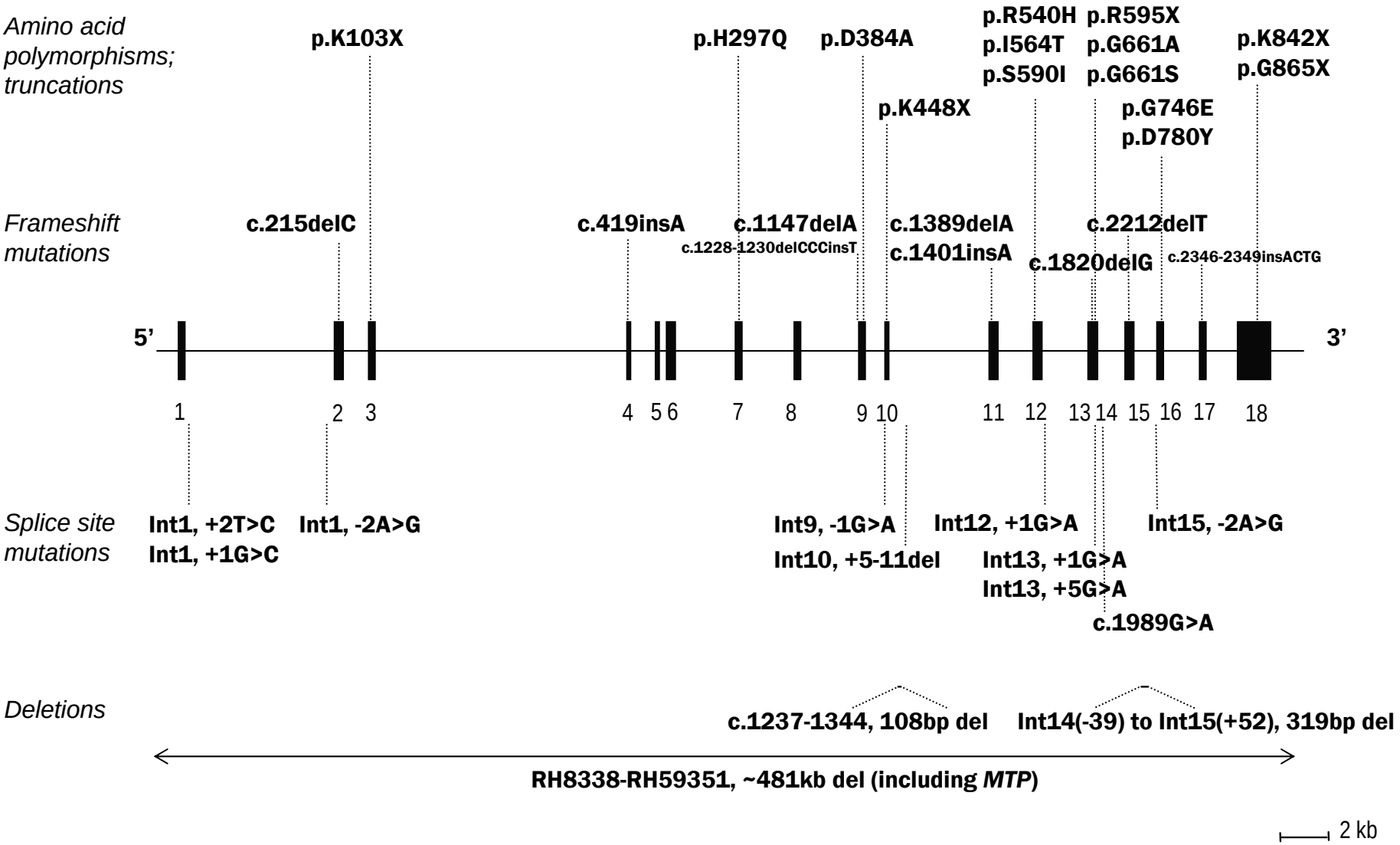
Amino acid polymorphisms; truncations
p.K103X
p.H297Q
p.D384A
p.R540H
p.I564T
p.S590I
p.R595X
p.G661A
p.G661S
p.K842X
p.G865X
p.K448X
p.G746E
p.D780Y
Frameshift mutations
c.215delC
c.419insA
c.1147delA
c.1228-1230delCCCinsT
c.1389delA
c.1401insA
c.1820delG
c.2212delT
c.2346-2349insACTG
5'
3'
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
Splice site mutations
Int1, +2T>C
Int1, +1G>C
Int1, -2A>G
Int9, -1G>A
Int10, +5-11del
Int12, +1G>A
Int13, +1G>A
Int13, +5G>A
c.1989G>A
Int15, -2A>G
Deletions
c.1237-1344, 108bp del
Int14(-39) to Int15(+52), 319bp del
RH8338-RH59351, ~481kb del (including MTP)
2 kb