

Human GP1BA: Published Nucleotide sequence and predicted amino acid sequence

Reported GP1BA Mutations and Polymorphisms highlighted. References listed below

The sequence numbering is according to the GP1BA sequence of Lopez et al "Cloning of the alpha chain of human platelet glycoprotein Ib: a transmembrane protein with homology to leucine-rich alpha 2-glycoprotein." Proc Natl Acad Sci U S A. 1987 84(16):5615-5619.

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                                     M P L L L L L L L L P S P L H P   -1
(-5T/C)(1)
GACGCTCTGTGCCTTCGGAGGTCTTTCTGCCTGCCTGTCCTCCTCTCTCTGTGCTCCTGCTGCCAAGCCCCTTACACCCC   90

H P I C E V S K V A S H L E V N C D K R N L T A L P P D L P   30
CACCCCATCTGTGAGGTCTCAAAGTGGCCAGCCACCTAGAAGTGAAGTGTGACAAGAGGAATCTGACAGCGCTGCCTCCAGACCTGCCTG   180

K D T T I L H L S E N L L Y T F S L A T L M P Y T R L T Q L   60
AAAGACACAACCATCCTCCACCTGAGTGAGAACCCTCCTGTACACCTTCTCCCTGGCAACCCTGATGCCTTACACTCGCCTCACTCAGCTG   270

(Leu70Phe)(2)
N L D R C E L T K L Q V D G T L P V L G T L D L S H N Q L Q   120
AACCTAGATAGGTGCGAGCTCACCAAGCTCCAGGTCGATGGGACGCTGCCAGTGTGGGGACCCCTGGATCTATCCACAATCAGCTGCAA   360

S L P L L G Q T L P A L T V L D V S F N R L T S L P L G A L   120
AGCCTGCCCTTGCTAGGGCAGACACTGCCTGCTCTCACCCTGCTGGACGCTCCTTCAACCGGCTGACCTCGCTGCCTCTTGGTGCCTGTG   450

R G L G E L Q E L Y L K G N E L K T L P P G L L T P T P K L   150
CGTGGTCTTGGCGAACTCCAAGAGCTCTACCTGAAAGGCAATGAGCTGAAGACCCCTGCCCCAGGGCTCCTGACGCCACACCCAAGCTG   540

(E Thr145Met)(3)
E K L S L A N N N L T E L P A G L L N G L E N L D T L L L Q   180
GAGAAGCTCAGTCTGGCTAACCAACAACCTTGACTGAGCTCCCCGCTGGGCTCCTGAATGGGCTGGAGAATCTCGACACCCTTCTCTCCAA   630

E N S L Y T I P K G F F G S H L L P F A F L H G N P W L C N   210
GAGAACTCGCTGTATAACAATCAAAAGGCTTTTTTGGGTCACCTCCTGCCCTTTTGTCTTTTCTCCACGGGAACCCCTGGTTATGCAAC   720

(Gly233Ser)(5,6) (Met239Val)(7,8)
C E I L Y F R R W L Q D N A E N V Y V W K Q G V D V K A M T   240
TGTGAGATCCTCTATTTTCGTCGCTGGCTGCAGGACAATGTGAAAATGTCTACGTATGGAAGCAAGTGTGGACGTCAAGGCCATGACC   810

S N V A S V Q C D N S D K F P V Y K Y P G K G C P T L G D E   270
TCTAACGTGGCCAGTGTGACGATGACAATTCAGACAAGTTTCCCGTCTACAAAATACCCAGGAAAGGGTGCACCCCTTGGTGATGAA   900

G D T D L Y D Y Y P E E D T E G D K V R A T R T V V K F P T   300
GGTGACACAGACCTATATGATTACTACCCAGAAGAGGACTGAGGGCGATAAGGTGCGTGCCACAAGGACTGTGGTCAAGTTCCCCACC   990

K A H T T P W G L F Y S W S T A S L D S Q M P S S L H P T Q   330
AAAGCCCATACAACCCCTGGGGTCTATTCTACTCATGGTCCACTGCTTCTCTAGACAGCCAAATGCCCTCCTCCTTGACATCCAACACAA   1080

E S T K E Q T T F P P R W T P N F T L H M E S I T F S K T P   360
GAATCCACTAAGGAGCAGACCACATTCACCTAGATGGACCCCAAAATTTACACTTCACATGGAATCCATCACATTCTCCAAAACCTCA   1170

M E S I T F S K T P T T S E P V P E P A P N M T T L E P T P   390
AAATCCACTACTGAACCAACCCCAAGCCGACACCTCAGAGCCCGTCCCGGAGCCCGCCCAACATGACCACCCCTGGAGCCCACTCCA   1260

(399) (VNTR length polymorphism) (411) (9) (27 bp del)(10)
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S P T T P E P T S E P A P S P T T P E P T P I P T I A T S P 420
 AGCCCGACCACCCAGAGCCACCTCAGAGCCCGCCCGAGCCGACCACCCCGAGCCCAATCCCGACCATCGCCACAAGCCCG 1350

T I L V S A T S L I T P K S T F L T T T K P V S L L E S T K 450
ACCATCCTGGTCTGCCACAAGCCTGATCACTCCAAAAAGCACATTTTTAACTACCACAAAACCCGTATCACTCTTAGAATCCACCAAA 1440

K T I P E L D Q P P K L R G V L Q G H L E S S R N D P F L H 480
 AAAACCATCCCTGAACTTGATCAGCCACCAAAGCTCCGTGGGGTGTCCAAGGGCATTGGAGAGCTCCAGAAATGACCCTTTTCTCCAC 1530

P D F C C L L P L G F Y V L G L F W L L F A S V V L I L L L 510
 CCCGACTTTTGTGCTCCTCCCCCTGGGCTTCTATGTCTTGGGTCTTCTGGGTGCTCTTGCCTCTGTGGTCTCATCTGCTGCTG 1620

S W V G H V K P Q A L D S G Q G A A L T T A T Q T T H L E L 540
 AGCTGGGTTGGGCATGTGAAACCACAGGCCCTGGACTCTGGCCAAGGTGCTGCTTGACCACAGCCACACAAACCACACACCTGGAGCTG 1710

Q R G R Q V T V P R A W L L F L R G S L P T F R S S L F L W 570
 CAGAGGGGACGGCAAGTGACAGTGCCTCCCGGCTGGTGTCTTCTCCTCAGAGTTGCTTCCCACTTTCCGCTCCAGCCTCTTCTGTGG 1800

V R P N G R V G P L V A G R R P S A L S Q G R G Q D L L S T 600
 GTACGGCCTAATGGCCGTGTGGGCTCTAGTGGCAGGAAGGAGGCCCTCAGCTCTGAGTCAGGGTCTGGTTCAGGACCTGCTGAGCACA 1890

V S I R Y S G H S L STOP
 GTGAGCATTAGGTA CTGGCCACAGCCTCTGA

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1. V. Afshar-Khargan et al. Kozak sequence polymorphism of the GPIba gene is a major determinant of the plasma membrane levels of the platelet GPIb-IX-V complex. *Blood*, 1999; 94 (1) 186–91.
 2. Yumiko Matsubara, Mitsuru Murata, Takanori Moriki, Kenji Yokoyama, Naohide Watanabe, Hideaki Nakajima, Makoto Handa, Koichi Kawano, Nobuo Aoki, Hideaki Yoshino, Yasuo Ikeda. A Novel Polymorphism, 70Leu/Phe, Disrupts a Consensus Leu Residue within the Leucine-rich Repeat Sequence of Platelet Glycoprotein Ib alpha. *Thromb Haemost* 2002; 87: 867–72
 3. Murata M, Furihata K, Ishida F, Russell S R, Ware J, Ruggeri ZM. Genetic and Structural Characterization of an Amino Acid Dimorphism in Glycoprotein Iba Involved in Platelet Transfusion Refractoriness. *Blood* 1992; 79: 3086-3090
 4. Miller JL, Cunningham D, Lyle VA, Finch CN. Mutation in the gene encoding the alpha chain of platelet glycoprotein Ib in platelet-type von Willebrand disease. *Proc Natl Acad Sci U S A*. 1991; 88: 4761–4765.
 5. Matsubara Y, Murata M, Sugita K, Ikeda Y. Identification of a novel point mutation in platelet glycoprotein Ibalpha, Gly to Ser at residue 233, in a Japanese family with platelet-type von Willebrand disease. *J Thromb Haemost*. 2003; 2198-2205.
 6. Nurden P, Lanza F, Bonnafous-Faurie C, Nurden A. A second report of platelet-type von Willebrand disease with a Gly233Ser mutation in the GPIBA gene. *Thromb Haemost*. 2007;97:319-321.
 7. Takahashi H, Murata M, Moriki T, Anbo H, Furukawa T Nikkuni K, Shibata A, Handa M, Kawai Y, Watanabe K, Ikeda Y: Substitution of Val for Met at residue

239 of in Japanese patients with platelet-type vonαplatelet glycoprotein Ib
Willebrand disease. Blood 1995;3727-3733

8. Russell, SD, Roth, GJ: von Willebrand disease: A mutation in the platelet gene associated with a hyperactive surface receptor. Bloodαglycoprotein Ib 1993;1787-1791
9. López JA, Ludwig EH, McCarthy BJ. Polymorphism of human glycoprotein Ib alpha results from a variable number of tandem repeats of a 13-amino acid sequence in the mucin-like macroglycopeptide region. Structure/function implications. J Biol Chem. 1992;267:10055-10061
10. Othman M, Notley C, Lavender FL, White HE, Byrne CD, Lillicrap D and O'Shaughnessy DF: Identification and functional characterisation of a novel 27bp deletion in the macroglycopeptide-coding region of the GPIb alpha gene resulting in platelet-type von Willebrand Disease. Blood 2005; 105:4330-4336