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The Role of Erythrocyte Membrane Proteins in Haemolytic Anaemias in South African Populations

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2006-11-16T10:35:04Z
http://hdl.handle.net/10539/1794

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Appendix A: Band 3 Mutations Resulting in Hereditary Spherocytosis

(adapted from Walensky *et al*, 2003)

Name	Affected Exon	Affected Codon	Designation	Genetic Mutation	Mutation Type	Consequence
Bohain	4	81	241delT	GC**T**GGG→CGGGG	Frameshift	PTC
Foggia	4	55	163delC	ACC**C**AC→ACCAC	Frameshift	PTC
Kagoshima	4	56	167delA	**A**AG→AG	Frameshift	PTC
Neapolis	2 nucleotides after exon 2	-	15+2t→c	T→c	Splicing	EITHER exon 2 skipping (initiation codon abolished) OR Retention of intron 2 (PTC)
Cape Town	5	90	E90K	**G**AG→**A**AG	Missense	Replacement of glutamic acid 90 with lysine
Fukayama I	5	112-113	336-337del2	AG**AG**TC→AGTC	Frameshift	PTC
Hodouin (Prague IV)	5	81	W81X	TG**G**→TG**A**	Nonsense	Replacement of tryptophan 81 with PTC
Napoli I	5	100	298-299ins1	TCTCT→TCT**T**CT	Frameshift	PTC
Fukuoka	6	130	G130R	**G**GA→**A**GA	Missense	Replacement of glycine 130 with arginine

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Lyon (Osnabrück II)	6	150	R150X	**C**GA→**T**GA	Nonsense	Replacement of arginine 150 with PTC
Fukuyama II	7	183	548-549insA	GAT→GA**A**T	Frameshift	PTC
Wilson	7	170-172	515delG	$G_{(6)}$→$G_{(5)}$	Frameshift	PTC
Worchester	7	170-172	515-516insG	$G_{(6)}$→$G_{(7)}$	Frameshift	PTC
Boston	9	285	A285D	G**C**T→G**A**T	Missense	Replacement of alanine 285 with aspartic acid
Campinas	**1nucleotide after exon 8**	204	694+1G>T	G→T	Splicing Frameshift	Eradication of splice site with the resultant skipping of exon 8 and PTC 13 amino acids downstream of the exon 7-exon 9 splice site as a result of a frameshift
Princeton	9	273-275	823-824insC	$C_{(6)}$→$C_{(7)}$	Frameshift	PTC
Noirterre	10	330	Q330X	**C**AG→**T**AG	Nonsense	Replacement of glutamine 330 with PTC

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Tuscaloosa	10	327	P327R	C**C**C→C**G**C	Missense	Replacement of proline 327 with arginine
Bruggen	11	418-419	1257delC	CACC**C**G→CACCG	Frameshift	PTC
Nachod (Hradec Kralove II)	Exon 5 to exon 6 splice site	117-121	350-3c→a	Replacement of a cytosine with an adenine in the acceptor splice site in intron 5 (three nucleotides downstream of position 350)	Splicing	Deletion of amino acids 117 to 121 as result of modified splicing
Yamagata	12	455	G455R	**G**GG→**A**GG	Missense	Replacement of glycine 455 with arginine
Benesov (Prague V)	12	455	G455E	G**G**G→G**A**G	Missense	Replacement of glycine 455 with glutamic acid
Bicetre II	12	456	1366delG 1475	CTGGGG**G**CT→CT GGGGCT	Frameshift	PTC at codon 1527 or 1565

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Pribram (Prague VI)	1 nucleotide after Intron 12	-	1431+1g→a	g→a	Splicing	Intron 12 is retained after codon 477, resulting in seven abnormal amino acids and a premature stop codon at triplet 8 of intron 12
Coimbra	13	488	V488M	**G**TG→**A**TG	Missense	Replacement of valine 488 with methionine
Bicetre I	13	490	R490C	**C**GC→**T**GC	Missense	Replacement of arginine 490 with cysteine
Pinhal (Bloemfontein)	13	490	R490H	C**G**C→C**A**C	Missense	Replacement of arginine 490 with histidine
Dresden	13	518	R518C	**C**G**C**→**T**GC	Missense	Replacement of arginine 518 with cysteine
Evry	13	496	1486delT	TTC**T**GG→TTCGG	Frameshift	PTC at position 1733
Milano	13	499	1497-1498ins69	Duplication of nucleotides 1432-1500	Insertion	Duplication of amino acids 478 to 499 and insertion of a glutamine
Smichov (Prague VII)	15	616	1848delC	AT**C**→AT	Frameshift	PTC

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Trutnov	15	628	Y628X	TA**C**→TA**A**	Nonsense	Replacement of a tyrosine with PTC
Hobart	16	646-647	1940delG	CGGG**G**C→CGGGC	Frameshift	PTC
Osnabruck II	16	663-664	DelM664	ATG**ATG**→ATG	Deletion	Deletion of methionine 664
Tochigi II	17 -5 nucleotides	-	2058-5delA	delA	Splicing	PTC
Most (Prague VIII)	17	707	L707P	C**T**G→C**C**G	Missense	Replacement of leucine 707 with a proline
Okinawa	17	714	G714R	GGG→A**A**G	Missense	Replacement of glycine 714 with arginine
Hradec Kralove (Tochigi I)	17	760	R760W	**C**GG→**T**GG	Missense	Replacement of arginine 760 with tryptophan
Kumamoto (Prague II)	17	760	R760Q	C**G**G→C**A**G	Missense	Replacement of arginine 760 with glutamine
Chur	18	771	G771D	G**G**C→G**A**C	Missense	Replacement of glycine 771 with aspartic acid
Napoli II	18	783	I783N	A**T**C→A**A**C	Missense	Replacement of isoleucine 783 with asparagine

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Jablonec	18	808	R808C	**C**GC→**T**GC	Missense	Replacement of arginine 808 with cysteine
Nara	18	808	R808H	C**G**C→C**A**C	Missense	Replacement of arginine 808 with histidine
Prague I	18	822	2464-2465ins10	TGTG→TG(10nt)TG	Frameshift	PTC
Birmingham	19	834	H834P	C**A**C→C**C**C	Missense	Replacement of histidine 834 with a proline
Nagoya	19	837	T837R	A**C**G→A**A**G	Missense	Replacement of threonine 837 with arginine
Philadelphia	19	837	T837M	A**C**G→A**T**G	Missense	Replacement of threonine 837 with methionine
Tokyo	19	837	T837A	**A**CG→**G**CG	Missense	Replacement of threonine 837 with alanine
Prague III	19	870	R870W	**C**GG→**T**GG	Missense	Replacement of arginine 870 with tryptophan
Vesuvio	20	894	2681delC	AC**C**→AC	Frameshift	Termination at codon 1026; elongated carboxy-terminal

PTC=premature termination codon

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