

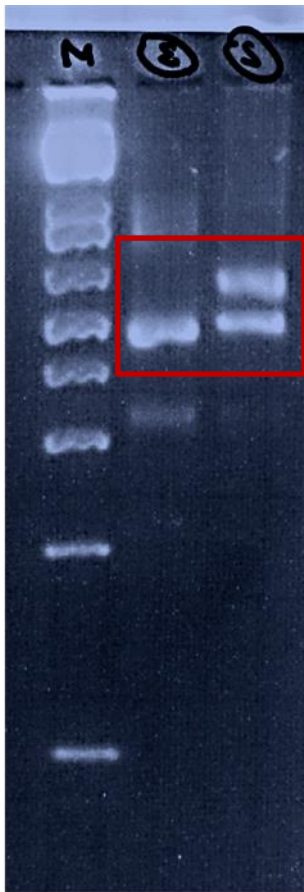
**A deep intronic mutation causes  
RAD50 deficiency through an unusual  
mechanism of distant exon activation**

Bousset K. et al.,

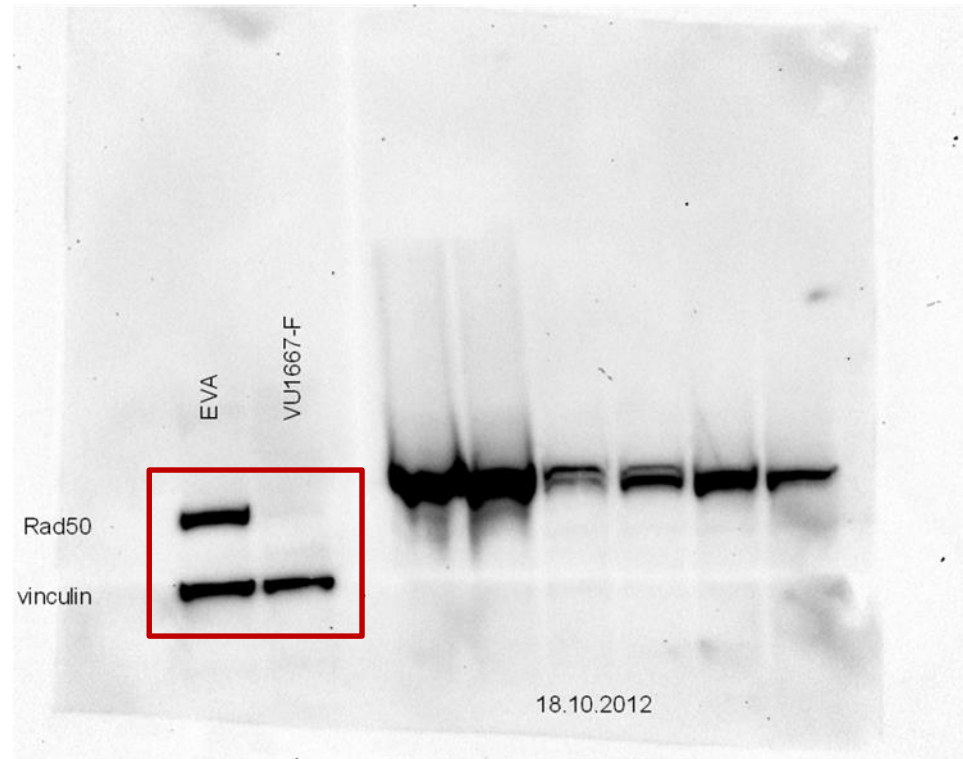
- Full unedited blots/gels -

Full unedited gel for Figure 1A:  
patient RNA  
cDNA analysis

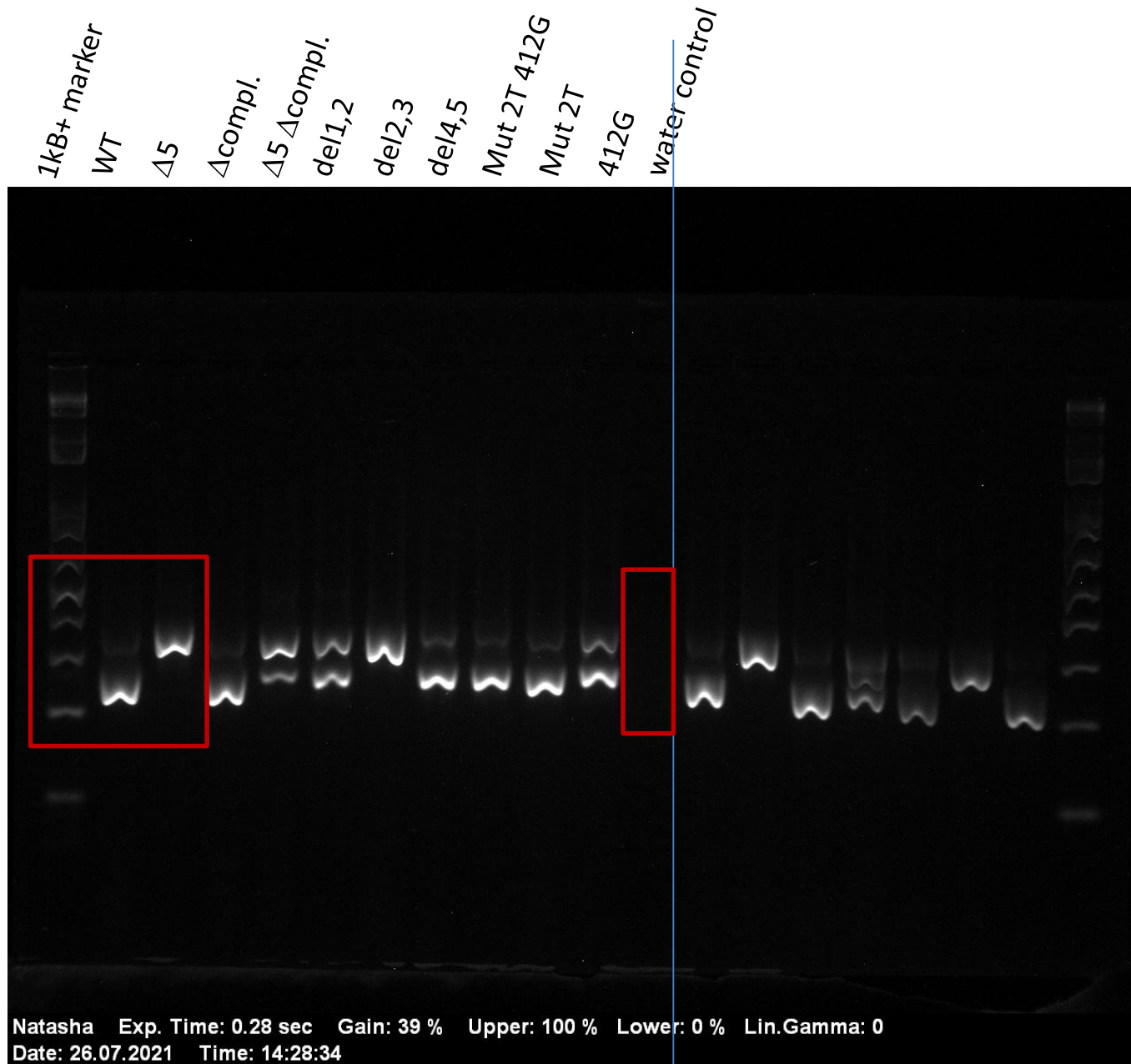
2= patient, 3= control



Full unedited blot for Figure 1B:  
Western blotting patient cells



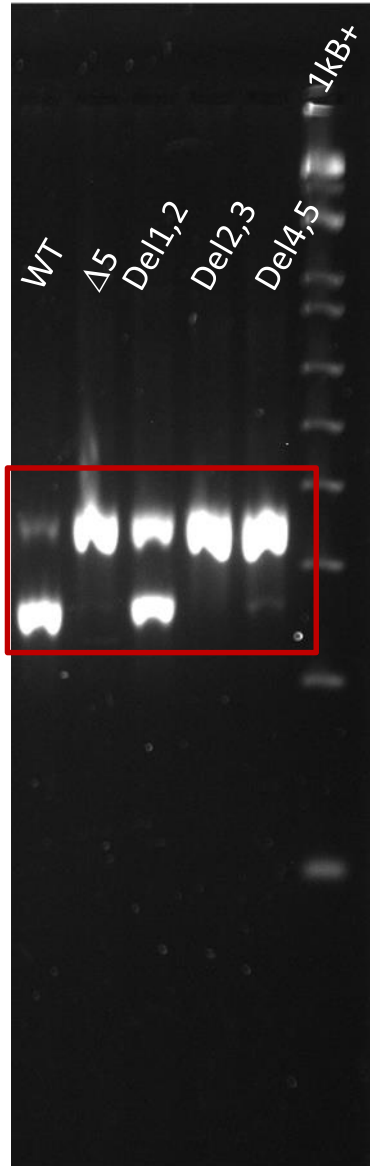
Full unedited gel for Figure 1B: RAD50 minigene (MG), agarose gel, PCR on cDNA



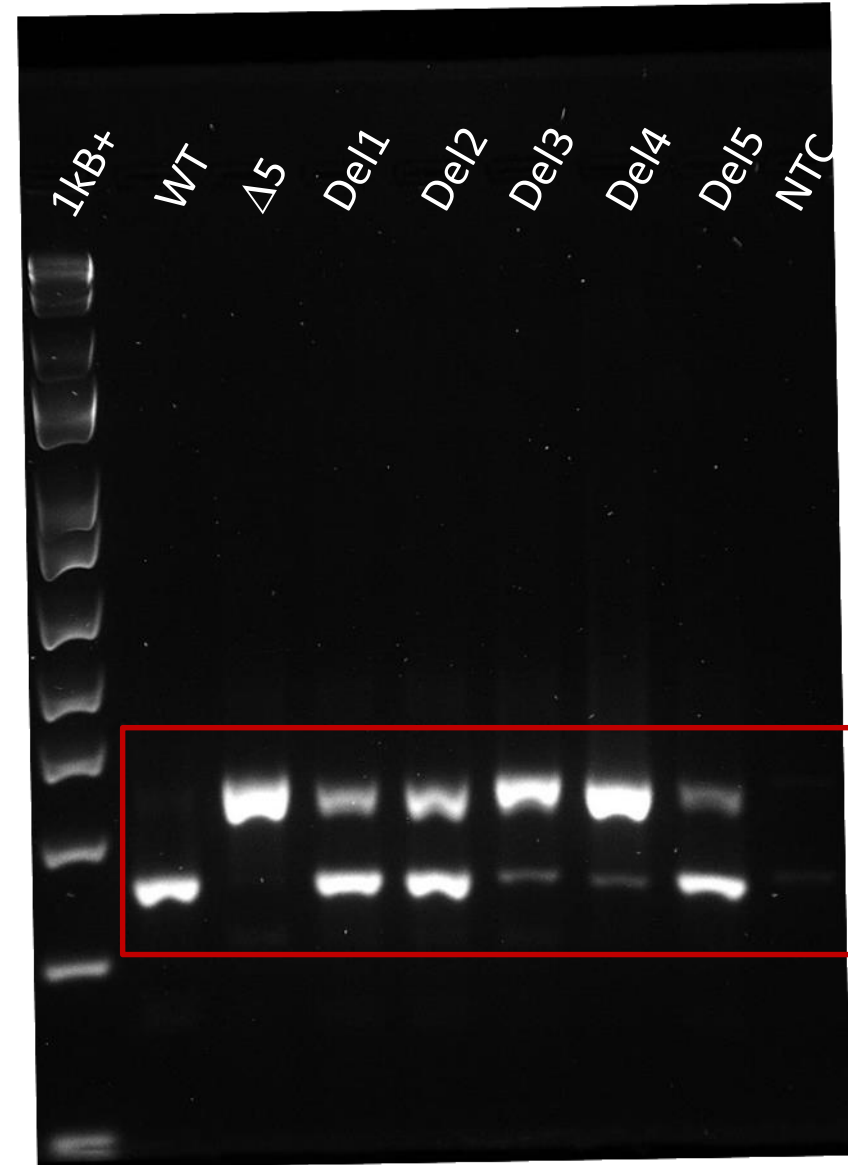
Full unedited gel for Figure 1C:

MG; agarose gel, PCR on cDNA

Double dels

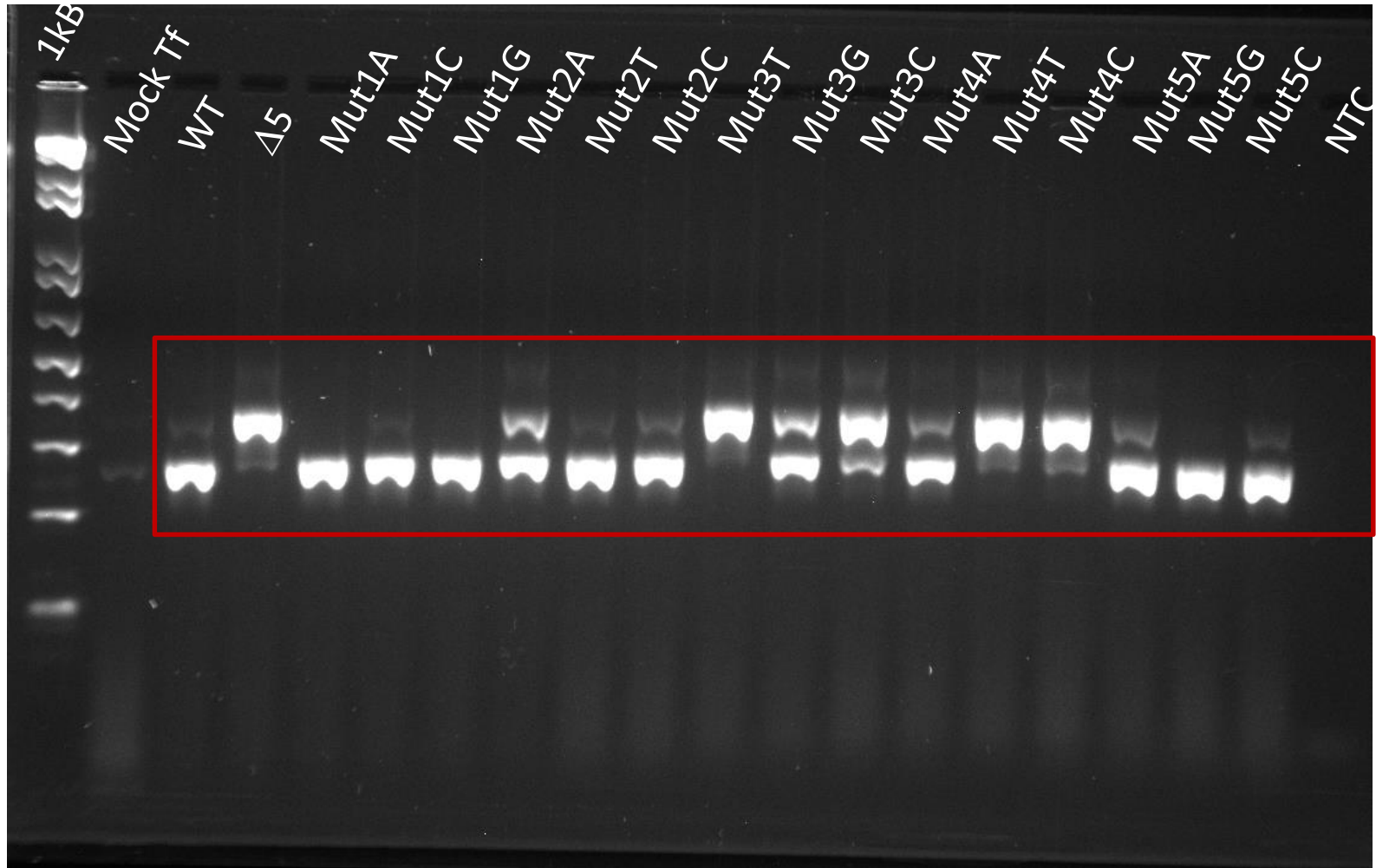


Single dels

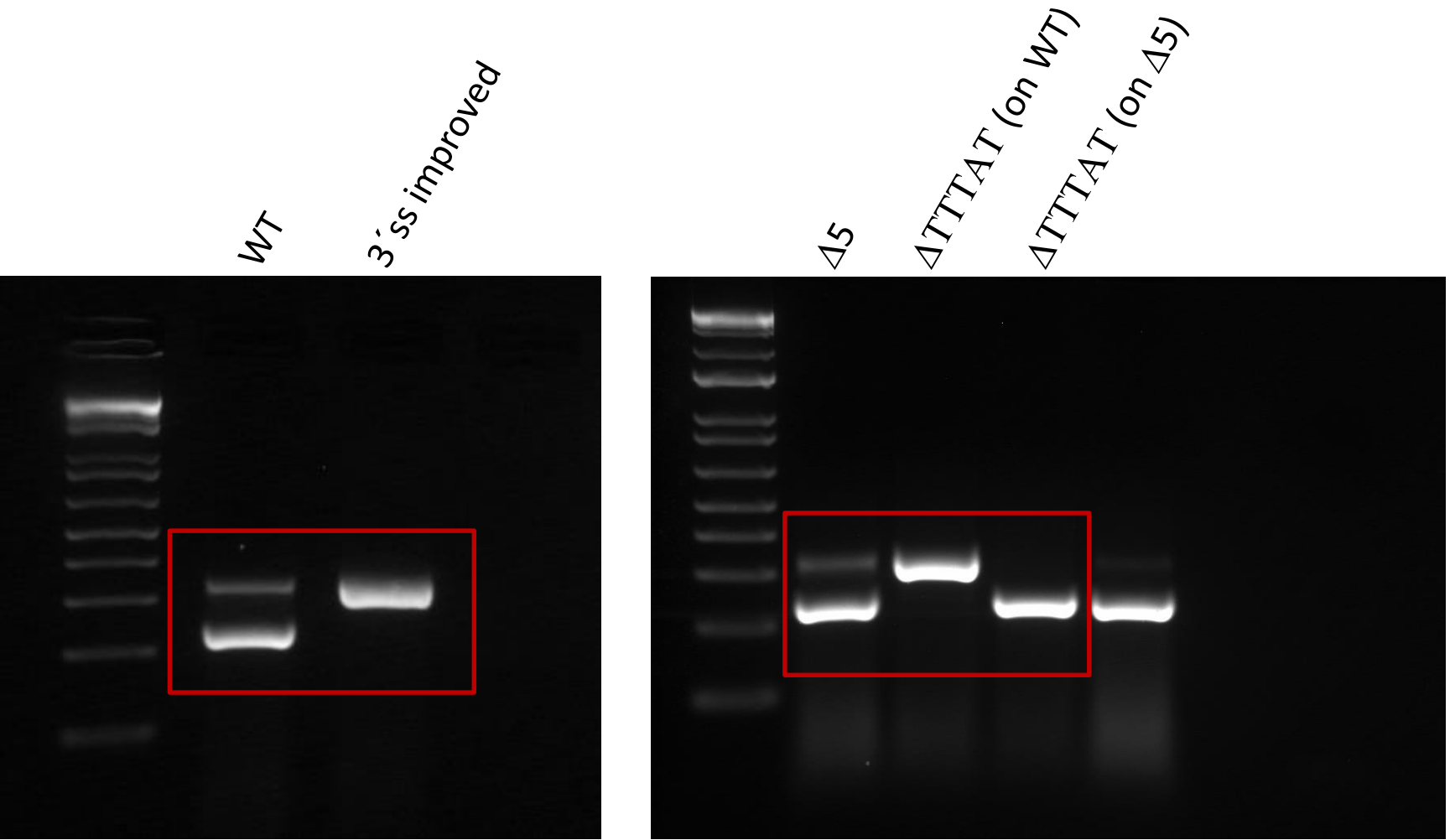


Full unedited gel for Figure 1C: continued

Single nucleotide substitutions MG; agarose gel, PCR on cDNA

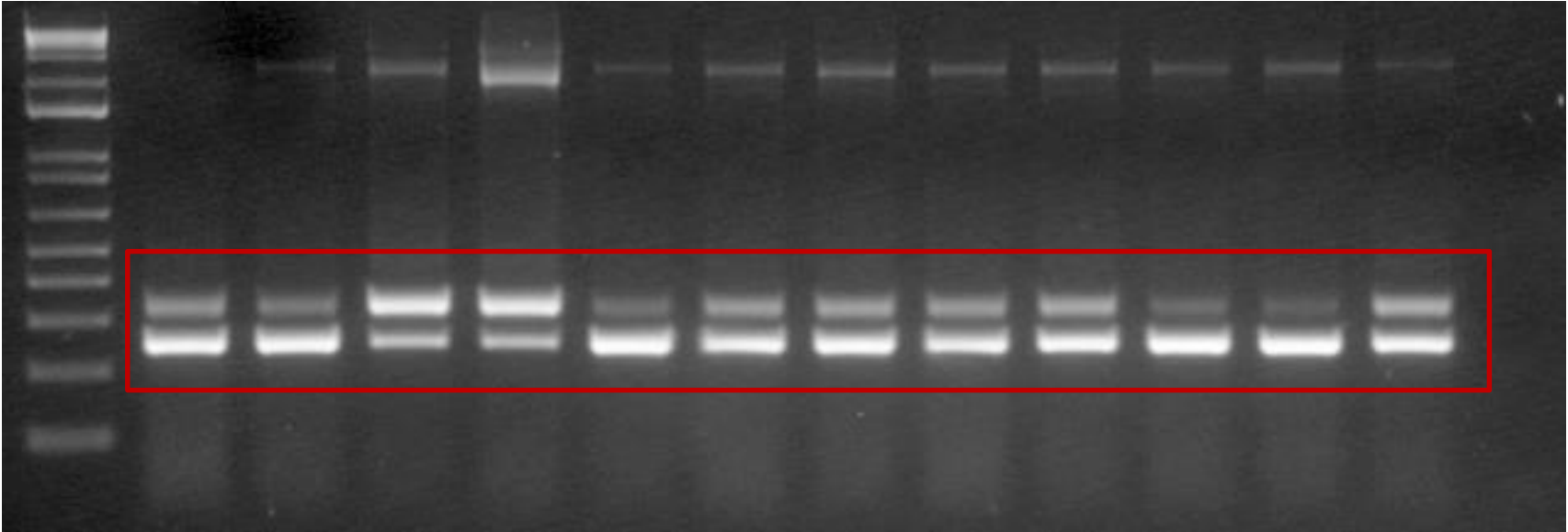


Full unedited gels for Figure 1E: PPT mutation MG; agarose gel, PCR on cDNA

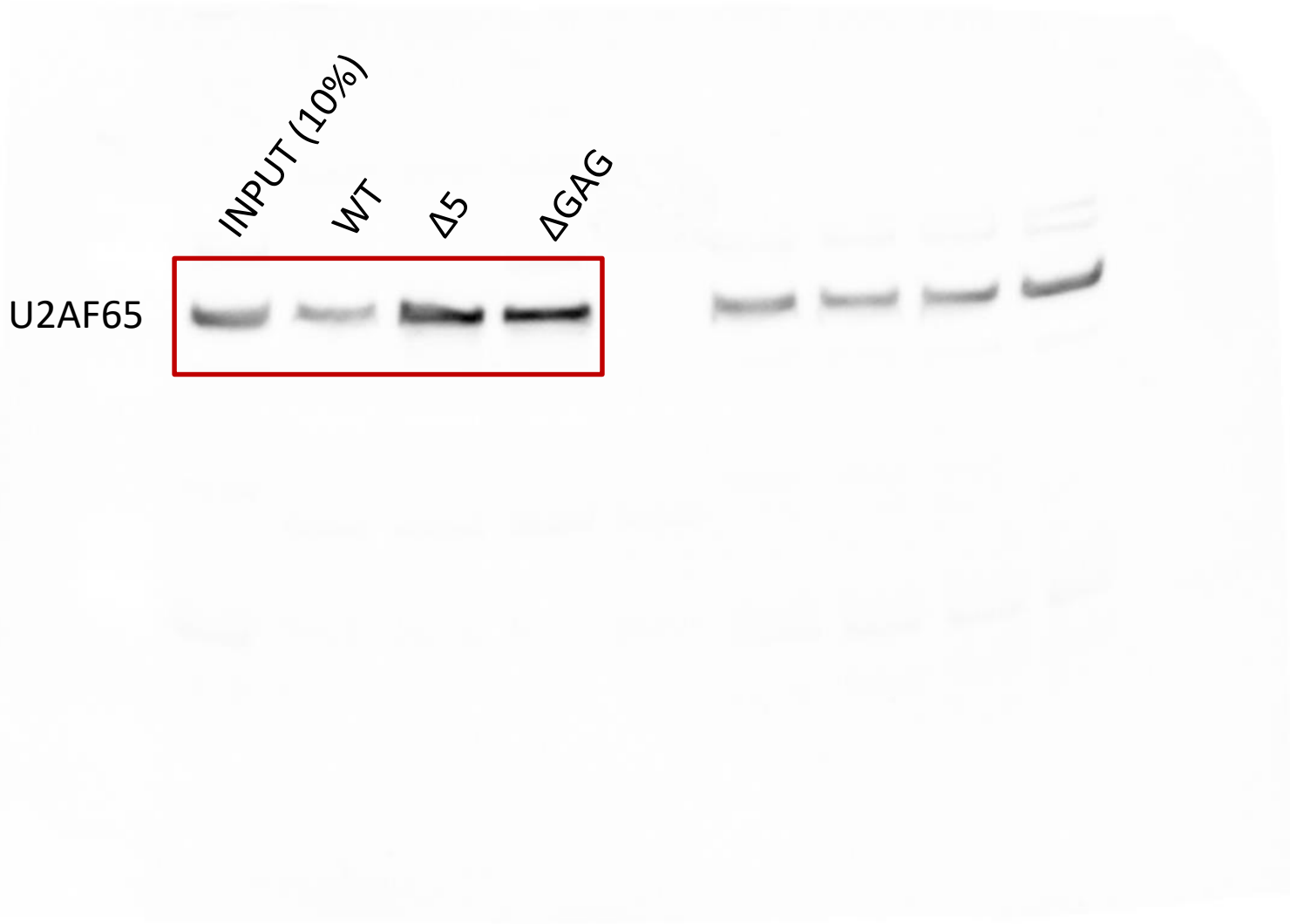


Full unedited gel for Figure 1F: splice factor titration, MG, agarose gel PCR on cDNA

WT \* SF7 \* U2AF35 \* U2AF65 \* hnRNPA1 \* hnRNPA2 \* hnRNPH \* PTB1 \* PTB4 \* CUGBP \* ETR3 \* CLF6

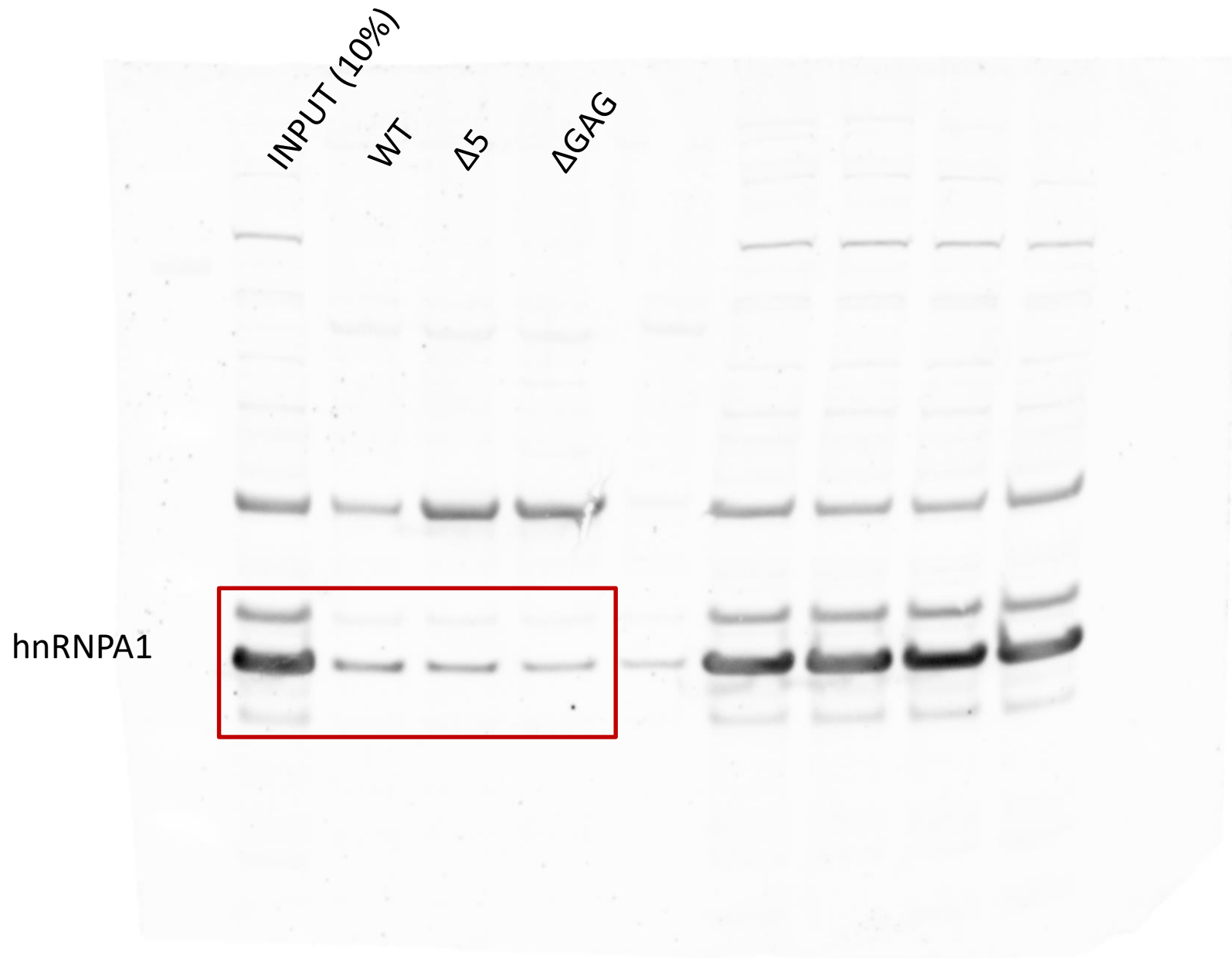


Full unedited blot for Figure 1G: pull down western, upper panel

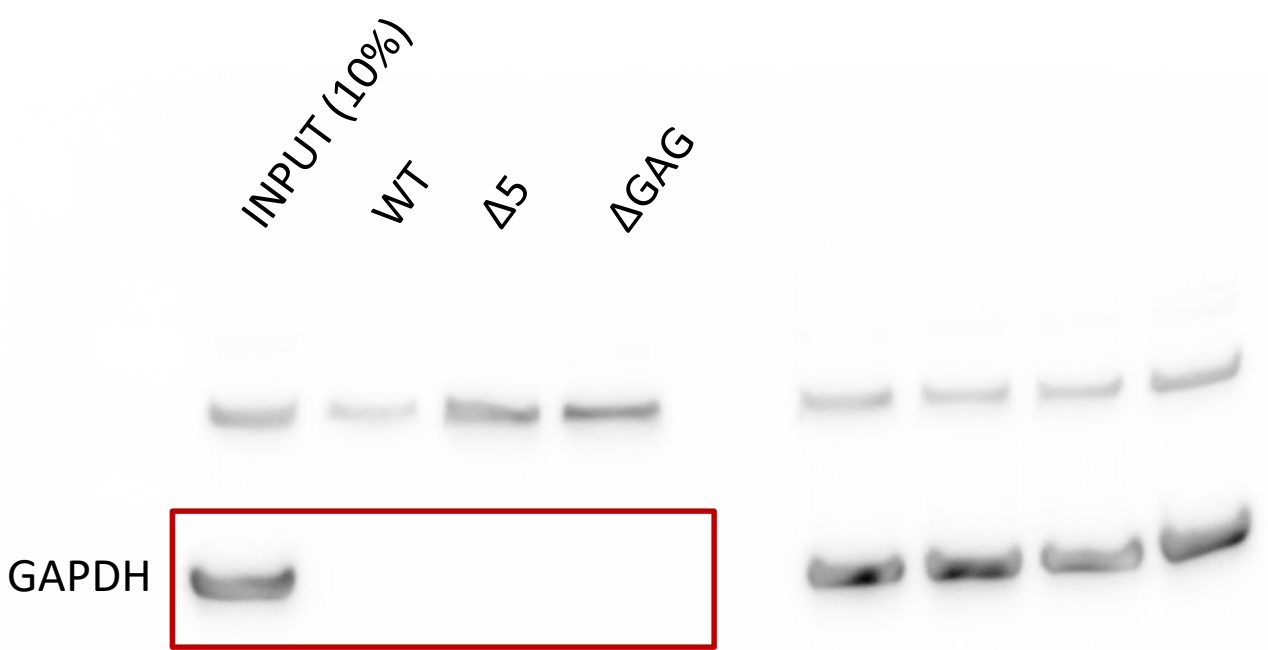




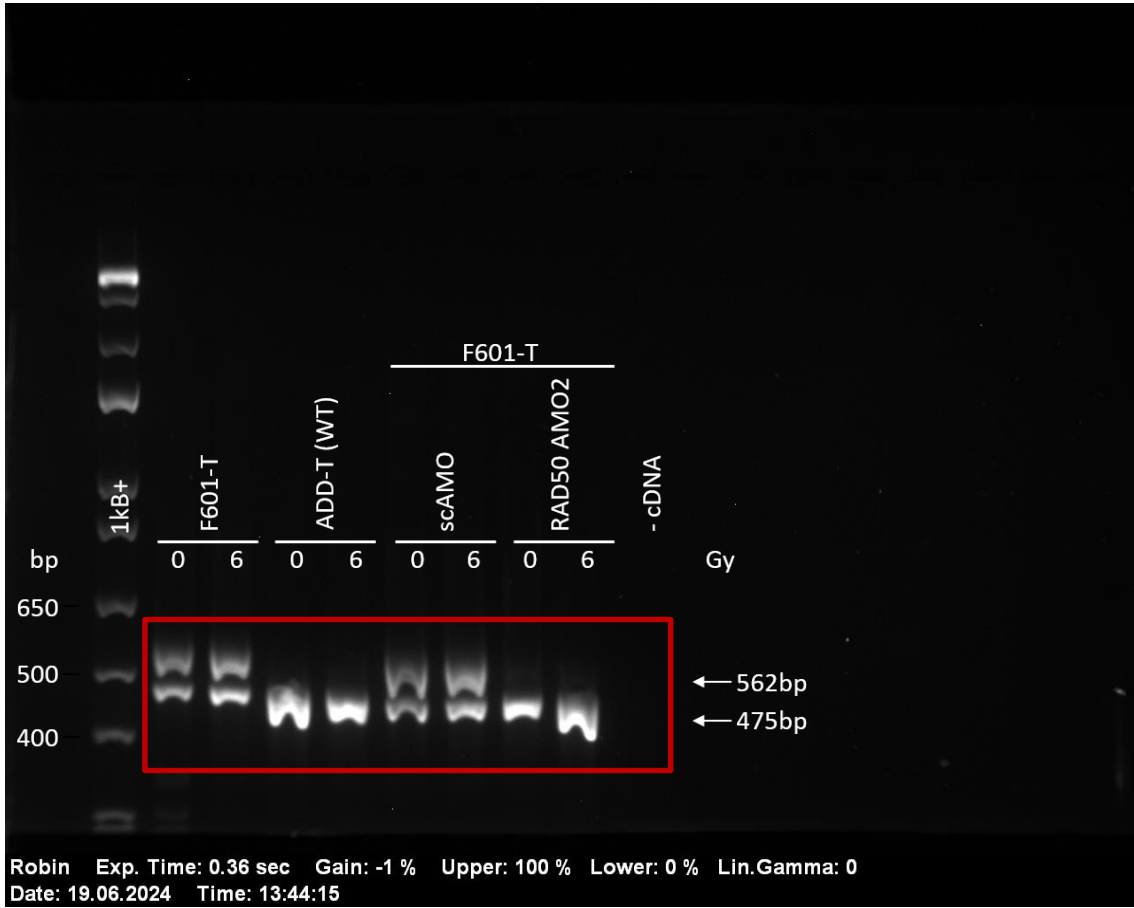
Full unedited blot for Figure 1G: pull down western, middle panel



Full unedited blot for Figure 1G: pull down western, lower panel

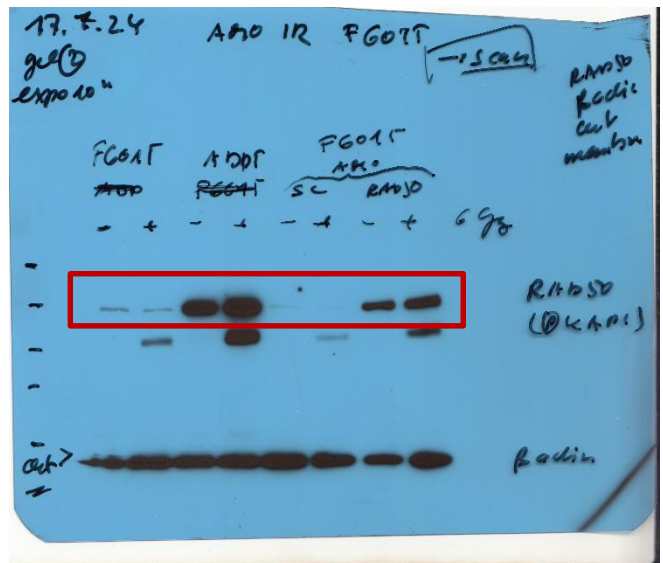


Full unedited gel for Figure 1I: IR AMO, agarose gel (PCR on cDNA)



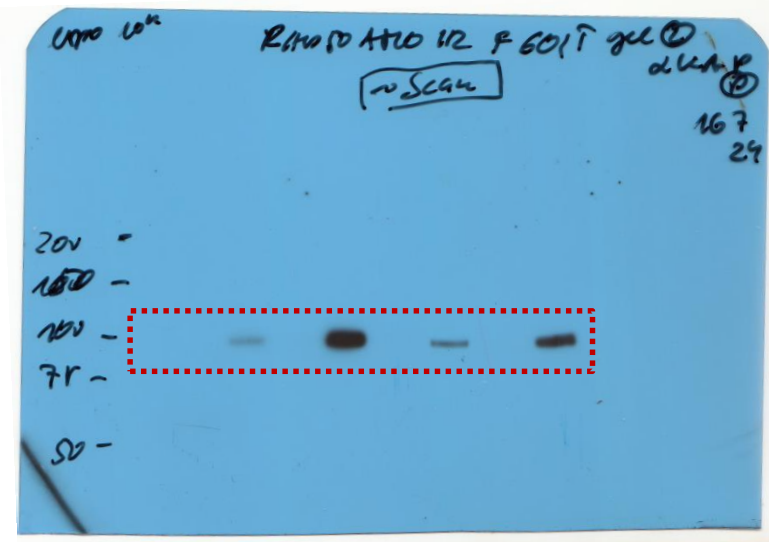
Full unedited blots for 1I: IR AMO, western blots partA

RAD50 &  $\beta$ -actin (cut membrane)



higher exposure

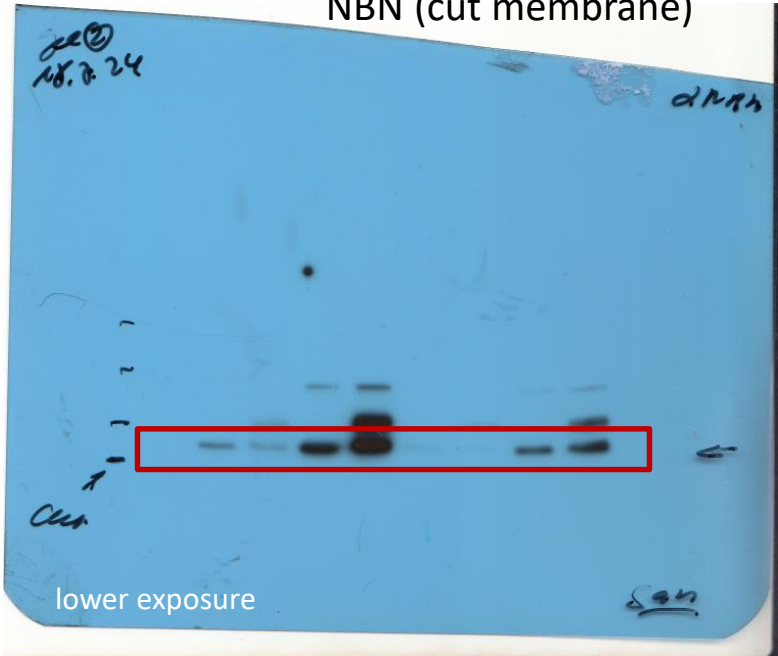
P-KAP (S824)



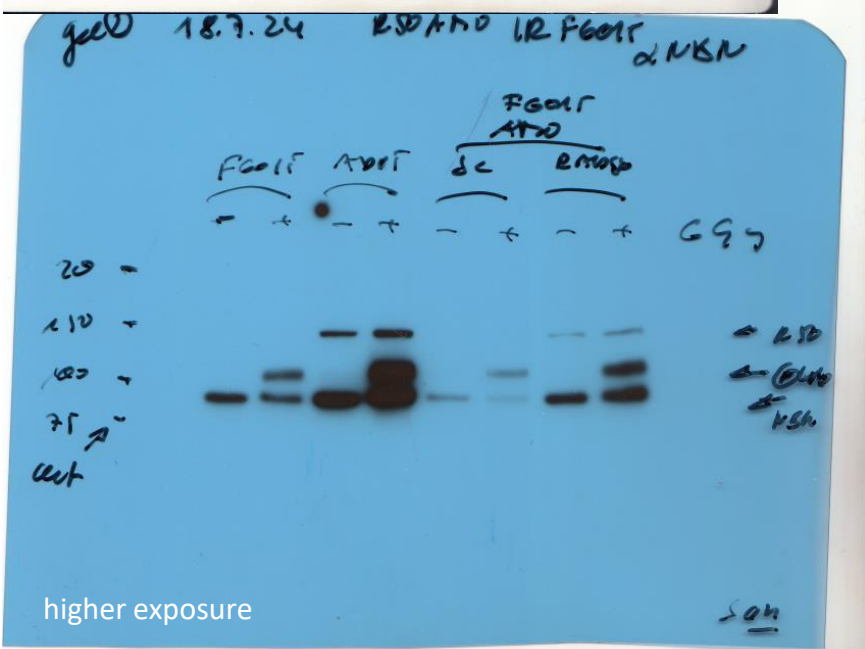
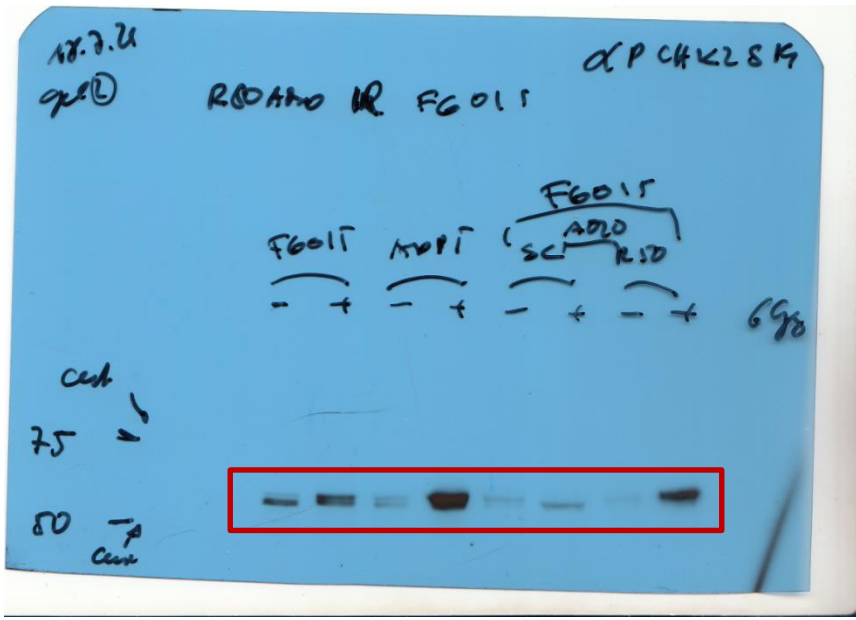
lower exposure

Full unedited blots for Figure 1I: IR AMO, western blots partB

NBN (cut membrane)

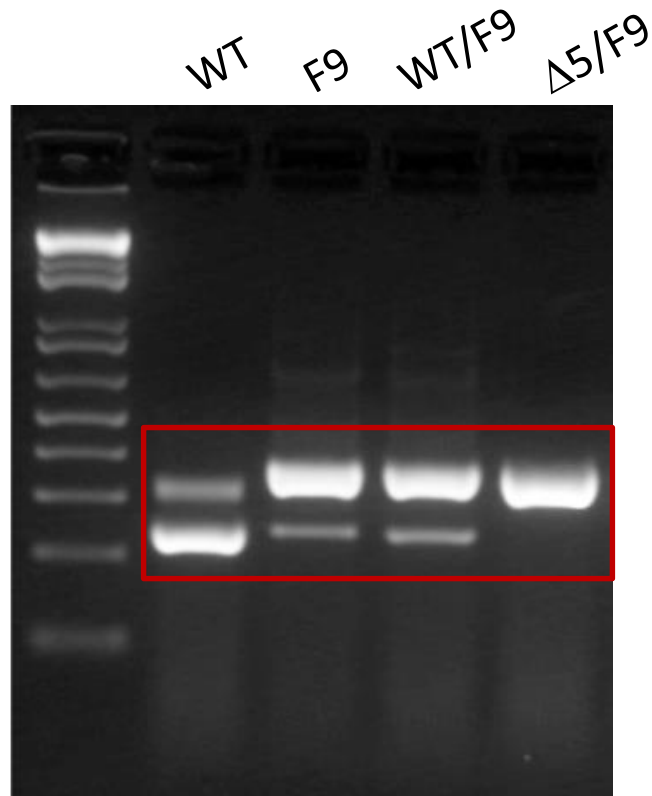


P-CHEK2 (S-19) (cut membrane)

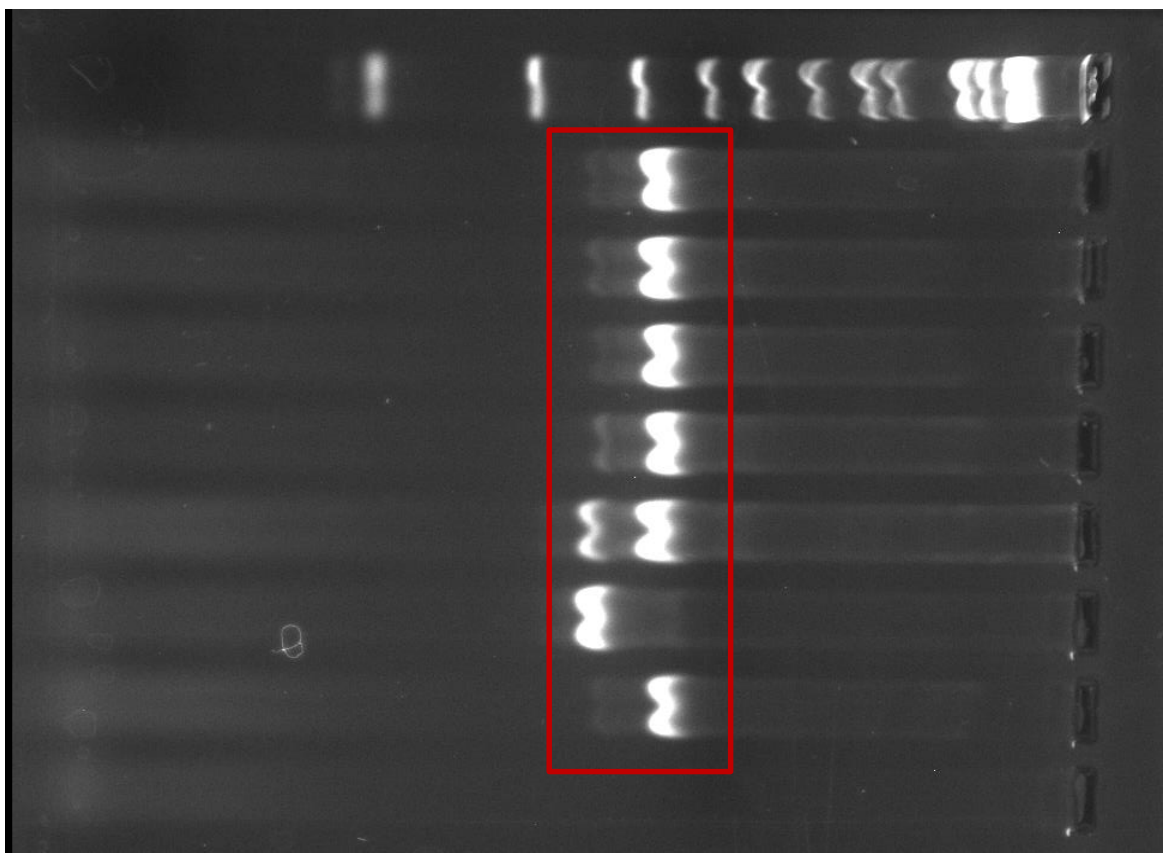


Full unedited gel for Suppl. Figure 2A

Transposition of RAD50 3' splice region to unrelated exon



Full unedited blot for Suppl. Figure 2F: Distinct deletions in the  $\Delta 5$  sequence



1kB+

Del1,2,3

Del1,2,3,4

Del2,3,4

Del3,4,5

Del3

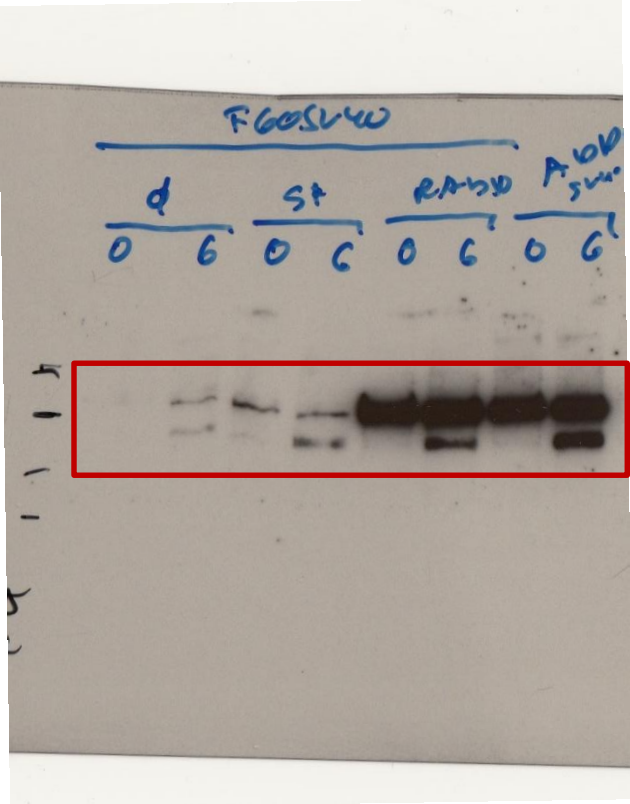
WT

$\Delta 5$

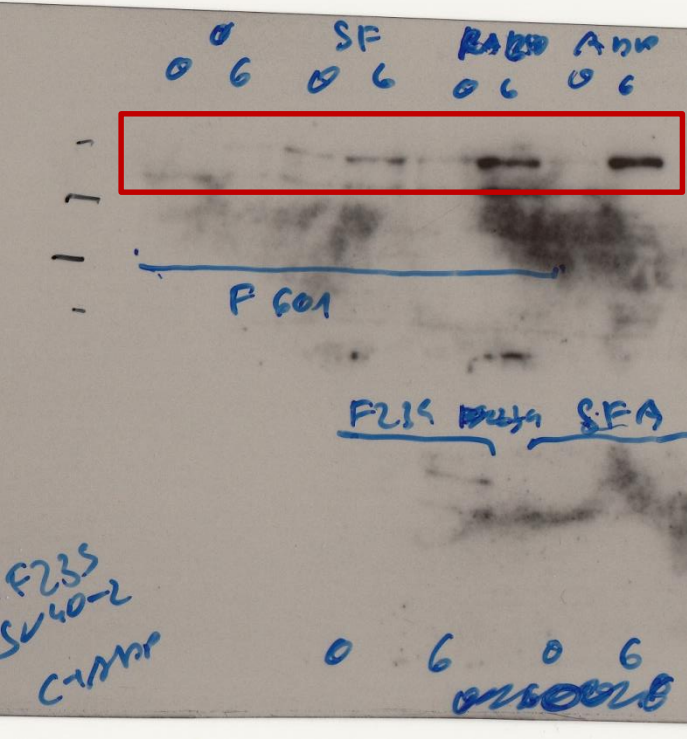
NTC

Full unedited blots for SUPPL Figure 4: : Functional complementation of RAD50-deficient fibroblasts partA

**RAD50  
P-KAP1 S-824**



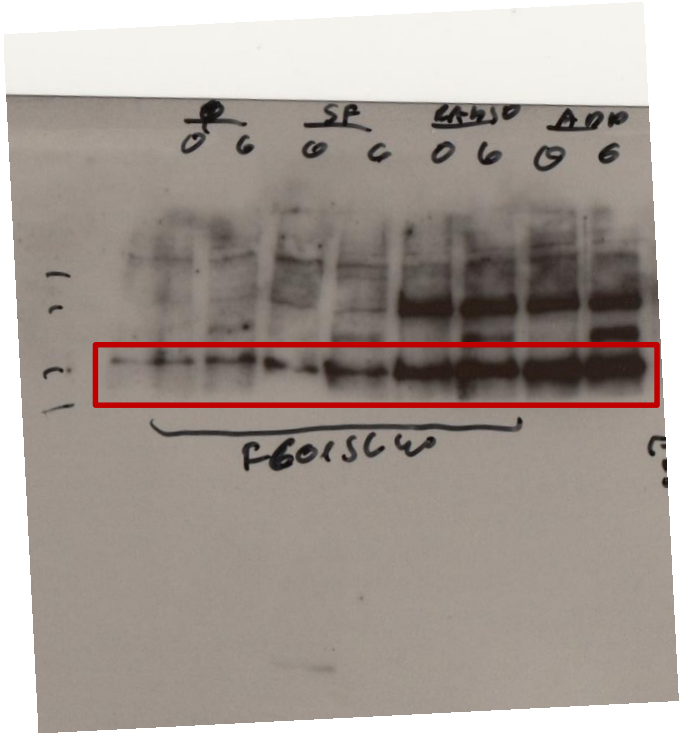
**P-CHK2 S-19**





Full unedited blots for SUPPL Figure 4: : Functional complementation of RAD50-deficient fibroblasts partB

NBN



RAD50  
P-KAP1 S-824  
NBN

ACTB

