

Table 1. Primers used for amplification and sequencing

Name	Sequence(5'-3')	Length (bp)	Annealing temperature
g.Ex_1F	GTCGGAGAAGAGCGAGAAGA	299	56°C
g.Ex_1R	ACAGTGACGAGGAGGACACC		
g.Ex_2F	AGAAGGTGGGGAGATGTGTG	364	56°C
g.Ex_2R	GCTCAAGAGCTCCAATGTCC		
g.Ex_3F	TCTTCATGGCAACACAGGT	476	65°C
g.Ex_3R	CCTTCAAGGGTCTCAGTGG		
g.Ex_4F	GTTCTGCTGCCCTCTCATTC	375	65°C
g.Ex_4R	CAGTCCCAATCTCACACCT		
g.Ex_5F	GACAGAGTCCCTGGACGGTA	382	65°C
g.Ex_5R	TGGTTTTAGAGGCAGCTTGG		
g.Ex_6F	TTGTCATCCCAAGAACAGAGG	394	65°C
g.Ex_6R	CATTCAATTCCAGGCTCACA		
g.Ex_7F	CTCCACCACCTGGAGCAC	300	65°C
g.Ex_7R	TGGCCACATAAGAGGCAGAG		
g.Ex_8F	TTTGCCGTTGTGGATAAAGAC	393	65°C
g.Ex_8R	CAAGAGGGCATCTCCATTA		
g.Ex_9F	TACTTGGTGAGGAGCCTGGA	345	65°C
g.Ex_9R	AACTGGAAAGACACATTTTCAGTCA		
g.Ex_10F	TTTGGCTTGTTTTCCCTCTG	479	65°C
g.Ex_10R	CTGCCCTAAGTTGAGAACCA		
g.Ex_11F	GGGATTCTGTAGTGGCATCG	285	65°C
g.Ex_11R	CACCCACCTCAGAGGAAGAG		

g.Ex_12F	GGCAGTTCTCTGCTGTCCTC`	375	56°C
g.Ex_12R	AACGCTTCTTCCAGCACACT		
g.Ex_13F	CCAGTTAAGGTCCTGGTTCG	395	65°C
g.Ex_13R	GAGGATGGAAGGGGAACCTA		
g.Ex_14F	TCAGAAAATGCTACCTGGATGA	280	65°C
g.Ex_14R	GCCCAGCATCTCTGTCTTTC		
g.Ex_15F	AGGTTGGTGTGTGGGTTTTG	396	65°C
g.Ex_15R	ATGTCAAGGCTGCTGTGATG		
g.Ex_16F	GCTGGAATCTTACCCCAGGT	381	65°C
g.Ex_16R	TGGGGAGTGTAAGTCCAGGT		
g.Ex_17F	TTCACATGCTGCAGGAAAAG	400	56°C
g.Ex_17R	AAAGCAGCCAAGAGGATGTG		
g.Ex_18F	GGGACTGTTCTTTGCTGCTC	388	56°C
g.Ex_18R	GGAAAATGGATTAATATCCCAGAA		
g.Ex_19F	GGATTCTGCATGGGAAGAAC	395	65°C
g.Ex_19R	CAGAGGAAGAGAGAAGCCAAG		
g.Ex_20F	GCATTTGCAGATGTCTCAGG	329	65°C
g.Ex_20R	TTGACAGAGGTTCACGTTGG		
g.Ex_21F	TTGACAGCTGATTCCCTGTG	586	60°C
g.Ex_21R	GGTCTGAGGACGTCTGTGCT		
g.Ex_22F	TGAGCATGCAATGATTGTGA	494	60°C
g.Ex_22R	GTGCACATGAGCCCAAGAC		
g.Ex_23F	TTTTCTTGAAAACAAAAGAAGC	350	60°C
g.Ex_23R	ATCCTTTCCCCACTGTAGCC		
g.Ex_24F	TCATGGGCAGCCTCTTAAC	357	60°C

g.Ex_24R	CACATGGACTGAGCATCACA		
g.Ex_25F	AGCCATCATTCCAAGAAGCA	367	60°C
g.Ex_25R	TGGTCACCATCAGCAGAAGA		
g.Ex_26F	TGGGAGGAGAGAGGAGACAG	369	60°C
g.Ex_26R	GACTTAGCAGGAGCACAATCC		
g.Ex_27F	GTGTTGGGTATTCACGCACA	283	65°C
g.Ex_27R	TGTTAGCAGACTGGGGCTCA		
g.Ex_28F	GGACAAAAGAAGGGGAGGAG	475	60°C
g.Ex_28R	CTCTGCCTTTTTGGAGGAGA		
g.Ex_29F	TGTTCTTTCCAGGCCTTTGT	381	65°C
g.Ex_29R	AACAGTAGGGCAGGGGTTCT		
g.Ex_30F	TTTTTCGTTTCCAAACCTTG	295	60°C
g.Ex_30R	AACTGTTCCCCGTGCTCTC		
g.Ex_31F	TGCTCCCCTGTTATGTTTCTG	370	65°C
g.Ex_31R	AGTTTTGCCAAAGGCAACC		
g.Ex_32F	GCACTGGTCACCTCCAACCT	286	60°C
g.Ex_32R	TGTAGGAAGAATGTGCAGAG		
g.Ex_33F	TGGAAGTTTGGAGAACCCAGT	392	65°C
g.Ex_33R	GGTTGTGCTGGGGATGGA		
g.Ex_34F	TGGGGAAATATTGAAAACACG	344	60°C
g.Ex_34R	TACAGCCAAACCCAGACACA		
g.Ex_35F	AGGGCTGCTCAGGAACACTA	298	60°C
g.Ex_35R	AAGACCCCACTGACCTCTGA		
g.Ex_36F	CCTGCCACCAAATAGATGA	389	60°C
g.Ex_36R	GCCTTCACTGGAAGGTTCTG		

g.Ex_37F	GCCCGGAGCACTAAATGAT	500	60°C
g.Ex_37R	GATGGAGACAAGGACTGTCAA		
g.Ex_38F	CCTGCAAAACACAACAGGTG	364	60°C
g.Ex_38R	AGGGTGGGAGTGCAACACTA		
g.Ex_39F	TGTGTTGTGGGCCTTGTCTA	385	60°C
g.Ex_39R	AAGTTACTCATGGGTGATACGG		
g.Ex_40_codingF	AGGGCTTCTCTCCCCACTAA	465	60°C
g.Ex_40_codingR	CCTCCACAGGCTGTCAATCT		

Table 2. Primers used for cDNA amplification.

Name	Sequence(5' -3')	Original length (bp)	Annealing temperature
c.Ex31F	GGCTGACCAACGAGAATCTG	464	60°C
c.Ex35R	GCGGCAGGTGTTGGATAAC		
c.Ex31F	GGCTGACCAACGAGAATCTG	575	60°C
c.Ex36R	CTGACGGTATTCGGTGAGGT		
g.Ex34F	TGGGGAAATATTGAAAACACG	307	60°C
c.Ex35R	GCGGCAGGTGTTGGATAAC		

Supplementary figure legends (Szperl et al.)

Supplementary Figure 1. The c.946G>A variant of exon 8 in patient 1 and the evolutionary conservation of the p.Gly316 residue.

Supplementary Figure 2. The c.2330_del G variant of exon 19 in patients 2 and 3 and the evolutionary conservation of the p.Gly777 residue.

Supplementary Figure 3. The c.2330_del G variant of exon 19 in patients 2 and 3 disturbs the reading frame and leads to a premature stop codon.

Supplementary Figure 4. The c.2246C>T variant of exon 19 in patient 6 and the evolutionary conservation of the p.Arg749 residue.

Supplementary Figure 5. The c.1367A>G variant of exon 11 in patient 7 and the evolutionary conservation of the p.Asn456 residue.

Supplementary Figure 6. The c.5062A>G variant of exon 36 in patient 7 and the evolutionary conservation of the p.Met1688 residue.

Supplementary Figure 7. Identification of cryptic splice acceptor sites in the intronic sequence upstream of the c.4460-1G>C mutation and exon 34. The upper panel shows the manually annotated consensus splice acceptor (SA) sites, while the lower panel indicates those selected by the software tool from www.fruitfly.org. Upper panel: SA

dinucleotide AG (yellow), consensus SA (pink), sequence primer g.E_34F (green).

Lower panel: Software selected consensus SA (yellow) with high ranking scores in bold.

Supplementary Figure 8. Sequence of the cDNA of patient 9 with the splice site mutation c.4460-1G>C, downstream of the sequence primer g.Ex_34F in intron 34 and including exon 34. Since the start position of the inclusion of intron 33 was not known, all possible reading frames were examined for stop codons. In total, nine stop codons were present (indicated in red), with premature termination affecting exon 34 in all three reading frames.

Supplementary table 1. Primers used for the amplification and sequencing of genomic DNA.

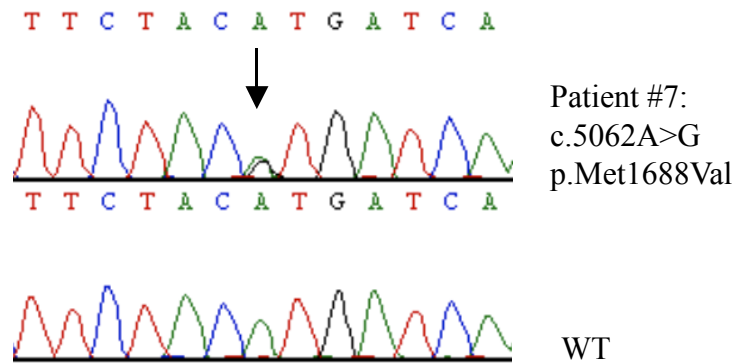
Supplementary table 2. Primers used for the amplification of cDNA.

Type of file: figure

Label: 10

Filename: Szperl_SupFig6-revised.pdf

Supplementary figure 6



Orthologues

p.Met1688

<i>Homo sapiens MYO5B</i>	L-QVFKQLFYMINAVTLNLL
<i>Pan troglodytes</i>	L-QVFKQLFYMINAVTLNLL
<i>Pongo pygmaeus</i>	L-QVFKQLFYMINAVTLNLL
<i>Macaca mulatta</i>	L-QVFKQLFYMINAVTLNLL
<i>Otolemur garnettii</i>	Q-QVFKQLFYMINAVTLNLL
<i>Tupaia belangeri</i>	L-QVFKQLFYMINAVTLNLL
<i>Equus caballus</i>	L-QVFKQLFYMINAVTLNLL
<i>Spermophilus tridecemlineatus</i>	L-QVFKQLFYMINAVTLNLL
<i>Loxodonta Africana</i>	Q-QVFKQLFYMINAVTLNLL
<i>Cavia porcellus</i>	L-QVFKQLFYMINAVTLNLL
<i>Bos taurus</i>	L-QVFRQLFYMINAVTLNLL
<i>Ochotona princeps</i>	IQQVFKQLFYVINAVTLNLL
<i>Myotis lucifugus</i>	L-QVFKQLFYMINAVTLNLL
<i>Canis familiaris</i>	L-QVFKQLFYMINAVTLNLL
<i>Erinaceus europaeus</i>	L-QVFKQLFYMINAVTLNLL
<i>Rattus norvegicus</i>	L-QVFKQLFYMINAVTLNLL
<i>Mus musculus</i>	L-QVFKQLFYMINAVTLNLL
<i>Echinops telfairi</i>	IQQVFKQLFYMVNAVTLNLL
<i>Monodelphis domestica</i>	IQQVFKQLFYMINAVTLNLL
<i>Xenopus tropicalis</i>	IQQVFKQLFYMINAVTLNLL
<i>Danio rerio</i>	SQQVLRQLFYMINAVTLNLL
<i>Tetraodon nigroviridis</i>	VGQVVKQLFHCINAVTLNLL
<i>Takifugu rubripes</i>	VGQVVKQLFHCINAVTLNLL
<i>Oryzias latipes</i>	MEQAFQQLTYLICASAINSL
<i>Ciona savignyi</i>	IRQVFRQVFYCITAHTLNNQL
<i>Ciona intestinalis</i>	IKQIFRQIFK-ITSHTVNNQL

Paralogues

<i>Homo sapiens MYO5A</i>	K-QVVKQMFYIIGAITLNNLL
<i>Homo sapiens MYO5C</i>	R-QAVKQLFFLIGAVTLNSLF

Type of file: figure

Label: 11

Filename: Szperl_supFig7.pdf

Intron 33 cryptic splice sites:

GTGGGTGTGGTCTCTGGAATCACATG 45626903
45626902 GGCTGTGTTCTAGACCTGGCTTCTTTTCTAACTAGCCGCACATGGCCGTGGCTGTGCTACT 45626843
45626842 CACCAGTGTTCCTTGTGTAGAGCACAGGAAAGGGGATCATGCCGCTGGCATGGATTGTGA 45626783
45626782 GAATGAAAGTTCGTAATTCGTTGCTCATAACATGGTCCCTGGACCAGCAGCATCAGCATCA 45626723
45626722 GCATCAGTATCACCCCTGGGAAGGGGTAGAGATGCAGAGTCTCGGGCTCCGTGCCAGGC 45626663
45626662 CTCTCAATTGCAAGGCACATAACAAGATGCCAGGTGACATTACAGGCTCATTAAGTG 45626603
45626602 AGGAATGCTGAGCTTCTGCGACTGTGTCTGGGATCACCCCTGGTCCCCTGTCAAAGTTTAC 45626543
45626542 CAGGGTTGGCTTTTACTGTCCTGCGATCATCTGCGAGTTTCTTATCCAGGCATGTGGCTG 45626483
45626482 AATCAGGGCTGGCCTGCCAGCCTCCCACCCCATGTGCTCCCTGGGGCCACCAGGCAGCT 45626423
45626422 GTGAGAACCACTGCTCAGAAATTCCTTCCCAGCTGGCTCCTAGGACTGAAAGACCTTCC 45626363
45626362 ACACCTAGCTGCCAAGAGGGCAGGGTTTTCTGTGAAAAGAGGAGCCCCCGCCACCCCTC 45626303
45626302 CGAGTCCCTCTTGGCTGCTCAGCAAGGGCCAGAGGGAGTCTGTGTGACCTGTGCAGCTT 45626243
45626242 GCTCAGGTTTGTGCCAGTCTGCTTTCCAGTCTGTAACCAGCTCCCCTCCCCTCCAGTCTT 45626183
45626182 GTGCCCATCTGCCACAGGCCACTGCAACAGGCCACCTCCCTGCTCTGTACCAACCCAGT 45626123
45626122 CCAGCATCCCCTTGCAGAGCCAGGAAGAGTCTTAGAGGAGAGAGTCCCTGTGGAAAGAGAG 45626063
45626062 AGAAATCTTTGGTGGTCTGATCCTCTGGACCCTGAGCCAGCAGCTCCCTTCTTATGGCA 45626003
45626002 CTTAGTGAATGGGATGATGACACTGACCTGCCATGATGTGAAGTGTGCAA 45625943
45625942 GCGGAAATGCTGTGTTAAAACCTGCAAAACCCCTTCTCAAATGTGAGGCTTTCAAAGCAGTG 45625883
45625882 GTAGCTTTTGCAGGAATATTTGAAGTTCAAATAAGAGGCAAGGCAAGCATTATTTAGAG 45625823
45625822 AAACCTATGTAAGACTGAGTCCGACTCTGCTGAGCACTGTGCCTAGGGCCAGGGACCC 45625763
45625762 AGGAAGAACAGATTGTGACATCTGCCTTAAACAGATTGCTCCAGCTGTTGGTGTGTGT 45625703
45625702 GTTTTTAGTAGCCTGCTTCTTAGATGATTTTGTCTTTTAAAAGCTTAATCTCCCTA 45625643
45625642 CAGTTGTCCAGCAATTAATTAATGTGGTGAAGTTTATCTTAAACTTCTTAAACAGAC 45625583
45625582 TGCATCTGTTCAAATGCAATTAGGAACAAACCTTTCATCTCTGGTGAATGAACTTTCA 45625523
45625522 ATAAATAAGCTGCTTGTGTCAGACAGGTGGCTCCCTCCTGCCATTTGCCAGGGTTTTTA 45625463
45625462 CAGAGAAAACCCCTGCCATCAGCATGATGACCTTGCTCAGGTGGCCCCAGGAAAGGTC 45625403
45625402 ATCTCCTGCTGAGCTACCTAGGAAAACTCTGCAGTCACTTCCCTCCAAAAGACCCCATC 45625343
45625342 CAAACAAATGCCAAAACTTGACTGCCACCCTACCTCTTAGTACCTGGCACTGGCCA 45625283
45625282 GTTGTGTGGAGGAGAAATAAACTAGTGTCTATGCTCTTCCCAAGATGCTAACATTT 45625223
45625222 AGCTGGGGCACCACAAACAAGCTTTATTTAATCACAATAGAACATTCCAAGACAGGACAAG 45625163
45625162 TTGTTTGAATCTGCACATGGATAGTTTGAAGTAAAGATTATGGATTGGAAGTCTGG 45625103
45625102 GCCTACAAAATACATTTCTGGATAAATGACTGGCATTCTAACTAGAGCCAAATGGCCAA 45625043
45625042 TCCACAGCCATCTGCTGGACCTATAAAATGGCCAAATGTCAACCATGTACTCAAGAGTAT 45624983
45624982 TATGGCCTACAGTTATCATTAACTAGAAGGATGCGTGGAGCAGGGACAATGTAACCCA 45624923
45624922 AGTACCAACTGGAGCTGCTGGCAGCAGAGCCCAGCCCAGGACCAGCCTGGTGAAGGTTT 45624863
45624862 GCAAGGATTACTGCCA CCTGCAGGTGGGAGGGAGTGGCCTCCCTTCATTGCCATTCTGA 45624803
45624802 GCCTACAGGTGGCTCATGAGCCCTCGTTTGGGAGGGTATGAGTGTAAAGAACAAGGGCTG 45624743
45624742 GAGATACAGTGCAGAGCTATGGTGAAGCCCTGGGGCCTTGTGCTCCTGCCCATTTCTCA 45624683
45624682 CCTGTCATCTTGCCCTTGCACCAGCCTTTCCCCTAGTTCTCTGGCCTTCTACCACCAC 45624623
45624622 CTGTGCCTTAAGTCTGGCTCAGCTGGCATGGTCTGGAATAGAAAATGGTCTGACAGCTG 45624563
45624562 CTTCTGTCCCCTCCAGGGCCTTTTGCAGAAAAGGGCCAAAGGGCTGAGTCTGCAGTTAAT 45624503
45624502 GGGGAGACTCACAAGGCCCTTCCAGTCCAGCCAGAGCAGCCCTTGGCCAGGCAGGGACAG 45624443
45624442 GAGTGTGGGATACAGAGGAGAGCAGTTGGGGGAAGTAAGAGAGCCGCTGGAGAGATGCAT 45624383
45624382 GCCAAGCAGCAGCACCAGGGTGGGGAAGGGGGCCCTGCTGATCTATTGGCTGTGAAGC 45624323
45624322 TGGCAAGCACGGCTGGTAATTTGTTCCTGAACCTGATGGCCAGAGATTGTTCCAACT 45624263
45624262 CCCTAGCCCTCAGCAGGCTGCCCTGTCTTTGGGCACATACAATTTGTCTAATTCTGGAG 45624203
45624202 CCAGCCCTTGCCTTCTGGAAGTGGTGGTGGCCACGAATCAGAGTTTCTGCCCCATGTG 45624143
45624142 GGAGCTAGCCCTGTCCTCAGCCTTCCACTTGCATGGGCCCCCTGTTGGCTCGGGACTGA 45624083
45624082 GCATCTGTGTAACCTTGCAGCCTTTCTTCCATATTCTTAGGCTGAGCATTAGAAGC 45624023
45624022 CTGTTTCAGGGTTATAGTGCAGACAATAGGAAATGGTTTTGTTTGTGTTTAA 45623963
45623962 CCACAGTGTCTGAAACTACTGGTAGGCATGCTCGGGAGTTTGGGTA CCGTGGTGGCTTC 45623903
45623902 TCTCCCAA TCTCTTTGAGCATCTATTCCTAGATGAGCTA TGGGAAATA TGAAGA 45623843
45623842 CTCTATTAGATGAGGATTCTGACAGCCTCCATCTGCCCATAGTCTAACTTGCAT 45623783
45623782 TTGCTGCTCTCGTTGCCCTAG

http://www.fruitfly.org/seq_tools/splice.html

GTGGGTGTTGCTCTCGAATCACATGGGCTGTGTTTCAGACCTGGCTTCTTTTCTAAGTCCGACATGGCCGTGGCTGT
GCTA**CTCACCAGTGTCTTGTGTAG**AGCACAGGAAAGGGGATCATGCCGTGGCATGAGTTGTGAGAATGAAAGTTCGTA
TTTTCGTTGCTCATACATGGTCCCTGGACCAGCAGCATCAGCATCAGTATCACCCTGGGAAGGGTGAGAGATGC
A**GAGTCTCGGGCTCCGTGCCAG**GCCTCCTCAATTGCAAGGCACATAACAAGATGCCAGGTGACATTACAGGCTCATTAA
AGTGAGGAATGCTGAGCTTCTGCAGTCTGTCTGGGATCACCTGGTCCACTGTCAAAGTTCATCAGGGTTGGCTTTTAC
TGTCCCTGCGAT**CATCTGCAGTTTCTTATCCAG**GCATGTGGCTGAATCAGGGCTGGCCTGCCAGCCTCCCACCCCATGT
GCTCCCTGGGGCCACCAGGCAGCTGTGAGAACCAC**TGCTCAGAATTCCTTCCCCAG**CTGGGCTCCTAGGACTGAAAGACC
TTCCACACTAGCTGCCAAGAGGGCAGGGTTTTCTCTGTGAAAAGAGGAGCCCCCGCCACCCCTCCGAGTCCCTCTTGCT
GCTCAGCAAGGGCCAGGAGGGAGTCTGTGTGACCTGTGCAGCTTGTCTCAGGTTTGTGCCAGTGTGCTTTGCCAGT**CGT**
ACCAGCTCCCTCCCTCCAGTCTTGTGCCCATCTGCCACAGGCCACTGCAACAGGCCACCTCCCTGCTCTGTACCAACC
CAGTCCAGCATCCCCCTGCAGAGCCAGGAAGAGTCTTAGAGGAGAGAGTCTGTGGGAAAGAGGAGAAATCTTTGGTGGT
CTGATCTCTGGACCCTGAGCCAGCAGCT**CCCTTCTTTATGGCACCTTAG**TGAAATGGGATGATGACACTGACCTGCCTG
CCCCATGATGTGAAGTGTGTCAAAGGGCGAAATGCTGTGTTAAAACCTGCAAAAACCTTCTCAAATGTGAGGCTTTCAAAG
AGTGGTAGCTTTTGCAGGAATATTTGAAGTTCAAATAAGAGGCAAGGCAAGCATTATTTTAGAGAACTTATGTAAGACT
GAAGTCCGACTCTGCTGAGCACTGTGCCAGGGCCAGGGACCCAGGAAGAACAGATTGTGACATCTGCCTTAAACAGAT
TTGCTCCAGCTGTT**TGGTTTGTGTGTTTTTAGTAGCCTGCTTCTTAG**ATGTATTTTGTCTTTTAA**AAAGCTTAATCTCC**
CCTACAGTTGTCCAGCAATTAATTAATGTGGTGAAGTTTATCTTAAACTTCCTTAAACAGACTGCATCTTGTTCAAAT
GCAATTAGGAACAAACCTTTCATCTCTGGTGAATGAACTTTCAATAAATAAGGCTGCTTGTGTGACAGAGTGG**CTCCC**
TCCTGCCATTTGCCAGGGTTTTTACAGAGAAAACCTGCCATCAGCATGAGATGACCCTTGTCTCAGGTGGCCCCAGGAAA
GGTCATCTCCTGCTGAGCTACCTAGGAAAATCCTGCAGTCATCTCCCTCCAAAAGACCCCATCCAAAACAAATGCCAAA
CTTGAC**TGCCGACCACCTACCTCTTAG**GTACCTGGCACTGGCAGTTGCTGTGGAGGAGAAATAAACTAG**TGTATCGT**
CTCTTCCCCAAGATGCTAACATTTAGCTGGGGCACCCAAACAAGCTTTATTTAATCACAATAGAACATTCCAAGACAGGA
CAAGTTGTTTGCATCTGCACATGGATCAGGTTTGAAGTAAGAAGTTATGGATTGGAAGTCTGGGCTACAAAATACATT
TTCTGGATAAATGACTGGCATTCTAACTAGAGCCAATGGCCAATCCACAGCCATCTGCTGGACCTATAAAATGGCCAAA
TGTCAACCATGTACTCAAGAGTATTATGGCCTACAGTTATCATTTAACTAGAAGGATGCGTTGGAGCAGGGACAATGTAA
CCCAAGTACCAACTGGAGCTGCTGGCAGCAGAGCCAGCCAGGACCCTGGTAGAAGGTTTGC**AAGGATACTGCCCA**
CCTGCAGGTGGGAGGGAGGTGGCCTCCCTTCATTGCCATTCTGAGCCTACAGGTGGCTCATGAGGCCTCGTTTTGGGAGGG
TATGAGTGTAAAGAACAAGGGCTGGAGATACAGTGCAGAGCTATGGTGAAGCCCTGGGGCTTGTGCTCCTGCCCAT
CTCACCT**GTCATCTTGCCCTTTGCACCAGGCCTTTCCCCCTAG**TTCTCTGGCCTTCTACCACCACCTGTTGCCTTAAGTCT
GGCTCAGCTGGCATGGTCTGGAATAGAAAATGGTCTGAC**AGCTGCTTCCCTGTCCCTCCAG**GGCCTTTTGAAGAAAGGGC
CAAAGGGCTGAGTCTGCAGTTAATGGGGAGACTCACAAGGCCCTTCCAGTCCAGCCAGAGCAGCCCTTGCCAGGCAGGG
ACAGGAGTGTGGGATACAGAGGAGAGCAGTTGGGGGAAGTAAGAGAGCCGCTGGAGAGATGCATGCCAAGCAGCAGCACC
GGGGTGGGGAAGGGGGCCTGCCTGATCTATTTGGCTGTGAAGCTGGGCAAGCAGCGCTGGGTAATTTGTTCTGAACCTG
ATGGGCCAGAGATTGTTCC**AACCTCCCTAGCCCTCAGCAG**GCTGGCCCTGTCTTTGGGCACATACAATTTGTCTAATTCT
GGAGCCAGCCCTTGCTTCTGGAAGTGGTGGTGGCCACGAATCAGGAGTTCCTGCCCCCATGTGGGAGCTAGGCCCTGTC
CTCAGCCTTCCACTTGCCATGGGCCCTGTTGGCTGCGGACTGAGGCATCTGTGTAACCTGCAGGC**ACTTTTCCCTCCATA**
TTGTTAGGCTGAGGCATTAGAAGCCTGTTTTCAGGGTTATAGTGCAGACAATTAGGAATGTGGTTTTGTTTGTGTTT
TTAACCACAGTCTGAAACTACTGGTAGGCATGCTCGGGAGTTTGGGTACCCTGGGTGGCTTCTCTCCAATCCT**CTTT**
GAGACATCTATTCTAGATGAGCTA**TGGGGAAATATTGAAAACAG**TCTATTGAGATGAGGATTTGAC**AGCCTCCATTG**
TCCCCATAGATCTAACTTGCCAT**TGCTGCTCTCGTTGCCCTTAG**acttgaagccccagatgctgtcgggacagtgccc
tgt

- Yellow, selected by software tool
- Bold yellow, best scores (> 0.88)
- Green, sequence primer Ex34F
- Small print, exon 34

Type of file: figure

Label: 12

Filename: Szperl_SupFig8.pdf

Intron 33/exon 34 splice site mutation in P9

Original reading frame (1 – 0)

tcccga~~aat~~attg~~aaa~~acacg~~t~~ctattcagatgaggattctgacagcctccattgtccc
catagatc~~t~~aa~~ct~~tggcatttggctgctctcg~~t~~ggccctta~~c~~ACTTGAAGCCCCAGATGCT
GTCGGGCACAGTGCCTGTCTCCCGCCTACATCCTCTACATGTGCATCCGGCACGCGGA
CTACACCAACGACGATCTCAAGGTGCACCTCCCTGCTGACCTCCACCATCAACGGCATTAA
GAAAGTCTGAAA

Alternative reading frame 1 (0 - 2)

tcccga~~aat~~at~~g~~aa~~aa~~cacg~~t~~ctattcagatgaggattctgacagcctccattgtccc
catagatc~~t~~aa~~ct~~tggcatttggctgctctcg~~t~~ggccctta~~c~~ACTTGAAGCCCCAGATGCT
GTCGGGCACAGTGCCTGTCTCCCGCCTACATCCTCTACATGTGCATCCGGCACGCGGA
CTACACCAACGACGATCTCAAGGTGCACCTCCCTGCTGACCTCCACCATCAACGGCATTAA
GAAAGTCTTGA~~AA~~

Alternative reading frame 2 (2 - 1)

tcccga~~aat~~attg~~aaa~~acacg~~t~~ctattcagatgaggattctgacagcctccattgtccc
catagatc~~t~~aa~~ct~~tggcatttggctgctctcg~~t~~ggccctta~~c~~ACTTGAAGCCCCAGATGCT
GTCGGGCACAGTGCCTGTCTCCCGCCTACATCCTCTACATGTGCATCCGGCACGCGGA
CTACACCAACGACGATCTCAAGGTGCACCTCCCTGCTGACCTCCACCATCAACGGCAT~~TAA~~
GAAAGTCTGAAA

TAA; **TAG**; **TGA**; stopcodons

C; splice acceptor site mutation IVS33+3753G>C

tcccga~~aat~~attg~~aaa~~acacg; primer g.Ex_34F

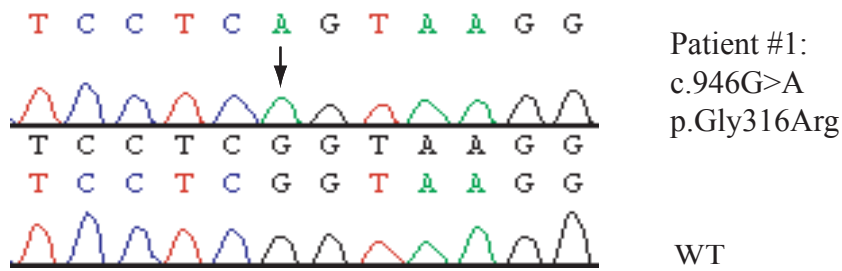
Capital letters indicate exon 34

Type of file: figure

Label: 5

Filename: Szperl_SupFig1-revised.pdf

Supplementary figure 1



Orthologues

p.Gly316

<i>Homo sapiens MYO5B</i>	DAEDFEKTRQAF TLL GVKESHQMSIFKIIASILHL
<i>Pan troglodytes</i>	DAEDFEKTRQAF TLL GVKESHQMSIFKIIASILHL
<i>Pongo pygmaeus</i>	DAEDFEKTRQAF TLL GVKESHQMSIFKIIASILHL
<i>Macaca mulatta</i>	DAEDFEKTRQAF TLL GVRESHQISIFKIIASILHL
<i>Otolemur garnettii</i>	DAEDFEKTRQAF ALL GVRESHQISIFKIIASILHL
<i>Tupaia belangeri</i>	DAKDFEKTRQAF TLL GVRESHQISIFKIIASILHL
<i>Equus caballus</i>	DAEDFEKTRQAF TLL GVRESHQISIFKIIASILHL
<i>Spermophilus tridecemlineatus</i>	DAEDFEKTRQAF TLL GVRESHQISIFKIIASILHL
<i>Loxodonta Africana</i>	XXXXFEKTRQAF TLL GVRESHQISIFKIIASILHL
<i>Cavia porcellus</i>	DAEDFEKTRQAF TLL GVRESHQINIFKIIASILHL
<i>Bos taurus</i>	-----
<i>Ochotona princeps</i>	XXXXXXXXXXXXXXXXXXXXVRESHQMSIFKIIASILHL
<i>Myotis lucifugus</i>	XX
<i>Canis familiaris</i>	-----
<i>Erinaceus europaeus</i>	DAEDFEKTRQAF TLL GVRESHQISIFKIIASILHL
<i>Rattus norvegicus</i>	DAEDFEKTRQAL TLL GVRESHQISIFKIIASILHL
<i>Mus musculus</i>	DADDFEKTRQAL TLL GVVDSDHQISIFKIIASILHL
<i>Echinops telfairi</i>	DAEDFEKTRQAF TLL GVRESHQMSIF-IIASILHL
<i>Monodelphis domestica</i>	DAEDFEKTRQAF TLL GVRESYQINIFKIIASILHL
<i>Xenopus tropicalis</i>	DAEDFEKTRQAF TLL GVKETHQMGIFKIVASILHL
<i>Danio rerio</i>	DAEDLVKTREAL TML GVKENHQMSIFKIIASILHL
<i>Takifugu rubripes</i>	DAEDFVKTREA FLL GIKESTQNNVFKIIASILHL
<i>Tetraodon nigroviridis</i>	DAEDFVKTREG FVFL GIKIDSTQNNVFKIIASILHL
<i>Oryzias latipes</i>	-----
<i>Ciona savigny</i>	DKQEFQETVHA FLL GVSSKHQSLIFRLLSAVLHM
<i>Ciona intestinalis</i>	DESEFKETIHA FLL GVSSKHQSLVFRLLSAILHM

Paralogues

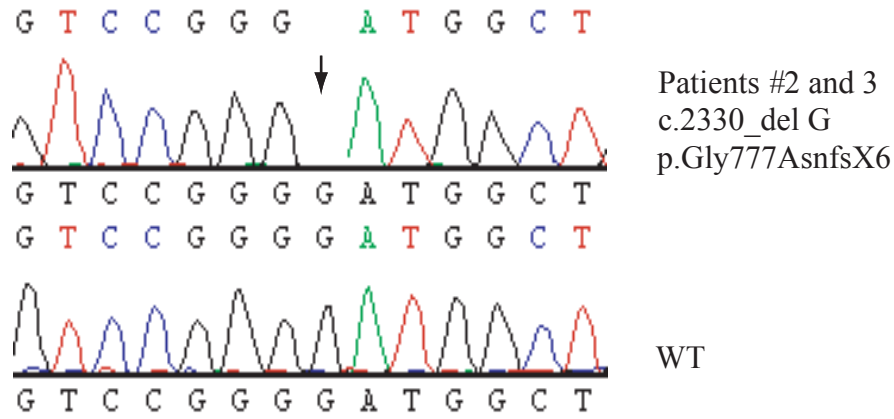
<i>Homo sapiens MYO5A</i>	DAKEMAHTRQACT L LGISESHQMGIFRILAGILHL
<i>Homo sapiens MYO5C</i>	DRAEMVETQKT FLL GFKEDFQMDVFKILAAIHL

Type of file: figure

Label: 6

Filename: Szperl_SupFig2-revised.pdf

Supplementary figure 2



Orthologues

p.Gly777

<i>Homo sapiens MY05B</i>	RADKFRTATIMIQT V R G W L Q K V K Y H R L K G A T L T L
<i>Pan troglodytes</i>	RADKFRTATIMIQT V R G W L Q K V K Y R R L K G A T L T L
<i>Pongo pygmaeus</i>	RADKFRTATIMIQT V R G W L Q K V K Y R R L K G A T L T L
<i>Macaca mulatta</i>	RADKFRTATIMIQT V R G W L Q K V K Y R R L K G A T L T L
<i>Otolemur garnettii</i>	RADRFRAATIMIQT V R G W L Q K V K Y H R L K G A T L T L
<i>Tupaia belangeri</i>	RADKFRAATIMIQT V R G W L Q K V K Y R R L K K A T L T L
<i>Equus caballus</i>	RADKFRAATIMIQT V R G W L Q R V K Y R R L K W A T L T L
<i>Spermophilus tridecemlineatus</i>	RADKFRAATIMIQT V R G W L Q K V K Y R R L K R A T L T L
<i>Loxodonta Africana</i>	RADKFRAATIMIQT V R G W L Q K V K Y R R L K R A T L T L
<i>Cavia porcellus</i>	RADKFRAATIMIQT V R G W L Q R V K Y N R L K K A T V T L
<i>Bos taurus</i>	RADKFRAATIMIQT V R G W L Q K V K Y R R L K G A T L I L
<i>Ochotona princeps</i>	RADKFRAATIMIQT V R G W L Q R V K Y R R L K G A A L T L
<i>Myotis lucifugus</i>	RADKFRAATIMIQT V R G W L Q R V K Y R R L K G A T L T L
<i>Canis familiaris</i>	-----
<i>Erinaceus europaeus</i>	RADKFRAATIMIQT V R G W L Q R V K Y Q R L K G A T L T L
<i>Rattus norvegicus</i>	RADKFREATIMIQT V R G W L Q R V K Y R R L R A A T L T L
<i>Mus musculus</i>	RADKFREATIMIQT V R G W L Q R V K Y R R L R A A T L S L
<i>Echinops telfairi</i>	RADRFRAATIMIQT V R G W L Q K V K Y R R L K R A T I T L
<i>Monodelphis domestica</i>	RADKFRAATIMIQT V R G W L Q K V K Y R R L R G A T L T L
<i>Xenopus tropicalis</i>	-----
<i>Danio rerio</i>	RADKFRAFACIKIQKT V R G W L Q R I R Y R K I R K S A I T L
<i>Tetraodon nigroviridis</i>	RADKFRAACIKIQKT V R G W L Q R V R Y R K I Y R A A V T L
<i>Takifugu rubripes</i>	RADKFRAACIKIQKT V R G W L Q R I R Y R K I C K A A I T L
<i>Oryzias latipes</i>	-----
<i>Ciona savigny</i>	RADKFRTATIMIQT V R G W L Q R N V R M W L Y R N K Y I R M K R S A V I I
<i>Ciona intestinalis</i>	RANKLRACAVI I Q K N T R M W L Q Y K R Y I R M K Q S A I V V

Paralogues

<i>Homo sapiens MY05A</i>	RADKLRAACIRIQKT I R G W L L R K K Y L R M R K A A I T M
<i>Homo sapiens MY05C</i>	RLDKLRQSCVMVQKHMRG W L Q R K K F L R R R A A L I I

Type of file: figure

Label: 7

Filename: Szperl_SupFig3-revised.doc

Supplementary figure 3.

c.2330_delG
p.Gly777AsnfsX6

WT

2300 CCACCATCATGATCCAGAAAAGTGTCCGGG**G**ATGGCTGCAGAAGGTGAAATATCACAGGC
767 A--T--I--M--I--Q--K--T--V--R--G--W--L--Q--K--V--K--Y--H--R--

Mutated

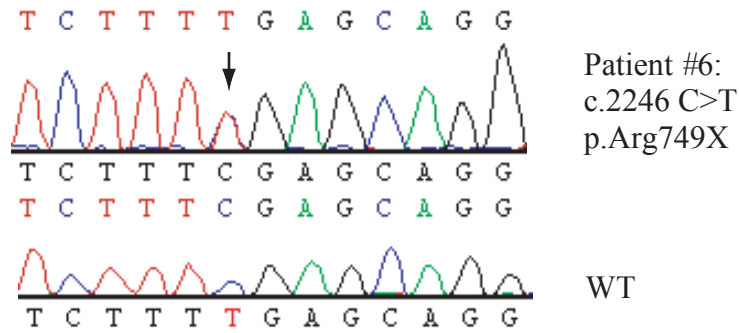
2300 CCACCATCATGATCCAGAAAAGTGTCCGGGAT**GGC**TGC**AGA**AGG**TGA**AAATATCACAGGC
767 A--T--I--M--I--Q--K--T--V--R--D--G--C--R--R--**X**--

Type of file: figure

Label: 8

Filename: Szperl_SupFig4-revised.pdf

Supplementary figure 4



Orthologues

p.Arg749

<i>Homo sapiens MYO5B</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRTAT
<i>Pan troglodytes</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRTAT
<i>Pongo pygmaeus</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRTAT
<i>Macaca mulatta</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRTAT
<i>Otolemur garnettii</i>	XXDPDKFQFGRTKIFFRAGQVAYLEKLRADRFRAAT
<i>Tupaia belangeri</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Equus caballus</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Spermophilus tridecemlineatus</i>	XXDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Loxodonta Africana</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Cavia porcellus</i>	IRDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Bos taurus</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Ochotona princeps</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Myotis lucifugus</i>	IKN-DK-QFGRTKI-FRAGQVAYLEKLRADKFRAAT
<i>Canis familiaris</i>	-----
<i>Erinaceus europaeus</i>	XXDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Rattus norvegicus</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFREAT
<i>Mus musculus</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFREAT
<i>Echinops telfairi</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADRFRAAT
<i>Monodelphis domestica</i>	IKDPDKFQFGRTKIFFRAGQVAYLEKLRADKFRAAT
<i>Xenopus tropicalis</i>	-----
<i>Danio rerio</i>	IKDPDKFQFGKTKIFFRAGQVAYLEKLRADKFRFAC
<i>Tetraodon nigroviridis</i>	IKEPDMFQFGKTKIFFRAGQVAYLEKLRADKFRAAC
<i>Takifugu rubripes</i>	IKGTRHVQFGKTKIFFRAGQVAYLEKIRADKFRAAC
<i>Oryzias latipes</i>	-----
<i>Ciona savigny</i>	IPEADKYQPGKNKIFFRAGQVAYLEKLRADKLRSCA
<i>Ciona intestinalis</i>	IPEEDKYQPGKNKIFFRAGQVAYLEKLRANKLRACA

Paralogues

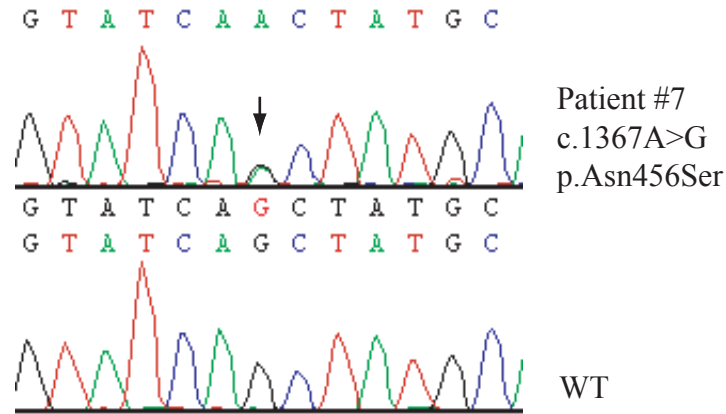
<i>Homo sapiens MYO5A</i>	ILDKDKYQFGKTKIFFRAGQVAYLEKLRADKLRAC
<i>Homo sapiens MYO5C</i>	IQDSNQYQFGKTKIFFRAGQVAYLEKLRADKLRQSC

Type of file: figure

Label: 9

Filename: Szperl_SupFig5-revised.pdf

Supplementary figure 5



Orthologues

p.Asn456

<i>Homo sapiens MYO5B</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Pan troglodytes</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Pongo pygmaeus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Macaca mulatta</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Microcebus marinus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Otolemur garnettii</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Tupaia belangeri</i>	EVNSFEQ CI Y ANEK L QQQF
<i>Felis catus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Equus caballus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Spermophilus tridecemlineatus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Loxodonta Africana</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Dasyus novemcinctus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Cavia porcellus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Bos taurus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Ochotona princeps</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Myotis lucifugus</i>	XXXXXXXXXXXXXXXXXXXXXXXXXXXX
<i>Canis familiaris</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Erinaceus europaeus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Rattus norvegicus</i>	EINSFEQFCIN Y ANEK L QQQF
<i>Mus musculus</i>	EINSFEQFCIN Y ANEK L QQQF
<i>Echinops telfairi</i>	ESNSFEQFCIN Y ANEK L QQQF
<i>Monodelphis domestica</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Ornithorhynchus anatinus</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Gallus gallus</i>	NLNSFEQFCIN Y ANEK L QQQF
<i>Danio rerio</i>	EINSFEQFCIN Y ANEK L QQQF
<i>Tetraodon nigroviridis</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Takifugu rubripes</i>	DINSFEQFCIN Y ANEK L QQQF
<i>Oryzias latipes</i>	ERNSFEQFCIN Y ANEK L QQQF
<i>Gasterosteus aculeatus</i>	NFKTFKSSFCLSAASRR L QQF
<i>Ciona savigny</i>	EVNSFEQFCIN Y ANEK L QQQF
<i>Ciona intestinalis</i>	ENNSFEQFCIN Y ANEK L QQQF

Paralogues

<i>Homo sapiens MYO5A</i>	EINSFEQFCIN Y ANEK L QQQF
<i>Homo sapiens MYO5C</i>	DVNSFEQFCIN Y ANEK L QQQF