

Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase II processing in transcription-coupled nucleotide-excision repair

Article (Unspecified)

Nakazawa, Yuka, Sasaki, Kensaku, Mitsutake, Norisato, Matsuse, Michiko, Shimada, Mayuko, Nardo, Tiziana, Takahashi, Yoshito, Ohyama, Kaname Ohyama, Ito, Kosei, Mishima, Hiroyuki, Nomura, Masayo, Kinoshita, Akira, Ono, Shinji, Takenaka, Katsuya, Masuyama, Ritsuko et al. (2012) Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase II processing in transcription-coupled nucleotide-excision repair. *Nature Genetics*, 44 (5). pp. 586-592. ISSN 1061-4036

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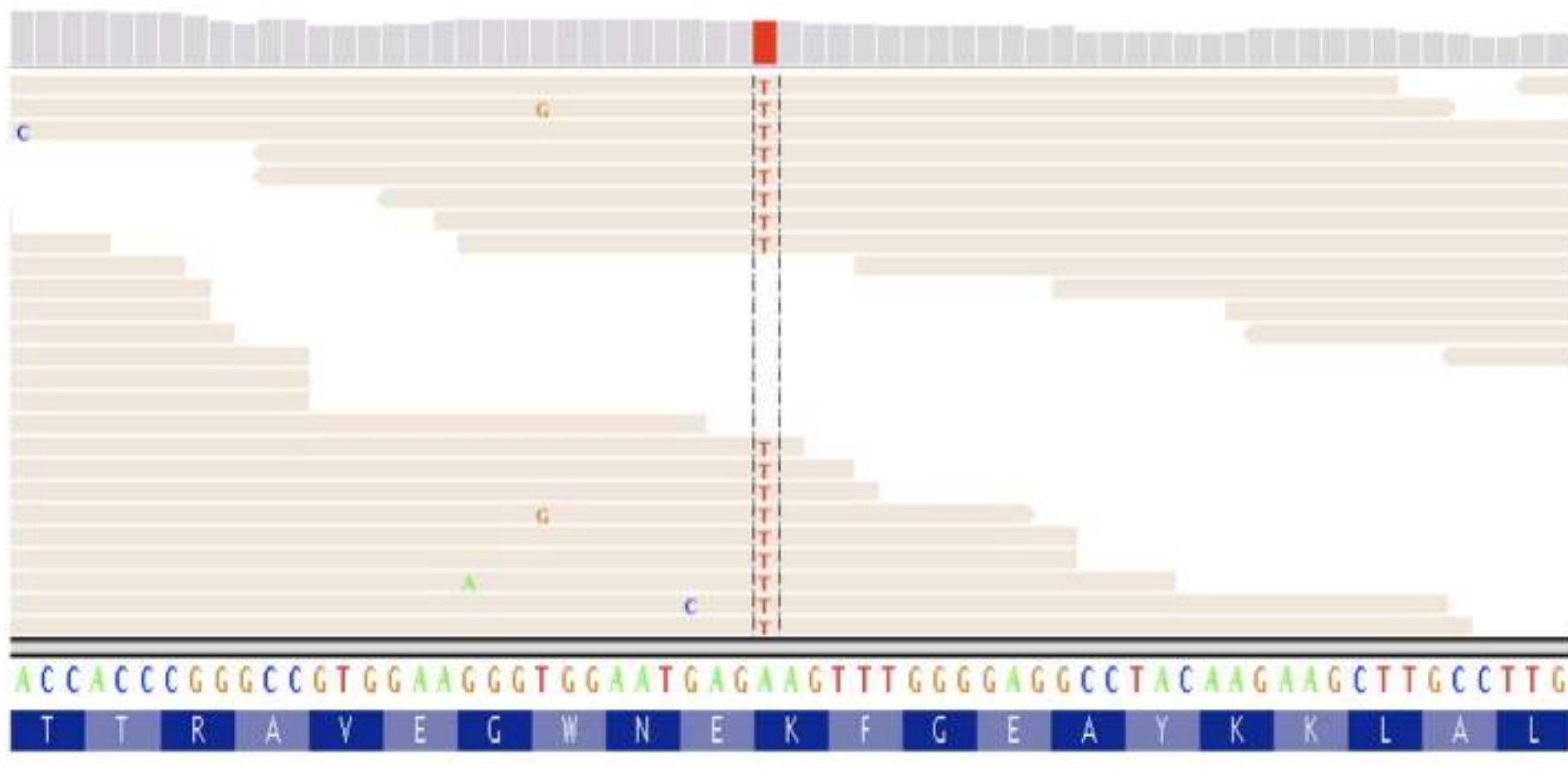
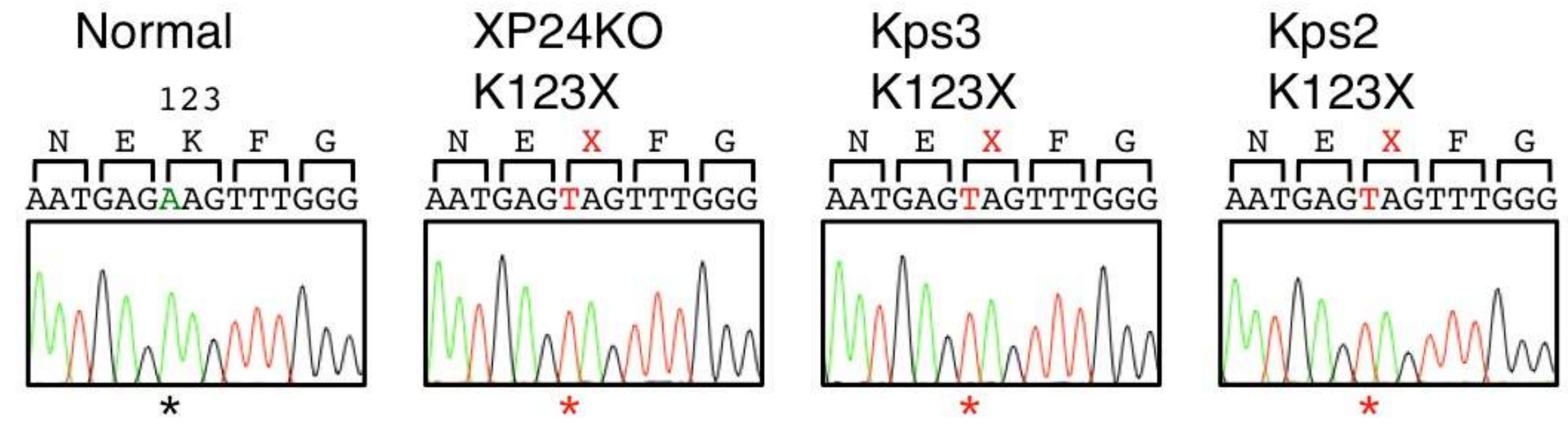
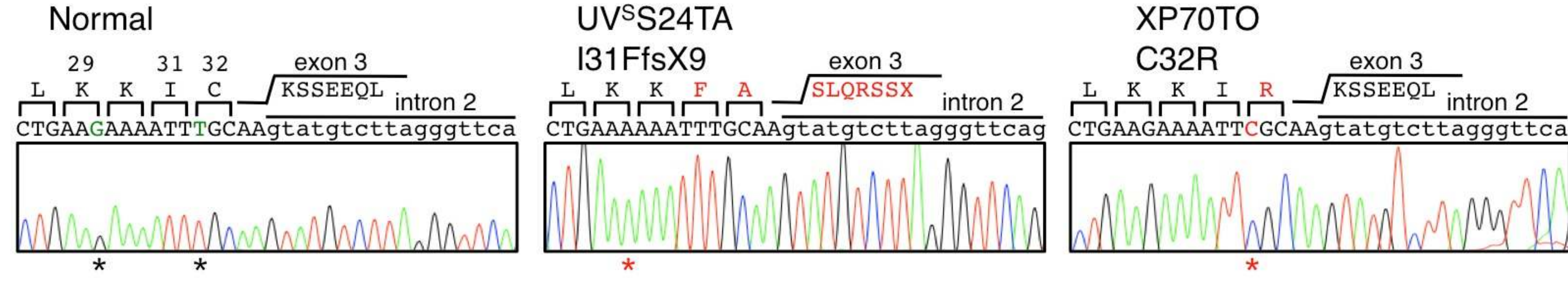
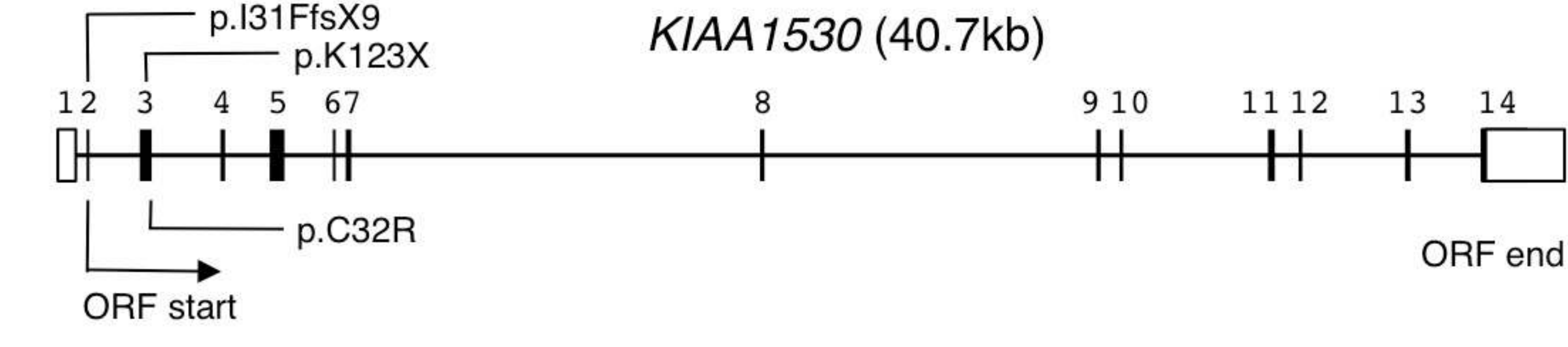
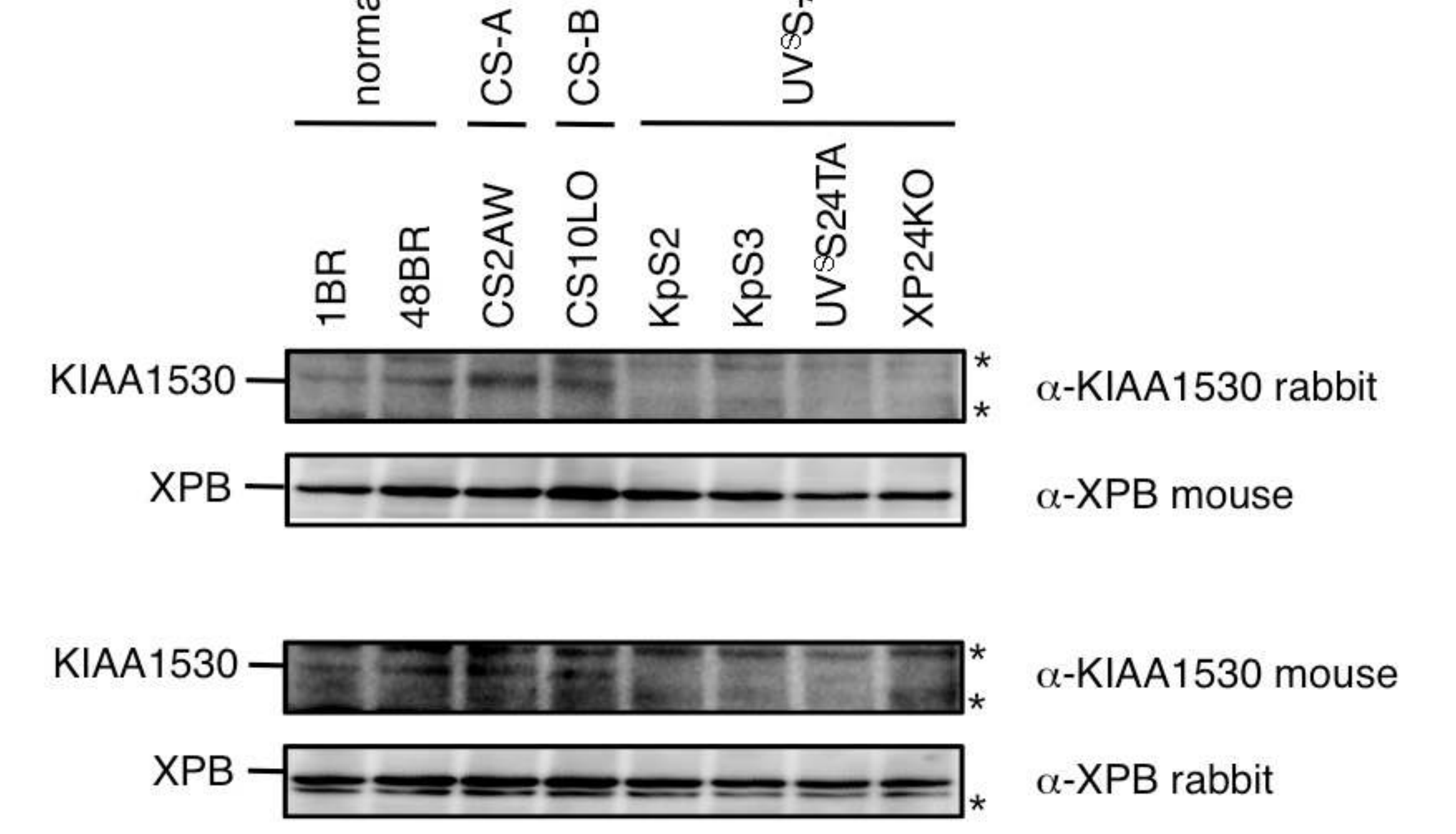
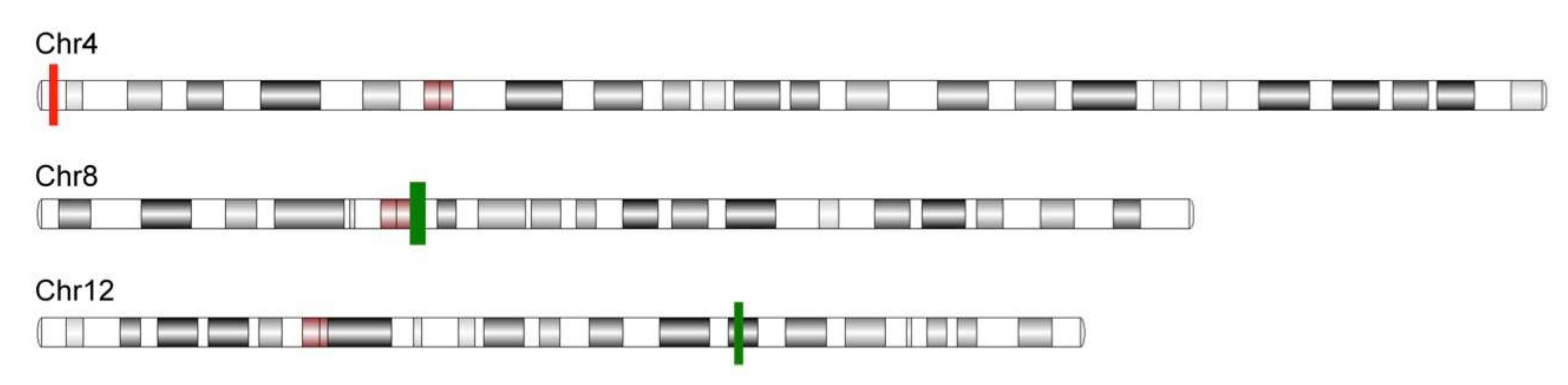
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a Kps3 *KIAA1530:c.367A>T***b****c****d****e****f****Fig.1**

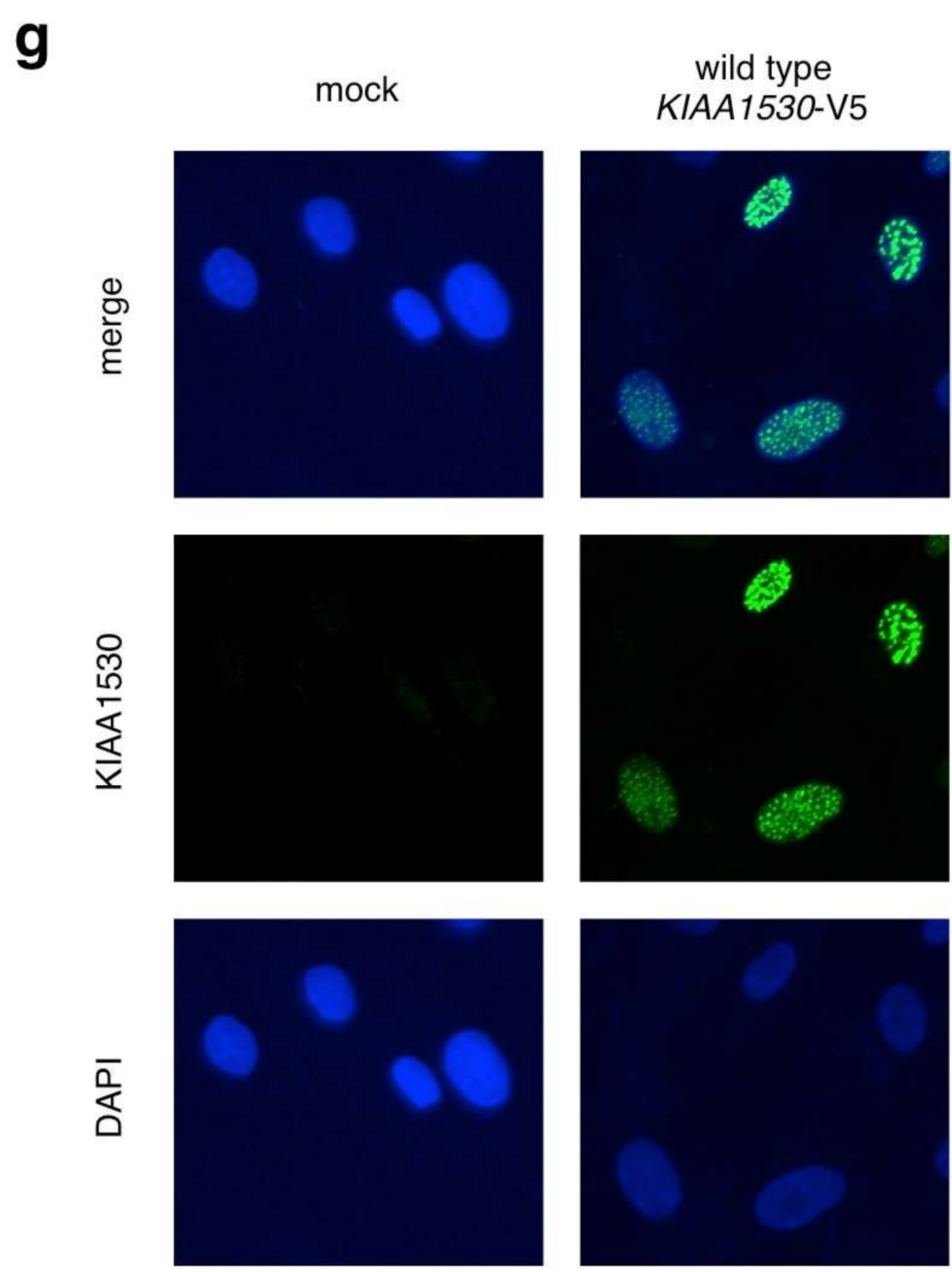
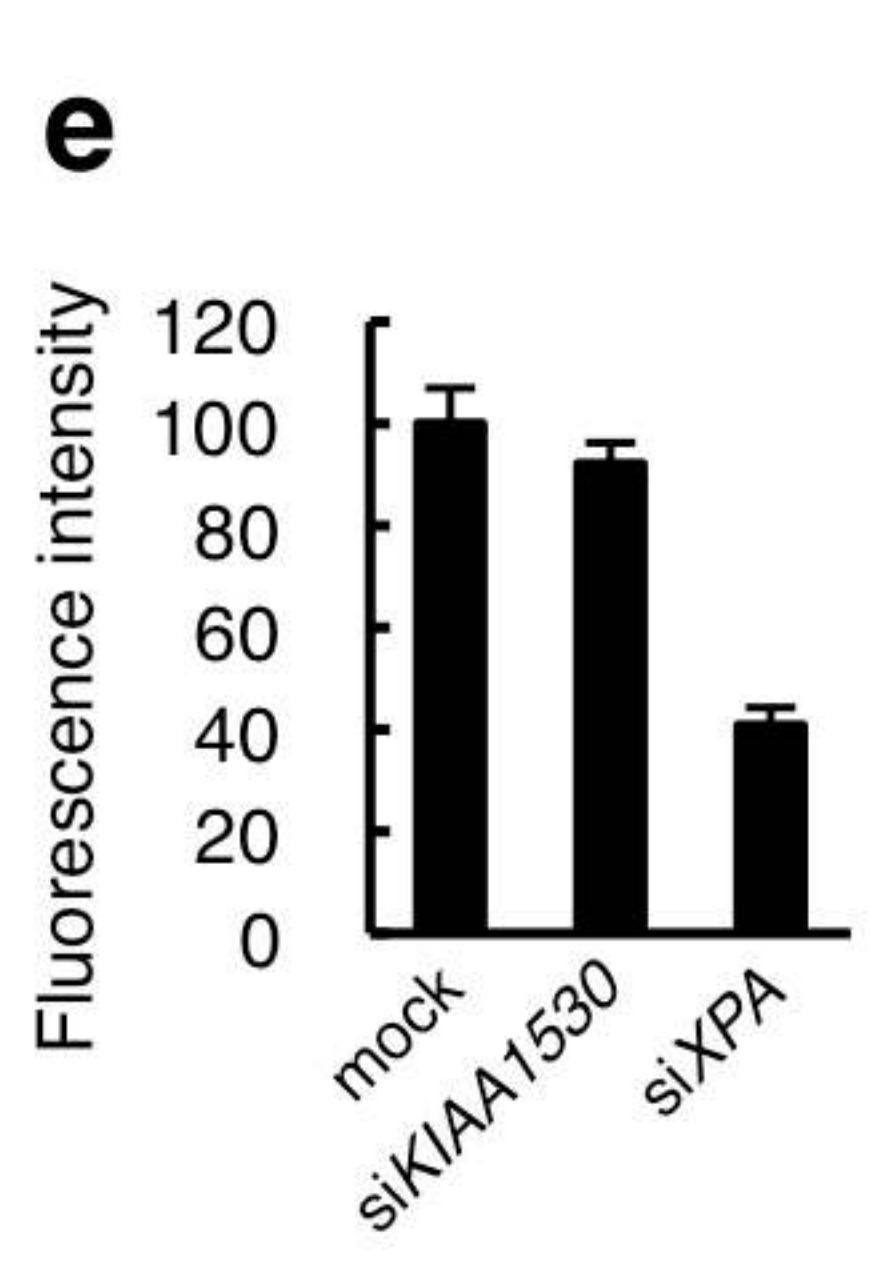
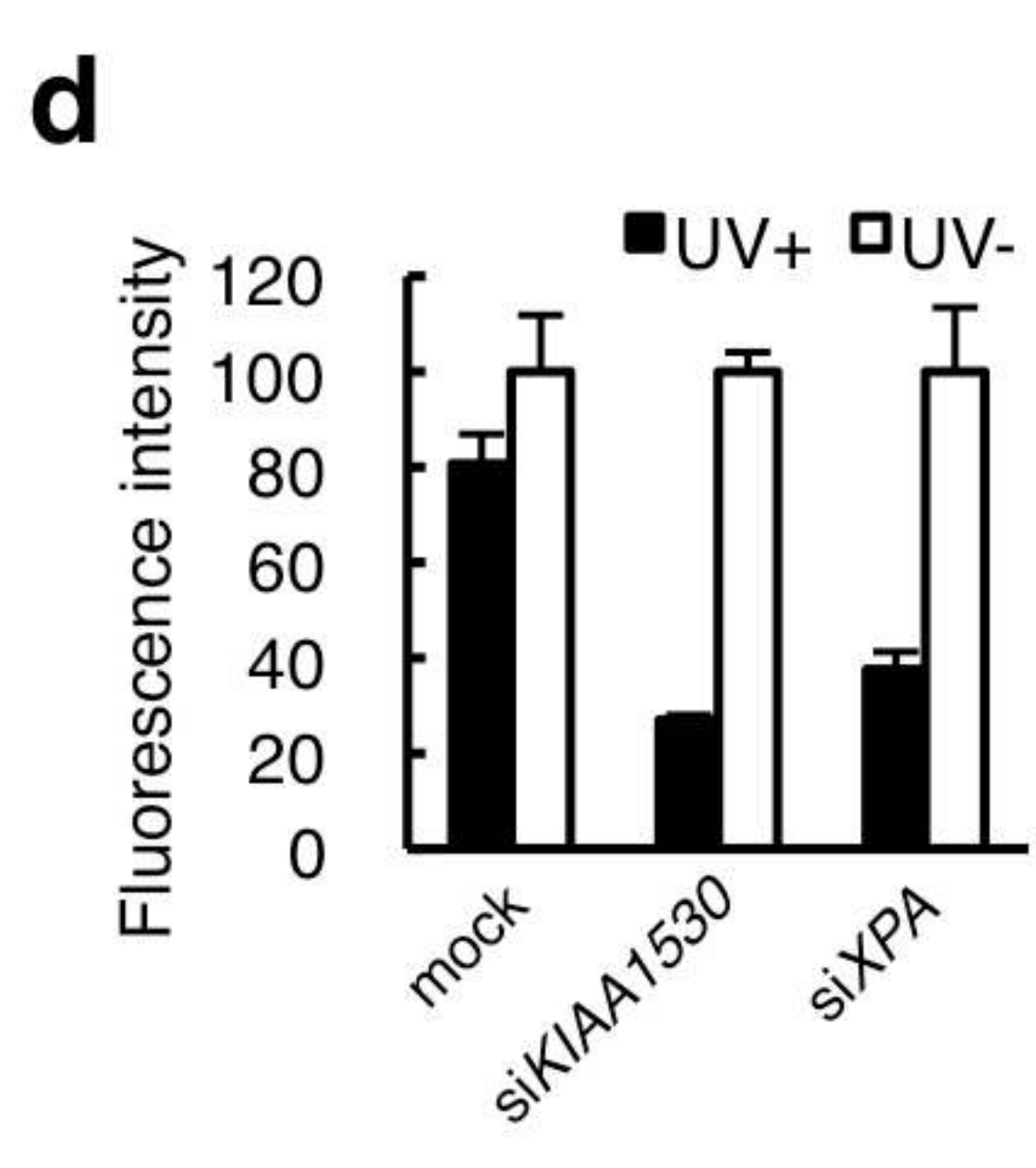
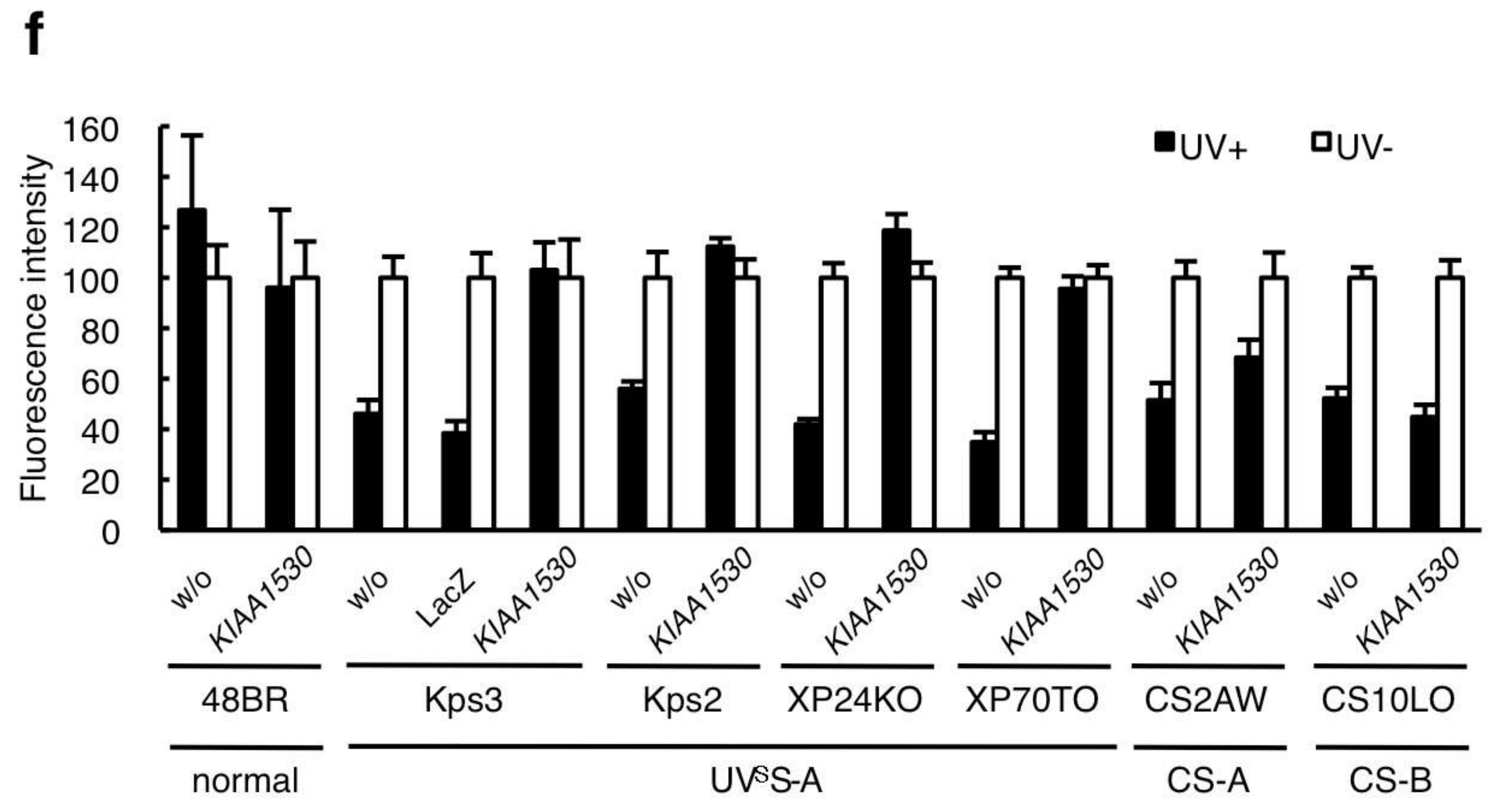
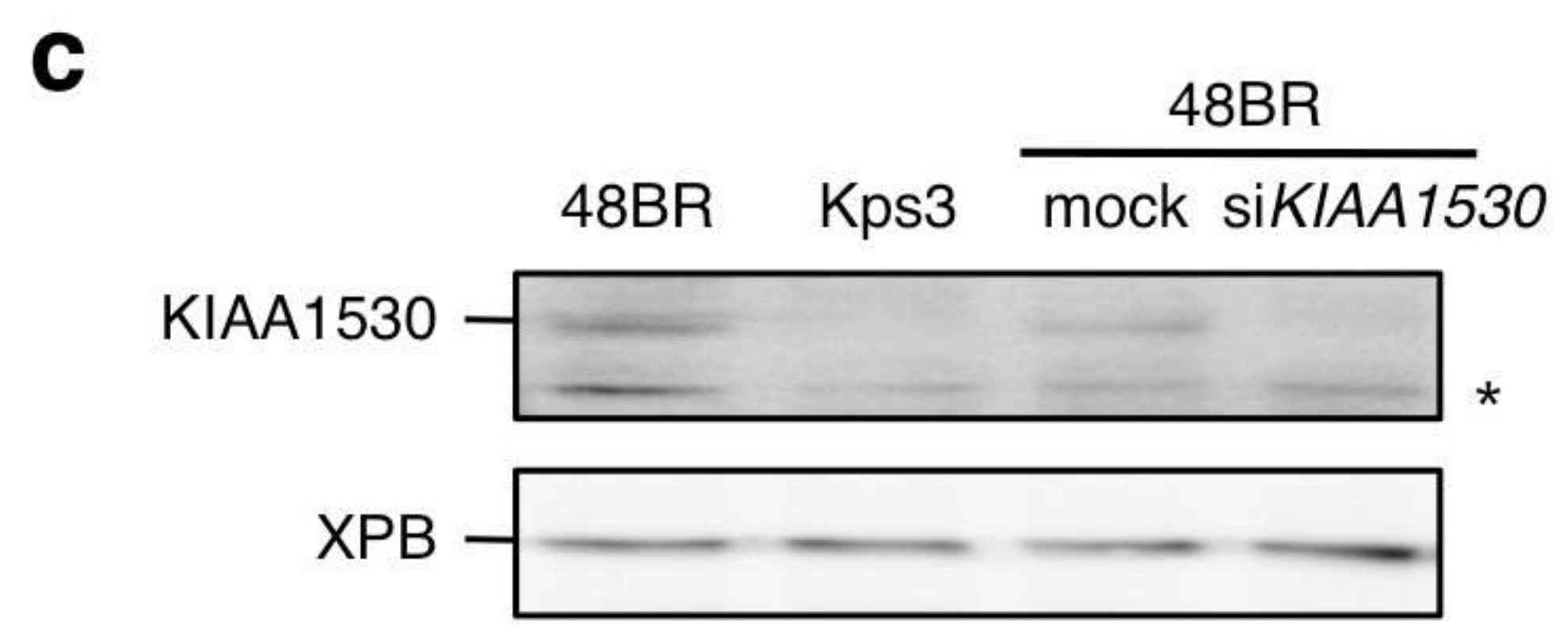
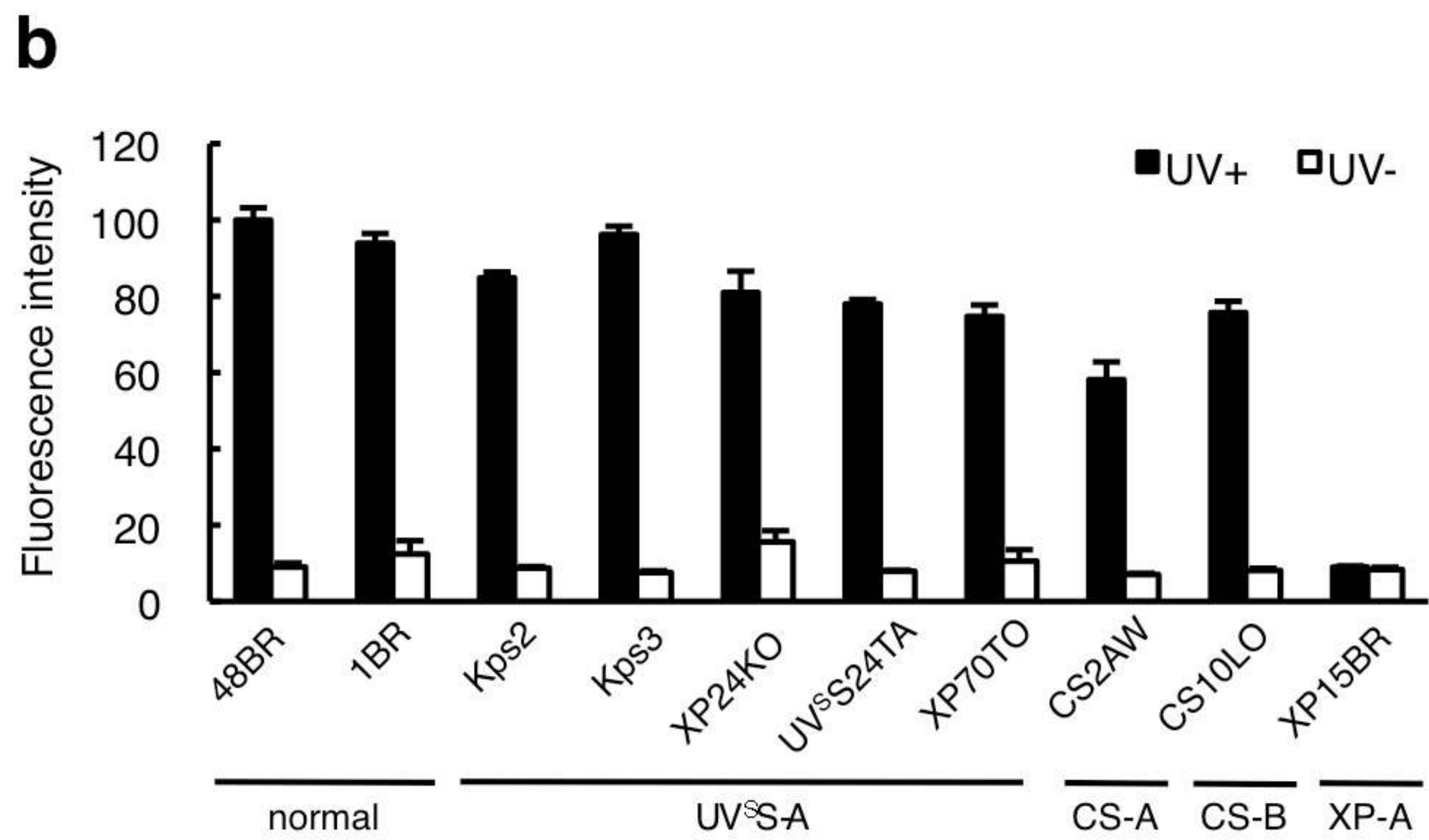
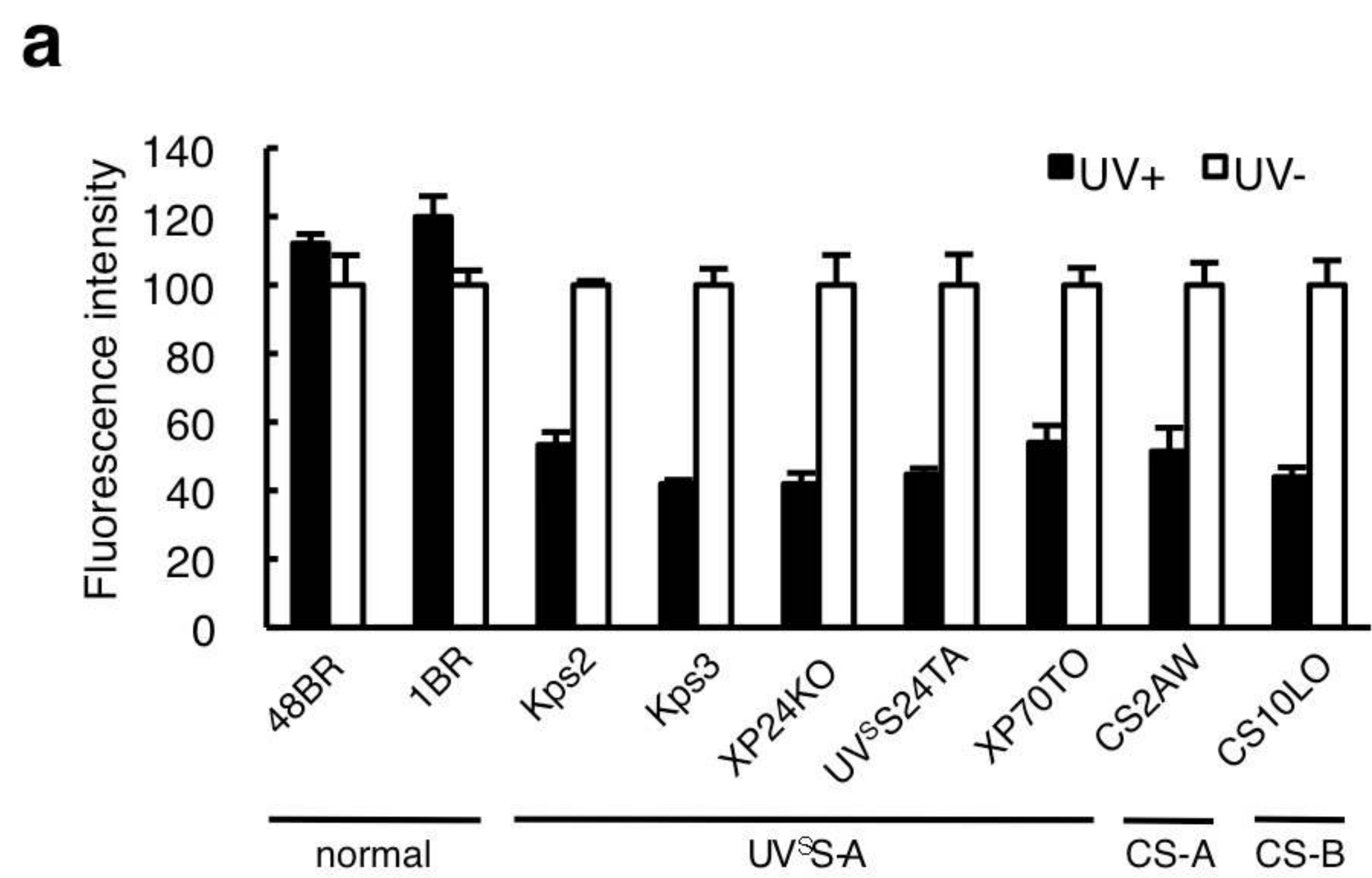


Fig.2

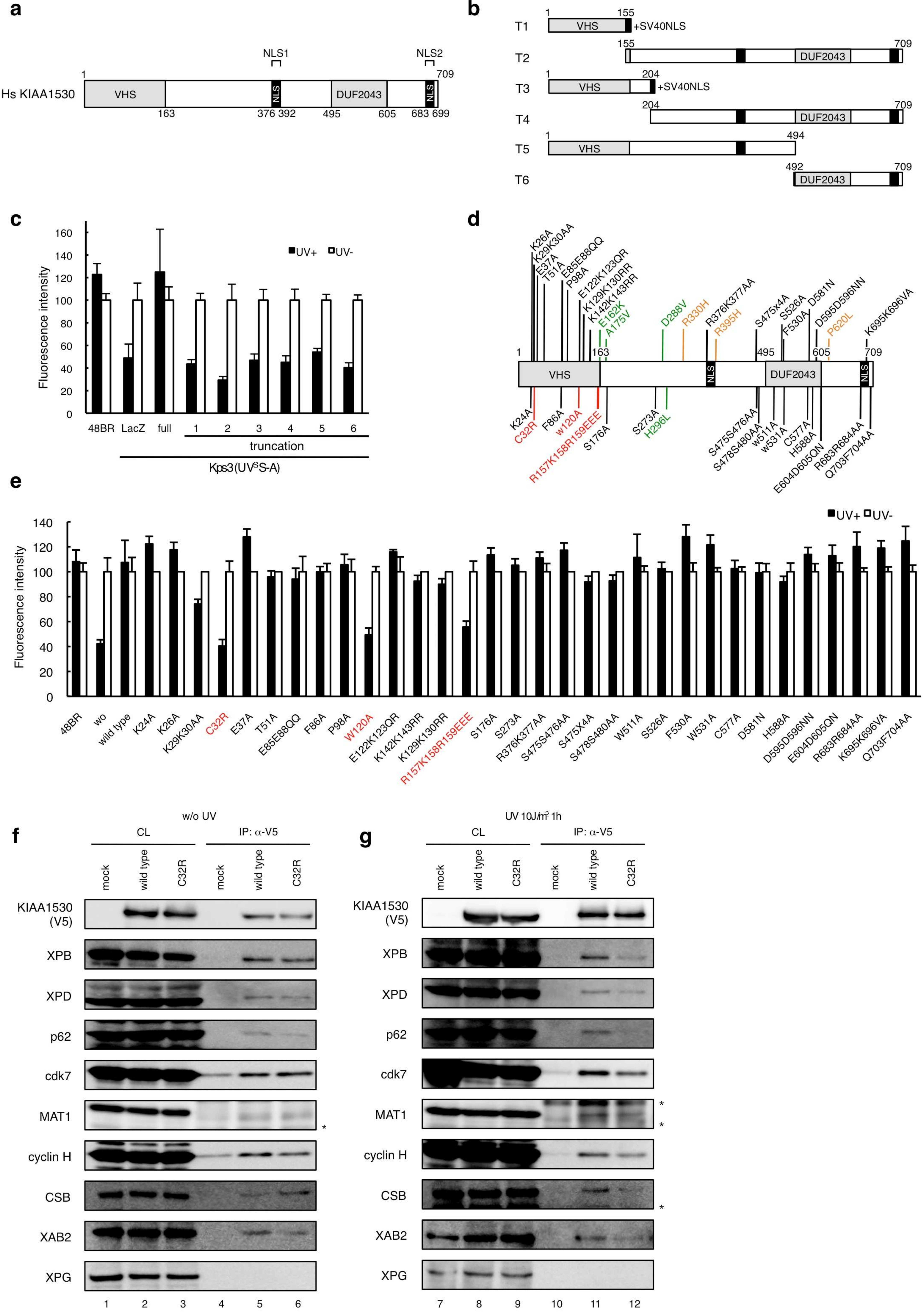


Fig.3

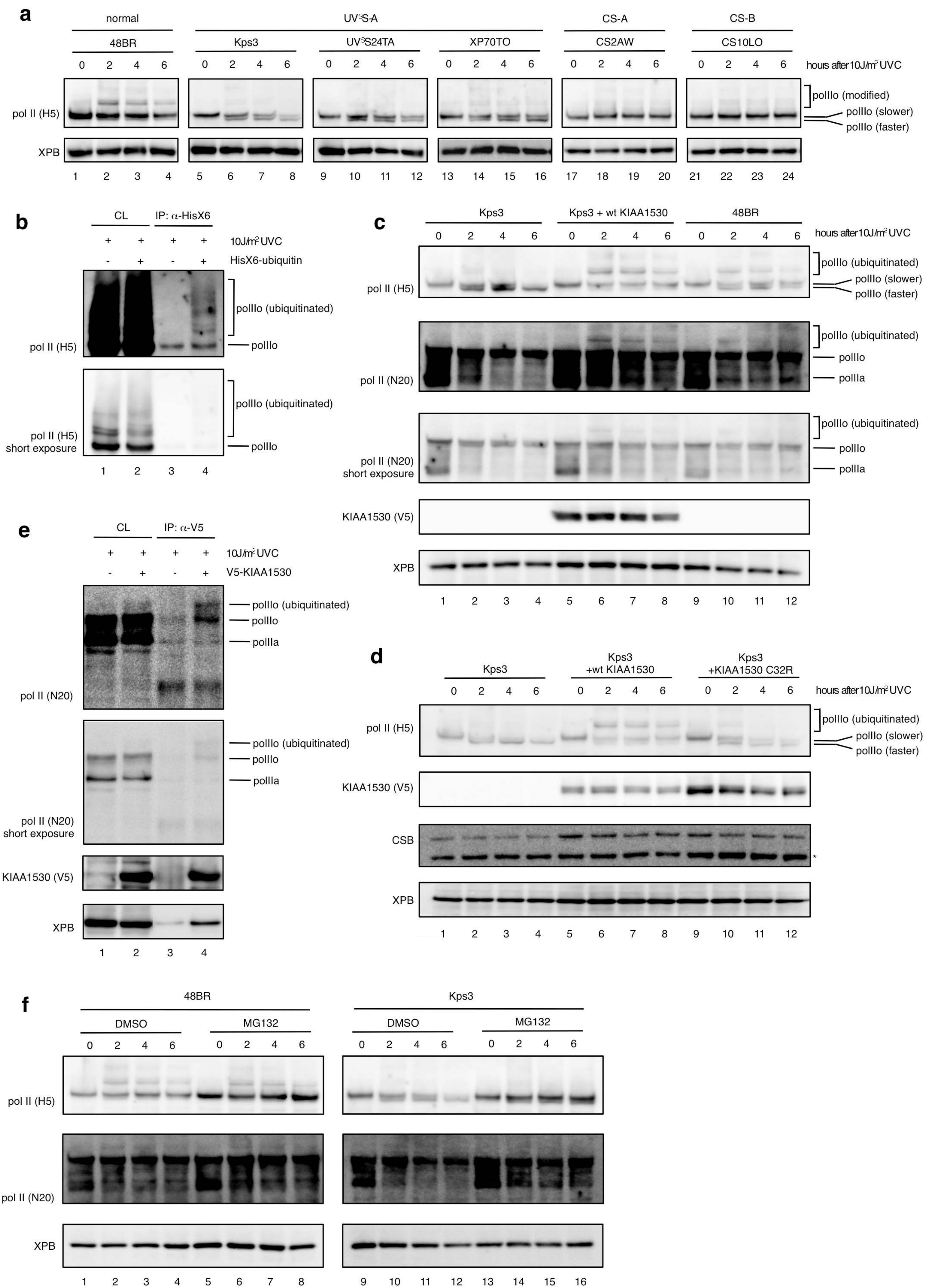
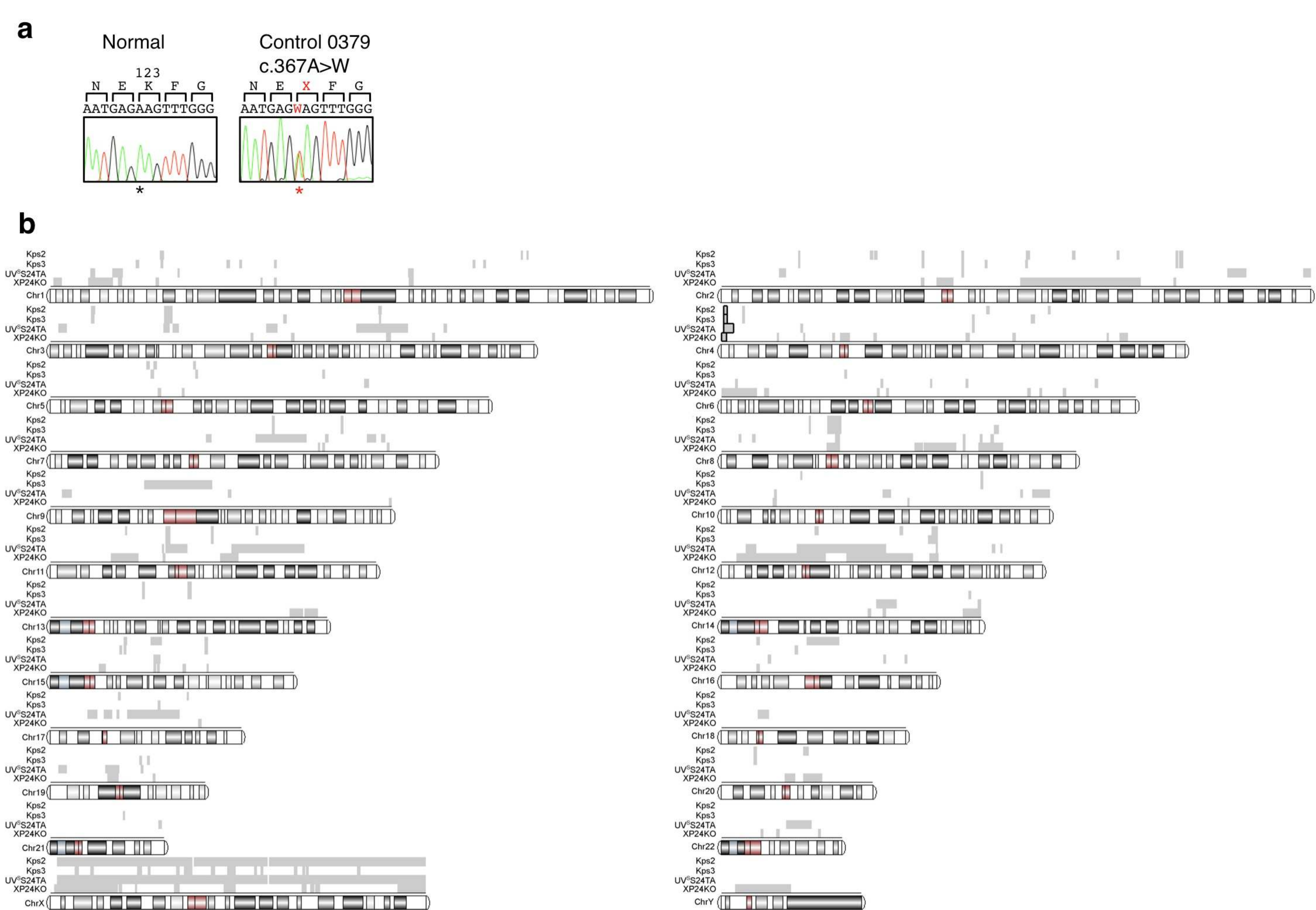
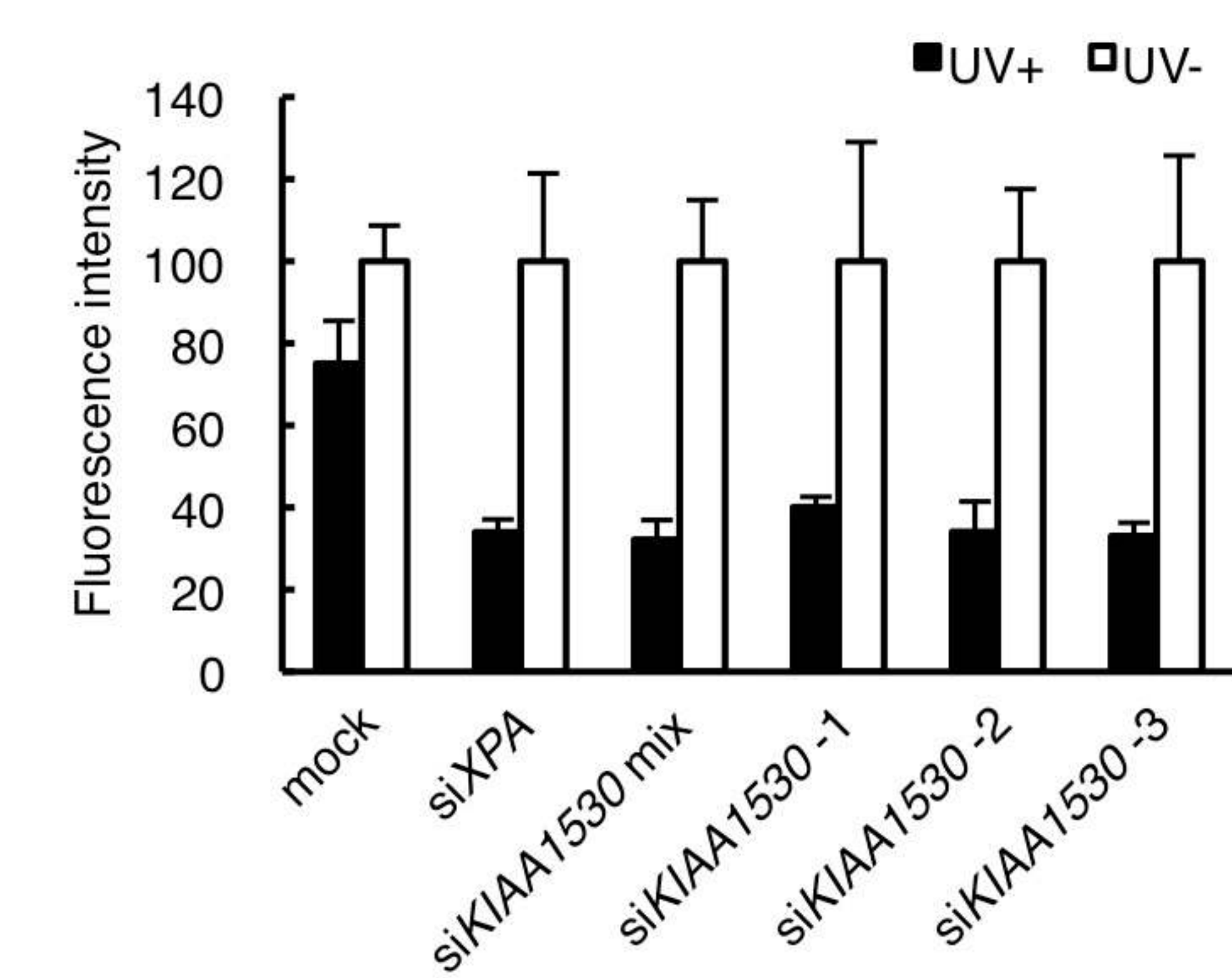


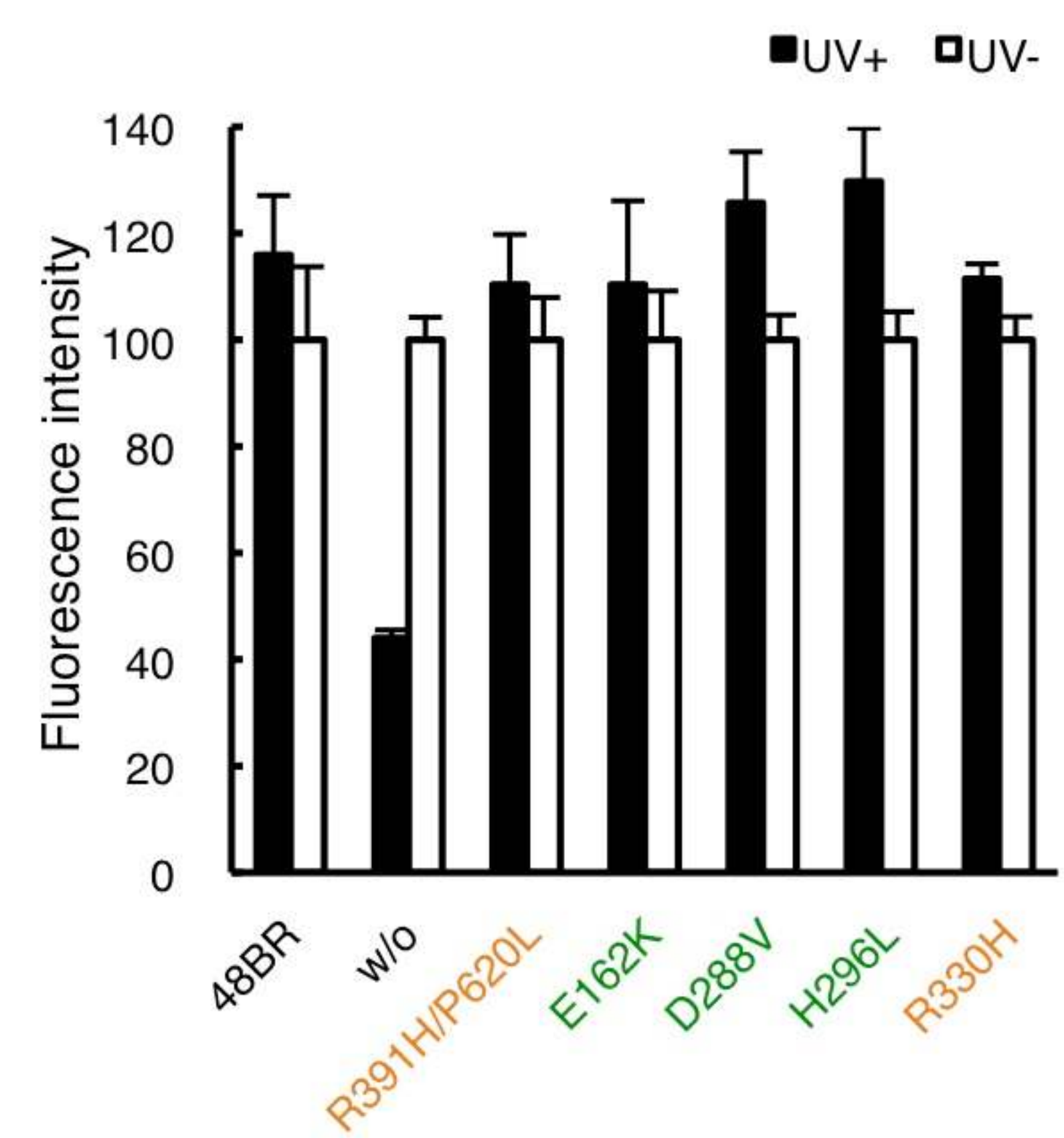
Fig.4



Supplementary figure S2



Supplementary figure S3



Supplementary figure S4

H. sapiens 1 -----MDQKLSKLVLEELTTSGEPRLNPEKMKELKKICKSSEEQ--LSRAYRLLIAQLTQEHAEIRLSAFQIVEELFVRSHQFRMLVVSNFQEFLELTLGTDPAQ 97
M. musculus 1 -----MDQKLSQLEIELTTSGESQLNAQKMKELKKICKSSEEQ--LSHAYRLITQLTQGHAEIRLSAFQIVDELFRSHQFRMLLVSDQEFLELTLGTDSDR 97
E. caballus 1 -----MDQKLSLVEELTTSGEPLNPEKMKELKKICKSSEEQ--LGHAYHLLKAQLSQEHAEIRLSAFQIVDELFRSHQFRMLVVISDFQEFLELTLGTDHEH 97
B. taurus 1 -----MDQKLSLVEELTTSGEPLNPEKMKELKKICKSSEEQ--LGHAYHLLMAQLSQEHAEIRLSAFQIVDELFRSHQFRMLVVISDFQEFLELTLGTDHER 97
X. tropicalis 1 -----MDQKLSLVEELTTSGEPLNPEKMKELKKICKSSEEQ--INHAYHLLMTQLNQEHAERLSAFQIVTELFARSHLFRMLLISNFQEFLELTLVETDHEQ 97
G. gallus 1 -----MDQRLAQLVEELTTSAGEPQLEPGRMKELKKICKSSEEQ--LSHAYHLLTRLHEEHAEMRFSAFQIVQELFRSHLFRMLLVSNFQEFLELTLVGTDHEQ 97
C. elegans 1 MLKRRQPKYCSFMSIDNSTTIRKKNLRFIRELTDGKLDIESIPYQNLQKEVANQDEEGCENVI ELLDTSRSGCPDRKLILQLFNSFFLQFPPIFRENLLNDPSFEFLMFMETNPIR 120
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H. sapiens 98 -PLPPPREEAQRRLQATTRAVERGN--EKFGAYKKLALGYHFLRHNKKVDFQDTNARSLAEKREEEKQKHLDKIYQERASQAEREMQEMSGEIESCLTEVESCFRLLV--FDLDPNPE 213
M. musculus 98 -PLPPPREEAQRRLQAAQAVEGN--EKFGAYKKLALGYHFLKHTKKVDFRDINVRTVAERKREEEKQKHLDKIHRESADRAKREMEEMDEIECCLTEVENCFKLLV--LDFVPCPE 213
E. caballus 98 -PLPPPREEAQRRLQATTRAVERGN--EKYGSAYKKLALGYHFLRHNKQVDFQDVNARTLAEKREEEKQKRLDKIYRERSERAQKMEEMSEIRRCLTEVESCFRLLV--FDLAPSPG 213
B. taurus 98 -PLPPPREEAQRRLQAAQAVEGN--EKYGAAYKKLALGFHFLKHSKQVDFQDVARTVAERKRAEERQKRLDKIYKERSWAAREMEEMSTEIRGCLTEVESCFRLLV--FDLAPSPG 213
X. tropicalis 98 -PLPPPREEAQRRLQAAQAVEGN--EKFGAYKKLALGYHFLKHTKKVDFQDVNARTLAEKREEEKQKRLDKIYRERSERAQKMEEMSEIRRCLTEVESCFRLLV--FDLAPSPG 213
G. gallus 98 -PLPPPREEAQRRLQAAQAVEGN--EKYGEAYKKLALGYHFLKHTKKVDFQDVHARTVAERKREEEKQKRLDKIYKERSWAAREMEEMSTEIRGCLTEVESCFRLLV--FDLAPSPG 213
C. elegans 121 NPLPGSKKHGNEKLVKVEAITVIKSEKVKVNDARMKCLVTLKTKTFVDYENGAKKIEAEKRRKKILEERKMKMIENS VNVYSSKYHEIKNDAETLSMELTTMQMLV----- 230
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H. sapiens 214 TESLG-----MASGMSD--ALRSSCAGQVPCRSRTPDPRDGEQPCSSRDL PASAGHPRAG--GGAQPSQTATGDPS--DEDESDL-----EEFVRSGLGSHKYTLDVLC 310
M. musculus 214 DKFFG-----EASSMTEGYAPCPLSPDLATPRESGLSGPQDEEQCCSKDLVASAYHVGSV--VGLKALPQTAMKDSRDEDEPSDP-----DDFLRSGLGSHKYTLDVLC 314
E. caballus 214 AAVP-----AAASSVSAEGRPHQAAAPNHEDEEQCCSKTLPTCARHP-----GATSREGPPSEDE--DEDESDQ-----EGFVRRGLGSHKYTLDVLC 298
B. taurus 214 -----AAWCPVPEKG-----DRDEEQCCSKSLAACAHHP-----GAMDAGGPPSEDE--DRDPD-----GFVRRGLGSRQYTLDVLC 281
X. tropicalis 216 EKDFASDMRTKPTSQSPSHSSTSSAYSSTSSQVDFNDDEQPCSSKLNLPFPSSCTSSASGAERSLGEKAKESDKSARKSDTDDSDGD---YEGSREAFRLDGLGSHAYSLSLEIS 332
G. gallus 215 -----TGLEPNEQTSADEDRSASSLPTVYGC--VDDEQPCSSKDLPLIS--QCVRTDQNEELADKQKPEQDLDLGDTCSEVESGVPVLGDDDYETVFRNGLISHKYALDLELS 320
C. elegans 231 -----SFTTADPEVPSTSTPSAISDS-----SFTTADPEVPSTSTPSAISDS-----SFTTADPEVPSTSTPSAISDS-----KSEFIFIPDL 263
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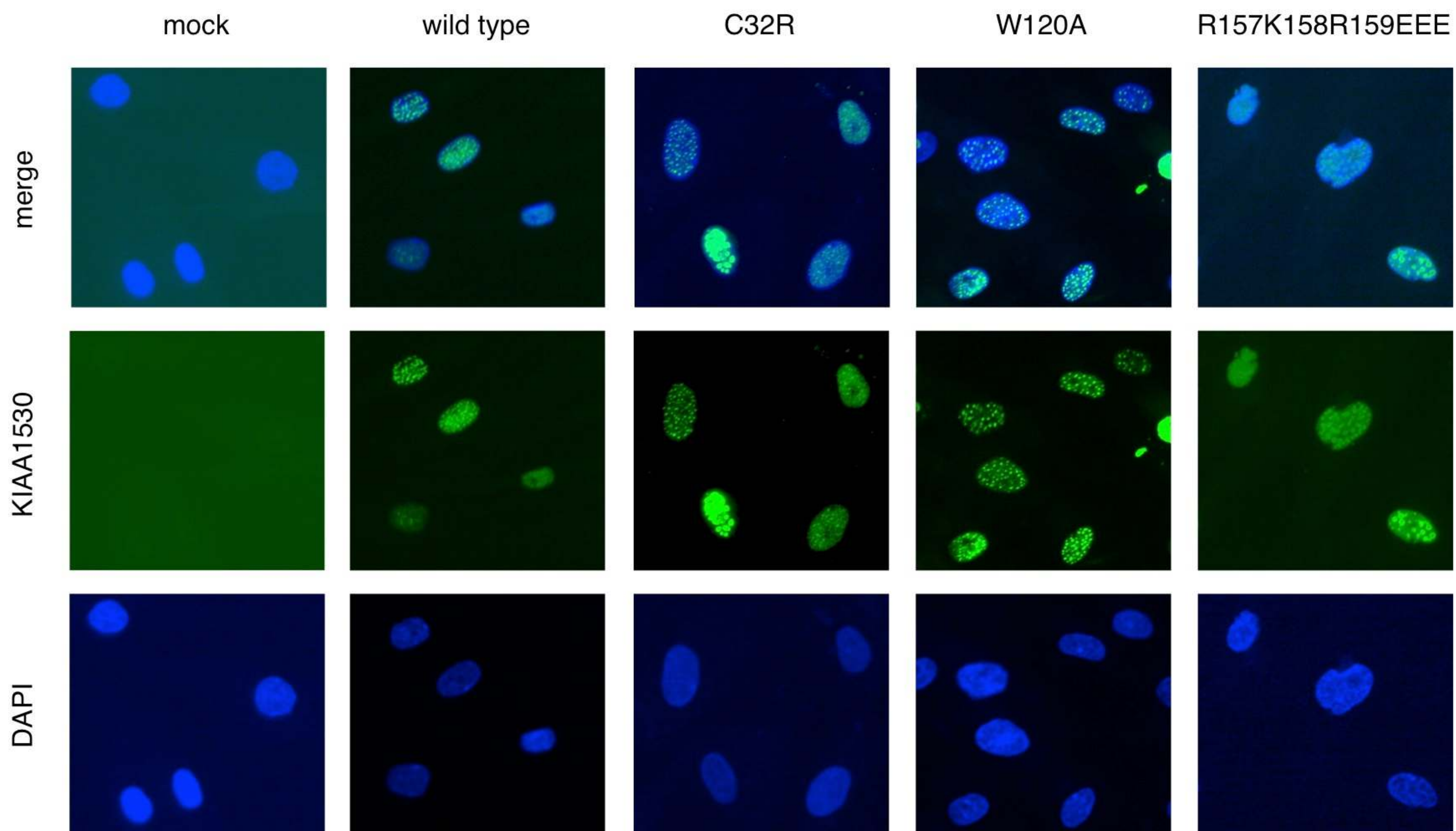
H. sapiens 311 SEGLKVQENEDNLALIHAAARDTLKLRNKLPAVCSWVQVTRVGTGTHGCLKRAIDLKAELELVLRYKELDIEPEGGERRTEALGD---AEDEDED--DEDFVEVPEKEGYEYEPHIDHL 425
M. musculus 315 SDGLKVQENEDNLAVLHAAARDSLKLIQNKFLPTVCSWVQVTRAGTYSAHLKQAIIDLKMELELALKKYEELNIEPGRGQRTEALED---SEDEDEQ---DFVEVPEKEGYEYEPHIDHL 427
E. caballus 299 SDSLVRVEDNCAVIHSARDALKLRNKLPAVCSWVQVTRAGVHGGHLEGAILKAELELALRRSQELDIEPEGVHRRETAAPGD---EDEDDEDEDDFVEVPEKEGYEACIPHIDHL 415
B. taurus 282 SDSLVRVEDNCAVIRAAARDALKLRNKLPAVCSWVQVTRAGTYGGHLEGAILKAELELALRRSQELDIEPEGVHRRETAAPGD---EDEDDEDEDDFVEVPEKEGYEACIPHIDHL 394
X. tropicalis 333 T-DLKVNENENNTDVLNMLDAHKLLKQKYWPAVQSWIQVFTKAGTNSSELKCAIDLKRLKAELELALRRSQELDIEPEGVHRRETAAPGD---EDEDDEDEDDFVEVPEKEGYEYEPHIDHL 443
G. gallus 321 TADIKVHENEEDNTAVINSVADAHKLRNKLPAVCSWVQVTRAGINDRRLRCAIDLKRLKAELELALRRSQELDIEPEGVHRRETAAPGD---EDEDDEDEDDFVEVPEKEGYEYEPHIDHL 436
C. elegans 264 TPEISVSENDAIVEAFLGAKLSLIHRVQTLRKLKRLQLLQKQPEK--LAQEI IDYRDGINKLVKLADELRI INPRPPKPKRKKSD--DFIDVDIS-----IDDIL 362
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H. sapiens 426 RPEYGLEAAPEKD-----TVVRCLR--TRTRMDEEVSDPTSAAAQLRQLRDHLP---PPSSASPSRALPEPQEAQKLAERARAPVVPYGVLDLHYWGQELPTAGKIVKSDSQHRFWKP 533
M. musculus 428 RAEYGLEPKAPLKTLEKGTAVCKLQER--TRMRREEASDPTSAAAQLRQLRDHLP---SPSSPS--TRVLPGPPEAQK--QAERARAPVVPYGVLDLHYWGQELPTAGKILKSDSQHRFWKP 540
E. caballus 416 RPEYGLQGXPKKD-----PAARDLEAR--KRTRRDEEACDPTSAAAQLRQLRDHLP---SPSSPS--TRVLPGPPEAQK--QAERARAPVVPYGVLDLHYWGQELPTAGKIFKSDSEHRFWKP 528
B. taurus 395 RPECGESPRAPREGLEEGLAAPGSQAR--KRPGSDMEAFDPTSAAAQLRQLRDHLP---SPSSPS--TRVLPGPPEAQK--QAERARAPVVPYGVLDLHYWGQELPTAGKIFKSDSEHRFWKP 512
X. tropicalis 444 REEYGLEPASKQPKTEVKRNPVQVPPSQKRIINDELNPTCAAATMKTMKDKMA--KALPGSSRNAGE--PKSKCPKRETDPSQAPVAPCGLDLHHWGQELPTAGKIFKSDSEHRFWKP 560
G. gallus 437 RKEYGEKAPAGPTP--LSSCAR-----LNRNEDELPTCAAATLKLIRDKLP--NESATTEPAALEEPDSKRRKLEERAKAPLMPFGLDLHYWGQELPTAGKIFKSDSEHRFWKP 543
C. elegans 363 MVQYAEKLEVDVK-----SKDESEKITESPEKHKIEMKNEK-----VKIKTVPFGLDLKHYWGQELPTAGKIFKSDSEHRFWKP 435
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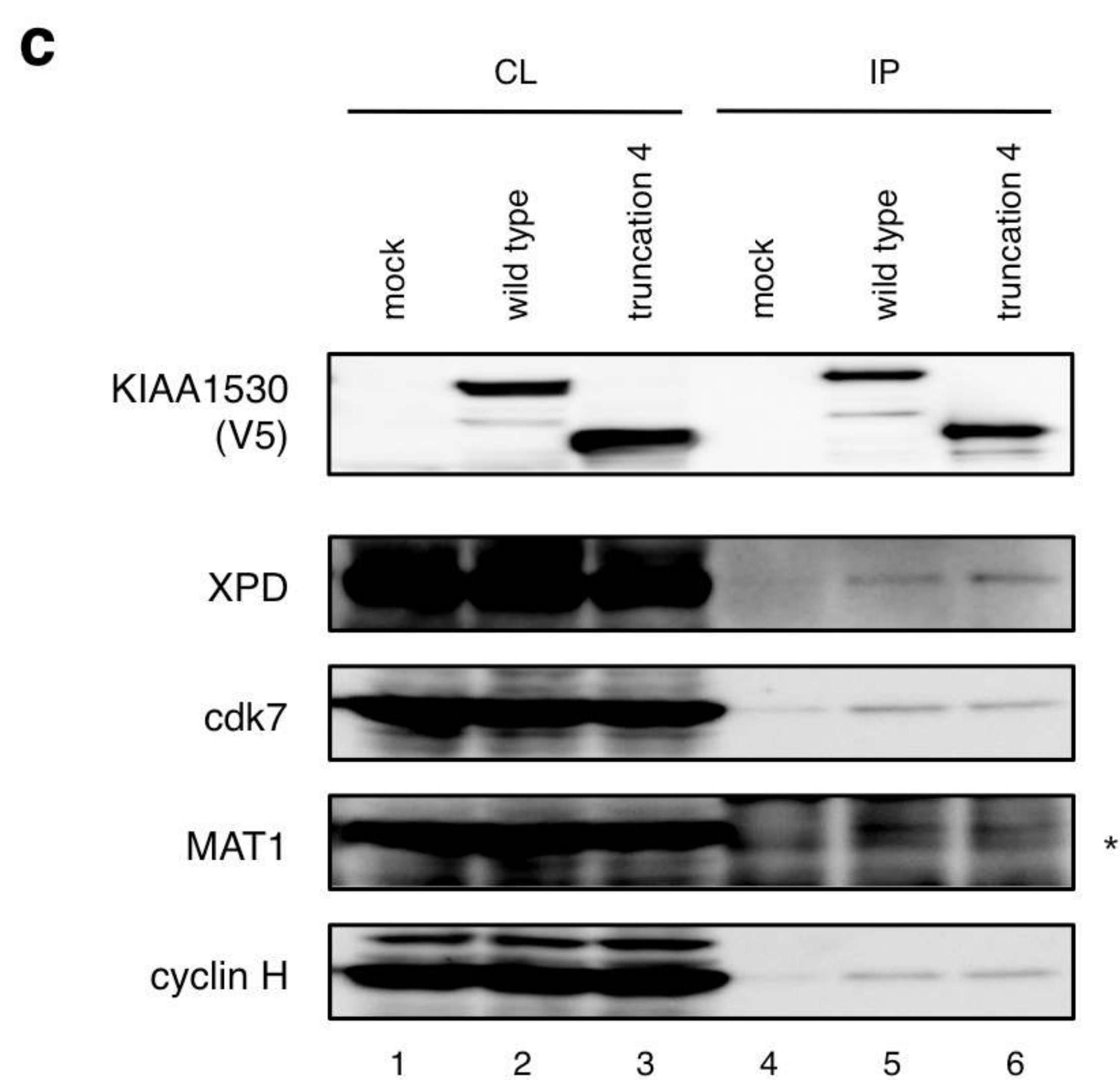
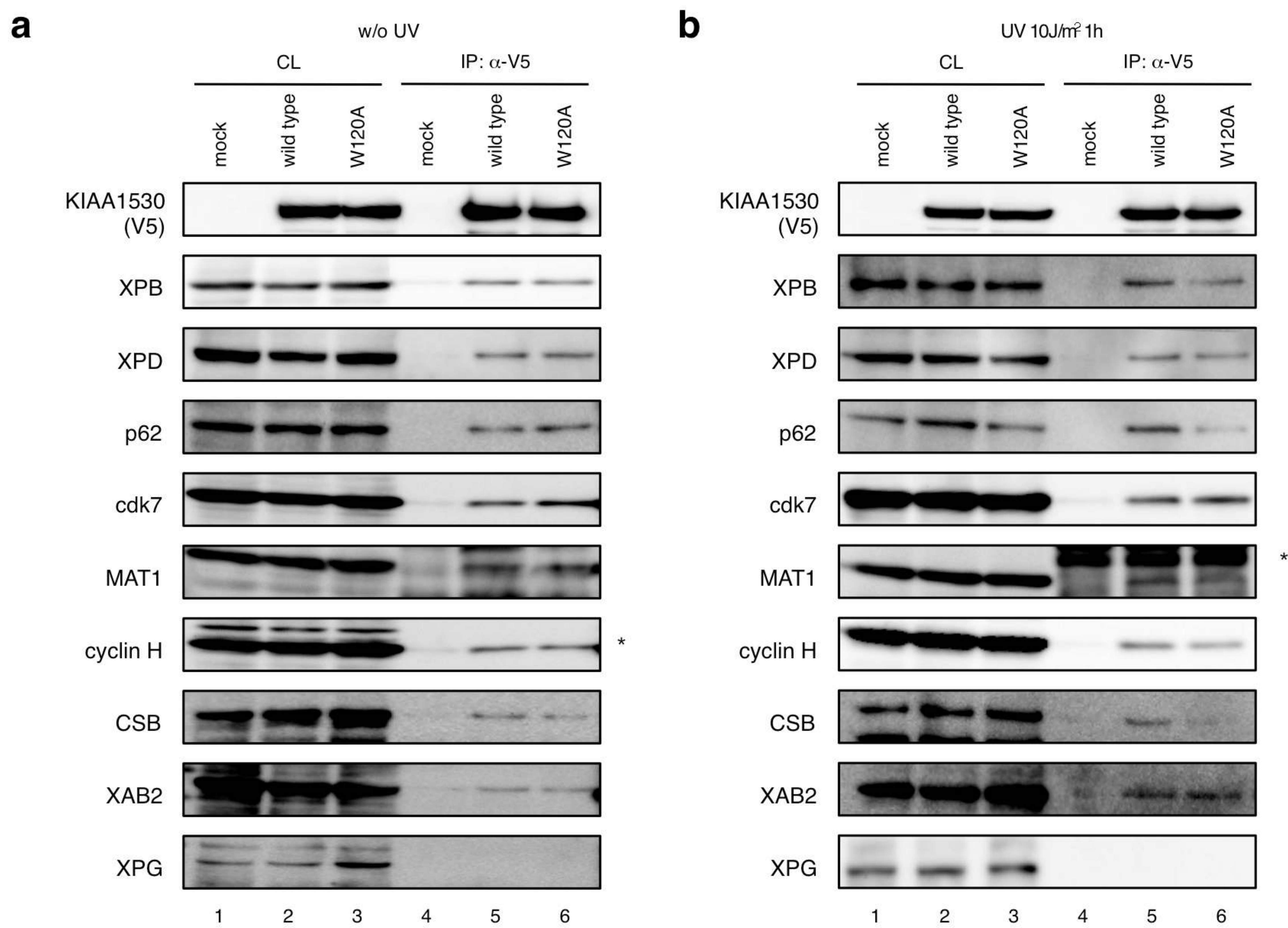
H. sapiens 534 SEVEEEVNADISEMLRSRHTFAGKFEFVQHWCRAPRPDGRLCERQDRKCPFHGKIVPRDDEGRPLDPEDRAREQRQLQKQER--PEWQDPELMDRVEAATGQDLGSSRSYSGKGRGK-- 651
M. musculus 541 SEVEEEVDNAHVSEMLRSRHTFAGKFEFVQHWCRALRPNGRLCERQDRKCPFHGKIIIPRDKGQPLNPEDRAREQRQLQKQER--PEWQDPELMDRVEAATGQDLGSSRSYSGKGRGK-- 659
E. caballus 529 SEVEEEVDNADVSETLRSRHTFAGKFEFVQHWCRAPRPDGRLCERQDRKCPFHGKIIIPRDNAGRPLNPEDRAREQRQLQKQER--PEWQDPELMDRVEAATGQDLGSSRSYSGKGRGK-- 647
B. taurus 513 CEVDAEVESASVSEALRSRHTFAGKFEFVQHWCRAPRPDGRLCERQDRKCPFHGKIIIPRDNAGRPLNPEDRAREQRQLQKQER--PEWQDPELMDRVEAATGQDLGSSRSYSGKGRGK-- 631
X. tropicalis 561 NEVDEEVESKELEALVKTRYITLPGKFEFVQHWCRAPRPDGRLCERQDRKCPFHGKIIIPRDECGVPINAEDRAREEAREKFEKQGEEDWRDPELMDRVEAATGQDLGSSKCPVKGKGRGK-- 680
G. gallus 544 SEVEEEVENKEITEMLKTRYITFAGKFEFVQHWCRAPRPDGRLCERQDRKCPFHGKIIIPRDECGVPINAEDRAREEAREKFEKQGEEDWRDPELMDRVEAATGQDLGSSKCPVKGKGRGK-- 659
C. elegans 436 SADEGTVAGKAQSIYTORQYTFIGKAPDNKRVCLAKMKSGLCPRKDYITCPLHGKIVDRDDEGRPINEEDRLEENYRKEQNHLKEDKIRQMIKEYESKT----- 538
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H. sapiens 652 -----KRRYPSLTNLKAQADTARARIGRVFAKAAVRRVVAAMNRMDQKKHEK--FSNQFNIALN 709
M. musculus 660 -----KKKHPNLTDLRERTNTARARLEKRVFAKAAVQRVVAAMNQMDQKKHEK--FANQFNIALN 717
E. caballus 648 -----RKKHPGLTDLKQADTARARISKKVFAKAAVQRVVTAMNQMDQKKHEK--FANQFNIALN 705
B. taurus 632 -----RRKHSGLTDLKQADTARARIAKKVFAKAAVQRVVTAMNQMDQKKHEK--FANQFNIALN 689
X. tropicalis 681 VKRNLKKKYPNLTDLKQANTARSRLKRVFNTGSGVKKRVISAMNQADKRRHEK--FANQFNIALN 743
G. gallus 660 GKKKGGKKYPNLTDLKQANTARSRLKRVFNTGSGVKKRVISAMNQADKRRHEK--FANQFNIALN 722
C. elegans 539 -----KRRKHVDVDTASEDVRNRLQKLLDPKTIQRVSADLDASRNRLKFNQGFQF-- 594
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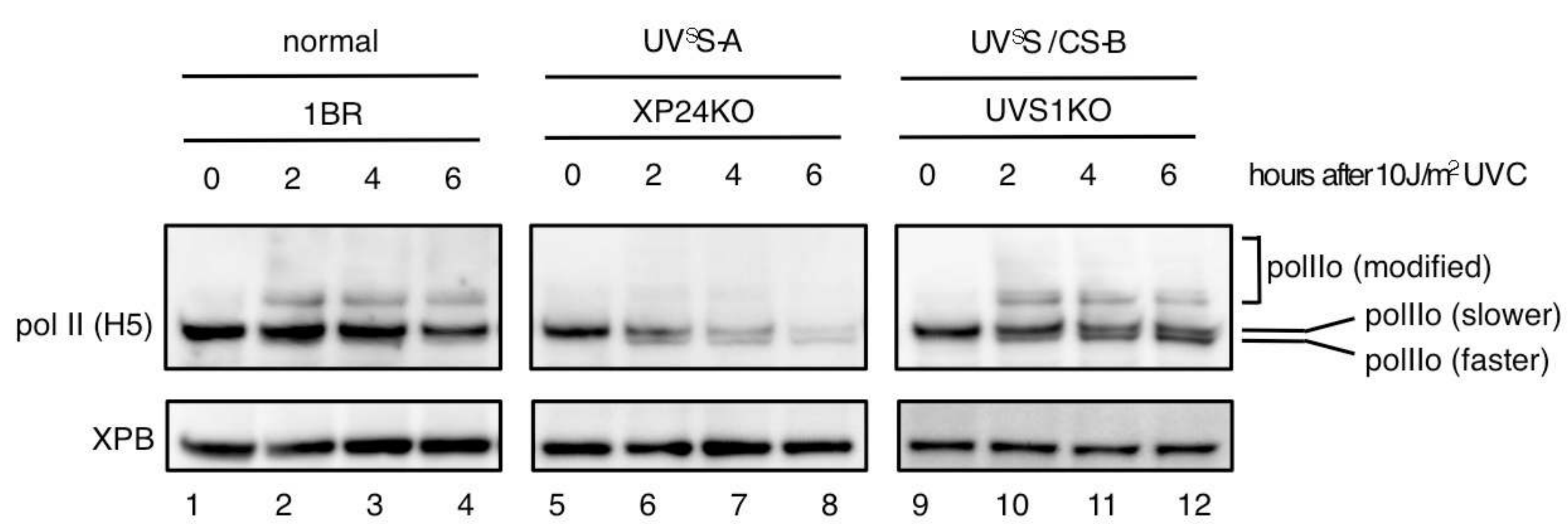
Supplementary figure S7



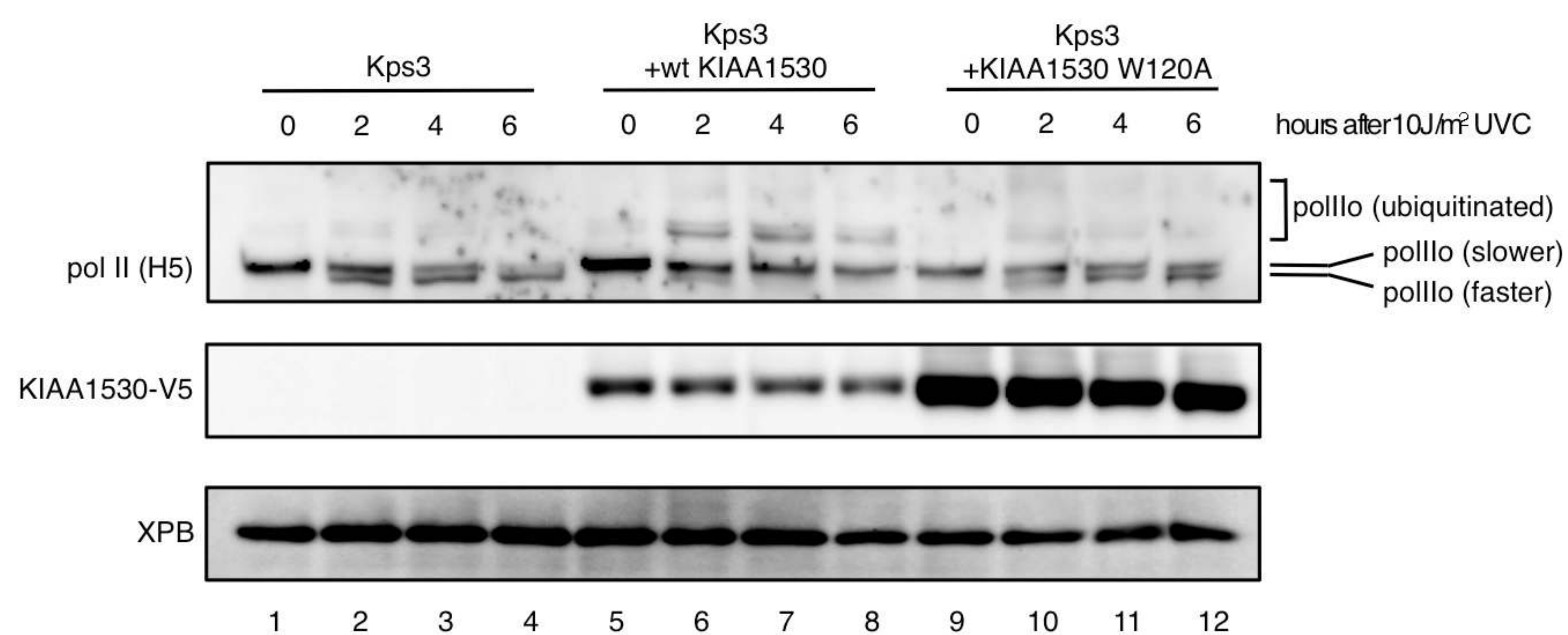
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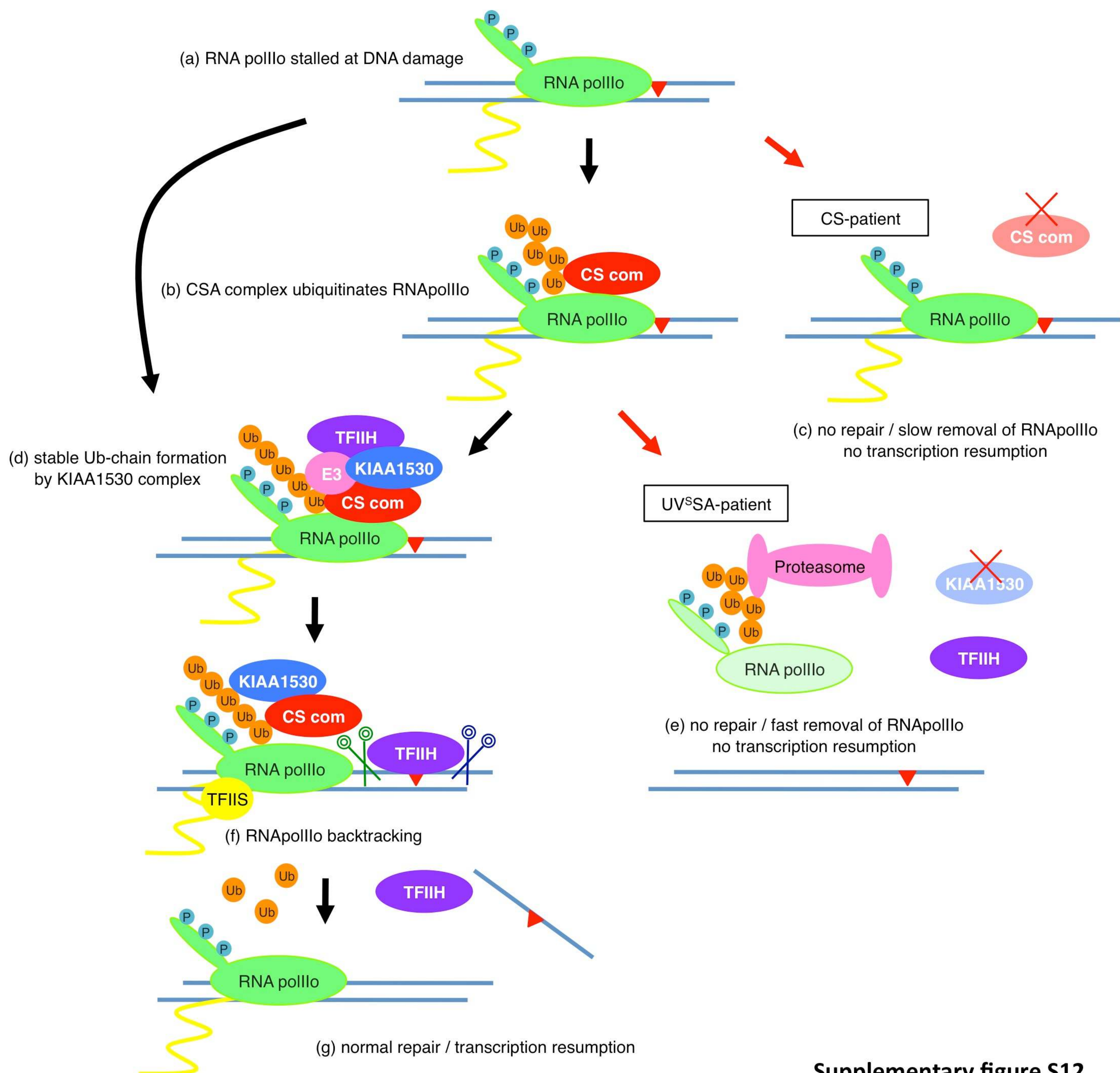
Supplementary figure S9



Supplementary figure S10



Supplementary figure S11



Supplementary figure S12