

Title: Sitosterolemia Mutations

Authors: Merkens L, Myrie S, Steiner R, Mymin D

Date: April 2013

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

Sitosterolemia mutations									
ref	total #	ABCG5		HMZ	# ABCG5	mutation #1	ABCG8	HMZ	# ABCG8
Lu 01									
	1 white					Trp361X	Arg412X		1
	2 white					Arg 543Ser	Gln172X		2
	3 white					Trp361X	Tyr658X		3
	4 white					Trp361X	IVS1-2A>G		4
	5 white					Trp361X	Trp361X	x	5
	6 white					Trp361X	Arg184His		6
	7 white					Trp361X	Leu501Pro		7
	8 white					Trp361X	Trp361X	x	8
	9 white					Leu596Arg	IVS1-2A>G		9
	10 white					Trp361X	Tyr658X		10
	11 white					Trp361X	Arg412X		11
	12 white					Arg121X	Arg121X	x	12
	13 white					Trp361X	Gly574Glu		13
	14 white					Trp361X			14
	15 Amish-Mennonite					Gly574Arg	Gly574Arg	x	15
	16 Amish-Mennonite					Gly574Arg	Gly574Arg	x	16
	17 white					Trp536X	Trp536X	x	17
	18 white					1798_1008 del TCC	Arg405His		18
	19 white					Trp361X	Trp361X	x	19
	20 white					Trp361X			20
	21 white					Trp361X	Trp361X	x	21
	22 white					Trp361X	Leu572Pro		22
	23 white					1568_1572delCTTT	IVS1-2A>G		23
	24 Asian					Arg164X	Arg121X		24
	25 Asian	Arg243X	Arg243X	x	1				
	26 Jap	Arg419His	Arg419His	x	2				
	27 Jap	Arg398His	Arg398His	x	3				
	28 Jap	del exon3	del exon3	x	4				
	29 Jap	Arg398His	Arg398His	x	5				
	30 Jap	Arg419His	Arg550Ser		6				
	31 Jap	Arg408X	Arg408X	x	7				
	32 Jap	Arg398His	Arg398His	x	8				
	33 white	Arg419Pro	Arg419Pro	x	9				
	34 African Am	Glu146Gln			10				
	35 white					Trp361X	Trp361X	x	25
	36 Amish					Gly574Arg	Gly574Arg	x	26
	37 white					Trp361X	Tyr658X		27

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ref	total #	mutation #1	ABCG5 mutation#2	HMZ	# ABCG5	mutation #1	ABCG8 mutation #2	HMZ	# ABCG8
	38 white					Trp361X	Arg412X		28
	39 white					Leu596Arg			29
	40 hispanic					Arg412X	del547C-191X		30
	41 white					Trp361X			31
	42 Chinese					Arg263Gln	Pro231Thr		32
	43 Chinese	Arg408X			11				
Wilund 04 1644									
	44 pacific islar	IVS1.1 G>T	IVS1.1 G>T	x	12				
Wang J 04 1669									
	45 Hutterite					S107X	S107X	x	33
	46 Jap/Canadi	eox3del388-428/i	eox3del388-428/ins388/408	x	13				
Sehayek E 04									
	47 Kosraens	Gln24His del X	Gln24His del X	x	14				
Heimerl S 02 1601									
	48 white	Arg408X			15				
	49 white					Leu195Gln			34
	50 white					Trp361X	Arg412X		35
	51 white					Trp361X	Trp361X	x	36
	52 white					Gly574Arg	Gly574Arg	x	37
Berge 00 381									
	53 white					Trp361X	Trp361X	x	38
	54 amish					Gly574Arg	Gly574Arg	x	39
	55 white					Trp361X	Tyr658X		40
	56 Chinese					Arg263Gln			41
	57 white					Trp361X	Arg412X		42
	58 white					Leu596Arg			43
	59 Mexican					Arg412X			44
	60 white					Trp361X			45
	61 Chinese	Arg408X			16				
Rios 10 1714									
	62	Q16X	R446X		17				
Hubacek 01 525									
	white					L596R	IVS1-2A>G	same individual as Berge #43	
	63 Chinese	c,335-336insA	lys437Asp		18				
Mannucci 07 1744									
	64 Iranian	Arg446X	Arg446X	x	19				

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ref	total #	ABCG5		HMZ	# ABCG5	ABCG8		HMZ	# ABCG8
		mutation #1	mutation#2			mutation #1	mutation #2		
Rees 05 1718									
	65	Bangladesh	E77X	x	20				
	66	European				IVS11+3insT	E146X		46
	67	European				Q271X	IV8-1G/A		47
	68	European				W361X	W361X	x	48
	69	European				W361X	W361X	x	49
Solca 05									
	70	Swiss				Gly574Arg	Gly574Arg	x	50
Su 06 1717									
	71	Chinese	Gln22X	x	21				
Tsubakio 10 1710									
	72	Japanese	R550S		22				
	73	Japanese	R389H		23				
Kratz, M 07									
	74	?				Q442X	L572P		51
	75	?	R446X		24				
Nui 10 1711									
	76	Chinese	Y329X		25				
	77	Chinese	R389H		26				
	78	Chinese	R389H	x	27				
	79	Chinese	R389H		28				
Togo M 09									
	80	Japanese	19 bp ins exon 7		29				
	81		19 bp ins exon 7		30				
Chong 201:									
	82	Hutterite				S107X	S107X	x	52
	83	Hutterite				S107X	S107X	x	53
	84	Hutterite				S107X	S107X	x	54
	85	Hutterite				S107X	S107X	x	55
Keller 2011 X		Arg406Gln				Thr400Lys			