Title: Bloom's Syndrome *GeneReview* Table 5

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 5. Bloom's Syndrome-Causing Mutations Identified in Registered Persons of Various Nationalities and Ethnic Groups

Nationality or Group	Nucleotide Change in the DNA	Amino Acid Change in the Protein
Sub-Saharan African	c.772_773delCT	p.Leu257fs
Ashkenazi Jewish	c.2207_2212delinsTAGATTC	p.Tyr736LeufsX5
	c.2407dupT	p.Trp803LeufsX4
Australian	c.2923delC	p.Gln975fs
Belgian	c.1088-2A>G	p.Ala363fs
Brazilian	c.2406+2T>G c.3278C>G c.3587delG del exons 20-22 [1]	p.770_802del p.Ser1093X p.Ser1196fs p.Glu1251fs
Dutch	c.2488_2489dupA c.3681delA	p.Thr830fs p.Lys1227fs
German	c.1933C>T c.3727_3728dupA c.3223_3224dupA	p.Gln645X p.Thr1243fs p.Arg1075fs
Hispanic	c.2887C>T c.2506_2507delAG c.3197G>A c.582delT c.2207_2212delinsTAGATTC	p.His963Tyr p.Arg836fs p.Cys1066Tyr p.Phe194fs p.Tyr736LeufsX5
Indian (Asian)	c.275delA	p.Asn92fs
Italian	c.2098C>T c.3164G>C c.311C>A c.3558+1G>A c.3847C>T c.2308-953_2555 + 4719del6126 c.3191A>T c.3475_3476delTT c.2855G>T	p.Gln700X p.Cys1055Ser p.Ser104X p.Ser1121fs p.Gln1283X p.Ile770fs p.Asp1064Val p.Lys1159fs p.Gly952Val
Japanese	c.557-559delCAA c.1544_1545dupA c.2074 + 1G>T	p.Ser186X p.Asn515fs p.628_691del
Portuguese	c.2406+2T>G del exons 20-22 [1]	p.770_802del p.Glu1251fs
Tunisian	c.3255-3256insT	p.Arg1086X
Turkish	c.1628T>A c.2643G>A	p.Leu543X p.Trp881X
Welsh	c.2193 + 2T>G	p.Gly692fs

1. The effect cannot be unambiguously inferred, because exon 22 contains the terminal codon and polyA addition signal. The first amino acid encoded by exon 20 is glutamic acid at 1251; therefore, an educated guess for the translational effect is p.Glu1251fs.