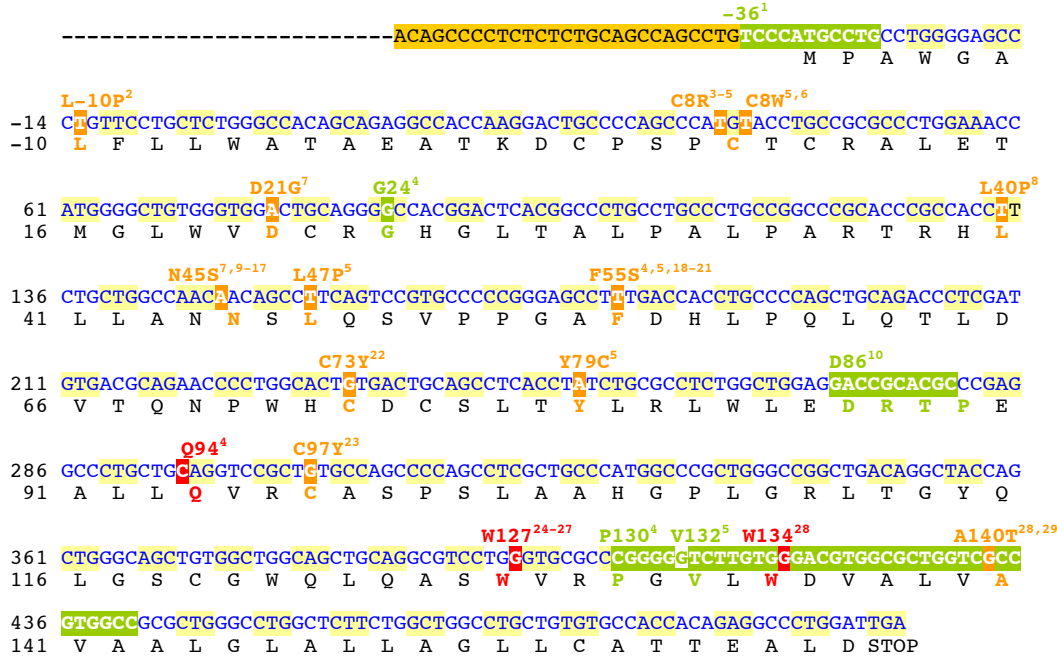


Mutations in the GP9 gene

Nucleotide and amino acid sequence for human GPIX 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 GP9 sequence of NCBI36 (NM_000174.2).



See mutation described in :

1. Sandrock K, Knofler R, Greinacher A, et al. *Novel Mutation in Bernard-Soulier Syndrome*. *Transfus Med Hemother*. 2010;37:278-284.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=21113250
2. Lanza F, De La Salle C, Baas MJ, et al. *A Leu7Pro mutation in the signal peptide of platelet glycoprotein (GP)IX in a case of Bernard-Soulier syndrome abolishes surface expression of the GPIb-V-IX complex*. *Br J Haematol*. 2002;118:260-266.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12100158
3. Rivera CE, Villagra J, Riordan M, Williams S, Lindstrom KJ, Rick ME. *Identification of a new mutation in platelet glycoprotein IX (GPIX) in a patient with Bernard-Soulier syndrome*. *Br J Haematol*. 2001;112:105-108.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=11167791
4. 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
5. 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
6. 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
7. Wright SD, Michaelides K, Johnson DJ, West NC, Tuddenham EG. *Double heterozygosity for mutations in the platelet glycoprotein IX gene in three siblings with Bernard-Soulier syndrome*. *Blood*. 1993;81:2339-2347.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=8481514
8. Noris P, Arbustini E, Spedini P, Belletti S, Balduini CL. *A new variant of Bernard-Soulier syndrome characterized by dysfunctional glycoprotein (GP) Ib and severely reduced amounts of GPIX and GPV*. *Br J Haematol*. 1998;103:1004-1013.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=9886312
9. Clemetson JM, Kyrle PA, Brenner B, Clemetson KJ. *Variant Bernard-Soulier syndrome associated with a homozygous mutation in the leucine-rich domain of glycoprotein IX*. *Blood*. 1994;84:1124-1131.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=8049428
10. Drouin J, Carson NL, Laneuville O. *Compound heterozygosity for a novel nine-nucleotide deletion and the Asn45Ser missense mutation in the glycoprotein IX gene in a patient with Bernard-Soulier syndrome*. *Am J Hematol*. 2005;78:41-48.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=15609295
11. Rand ML, Wang H, Bang KW, et al. *Phosphatidylserine exposure and other apoptotic-like events in Bernard-Soulier syndrome platelets*. *Am J Hematol*. 2010;85:584-592.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=20658588

12. Dagistan N, Kunishima S. *First Turkish case of Bernard-Soulier syndrome associated with GPIX N45S*. *Acta Haematol*. 2007;118:146-148.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=17804902
13. Liang HP, Morel-Kopp MC, Clemetson JM, et al. *A common ancestral glycoprotein (GP) 9 1828A>G (Asn45Ser) gene mutation occurring in European families from Australia and Northern Europe with Bernard-Soulier Syndrome (BSS)*. *Thromb Haemost*. 2005;94:599-605.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=16268478
14. Sachs UJ, Kroll H, Matzdorff AC, Berghofer H, Lopez JA, Santoso S. *Bernard-Soulier syndrome due to the homozygous Asn-45Ser mutation in GPIX: an unexpected, frequent finding in Germany*. *Br J Haematol*. 2003;123:127-131.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=14510954
15. Vanhoorelbeke K, Schlammadinger A, Delville JP, et al. *Occurrence of the Asn45Ser mutation in the GPIX gene in a Belgian patient with Bernard Soulier syndrome*. *Platelets*. 2001;12:114-120.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=11297032
16. Koskela S, Javela K, Jouppila J, et al. *Variant Bernard-Soulier syndrome due to homozygous Asn45Ser mutation in the platelet glycoprotein (GP) IX in seven patients of five unrelated Finnish families*. *Eur J Haematol*. 1999;62:256-264.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=10227459
17. Donner M, Karpman D, Kristofferson AC, Winqvist I, Holmberg L. *Recurrent mutation Asn45-->Ser of glycoprotein IX in Bernard-Soulier syndrome*. *Eur J Haematol*. 1996;57:178-179.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=8856096
18. Suzuki K, Hayashi T, Yahagi A, et al. *Novel point mutation in the leucine-rich motif of the platelet glycoprotein IX associated with Bernard-Soulier syndrome*. *Br J Haematol*. 1997;99:794-800.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=9432024
19. Afrasiabi A, Lecchi A, Artoni A, et al. *Genetic characterization of patients with Bernard-Soulier syndrome and their relatives from Southern Iran*. *Platelets*. 2007;18:409-413.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=17763149
20. Suzuki K, Hayashi T, Akiba J, Satoh S, Kato T. *Phenotypic consequence of the gene abnormality in the platelet glycoprotein IX gene observed in a patient with Bernard-Soulier syndrome through mammalian cell expression system*. *Thromb Res*. 1999;95:295-302.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=10527407
21. Noris P, Simsek S, Stibbe J, von dem Borne AE. *A phenylalanine-55 to serine amino-acid substitution in the human glycoprotein IX leucine-rich repeat is associated with Bernard-Soulier syndrome*. *Br J Haematol*. 1997;97:312-320.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=9163595
22. Noda M, Fujimura K, Takafuta T, et al. *A point mutation in glycoprotein IX coding sequence (Cys73 (TGT) to Tyr(TAT)) causes impaired surface expression of GPIb/IX/V complex in two families with Bernard-Soulier syndrome*. *Thromb Haemost*. 1996;76:874-878.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=8972003
23. Kunishima S, Tomiyama Y, Honda S, et al. *Cys97-->Tyr mutation in the glycoprotein IX gene associated with Bernard-Soulier syndrome*. *Br J Haematol*. 1999;107:539-545.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=10583255
24. Noda M, Fujimura K, Takafuta T, et al. *Heterogeneous expression of glycoprotein Ib, IX and V in platelets from two patients with Bernard-Soulier syndrome caused by different genetic abnormalities*. *Thromb Haemost*. 1995;74:1411-1415.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=8772211
25. Kunishima S, Yamada T, Hamaguchi M, Saito H. *Bernard-Soulier syndrome due to GPIX W127X mutation in Japan is frequently misdiagnosed as idiopathic thrombocytopenic purpura*. *Int J Hematol*. 2006;83:366-367.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=16757441
26. Toyohama T, Nagasaki A, Gushi K, Tamaki K, Masuda M, Takasu N. *Recurrent mutation Trp126 --> stop of glycoprotein IX in Japanese Bernard-Soulier syndrome*. *Platelets*. 2003;14:197-198.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=12850844
27. Iwanaga M, Kunishima S, Ikeda S, Tomonaga M, Naoe T. *Vulnerable mutation Trp126-->stop of glycoprotein IX in Japanese Bernard-Soulier syndrome*. *Eur J Haematol*. 1998;60:264-266.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=9579882
28. Xu LM, Sun GB, Wang P, et al. *Single novel mutation in transmembrane region of glycoprotein (GP) IX affects GP Ib-IX complex expression and causes Bernard-Soulier syndrome*. *Br J Haematol*. 2010;150:627-629.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=20497174
29. Wang Z, Zhao X, Duan W, et al. *A novel mutation in the transmembrane region of glyco-protein IX associated with Bernard-Soulier syndrome*. *Thromb Haemost*. 2004;92:606-613.
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=PubMed&dopt=Citation&list_uids=15351858