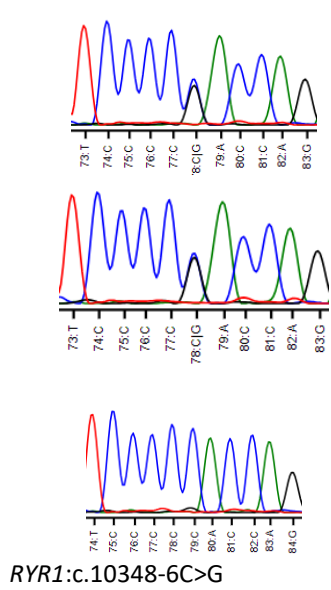
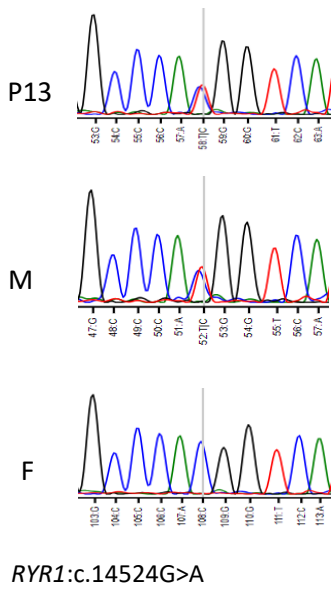
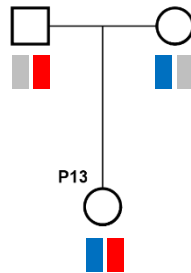
*RYR1:c.6797-1G>T*

Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Father)

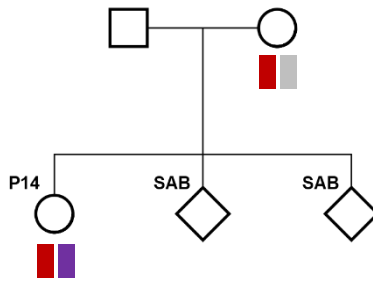
**P12 and Relative**



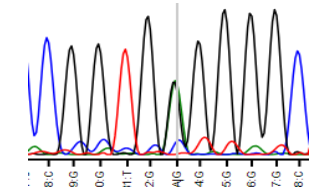
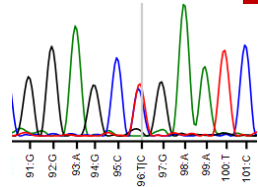
**P13 and Relatives**



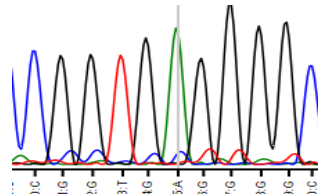
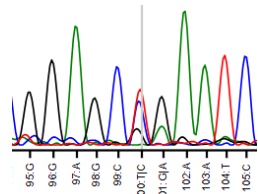
**P14 and Relative**



P14



M

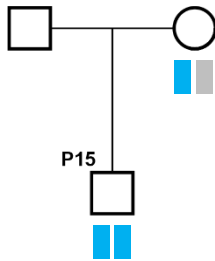


*RYR1:c.10280C>T*

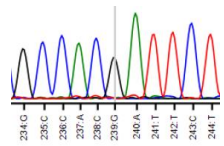
*RYR1:c.2682+4A>G*

**STAC3-KDS Genotypes**

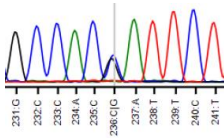
**P15 and Relative**



P15

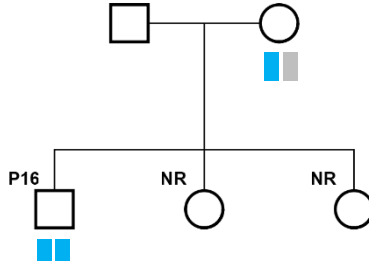


M

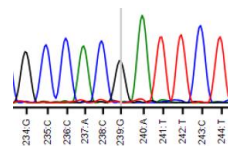


*STAC3:c.851G>C*

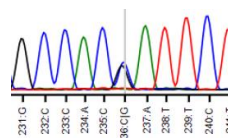
**P16 and Relative**



P16

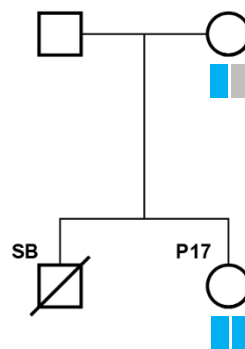


M

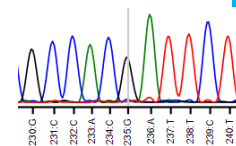


*STAC3:c.851G>C*

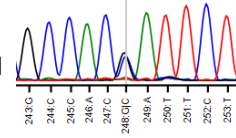
**P17 and Relative**



P17

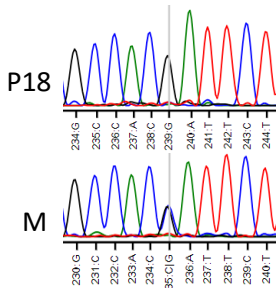
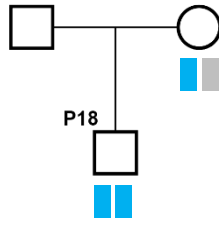


M



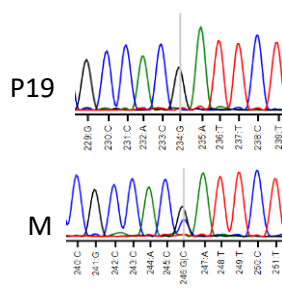
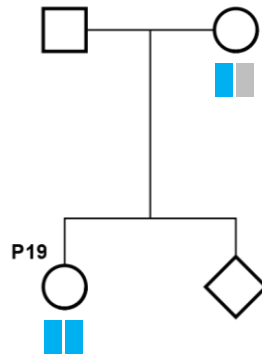
*STAC3:c.851G>C*

**P18 and Relative**



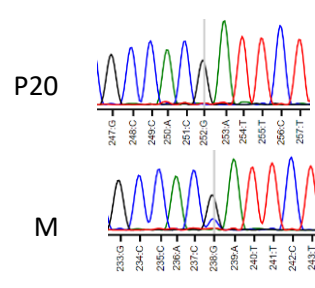
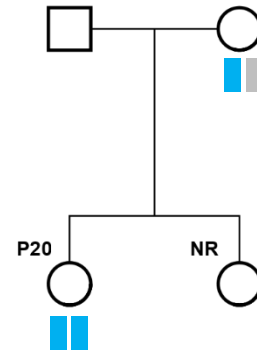
STAC3:c.851G>C

**P19 and Relative**



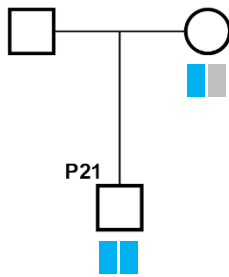
STAC3:c.851G>C

**P20 and Relative**



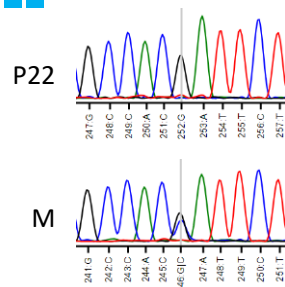
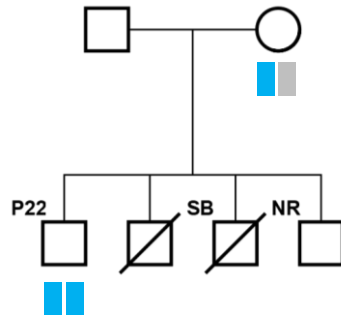
STAC3:c.851G>C

**P21 and Relative**



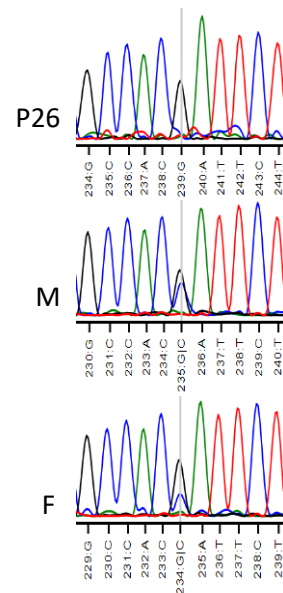
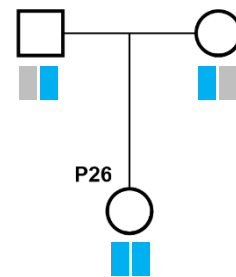
Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Proband and Mother)

**P22 and Relative**

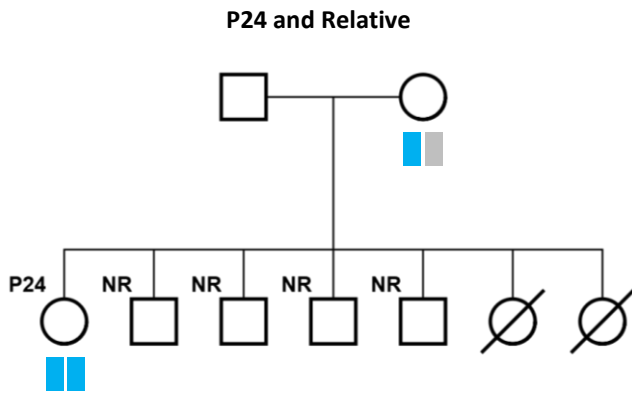
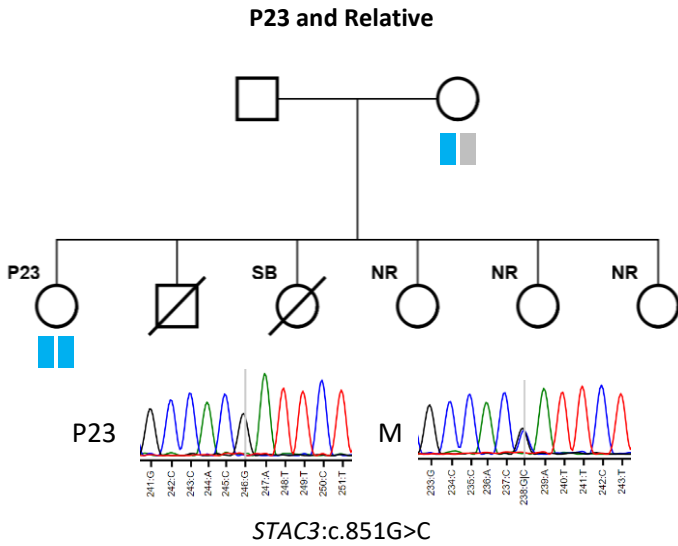


STAC3:c.851G>C

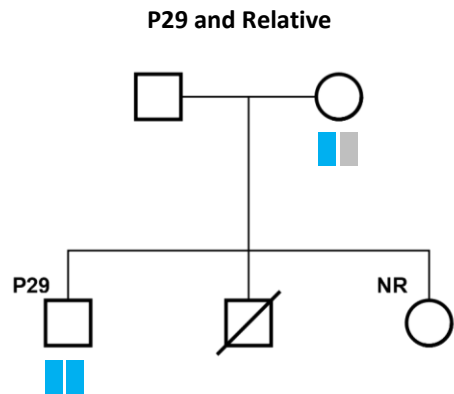
**P26 and Relatives**



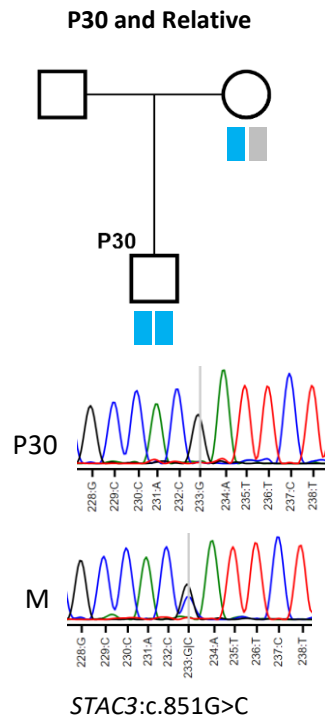
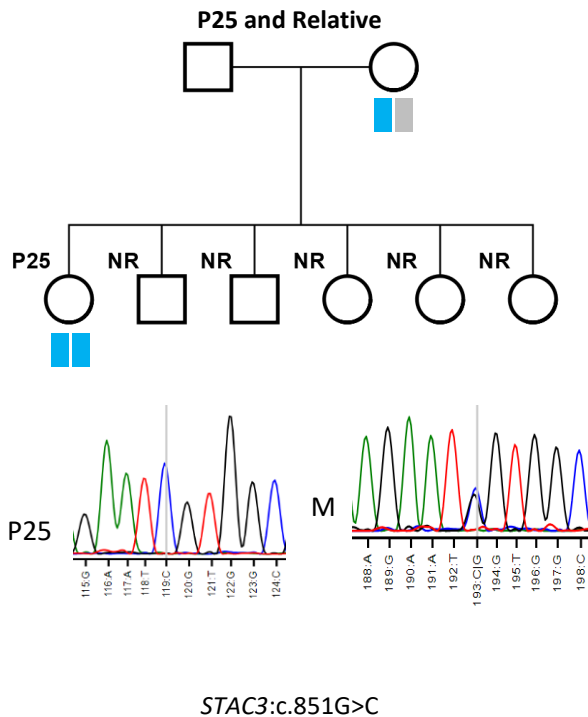
STAC3:c.851G>C



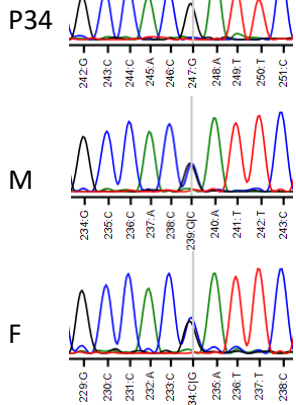
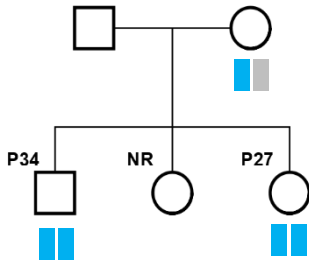
Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Proband and Mother)



Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Proband and Mother)

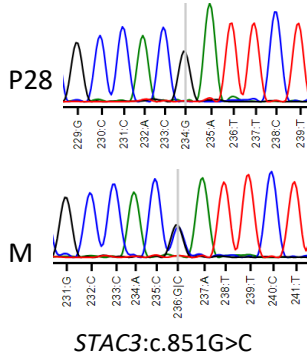
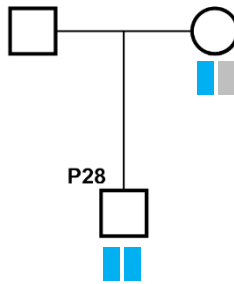


**P34 and P27 and Relatives**



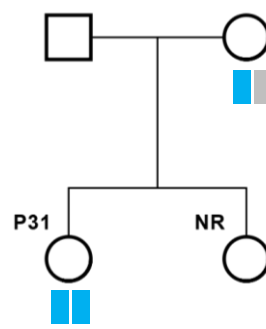
Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (P27)  
**STAC3:c.851G>C**

**P28 and Relative**



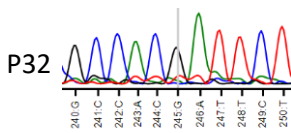
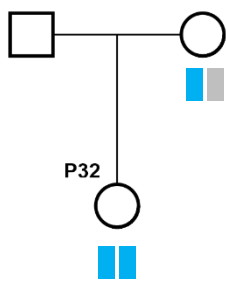
**STAC3:c.851G>C**

**P31 and Relative**



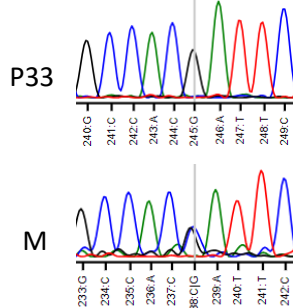
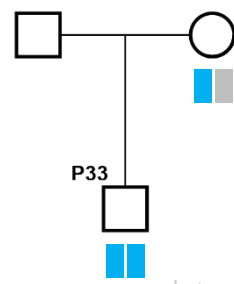
Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Proband and Mother)

**P32 and Relative**



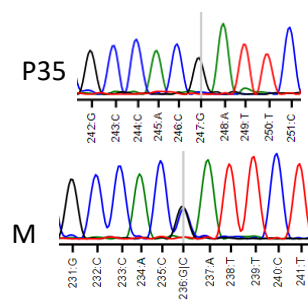
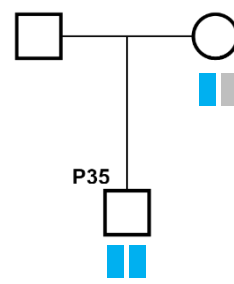
Detected on WES, no Sanger confirmation done due to lack of whole blood/gDNA samples (Mother).  
**STAC3:c.851G>C**

**P33 and Relative**

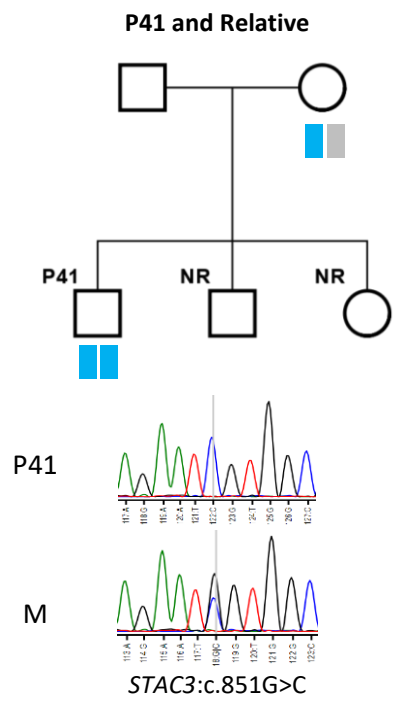
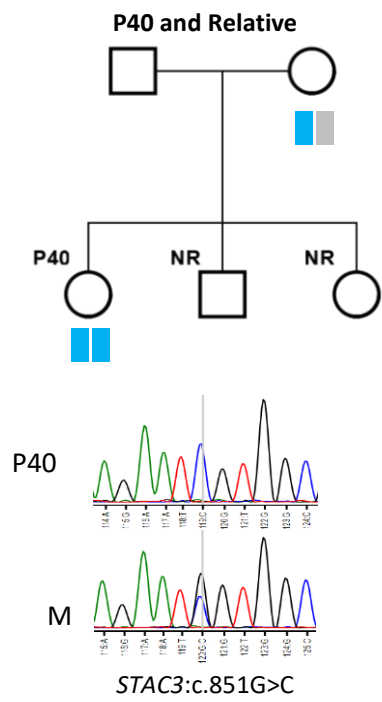
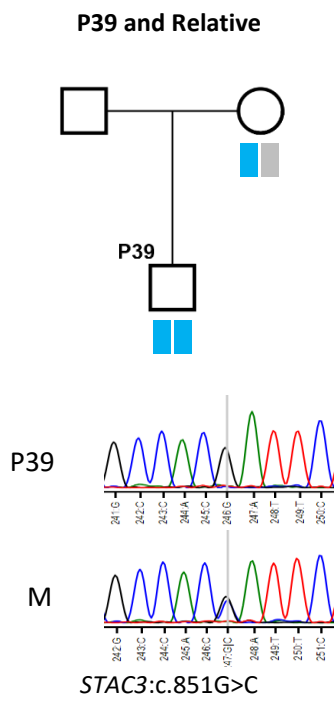
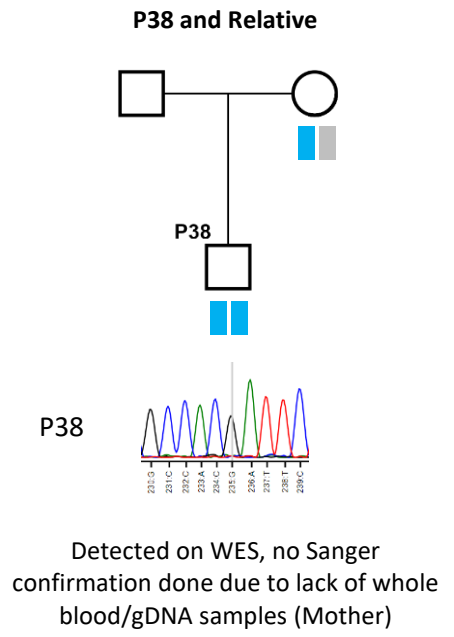
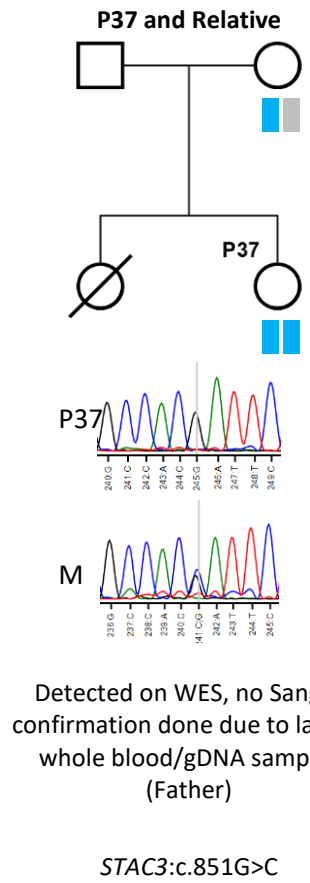
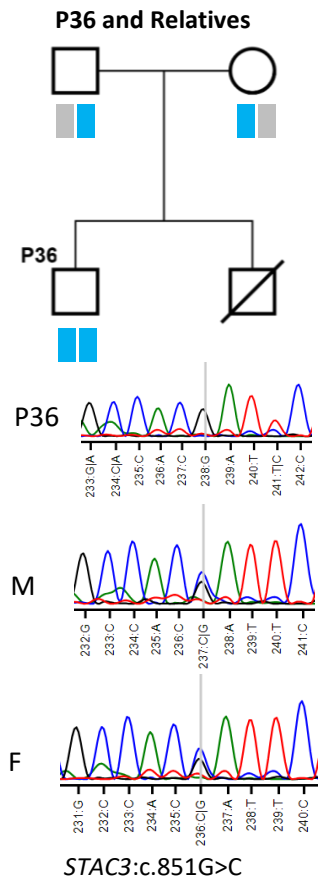


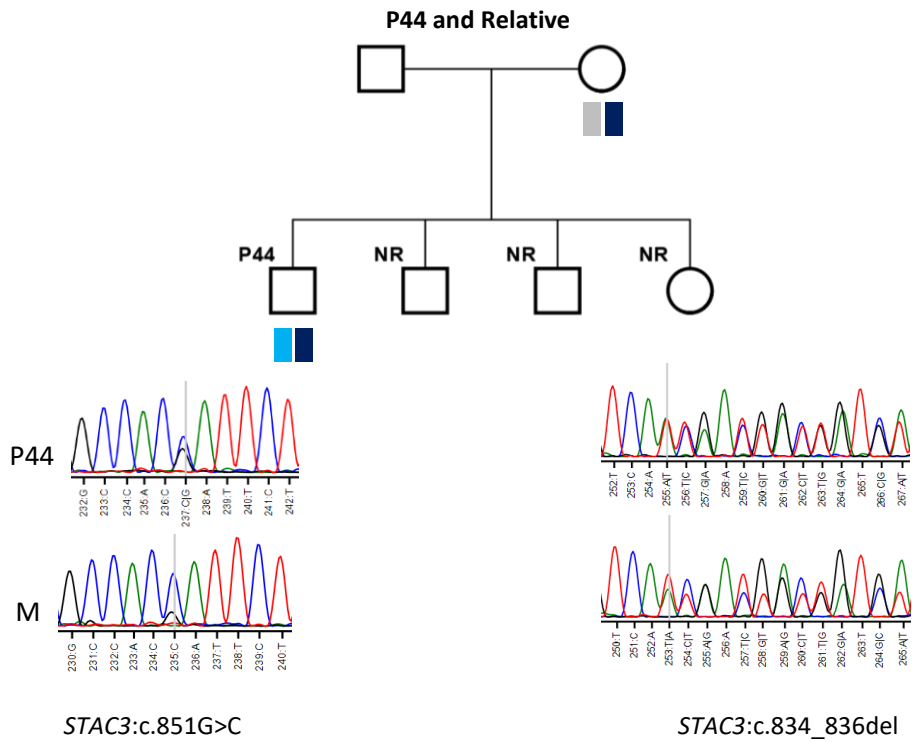
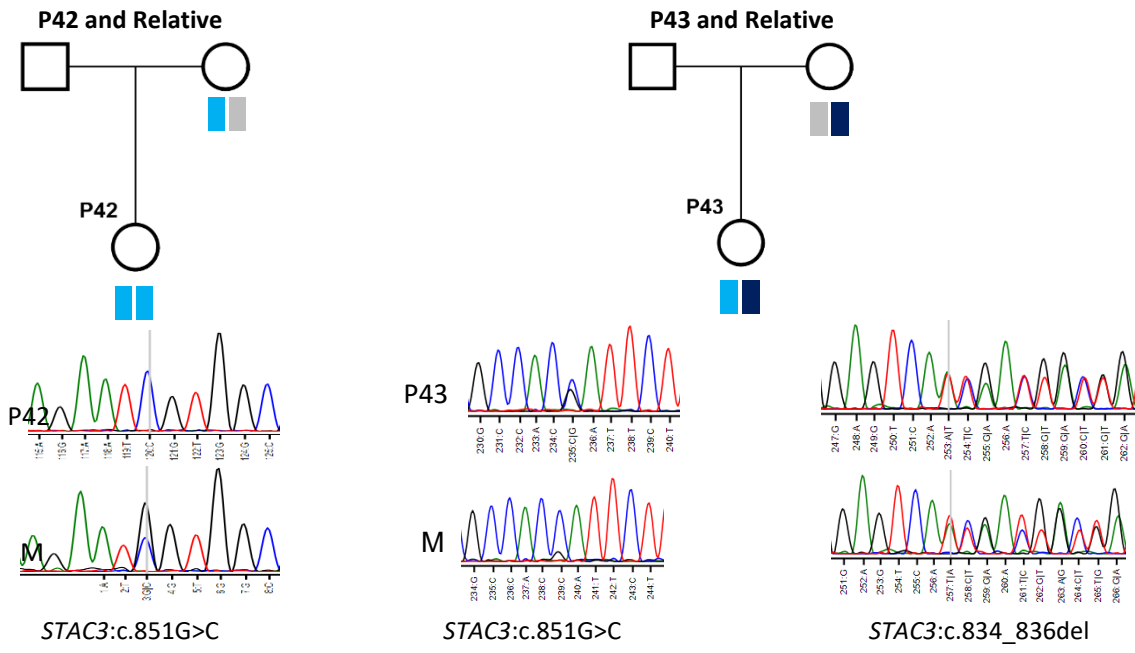
**STAC3:c.851G>C**

**P35 and Relative**



**STAC3:c.851G>C**





Abbreviations: NR, Not recruited; SB, Still birth; SAB, Spontaneous abortion

- |  |   |  |                                       |
|--|---|--|---------------------------------------|
|  | Wildtype                                |  | <i>RYR1</i> :c.6797-1G>T              |
|  | <i>RYR1</i> :[c.10348-6C>G, c.14524G>A] |  | <i>RYR1</i> :c.2870+1G>T              |
|  | <i>RYR1</i> :c.8342_8343delTA           |  | <i>RYR1</i> :c.12814_12815insCGCGGAGT |
|  | <i>RYR1</i> :c.12625-1G>A               |  | <i>RYR1</i> :c.10280C>T               |
|  | <i>RYR1</i> :c.2682+4A>G                |  | <i>STAC3</i> :c.851G>C                |
|  | <i>STAC3</i> :c.834_836del              |  |                                       |