Mutations in the *GP1BA* gene

Nucleotide and amino acid sequence for human GPIbα 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 *GP1BA* sequence of NCBI36 (NM_000173.4).

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   1 GACGCTCTGTGCCTTCGGAGGTCTTTCTGCCTGCCTGTCCTCATGCCTCTCCTCCTCTTGCTGCTCCTGCTGCCAAGC
     ..........................................-M--P--L--L--L--L--L--L--L--L--P--S-

                                                                       K19[1]
  79 CCCTTACACCCCCACCCCATCTGTGAGGTCTCCAAAGTGGCCAGCCACCTAGAAGTGAACTGTGACAAGAGGAATCTG
  -4 -P--L--H--P--H--P--I--C--E--V--S--K--V--A--S--H--L--E--V--N--C--D--K--R--N--L-

                                            L36[2]  S39[3,4] N41H[5]
 157 ACAGCGCTGCCTCCAGACCTGCCGAAAGACACAACCATCCTCCACCTGAGTGAGAACCTCCTGTACACCTTCTCCCTG
  23 -T--A--L--P--P--D--L--P--K--D--T--T--I--L--H--L--S--E--N--L--L--Y--T--F--S--L-

                   Y54D[6]   L57F[7]                C65R[8]
 235 GCAACCCTGATGCCTTACACTCGCCTCACTCAGCTGAACCTAGATAGGTGCGAGCTCACCAAGCTCCAGGTCGATGGG
  49 -A--T--L--M--P--Y--T--R--L--T--Q--L--N--L--D--R--C--E--L--T--K--L--Q--V--D--G-

                 L76[9]                                                 T98[10]
 313 ACGCTGCCAGTGCTGGGGACCCTGGATCTATCCCACAATCAGCTGCAAAGCCTGCCCTTGCTAGGGCAGACACTGCCT
  75 -T--L--P--V--L--G--T--L--D--L--S--H--N--Q--L--Q--S--L--P--L--L--G--Q--T--L--P-

                               N110D[11,12]
 391 GCTCTCACCGTCCTGGACGTCTCCTTCAACCGGCTGACCTCGCTGCCTCTTGGTGCCCTGCGTGGTCTTGGCGAACTC
 101 -A--L--T--V--L--D--V--S--F--N--R--L--T--S--L--P--L--G--A--L--R--G--L--G--E--L-

          L129P[13-15]
 469 CAAGAGCTCTACCTGAAAGGCAATGAGCTGAAGACCCTGCCCCCAGGGCTCCTGACGCCCACACCCAAGCTGGAGAAG
 127 -Q--E--L--Y--L--K--G--N--E--L--K--T--L--P--P--G--L--L--T--P--T--P--K--L--E--K-

             A156V[16-18]                                 Δ169-180 (E81K)[17]
 547 CTCAGTCTGGCTAACAACAACTTGACTGAGCTCCCCGCTGGGCTCCTGAATGGGCTGGAGAATCTCGACACCCTTCTC
 153 -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-

     L179[19,20]
 625 CTCCAAGAGAACTCGCTGTATACAATACCAAAGGGCTTTTTTGGGTCCCACCTCCTGCCTTTTGCTTTTCTCCACGGG
 179 -L--Q--E--N--S--L--Y--T--I--P--K--G--F--F--G--S--H--L--L--P--F--A--F--L--H--G-

       W207G[21] W207[22] C209S[11,23-26]
 703 AACCCCTGGTTATGCAACTGTGAGATCCTCTATTTTCGTCGCTGGCTGCAGGACAATGCTGAAAATGTCTACGTATGG
 205 -N--P--W--L--C--N--C--E--I--L--Y--F--R--R--W--L--Q--D--N--A--E--N--V--Y--V--W-

 781 AAGCAAGGTGTGGACGTCAAGGCCATGACCTCTAACGTGGCCAGTGTGCAGTGTGACAATTCAGACAAGTTTCCCGTC
 231 -K--Q--G--V--D--V--K--A--M--T--S--N--V--A--S--V--Q--C--D--N--S--D--K--F--P--V-

 859 TACAAATACCCAGGAAAGGGGTGCCCCACCCTTGGTGATGAAGGTGACACAGACCTATATGATTACTACCCAGAAGAG
 257 -Y--K--Y--P--G--K--G--C--P--T--L--G--D--E--G--D--T--D--L--Y--D--Y--Y--P--E--E-

                                        V295[27]
 937 GACACTGAGGGCGATAAGGTGCGTGCCACAAGGACTGTGGTCAAGTTCCCCACCAAAGCCCATACAACCCCCTGGGGT
 283 -D--T--E--G--D--K--V--R--A--T--R--T--V--V--K--F--P--T--K--A--H--T--T--P--W--G-

1015 CTATTCTACTCATGGTCCACTGCTTCTCTAGACAGCCAAATGCCCTCCTCCTTGCATCCAACACAAGAATCCACTAAG
 309 -L--F--Y--S--W--S--T--A--S--L--D--S--Q--M--P--S--S--L--H--P--T--Q--E--S--T--K-

                           W343[28] W343[29]    L365[30]
1093 GAGCAGACCACATTCCCACCTAGATGGACCCCAAATTTCACACTTCACATGGAATCCATCACATTCTCCAAAACTCCA
 335 -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-

1171 AAATCCACTACTGAACCAACCCCAAGCCCGACCACCTCAGAGCCCGTCCCGGAGCCCGCCCCAAACATGACCACCCTG
 261 -K--S--T--T--E--P--T--P--S--P--T--T--S--E--P--V--P--E--P--A--P--N--M--T--T--L-

                                                       S403[10]
1249 GAGCCCACTCCAAGCCCGACCACCCCAGAGCCCACCTCAGAGCCCGCCCCCAGCCCGACCACCCCGGAGCCCACCCCA
 287 -E--P--T--P--S--P--T--T--P--E--P--T--S--E--P--A--P--S--P--T--T--P--E--P--T--P-

1327 ATCCCGACCATCGCCACAAGCCCGACCATCCTGGTGTCTGCCACAAGCCTGATCACTCCAAAAAGCACATTTTTAACT
 313 -I--P--T--I--A--T--S--P--T--I--L--V--S--A--T--S--L--I--T--P--K--S--T--F--L--T-

              V443[26,27] S444[31]    K450[27,32,33]
1405 ACCACAAAACCCGTATCACTCTTAGAATCCACCAAAAAAACCATCCCTGAACTTGATCAGCCACCAAAGCTCCGTGGG
 339 -T--T--K--P--V--S--L--L--E--S--T--K--K--T--I--P--E--L--D--Q--P--P--K--L--R--G-

1483 GTGCTCCAAGGGCATTTGGAGAGCTCCAGAAATGACCCTTTTCTCCACCCCGACTTTTGCTGCCTCCTCCCCCTGGGC
 365 -V--L--Q--G--H--L--E--S--S--R--N--D--P--F--L--H--P--D--F--C--C--L--L--P--L--G-
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     Y492[22] Y492[4,10,15,34,35]  W498[8,36]
1561 TTCTATGTCTTGGGTCTCTTCTGGCTGCTCTTTGCCTCTGTGGTCCTCATCCTGCTGCTGAGCTGGGTTGGGCATGTG
 391 -F--Y--V--L--G--L--F--W--L--L--F--A--S--V--V--L--I--L--L--L--S--W--V--G--H--V-

1639 AAACCACAGGCCCTGGACTCTGGCCAAGGTGCTGCTCTGACCACAGCCACACAAACCACACACCTGGAGCTGCAGAGG
 417 -K--P--Q--A--L--D--S--G--Q--G--A--A--L--T--T--A--T--Q--T--T--H--L--E--L--Q--R-

1717 GGACGGCAAGTGACAGTGCCCCGGGCCTGGCTGCTCTTCCTTCGAGGTTCGCTTCCCACTTTCCGCTCCAGCCTCTTC
 443 -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-

1795 CTGTGGGTACGGCCTAATGGCCGTGTGGGGCCTCTAGTGGCAGGAAGGAGGCCCTCAGCTCTGAGTCAGGGTCGTGGT
 469 -L--W--V--R--P--N--G--R--V--G--P--L--V--A--G--R--R--P--S--A--L--S--Q--G--R--G-

1873 CAGGACCTGCTGAGCACAGTGAGCATTAGGTACTCTGGCCACAGCCTCTGAGGGTGGGAGGTTTGGGGACCTTGAGAG
 495 -Q--D--L--L--S--T--V--S--I--R--Y--S--G--H--S--L--*-.........................
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