

Mutations in the *GP1BB* gene

Nucleotide and amino acid sequence for human GPIIb subunit.

Mutations are highlighted and described in the corresponding references listed below. Missense mutations are indicated in orange, nonsense mutations in red, and mutations causing a frameshift in green.

The sequence numbering is according to the *GP1BB* sequence of NCBI36 (NM_000407.4).

P-8^{2,3}

-133⁴ C.....TGCCGTCCTTCTCGCCATGGGGTCCGGGCGCGGGGGCGCTGAGCTTACTGCTCCTGCTGCTGGCCCCGCCGAGCC

-25--M--G--S--G--P--R--G--A--L--S--L--L--L--L--L--L--L--A--P--P--S--

84 GCCCGGCCGCAGGTGCCCGGCCCTGT^{C5Y¹}AGTCGCGGGGACGCTCGTGGACTGCGGGCGCR17C⁵ R17⁶ W21⁷ S23^{8,9}

-5 -R--P--A--A--G--C--P--A--P--C--S--C--A--G--T--L--V--D--C--G--R--R--G--L--T--W--A--S--L-

170 CCGACCGCCTTCC^{P29L¹⁰}GTTCTGTCGACACAACCGAGCTGTGTGCTGACCGGCAACAACTGACGGCCTGCCCGGGGCTGCTGGACCGCCTG

25 -P--T--A--F--P--V--D--T--T--E--L--V--L--T--G--N--N--L--T--A--L--L--P--P--G--L--L--D--A--L-

256 CCCGCGCTGCGCACCGCACACCTGGCGCCA^{N64T¹¹ P65R⁶}ACCGCTGGCGTGCGACTGCGCCTTGTG^{P74R¹²}CGCTGCGCGCTGGCTGGCCCCGCCG

54 -P--A--L--R--T--A--H--L--G--A--N--P--W--R--C--D--C--R--L--V--P--L--R--A--W--L--A--G--R-

342 CCCGAGCGT^{Y88C^{14,15}}GCGCCTACCGGACCTGCGTTGCGTGCCGCGCCCA^{A108P¹⁴}GCGCGCGCCTGCTGCCC^{A108P¹⁴}TATCTGCCGAGGACGAG

83 -P--E--R--A--P--Y--R--D--L--R--C--V--A--P--P--A--L--R--G--R--L--L--P--Y--L--A--E--D--E-

428 CTGCGCGCCGCTTGCGCT^{C122S¹⁶ W123¹⁶ W123¹⁷}TGGCGCGCGCGCCCA^{Q129H⁶ A131¹⁸ L132P⁶}GCGCGCGCCTGGGCTGGGCTGCTGCA^{H139^{19,20}}ACGG

112 -L--R--A--A--C--A--P--G--P--L--C--W--G--A--L--A--A--O--L--A--L--L--G--L--G--L--L--H--A-

514 TTGCTGCTGGTGTGCTGCTGCTGTGC^{C122S¹⁶ W123¹⁶ W123¹⁷}CGCCTGCGGAGGCTGCGGGCCCGCGCGCTCGCGCCGAGCCCGGCTGCTG^{H139^{19,20}}ACCGAC

141 -L--L--L--V--L--L--L--C--R--L--R--R--L--R--R--A--R--R--A--R--A--R--L--S--L--T--D-

600 CCGCTGGTGGCCGAGCGAGCCGGAACCGACGAGTCCTGA

170 -P--L--V--A--E--R--A--G--T--D--E--S--*--

See mutation described in :

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