

Gene	Mutant designation	Nucleotide aberration	Location in gene
ALDH5A1	C93_R99dup	c.278_298dup	Exon 1
ALDH5A1	P382L	c.1145C>T	Exon 7
ALDH7A1	R82X	c.244C>T	Exon 4
ALDH7A1	A171V	c.XXXC>T	Exon 6
ALDH7A1	Y380X	c.XXXT>G	Exon 14
ALDH7A1	E399Q	c.1195G>C	Exon 14
ALDH7A1	IVS3+2T>A	c.230T>A	Intron 3
ALDH7A1	IVS5-1G>C	c.433G>C	Intron 5
ALDH7A1	c.1512delG		
ALDH7A1	N273I	c.818A>T	Exon 10
ALDH7A1	R307Q	c.920G>A	Exon 11
ALDH7A1	S430N	c.1289G>A	Exon 15
ALDH7A1	K120fs	c.567_611del	Exon 7
ALDH7A1	IVS17-1G>T	c.1481G>T	Intron 17
ALDH7A1	R307X	c.919C>T	Exon 11
ALDH7A1	G174V	c.521G>T	Exon 6
ALDH7A1	V367G	c.1100T>G	Exon 13
ALDH7A1	G263E	c.788G>A	Exon 10
ALDH7A1	IVS1+3A>T	c.108+3A>T	Intron 1
ALDH7A1	P403L	c.1208C>T	Exon 4
ALDH7A1	G378R	c.1132G>A	Exon 14
ALDH7A1	D499N	c.1345G>A	Exon 16
ALDH7A1	W31X	c.93G>A	Exon 1
ALDH7A1	1937-bp del		
DBH	V87M	c.259G>A	Exon 1
DBH	IVS1+2T>C	c.297+2T>C	Intron 1
DBH	D100E	c.300C>A	Exon 2
DBH	G192fs	c.575_576delA	Exon 3
DBH	C255F	c.764G>T	Exon 4
DBH	D331N	c.991C>A	Exon 6
DBH	Y542C	c.1625A>G	Exon 11
DDC	P47H	c.140C>A	Exon 2
DDC	A91V	c.272C>T	Exon 3
DDC	G102S	c.304G>A	Exon 3
DDC	S147R	c.439A>C	Exon 5
DDC	S250F	c.749C>T	Exon 7
DDC	A275T	c.823G>A	Exon 8
DDC	F309F	c.925C>T	Exon 9
DDC	R347Q	c.1040G>A	Exon 11
DDC	L408I	c.1222C>T	Exon 13
DDC	delGAGA	c.-64_-61delGAGA	
DDC	R285W	c.853C>T	Exon 8
DDC	E25K		Exon 2
DDC	V460G		
DDC	R447H	c.1340G>A	Exon 14
DDC	R358H		
DDC	IVS6+4A>T	c.1297_1298insA	Intron 6
DDC	R462P		Exon 14
DDC	R7X	c.19C>T	Exon 2

DDC	I42fs	c.127delC	Exon 2
DDC	L38P	c.113T>C	Exon 2
DDC	R412W	c.1234C>T	Exon 13
DDC	A110Q	c.329C>A	Exon 4
DDC	I433fs		Exon 14
DDC	H72Y	c.214C>T	Exon 3
DDC	T69M	c.206C>T	Exon 3
DDC	H70T	c.208C>T	Exon 3
DDC		c.236A>G	
DDC	AA 357		Exon 12
DDC	Y79C	c.236A>G	Exon 3
GCH1	F234S	c.701T>C	Exon 6
GCH1	I171fs	c.512_524del13	Exon 4
GCH1	Q110X	c.328C>T	Exon 1
GCH1	S114X	c.341C>A	Exon 1
GCH1	IVS1-1G>A	c.344-1G>A	Intron 1
GCH1	D134V	c.401A>T	Exon 2
GCH1	R88W	c.262C>T	Exon 1
GCH1	H153P	c.458A>C	Exon 3
GCH1	L79P	c.236T>C	Exon 1
GCH1	K224R	c.671A>G	Exon 6
GCH1	R216X	c.646C>T	Exon 6
GCH1	M211I	c.633G>A	Exon 6
GCH1	G203R	c.607G>A	Exon 5
GCH1	G201E	c.602G>A	Exon 5
GCH1	R184H	c.551G>A	Exon 5
GCH1	IVS2+1G>C	c.453+1G>C	Intron 2
GCH1	E2fs	c.3_4insGG	Exon 1
GCH1	E65X	c.193G>T	Exon 1
GCH1	R88P	c.263G>C	Exon 1
GCH1	R178S	c.534A>C	Exon 4
GCH1	T186fs	c.557C>A	Exon 5
GCH1	H144P	c.431A>C	Exon 2
GCH1	P23L	c.68C>T	Exon 1
GCH1	M102K	c.305T>A	Exon 1
GCH1	C141W	c.423T>G	Exon 2
GCH1	Q182X	c.544C>T	Exon 5
GCH1	IVS1-2A>G	c.344-2A>G	Intron 1
GCH1	IVS2-2A>G	c.454-2A>G	Intron 2
GCH1	K224X	c.670A>T	Exon 6
GCH1	D115N	c.343G>A	Exon 1
GCH1	M1X	c.3G>C	Exon 1
GCH1	Q103fs	c.309delG	Exon 1
GCH1	M221T	c.662T>C	Exon 6
GCH1	A50fs	c.149delC	Exon 1
GCH1	L71fs	c.212delT	Exon 1
GCH1	S77_L82del	c.229_246del18	Exon 1
GCH1	L117fs	c.351delA	Exon 2
GCH1	IVS3-2G>A	c.510-2G>A	Intron 3
GCH1	Q110fs	c.328_329insA	Exon 1

GCH1	G108D	c.323G>A	Exon 1
GCH1	S176T	c.527G>C	Exon 4
GCH1	E61X	c.181G>T	Exon 1
GCH1	IVS5+1G>A	c.626+1G>A	Intron 5
GCH1	E56X	c.166G>T	Exon 1
GCH1	L71Q	c.212T>A	Exon 1
GCH1	A74V	c.221C>T	Exon 1
GCH1	-39C>T / -132C>T	-39C>T / -132C>T	5'-UTR
GCH1	V191I	c.571G>A	Exon 5
GCH1	R241W	c.721C>T	Exon 6
GCH1	M211V	c.631A>G	Exon 6
GCH1	M211fs	c.631_632delAT	Exon 6
GCH1	I135K	c.404T>A	Exon 2
GCH1	S196A	c.586G>T	Exon 5
GCH1	G108V	c.323G>T	Exon 1
GCH1	W96X	c.288G>A	Exon 1
GCH1	T94K	c.281C>A	Exon 1
GCH1	R249S	c.747C>G	Exon 6
GCH1	G83A	c.248G>C	Exon 1
GCH1	M102R	c.305T>G	Exon 1
GCH1	C141R	c.421T>C	Exon 2
GCH1	Q180X	c.538C>T	Exon 4
GCH1	A190fs	c.568delG	Exon 5
GCH1	-22C>T	-22C>T	5'-UTR
GCH1	R88_Q89del	c.261_266delGCGGCA	Exon 1
GCH1	R178S	c.534A > T	Exon 4
GCH1	Q180R	c.539A > G	Exon 4
GCH1	P199L	c.596C > T	Exon 5
GCH1	L185R	c.554T>G	Exon 5
GCH1	IVS4+1G>C	c.541+1G>C	Intron 4
GCH1	Y75X	c.225C>A	Exon 1
GCH1	IVS5+3insT	c.626+2insT	Intron 5
GCH1	L163R	c.488T>G	Exon 3
GCH1	N159fs	c.477delC	Exon 3
GCH1	IVS5+5G>A	c.626+5G>A	Intron 5
GCH1	IVS3+1G>T	c.509+1G>T	Intron 3
GCH1	Q89X	c.265C>T	Exon 1
GCH1	M213T	c.638T>C	Exon 6
GCH1	Q48X	c.142C>T	Exon 1
GCH1	X251R	c.751T>C	Exon 6
GCH1	V226A	c.677T>C	Exon 6
GCH1	Q87P	c.260A>C	Exon 1
GCH1	del exon 1	?	Exon 1
GCH1	del exon 1-3	?	
GCH1	P199A	c.595C>G	Exon 5
GCH1	S114X	c.341C>G	Exon 1
GCH1	E243fs	c.726_727insTTCCC	Exon 6
GCH1	V205G	c.614T>G	Exon 5
GCH1	IVS5+1G>C	c.626+1G>C	Intron 5
GCH1	L231fs	c.693delG	Exon 6

GCH1	P199S	c.595C>T	Exon 5
GCH1	del exon 1-4	?	
GCH1	del exon 1-6	?	
GCH1	A8X	c.22_23GC>TA	Exon 1
GCH1	R88G	c.262C>G	Exon 1
GCH1	L91R	c.272T>G	Exon 1
GCH1	G103P	c.308A>C	Exon 1
GCH1	T106ins	c.316insTTC	Exon 1
GCH1	R178G	c.532A>G	Exon 4
GCH1	IVS4+1G>A	c.541+1G>A	Intron 4
GCH1	R184C	c.550C>T	Exon 5
GCH1	G201R	c.601G>A	Exon 5
GCH1	V202I	c.604G>A	Exon 5
GCH1	V218fs	c.654insT	Exon 6
GCH1	S223R	c.669C>A	Exon 6
GCH1	del exon 4-6	?	Exon 4-6
GCH1	del exon 2-6	?	Exon 2-6
GCH1	T106I	c.317C>T	Exon 1
GCH1	H210Q	c.630C>G	Exon 6
GCH1	del exon 3		Exon 3
GCH1	del exon 3-4		Exon 3-4
GCH1	I78fs	c.233delT	Exon 1
GCH1	E183K	c.547G>A	Exon 5
GCH1	G21fs	c.64_65delGGinsAACC	Exon 1
GCH1	del exon 3-6	?	
PCBD	E87X	g.9214G>T; c.282G>T	Exon 4
PCBD	C82R	g.9199T>C; c.267T>C	Exon 4
PCBD	Q98X	g.9247C>T; c.315C>T	Exon 4
PCBD	T79I	g.9191C>T; c.259C>T	Exon 4
PCBD	E27X	g.7367G>T; c.102G>T	Exon 2
PCBD	R88Q	g.9218G>A; c.286G>A	Exon 4
PCBD	E97K	g.9244G>A; c.312G>A	Exon 4
PCBD	E58K	g.8025G>A; c.195G>A	Exon 3
PCBD	K72del	g.8066_8068delCCA; c.236_238delCCA	Exon 3
PNPO	E50K	c.XXXG>A	Exon 2
PNPO	R229W	c.XXXC>T	Exon 7
PNPO	X262Q	c.XXXT>C	Exon 7
PNPO	IVS3-1G>A		Intron 3
PTS	D96N	g.7596G>A; c.286G>A	Exon 5
PTS	P87L	g.7570C>T; c.260C>T	Exon 5
PTS	P87S	g.7569C>T; c.259C>T	Exon 5
PTS	T67M	g.5030C>T; c.200C>T	Exon 4
PTS	V57del	g.4604_4606delGTG; c.169_171delGTG	Exon 3
PTS	V56M	g.4601G>A; c.166G>A	Exon 3
PTS	IVS2-7T>A	c.164-7T>A; r.164_186del exon 3	Intron 2
PTS	N52S	g.3059A>G; c.155A>G	Exon 2
PTS	IVS1-3C>G	c.84-3C>G	Intron 1
PTS	R25Q	g.961G>A; c.74G>A	Exon 1
PTS	R16C	g.933C>T; c.46C>T	Exon 1
PTS	I114V	g.7848A>G; c.340A>G	Exon 6

PTS	K120fs	g.7869_7882del14, c.361_374del14	Exon 6
PTS	K129E	g.7893G>A; c.385A>G	Exon 6
PTS	D136V	g.7915A>T; c.407A>T	Exon 6
PTS	T106M	g.7525C>T; c.317C>T	Exon 6
PTS	IVS3+1G>A	c.186+1G>A	Intron 3
PTS	R25G	g.960C>G; c.73C>G	Exon 1
PTS	N138H	g.7930A>C	Exon 6
PTS	F40fs	g.3020_3023delTGTT; c.116_119delTGTT	Exon 2
PTS	V70D	g.5039T>A; c.209T>A	Exon 4
PTS	N36K	g.3012C>G; c.108C>G	Exon 2
PTS	E35G	g.3008A>G; c.104A>G	Exon 2
PTS	F100V	g.7608T>G; c.298T>G	Exon 5
PTS	A101V	g.7612C>T	Exon 5
PTS	L76F	g.5056C>T; c.226C>T	Exon 4
PTS	G125R	g.7581G>A	Exon 6
PTS	L26F	g.965G>T; c.78G>T	Exon 1
PTS	V124L	g.7878G>T; c.370G>T	Exon 6
PTS	D136G	g.7915A>G; c.407A>G	Exon 6
PTS	R16ins	g.932_933insGGC; c.45_46insGGC	Exon 1
PTS	N47D	g.3043A>G; c.139A>G	Exon 2
PTS	D116G	g.7855A>G; c.347A>G	Exon 6
PTS	E81E	g.5073G>A; c.243G>A and del187_243 exon 4	Exon 4
PTS	Y99C	g.7606A>G; c.296A>G	Exon 5
PTS	N72L	g.5046T>A; c.216T>A	Exon 4
PTS	Y113C	g.7846A>G; c.338A>G	Exon 6
PTS	Y99X	g.7607C>A; c.297C>A	Exon 5
PTS	G144R	g.7938G>C; c.430G>C	Exon 6
PTS	F40L	g.3024T>G; c.120T>G	Exon 2
PTS	A22G	g.952C>G	Exon 1
PTS	K131fs	g.7901delA; c.393delA	Exon 6
PTS	F40fs	g.3022_3025delTTTG; c.118_121delTTTG	Exon 2
PTS	IVS1-323A>T	c.84-323A>T	Intron 1
PTS	A12G	c35C>G	Exon 1
PTS	W115X	c.345G>A	Exon 6
PTS	M80V	c.238A>G	Exon 4
PTS	V132A	c.395T>C	Exon 6
PTS	K131fs	c.391_395delAAAGT	Exon 6
PTS	Q86X	c.256C>T	Exon 5
PTS	P123S	c.367C>T	Exon 6
PTS	IVS3+6T>C	c.186+6T>C	Intron 3
QDPR	H158Y	c.472C>T	Exon 5
QDPR	IVS5+1G>A	c.626+1G>A	Intron 5
QDPR	IVS4+1G>C	c.436+1G>C	Intron 4
QDPR	R221X	c.661C>T	Exon 7
QDPR	F212C	c.635T>G	Exon 7
QDPR	L205X	c.614T>G	Exon 6
QDPR	G23D	c.68G>A	Exon 1
QDPR	W36R	c.106T>C	Exon 2
QDPR	G151S	c.451G>A	Exon 5
QDPR	P145L	c.434C>T	Exon 4

QDPR	L132L	c.396G>A	Exon 4
QDPR	T123ins	c.366_367insACT	Exon 4
QDPR	W108G	c.322T>G	Exon 4
QDPR	C85C	c.255C>T	Exon 3
QDPR	P182fs	c.546delG	Exon 6
QDPR	Y150C	c.449A>G	Exon 5
QDPR	G170S	c.508G>A	Exon 5
QDPR	IVS4+2.6kbA>G	c.436_437ins152bp	Intron 4
QDPR	L74P	c.221T>C	Exon 3
QDPR	G218ins	c.653_654insGGATCACAG	Exon 7
QDPR	S39R	c.115A>C	Exon 2
QDPR	G149R	c.445G>A	Exon 5
QDPR	G17R	c.49G>C	Exon 1
QDPR	G18D	c.53G>A	Exon 1
QDPR	A32A	c.96C>T	Exon 1
QDPR	W35fs	c.105delGgta/insAC	Exon 1
QDPR	S97fs	c.291delC	Exon 3
QDPR	W90X	c.270G>A	Exon 3
QDPR	Q66R	c.197A>G	Exon 2
QDPR	L14P	c.41T>C	Exon 1
QDPR	G17V	c.50G>T	Exon 1
QDPR	IVS4-1G>A	c.437-1G>A	Intron 4
QDPR	S115S	c.345G>A	Exon 4
QDPR	T51S	c.152CG>GC	exon 2
QDPR	V27M	c.79G>A	Exon 1
SPR	Q119X	g.1303_1304TC>CT; c.354_355TC>CT	Exon 2
SPR	P163L	g.1437C>T; c.488C>T	Exon 2
SPR	R150fs	g.1397_1401delAGAAC	Exon 2
SPR	R150G	g.1397A>G; c.448A>G	Exon 2
SPR	K251X	c.751A>T	Exon 3
SPR	IVS2-2A>G	IVS2-2A>G	Intron 2
SPR	-13G>A	g.359G>A	5'-UTR
SPR	V138D	c.413T>A	Exon 2
SPR	K230X	c.688A>T	Exon 3
SPR	N127K	c.381C>G	Exon 2
SPR	Q119X	g.1303_1304TC>CT; c.354_355TC>CT	Exon 2
SPR	P163L	g.1437C>T; c.488C>T	Exon 2
SPR	R150fs	g.1397_1401delAGAAC	Exon 2
SPR	R150G	g.1397A>G; c.448A>G	Exon 2
SPR	K251X	c.751A>T	Exon 3
SPR	IVS2-2A>G	IVS2-2A>G	Intron 2
SPR	-13G>A	g.359G>A	5'-UTR
SPR	V138D	c.413T>A	Exon 2
SPR	K230X	c.688A>T	Exon 3
SPR	R219X	c.655C>T	Exon 3
TH	D97fs	c.291delC	Exon 3
TH	L99fs	c.296delT	Exon 3
TH	R233H	c.698G>A	Exon 6
TH	L236P	c.707C>T	Exon 6
TH	H246Y	c.736C>T	Exon 6

TH	T276P	c.826A>C	Exon 8
TH	F309S	c.926T>C	Exon 8
TH	T314M	c.941C>T	Exon 9
TH	R328W	c.982C>T	Exon 9
TH	R337H	c.1010G>A	Exon 9
TH	C359F	c.1076G>T	Exon 10
TH	A376V	c.1127C>T	Exon 10
TH	T399M	c.1196C>T	Exon 11
TH	IVS11-24T>A	c.1198-24T>A	Intron 11
TH	Q412K	c.1234C>A	Exon 12
TH	T494M	c.1481C>T	Exon 14
TH	D498G	c.1493A>G	Exon 14
TH	G414R	c.1240G>A; g.7008G>A	Exon 12
TH	L510Q	c.1529T>A; g.7495T>A	Exon 14
TH	c.1-71C>T	c.1-71C>T	3'-region