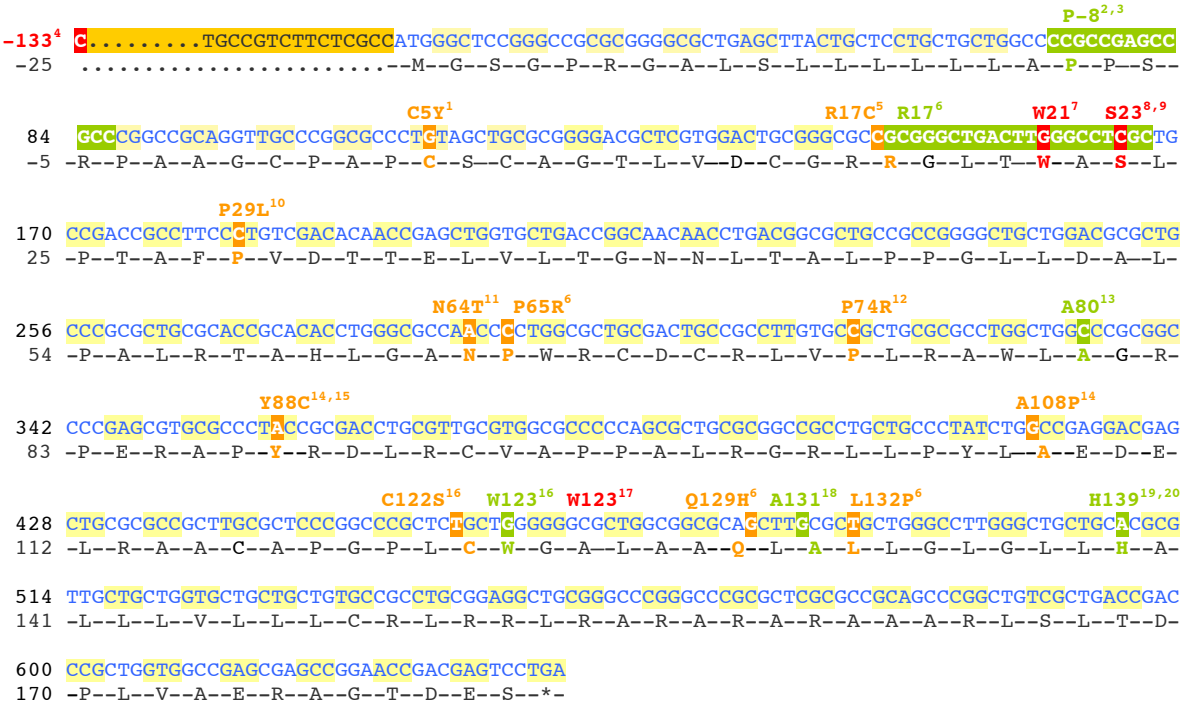
Mutations in the *GP1BB* gene

Nucleotide and amino acid sequence for human GPIbβ 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).



See mutation described in :

1. Gonzalez-Manchon C, Butta N, Iruin G, Alonso S, Ayuso MS, Parrilla R. *Disruption of the Cys5-Cys7 disulfide bridge in the platelet glycoprotein Ibbeta prevents the normal maturation and surface exposure of GPIb-IX complexes*. Thromb Haemost. 2003;90:456-464.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12958615
2. Watanabe R, Ishibashi T, Saitoh Y, et al. *Bernard-soulier syndrome with a homozygous 13 base pair deletion in the signal peptide-coding region of the platelet glycoprotein Ib(beta) gene*. Blood Coagul Fibrinolysis. 2003;14:387-394.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12945881
3. Strassel C, Alessi MC, Juhan-Vague I, Cazenave JP, Lanza F. *A 13 base pair deletion in the GPIbbeta gene in a second unrelated Bernard-Soulier family due to slipped mispairing between direct repeats*. J Thromb Haemost. 2004;2:1663-1665.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=15333045
4. Ludlow LB, Schick BP, Budarf ML, et al. *Identification of a mutation in a GATA binding site of the platelet glycoprotein Ibbeta promoter resulting in the Bernard-Soulier syndrome*. J Biol Chem. 1996;271:22076-22080.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=8703016
5. Kunishima S, Naoe T, Kamiya T, Saito H. *Novel heterozygous missense mutation in the platelet glycoprotein Ib beta gene associated with isolated giant platelet disorder*. Am J Hematol. 2001;68:249-255.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=11754414
6. Sumitha E, Jayandharan GR, David S, et al. *Molecular basis of Bernard-Soulier syndrome in 27 patients from India*. J Thromb Haemost. 2011;9:1590-1598.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=21699652
7. Moran N, Morateck PA, Deering A, et al. *Surface expression of glycoprotein ib alpha is dependent on glycoprotein ib beta: evidence from a novel mutation causing Bernard-Soulier syndrome*. Blood. 2000;96:532-539.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=10887115
8. Hadjkacem B, Elleuch H, Trigui R, Gargouri J, Gargouri AF. *The same genetic defect in three Tunisian families with Bernard Soulier syndrome: a probable founder Stop mutation in GPIbbeta*. Ann Hematol. 2009.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=19484238
9. Hadjkacem B, Elleuch H, Gargouri J, Gargouri A. *Bernard-Soulier syndrome: novel nonsense mutation in GPIbbeta gene affecting GPIb-IX complex expression*. Ann Hematol. 2009;88:465-472.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=18825380
10. Hillmann A, Nurden A, Nurden P, et al. *A novel hemizygous Bernard-Soulier Syndrome (BSS) mutation in the amino terminal domain of glycoprotein (GP)Ibbeta--platelet characterization and transfection studies*. Thromb Haemost. 2002;88:1026-1032.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12529755
11. Strassel C, Pasquet JM, Alessi MC, et al. *A novel missense mutation shows that GPIbbeta has a dual role in controlling the processing and stability of the platelet GPIb-IX adhesion receptor*. Biochemistry. 2003;42:4452-4462.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12693941
12. Kunishima S, Tomiyama Y, Honda S, et al. *Homozygous Pro74-->Arg mutation in the platelet glycoprotein Ibbeta gene associated with Bernard-Soulier syndrome*. Thromb Haemost. 2000;84:112-117.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=10928480

13. Kenny D, Morateck PA, Gill JC, Montgomery RR. *The critical interaction of glycoprotein (GP) IBbeta with GPIX-a genetic cause of Bernard-Soulier syndrome*. Blood. 1999;93:2968-2975.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=10216092
14. Kunishima S, Lopez JA, Kobayashi S, et al. *Missense mutations of the glycoprotein (GP) Ib beta gene impairing the GPIb alpha/beta disulfide linkage in a family with giant platelet disorder*. Blood. 1997;89:2404-2412.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=9116284
15. Kurokawa Y, Ishida F, Kamijo T, et al. *A missense mutation (Tyr88 to Cys) in the platelet membrane glycoprotein Ibbeta gene affects GPIb/IX complex expression--Bernard-Soulier syndrome in the homozygous form and giant platelets in the heterozygous form*. Thromb Haemost. 2001;86:1249-1256.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=11816714
16. Kunishima S, Sako M, Yamazaki T, Hamaguchi M, Saito H. *Molecular genetic analysis of a variant Bernard-Soulier syndrome due to compound heterozygosity for two novel glycoprotein Ibbeta mutations*. Eur J Haematol. 2006;77:501-512.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=16978236
17. Kunishima S, Matsushita T, Ito T, Kamiya T, Saito H. *Novel nonsense mutation in the platelet glycoprotein Ibbeta gene associated with Bernard-Soulier syndrome*. Am J Hematol. 2002;71:279-284.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12447957
18. Strassel C, David T, Eckly A, et al. *Synthesis of GPIb beta with novel transmembrane and cytoplasmic sequences in a Bernard-Soulier patient resulting in GPIb-defective signaling in CHO cells*. J Thromb Haemost. 2006;4:217-228.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=16409472
19. Balduini A, Malara A, Balduini CL, Noris P. *Megakaryocytes derived from patients with the classical form of Bernard-Soulier syndrome show no ability to extend proplatelets in vitro*. Platelets. 2011;22:308-311.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=21322749
20. Savoia A, Pastore A, De Rocco D, et al. *Clinical and genetic aspects of Bernard-Soulier syndrome: searching for genotype/phenotype correlations*. Haematologica. 2011;96:417-423.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=21173099