Normalized Melting Curves

Difference Plots

Exon 2b

c.498_596del
p.Ile167_Gly169del

Exon 2b

c.498_596del
p.Ile167_Gly169del

Exon 7b
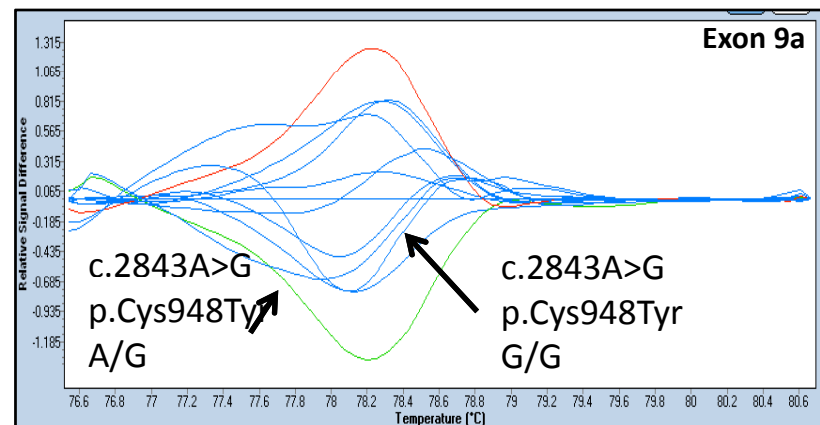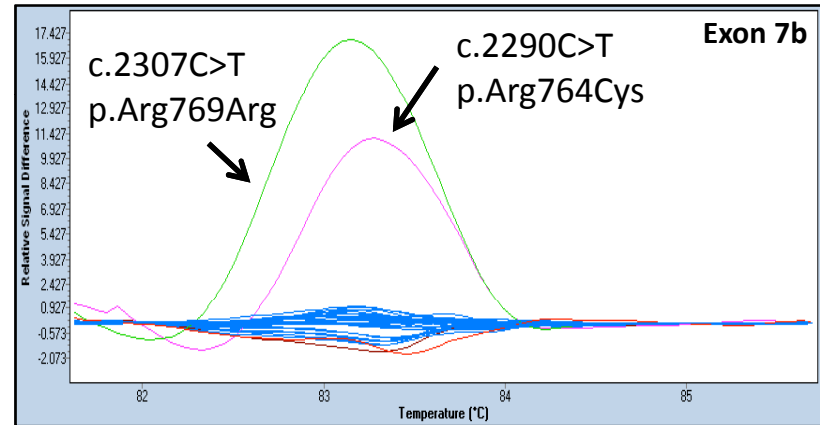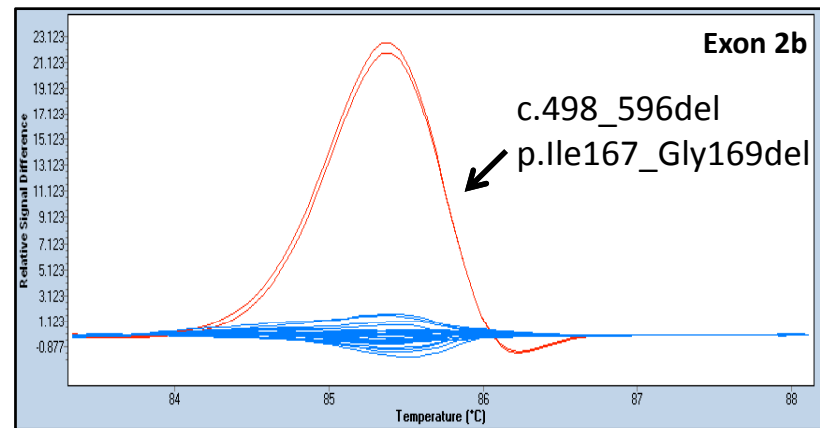
c.2290C>T
p.Arg764Cys

c.2307C>T
p.Arg769Arg

Exon 7b

c.2307C>T
p.Arg769Arg

c.2290C>T
p.Arg764Cys

Exon 9a

Exon 9a

c.2843A>G
p.Cys948Tyr
A/G

c.2843A>G
p.Cys948Tyr
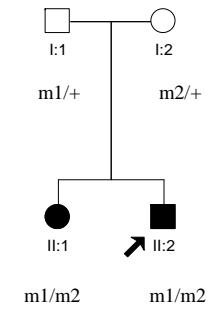G/G
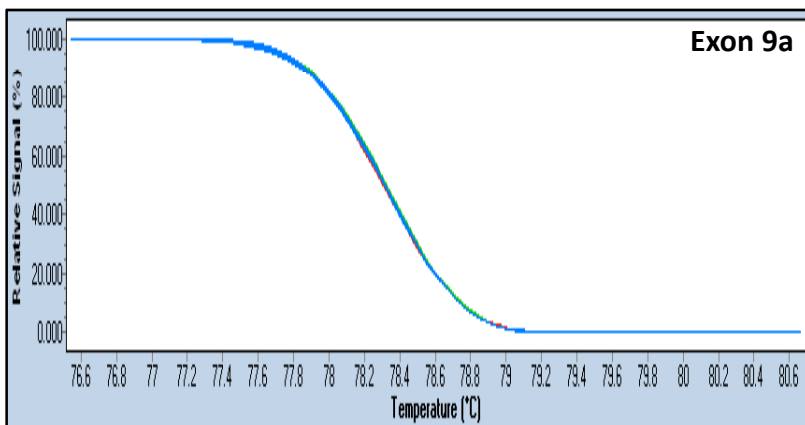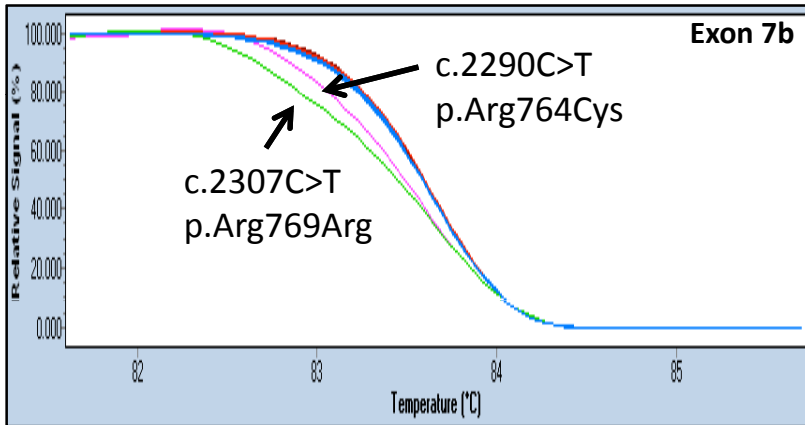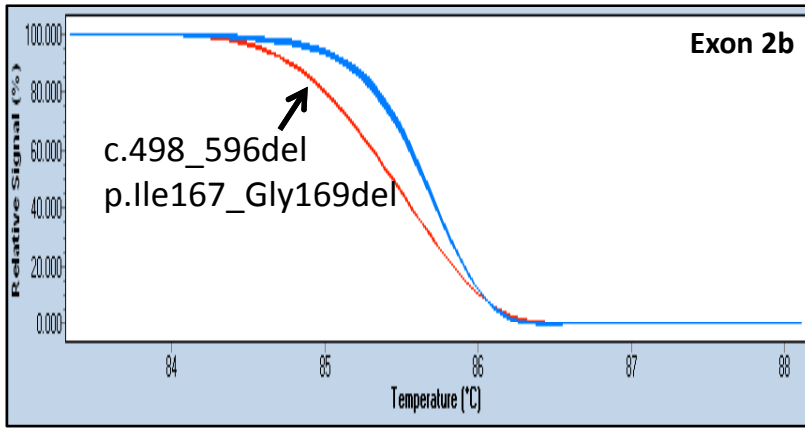
LCA-0004
m1: *CRB1* p.Cys896*
m2: *CRB1* p.Cys948Tyr

I:1 m1/+
I:2 m2/+
II:1 m1/m2
II:2
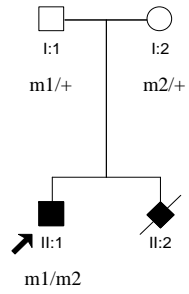
LCA-0010
m: *CRB1* p.Ala161Glyfs*8

I:1
I:2 m/+
II:1 m/m
II:2 m/m
II:3 m/+
II:4 m/m
II:5 m/+
II:6 +/+
II:7 m/+
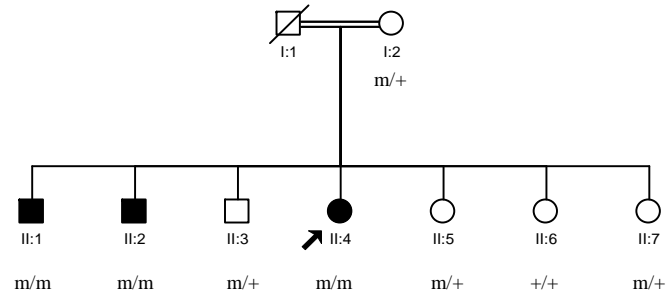
LCA-0011
m1: *CRB1* p.Ser749del
m2: *CRB1* p.Cys948Tyr

I:1 m1/+
I:2 m2/+
II:1 m1/m2
II:2 m1/m2

LCA-0017
m: *CRB1* p.Lys801*

I:1 m/+
I:2 m/+
II:1 m/m
II:2 m/+
II:3 m/m
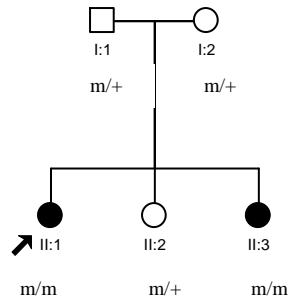
LCA-0019
m1: *CRB1* p.Ile205Aspfs*13
m2: *CRB1* p.Val743Serfs*11

I:1 m1/+
I:2 m2/+
II:1 m1/m2
II:2 m1/m2
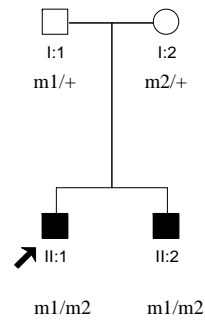
LCA-0027
m1: *CRB1* p.Cys948Tyr
m2: *CRB1* p.Glu1330*

I:1 m2/+
I:2 m1/+
II:1 m1/m2
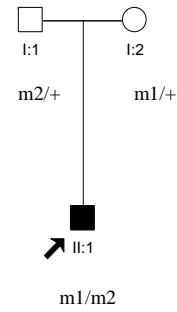
LCA-0028
m1: *CRB1* p.Cys948Tyr
m2: *CRB1* p.Ile1100Thr

I:1 m1/+
I:2 m2/+
II:1 m1/m2
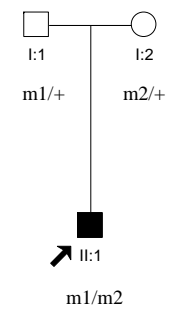
Supplemental Figure S2.

LCA-0032
m1: *CRB1* p.Leu535Pro
m2: *CRB1* p.Cys948Tyr

I:1 m2/+   I:2 m1/+

II:1 m1/m2   II:2 m2/+
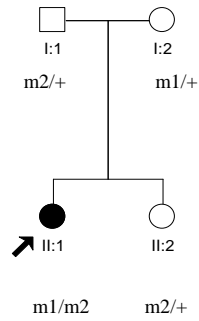
LCA-0050
m: *CRB1* p.Cys948Tyr

I:1 m/+   I:2 m/+

II:1 m/+   II:2 m/+   II:3 m/m   II:4
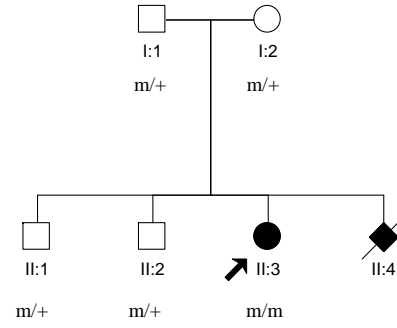
LCA-0051
m1: *CRB1* p.Ile205Aspfs*13
m2:*CRB1* p.Cys948Tyr

I:1   I:2

	I:1		I:2	
D1S408	186	186	182	186
D1S2757	258	266	254	266
D1S2816	250	252	252	250
p.Ile205Aspfs*13	+	+	m1	+
p.Cys948Tyr	m2	+	+	+
D1S1660	240	256	248	236

II:1   II:2   II:3   II:4   II:5

	II:1		II:2		II:3		II:4		II:5	
D1S408	186	186	186	182	186	186	186	186	186	182
D1S2757	258	266	266	254	266	266	266	258	266	254
D1S2816	250	250	252	252	250	252	250	250	250	252
p.Ile205Aspfs*13	+	+	+	m1	+	+	+	+	+	m1
p.Cys948Tyr	m2	+	+	+	+	+	+	+	m2	+
D1S1660	240	236	256	248	256	236	256	236	240	248

LCA-0060
m1: *CRB1* p.Gly770Val
m2: *CRB1* p.His935Glnfs*13

I:1 m1/+   I:2

II:1 m1/m2
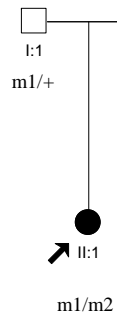
LCA-0063
m1: *CRB1* p.Ile205Aspfs*13
m2: c.4005+1G>A

I:1 m1/+   I:2 m2/+

II:1 m1/m2
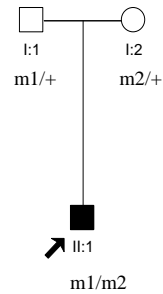
LCA-0099
m1: *CRB1* p.Asp564Tyr
m2: *CRB1* p.Cys896*

I:1 m1/+   I:2 m2/+

II:1 m1/m2
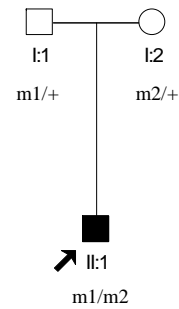
LCA-0101
m1: *CRB1* p.Tyr1051*
m2: *CRB1* p.Val1334Trpfs*

I:1   I:2

II:1   II:2 m1/m2   II:3
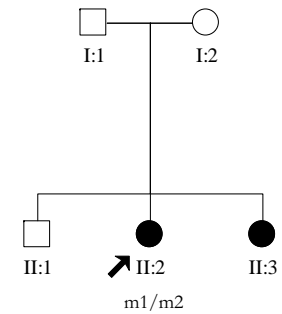
Supplemental Figure S2 (Continued).

RP-0025
m: *CRB1* p.Ile1100Thr

I:1 I:2

II:1 II:2 II:3 II:4 II:5 II:6
m/m
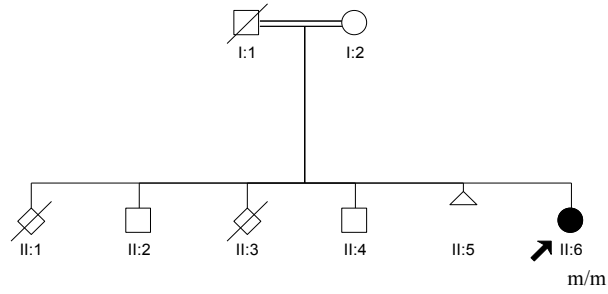
RP-0091
m1: *CRB1* p.Ala161Glyfs*8

I:1 I:2

II:1 II:2
m1/+
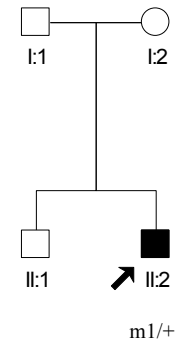
MD-0092
m1: *CRB1* p.Ile167_Gly169del
m2: *CRB1* p.Cys948Tyr

I:1 I:2 I:3 I:4

II:1 II:3 II:4 II:6
+/+ m1/m2 m2/m2
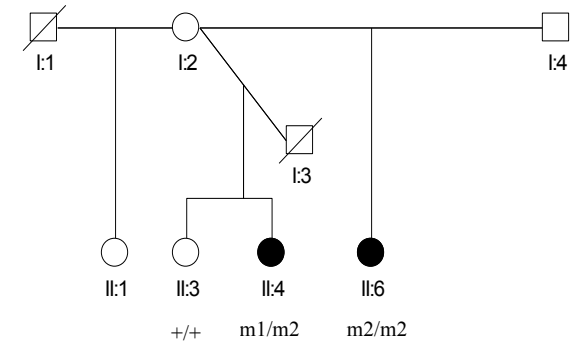
RP-0280 &
m1: *CRB1* p.Trp822*
m2: *CRB1* p.Cys948Tyr

I:1 I:2
m2/+ m1/+

II:1 II:2 II:3 II:4
m2/+ m2/+ m2/+ m1/m2
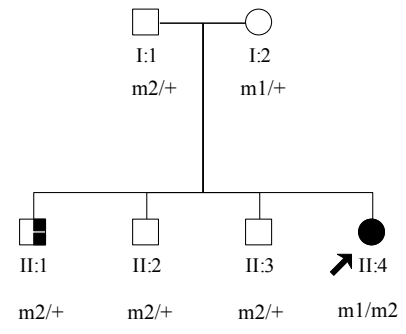
MD-0351
m1: *CRB1* p.Cys948Tyr
m2: *CRB1* p.Met1053Val

I:1 I:2
m2/

II:1 II:3 II:2
m1/m2 m1/+

m2/+
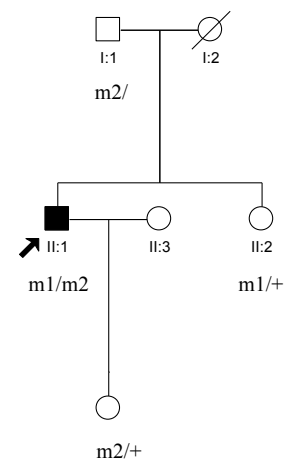
RP-0487
m1: *CRB1* p.Arg764Cys
m2: *CRB1* p.Glu1330*

I:1 I:2

II:1 II:2 II:3 II:4
m1/m2

Supp Figure S3.

RP-0561
m1: *CRB1* p.Thr745Met
m2: *CRB1* p.Glu1330*

I:1 m1/+   I:2 m2/+

II:1 m1/m2   II:2 m1/m2   II:3 m1/+

MD-0643
m1: CRB1 p.Thr745Met
m2: p.Cys948Tyr

I:1   I:2

II:1 m1/m2

RP-0745
m1: *CRB1* p.Ile167_Gly169del
m2: *CRB1* p.Arg764Cys

I:1 m1/+   I:2 m2/+

II:1 m1/m2   II:2 m2/+

RP-1017
m1: *CRB1* p.Thr745Met
m2: *CRB1* p.Glu806*

I:1 m1/+   I:2 m2/+

II:1   II:2 m1/m2

RP-1212
m: *CRB1* p.Ile1100Thr

I:1   I:2

II:1 m/m

RP-1311
m1: *CRB1* p.Ile205Aspfs*13

I:1   I:2

II:1 m1/+

RP-1426
m: *CRB1* p.Ile167_Gly169del

I:1   I:2

II:1   II:2   II:3   II:4   II:5 m/m   II:6

RP-1440
m1: *CRB1* c.3878+2insT
m2: *CRB1* p.Gly899Ala

I:1   I:2

II:1   II:2   II:3   II:4   II:5   II:6

	II:1	II:2	II:3	II:4	II:5	II:6
D1S408	180	176 176	176 180	176 176	184 176	176 176	176
D1S2757	256	258 260	258 256	258 260	262 260	258 260	258
D1S2816	246	246 244	246 246	246 244	246 244	246 244	246
c.3878+2insT	+	+ m1	+ +	+ m1	+ m1	+ m1	+
p.Gly899Ala	+	+ +	+ +	+ m2	+ +	+ +	+
D1S1660	230	242 242	242 230	242 234	242 242	242 242	242

RP-1504
m1: *CRB1* p.His568Tyr
m2: p.Pro1381Leu

I:1   I:2

II:1   II:2 m1/m2

Supp Figure S3. (Continued).

RP-1535
m1: *CRB1* p.Asp564Tyr
m2: *CRB1* p.Asp1005Val
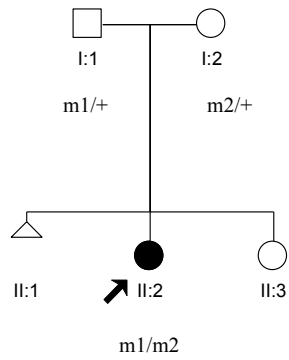
RP-1558
m1: *CRB1* p.Cys948Tyr
m2: *CRB1* p.Glu1203*
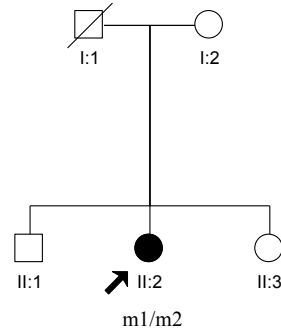
RP-1586
m: *CRB1* p.Thr745Met
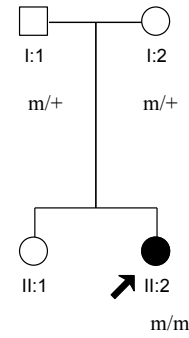
RP-1611
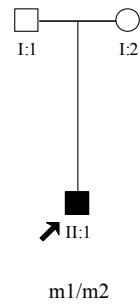m1: *CRB1* p.Ile167_Gly169del
m2: *CRB1* p.C383Sfs*66
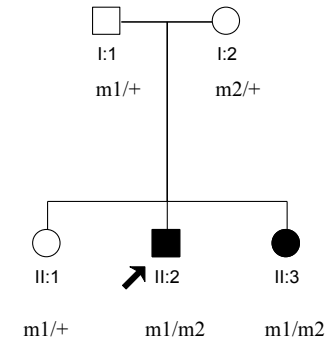
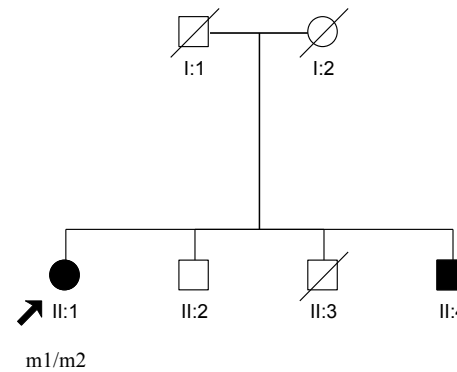**RP-1615**
m1*: CRB1* p.Ile1100Thr
m2: c.3749+1_3749+2delGT
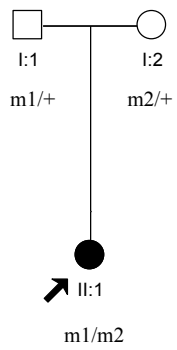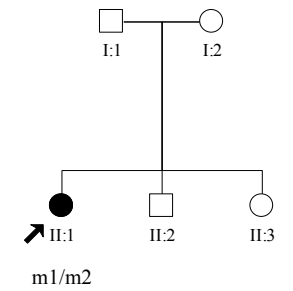
RP-1625
m1: *CRB1* p.Cys948Tyr
m2: *CRB1* p.Met1053Val
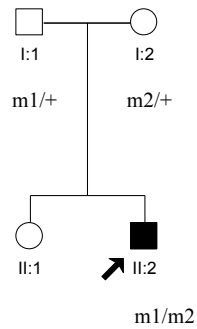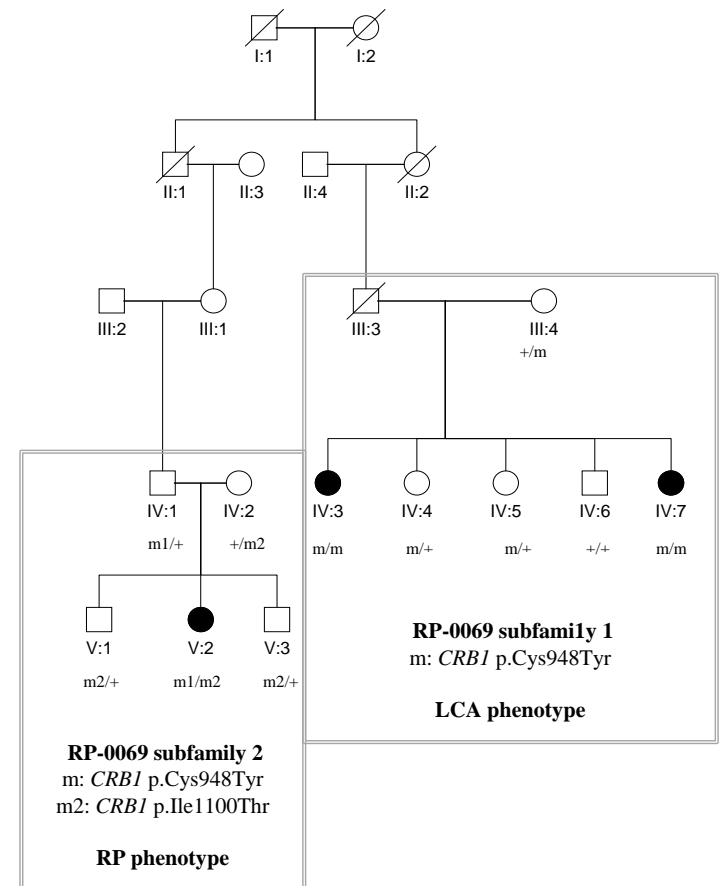
RP-1689
m1: *CRB1* p.Arg764His
m2: p.Arg1390*

RP-1779
m1: *CRB1* p.Arg764Cys
m2: *CRB1* p.Ile1100Thr

RP-2004
m1: *CRB1* p.Ile167_Gly169del
m2: p.Thr745Met

Supp Figure S3. (Continued).

Supp Figure S4

Supp Figure S5

Suppl Figure S6