TREX1 Dominant Mutations in Lupus and Aicardi-Goutieres Syndrome

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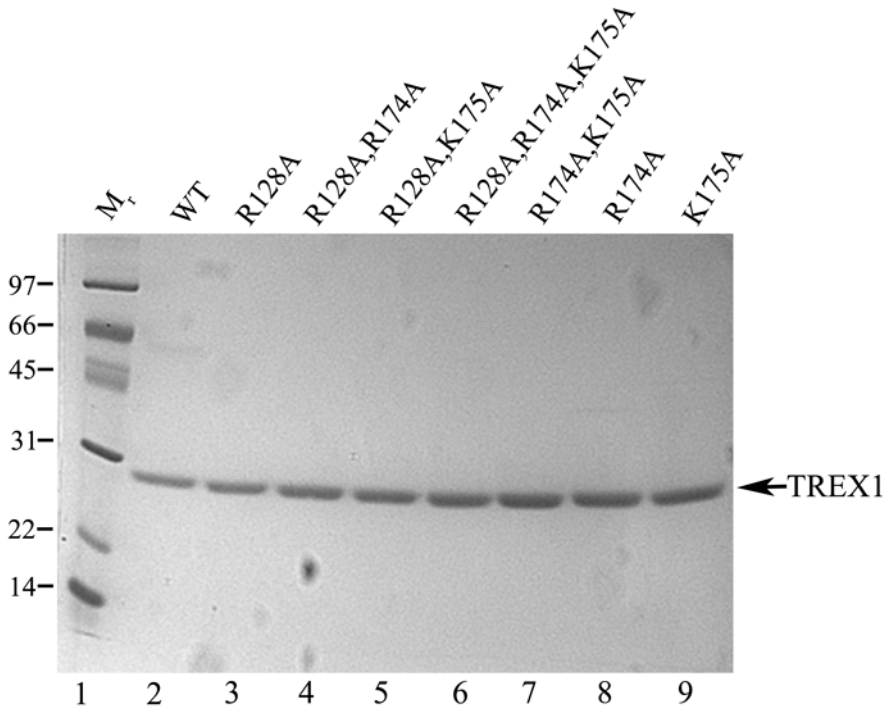
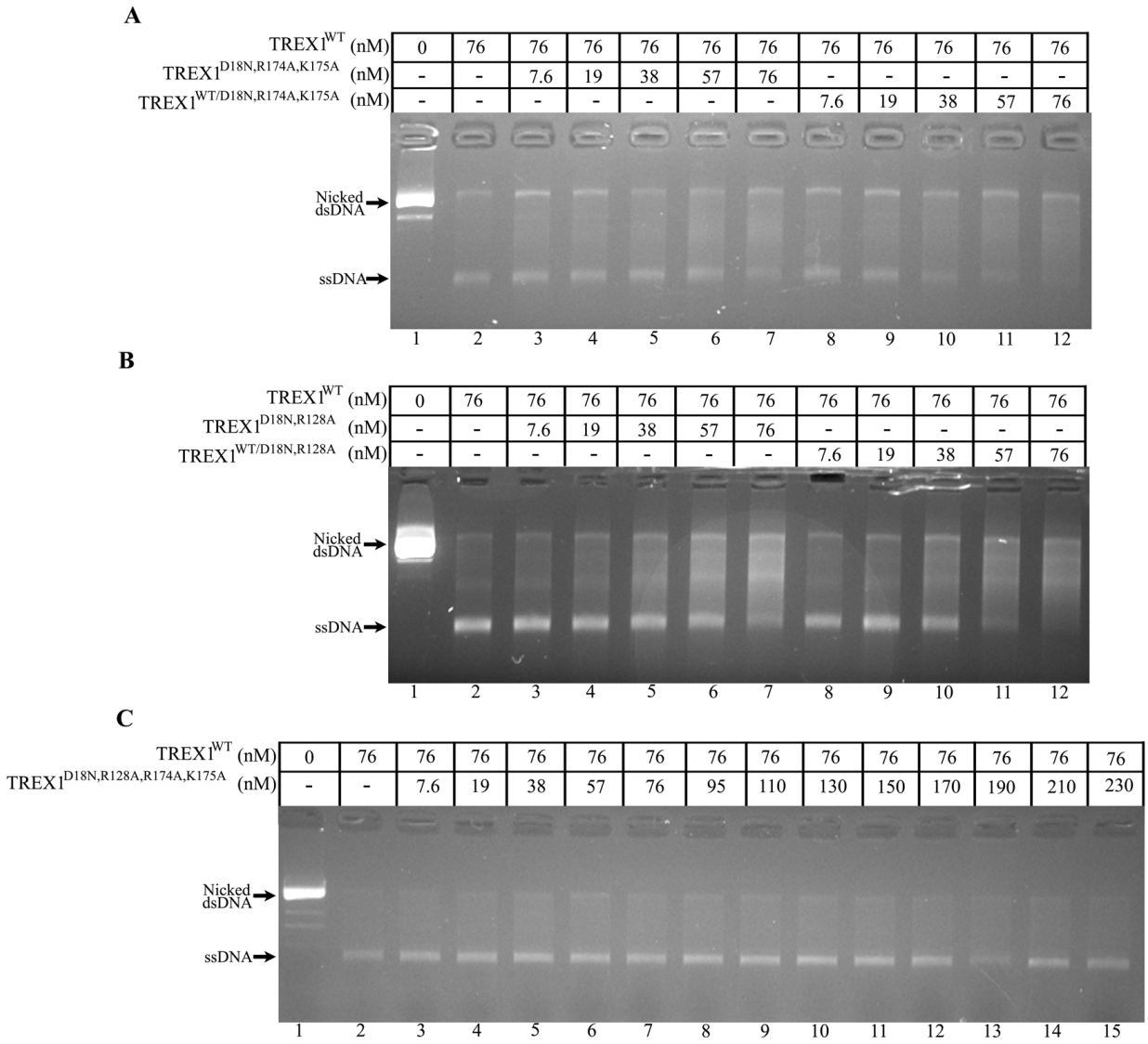
Supplemental Information

Fig. S1



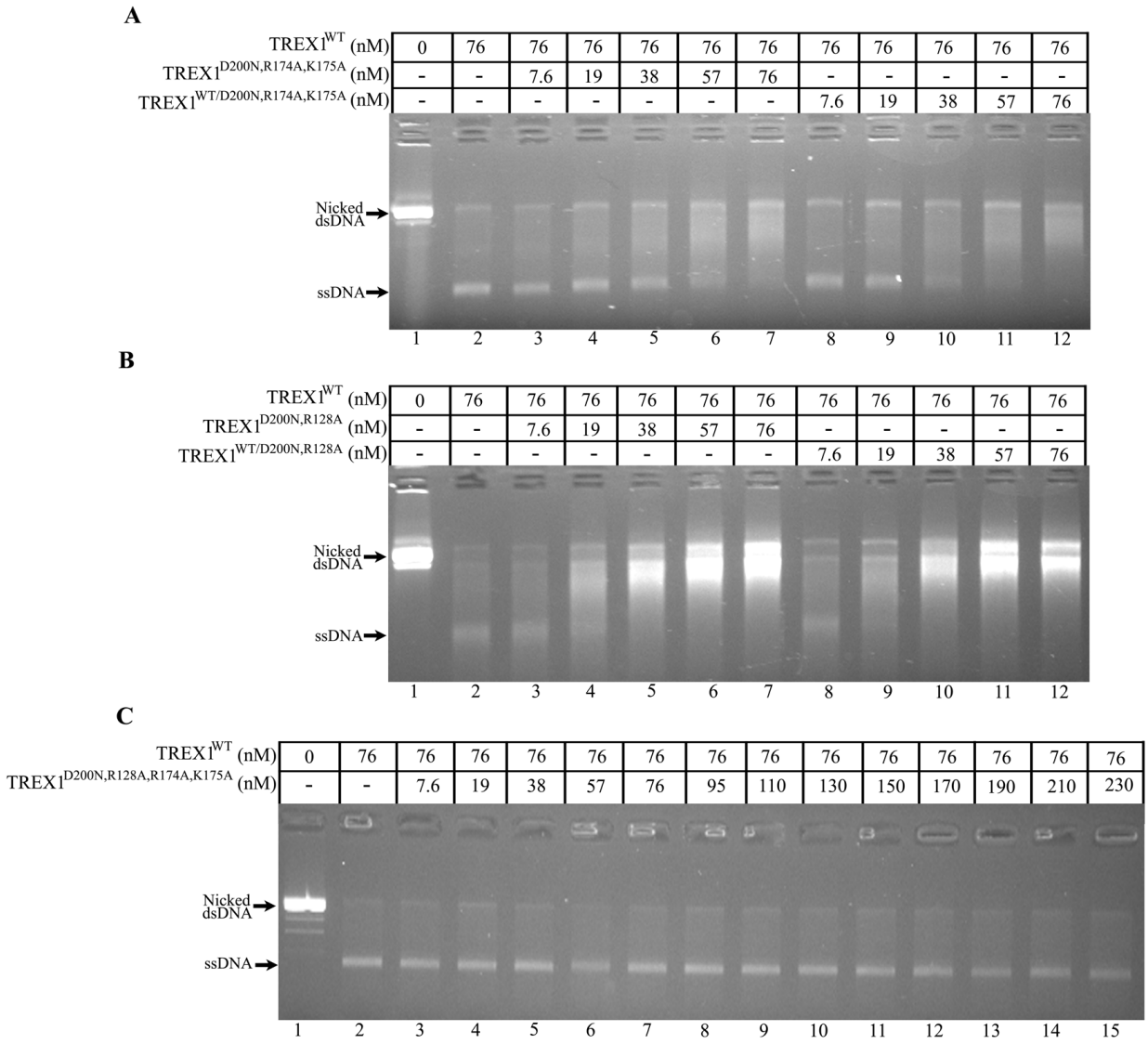
Supplemental Fig. S1. **SDS-PAGE analysis of the purified human TREX1^{WT} and mutant proteins.** Site-directed mutations were introduced into the TREX1 gene by polymerase chain reaction and cloned into the pLM303x plasmid to generate the TREX1 expression constructs. The TREX1 enzymes were generated as described in the text. Approximately 3 ug of each protein was subjected to 12% SDS-PAGE, and the gel was stained with Coomassie Brilliant Blue. The positions of migration of the molecular weight standards (lane 1) are indicated.

Fig. S2



Supplemental Fig. S2. **DNA binding residues in the TREX1 D18N dominant mutant.** The exonuclease reactions contained nicked dsDNA plasmid 1 and no enzyme (*A, B, C; lane 1*) or the indicated concentration of TREX1^{WT} only (*A, B, C; lane 2*) or a mixture of TREX1^{WT} with the indicated increased concentrations of TREX1^{D18N,R174A,K175A} (*A;lanes 3–7*), TREX1$^{WT/D18N,R174A,K175A}$ (*A;lanes 8–12*), TREX1^{D18N,R128A} (*B;lanes 3–7*), TREX1$^{WT/D18N,R128A}$ (*B;lanes 8–12*), or TREX1^{D18N,R128A,R174A,K175A} (*C;lanes 3–15*). The reactions were 30 min and products were subjected to agarose gel electrophoresis. The positions of migration of Form II (*Nicked dsDNA*) and circular ssDNA (*ssDNA*) are indicated.

Fig. S3



Supplemental Fig. S3. **DNA binding residues in the TREX1 D200N dominant mutant.** The exonuclease reactions contained nicked dsDNA plasmid 1 and no enzyme (*A, B, C; lane 1*) or the indicated concentration of TREX1^{WT} only (*A, B, C; lane 2*) or a mixture of TREX1^{WT} with the indicated increased concentrations of TREX1^{D200N,R174A,K175A} (*A;lanes 3–7*), TREX1$^{WT/D200N,R174A,K175A}$ (*A;lanes 8–12*), TREX1^{D200N,R128A} (*B;lanes 3–7*), TREX1$^{WT/D200N,R128A}$ (*B;lanes 8–12*), or TREX1^{D200N,R128A,R174A,K175A} (*C;lanes 3–15*). The reactions were 30 min and products were subjected to agarose gel electrophoresis. The positions of migration of Form II (*Nicked dsDNA*) and circular ssDNA (*ssDNA*) are indicated.