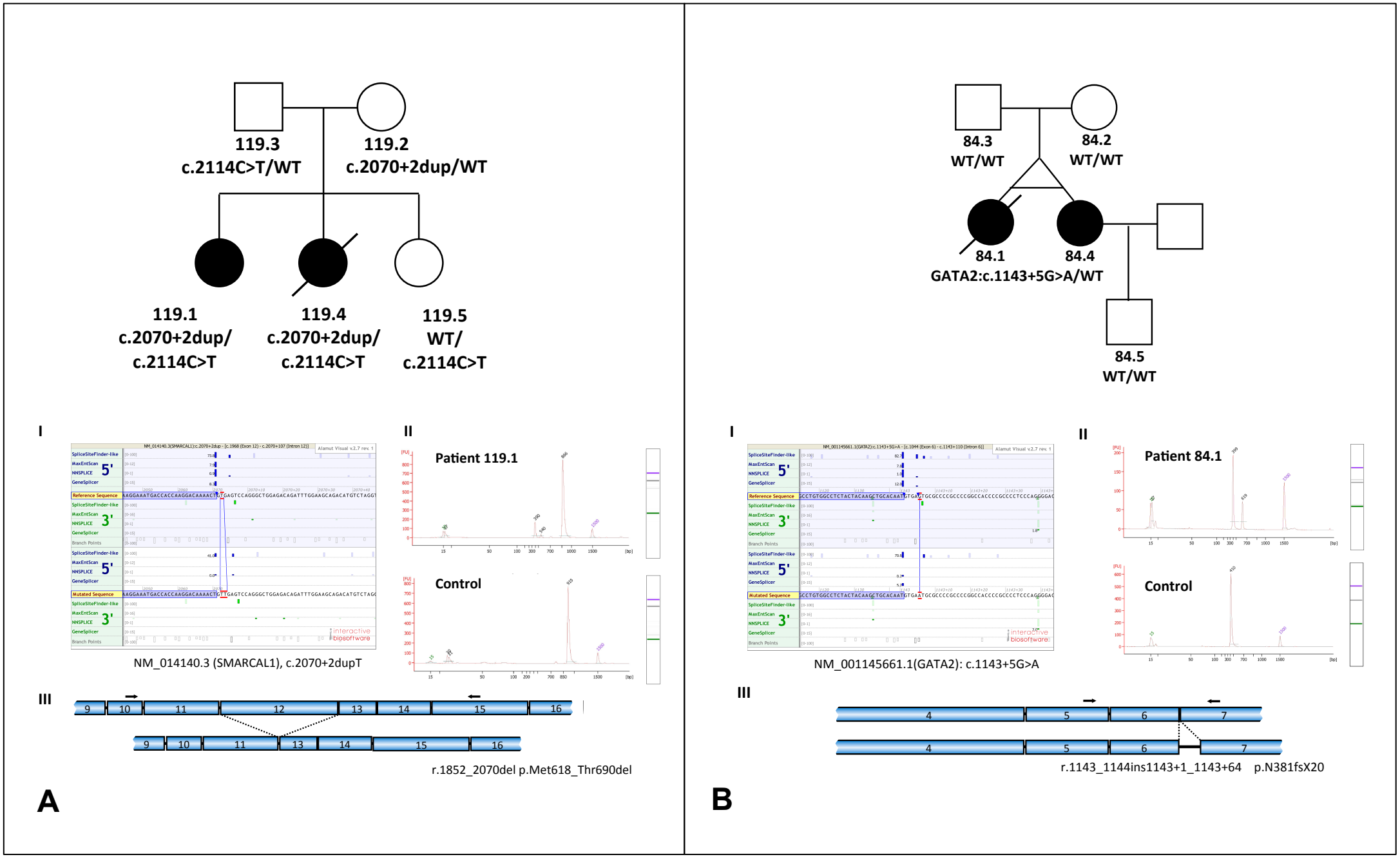
119.3
c.2114C>T/WT
119.2
c.2070+2dup/WT
119.1
c.2070+2dup/
c.2114C>T
119.4
c.2070+2dup/
c.2114C>T
119.5
WT/
c.2114C>T
I
II
Patient 119.1
Control
NM_014140.3 (SMARCAL1), c.2070+2dupT
III
r.1852_2070del p.Met618_Thr690del
A
84.3
WT/WT
84.2
WT/WT
84.1
84.4
GATA2:c.1143+5G>A/WT
84.5
WT/WT
I
II
Patient 84.1
Control
NM_001145661.1(GATA2): c.1143+5G>A
III
r.1143_1144ins1143+1_1143+64 p.N381fsX20
B

Figure E6.