

Myosin VIIA

Homo sapiens (Q13402)
Gallus gallus (XM417277)
Danio rerio (Q9DGG9)
Drosophila pseudoobscura (Q29P71)
Caenorhabditis elegans (CE13575)
Lottia gigantea (Lotgi:187114)
Nematostella vectensis (Nemvel:30268)
Ciona savignyi (SINCAVP00000010178)

G163 and T165

ISGESGAGKTESTKLLIQFL

K268

NYHVFCMLGMSDDQKKL
NYHVFCMLRGMTMEQKKL
NYHIFCYMLKGMTPDQKQL
NYHVFCYLLAGLSSEESRL
NYHIFCYLLAGLSREESL
NYHIFCYMLAGMKNKEDNKL
NYHIFYRMLAGMTPKEALL
NYHVFCYMLAGMAANKQEL

A457

SFEQLCINFANEHLQOFFVR
SFEQLCINFANENLQOFFVR
SFEQLCINFANENLQOFFVR
SFEQFCINYNENLQOFFVQ
SFEQLCINFANETLQOFFVH
SFEQLCINYNENLQOFFVR
SFEQLCINYNENLQOFFVR
SFEQLCINYNENLQOFFVR
SFEQLCINYNENLQOFFVR
SFEQLCINYNENLQOFFVR

R1240

APYCEERLRRTFVNGTETQP
APYCEERLRRTFANGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP
APYCEERLRRTFVNGTETQP

R1883

Homo sapiens (Q13402)
Gallus gallus (XM417277)
Danio rerio (Q9DGG9)
Drosophila pseudoobscura (Q29P71)
Caenorhabditis elegans (CE13575)
Lottia gigantea (Lotgi:187114)
Nematostella vectensis (Nemvel:122088)
Ciona savignyi (SINCAVP00000010178)

LRNGSRKYPHLLVEVEAIQH

Harmonin

Danio rerio (Q29RC0)
Gallus gallus (XP421006.2)
Homo sapiens (Q7RTU8)
Culex quinquefasciatus (Box051)
Lottia gigantea (Lotgi:159301)
Nematostella vectensis (Nemvel:95751)

R103

LGLSVRGGLEFGSGLFISQI
LGLSVRGGAEFGSGLFISQL
LGLSVRGGLEFGSGLFISHL
FGFSIRGGLEYGTGFVSAI
LGFVVRGGFEHGVGVFVSDV
FGFCMRGGSEHGVLVYSSI

R731

SYEETGGGFCYVDDFDEH
SFRSNKQGFQKYVEDFDEH
VYQTAFRQDFRKYEEGFDPY
PKPVRPPSYFPCQGTGA

Cadherin 23

Homo sapiens (Q9H251)
Gallus gallus (XM421595.2)
Danio rerio (Q6QOE1)
Drosophila melanogaster (NP_477497)
Lottia gigantea (Lotgi:91169)
Ciona savignyi (SINCAVP00000012941)
Nematostella vectensis (Nemvel:30053)

H755

DGGVGHQKRTGIATVNIITLL
DGGVGHQKRTGIAMVNIITLL
DGGVGPQKRTGIATVNIITVL
DQGIPLSSSTTINIRVLDV

R1189

LGPMR---SSVRVIVYVEDI
HGPMR---SSVRVIVYVEDV
LGPMR---STVRVIVYVEDV
QPSEQLSTKLVITVIVEDI

R1379

GVITVQGLVDREKG--DFYT
GVITVKGQVDRKKG--DFYT
GRITVTGLLDREKG--DSYT
GILSTAELDREAGPEYEV

A3317

ITAGNRERFFINATGIVT
ITAGNKESAFYINSTGIIY
ITAGNSDGAFYINETSQVVO
TVLGDQNHFSISEDTGVVVR
ITGDSKRYFTIGKTTGAT
IDD--VDKTFSTNKKSGQIY
IISGNDKPKKINSTSGVIM

R2066

Homo sapiens (Q9H251)
Gallus gallus (XM421595.2)
Danio rerio (Q6QOE1)
Drosophila melanogaster (NP_477497)
Ciona savignyi (SINCAVP00000012941)

ITILDNDNRPTFSEATLIV
VTILDNDNRPVFQPVSIITA
ITILDVNDNPPVFNSSFTV
VEVQDINDNDPVFELQSYHA
VTVKDLNDNPPRFASRRVRT

A2110

LVYRIEAGAQDRFLIHLVTG
LDYRIE GGGQDRFLIDATG
LQYRLDSGAQDRFIVDALSG
LRFNLLGEMHRRFHDSEITG
LFYITVVSGLHDRVVDRETG

I2199

VLESAPGTVIANITALDHD
VPESATPGTVVAEVTALDRD
VNESTPPGVVATLAAEDPD
VPERISKGEFVFGARALDLD

R2270

SVIDNASDLPEISVSVPNK
SVHDNASVTDSSVSVPNK
SVHDNASDVIEMSVSVPNK
VSATS PKKGLV GKIRYAIAG
FIVDDVDQGLNCDVYIEIGG

T2290

Homo sapiens (Q9H251)
Gallus gallus (XM421595.2)
Danio rerio (Q6QOE1)
Drosophila melanogaster (NP_477497)
Ciona savignyi (SINCAVP00000012941)

VNVLDVNDNTPQFKPFQITY
VNVLDVNDNTPRFRPPGVITY
INVLDVNDNAPRFRPPGVAN
LITLNVTDANDNAPVMEQLI
FIVDDVDQGLNCDVYIEIGG

D2639

VYATDKDEGLNGAVRYSFL
VYATDKDEGLNGAVLYSL
VFATDEDEGVNGEVRYSEFL
AHAAADKDSPKNAI IHYAEFL

D3017

LLIHVVNRDITNRIILDVDRVI
LLIHVVNRITNRIILDVERVI
VLIHVVNKQTNRIILDVEKVI
QLLMAVRLAKTSYQQPKYLL

A3317

QKGLGRSLETLLAEATAFE
QKVLGRSLETLLDCG-AYD
QRALARSLEALHAGGLYAE
QHRHTAPFVTRNQGQPPPP

Protocadherin 15

Homo sapiens (Q96QU1)
Gallus gallus (NP_001038119.1)
Danio rerio (NP_001012500.1)
Drosophila melanogaster (NP_733314.1)

R1273

EKKIEDTTEILDVYVQEQIF
EQNKDQLIGILEYVQDQIP
EENKEQLISILEYVQDQIP
PSDLRNYYTELEBLLDKKTG

Usherin

Homo sapiens (O75445)
Gallus gallus (XM419417)
Danio rerio (Q1MTD2)
Lottia gigantea (Lotgil :161377)
Nematostella vectensis (Nemvel:199904)
Ciona savignyi (ENSCSAVAT0000000386)

R303

RLHAQSHCRCPGSHPRVHPL
HLHTQSECRCPGSHPRVHPL
HLHSQSTGLCPASHPRVHPL
EARIQTDCCPPHPRVKEQ
KAYIATNCRCPGYPKISTG
EVDIQPNCLCEGFPVHPL

T352

AHPLSFVNDNDVGTISVVSIV
AHPLYYINDDDIGTTWISSV
AHPLDYINDNDIGTTWISSI
AHPLFANDGDSNVVWIS -
AHPSEYMTDADTTTWQSAV
AHPLAYINDDDSSTTWISSL

C536

CDTTSQPYRCLCSQESFTEG
CDTAMEPYRCLCIKESYTEG
CDTSVSPYRCACLPESHTEG
CDMSTLPYKCNCLPASNTQG
-----RCECDPSFTTQG
CNTTVNPFQCNCLLEYNTDG

C575

YAFNCKPCQCNHSHKSCCHYN
HAYNCKPCQCYSHAVSCHYD
QAFNCRPCQCYGHASSCHYN
KDYNCQLCCYNHATSCYYN
DPYPCVKCECNHATACYYN
SSYDCQQCECHHALSCHYE

Homo sapiens (O75445)

Gallus gallus (XM419417)
Danio rerio (Q1MTD2)
Lottia gigantea (Lotgil :161377)
Nematostella vectensis (Nemvel:199904)
Ciona savignyi (ENSCSAVAT00000003863)

G2752

VQVTWKPPLIQNGDILSYEI
VKVSWKPLIQNGEILNYEI
VQVTWAAPLIPNGEIERYEI

ATLNWTQPTQPNGL-----
ATIRWQSEPTQENGPIIRYEI

V3003

YWIFISVFNQVHSINSAGLH
YQIFPQVFNQVHSINSAGLH
YKLSLTVSNGPHNITSPEVN
YNMSVEACTAGGCTKSSPIL
YEFVFNASNTVGYILSNIIIS
YVFTLVDNNGAHNVTSPPVQ

T3571

EYSYQLKACTVAGCATSSKV
EYSYQLRACTVAGCADSSKV
EYIYQLRACTSAGCTDSTKV
SYTYQIEA INAFGSVRSFPV
FYTFVAVNACTTVGCSRSQDA
EYEVTLRACISVGCSSSIPI

T4809

TNYSIGVEACTCFNCCSKGP
TTYSIGVEACTCFNCCSKGP
TVYVSVGVEVCTCFNCCSRGP
TKYNMSVEACTAGGCTKSSP
TFYTFVAVNACTTVGCSRSQDA
TTYRVAVEMCTLGSMCTRGP

VLGR1

Homo sapiens (Q8WXG9)
Mus musculus (NP_473394.3)
Danio rerio (ENSDARP000000020853)

Q345

HESHLLKQIVDDTIPEIAES
HESHLLKQIIDDLIPEIAES
HETDLRFISIRDDNIPEIAES

V754

LSLDWVNVENQVLSKGYTSR
LSLDRVSVDSVLSKGYTSR
LTLDRTNVDNQILKPGFTSR

S1161

VSVSWQLFQNDALQPGQEF
VSVAWQLFQN-ASLQPGQEF
VTIYWQLFANDTPLEEYQEF

D1944

NITVEILPDEPELQKAFSV
SITVEILSDHEPELQKALTV
SIYLLISQDVPPELQKTFKV

Homo sapiens (Q8WXG9)

Mus musculus (NP_473394.3)
Danio rerio (ENSDARP000000020853)

H3399

FRWNGGSEVLHQKLPVRGVL
FRWNGNFAWHQTLPLVRGVL
FVWNRGSEFLHQSLLELQDII

D3477 and Q3497

GQNSIDIFIWEMGQSSFRYFQS
GNQNAIDIFVWEMGHSSLRYPQS
GLNPAQCEVFLWGSQQTFFVQQTQS

D4707

GFFTADGSEASFDVHLLP
GFFTADGSEASFDVHLLP
GSVITLAGQRLAEIILITLLE

N4885

AVLPVSEKAANSQVGFESTA
AILSVPEEAANSQVGFESAA
AVVSVPEVAANPVVGFASLA

Homo sapiens (Q8WXG9)

Mus musculus (NP_473394.3)
Danio rerio (ENSDARP000000020853)

Q5459

ELKPEKVPQVEVYFFVELYE
ELNPKKVHQVEMPPFFVELYD
EVQPDKNPEYEVWFLVEVYK

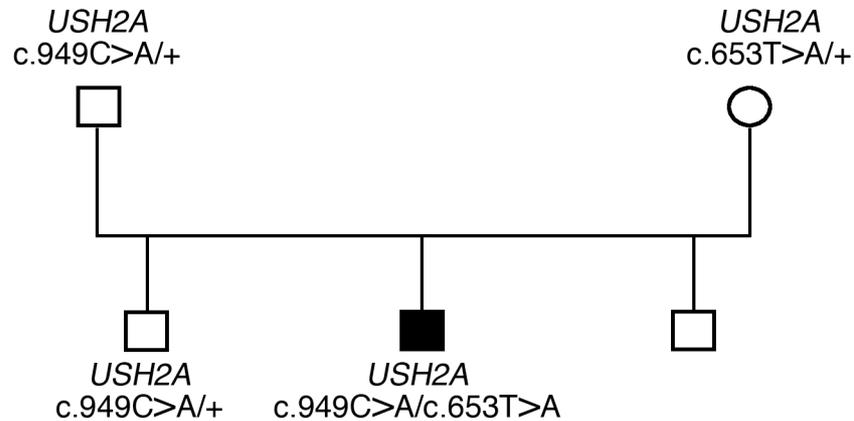
Whirlin

Homo sapiens (Q9P202)
Gallus gallus (ENSGALP00000010189)
Danio rerio (XP_685827.3)
Caenorhabditis elegans (CE43560)
Lottia gigantea (Lotgil:116246)
Nematostella vectensis (Nemvel:95751)
Ciona savignyi (SINCSAVP00000007641)

L327

GSEAEAGSLK-VGDQILEVN
GSEPTAGGMATVGDQILEVN
GSAAEYSGLK-VGDQILEVN
GGAABEDGRLQEGEIVEID
FSVAENAGIK-VGDQILDVN
HSAASRAGLK-CGDQIMDVN
GSAABNAGLK-AGDEILEVN

				Splice site prediction Score for splice sites (0.4 to 1)
Usherin				
	V218			
<i>Homo sapiens</i> (O75445)	HLSVQVHQTKISFFINGVEK	Intron 3 ▼ Exon 4	normal allele	0.92
<i>Gallus gallus</i> (XM419417)	HLSVQVHHSRISFFLINGWED	tcttacaggTgcatcag	mutated allele	0.76
<i>Danio rerio</i> (Q1MTD2)	HLALQIHETSVSFLFNGNEE			
<i>Lottia gigantea</i> (Lotgi1:161377)	HLAIQVYRTTISFFLINGPRV			
<i>Ciona savignyi</i> (ENSCSAVAT00000003863)	HLAIQVHKTDISVFINGVES			
R317				
<i>Homo sapiens</i> (O75445)	HPRVHPLAQRYCIPNDAGDT	Exon 6	normal allele	0
<i>Gallus gallus</i> (XM419417)	HPRVHPLIQRYCIPNGADDT	ggcacagCg gtactgcatt	mutated allele	0.85
<i>Danio rerio</i> (Q1MTD2)	HPRVHPLVEFYCIPNAANDT	ggcacag Aggtactgcatt		
<i>Ciona savignyi</i> (ENSCSAVAT00000003863)	FPRVHPLLEPRYCIPNGV PDT			
Myosin VIIA				
	L2186			
<i>Homo sapiens</i> (Q13402)	KLLCETSLGYKMDDLTSYI	Exon 48 ▼ Intron 48	normal allele	0.98
<i>Gallus gallus</i> (XM417277)	KLLCETSLGYKMDDLTSYI	gcgagacgtcacTggtgagggcgcat	mutated allele	0.82
<i>Danio rerio</i> (Q9DGG9)	KLLCETSLGYKMDDLTSYI			
<i>Drosophila pseudoobscura</i> (Q29P71)	KLLCETSLGYKMDDLTSYI	gcgagacgtcacCggtgagggcgcat		
<i>Caenorhabditis elegans</i> (CE13575)	KLLLDTVGYKMDDLTSYI			
<i>Lottia gigantea</i> (Lotgi:187114)	KLLCETSLGYKMDDLTSYI			
<i>Nematostella vectensis</i> (Nemvel:122088)	KLLCETSLGYKMDDLTSYI			
<i>Ciona savignyi</i> (SINCAVP00000010178)	KLLCETSLGYKMDDLTSYI			
Sans				
	L16			
<i>Homo sapiens</i> (Q495M9)	DQYHRAARDGYLLELLKEATR	Exon 1	normal allele	0
<i>Gallus gallus</i> (E1C0X4)	DQYHRAARDGYLLELLKEATK	ggctacctggagCtcctcaag	mutated allele	0.75
<i>Drosophila melanogaster</i> (NP_610829)	DRFHKAARDGLLDVLAATR	ggctacctggag Gtcctcaag		
<i>Lottia gigantea</i> (Lotgi:71589)	DRFIAAARDGYLLELLQSATK			
<i>Nematostella vectensis</i> (Nemvel:171827)	DLYFKAARDGNLNILKNATK			



Segregation analysis of the USH2A mutations in family U51: The isocoding variation c.949C>A (p.R317R) has been predicted to be pathogenic by Pennings et al [1], and considered as non-pathogenic by Dreyer et al [2]. Segregation analysis of this variation in family U51 is compatible with a pathogenic mutation.

References

1. Pennings RJ, Te Brinke H, Weston MD, Claassen A, Orten DJ, Weekamp H, Van Aarem A, Huygen PL, Deutman AF, Hoefsloot LH, Cremers FP, Cremers CW, Kimberling WJ, Kremer H: **USH2A mutation analysis in 70 Dutch families with Usher syndrome type II.** *Hum Mutat* 2004, **24**:185.
2. Dreyer B, Brox V, Tranebjaerg L, Rosenberg T, Sadeghi AM, Moller C, Nilssen O: **Spectrum of USH2A mutations in Scandinavian patients with Usher syndrome type II.** *Hum Mutat* 2008, **29**:451.