

Distinct frequencies and mutation spectrums of genetic thrombophilia in Korea in comparison with other Asian countries both in patients with thromboembolism and in the general population

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Manuscript received on May 24, 2013. Manuscript accepted on October 18, 2013.

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Supplementary Figures

The comparative genomic analyses to assess the degree of conservation of the mutated residues in novel missense mutations in the *PROC*, *PROS1*, and *SERPINC1* genes identified in this study.

PROC: c.790A>G (p.Arg264Gly)

Arg264

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Homo_sapiens      KLACGAVLIHPSWVLTAAHCMDESSKKLLVRLGEYDLRRWEKWELDLDIKEV FVHPNYSKS
Pan_troglodytes  KLACGAVLIHPSWVLTAAHCMDESSKKLLVRLGEYDLRRWEKWELDLDIKEV FVHPNYSKS
Mus_musculus     KLACGGVLIHTSWVLTAAHCVEGTKKLTVRLGEYDLRRRDHWELDLDIKEI LVHPNYTRS
Rattus_norvegicus KLACGGVLIHTSWVLTAAHCLESSKLTVRLGEYDLRRRDPWELDLDIKEV LVHPNYTRS
Bos_taurus       KLVCGAVLIHVSWVLTVAHCLDSRKKLIVRLGEYDMRRWESWEVDLDIKEV IHPNYTKS
Canis_familiaris KLACGAVLIHTSWVLTAAHCMEDSKKLIVRLGEYDLRRWEKGEMDVDIKEV LIHPNYSKS
Macaca_mulatta   KLACGAVLIHPSWVLTAAHCMEESKLLVRLGEYDLRRWEKWELDLDI EEFVFIHPNYTKS
Ovis_aries       KLVCGAVLIHVSWVLTVAHCLESHKKLIVRLGEYDMRRWESWEVDLDIKEV IVHPNYTKS
Sus_scrofa       KLACGAVLIHVSWVLTAAHCLDDYKKLTVRLGEYDLRRREKWEVDLDIKE FLVHPNYTRS
Gallus_gallus    KFLCGGVLIHPSWVLTAAHCVETGETLKVRLGKYHRLRIENSEQTIRVDKYVRHENYTKL
Danio_rerio      RFHCGGVLI DENWVLTAAHCLETSSKFSVRLGDYQRFKFEGSEVTLPVKQHISHPQYNPI
Xenopus_laevis   KMKCGGVLIHPSWVLTAAHCVTVTGTKYSVRLGEYDIRKLEDETEQQFAVVKI IIPPEYRSD
Xenopus_tropicalis KLKCGGVLIHPFWVLTAAHCVTHAGKYTVRLGEYDIRKLEDETEQQFAVIKI IIPPEYESN
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PROC: c.796G>C (p.Gly266Arg)

Gly266

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Homo_sapiens      KLACGAVLIHPSWVLTAAHCMDESKKLLVRI GEYDLRRWEKWELDLDIKEVFPVHPNYSKS
Pan_troglodytes  KLACGAVLIHPSWVLTAAHCMDESKKLLVRI GEYDLRRWEKWELDLDIKEVFPVHPNYSKS
Mus_musculus     KLACGGVLIHTSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
Rattus_norvegicus KLACGGVLIHTSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
Bos_taurus       KLACGAVLIHPSWVLTAAHCVETGKTLVRI  GEYDLRRWEKWELDLDIKEVFPVHPNYSKS
Canis_familiaris KLACGAVLIHPSWVLTAAHCMDESKKLLVRI GEYDLRRWEKWELDLDIKEVFPVHPNYSKS
Macaca_mulatta   KLACGAVLIHPSWVLTAAHCMDESKKLLVRI GEYDLRRWEKWELDLDIKEVFPVHPNYSKS
Ovis_aries       KLACGAVLIHPSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
Sus_scrofa       KLACGGVLIHTSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
Gallus_gallus    KFLCGGVLIHPSWVLTAAHCVETGKTLVRI  GKYHRLRIENSEQTIRVDKYVRHENYTKL
Danio_rerio      RFHCGGVLIHTSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
Xenopus_laevis   KMKCGGVLIHTSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
Xenopus_tropicalis KLKCGGVLIHTSWVLTAAHCVETGKTLVRI  GEYDLRRRDHWELDLDIKEILVHPNYTRS
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PROS1: c.947G>A (p.Arg316His)

Arg316

Homo_sapiens	TKYELLYLAEQFAGV-VLYLKFR	LPEISRFSAEFD	FRTYDSEGVILYAESIDHSAWLLIA
Pan_troglodytes	TKYELLYLAEQFAGV-VLYLKFR	LPEISRFSAEFD	FRTYDSEGVILYAESIDHSAWLLIA
Mus_musculus	KNYELLYLAEQFAGV-VLYLKFR	LPDITRFSAEFD	FRTYDSEGIILYAESLDHSNWLLIA
Rattus_norvegicus	KNYELLYLAEQFAGV-VLYLKFR	LPDITRFSAEFD	FRTYDSEGIILYAESLDHSNWLLIA
Bos_taurus	KNYELLYLAEQFVGV-VLYLKFR	LPETTRFSAEFD	FRTYDSEGVILYAESSDHSWFLIA
Canis_familiaris	KNYELLYLAEQFVGV-VLYLKFR	LPEITRFSAEFD	FRTYDSEGVILYAESLDRSAWFLIA
Macaca_mulatta	TKYELLYLAEQFAGV-VLYLKFR	LPEISRFSAEFD	FRTYDSQGVILYAESIDHSAWLLIA
Ovis_aries	KNYELLYLAEQFVGV-VLYLKFR	LPETTRFSAEFD	FRTYDSEGVILYAESSDHSSWFLIA
Gallus_gallus	RNYELLYLAEHFTGIYVLYLKFK	LPNVRFSAEFD	FRTYDSEGIILYAESFDNTAWILLA
Danio_rerio	KHKEMLYLGEQFSGLPVIYLR	FRLQPHTRFAAE	FDPEGVILYAESSP-DSWIMLG
Xenopus_tropicalis	TNYELLYLAEQFTGIPVVYLR	FKLPDVSRFSAE	FDRTYDGEGVILYAESADSTSWFLIA
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PROS1: c.974C>G (p.Ala325Glu)

Ala325

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Homo_sapiens      TKYELLYLAEQFAGV-VLYLKFRLLPEISRFSAEFDRTYDSEGVILYAESIDHSAWLLIA
Pan_troglodytes   TKYELLYLAEQFAGV-VLYLKFRLLPEISRFSAEFDRTYDSEGVILYAESIDHSAWLLIA
Mus_musculus      KNYELLYLAEQFAGV-VLYLKFRLLPDITRFSAEFDRTYDSEGIILYAESLDHSNWLLIA
Rattus_norvegicus KNYELLYLAEQFAGV-VLYLKFRLLPDITRFSAEFDRTYDSEGIILYAESLDHSNWLLIA
Bos_taurus        KNYELLYLAEQFVGV-VLYLKFRLLPETTRFSAEFDRTYDSEGVILYAESSDHSWFLIA
Canis_familiaris KNYELLYLAEQFVGV-VLYLKFRLLPEITRFSAEFDRTYDSEGVILYAESLDRSAWFLIA
Macaca_mulatta    TKYELLYLAEQFAGV-VLYLKFRLLPEISRFSAEFDRTYDSQGVILYAESIDHSAWLLIA
Ovis_aries        KNYELLYLAEQFVGV-VLYLKFRLLPETTRFSAEFDRTYDSEGVILYAESSDHSWFLIA
Gallus_gallus     RNYELLYLAEHFTGIYVLYLKFKLPNVTFRFSAEFDRTYDSEGIILYAESFDNTAWILLA
Danio_rerio       KHKEMLYLGEQFSGLPVIYLRFRLLQPHTRFAAEFDRTFDPEGVILYAESSP-DSWIMLG
Xenopus_tropicalis TNYELLYLAEQFTGIPVVYLRFKLLPDVSRFSAEFDRTYDSEGVILYAESADSTSWFLIA
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SERPINC1: c.1057C>G (p.Pro353Ala)

Pro353

Homo_sapiens	PEVLQEWLDELEEMMLVYHMPRFRIEDGFSLKEQLQDMGLVOLFSPPEKSKLPGLVAEG-R
Pan_troglodytes	PEVLQEWLDELEEMMLVYHMPRFRIEDSFSLKEQLQDMGLVOLFSPPEKSKLPGLVAEG-R
Mus_musculus	PELLQEWLDELSETMLVYHMPRFRTEDEGFSLKEQLQDMGLIDLFSPPEKSQLPGLVAGG-R
Rattus_norvegicus	PELLQEWLDELSEVMLVYHMPRFRIEDSFSLKEQLQDMGLVOLFSPPEKSQLPGLVAEG-R
Bos_taurus	PDMLQEWLDELTEPLLVYHMPRFRIEDSFSVKEQLQDMGLEDLFSPPEKSRLPGLVAEG-R
Canis_familiaris	PEVLQEWLDEMTETPLLVYHMPCFRMEDEFSVKEQLQDMGLVOLFNPPEKSRLPGLVAEG-R
Macaca_mulatta	PEVLQEWLDELEEMMLVYHMPRFRIEDGFSLKEQLQDMGLVOLFSPPEKSKLPGLVAEG-R
Ovis_aries	PDMLQEWLDELTEPLLVYHMPFRFRIEDSFSVKEQLQDMGLEDLFSPPEKSRLPGLVAEG-R
Sus_scrofa	PEVLQEWLDELADTPLLVYHMPRFRHIEDSFSVKEQLQDMGLEDLFPEKAKLPGLVAEG-R
Gallus_gallus	SDKLQDWIDSMMEVSLTVSFRFRVEDSFSVKEKLRKMGLEDLFSPENAKLPGLVAGD-R
Danio_rerio	LKKLVGWLHAMKETTVAVQIPRFRVEDSFSLKEQLTKMGLEDLFSPANASLPGMVADAEG
Xenopus_laevis	LEKLGHWLQKSRELQLSVYLPFRFRVEDSFSVKEKLEMGVOLFDPNSAKLPGLVAGG-R
Xenopus_tropicalis	LEKLGHWLQKSRELQLSVYLPFRFRVEDSFSVKEKLEMGVOLFDPNSAKLPGLVAGG-R
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SERPINC1: c.377C>A (p.Ala126Asp)

Ala126

Homo_sapiens	SISTAFAMTKLGA C NDTLQQLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS K L V
Pan_troglodytes	SISTAFAMTKLGA C NDTLQQLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS K L V
Mus_musculus	SISTAFAMTKLGA C NDTL K QLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS D L V
Rattus_norvegicus	SISTAFAMTKLGA C NN T L K QLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS N L V
Bos_taurus	SISTAFAMTKLGA C D N T L KQLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS S E L V
Canis_familiaris	SISTAFAMTKLGA C NN T L K QLMEVFKFDI I SEKTS D Q V HFFFAK L NCRL Y RK A NKSS S E L V
Macaca_mulatta	SVSTAFAMTKLGA C NDTL K QLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS K L V
Ovis_aries	SISTAFAMTKLGA C NN T L K QLMEVFKFDI I SEKTS D Q I HFFFAK L NCRL Y RK A NKSS S E L V
Sus_scrofa	SISTAFAMTKLGA C D N T L KQLMEVFKFDI I SEKTS D Q V HFFFAK L NCRL Y RK A NKSS S E L V
Gallus_gallus	SISTAFAMTKLGA C G D T L QQLMEV F Q F DI I SEKTS D Q V HFFFAK L NCRL Y K K A N KSS E L I
Danio_rerio	SISTAFAMTKLGA C N T T L E Q LM K V F Q F DI I KEKTS D Q V HFFFAK L NCRL Y R K K H E T T E L I
Xenopus_laevis	SISQAFTMAKLGA C NN T L K QLMEV F H F DT V SE R AS D Q I H Y FFAK L NCRL F R K A N KSS E L V
Xenopus_tropicalis	SISQAFTMAKLGA C NN T L K ELMEV F Y F DT I SE R AS D Q I H Y FFAK L NCRL F R K A N KSS E L V
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SERPINC1: c.851T>G (p.Met284Arg)

Met284

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Homo_sapiens      IPSEAINELTVLVLVNTIYFKGLWKSKFSPENTRKELFYKADGESCSASMMYQEGKFRYR
Pan_troglodytes  IPSEAINELTVLVLVNTIYFKGLWKSKFSPENTRKELFYKADGESCSASMMYQEGKFRYR
Mus_musculus     IPQGAINELTALVLVNTIYFKGLWKSKFSPENTRKEPFYKVDGQSCPVPMMYQEGKFKYR
Rattus_norvegicus IPQGAIDELTALVLVNTIYFKGLWKSKFSPENTRKEPFHKVDGQSCLVPMYQEGKFKYR
Bos_taurus       IPPQAINEFTVLVLVNTIYFKGLWKSKFSPENTRKELFYKADGESCSVLMYQESKFRYR
Canis_familiaris VPPDAIDELTVLVLVNTIYFKGLWKSKFSPENTRKELFYKAGGESCSVSMYQEGKFRYR
Macaca_mulatta   IPPEAINELTVLVLVNTIYFKGLWKSKFSPENTRMEPFYKADGESCSASMMYQEGKFCYR
Ovis_aries       IPPQAIDEFTVLVLVNTIYFKGLWKSKFSPENTKKELFYKADGESCSVPMYQEGKFRYR
Sus_scrofa       IPPEAINELTVLVLVNTIYFKGRWKSEFSSENTRKELFYKANGESCSVSMYQESKFRYR
Gallus_gallus    IPEKGIDDLTVLVLVNTIYFKGHWKSQFPAPNTRLDFHKANGETCNVPIYQESRFRYA
Danio_rerio      LPEGSIDTNTILVLVNAIYFKGQWKNKFDKQNVMKLDFHVSPTHKCPVPMYQEKQYA
Xenopus_laevis   IPKDAITPDTVLVLINAIYFKGLWKSKFNSENTKMDQFHPAKNSNCLTATMYQEGTFRYG
Xenopus_tropicalis IPVGVITPDTVLVLINAIYFKGLWKSKFNSENTKMEQFYPDESNHCLAATMYQEGIFRYS
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