

Supplemental_Figure_S2

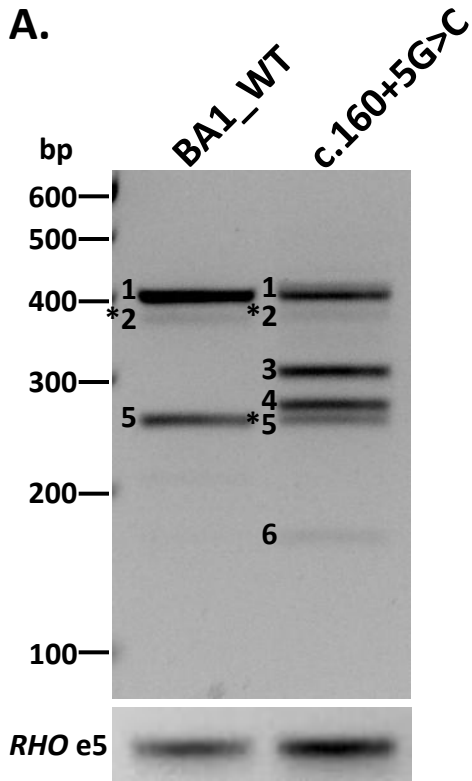
Supplemental Figure S2. RT-PCR analysis for the non-canonical splice site variants in *ABCA4*. Forty-seven variants were inserted in 18 BA constructs and the MG30 minigene. RT-PCR was performed by using *ABCA4* exonic primers (Supplemental Table S10) and for four variants (BA1_c.160+5G>C, BA4_c.768G>T, BA25_c.5312+3A>T, MG30_c.6729+5_6729+19del) by using one or both rhodopsin (*RHO*) primers.

The fragments for which we could not provide sequence validation are marked with an asterisk (*). Exon elongations are represented as gray rectangles of different length delimited at one of the two edges by a stippled line, while intron retentions given by gray rectangles are delimited by closed lines (e.g. fragment 5 in BA17). For wild-type constructs BA1, BA12 and BA26, showing alternative spliced products, RT-PCR on human retina was performed as depicted in panels with a grey background.

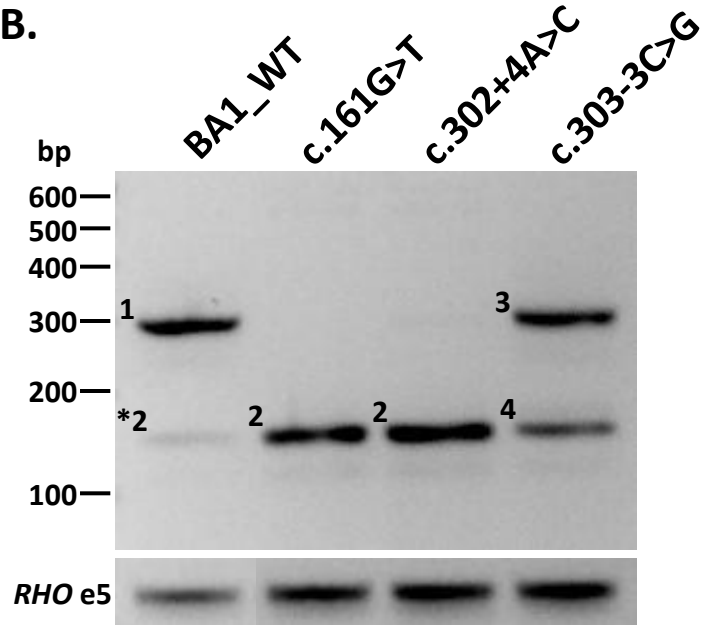
BA20, panel (D), depicts the special characteristics of cryptic splice acceptor and donor sites that reside next to each other in exon 30. The non-canonical splice variants c.4538A>G or c.4538A>C, both affecting the penultimate nucleotide of exon 30, result in exon 30 elongation but also in partial exon 30 skipping. The first 114 nt of exon 30 are skipped if the cryptic splice acceptor site is used; the last 73 nt of exon 30 are skipped if the cryptic splice donor site is used.

BA1

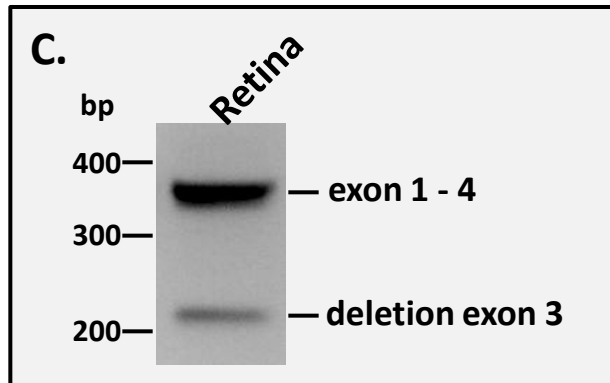
A.



B.



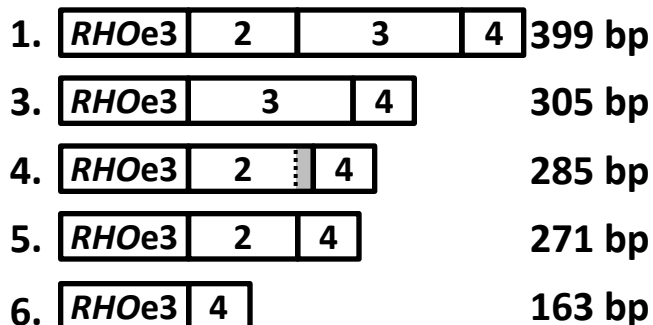
C.



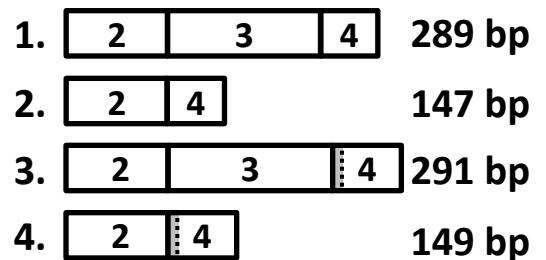
D.



E.

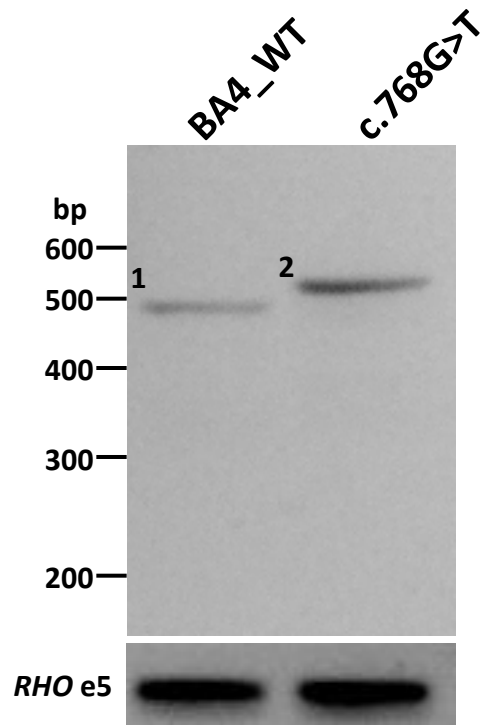


F.



BA4

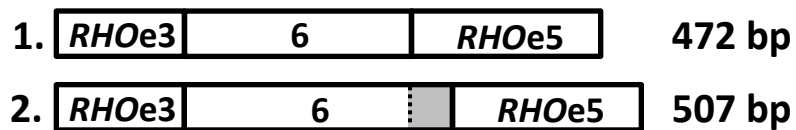
A.



B.

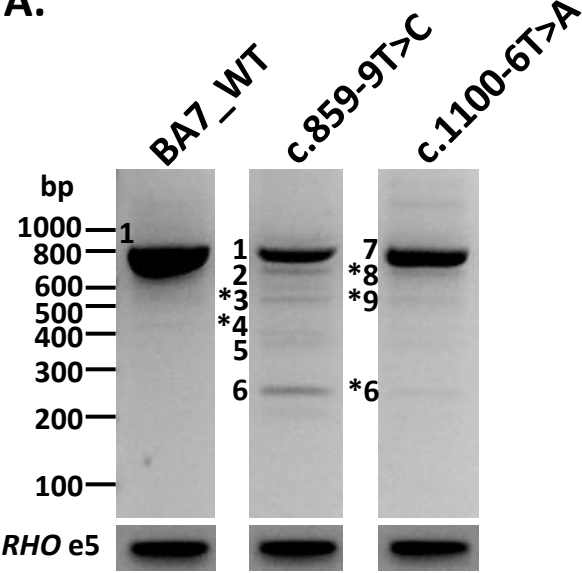


C.

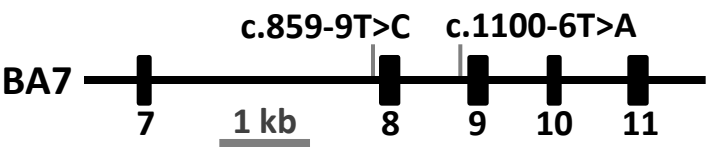


BA7

A.



B.

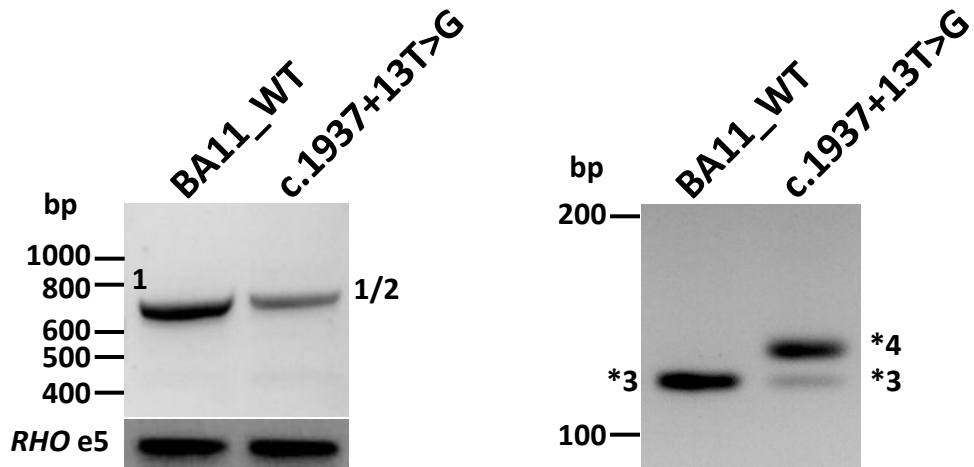


C.

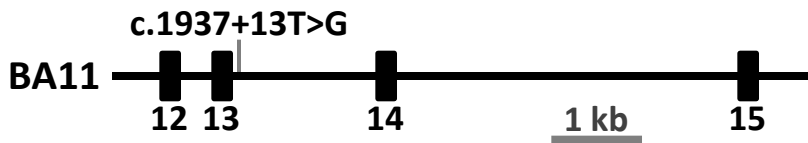
1.	7	8	9	10	11	748 bp
2.	7	8	9	11		631 bp
5.	7	10	11			367 bp
6.	7	11				250 bp
7.	7	8	9	10	11	752 bp

BA11

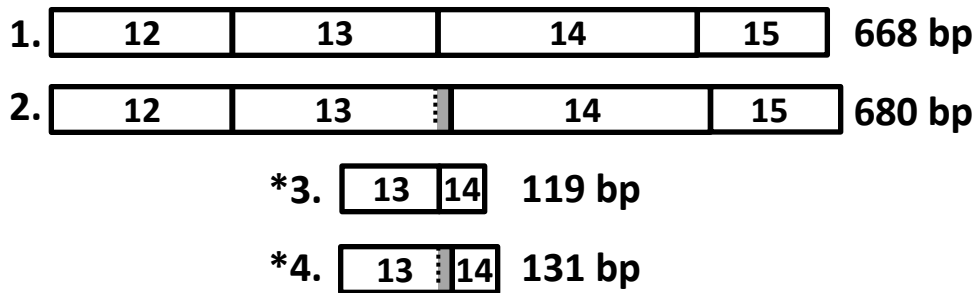
A.



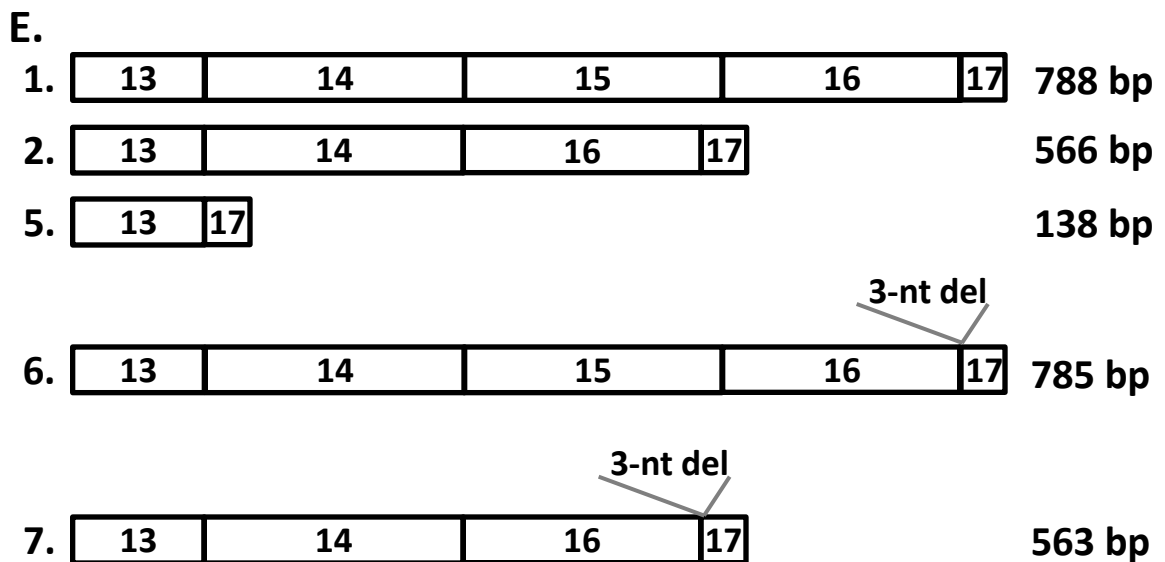
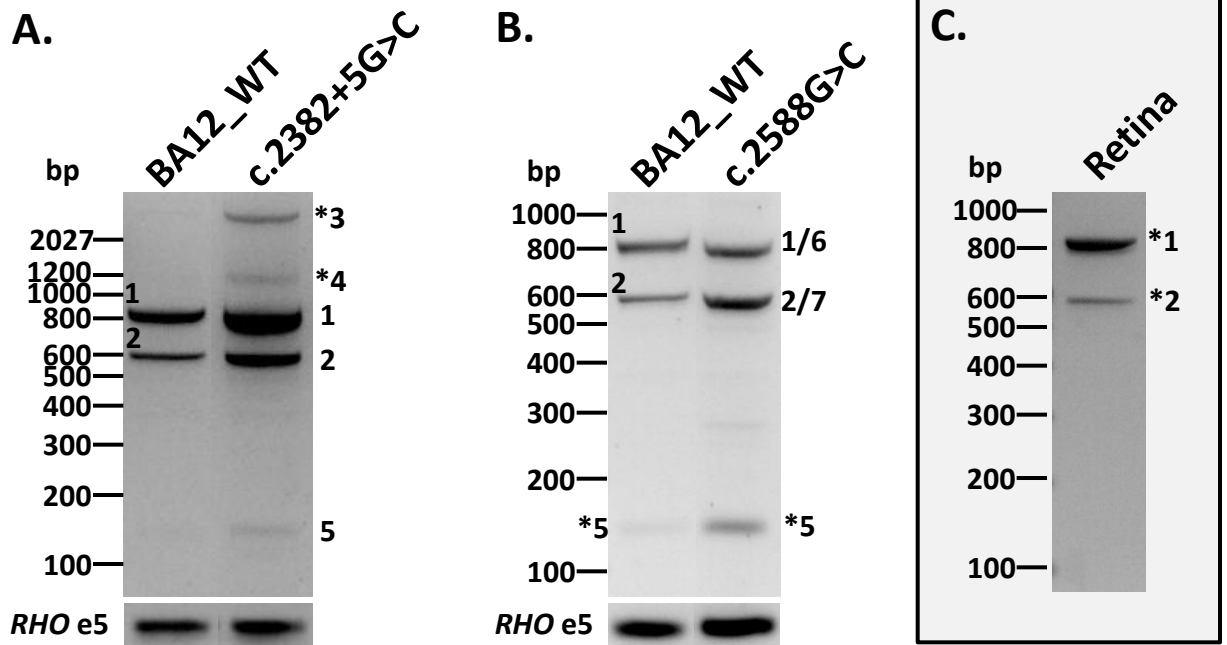
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C.

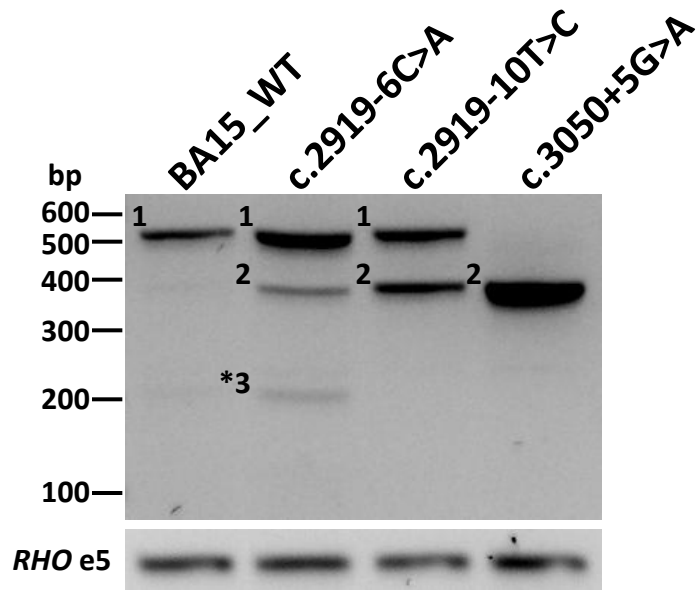


BA12

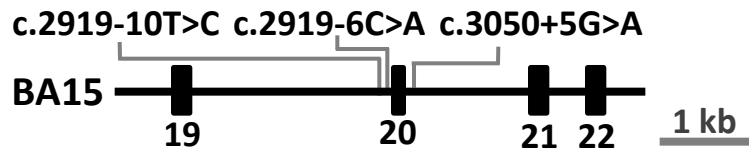


BA15

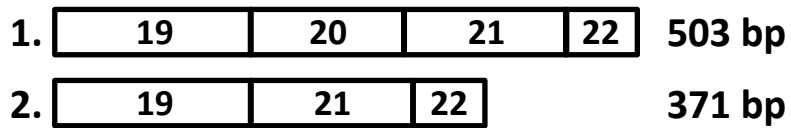
A.



B.

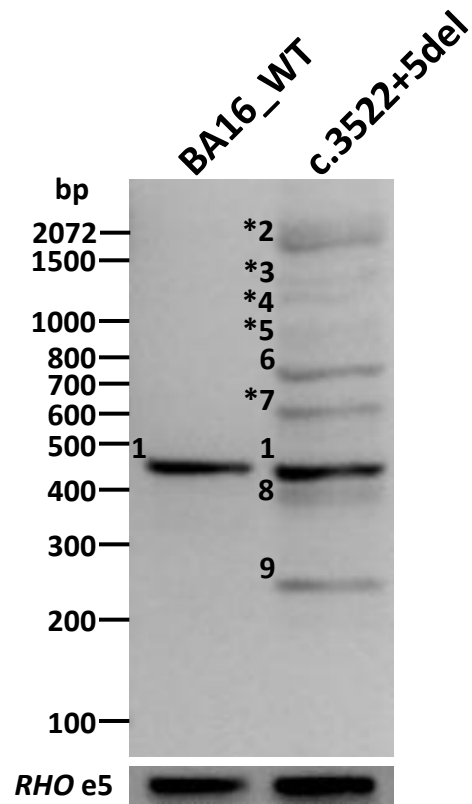


C.

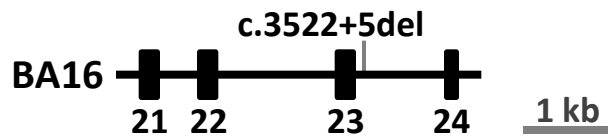


BA16

A.



B.

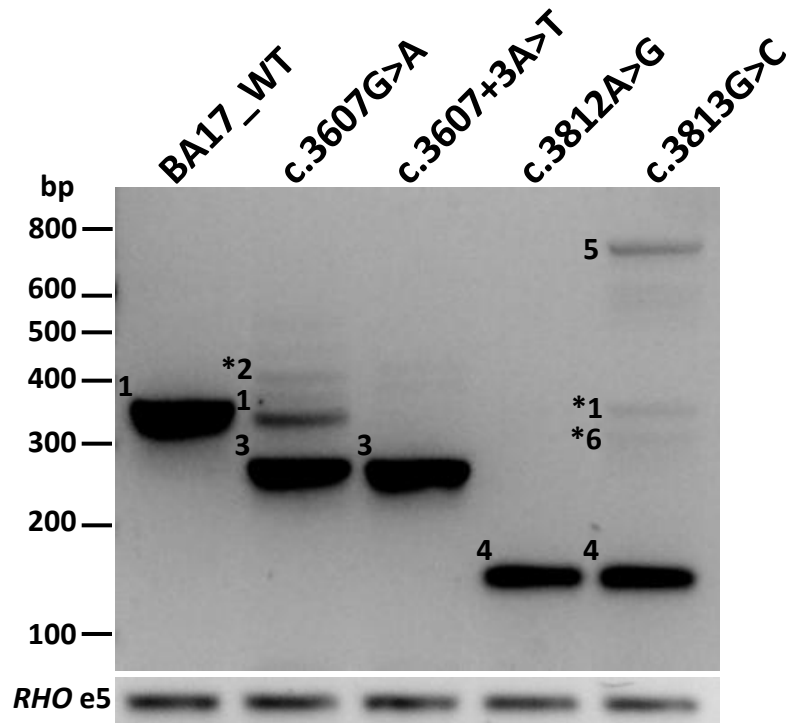


C.

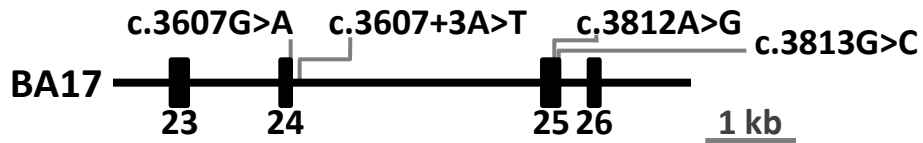
1.	21	22	23	24	440 bp
8.	21	22	23	24	395 bp
9.	21	22	24		246 bp
6.	21	22	23	24	713 bp

BA17

A.



B.

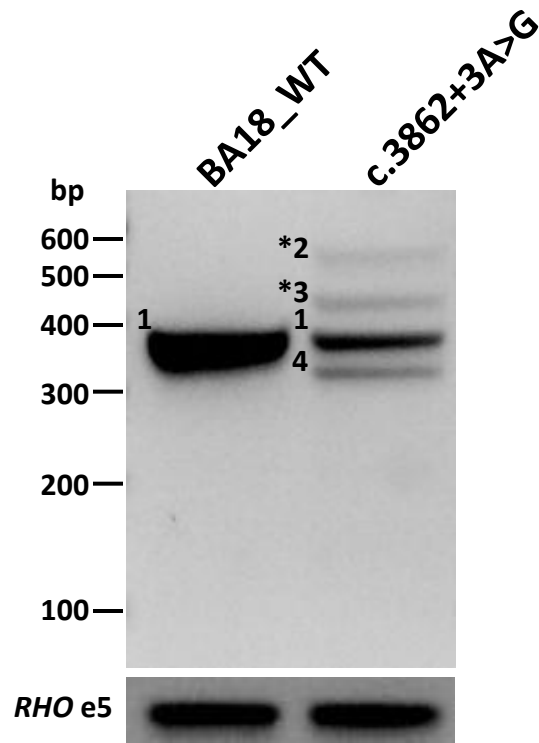


C.

1.	23	24	25	26	357 bp	
3.	23	25	26	272 bp		
4.	23	24	26	151 bp		
5.	23	24	25	intron 25	26	713 bp

BA18

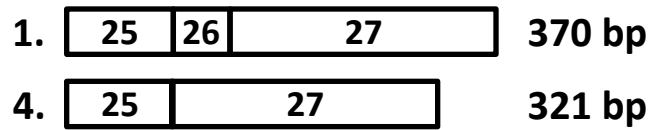
A.



B.

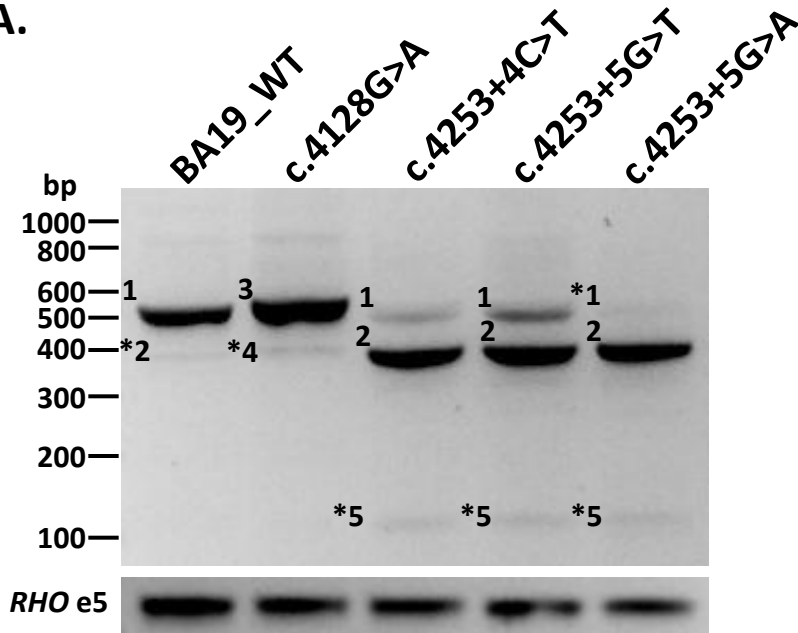


C.



BA19

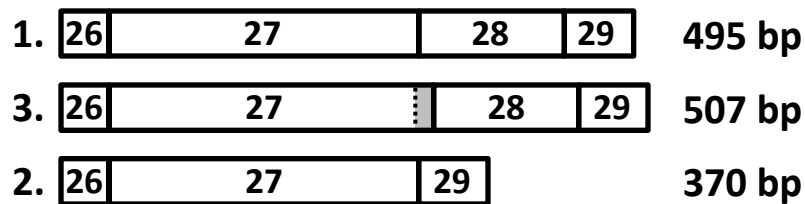
A.



B.

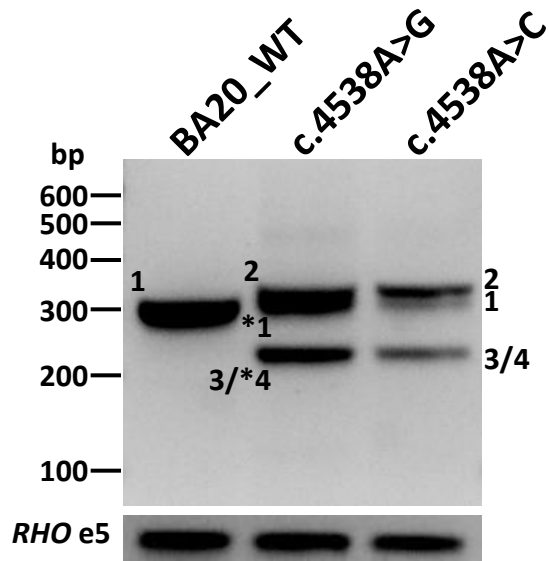


C.



BA20

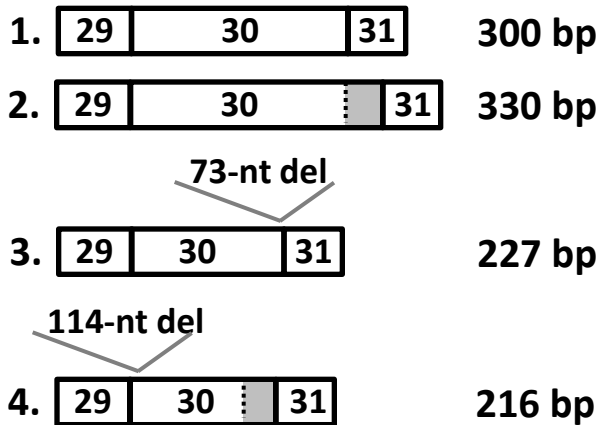
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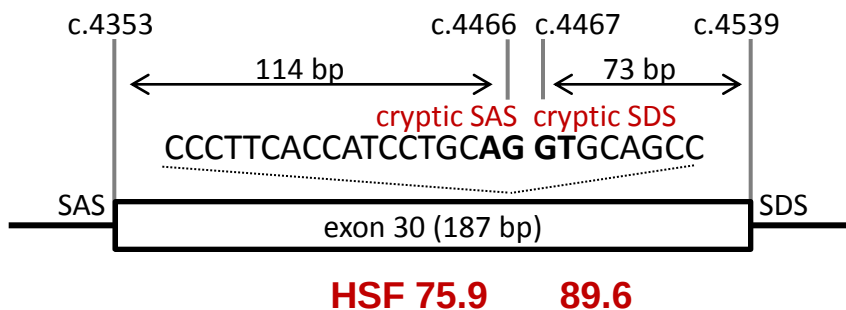
B.



C.

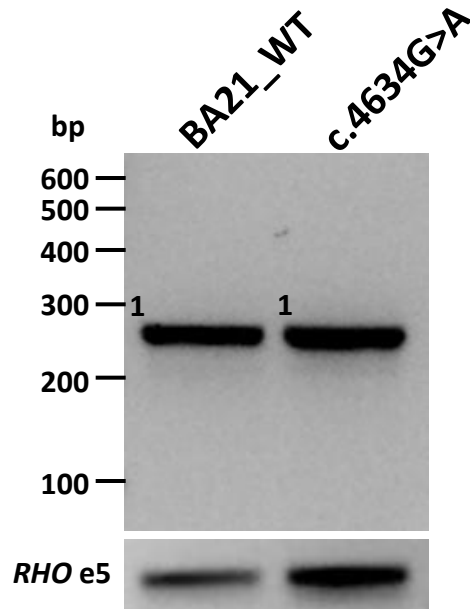


D.

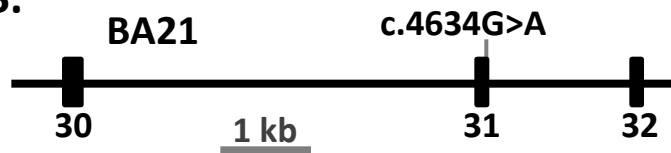


BA21

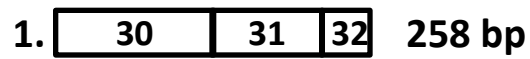
A.



B.

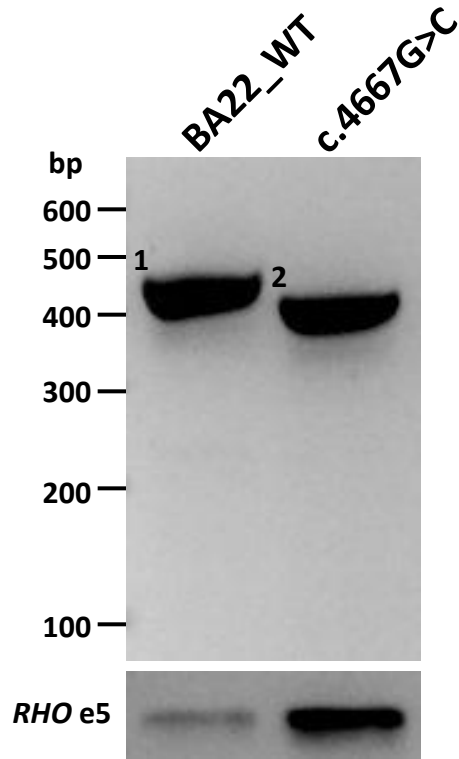


C.

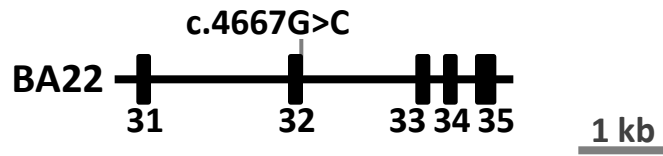


BA22

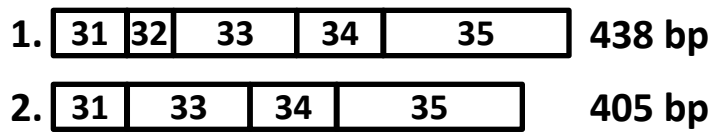
A.



B.

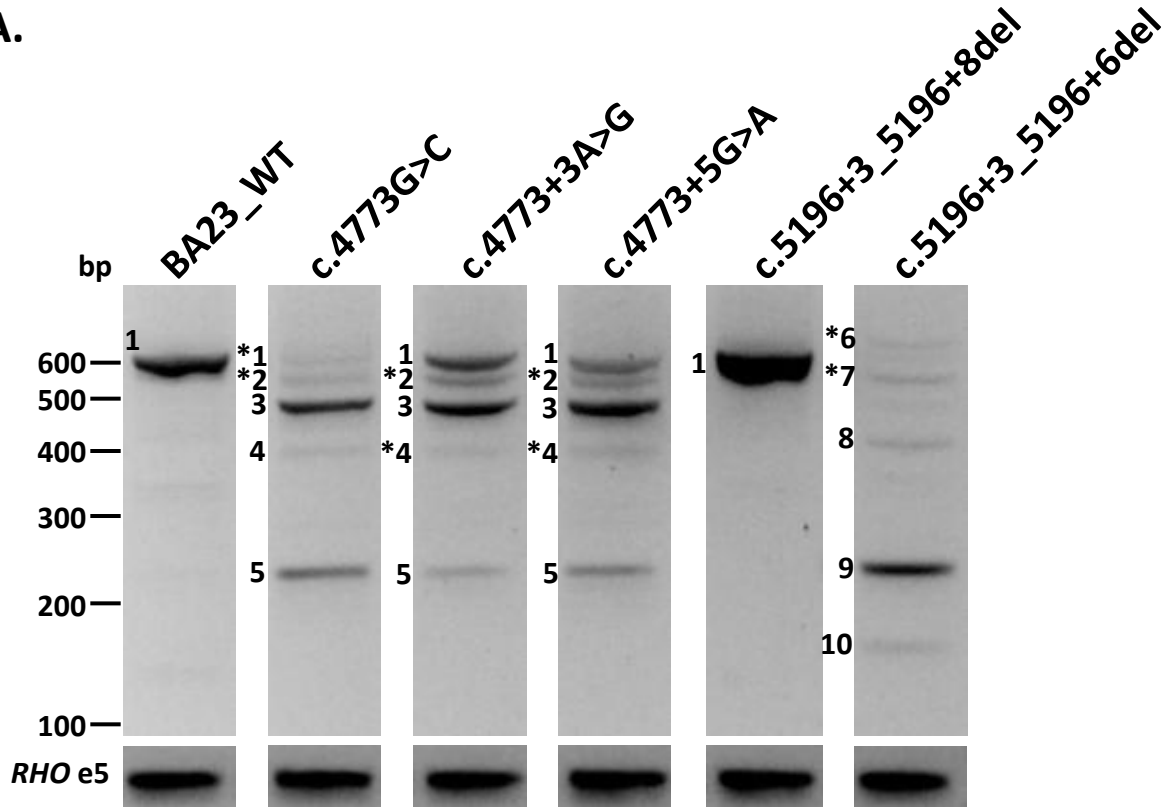


C.

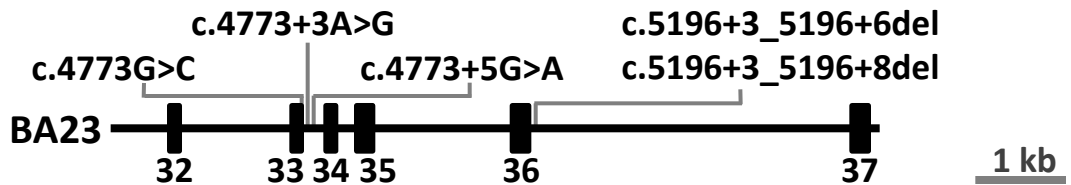


BA23

A.



B.

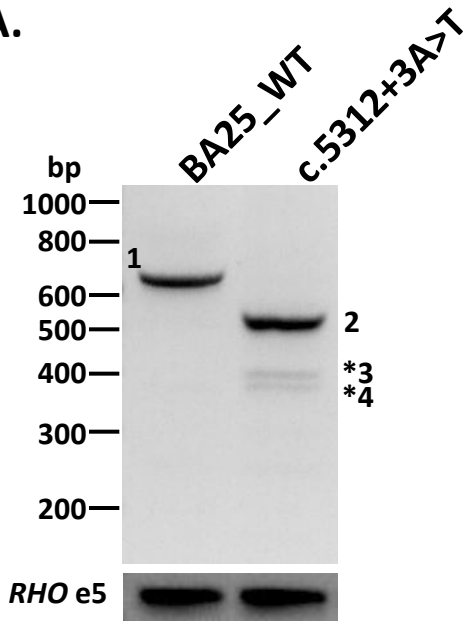


C.

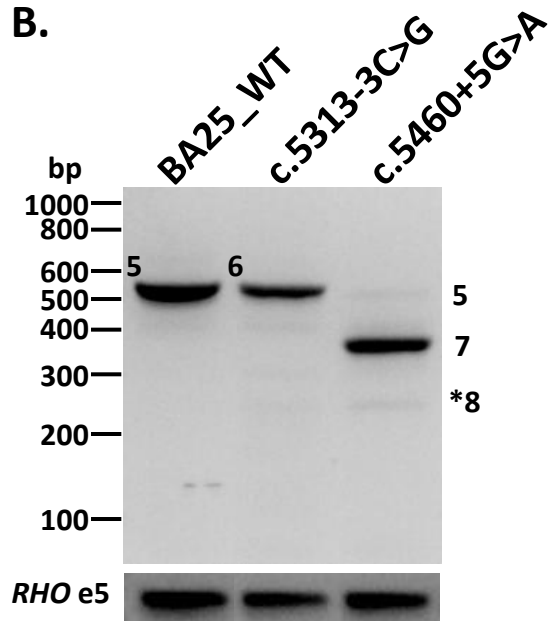
1.	32	33	34	35	36	37	578 bp
3.	32	34	35	36	37		472 bp
4.	32	35	36	37			397 bp
5.	32	36	37				227 bp
8.	32	33	34	35	37		400 bp
9.	32	33	34	37			230 bp
10.	32	33	37				155 bp

BA25

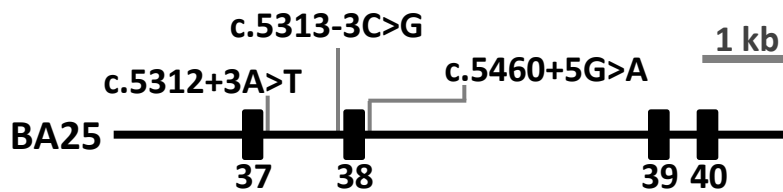
A.



B.



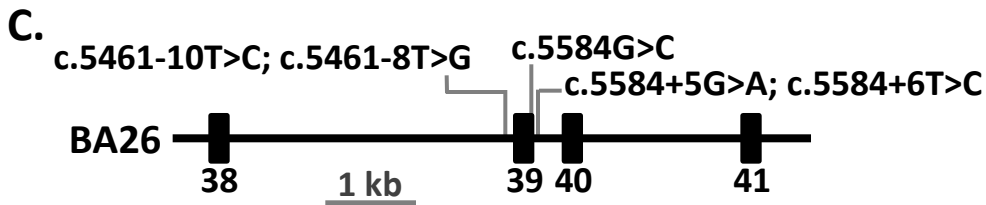
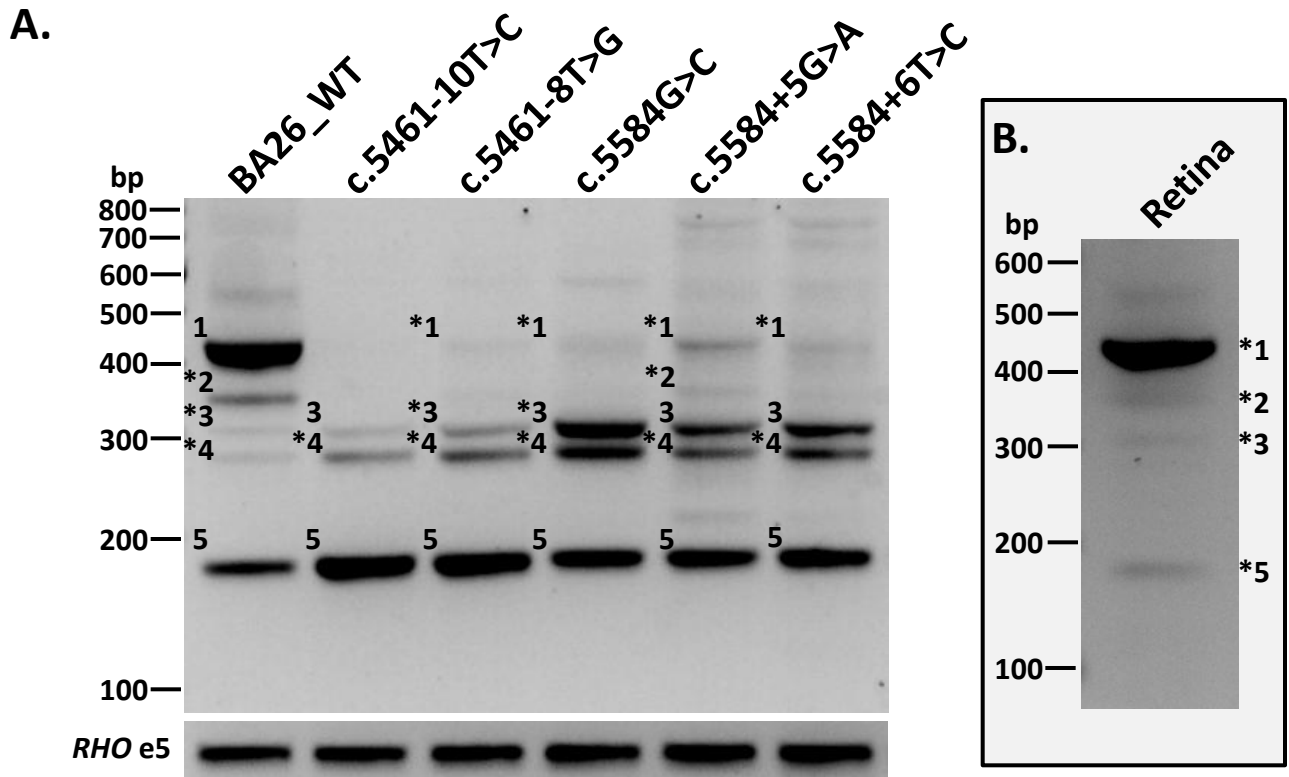
C.



D.

1.	RHOe3	37	38	39	40	627 bp
2.	RHOe3	38	39	40		511 bp
5.	37	38	39	40		517 bp
6.	37	...	38	39	40	519 bp
7.	37	39	40			369 bp

BA26

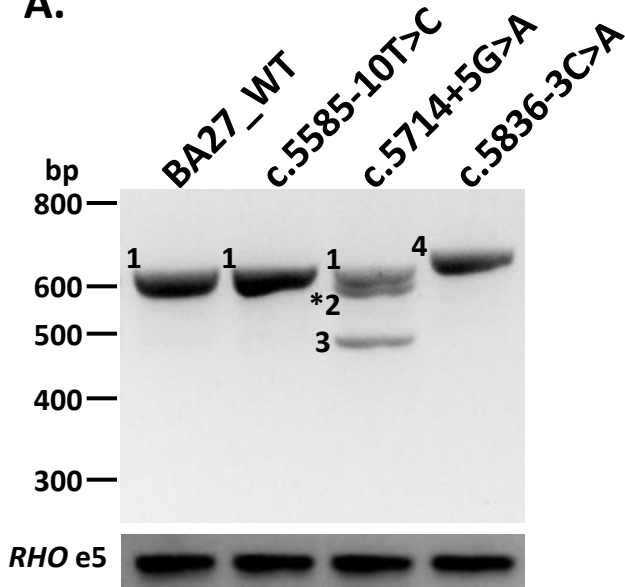


D.

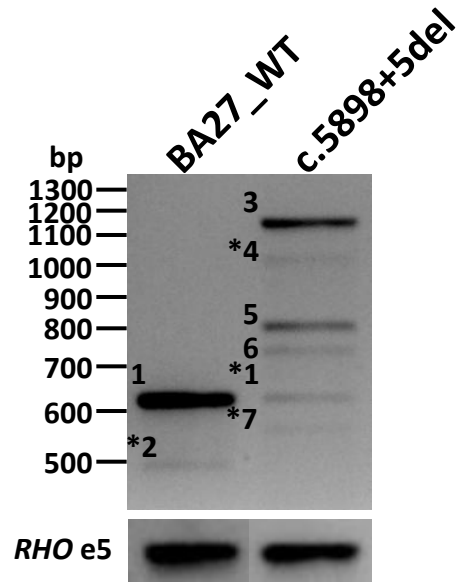
1.	38	39	40	41	429 bp
3.	38	40	41		305 bp
4.	38	39	41		299 bp
5.	38	41			175 bp

BA27

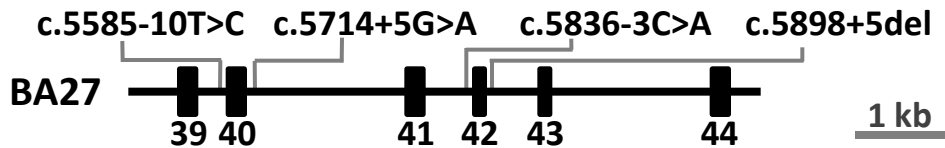
A.



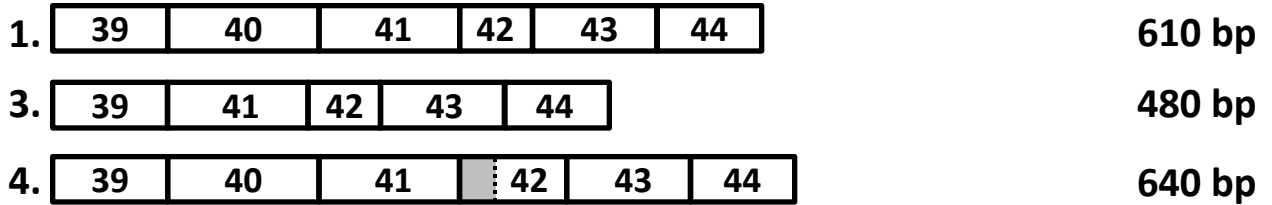
B.



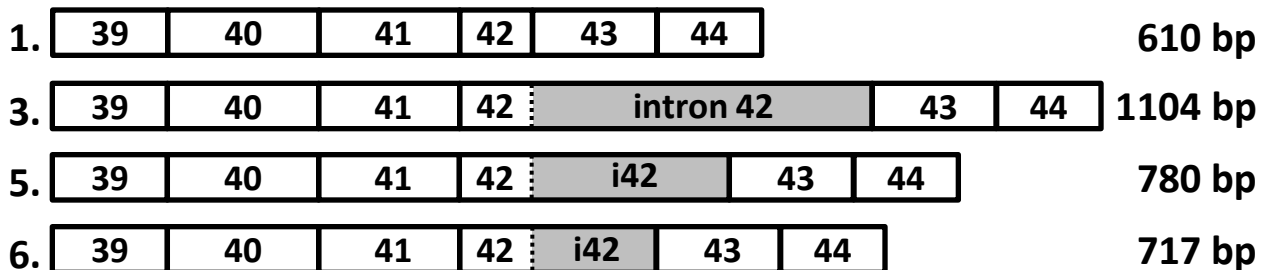
C.



D.

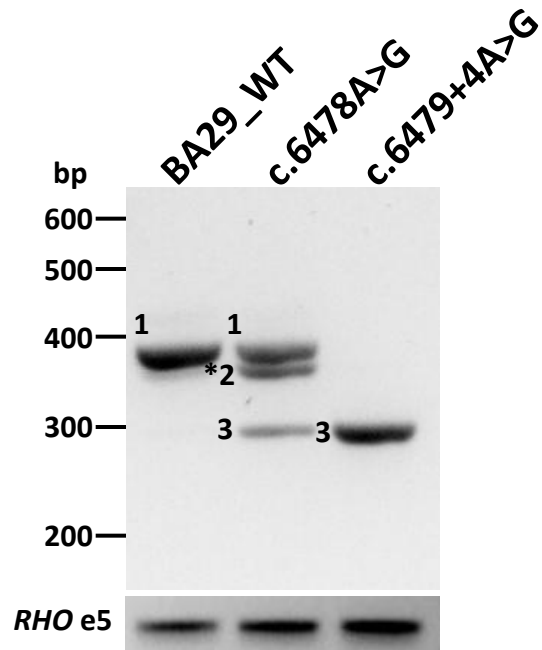


E.



BA29

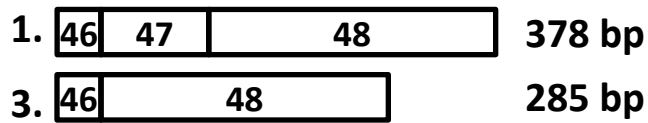
A.



B.

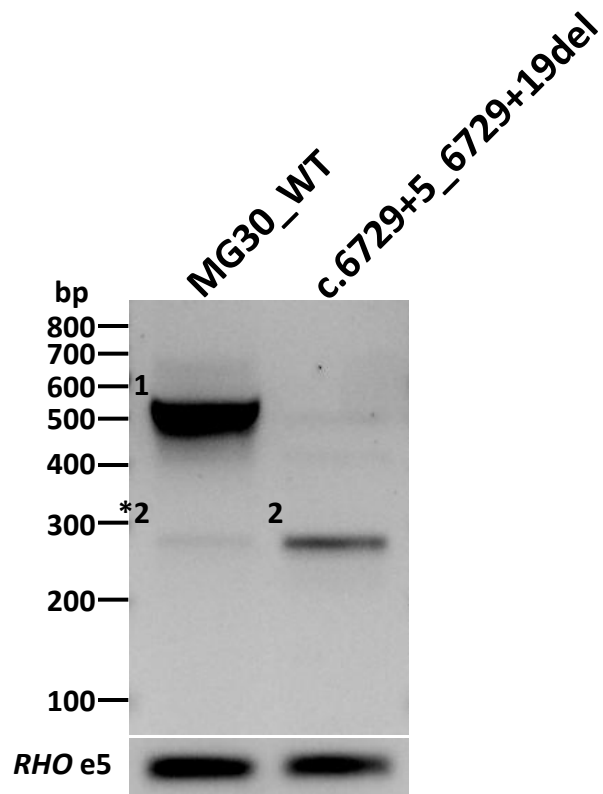


C.

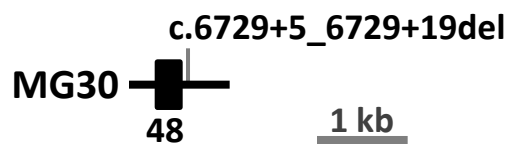


MG30

A.



B.



C.

