

Table 1: Genetic defects in Bernard-Soulier syndrome

Gene	Missense mutations or short deletions	Nonsense mutations	Frameshift ->Stop	Others	References	
GPIbα	Leu57->Phe				Miller HL <i>et al</i> , 1992	
	Cys65->Arg				Kenny D <i>et al</i> , 1998	
	Leu 123->Pro				Proulle V <i>et al</i> , unpublished	
	Leu126->Phe				Furihata K <i>et al</i> , unpublished	
	Leu129->Pro*				Li C <i>et al</i> , 1995 ; Antonucci JV <i>et al</i> , 2000 ; Koskela S <i>et al</i> , 1999	
	Ala156->Val*				Ware J <i>et al</i> , 1993; Lanza <i>et al</i> , unpublished; Margaglione M <i>et al</i> , 1999 ; Savoia A <i>et al</i> , 2001	
	Δ Leu179*				de la Salle C <i>et al</i> , 1995; Ulsemer P <i>et al</i> , 2000 ; Ulsemer P <i>et al</i> , 2001	
	Δ 169-180/Glu81->Lys				Margaglione M <i>et al</i> , 1999	
	Cys209->Ser*				Simsek S <i>et al</i> , 1994; Gonzalez-Manchon C <i>et al</i> , 2001	
			Thr294->Stop			
			Trp343->Stop			Ware J <i>et al</i> , 1990
			Ser444->Stop*			Kunishima S <i>et al</i> , 1994
			Trp498->Stop*			Holmberg L <i>et al</i> , 1997
				Lys19->Arg + 2AA->Stop		Li C <i>et al</i> , 1996
				Δ Ser39Glu40 + 51AA->Stop		Afshar-Kharghan V <i>et al</i> , 2000
				Arg->Leu76 + 19AA->Stop		Simsek S <i>et al</i> , 1994
			Val295->Gly + 39AA->Stop		Kanaji T <i>et al</i> , 1997	
			Ser444->Ile + 11AA->Stop (het)		Kanaji T <i>et al</i> , 1997; Gonzalez-Manchon C <i>et al</i> , 2001	
			Thr->Pro 452 + 58 AA->Stop		Noda M <i>et al</i> , 1995 ;1996 ; Mitsui T <i>et al</i> , 1998 ; Kanaji T <i>et al</i> , 1997	
			Tyr492->Cys + 80AA->Stop		Afshar-Kharghan V and Lopez JA, 1997; Kenny D <i>et al</i> , 1997; Koskela S <i>et al</i> , 1999	
GPIbβ	Cys5->Tyr				Gonzalez-Manchon C <i>et al</i> , 2003	
	Arg17->Cys (Giant platelets)				Kunishima S <i>et al</i> , 2001	
	Pro29->Leu (het)				Hillmann A <i>et al</i> , 2002	
	Asn64->Thr				Strassel C <i>et al</i> , 2003	
	Pro74->Arg				Kunishima S <i>et al</i> , 2000	
	Tyr88->Cys				Kurokawa Y <i>et al</i> , 2001	
	Tyr88->Cys +Ala108-Pro (het) (Giant platelets)				Kunishima S <i>et al</i> , 1997	
	Pro96->Ser + Di George-VCF				Tang J <i>et al</i> , 2004	
			Trp21->Stop			Moran N <i>et al</i> , 2000
			Cys116->Stop			de la Salle C <i>et al</i> , 1997
		Trp123->Stop			Kunishima S <i>et al</i> , 2002	

			Gly81->Ala + 86AA->Stop +		Kenny D <i>et al</i> , 1999
			Δ13bp Signal sequence + 22AA->Stop		Watanabe R <i>et al</i> , 2003; Strassel <i>et al</i> , 2004
			Ala131->Gly +153AA->Stop		Strassel <i>et al</i> , unpublished
				Promoter Mutation GATA + Di George-VCF	Budarf ML <i>et al</i> , 1995 ; Ludlow LB <i>et al</i> , 1996
GPIX	Leu7->Pro				Lanza F <i>et al</i> , 2002
	Cys8->Arg				Rivera CE <i>et al</i> , 2001
	Asp21->Gly (het)				Wright SD <i>et al</i> , 1993
	Leu40->Pro				Noris P <i>et al</i> , 1998
	Asn45->Ser (homo and het)				Wright SD <i>et al</i> , 1993; Clemetson JM <i>et al</i> , 1994 ; Donner M <i>et al</i> , 1996 ; de la Salle <i>et al</i> , 1997 ; Koskela S <i>et al</i> , 1999 ; Vanhoorelbeke K <i>et al</i> , 2001; Sachs UJ <i>et al</i> , 2003 ; Drouin J <i>et al</i> , 2005; Lanza F <i>et al</i> , unpublished
	Phe55->Ser				Noris P <i>et al</i> , 1997 ; Suzuki K <i>et al</i> , 1997 ; Suzuki K <i>et al</i> , 1999
	Cys73->Tyr				Noda M <i>et al</i> , 1996
	Asn86->Ala ΔArg87-Pro89 (het)				Drouin J <i>et al</i> , 2005
	Cys97->Tyr				Kunishima S <i>et al</i> , 1999
	Ala140->Thr				Wang Z <i>et al</i> , 2004
		Trp126->Stop			Noda M <i>et al</i> , 1995 ; Iwanaga M <i>et al</i> , 1998 ; Toyohama T <i>et al</i> , 2003

Δ : deletion

Amino acids (AA) are in three letter code

(->) change from wild type to mutated AA sequence

Numbering is based on the mature sequence (except for the GPIX Leu7->Pro mutation)

Giant platelets: variant phenotype not a BSS

(het) compound heterozygote

di George-VCF: di George (velo-cardio-facial syndrome) deletion of chromosome region 22q11.2 on the second allele