

Appendix 1. A list of members of 99 Lives Cat Genome Sequencing Consortium, and detailed information on magnetic resonance imaging sequences, whole genome sequencing, fibroblast cell culture, feline *COL6A1* cDNA analysis, and histopathology and transmission electron microscopic analysis.

A list of members of 99 Lives Cat Genome Sequencing Consortium (Domestic cat analysis – whole genome sequencing data from 362 cats and whole exome sequencing data from 52 cats)

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Magnetic resonance imaging sequences obtained for the brain and spinal cord

The sequences for brain magnetic resonance imaging (MRI) obtained are as follows: T2-weighted, fluid-attenuated inversion recovery, T1-weighted, and contrast-enhanced T1-weighted images in sagittal, dorsal, and transverse planes. The spinal MRI sequences obtained were as follows: T2-weighted and T1-weighted images in sagittal and transverse planes, and transverse sections were obtained at levels between the T4–T12 vertebrae.

Detailed information regarding whole genome sequencing and its analysis

DNA was extracted from heparinized whole blood using Genra Puregene Blood Kit (Qiagen, Hilden, Germany) and submitted to Macrogen Japan (Tokyo, Japan), where DNA library preparation and whole genome sequencing (WGS) were performed. In brief, DNA library was prepared using TruSeq DNA PCR-Free (Illumina, San Diego, CA, USA) with 350 bp of the target insert size. Then WGS was conducted for the case using illumina HiSeq X Ten system (Illumine) with 2×150 base paired-end reads. Read mapping, realignment, and variant calling were carried out as described (1), with respect to a feline genome assembly (Felis_catus_9.0) (2) and RefSeq 104 annotation. The sequence data was included in the 99 Lives Cat Genome Sequencing Consortium (<http://felinegenetics.missouri.edu/99lives>) and submitted to the NCBI

short read archive under BioProject: PRJNA614458; BioSample: SAMN14425582. The 99 Lives Cat Genome Sequencing Consortium members are listed at the top of this file.

Approximately 25× genomic coverage was generated for this cat. This case's WGS data were compared with the 99 Lives Cat Genome Sequencing Consortium dataset was conducted using VarSeq v2.2.5 software (Golden Helix, Bozeman, Montana, USA), which included 362 cats' WGS and 52 cats' whole exome sequencing data. Variants affecting the coding sequence of genes or existing in 10 bp regions flanking exons in this cat were filtered, for which the cat is privately homozygous; heterozygosity in up to one additional cat was permitted. After that, unique candidate gene variants were assessed, including previously known causative genes of human or animal Ehlers-Danlos Syndromes (EDS) or congenital scoliosis based on the published literature. Those for EDS initially included the following 20 orthologous candidate genes: *COL5A1* (XM_023242950.1), *COL5A2* (XM_003990962.4), *COL1A1* (XM_003996699.4), *COL3A1* (XM_003990961.3), *COL1A2* (XM_003982764.5), *ADAMTS2* (XM_023254116.1), *PLOD1* (XM_003989499.3), *FKBP14* (XM_003982902.4), *TNXB* (XM_023254711.1), *COL12A1* (XM_019830870.1), *CHST14* (XM_003987275.4), *DSE* (XM_019831105.2), *B4GALT7* (XM_023254211.1), *B3GALT6* (XM_011279777.2), *SLC39A13* (*ZIP13*) (XM_023239820.1), *ZNF469* (XM_023245050.1), *PRDM5* (XM_023252840.1), *C1R* (XM_019834454.2), *C1S* (XM_006933467.4), *AEBP1* (*ACLP*) (XM_023250110.1) (3,4). Those for congenial scoliosis included the following 37 orthologous candidate genes, including *DLL3* (XM_003997806.5), *MESP2* (XM_019833746.2), *LFNG* (XM_023246563.1), *HES7* (XM_003996191.4), *NOTCH2* (XM_011285094.3), *NOTCH3* (XM_023246608.1), *SLC35A3* (XM_003990360.4), T (*TBXT*) (XM_003986708.3), *TBX6* (XM_011290514.3), *FBNI* (XM_023255387.1), *SHH* (XM_023242357.1), *SOX9* (XM_023243815.1), *FLNB*

(XM_019823423.2), *PTK7* (XM_003986191.4), *HSPG2* (XM_023258277.1), *KIAA1217* (XM_023256446.1), *DCC* (XM_019815281.2), *DVL3* (XM_023260347.1), *FGFR3* (XM_023253489.1), *RECQL4* (XM_019823278.2), *ROBO3* (XM_019811488.2), *WNT7A* (XM_011280231.3), and *CHRNA3* (XM_003991260.4), all which or some of which were previously used elsewhere (5,6), and recently reported candidate gene, named *FGFR1* (XM_011281522.3) (7). Subsequently, the number of genes studied was expanded to 55, including hereditary connective tissue disorders overlapping phenotype genes in addition to genes associated with EDS (8). Those 55 orthologous genes were the following: *ADAMTS2* (XM_023254116.1), *B3GALT6* (XM_011279777.2), *CHST14* (XM_003987275.4), *COL1A1* (XM_003996699.4), *COL1A2* (XM_003982764.5), *COL3A1* (XM_003990961.3), *COL5A1* (XM_023242950.1), *COL5A2* (XM_003990962.4), *DSE* (XM_019831105.2), *FKBP14* (XM_003982902.4), *PLOD1* (XM_003989499.3), *TNXB* (XM_023254711.1), *COL12A1* (XM_019830870.1), *C1R* (XM_019834454.2), *C1S* (XM_006933467.4), *SLC39A13* (XM_023239820.1), *ZNF469* (XM_023245050.1), *PRDM5* (XM_023252840.1), *LZTS1* (XM_003984723.4), *FLNA* (XM_023249411.1), *COL4A1* (XM_023253172.1), *COL6A1* (XM_011285711.3), *COL6A2* (XM_023238595.1), *COL6A6* (XM_011286221.3), *ELN* (XM_019820582.2), *FBLN5* (XM_019833518.1), *FBNI* (XM_023255387.1), *LOX* (XM_023254614.1), *MYH11* (XM_023246707.1), *RYR1* (XM_023245508.1), *SEPNI* (XM_023258321.1), *SGCB* (XM_023253126.1), *myosin-7 (MYH7)* (XM_006932746.4), *TTN* (XM_023259509.1), *SMAD2* (XM_023241966.1), *SMAD3* (XM_023255153.1), *COL2A1* (XM_023256993.1), *COL11A1* (XM_023258940.1), *COL11A2* (XM_003985999.5), *COL9A1* (XM_003986294.4), *COL9A2* (XM_023258522.1), *COL9A3* (XM_023251026.1), *MED12* (XM_004000604.5), *FLNB* (XM_019823423.2), LOC105261197 (*CANT1*) (XM_019818343.2),

SLC2A10 (XM_006929697.2), *ABCC6* (XM_011290585.3), *GGCX* (XM_011281186.3), *ENPPI* (XM_023254376.1), *AEBPI* (XM_023250110.1), *SKI* (XM_023257965.1), *TGFB2* (XM_003999507.5), *TGFB3* (XM_003987851.3), *TGFBRI* (XM_023242499.1), *TGFBRI* (XM_023260647.1). Furthermore, the presence of a *HES7* variant (XM_003996191.5; c.5A>G; p.Val2Ala [NC_058381.1:chromosome E1:g.2918735A>G]) and *T* variants (XM_003986708.3; c.1196del,c.1166del,c.995delT,c.[995_1011dup;1011_1014del] [NC_058372.1:chromosome B2:g.152019395del, g.152019424del, g.152021379del,g.{152021360_152021362del;152021363_152021379dup}}) were checked, which were known to cause kinked-tail phenotype in Japanese Bobtail cats and Manx cats, respectively (9,10). Those genes and corresponding phenotypes were summarized in **Table S1**.

Method of fibroblast cell culture

Fibroblasts, derived from skin biopsy samples, were grown in Dulbecco's Modified Eagle's Medium - low glucose (Sigma-Aldrich, St. Louis, MO, USA) supplemented with 10% fetal bovine serum and 1% penicillin-streptomycin-amphotericin B at 37 °C and 5% CO₂.

Skin biopsy from healthy cats for cDNA sequence analysis, histopathology, and transmission electron microscopic analysis

Ethical approval for the skin biopsy from healthy cats was obtained from the Animal Care and Use Committee of Nippon Veterinary and Life Science University (Accession No. 2019S-32, S2019S-32). Healthy cats were part of another study by the Laboratory of Veterinary Radiology, Nippon Veterinary and Life Science University (Accession Nos. 2019K-2, S2019K-2).

For cDNA analysis, a skin biopsy was performed on 1 healthy cat (sex: female; age: 57 mo; body weight: 5.9 kg) from the caudal cervical region. The skin sample was cut into pieces and immediately snap-frozen in liquid nitrogen. For histopathology, and transmission electron microscopic analysis, a skin biopsy was conducted on another healthy cat (sex: female; age: 106 mo; body weight: 3.6 kg) from the caudal cervical region. The skin biopsies in each cat were performed under general anesthesia or sedation, respectively.

Methods of RNA extraction, reverse transcription PCR, and cDNA sequence analysis of feline *COL6A1*

Reverse transcription PCR (RT-PCR) and Sanger sequencing were performed to validate the in-frame deletion in *COL6A1* in the affected cat. Cultured cells established from the affected cat's dermal fibroblast were used as material for RNA extraction.

Total RNA of the affected cat and the healthy cat was extracted from the cultured cells and a skin sample using a FastGene RNA Premium Kit (NIPPON Genetics, Tokyo, Japan). RT-PCR was performed using ReverTra Ace (TOYOBO, Osaka, Japan) with the Oligo(dT)20 Primer (TOYOBO). Then, PCR was conducted using the synthesized cDNA, the primer sets, and Tks Gflex DNA Polymerase (Takara Bio, Shiga, Japan) with a 3-step PCR protocol, following the manufacturer's instructions. The PCR conditions and the primer sequences used are summarized in **Table S5**. Purification of PCR products and the subsequent Sanger sequencing was performed as described (11).

Sanger sequencing of the cDNA confirmed that the affected cat had an in-frame 3-bp deletion predicted to delete one amino acid residue (XM_011285711.3:c.1678_1680del; XP_011284013.1:p.Asn560del) in *COL6A1*, which suggested that the affected cat had a 1026

amino acid sequence (3081 nucleotides) whereas a normal cat had a 1027 amino acid sequence (3084 nucleotides). The *COL6A1* coding sequences of both the affected cat and a normal cat were submitted to the DNA Data Bank of Japan (DDBJ) (Accession Nos. LC750709 and LC750710, respectively).

Methods of histopathology and transmission electron microscopic analysis

For histopathological analysis, skin biopsy samples were fixed in 10% neutral buffered formalin, processed routinely, and embedded in paraffin wax. Sections (4 μ m) were stained with hematoxylin and eosin (H&E) and Masson's trichrome. For transmission electron microscopy analysis, small pieces of cat skin samples were prefixed in 4% paraformaldehyde and 1% glutaraldehyde in 0.1 M phosphate buffer, postfixed in 1% osmium tetroxide in 0.1 M phosphate buffer, dehydrated through a graded ethanol series, transferred to 100% acetone, and then embedded in epoxy resin. Using a transmission electron microscope (JEM1011; JEOL, Tokyo, Japan), ultrathin sections were examined after staining with uranyl acetate and lead citrate.

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Table S1. Private homozygous genetic variants were identified in the case of 414 cats of the 99 Lives Cat Genome Sequencing Consortium dataset, including WGS data from 362 cats and whole exome sequencing data from 52 cats. Variant calling was performed with respect to the *Felis_catus_9.0* reference genome assembly. Variants listed are those for which the cat is homozygous privately, but heterozygosity in up to 1 other cat was permitted. Presented are variants affecting the coding sequence of genes or existing in 10 bp regions flanking exons.

Chr :Pos	Ref/Alt	Filter	Allele Counts	Allele Frequencies	# Alt Alleles	# H om o z y g o u s	# H o m o z y g o u s i n o t h e r c a t	# S a m p l e s	Gene Names	Sequence Ontology (Combined)	Effect (Com bined)	Transcript Name (Clinically Relevant)	HGVS c. (Clinically Relevant)	HGVS p. (Clinical Relevant)	Variant Allele Freq of the Case	Allelic Depths (AD) of the Case	Read Depths (DP) of the Case	Genotype Qualities (GQ) of the Case
E1: 578 118 82	/TGGGG GACCGC AGGGGG CT	PASS	2	0.002 41546	82 8	0	1	1	<i>FBF1</i>	splice_acc eptor_varia nt	LoF	XM_019818 272.2	XM_019818272.2:c.2411- 20_2411-2dup		1	0,13	13	39
B2: 415 638 0	A/-	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC10 108520 7</i>	frameshift _variant	LoF	XM_023253 584.1	XM_023253584.1:c. 458delA	p.Lys153 Argfs*7	1	0,21	21	63
C1: 105 127 310	-/A	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC10 108842 3</i>	frameshift _variant	LoF	XM_006935 068.4	XM_006935068.4:c. 56dupA	p.His19Glnfs*57	1	0,24	24	72
C1: 127 706 894	G/A	PASS	3	0.003 62319	82 8	1	1	2	<i>LOC10 526085 1</i>	splice_acc eptor_varia nt	LoF	XR_0027447 67.1	XR_002744767.1:n.2405-1G>A XR_002159997.1:n.		1	0,26	26	78
B4: 122 551 848	C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>ANKS1 B,FAM 71C</i>	missense_ variant	Misse nse	XR_0021599 97.1, XM_01 9835379.1	1309+4955G>A, X M_019835379.1:c.3 83C>T	p.Thr128 Met	1	0,26	26	78
E1: 398 550 60	G/A	PASS	3	0.003 62319	82 8	1	1	2	<i>ARHGA P23 ARID1 B,LOC 111560 543</i>	missense_ variant	Misse nse	XM_023244 567.1	XM_023244567.1:c. 967G>A XM_023254522.1:c. 709T>G, XR_00274 522.1, XR_00 2742522.1	p.Asp323 Asn	1	0,19	19	57
B2: 144 037 370	T/G	VQSRTran cheSNP99. 90to100.00	2	0.002 41546	82 8	0	1	1	<i>111560 543</i>	missense_ variant	Misse nse	XM_023254 522.1, XR_00 2742522.1	2522.1:n.87+601A> C	p.Phe237 Val,	1	0,2	2	7
A2: 126 523 252	A/C	PASS	2	0.002 41546	82 8	0	1	1	<i>BMPER</i>	missense_ variant	Misse nse	XM_003982 930.5	XM_003982930.5:c. 232A>C	p.Thr78Pr o	1	0,23	23	69
E3: 112 589 67	C/A	PASS	2	0.002 41546	82 8	0	1	1	<i>CASTO R2</i>	missense_ variant	Misse nse	XM_023246 384.1	XM_023246384.1:c. 592G>T	p.Ala198S er	1	0,20	20	60

B4: 119 334 703 D4: 269 302 19 B3: 207 175 1 X:4 109 184 3 A2: 119 192 88 C2: 838 840 A1: 188 662 224 A1: 711 263 0 B1: 844 872 37 C1: 104 653 076 X:4 617 580 9 C1: 103 358 745 A1: 599 017 8 B4: 416 271 64 A2: 122 664 33 C1: 557	PASS,VQS RTrancheS NP99.00to9 9.90	3	0.003 62319	82 8	1	1	2	<i>CCDC38</i>	missense_ variant	Misse nse	XM_011284 160.3	XM_011284160.3:c. 269T>C	p.Phe90Se r	1	0.20	20	60
	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>CD4H9 orf153</i>	missense_ variant	Misse nse	XM_019815 828.2	XM_019815828.2:c. 553G>A	p.Gly185 Arg	1	0.20	20	60
	PASS	2	0.002 41546	82 8	0	1	1	<i>CEMIP</i>	missense_ variant	Misse nse	XM_023254 807.1	XM_023254807.1:c. 382G>A	p.Val128Il e	1	0.21	21	63
	VQSRTran cheSNP99. 90to100.00	2	0.002 41546	82 8	0	1	1	<i>CHST7</i>	missense_ variant	Misse nse	XM_023248 942.1	XM_023248942.1:c. 559A>C	p.Thr187P ro	1	0.5	5	15
	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>CIB3</i>	missense_ variant	Misse nse	XM_023241 207.1	XM_023241207.1:c. 469G>C	p.Gly157 Arg	1	0.14	14	42
	PASS	3	0.003 62319	82 8	1	1	2	<i>COL6A1</i>	inframe_de letion	Misse nse	XM_011285 711.3	XM_011285711.3:c. 1678_1680delAAC	p.Asn560d el	1	0.20	20	60
	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>GABRG2</i>	missense_ variant	Misse nse	XM_003981 341.5	XM_003981341.5:c. 8C>T	p.Pro3Leu	1	0.12	12	36
	VQSRTran cheSNP99. 90to100.00	2	0.002 41546	82 8	0	1	1	<i>GTF3A</i>	missense_ variant	Misse nse	XM_011282 105.2	XM_011282105.2:c. 1247G>A	p.Cys416 Tyr	1	0.19	19	57
	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>HHIP</i>	missense_ variant	Misse nse	XM_003984 977.5	XM_003984977.5:c. 590A>G	p.Asp197 Gly	1	0.29	29	87
	PASS	2	0.002 41546	82 8	0	1	1	<i>ITGA10</i>	missense_ variant	Misse nse	XR_0021612 63.2	XR_002161263.2:n.88C>T		1	0.25	25	75
	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>KDM5C</i>	missense_ variant	Misse nse	XM_023249 335.1	XM_023249335.1:c. 4060G>A	p.Gly1354 Arg	1	0.8	8	24
	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>LOC101093537</i>	missense_ variant	Misse nse	XM_006935 036.4	XM_006935036.4:c. 2557A>G	p.Met853 Val	1	0.29	29	87
	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC101096779</i>	missense_ variant	Misse nse	XM_019821 051.2	XM_019821051.2:c. 883A>T	p.Ile295Le u	1	0.18	18	54
	PASS	3	0.003 62319	82 8	1	1	2	<i>LOC109501438,NCA PD2</i>	missense_ variant	Misse nse	XR_0021594 88.2, XM_019834405.2	XR_002159488.2:n. 82+2075C>T, XM_019834405.2:c. 4141G>A	,p.Asp138 IAsn	1	0.16	16	48
	PASS	3	0.003 62319	82 8	1	1	2	<i>MED26</i>	missense_ variant	Misse nse	XM_023246 873.1	XM_023246873.1:c. 301G>A	p.Ala101T hr	1	0.25	25	73
	PASS	3	0.003 62319	82 8	1	1	2	<i>MIER1</i>	missense_ variant	Misse nse	XM_019837 205.1	XM_019837205.1:c. 812G>A	p.Cys271 Tyr	1	0.28	28	84

334																			
58																			
B4:																			
167																			
369				0.003	82														
66	A/G	PASS	3	62319	8	1	1	2	<i>MRC1</i>	missense_	Misse	XM_011283	XM_011283622.3:c.	p.Asp63Gly	1	0,17	17	51	
D1:																			
106																			
982		VQSRTrancheSNP99.00to99.90	3	0.003	82	1	1	2	<i>MS4A15</i>	missense_	Misse	XM_003993	XM_003993487.4:c.	p.Ala49Thr	1	0,15	15	45	
531	G/A																		
C1:																			
113																			
590		VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>MYO7B</i>	missense_	Misse	XM_019837	XM_019837914.2:c.	p.Val1691Met	0.928571	1,13	14	32	
948	C/T																		
A1:																			
210																			
532																			
841	C/T	PASS	3	0.003	82	1	1	2	<i>OSMR</i>	missense_	Misse	XM_023238	XM_023238963.1:c.	p.Val697Met	1	0,21	21	63	
E1:																			
181																			
026																			
61	A/G	PASS	2	0.002	82	0	1	1	<i>PROCA1</i>	missense_	Misse	XM_006940	XM_006940026.4:c.	p.Glu198Gly	1	0,14	14	42	
C2:																			
426																			
893		VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>RIPK4</i>	missense_	Misse	XM_023238	XM_023238708.1:c.	p.Ala699Val	1	0,18	18	53	
2	C/T																		
D3:																			
834																			
279		VQSRTrancheSNP99.00to99.90	3	0.003	82	1	1	2	<i>SERP1NB5</i>	missense_	Misse	XM_003995	XM_003995237.4:c.	p.Gln71Glu	1	0,17	17	51	
65	C/G																		
A2:																			
129																			
435		VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>SLC27A1</i>	missense_	Misse	XM_011290	XM_011290625.2:c.	p.Gly561Ser	1	0,23	23	69	
80	G/A																		
E1:																			
579																			
367		VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>SRP68</i>	missense_	Misse	XM_003997	XM_003997170.4:c.	p.Val224Leu	1	0,26	26	78	
61	C/A																		
A2:																			
113																			
332		VQSRTrancheSNP99.00to99.90	3	0.003	82	1	1	2	<i>SYDE1</i>	missense_	Misse	XM_023241	XM_023241193.1:c.	p.Pro184Ala	1	0,21	21	63	
17	C/G																		
B4:																			
415																			
675																			
58	T/A	PASS	2	0.002	82	0	1	1	<i>TAPBP L</i>	missense_	Misse	XM_023256	XM_023256705.1:c.	p.Val131Glu	1	0,20	20	60	
B3:																			
124																			
328																			
158	G/A	PASS	2	0.002	82	0	1	1	<i>TMEM63C</i>	missense_	Misse	XR_440592.3	XR_440592.3:n.1465G>A		1	0,25	25	75	
B4:																			
166																			
627																			
34	A/T	PASS	3	0.003	82	1	1	2	<i>TMEM236</i>	missense_	Misse	XM_019834	XM_019834003.2:c.	p.Thr34Ser	1	0,26	26	78	
C2:																			
160		PASS,VQSRTrancheSNP99.00to99.90	3	0.003	82	1	1	2	<i>TOPAZ1</i>	missense_	Misse	XM_023260	XM_023260821.1:c.	p.Asn149Ser	1	0,26	26	78	
653	A/G																		
073																			
C1:																			
381																			
515																			
02	C/T	PASS	2	0.002	82	0	1	1	<i>TRABD2B</i>	missense_	Misse	XR_0021608	XR_002160835.2:n.1574G>A		1	0,13	13	39	

D3: 930 048 54 E2: 713 294 9 B2: 131 009 829 A2: 115 779 39 B3: 310 817 3 B3: 310 868 8 B3: 310 893 6 D4: 264 510 25 C1: 113 356 106 A1: 233 694 164 B2: 127 526 437 C1: 114 125 572 C1: 114 137 697 C1: 114 147 359 D1: 115 423 505 B3: 208	C/T	VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>TSHZ1</i>	missense_variant	Mis nse	XM_023242110.1	XM_023242110.1:c.2033C>T	p.Ala678 Val	1	0.21	21	62
	G/A	PASS	2	0.002 41546	82 8	0	1	1	<i>VSIG10L</i>	missense_variant	Mis nse	XM_023245113.1	XM_023245113.1:c.1880G>A	p.Arg627 His	1	0.22	22	66
	G/A	PASS	2	0.002 41546	82 8	0	1	1	<i>VTA1</i>	missense_variant	Mis nse	XM_003986613.5	XM_003986613.5:c.575G>A	p.Arg192 Gln	1	0.24	24	72
	G/A	VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>WIZ</i>	missense_variant	Mis nse	XM_023246657.1	XM_023246657.1:c.5342C>T	p.Pro1781 Leu	1	0.17	17	51
	G/C	VQSRTrancheSNP99.90to100.00	2	0.002 41546	82 8	0	1	1	<i>ADAM TSL3</i>	3_prime_UTR_variant	Other	XM_023254830.1	XM_023254830.1:c.*784C>G		1	0.24	24	72
	C/T	VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>ADAM TSL3</i>	3_prime_UTR_variant	Other	XM_023254830.1	XM_023254830.1:c.*269G>A		1	0.15	15	45
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>ADAM TSL3</i>	3_prime_UTR_variant	Other	XM_023254830.1	XM_023254830.1:c.*21G>A		1	0.20	20	60
	A/G	PASS	3	0.003 62319	82 8	1	1	2	<i>AGTPB PI</i>	synonymous_variant	Other	XM_023242223.1	XM_023242223.1:c.3579T>C	p.Asn1193 =	1	0.24	24	72
	A/G	VQSRTrancheSNP99.00to99.90	3	0.003 62319	82 8	1	1	2	<i>AMME CRIL</i>	splice_region_variant	Other	XM_003990674.4	XM_003990674.4:c.518+7A>G		1	0.25	25	75
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>ANKR D33B</i>	3_prime_UTR_variant	Other	XM_011287289.3	XM_011287289.3:c.*91G>A		1	0.19	19	56
	C/T	VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>ARFGE F3</i>	synonymous_variant	Other	XM_023254439.1	XM_023254439.1:c.1608C>T	p.Leu536 =	1	0.24	24	72
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>BIN1</i>	synonymous_variant	Other	XM_023259216.1	XM_023259216.1:c.222C>T	p.Ala74=	1	0.22	22	66
	G/A	PASS	2	0.002 41546	82 8	0	1	1	<i>BIN1</i>	synonymous_variant	Other	XM_023259216.1	XM_023259216.1:c.972G>A	p.Pro324=	1	0.18	18	54
	-/C	VQSRTrancheINDEL99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>BIN1</i>	3_prime_UTR_variant	Other	XM_023259216.1	XM_023259216.1:c.*137dupC		1	0.8	8	30
	G/T	VQSRTrancheSNP99.00to99.90	3	0.003 62319	82 8	1	1	2	<i>CDKN1C</i>	5_prime_UTR_variant	Other	XM_023240183.1	XM_023240183.1:c.-56G>T		1	0.15	15	45
	G/A	PASS,VQSRTrancheS	3	0.003 62319	82 8	1	1	2	<i>CEMIP</i>	synonymous_variant	Other	XM_023254807.1	XM_023254807.1:c.1674G>A	p.Ala558=	1	0.24	24	72

920 4 A3: 189 658 55 E1: 426 715 62 C2: 744 671 64 E1: 598 468 20 C1: 447 230 94 C2: 226 189 0 D3: 583 900 10 D3: 584 506 64 C1: 154 016 015 C1: 154 016 024 B4: 416 330 60 A3: 174 768 6 X:1 122 459 52 C1: 113 570 748 C1: 112 980 091		NP99.00to9 9.90																	
A/G	PASS	3	0.003 62319	82 8	1	1	2	<i>CHD6</i>	splice_regi on_variant	Other	XM_019826 498.2	XM_019826498.2:c.975-4A>G	1	0,20	20	60			
T/G	PASS	2	0.002 41546	82 8	0	1	1	<i>CNP</i>	3_prime_U TR_variant	Other	XM_003996 870.5	XM_003996870.5:c.*745T>G	1	0,19	19	57			
T/C	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>CPN2</i>	synonymo us_variant	Other	XM_003991 783.3	XM_003991783.3:c. 1354T>C	p.Leu452 =	1	0,39	39	99		
C/T	PASS	3	0.003 62319	82 8	1	1	2	<i>CYTH1</i>	3_prime_U TR_variant	Other	XM_023244 156.1	XM_023244156.1:c.*96G>A	1	0,20	20	59			
C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>DHCR2 4</i>	3_prime_U TR_variant	Other	XM_023258 735.1	XM_023258735.1:c.*912G>A	1	0,19	19	56			
G/C	PASS	3	0.003 62319	82 8	1	1	2	<i>DNMT 3L</i>	3_prime_U TR_variant	Other	XM_023260 865.1	XM_023260865.1:c.*575G>C	1	0,22	22	66			
GGT/-	VQSRTran cheINDEL9 9.00to99.90 PASS,VQS RTrancheS NP99.00to9 9.90	2	0.002 41546	82 8	0	1	1	<i>DTNA</i>	splice_regi on_variant	Other	XR_0021476 25.2	XR_002147625.2:n.986+6_986+8d eITGG	1	0,3	3	9			
C/A		3	0.003 62319	82 8	1	1	2	<i>DTNA</i>	synonymo us_variant	Other	XR_0021476 25.2	XR_002147625.2:n.2226C>A	1	0,21	21	63			
G/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>FAP</i>	synonymo us_variant	Other	XM_011285 347.2	XM_011285347.2:c. 687C>A	p.Thr229=	1	0,23	23	69		
A/G	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>FAP</i>	synonymo us_variant	Other	XM_011285 347.2	XM_011285347.2:c. 678T>C	p.Phe226=	1	0,22	22	66		
A/G	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>GAPD H</i>	synonymo us_variant	Other	NM_001009 307.1	NM_001009307.1:c. 513A>G	p.Gly171=	1	0,20	20	60		
C/-	VQSRTran cheINDEL9 9.00to99.90	2	0.002 41546	82 8	0	1	1	<i>GATA5</i>	splice_regi on_variant	Other	XR_0027408 78.1	XR_002740878.1:n.919-3delC	1	0,21	21	63			
A/G	PASS	2	0.002 41546	82 8	0	1	1	<i>GPC4</i>	3_prime_U TR_variant	Other	XM_004000 909.5	XM_004000909.5:c.*945T>C	1	0,11	11	33			
G/A	PASS	3	0.003 62319	82 8	1	1	2	<i>GPR17, LIMS2</i>	synonymo us_variant	Other	XM_006935 193.3, XM_0 23259205.1	XM_006935193.3:c. 867C>T, XM_02325 9205.1:c.434+4155 G>A	p.Asn289 =,	1	0,19	19	57		
G/A	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>HS6ST 1</i>	synonymo us_variant	Other	XM_023259 197.1	XM_023259197.1:c. 849G>A	p.Lys283=	1	0,21	21	63		

E1: 431 199 46 C1: 154 070 791 E2: 714 340 7 C1: 100 108 969 C1: 113 705 553 C1: 113 705 781 E1: 584 615 21 E1: 584 621 22 E1: 584 623 51 E1: 584 626 77 E1: 584 640 96 E1: 584 642 75 E1: 584 643 67 E1: 584 646 49 C2: 771 606 9	C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>HSD17 BI</i>	synonymo us_variant	Other	XM_006940 403.4	XM_006940403.4:c. 291C>T	p.Leu97=	1	0,18	18	52
	TA/-	PASS	2	0.002 41546	82 8	0	1	1	<i>IFIH1</i>	3_prime_U TR_variant	Other	XM_006935 321.4	XM_006935321.4:c.*133_*134del TA		1	0,25	25	75
	G/A	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>IGLON 5</i>	synonymo us_variant	Other	XM_023245 114.1	XM_023245114.1:c. 600C>T	p.Gly200=	1	0,30	30	88
	GTCT/-	PASS	3	0.003 62319	82 8	1	1	2	<i>IGSF3</i>	3_prime_U TR_variant	Other	XM_019837 688.2	XM_019837688.2:c.*168_*171del AGAC		1	0,20	20	60
	C/T	PASS	3	0.003 62319	82 8	1	1	2	<i>IWS1</i>	synonymo us_variant	Other	XR_0021613 35.2	XR_002161335.2:n.825C>T		1	0,22	22	66
	C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>IWS1</i>	synonymo us_variant	Other	XR_0021613 35.2	XR_002161335.2:n.1053C>T		1	0,17	17	51
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*3168G>A		1	0,21	21	63
	C/G	PASS	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*2567G>C		1	0,24	24	72
	A/G	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*2338T>C		1	0,19	19	57
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*2012G>A		1	0,21	21	63
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*593G>A		1	0,10	10	29
	C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*414G>A		1	0,14	14	42
	C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*322G>A		1	0,18	18	54
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>JMJD6</i>	3_prime_U TR_variant	Other	XM_003997 181.5	XM_003997181.5:c.*40G>A		1	0,24	24	72
	G/A	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>KCNJ6</i>	5_prime_U TR_variant	Other	XM_003991 434.5	XM_003991434.5:c.-327G>A		1	0,19	19	57

D1: 115 459 989 D4: 860 385 3 E2: 740 371 4 E2: 740 375 6 E1: 423 648 89 C1: 113 544 973 C1: 113 582 919 C1: 113 583 215 C1: 113 583 592 A2: 105 359 05 E1: 424 086 41 E2: 355 362 3 C1: 103 362 532 E2: 518 602 66 A1: 583 696 4	G/A	VQSRTrancheSNP99.00to99.90 PASS,VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0 0	1 1	1 1	<i>KCNQ1</i>	3_prime_UTR_variant	Other	XM_011287088.3	XM_011287088.3:c.*154C>T	1	0,18	18	54
	C/T		3	0.003 62319	82 8	1 1	1 1	2 2	<i>KCNV2</i>	synonymous_variant	Other	XM_023243303.1	XM_023243303.1:c.552G>A p.Pro184=	1	0,28	28	84
	C/T	PASS	3	0.003 62319	82 8	1 1	1 1	2 2	<i>KLK6</i>	3_prime_UTR_variant	Other	XM_011289717.3	XM_011289717.3:c.*472C>T	1	0,30	30	93
	C/A	PASS	3	0.003 62319	82 8	1 1	1 1	2 2	<i>KLK6</i>	3_prime_UTR_variant	Other	XM_011289717.3	XM_011289717.3:c.*514C>A	1	0,25	25	78
	G/A	VQSRTrancheSNP99.00to99.90	3	0.003 62319	82 8	1 1	1 1	2 2	<i>KRT14</i>	synonymous_variant	Other	XM_003996860.5	XM_003996860.5:c.84C>T p.Ser28=	1	0,12	12	36
	G/C	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LIMS2</i>	5_prime_UTR_variant	Other	XM_023259204.1	XM_023259204.1:c.-33G>C	1	0,19	19	57
	G/C	VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LIMS2</i>	3_prime_UTR_variant	Other	XM_023259205.1	XM_023259205.1:c.*67G>C	1	0,27	27	81
	C/T	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LIMS2</i>	3_prime_UTR_variant	Other	XM_023259205.1	XM_023259205.1:c.*363C>T	1	0,15	15	45
	C/G	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LIMS2</i>	3_prime_UTR_variant	Other	XM_023259205.1	XM_023259205.1:c.*740C>G	1	0,17	17	51
	G/A	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LOC101083497</i>	synonymous_variant	Other	XM_023246468.1	XM_023246468.1:c.465C>T p.Phe155=	1	0,14	14	42
	C/T	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LOC101088377</i>	synonymous_variant	Other	XM_003996938.3	XM_003996938.3:c.69G>A p.Pro23=	1	0,19	19	56
	C/T	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LOC101093312</i>	synonymous_variant	Other	XM_006940863.3	XM_006940863.3:c.420C>T p.Asn140=	1	0,27	27	81
	C/G	PASS	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LOC101093537</i>	synonymous_variant	Other	XM_006935036.4	XM_006935036.4:c.2778C>G p.Leu926=	1	0,24	24	72
	G/A	VQSRTrancheSNP99.00to99.90	2	0.002 41546	82 8	0 0	1 1	1 1	<i>LOC101096371</i>	synonymous_variant	Other	XM_023246162.1	XM_023246162.1:c.387G>A p.Pro129=	1	0,25	25	75
	G/A	PASS	3	0.003 62319	82 8	1 1	1 1	2 2	<i>LOC101096779</i>	3_prime_UTR_variant	Other	XM_019821051.2	XM_019821051.2:c.*358C>T	1	0,27	27	81

C1: 114 016									<i>LOC10</i> <i>109690</i>	3_prime_U TR_variant	Other	XM_023259 212.1	XM_023259212.1:c.1293+3598A> G					
114 C2: 161 050	A/G	PASS	2	0.002 41546	82 8	0	1	1	8					1	0,14		14	42
757 E3: 101 643									<i>LOC10</i> <i>109965</i>	synonymo us_variant	Other	XM_019811 045.2	XM_019811045.2:c. 2835G>A					
39 D1: 257 908	G/A	PASS	3	0.003 62319	82 8	1	1	2	1				p.Gly945= p.Gly945=	1	0,25		25	75
10 C2: 160 997									<i>LOC10</i> <i>290125</i>	non_codin g_exon_va riant	Other	XR_442127. 3	XR_442127.3:n.456G>A					
930 E1: 374 455	G/A	PASS	2	0.002 41546	82 8	0	1	1	3					1	0,18		18	53
90 A3: 134 145									<i>LOC10</i> <i>290188</i>	non_codin g_exon_va riant	Other	XR_441265. 4	XR_441265.4:n.2097C>T					
471 B2: 129 117	G/A	PASS	2	0.002 41546	82 8	0	1	1	1					1	0,21		21	63
505 B2: 129 118	A/G	VQSRTran cheSNP99. 90to100.00	2	0.002 41546	82 8	0	1	1	8	5_prime_U TR_variant	Other	XM_023260 827.1	XM_023260827.1:c.-50T>C	1	0,2		2	6
101 C1: 593 869	T/C	PASS	3	0.003 62319	82 8	1	1	2	<i>LOC10</i> <i>949433</i>	3_prime_U TR_variant	Other	XR_0027379 81.1, XM_00 3996700.4	XR_002737981.1:n. 6246T>C, XM_0039 96700.4:c.*119A>G	1	0,19		19	57
75 C2: 747 788									<i>LOC10</i> <i>949841</i>	non_codin g_exon_va riant	Other	XR_0027413 23.1	XR_002741323.1:n.2140C>T					
89 C2: 746 820	G/A	PASS	3	0.003 62319	82 8	1	1	2	0					1	0,8		8	24
90 C2: 746 854	A/G	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>LOC10</i> <i>949992</i>	non_codin g_exon_va riant	Other	XR_0021575 11.2	XR_002157511.2:n.75A>G	1	0,29		29	87
68 C2: 746 854	T/G	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>LOC10</i> <i>949992</i>	non_codin g_exon_va riant	Other	XR_0021575 11.2	XR_002157511.2:n.671T>G	1	0,25		25	75
003 65	G/C	PASS	3	0.003 62319	82 8	1	1	2	<i>LOC10</i> <i>950239</i>	non_codin g_exon_va riant	Other	XR_0021609 62.1	XR_002160962.1:n.550G>C	1	0,27		27	81
	C/G	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC10</i> <i>950350</i>	non_codin g_exon_va riant	Other	XR_0027455 73.1	XR_002745573.1:n.427C>G	1	0,19		19	57
	G/A	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC10</i> <i>950350</i>	non_codin g_exon_va riant	Other	XR_0027452 98.1	XR_002745298.1:n.102G>A	1	0,16		16	48
	A/-	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>LOC10</i> <i>950350</i>	non_codin g_exon_va riant	Other	XR_0027452 98.1	XR_002745298.1:n.1494delA	1	0,24		24	72
	AA/-	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC10</i> <i>950350</i>	non_codin g_exon_va riant	Other	XR_0027452 98.1	XR_002745298.1:n.1493_1494del AA	1	0,24		24	72
	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>LOC10</i> <i>950350</i>	non_codin g_exon_va riant	Other	XR_0027452 99.1	XR_002745299.1:n.3095C>T	1	0,19		19	57

B2: 908		VQSRTrancheSNP99.00to99.90	3	0.003	82	1	1	2	<i>LOC11560707</i>	non_coding_exon_variant	Other	XR_002742701.1	XR_002742701.1:n.57A>C	1	0.20	20	60	
266	A/C																	
76																		
C2: 803																		
192	C/T	PASS	3	0.003	82	1	1	2	<i>LOC11562253</i>	non_coding_exon_variant	Other	XR_002745306.1	XR_002745306.1:n.609C>T	1	0.25	25	75	
10																		
C2: 143	-	VQSRTrancheINDEL9.90to100.00	3	0.003	82	1	1	2	<i>LOC11562308</i>	3_prime_UTR_variant	Other	XR_002745474.1	XR_002745474.1:n.2817_2823dupGGGGGGG	1	0.6	6	21	
215	/GGGGGG																	
890	GG																	
D1: 112																		
868	G/A	VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>LRP5</i>	synonymous_variant	Other	XM_023240160.1	XM_023240160.1:c.2694G>A	p.Thr898=	1	0.26	26	78
800																		
C2: 744																		
523	G/A	PASS	3	0.003	82	1	1	2	<i>LRRC15</i>	synonymous_variant	Other	XM_006936178.2	XM_006936178.2:c.1482G>A	p.Pro494=	1	0.24	24	72
86																		
C1: 106																		
788	A/G	PASS	2	0.002	82	0	1	1	<i>MRPL9</i>	synonymous_variant	Other	XM_006935152.4	XM_006935152.4:c.552T>C	p.Asn184=	1	0.20	20	60
800																		
A1: 236		VQSRTrancheINDEL9.90to100.00	3	0.003	82	1	1	2	<i>MTRR</i>	3_prime_UTR_variant	Other	XM_019812938.2	XM_019812938.2:c.*1117_*1118insT		1	0.8	8	24
034	-/A																	
049																		
C1: 150																		
157	G/A	VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>MUL1</i>	3_prime_UTR_variant	Other	XM_003989627.5	XM_003989627.5:c.*17C>T		0.9375	1.15	16	17
02																		
A2: 128																		
830	G/A	VQSRTrancheSNP99.00to99.90	3	0.003	82	1	1	2	<i>MVB12A</i>	synonymous_variant	Other	XM_006928584.3	XM_006928584.3:c.171G>A	p.Ser57=	1	0.22	22	66
69																		
C1: 113																		
610	G/A	VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>MYO7B</i>	synonymous_variant	Other	XM_019837914.2	XM_019837914.2:c.2610C>T	p.Ala870=	1	0.25	25	74
903																		
C1: 113																		
631	G/A	VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>MYO7B</i>	synonymous_variant	Other	XM_019837914.2	XM_019837914.2:c.1222C>T	p.Leu408=	1	0.22	22	66
747																		
B2: 127																		
697	C/T	VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>NHSL1</i>	synonymous_variant	Other	XM_023254444.1	XM_023254444.1:c.3513G>A	p.Pro1171=	1	0.23	23	69
302																		
A3: 135																		
313	G/A	VQSRTrancheSNP99.00to99.90	2	0.002	82	0	1	1	<i>NOL10</i>	3_prime_UTR_variant	Other	XM_003984501.5	XM_003984501.5:c.*63G>A		1	0.28	28	84
065																		
A3: 135																		
313	C/T	PASS	2	0.002	82	0	1	1	<i>NOL10</i>	3_prime_UTR_variant	Other	XM_003984501.5	XM_003984501.5:c.*81C>T		1	0.24	24	72
083																		
C2: 750	GATTAT																	
342	ATTATG	PASS	2	0.002	82	0	1	1	<i>OPA1</i>	3_prime_UTR_variant	Other	XR_002162522.2	XR_002162522.2:n.3286_3299delCTCATAATATAATC		1	0.26	26	78
21	AG/-																	

B4: 220 303 17 C2: 802	C/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>OTUD1</i>	synonymo us_variant	Other	XM_003988 183.5	XM_003988183.5:c. 552C>T	p.Gly184=	1	0.23	23	67
507 57 C2: 802	A/G	PASS	2	0.002 41546	82 8	0	1	1	<i>P3H2</i>	synonymo us_variant	Other	XM_003991 795.3	XM_003991795.3:c. 855A>G	p.Glu285=	1	0.20	20	60
781 84 C2: 802	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>P3H2</i>	3_prime_U TR_variant	Other	XM_003991 795.3	XM_003991795.3:c.*476C>T		1	0.15	15	45
784 06 C1: 106	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>P3H2</i>	3_prime_U TR_variant	Other	XM_003991 795.3	XM_003991795.3:c.*698C>T		1	0.22	22	66
504 469 C1: 113	G/A	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>POGZ</i>	synonymo us_variant	Other	XM_011285 146.3	XM_011285146.3:c. 1302C>T	p.Ser434=	1	0.22	22	66
787 307 C1: 113	C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>PROC</i>	3_prime_U TR_variant	Other	XM_023259 207.1	XM_023259207.1:c.*310G>A		1	0.14	14	42
795 528 B4: 232	C/A	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>PROC</i>	synonymo us_variant	Other	XM_023259 207.1	XM_023259207.1:c. 297G>T	p.Leu99=	1	0.18	18	52
463 14 C1: 106	ACAA/-	PASS	2	0.002 41546	82 8	0	1	1	<i>PRTFD CI</i>	3_prime_U TR_variant	Other	XM_006933 313.4	XM_006933313.4:c.*518_*521del TTGT		1	0.10	10	30
152 630 C1: 100	C/T	PASS	3	0.003 62319	82 8	1	1	2	<i>PRUNE I</i>	synonymo us_variant	Other	XM_003990 600.5	XM_003990600.5:c. 870C>T	p.Asn290 =	1	0.19	19	57
405 365 D1: 468	G/T	VQSRTran cheSNP99. 00to99.90	3	0.003 62319	82 8	1	1	2	<i>PTGFR N</i>	synonymo us_variant	Other	XM_023259 040.1	XM_023259040.1:c. 363G>T	p.Ala121=	1	0.16	16	47
232 30 C1: 144	G/T	PASS	2	0.002 41546	82 8	0	1	1	<i>RAB38</i>	5_prime_U TR_variant	Other	XM_003992 646.4	XM_003992646.4:c.-209G>T		1	0.22	22	66
526 754 C1: 113	C/T	PASS	3	0.003 62319	82 8	1	1	2	<i>RIF1</i>	synonymo us_variant	Other	XR_0027448 01.1	XR_002744801.1:n.2938C>T		1	0.31	31	92
281 402 C1: 984	G/A	PASS	3	0.003 62319	82 8	1	1	2	<i>SAP130</i>	synonymo us_variant	Other	XM_003990 673.4	XM_003990673.4:c. 2493G>A	p.Pro831=	1	0.16	16	48
969 14 C1: 984	T/A	PASS	2	0.002 41546	82 8	0	1	1	<i>SIKE1</i>	splice_regi on_variant	Other	XM_004001 118.5	XM_004001118.5:c.159+8A>T		1	0.11	11	33
969 15 F2:8	G/T	VQSRTran cheSNP99. 00to99.90	2	0.002 41546	82 8	0	1	1	<i>SIKE1 SLC45</i>	splice_regi on_variant	Other	XM_004001 118.5	XM_004001118.5:c.159+7C>A		1	0.11	11	33
297	G/A	PASS	2	0.002 41546	82 8	0	1	1	<i>A4</i>	synonymo us_variant	Other	XM_019823 200.2	XM_019823200.2:c. 2094C>T	p.Arg698 =	1	0.22	22	66

447
0
B4:
117
303
134
B4:
117
303
157
B4:
117
303
189
E1:
581
993
10
E1:
181
053
93
E1:
377
294
63
E3:
953
473
1
A3:
167
852
75
C2:
803
322
63
C2:
803
322
67
C2:
803
323
84
C2:
803
323
87
C2:
803
324
70
C2:
803
330
97
C2:
803
331
82

C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>SOCS2</i>	5_prime_UTR_variant	Other	XM_011284154.3	XM_011284154.3:c.-220+423C>T	1	0,17	17	51
T/C	PASS	2	0.002 41546	82 8	0	1	1	<i>SOCS2</i>	5_prime_UTR_variant	Other	XM_011284154.3	XM_011284154.3:c.-220+446T>C	1	0,17	17	51
C/G	PASS	2	0.002 41546	82 8	0	1	1	<i>SOCS2</i>	5_prime_UTR_variant	Other	XM_011284154.3	XM_011284154.3:c.-220+478C>G	1	0,19	19	56
C/T	PASS	3	0.003 62319	82 8	1	1	2	<i>SPHK1</i>	synonymous_variant	Other	XM_019818292.2	XM_019818292.2:c.1419C>T	1	0,26	26	78
G/T	VQSRTranschedeSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>SUPT6H</i>	synonymous_variant	Other	XM_003996485.5	XM_003996485.5:c.5118C>A	1	0,20	20	60
C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>TAC4</i>	synonymous_variant	Other	XM_006940216.4	XM_006940216.4:c.51C>T	1	0,21	21	63
G/A	PASS	3	0.003 62319	82 8	1	1	2	<i>TMEM120A</i>	splice_region_variant	Other	XM_023246748.1	XM_023246748.1:c.563+7G>A	1	0,15	15	45
C/T	PASS	2	0.002 41546	82 8	0	1	1	<i>TOX2</i>	synonymous_variant	Other	XM_023251217.1	XM_023251217.1:c.333G>A	1	0,29	29	87
G/T	VQSRTranschedeSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*1640C>A	1	0,20	20	60
-/A	PASS	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*1636dupT	1	0,19	19	57
-/TCCCCCT	PASS	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*1519_*1520insAGGGGA	1	0,28	28	90
T/C	PASS	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*1516A>G	1	0,29	29	87
T/C	VQSRTranschedeSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*1433A>G	1	0,16	16	48
T/C	VQSRTranschedeSNP99.00to99.90	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*806A>G	1	0,20	20	60
T/C	PASS	2	0.002 41546	82 8	0	1	1	<i>TP63</i>	3_prime_UTR_variant	Other	XM_003991796.5	XM_003991796.5:c.*721A>G	1	0,22	22	66

C2: 803 338 10 C2: 804 143 97 C2: 211 073 9 C2: 211 120 6 D3: 930 049 57 C1: 113 033 585 C1: 113 510 845 A1: 186 050 8 B2: 221 705 2 B2: 211 569 3	G/- G/A C/A G/A C/T G/A TC/- A/T G/A A/G	PASS VQSRTrancheSNP99.00to99.90 PASS VQSRTrancheSNP99.00to99.90 PASS PASS PASS VQSRTrancheSNP99.00to99.90	2 2 3 3 2 2 2 2	0.002 0.002 0.003 0.003 0.002 0.002 0.002 0.002	82 82 82 82 82 82 82 82	0 0 1 1 0 0 1 0	1 1 1 1 1 1 1 1	1 1 2 2 1 1 1 1	<i>TP63</i> <i>TP63</i> <i>TRPM2</i> <i>TRPM2</i> <i>TSHZ1</i> <i>UGGT1</i> <i>WDR33</i> <i>XPO4</i> <i>ZSCAN9</i> <i>ZSCAN31</i>	3_prime_UTR_variant synonymous_variant 3_prime_UTR_variant 3_prime_UTR_variant synonymous_variant 3_prime_UTR_variant synonymous_variant synonymous_variant	Other Other Other Other Other Other Other Other	XM_003991796.5 XM_003991796.5 XM_011285792.3 XM_011285792.3 XM_023242110.1 XM_023259198.1 XM_003990676.4 XM_003980253.5 XM_019831673.2 XM_006931335.3	XM_003991796.5:c.*93delC XM_003991796.5:c.570C>T XM_011285792.3:c.*749G>T XM_011285792.3:c.*282C>T XM_023242110.1:c.2136C>T XM_023259198.1:c.*108C>T XM_003990676.4:c.*250_*251delCT XM_003980253.5:c.747T>A XM_019831673.2:c.399C>T XM_006931335.3:c.1134A>G	p.Gly190= p.Thr712= p.Leu133= p.Pro378=	1 1 1 1 1 1 1 1 1 1	0.17 0.25 0.18 0.12 0.24 0.18 0.13 0.35 0.14 0.30	17 25 18 12 24 18 13 35 14 30	51 74 53 36 72 54 38 99 42 90
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Table S2. Private heterozygous genetic variants identified uniquely in the case of 414 cats of the 99 Lives Cat Genome Sequencing Consortium dataset including WGS data from 362 cats and whole exome sequencing data from 52 cats. Variant calling was performed with respect to the *Felis_catus_9.0* reference genome assembly. Presented are variants affecting the coding sequence of genes or existing in 10 bp regions flanking exons.

Chr:POS	Ref/Alt	Filter	Allele Frequency	# Heterozygotes	# Homozygotes	# Homozygotes	Gene Names	Sequence Ontology (Combined)	Effect (Combined)	Transcript Name (Clinically Relevant)	HGVS c. (Clinically Relevant)	HGVS p. (Clinically Relevant)	Variant Allele Freq of the Case	Allelic Depths (AD) of the Case	Read Depths (DP) of the Case	Genotype Qualities (GQ) of the Case	
X:1758	GCGTGCT/-	PASS	0.0073	8	2	1	<i>ARSD</i>	frameshift_variant	LoF	XM_023249531.1	XM_023249531.1:c.1398_1404delAGCACGC	p.Ala67Glyfs*15	0.61538	5	10,16	26	99
A2:129	TTTTC/-	PASS	0.0073	8	2	1	<i>DNAJB9</i>	frameshift_variant	LoF	XM_003982962.5	XM_003982962.5:c.530_534delCTTTT	p.Ser177Tyrfs*4	0.52941	2	8,9	17	99
A2:120	-TTTTTTA	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>EVXI</i>	frameshift_variant	LoF	XM_023250457.1	XM_023250457.1:c.726_727insTTTTTTA	p.Thr243Phefs	0.22222	2	14,4	18	99
A2:120	CG/-	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>EVXI</i>	frameshift_variant	LoF	XM_023250457.1	XM_023250457.1:c.728_729delCG	p.Thr243Metfs	0.21052	6	15,4	19	99
A2:120	GCCGC/-	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>EVXI</i>	frameshift_variant	LoF	XM_023250457.1	XM_023250457.1:c.732_736delGCCGC	p.Trp244Ter	0.21052	6	15,4	19	99
A2:120	C/-	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>EVXI</i>	frameshift_variant	LoF	XM_023250457.1	XM_023250457.1:c.744delC	p.Asp249Thrfs*8	0.23529	4	13,4	17	99
A2:120	-A	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>EVXI</i>	frameshift_variant	LoF	XM_023250457.1	XM_023250457.1:c.748_749insA	p.Pro250Hisfs	0.23529	4	13,4	17	99
A3:185	-TTTTTTAA	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>LAMA5</i>	frameshift_variant	LoF	XM_023251034.1	XM_023251034.1:c.1033_1034insTTTTTTAA	p.Trp345Phefs*64	0.21428	6	11,3	14	90
A3:185	-T	VQSRTTrancheINDEL99.9	0.0073	8	2	1	<i>LAMA5</i>	frameshift_variant	LoF	XM_023251034.1	XM_023251034.1:c.1036_1037insT	p.Lys346Ilefs*61	0.23076	9	10,3	13	93

		0to100.0	0															
A3:		VQSRTr																
185		ancheIN																
202		DEL99.9	0.00	8														
5	GC/-	0to100.0	1207	2	1	0	1	LAMA5	frameshift_v	LoF	XM_0232	XM_023251034.1:c.1038_1	p.Lys34	0.23076				
D1:		0	73	8	1	0	1		ariant		51034.1	039delG	6Asnfs*	9	10,3		13	93
875		VQSRTr																
737		ancheSN	0.00	8														
30	G/A	P99.0to	1207	2	1	0	1	LOC10			XM_0198	XM_019812218.2:c.661C>	p.Arg22	0.60869				
E1:		99.90	73	8	1	0	1	108600	stop_gained	LoF	12218.2	T	1Ter	6	9,14		23	99
306		VQSRTr																
563		ancheIN	0.00	8														
35	-/CCGT	DEL99.9	1207	2	1	0	1	LOC10	frameshift_v	LoF	XM_0039	XM_003996638.3:c.845_84	p.Asn28	0.15789				
603		0to100.0	1207	2	1	0	1	109817	ariant		96638.3	6insACGG	3Argfs*	5	16,3		19	76
175		0	73	8	1	0	1	7					28					
74	-/T	PASS	0.00	8	1	0	1	LOC11	frameshift_v	LoF	XM_0232	XM_023254723.1:c.565_56	p.Ala18	0.55	9,11		20	99
E1:		VQSRTr																
113		ancheSN	0.00	8														
295		P99.0to	1207	2	1	0	1	9			XR_00273			0.58823				
12	C/A	99.90	73	8	1	0	1	MIEF2	stop_gained	LoF	8041.1	XR_002738041.1:n.2301G>T		5	7,10		17	99
216		VQSRTr																
815		ancheIN	0.00	8														
13	CA/-	DEL99.9	1207	2	1	0	1	RREB1	frameshift_v	LoF	XM_0112	XM_011282167.3:c.5210_5	p.Val17	0.52173				
B2:		0to100.0	73	8	1	0	1		ariant		82167.3	211delTG	37Alafs	9	11,12		23	99
216		0	73	8	1	0	1											
815	-/TCTT	PASS	0.00	8	1	0	1	RREB1	frameshift_v	LoF	XM_0112	XM_011282167.3:c.5205_5	p.Leu17	0.52173				
19		VQSRTr																
E1:		ancheSN	0.00	8														
621		P99.0to	1207	2	1	0	1	SLC25A	frameshift_v	LoF	XM_0232	XM_023244311.1:c.140_14	p.Leu49	0.5	9,9		18	99
728	-/TGGC	99.90	73	8	1	0	1	10	ariant		44311.1	3dupTGGC	Glyfs*3					
52		PASS	0.00	8	1	0	1						6	0.5	9,9		18	99
C2:		VQSRTr																
160		ancheIN	0.00	8														
701		DEL99.9	1207	2	1	0	1	TOPAZ	frameshift_v	LoF	XM_0232	XM_023260821.1:c.3585_3	p.Val11	0.25	6,2		8	66
824	-/TTTTTTTTTT	0to100.0	73	8	1	0	1	1	ariant		60821.1	586insTTTTTTTTTT	*5					
C1:		0	73	8	1	0	1											
200		VQSRTr																
587		ancheSN	0.00	8														
034	T/C	P99.90to	1207	2	1	0	1	ABCA1	missense_va	Miss	XM_0198	XM_019838638.1:c.6110A>	p.Asn20	0.26315				
E2:		100.00	73	8	1	0	1	2	ariant	ense	38638.1	G	37Ser	8	14,5		19	99
367		VQSRTr																
535		ancheIN	0.00	8														
39	G/A	DEL99.9	1207	2	1	0	1	ADGR	missense_va	Miss	XM_0198	XM_019819797.2:c.1006G>	p.Glu33	0.5	6,6		12	99
E2:		0to100.0	73	8	1	0	1	G3	ariant	ense	19797.2	A	6Lys					
366		0	73	8	1	0	1											
431	G/A	PASS	0.00	8	1	0	1	ADGR	missense_va	Miss	XR_00273	XR_002738471.1:n.537G>A		0.57142				
76		VQSRTr																
C1:		ancheSN	0.00	8														
204		P99.90to	1207	2	1	0	1	ANKZF	missense_va	Miss	XM_0198	XM_019838707.1:c.1883G>	p.Arg62	0.63636				
228	G/A	100.00	73	8	1	0	1	1	ariant	ense	38707.1	A	8Gln	4	8,14		22	99
319		VQSRTr																
B2:		ancheIN	0.00	8														
421		DEL99.9	1207	2	1	0	1	APOBE	inframe_dele	Miss	XM_0039	XM_003986124.5:c.30_38d	p.Ala13	0.69230				
077	GGCCGCTGC/-	99.90	73	8	1	0	1	C2	tion	ense	86124.5	eICGCTGCGGC	_Ala15d	8	4,9		13	99
35		0to100.0	73	8	1	0	1						el					

A2: 916 294 1 B1: 114 930 572 C1: 206 221 767 A2: 120 655 694 A2: 120 655 707 A2: 120 655 709 D1: 107 814 795 A2: 919 859 2 D4: 919 856 A3: 574 946 53 D4: 753 562 50 A3: 401 121 10 C1: 200 911 040 B1: 685 376 56 C2: 118 325 93 C1: 204	C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>DNASE2</i>	missense_variant	Missense	XM_003981919.4	XM_003981919.4:c.407G>A	p.Arg136His	0.48	13,12	25	99
G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>ENPEP</i>	missense_variant	Missense	XM_003985081.5	XM_003985081.5:c.2240C>T	p.Ala747Val	0.47619	11,10	21	99	
C/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>EPHA4</i>	missense_variant	Missense	XR_002161778.1	XR_002161778.1:n.1915G>T		0.272727	16,6	22	99	
C/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>EVX1</i>	missense_variant	Missense	XM_023250457.1	XM_023250457.1:c.739C>A	p.Pro247Thr	0.235294	13,4	17	99	
C/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>EVX1</i>	missense_variant	Missense	XM_023250457.1	XM_023250457.1:c.752C>A	p.Ala251Asp	0.266667	11,4	15	99	
T/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>EVX1</i>	missense_variant	Missense	XM_023250457.1	XM_023250457.1:c.754T>A	p.Phe252Ile	0.25	12,4	16	99	
G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FADS1</i>	missense_variant	Missense	XM_003993434.5	XM_003993434.5:c.923C>T	p.Pro308Leu	0.529412	8,9	17	99	
G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FARSA</i>	missense_variant	Missense	XM_019818179.2	XM_019818179.2:c.1208C>T	p.Thr403Met	0.73913	6,17	23	99	
G/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FGD3</i>	missense_variant	Missense	XM_003995335.5	XM_003995335.5:c.317G>C	p.Gly106Ala	0.35	13,7	20	99	
C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FHL2</i>	missense_variant	Missense	XM_003983876.5	XM_003983876.5:c.209C>T	p.Ser70Leu	0.6	10,15	25	99	
C/G	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FKBP15</i>	missense_variant	Missense	XM_003995763.5	XM_003995763.5:c.154G>C	p.Gly52Arg	0.545455	10,12	22	99	
C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FLRT3, MACROD2</i>	missense_variant	Missense	XM_006929960.4, XM_023251484.1	XM_006929960.4:c.1331G>A, XM_023251484.1:c.272-163142C>T	p.Gly44Asp,	0.375	15,9	24	99	
G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FNI</i>	missense_variant	Missense	XM_003991118.4	XM_003991118.4:c.5474C>T	p.Thr1825Ile	0.590909	9,13	22	99	
A/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>FSTL5</i>	missense_variant	Missense	XM_023252713.1	XM_023252713.1:c.415A>T	p.Asn139Tyr	0.6	10,15	25	99	
A/G	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>GART</i>	missense_variant	Missense	XM_003991457.5	XM_003991457.5:c.1189A>G	p.Ile397Val	0.590909	9,13	22	99	
C/G	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>GLB1L</i>	missense_variant	Missense	XM_003991175.5	XM_003991175.5:c.571G>C	p.Ala191Pro	0.4	12,8	20	99	

232																			
351																			
B1:		VQSRTr																	
130		ancheSN		0.00	8														
318		P99.00to		1207	2														
500	G/A	99.90	1	73	8	1	0	1	GRID2	missense_v	Miss	XM_0198	XM_019829242.1:c.337C>	p.His11					
D1:																			
893				0.00	8														
512				1207	2														
89	C/T	PASS	1	73	8	1	0	1	HIPK3	missense_v	Miss	XM_0198	XM_019812241.2:c.266C>	p.Ala89	0.41666				
D2:																			
323				0.00	8														
449				1207	2														
06	C/A	PASS	1	73	8	1	0	1	HKDC1	missense_v	Miss	XM_0039	XM_003994002.4:c.638C>	p.Thr21					
E2:		VQSRTr																	
339		ancheSN		0.00	8														
642		P99.00to		1207	2														
43	C/T	99.90	1	73	8	1	0	1	IRX3	missense_v	Miss	XM_0232	XM_023245118.1:c.475G>	p.Gly15					
A2:		VQSRTr																	
850		ancheSN		0.00	8														
158		P99.00to		1207	2														
8	C/T	99.90	1	73	8	1	0	1	KANK2	missense_v	Miss	XM_0069	XM_006928415.4:c.1229G>	p.Arg41	0.46428				
E3:																			
225				0.00	8														
590				1207	2				KIAA05	missense_v	Miss	XM_0069	XM_006942221.3:c.4609G>	p.Asp15					
85	C/T	PASS	1	73	8	1	0	1	56	riant	ense	42221.3	A	37Asn		0.4	9,6		15
E2:																			
368				0.00	8														
478				1207	2														
00	C/T	PASS	1	73	8	1	0	1	KIFC3	missense_v	Miss	XM_0232	XM_023245836.1:c.484G>	p.Ala16					
A3:		VQSRTr																	
185		ancheIN		0.00	8														
201		DEL99.9		1207	2														
3	GCAGCC/-	0to100.0	1	73	8	1	0	1	LAMA5	inframe_dele	Miss	XM_0232	XM_023251034.1:c.1027_1	p.Gln34					
A3:		VQSRTr																	
185		ancheSN		0.00	8														
202		P99.90to		1207	2														
1	G/T	100.00	1	73	8	1	0	1	LAMA5	missense_v	Miss	XM_0232	XM_023251034.1:c.1034G>	p.Trp34	0.21428				
D1:																			
108				0.00	8														
925				1207	2														
605	G/A	PASS	1	73	8	1	0	1	LGALS12	missense_v	Miss	XR_00273	XR_002736394.1:n.1113G>A						
D1:																			
871				0.00	8														
529				1207	2				LOC10	missense_v	Miss	XM_0198	XM_019812218.2:c.5329C>	p.Arg17	0.57142				
35	G/A	PASS	1	73	8	1	0	1	1086002	riant	ense	12218.2	T	77Cys		9	6,8		14
D1:																			
102				0.00	8														
461				1207	2				LOC10	missense_v	Miss	XM_0039	XM_003993298.4:c.398T>	p.Ile133	0.55555				
643	T/C	PASS	1	73	8	1	0	1	1089895	riant	ense	93298.4	C	Thr		6	8,10		18
C1:																			
194				0.00	8														
526				1207	2				LOC10	missense_v	Miss	XM_0039							
654	T/C	PASS	1	73	8	1	0	1	1093129	riant	ense	91080.3	XM_003991080.3:c.50A>G	p.Tyr17					
B2:																			
108				0.00	8														
948				1207	2				LOC10	missense_v	Miss	XM_0232	XM_023254289.1:c.433C>	p.Leu14					
452	C/T	PASS	1	73	8	1	0	1	1093938	riant	ense	54289.1	T	5Phe		0.6	6,9		15
E1:		VQSRTr																	
306	G/T	ancheSN	1	73	8	1	0	1	LOC101098177	missense_v	Miss	XM_0039	XM_003996638.3:c.844C>	p.Leu28	0.11111				

563		P99.90to																	
36		100.00																	
B2:																			
571			0.00	8															
894			1207	2															
12	G/A	PASS	1	73	8	1	0	1	4	missense_v	Miss	XM_0232	XM_023254076.1:c.816G>	p.Met27	0.52777	8	17,19	36	99
E2:																			
358			0.00	8															
853			1207	2															
72	G/C	PASS	1	73	8	1	0	1	41	missense_v	Miss	XM_0232	XM_023246150.1:c.17G>C,	p.Arg6P	0.55555	6	12,15	27	99
A2:		VQSRTr																	
368		ancheSN																	
902		P99.90to																	
23	A/C	100.00	1	73	8	1	0	1	5	missense_v	Miss	XR_00274	XR_002740338.1:n.766A>C		0.11965	8	206,28	234	99
B2:																			
603			0.00	8															
175			1207	2															
74	C/A	PASS	1	73	8	1	0	1	9	missense_v	Miss	XM_0232	XM_023254723.1:c.565G>	p.Ala18	0.52631	6	9,10	19	99
E3:																			
319			0.00	8															
114			1207	2															
69	G/A	PASS	1	73	8	1	0	1	MARF1	missense_v	Miss	XM_0232	XM_023247059.1:c.2803G>	p.Gly93	0.46666	7	8,7	15	99
C1:																			
186			0.00	8															
523			1207	2															
332	C/G	PASS	1	73	8	1	0	1	MARS2,	missense_v	Miss	XM_0039	XM_003990998.5:c.617C>	p.Thr20	0.66666	7	9,18	27	99
A3:		VQSRTr																	
103		ancheSN																	
281		P99.00to																	
004	T/C	99.90	1	73	8	1	0	1	5	missense_v	Miss	XR_43997	XR_439970.4:n.332T>C		0.45454	5	6,5	11	99
B1:		VQSRTr																	
124		ancheSN																	
459		P99.00to																	
394	C/T	99.90	1	73	8	1	0	1	MTTP	missense_v	Miss	XM_0198	XM_019829170.2:c.625G>	p.Gly20	0.55172	4	13,16	29	99
B1:																			
174			0.00	8															
314			1207	2															
632	T/C	PASS	1	73	8	1	0	1	N4BP2	missense_v	Miss	XR_00215	XR_002156413.2:n.2445A>G		0.45	11,9	20	99	
B3:																			
767			0.00	8															
766			1207	2															
86	G/A	PASS	1	73	8	1	0	1	NOP9	missense_v	Miss	XM_0039	XM_003987519.4:c.452G>	p.Arg15	0.40625	19,13	32	99	
B2:																			
142			0.00	8															
818			1207	2															
834	C/T	PASS	1	73	8	1	0	1	NOX3	missense_v	Miss	XM_0232	XM_023254521.1:c.964G>	p.Val32	0.43478	3	13,10	23	99
A3:		VQSRTr																	
278		ancheSN																	
838		P99.00to																	
64	C/T	99.90	1	73	8	1	0	1	NRSN2	missense_v	Miss	XM_0039	XM_003983669.3:c.233C>	p.Ala78	0.73684	2	5,14	19	99
A3:																			
128			0.00	8															
790			1207	2															
460	G/A	PASS	1	73	8	1	0	1	NT5C1	missense_v	Miss	XM_0039	XM_003984482.5:c.872G>	p.Arg29	0.29411	8	12,5	17	99
A1:																			
120			0.00	8															
031			1207	2															
912	C/G	PASS	1	73	8	1	0	1	PCDH1	missense_v	Miss	XM_0039	XM_003980868.4:c.154G>	p.Glu52	0.46153	8	14,12	26	99
B4:																			
536	A/C	PASS	1	73	8	1	0	1	PIK3C2	missense_v	Miss	XM_0069	XM_006933551.3:c.590A>	p.Asn19	0.33333	3	12,6	18	99

936																			
30																			
B4: 144		VQSRTr		0.00	8														
058		ancheSN		1207	2														
679	C/T	P99.00to 99.90	1	73	8	1	0	1	<i>PLXNB</i> 2	missense_v ariant	Miss ense	XM_0198 35766.1	XM_019835766.1:c.40G>A	p.Val14 Met	0.5625	7,9	16	99	
A2: 763		VQSRTr		0.00	8														
141		ancheSN		1207	2														
50	G/A	P99.90to 100.00	1	73	8	1	0	1	<i>POUF6</i> 2	missense_v ariant	Miss ense	XM_0232 50204.1	XM_023250204.1:c.646C> T	p.Pro21 6Ser	0.44444	4	5,4	9	99
A2: 555		VQSRTr		0.00	8														
733		ancheSN		1207	2														
8	C/A	P99.00to 99.90	1	73	8	1	0	1	<i>PRAMI</i>	missense_v ariant	Miss ense	XR_00214 8534.2	XR_002148534.2:n.4808C>A		0.5	10,10	20	99	
D1: 107				0.00	8														
960				1207	2														
526	G/A	PASS	1	73	8	1	0	1	<i>RAB3IL</i> 1	missense_v ariant	Miss ense	XM_0069 37395.3	XM_006937395.3:c.632C> T	p.Thr21 1Met	0.59259	3	11,16	27	99
D4: 915				0.00	8														
651				1207	2														
48	C/T	PASS	1	73	8	1	0	1	<i>RAPGE</i> F1	missense_v ariant	Miss ense	XM_0112 88359.3	XM_011288359.3:c.2515G> A	p.Gly83 9Ser	0.56	11,14	25	99	
B1: 165		VQSRTr		0.00	8														
621		ancheSN		1207	2														
779	C/A	P99.00to 99.90	1	73	8	1	0	1	<i>RASL11</i> B	missense_v ariant	Miss ense	XM_0039 85387.5	XM_003985387.5:c.439G> T	p.Val14 7Leu	0.42105	3	22,16	38	99
B2: 216				0.00	8														
815				1207	2														
11	T/G	PASS	1	73	8	1	0	1	<i>RREB1</i>	missense_v ariant	Miss ense	XM_0112 82167.3	XM_011282167.3:c.5213A> C	p.Gln17 38Pro	0.57142	9	9,12	21	99
B2: 216				0.00	8														
815				1207	2														
17	A/G	PASS	1	73	8	1	0	1	<i>RREB1</i>	missense_v ariant	Miss ense	XM_0112 82167.3	XM_011282167.3:c.5207T> C	p.Leu17 36Pro	0.52173	9	11,12	23	99
A1: 247				0.00	8														
427				1207	2														
15	G/A	PASS	1	73	8	1	0	1	<i>RUBCN</i> L	missense_v ariant	Miss ense	XM_0069 27291.4	XM_006927291.4:c.529G> A	p.Gly17 7Ser	0.57142	9	9,12	21	99
B3: 712		VQSRTr		0.00	8														
907		ancheSN		1207	2														
62	G/A	P99.00to 99.90	1	73	8	1	0	1	<i>RYR3</i>	missense_v ariant	Miss ense	XR_00274 2989.1	XR_002742989.1:n.5150G>A		0.36	16,9	25	99	
D4: 940		VQSRTr		0.00	8														
244		ancheSN		1207	2														
70	G/A	P99.00to 99.90	1	73	8	1	0	1	<i>SARDH</i>	missense_v ariant	Miss ense	XM_0198 16743.2	XM_019816743.2:c.2359G> A	p.Gly78 7Ser	0.39130	4	14,9	23	99
B3: 139				0.00	8														
663				1207	2														
555	C/T	PASS	1	73	8	1	0	1	<i>SERPIN</i> A11	_start_codon _gain_varian t	Miss ense	XM_0069 33077.2	XM_006933077.2:c.- 62G>A	p.Met1e xt-20	0.57142	9	9,12	21	99
B2: 147		VQSRTr		0.00	8														
049		ancheSN		1207	2														
241	C/T	P99.00to 99.90	1	73	8	1	0	1	<i>SLC22A</i> 1	missense_v ariant	Miss ense	XM_0039 86693.5	XM_003986693.5:c.872C> T	p.Ser29 1Leu	0.41666	7	14,10	24	99
D1: 110				0.00	8														
406				1207	2														
583	A/G	PASS	1	73	8	1	0	1	<i>SLC22A</i> 20P	missense_v ariant	Miss ense	XM_0039 93589.3	XM_003993589.3:c.1580A> G	p.Gln52 7Arg	0.38095	2	13,8	21	99
E1: 591				0.00	8														
718	CCCCGGCA/-	PASS	1	73	8	1	0	1	<i>SPNS2</i>	inframe_dele tion	Miss ense	XR_00273 8273.1	XR_002738273.1:n.313_321delACCCC CGGC		0.33333	3	6,3	9	99

A2: 648		VQSRT ancheSN		0.00	8														
381		P99.00to		1207	2														
86	C/A	99.90	1	73	8	1	0	1	<i>ABCA1</i>	synonymous _variant	Othe r	XM_0232 41981.1	XM_023241981.1:c.14145C >A	p.Ile471 5=	0.44444	4	10,8	18	99
E2: 366		VQSRT ancheSN		0.00	8														
542		P99.00to		1207	2														
10	C/T	99.90	1	73	8	1	0	1	<i>ADGR</i> <i>G5.LO</i> <i>C11155</i>	synonymous _variant	Othe r	XM_0069 41542.2.X R_002738 472.1	XM_006941542.2:c.1461C> T,XR_002738472.1:n.107+1 418G>A	p.Tyr48 7=,	0.5	13,13	26	99	
D4: 878		VQSRT ancheSN		0.00	8														
479		P99.00to		1207	2														
77	G/A	99.90	1	73	8	1	0	1	<i>ANGPT</i> <i>L2,RAL</i> <i>GPS1</i>	synonymous _variant	Othe r	XM_0069 39476.4.X M_019816 547.2	XM_006939476.4:c.339C> T,XM_019816547.2:c.610+ 37902G>A	p.Thr11 3=,	0.5	10,10	20	99	
A1: 806		VQSRT ancheSN		0.00	8														
990		P99.00to		1207	2														
88	G/A	99.90	1	73	8	1	0	1	<i>ANKRD</i> <i>10</i>	synonymous _variant	Othe r	XM_0232 53212.1	XM_023253212.1:c.1152C> T	p.Ala38 4=	0.76470	6	4,13	17	73
D4: 119		VQSRT ancheSN		0.00	8														
998		P99.00to		1207	2														
85	A/T	99.90	1	73	8	1	0	1	<i>APBA1</i>	3_prime_UT R_variant	Othe r	XM_0232 43333.1	XM_023243333.1:c.*183T>A		0.55555	6	8,10	18	99
F2: 197				0.00	8														
313				1207	2														
39	T/C	PASS	1	73	8	1	0	1	<i>ARFGE</i> <i>F1</i>	synonymous _variant	Othe r	XM_0112 91364.3	XM_011291364.3:c.4752A> G	p.Arg15 84=	0.41176	5	10,7	17	99
D2: 176		VQSRT ancheSN		0.00	8														
397		P99.90to		1207	2														
09	G/C	100.00	1	73	8	1	0	1	<i>ARV1</i>	3_prime_UT R_variant	Othe r	XM_0232 40424.1	XM_023240424.1:c.*835C>G		0.16363	6	46,9	55	91
D2: 176				0.00	8														
398				1207	2														
91	G/A	PASS	1	73	8	1	0	1	<i>ARV1</i>	3_prime_UT R_variant	Othe r	XM_0232 40424.1	XM_023240424.1:c.*653C>T		0.19642	9	45,11	56	99
D2: 176		VQSRT ancheSN		0.00	8														
402		P99.90to		1207	2														
52	C/T	100.00	1	73	8	1	0	1	<i>ARV1</i>	3_prime_UT R_variant	Othe r	XM_0232 40424.1	XM_023240424.1:c.*292G>A		0.16666	7	30,6	36	74
C1: 220		VQSRT ancheSN		0.00	8														
138		P99.00to		1207	2														
332	T/A	99.90	1	73	8	1	0	1	<i>ASB1</i>	5_prime_UT R_variant	Othe r	XM_0232 59934.1	XM_023259934.1:c.244+66T>A		0.5	7,7	14	99	
C1: 999		VQSRT ancheIN		0.00	8														
207		DEL99.0		1207	2														
69	AG/-	0to99.90	1	73	8	1	0	1	<i>ATP1A</i> <i>1</i>	splice_regio n_variant	Othe r	XM_0112 85086.3	XM_011285086.3:c.1216+3_1216+4del AG		0.4375	9,7	16	99	
A2: 515				0.00	8														
086				1207	2														
13	G/A	PASS	1	73	8	1	0	1	<i>ATP2B</i> <i>2</i>	synonymous _variant	Othe r	XM_0232 50008.1	XM_023250008.1:c.2820C> T	p.Tyr94 0=	0.33333	3	8,4	12	99
B3: 249				0.00	8														
966				1207	2														
86	C/T	PASS	1	73	8	1	0	1	<i>ATP10</i> <i>A</i>	synonymous _variant	Othe r	XM_0232 55028.1	XM_023255028.1:c.3438G> A	p.Gln11 46=	0.36363	6	14,8	22	99
B3: 250				0.00	8														
111				1207	2														
30	G/A	PASS	1	73	8	1	0	1	<i>ATP10</i> <i>A</i>	synonymous _variant	Othe r	XM_0232 55028.1	XM_023255028.1:c.2485C> T	p.Leu82 9=	0.6	8,12	20	99	
B3: 250				0.00	8														
195				1207	2														
21	G/A	PASS	1	73	8	1	0	1	<i>ATP10</i> <i>A</i>	synonymous _variant	Othe r	XM_0232 55028.1	XM_023255028.1:c.1920C> T	p.Thr64 0=	0.48148	1	14,13	27	99

B3: 250 309				0.00	8														
04 B3: 250 314 10	C/T	PASS	1	73	8	1	0	1	<i>ATP10</i>	synonymous	Othe	XM_0232	XM_023255028.1:c.948G>	p.Val31	0.47058	8	9,8	17	99
B3: 250 314 10		VQSRT ancheSN		0.00	8				<i>ATP10</i>	synonymous	Othe	XM_0232	XM_023255028.1:c.834G>	p.Thr27	0.57692	3	11,15	26	99
10 B3: 250 971	C/T	P99.00to	1	73	8	1	0	1	<i>ATP10</i>	synonymous	Othe	XM_0232	XM_023255028.1:c.834G>	p.Thr27	0.57692	3	11,15	26	99
250 971		99.90		0.00	8				<i>ATP10</i>	synonymous	Othe	XM_0232	XM_023255028.1:c.660C>	p.Gly22	0.61111	1	7,11	18	99
24 C2: 102	G/A	PASS	1	73	8	1	0	1	<i>ATP10</i>	synonymous	Othe	XM_0232	XM_023255028.1:c.660C>	p.Gly22	0.61111	1	7,11	18	99
088 182 B4: 133 006				0.00	8				<i>BCHE,</i>			NM_001009364.1:c.367C>							
097 D2: 178				0.00	8				<i>LOC10</i>			NM_001009364.1:c.367C>							
525 99 C1: 181 255 189 C1: 194	C/T	PASS	1	73	8	1	0	1	<i>949140</i>	synonymous	Othe	R_002745	NM_001009364.1:c.367C>	p.Leu12	0.39130	4	14,9	23	99
166 A2: 325 203 0				0.00	8				<i>3</i>			R_002745	NM_001009364.1:c.367C>	p.Leu12	0.39130	4	14,9	23	99
0 C1: 185 632 533 D4: 619 090 04 B4: 965 655 47	-/A	PASS	1	73	8	1	0	1	<i>CACNG</i>	5_prime_UT	Othe	XM_0112	XM_011284199.3:c.-76dupT		0.73913	6,17	23	60	
188 796 870 D2: 647 315 38 D1: 108 617 270				0.00	8				<i>2</i>			XM_0112	XM_011284199.3:c.-76dupT		0.73913	6,17	23	60	
971				0.00	8				<i>CAPN9</i>	splice regio	Othe	XM_0039	XM_003993929.4:c.1482-3C>T		0.63157	9	7,12	19	99
250 971	G/A	PASS	1	73	8	1	0	1	<i>CAPN9</i>	splice regio	Othe	XM_0039	XM_003993929.4:c.1482-3C>T		0.63157	9	7,12	19	99
24 C2: 102				0.00	8				<i>CAVIN</i>	synonymous	Othe	XM_0039	XM_003990983.5:c.1263G>	p.Val42	0.34782	6	15,8	23	99
088 182 B4: 133 006	C/T	P99.90to	1	73	8	1	0	1	<i>2</i>	synonymous	Othe	XM_0039	XM_003990983.5:c.1263G>	p.Val42	0.34782	6	15,8	23	99
097 D2: 178				0.00	8				<i>CCIH2</i>	5_prime_UT	Othe	XM_0198	XM_019838554.2:c.-72G>A		0.4	15,10	25	99	
525 99 C1: 181 255 189 C1: 194	C/T	PASS	1	73	8	1	0	1	<i>orf80</i>	5_prime_UT	Othe	XM_0198	XM_019838554.2:c.-72G>A		0.4	15,10	25	99	
166 A2: 325 203 0				0.00	8				<i>CCDC9</i>	synonymous	Othe	XM_0232	XM_023243963.1:c.195C>	p.Asn65	0.61538	5	10,16	26	99
0 C1: 185 632 533 D4: 619 090 04 B4: 965 655 47	C/T	PASS	1	73	8	1	0	1	<i>4</i>	synonymous	Othe	XM_0232	XM_023243963.1:c.195C>	p.Asn65	0.61538	5	10,16	26	99
188 796 870 D2: 647 315 38 D1: 108 617 270				0.00	8				<i>CCDC1</i>	splice regio	Othe	XR_00216	XR_002161646.1:n.2207-4T>C		0.48	13,12	25	99	
971	T/C	PASS	1	73	8	1	0	1	<i>50</i>	splice regio	Othe	XR_00216	XR_002161646.1:n.2207-4T>C		0.48	13,12	25	99	
250 971				0.00	8				<i>CCDC1</i>	non_coding_	Othe	XR_00273	XR_002737518.1:n.3234C>T		0.48148	1	14,13	27	99
24 C2: 102				0.00	8				<i>80</i>	non_coding_	Othe	XR_00273	XR_002737518.1:n.3234C>T		0.48148	1	14,13	27	99
088 182 B4: 133 006	C/T	P99.00to	1	73	8	1	0	1	<i>CCDC1</i>	non_coding_	Othe	XR_00273	XR_002737518.1:n.3234C>T		0.48148	1	14,13	27	99
097 D2: 178				0.00	8				<i>3</i>			XR_00273	XR_002737518.1:n.3234C>T		0.48148	1	14,13	27	99
525 99 C1: 181 255 189 C1: 194	G/A	PASS	1	73	8	1	0	1	<i>CCT2</i>	3_prime_UT	Othe	XM_0039	XM_003989035.4:c.*87G>A		0.48	13,12	25	99	
166 A2: 325 203 0				0.00	8				<i>R_variant</i>			XM_0039	XM_003989035.4:c.*87G>A		0.48	13,12	25	99	
0 C1: 185 632 533 D4: 619 090 04 B4: 965 655 47				0.00	8				<i>2</i>			XM_0039	XM_003989035.4:c.*87G>A		0.48	13,12	25	99	
188 796 870 D2: 647 315 38 D1: 108 617 270				0.00	8				<i>synonymous</i>			XM_0198	XM_019838488.2:c.735T>	p.Gly24	0.44444	4	15,12	27	99
971	T/A	P99.00to	1	73	8	1	0	1	<i>CDK15</i>	synonymous	Othe	XM_0198	XM_019838488.2:c.735T>	p.Gly24	0.44444	4	15,12	27	99
250 971				0.00	8				<i>R_variant</i>			XM_0198	XM_019838488.2:c.735T>	p.Gly24	0.44444	4	15,12	27	99
24 C2: 102				0.00	8				<i>3</i>			XR_00273	XR_002736911.1:n.2810A>T		0.44444	4	10,8	18	99
088 182 B4: 133 006	A/T	PASS	1	73	8	1	0	1	<i>8</i>	3_prime_UT	Othe	XR_00273	XR_002736911.1:n.2810A>T		0.44444	4	10,8	18	99
097 D2: 178				0.00	8				<i>R_variant</i>			XR_00273	XR_002736911.1:n.2810A>T		0.44444	4	10,8	18	99
525 99 C1: 181 255 189 C1: 194				0.00	8				<i>5</i>			XM_0069	XM_006937437.4:c.-9C>A		0.6875	5,11	16	99	
166 A2: 325 203 0				0.00	8				<i>R_variant</i>			XM_0069	XM_006937437.4:c.-9C>A		0.6875	5,11	16	99	
0 C1: 185 632 533 D4: 619 090 04 B4: 965 655 47	G/T	P99.00to	1	73	8	1	0	1	<i>CHRM1</i>	5_prime_UT	Othe	XM_0069	XM_006937437.4:c.-9C>A		0.6875	5,11	16	99	

B1: 610			0.00	8														
093			1207	2														
46	A/G	PASS	1	73	8	1	0	1	<i>CLCN3</i>	synonymous _variant	Othe r	XM_0039 84867.5	XM_003984867.5:c.159T> C	p.Val53 =	0.5	13,13	26	99
D2: 590		VQSRTr ancheSN P99.00to 99.90		0.00	8													
369	C/T		1	1207	2	1	0	1	<i>CRTAC</i> <i>I</i>	synonymous _variant	Othe r	XM_0198 13654.2	XM_019813654.2:c.864G> A	p.Pro28 8=	0.46153 8	7,6	13	99
11				0.00	8													
C2: 117			1	1207	2	1	0	1	<i>CRYZL</i> <i>I</i>	3_prime_UT R_variant	Othe r	XM_0039 91454.5	XM_003991454.5:c.*26G>C		0.66666 7	10,20	30	99
712	G/C	PASS	1	73	8	1	0	1										
B1: 141				0.00	8													
593	A/C	PASS	1	1207	2	1	0	1	<i>DCTD</i>	3_prime_UT R_variant	Othe r	XM_0232 52314.1	XM_023252314.1:c.*268T>G		0.61904 8	8,13	21	99
53				0.00	8													
B3: 767			1	1207	2	1	0	1	<i>DHRS1</i>	synonymous _variant	Othe r	XM_0039 87518.5	XM_003987518.5:c.885G> A	p.Leu29 5=	0.34782 6	15,8	23	99
703	C/T	PASS	1	73	8	1	0	1										
D3: 537		VQSRTr ancheSN P99.00to 99.90		0.00	8													
244	G/A		1	1207	2	1	0	1	<i>DHX37</i>	synonymous _variant	Othe r	XM_0232 41282.1	XM_023241282.1:c.1170G> A	p.Ala39 0=	0.75	2,6	8	40
7				0.00	8													
A2: 851			1	1207	2	1	0	1	<i>DOCK6</i>	synonymous _variant	Othe r	XM_0232 45498.1	XM_023245498.1:c.4626C> T	p.Thr15 42=	0.42857 1	8,6	14	99
587	G/A	PASS	1	73	8	1	0	1										
B1: 115		VQSRTr ancheSN P99.00to 99.90		0.00	8													
461	T/C		1	1207	2	1	0	1	<i>EGF</i>	synonymous _variant	Othe r	NM_0010 09381.1	NM_001009381.1:c.423A> G	p.Thr14 1=	0.24	19,6	25	99
553				0.00	8													
A3: 760		VQSRTr ancheSN P99.00to 99.90		0.00	8													
888	C/T		1	1207	2	1	0	1	<i>EML6</i>	synonymous _variant	Othe r	XR_00274 1131.1	XR_002741131.1:n.4908C>T		0.5	13,13	26	99
30				0.00	8													
A2: 862			1	1207	2	1	0	1	<i>EPOR</i>	synonymous _variant	Othe r	XM_0232 45578.1	XM_023245578.1:c.63G>T	p.Gly21 =	0.46153 8	7,6	13	99
320	C/A	PASS	1	73	8	1	0	1										
8				0.00	8													
A1: 178			1	1207	2	1	0	1	<i>ERGIC</i> <i>I</i>	synonymous _variant	Othe r	XM_0232 59579.1	XM_023259579.1:c.1111C> T	p.Leu37 =	0.55555 6	8,10	18	99
981	G/A	PASS	1	73	8	1	0	1										
295				0.00	8													
A2: 120			1	1207	2	1	0	1	<i>EVX1</i>	synonymous _variant	Othe r	XM_0232 50457.1	XM_023250457.1:c.738C> T	p.His24 6=	0.23529 4	13,4	17	99
655	C/T	PASS	1	73	8	1	0	1										
693				0.00	8													
A2: 120			1	1207	2	1	0	1	<i>EVX1</i>	synonymous _variant	Othe r	XM_0232 50457.1	XM_023250457.1:c.753C> G	p.Ala25 1=	0.26666 7	11,4	15	99
655	C/G	PASS	1	73	8	1	0	1										
708				0.00	8													
D2: 176			1	1207	2	1	0	1	<i>FAM89</i> <i>A</i>	3_prime_UT R_variant	Othe r	XM_0232 40417.1	XM_023240417.1:c.*281G>A		0.2	52,13	65	99
276	G/A	PASS	1	73	8	1	0	1										
08				0.00	8													
D2: 176		VQSRTr ancheSN P99.90to 100.00		0.00	8													
276	C/T		1	1207	2	1	0	1	<i>FAM89</i> <i>A</i>	3_prime_UT R_variant	Othe r	XM_0232 40417.1	XM_023240417.1:c.*368C>T		0.14814 8	46,8	54	99
95				0.00	8													

B3: 334				0.00	8														
052				1207	2														
92	TTC/-	PASS	1	73	8	1	0	1	<i>FAM21 9B</i>	3_prime_UT R_variant	Othe r	XM_0039 86942.5	XM_003986942.5:c.*1291_*1294delCT TT		0.54166 7	11,13	24	99	
D4: 946				0.00	8														
542	G/A	PASS	1	73	8	1	0	1	<i>FGD3</i>	3_prime_UT R_variant	Othe r	XM_0039 95335.5	XM_003995335.5:c.*537G>A		0.69565 2	7,16	23	99	
C2: 118				0.00	8														
445				1207	2														
74	C/T	PASS	1	73	8	1	0	1	<i>GART</i>	3_prime_UT R_variant	Othe r	XM_0039 91457.5	XM_003991457.5:c.*13C>T		0.47368 4	10,9	19	99	
E1: 635				0.00	8														
181	T/C	PASS	1	73	8	1	0	1	<i>GGT6</i>	3_prime_UT R_variant	Othe r	XM_0198 17018.2	XM_019817018.2:c.*238A>G		0.64	9,16	25	99	
B1: 433				0.00	8														
469				1207	2														
61	A/G	PASS	1	73	8	1	0	1	<i>GPAT4</i>	5_prime_UT R_variant	Othe r	XM_0039 84761.5	XM_003984761.5:c.*60T>C		0.55	9,11	20	99	
E2: 200				0.00	8														
324				1207	2				<i>GPATC</i>	synonymous _variant	Othe r	XM_0039 97950.5	XM_003997950.5:c.1218A> T	p.Pro40 6=	0.60869 6	9,14	23	99	
78	T/A	PASS	1	73	8	1	0	1	<i>H1</i>										
D2: 754		VQSRTr ancheSN		0.00	8														
626		P99.00to 99.90	1	1207	2	1	0	1	<i>HSPA1 2A</i>	3_prime_UT R_variant	Othe r	XM_0232 40980.1	XM_023240980.1:c.*14C>T		0.64705 9	6,11	17	99	
50	G/A			0.00	8														
B3: 173		VQSRTr ancheSN		0.00	8														
017		P99.00to 99.90	1	1207	2	1	0	1	<i>IGF1R</i>	synonymous _variant	Othe r	XM_0232 54966.1	XM_023254966.1:c.3453C> T	p.Ile115 1=	0.5	13,13	26	99	
78	G/A			0.00	8														
B3: 173		VQSRTr ancheSN		0.00	8														
063		P99.00to 99.90	1	1207	2	1	0	1	<i>IGF1R</i>	synonymous _variant	Othe r	XM_0232 54966.1	XM_023254966.1:c.3114C> T	p.Ala10 38=	0.38095 2	13,8	21	99	
66	G/A			0.00	8														
C1: 201		VQSRTr ancheSN		0.00	8														
986		P99.00to 99.90	1	1207	2	1	0	1	<i>IGFBP 2</i>	5_prime_UT R_variant	Othe r	XM_0112 85583.3	XM_011285583.3:c.*41C>G		0.55	9,11	20	99	
084	C/G			0.00	8														
A2: 633		VQSRTr ancheSN		0.00	8														
333		P99.00to 99.90	1	1207	2	1	0	1	<i>IGFBP 3</i>	synonymous _variant	Othe r	XM_0232 50143.1	XM_023250143.1:c.33G>A	p.Ala11 =	0.375	10,6	16	99	
84	C/T			0.00	8														
B2: 526				1207	2														
611				0.00	8														
45	C/A	PASS	1	73	8	1	0	1	<i>IL17F</i>	3_prime_UT R_variant	Othe r	XM_0232 54749.1	XM_023254749.1:c.*245G>T		0.5625	7,9	16	99	
D1: 877				0.00	8														
236				1207	2														
29	C/A	PASS	1	73	8	1	0	1	<i>IMMP1 L</i>	5_prime_UT R_variant	Othe r	XM_0069 37165.3	XM_006937165.3:c.*29-23901G>T		0.48	13,12	25	99	
D1: 158				0.00	8														
312				1207	2														
73	C/T	PASS	1	73	8	1	0	1	<i>JAML</i>	synonymous _variant	Othe r	XM_0198 11359.2	XM_019811359.2:c.711G> A	p.Ala23 7=	0.44444 4	10,8	18	99	
D1: 158				0.00	8														
481				1207	2														
86	G/A	PASS	1	73	8	1	0	1	<i>JAML</i>	5_prime_UT R_variant	Othe r	XM_0069 36675.4	XM_006936675.4:c.*35C>T		0.5	7,7	14	99	
E2: 368				0.00	8														
095				1207	2														
16	G/A	PASS	1	73	8	1	0	1	<i>KATNB 1</i>	synonymous _variant	Othe r	XM_0039 98077.5	XM_003998077.5:c.1758G> A	p.Leu58 6=	0.72	7,18	25	99	

B1: 124 409 361 B1: 124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 290224 1	3_prime_UT R_variant	Othe r	XM_0069 30945.4	XM_006930945.4:c.*1595C>T	0.4	15,10	25	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 290224 1	3_prime_UT R_variant	Othe r	XM_0069 30945.4	XM_006930945.4:c.*2514G>T	0.44444 4	10,8	18	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 525965 5	synonymous _variant	Othe r	XM_0232 45891.1	XM_023245891.1:c.268C>T	p.Leu90 =	0.25714 3	78,27	105	99
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 525972 2	3_prime_UT R_variant	Othe r	XM_0232 41195.1	XM_023241195.1:c.*158C>T	0.23529 4	91,28	119	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 526134 7	non_coding_ exon_variant	Othe r	XR_00273 9338.1	XR_002739338.1:n.1894C>G	0.65517 2	10,19	29	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	T/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949242 8	non_coding_ exon_variant	Othe r	XR_00214 6284.2	XR_002146284.2:n.1929_1964del	0.57142 9	12,16	28	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	T/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949293 1	non_coding_ exon_variant	Othe r	XR_00273 6633.1	XR_002736633.1:n.480A>G	0.57894 7	8,11	19	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	A/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949488 6,LOC1 115581 41	synonymous _variant	Othe r	XM_0232 46150.1,X R_002738 727.1	XM_023246150.1:c.25A>C, XR_002738727.1:n.31T>G	p.Arg9= ,	0.56	11,14	25	99
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	A/G	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949863 5	non_coding_ exon_variant	Othe r	XR_00215 5471.2	XR_002155471.2:n.961T>C	0.56521 7	10,13	23	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949927 6	non_coding_ exon_variant	Othe r	XR_00274 1540.1	XR_002741540.1:n.148C>T	0.45454 5	12,10	22	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	T/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949934 1	non_coding_ exon_variant	Othe r	XR_00274 1469.1	XR_002741469.1:n.256T>A	0.52631 6	9,10	19	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 949968 8	non_coding_ exon_variant	Othe r	XR_00274 2293.1	XR_002742293.1:n.847G>A	0.73913	6,17	23	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	CTTC/-	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 950101 4	non_coding_ exon_variant	Othe r	XR_00274 3257.1	XR_002743257.1:n.883_886delTCCT	0.66666 7	6,12	18	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 950116 2	non_coding_ exon_variant	Othe r	XR_00215 9119.2	XR_002159119.2:n.181C>T	0.57142 9	9,12	21	99	
124 410 280 E2: 569 822 5 A2: 117 037 24 A2: 159 585 30 D2: 177 438 66 D2: 136 842 0	G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	1	LOC10 950280 0	non_coding_ exon_variant	Othe r	XR_00216 1645.2	XR_002161645.2:n.3C>T	0.60869 6	9,14	23	99	

B2: 347		VQSRTr		0.00	8														
761		ancheSN		1207	2			<i>LOC11</i>											
44	C/T	P99.90to	1	73	8	1	0	1	6	synonymous	Othe	XM_0232	XM_023253817.1:c.144C>	p.Asn48					
E1: 545		100.00								_variant	r	53817.1	T	=	0.75	1,3	4	20	
659		VQSRTr		0.00	8														
87	G/A	ancheSN	1	1207	2	1	0	1	4	non_coding_	Othe	XR_00273			0.60869				
B1: 161		P99.00to		73	8	1	0	1		exon_variant	r	8129.1	XR_002738129.1:n.631C>T		6	9,14	23	99	
031		99.90																	
87	G/A	VQSRTr	1	0.00	8	1	0	1	3	non_coding_	Othe	XR_00274	XR_002741614.1:n.657G>A		2	12,14	26	99	
B3: 405		ancheSN		1207	2					exon_variant	r	1614.1							
375		P99.00to	1	73	8	1	0	1	7	non_coding_	Othe	XR_00274			0.42857				
04	C/T	99.90								exon_variant	r	2887.1	XR_002742887.1:n.146G>A		1	12,9	21	99	
B3: 148		VQSRTr		0.00	8														
757		ancheSN		1207	2														
733	G/A	P99.00to	1	73	8	1	0	1	5	5_prime_UT	Othe	XM_0232			0.55555				
		99.90								R_variant	r	56127.1	XM_023256127.1:c.-298C>T		6	12,15	27	99	
		TTTTCTTCTTCTTT																	
B3: 134		CTTCTTCTTCTTCT		0.00	8														
812		TCTTCTTCTTCTTCT		1207	2														
023	T/-	TCTTCTTCTTCTTCT	1	73	8	1	0	1	3	non_coding_	Othe	XR_00274	XR_002743403.1:n.105_107delAGA		4	10,8	18	99	
A2: 135		PASS								exon_variant	r	3403.1							
101		VQSRTr		0.00	8														
42	T/A	ancheSN	1	1207	2	1	0	1		3_prime_UT	Othe	XM_0039	XM_003982075.5:c.*201A>T		0.1875	13,3	16	68	
A2: 135		100.00								R_variant	r	82075.5							
101		VQSRTr		0.00	8														
49	T/A	ancheSN	1	1207	2	1	0	1		3_prime_UT	Othe	XM_0039	XM_003982075.5:c.*194A>T		0.1875	13,3	16	68	
A2: 159		P99.90to		73	8	1	0	1		R_variant	r	82075.5							
678		100.00																	
12	G/A	PASS	1	0.00	8	1	0	1		synonymous	Othe	XM_0112	XM_011291127.2:c.1888C>	p.Leu63					
D2: 132		99.90to		1207	2					_variant	r	91127.2	T	0=	0.5	10,10	20	99	
166		100.00																	
77	G/A	PASS	1	0.00	8	1	0	1		synonymous	Othe	NM_0012	NM_001290242.1:c.1566G>	p.Ser52	0.54545				
E1: 440		99.90to		73	8	1	0	1		_variant	r	90242.1	A	2=	5	10,12	22	99	
043		100.00																	
79	C/T	PASS	1	0.00	8	1	0	1		synonymous	Othe	XM_0232	XM_023243721.1:c.402G>	p.Val13	0.59090				
E1: 122		99.90to		1207	2					_variant	r	43721.1	A	4=	9	9,13	22	99	
560		100.00																	
51	G/A	PASS	1	0.00	8	1	0	1		3_prime_UT	Othe	XM_0232	XM_023244645.1:c.6900+592C>T		0.44	14,11	25	99	
B1: 124		VQSRTr		1207	2					R_variant	r	44645.1							
428		ancheSN		73	8	1	0	1											
617	C/T	P99.00to	1	73	8	1	0	1		3_prime_UT	Othe	XM_0198	XM_019829170.2:c.*214G>A		0.45454				
B1: 124		99.90								R_variant	r	29170.2			5	12,10	22	99	
428		100.00																	
822	CAAT/-	PASS	1	0.00	8	1	0	1		3_prime_UT	Othe	XM_0198	XM_019829170.2:c.*6_*9delATTG		0.5	9,9	18	99	
A3: 894		VQSRTr		1207	2					R_variant	r	29170.2							
375		ancheIN		73	8	1	0	1											
85	C/-	DEL99.0	1	0.00	8	1	0	1		3_prime_UT	Othe	XM_0039	XM_003984090.5:c.*85delC		0.47368				
		Oto99.90		1207	2					R_variant	r	84090.5			4	10,9	19	99	

D2: 696 784 20	C/T	VQSRTr ancheSN P99.90to 100.00 VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>MXII</i>	5_prime_UT R_variant	Othe r	XM_0039 94402.5	XM_003994402.5:c.263-1490C>T	0.29411 8	12,5	17	99	
X:2 001 064 A1: 513 582 48 A1: 513 844 78 A1: 514 901 60 A1: 515 251 60 B4: 132 656 315 D2: 130 335 76 D2: 130 389 91 A2: 778 888 89 E1: 450 227 41 B3: 767 759 51 B3: 767 764 64 E2: 360 463 41 B3: 425 292 52	G/A	VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>MXRA5</i>	synonymous _variant	Othe r	XM_0040 00245.5	XM_004000245.5:c.4284C>T	p.Ser14 28=	0.57142 9	9,12	21	99
	A/G	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>MYCBP 2</i>	synonymous _variant	Othe r	XM_0112 80436.3	XM_011280436.3:c.8004T>C	p.Leu26 68=	0.475	21,19	40	99
	G/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>MYCBP 2</i>	synonymous _variant	Othe r	XM_0112 80436.3	XM_011280436.3:c.7105C>A	p.Arg23 69=	0.5	17,17	34	99
	C/T	VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>MYCBP 2</i>	synonymous _variant	Othe r	XM_0112 80436.3	XM_011280436.3:c.2544G>A	p.Gly84 8=	0.46153 8	14,12	26	99
	C/T	VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>MYCBP 2</i>	synonymous _variant	Othe r	XM_0112 80436.3	XM_011280436.3:c.642G>A	p.Thr21 4=	0.72727 3	6,16	22	99
	G/A	VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>MYH9</i>	synonymous _variant	Othe r	XM_0232 57448.1	XM_023257448.1:c.4875C>T	p.His16 25=	0.61111 1	7,11	18	99
	C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>NID1</i>	synonymous _variant	Othe r	XM_0039 93899.5	XM_003993899.5:c.843C>T	p.Leu28 1=	0.56521 7	10,13	23	99
	C/T	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>NID1</i>	synonymous _variant	Othe r	XM_0039 93899.5	XM_003993899.5:c.1305C>T	p.Asn43 5=	0.5	14,14	28	99
	G/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>NME8</i>	3_prime_UT R_variant	Othe r	XM_0198 25295.2	XM_019825295.2:c.*690C>G	0.55555 6	8,10	18	99	
	-/TA	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>NMT1</i>	3_prime_UT R_variant	Othe r	XM_0039 96998.5	XM_003996998.5:c.*340_*341dupAT	0.46666 7	8,7	15	99	
	A/G	VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>NOP9</i>	5_prime_UT R_variant	Othe r	XM_0039 87519.4	XM_003987519.4:c.-71A>G	0.59090 9	9,13	22	99	
	G/C	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>NOP9</i>	splice_regio n_variant	Othe r	XM_0039 87519.4	XM_003987519.4:c.236-6G>C	0.4	15,10	25	99	
	A/G	VQSRTr ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>NUP93</i>	synonymous _variant	Othe r	XM_0069 41526.4	XM_006941526.4:c.1728A>G	p.Glu57 6=	0.4	18,12	30	99
	G/A	PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>OAZ2</i>	3_prime_UT R_variant	Othe r	NM_0012 90005.1	NM_001290005.1:c.*54G>A	0.5	10,10	20	99	

A3: 128				0.00	8															
812				1207	2															
051	AA/-	PASS	1	73	8	1	0	1	<i>RDH14</i>	3_prime_UT R_variant	Othe r	XM_0039 84483.4	XM_003984483.4:c.*185_*186delAA	0.57894 7	8,11	19	99			
A3: 103		VQSRTr ancheSN		0.00	8															
292		P99.00to		1207	2															
529	A/C	99.90	1	73	8	1	0	1	<i>REEP1</i>	3_prime_UT R_variant	Othe r	XM_0232 51848.1	XM_023251848.1:c.*162T>G	0.33333 3	18,9	27	99			
D4: 757		VQSRTr ancheSN		0.00	8															
003		P99.00to		1207	2															
56	G/A	99.90	1	73	8	1	0	1	<i>RGS3</i>	3_prime_UT R_variant	Othe r	XM_0198 16366.2	XM_019816366.2:c.*622G>A	0.60869 6	9,14	23	99			
A1: 120				0.00	8															
047				1207	2															
366	C/T	PASS	1	73	8	1	0	1	<i>RNF14</i>	5_prime_UT R_variant	Othe r	XM_0039 80869.5	XM_003980869.5:c.-11C>T	0.41379 3	17,12	29	99			
A1: 120		VQSRTr ancheSN		0.00	8															
059		P99.00to		1207	2															
545	G/A	99.90	1	73	8	1	0	1	<i>RNF14</i>	3_prime_UT R_variant	Othe r	XM_0039 80869.5	XM_003980869.5:c.*176G>A	0.44444 4	10,8	18	99			
A1: 120				0.00	8															
059				1207	2															
695	AGTTAA/-	PASS	1	73	8	1	0	1	<i>RNF14</i>	3_prime_UT R_variant	Othe r	XM_0039 80869.5	XM_003980869.5:c.*331_*336delAAGT TA	0.59375	13,19	32	99			
B3: 478		VQSRTr ancheSN		0.00	8															
280		P99.00to		1207	2															
31	C/G	99.90	1	73	8	1	0	1	<i>RNF11 1</i>	synonymous _variant	Othe r	XM_0039 87100.5	XM_003987100.5:c.48G>C	p.Val16 =	0.5	10,10	20	99		
742				0.00	8															
823				1207	2															
50	C/T	PASS	1	73	8	1	0	1	<i>RPGRI P1</i>	synonymous _variant	Othe r	XM_0198 32776.2	XM_019832776.2:c.1626C> T	p.Tyr54 2=	0.38888 9	11,7	18	99		
B3: 713		VQSRTr ancheSN		0.00	8															
392		P99.00to		1207	2															
26	A/G	99.90	1	73	8	1	0	1	<i>RYR3</i>	synonymous _variant	Othe r	XR_00274 2989.1	XR_002742989.1:n.6879A>G	0.45	11,9	20	99			
B3: 334		VQSRTr ancheSN		0.00	8															
363		P99.90to		1207	2															
59	C/T	100.00	1	73	8	1	0	1	<i>SCAMP 2</i>	synonymous _variant	Othe r	XM_0198 32043.2	XM_019832043.2:c.381C> T	p.Ser12 7=	0.2	24,6	30	92		
D1: 101				0.00	8															
390				1207	2															
191	G/A	PASS	1	73	8	1	0	1	<i>SLC39A 13</i>	3_prime_UT R_variant	Othe r	XM_0232 39820.1	XM_023239820.1:c.*9G>A	0.33333 3	14,7	21	99			
E1: 661		VQSRTr ancheSN		0.00	8															
491	C/T	P99.00to	1	73	8	1	0	1	<i>SMTNL 2</i>	5_prime_UT R_variant	Othe r	XM_0232 43730.1	XM_023243730.1:c.-26C>T	0.33333 3	8,4	12	99			
E1: 669		VQSRTr ancheSN		0.00	8															
606	C/T	P99.00to	1	73	8	1	0	1	<i>SMTNL 2</i>	synonymous _variant	Othe r	XM_0232 43730.1	XM_023243730.1:c.756C> T	p.Ser25 2=	0.69230 8	4,9	13	83		
E1: 669		VQSRTr ancheSN		0.00	8															
654	C/T	P99.00to	1	73	8	1	0	1	<i>SMTNL 2</i>	synonymous _variant	Othe r	XM_0232 43730.1	XM_023243730.1:c.804C> T	p.Ser26 8=	0.58333 3	5,7	12	99		
C2: 117				0.00	8															
995				1207	2															
51	C/A	PASS	1	73	8	1	0	1	<i>SON</i>	3_prime_UT R_variant	Othe r	XM_0232 38789.1	XM_023238789.1:c.6933+431G>T	0.47619	11,10	21	99			

D4: 112 869 3 E1: 191 542 92 B1: 201 541 386 B1: 201 541 515 B4: 102 578 953 B1: 200 870 224		VQSRT ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>WNK2</i>	synonymous _variant	Othe r	XM_0232 43243.1	XM_023243243.1:c.2187G> A	p.Ala72 9=	0.38888 9	11,7	18	99
	G/A																	
		PASS VQSRT ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>WSBI</i>	synonymous _variant	Othe r	XM_0039 96524.5	XM_003996524.5:c.678G> A	p.Ser22 6=	0.53846 2	12,14	26	99
	C/T																	
		PASS VQSRT ancheSN P99.00to 99.90	1	0.00 1207 73	8 2 8	1	0	1	<i>ZBTB49</i>	synonymous _variant	Othe r	XM_0232 53313.1	XM_023253313.1:c.528C> T	p.His17 6=	0.46153 8	14,12	26	99
	C/T																	
		PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>ZBTB49</i>	synonymous _variant	Othe r	XM_0232 53313.1	XM_023253313.1:c.657C> T	p.Tyr21 9=	0.40909 1	13,9	22	99
	C/T																	
		PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>ZDHH C17</i>	3_prime_UT R_variant	Othe r	XM_0069 33902.3	XM_006933902.3:c.*1111delA		0.54166 7	11,13	24	99
	A/-																	
		PASS	1	0.00 1207 73	8 2 8	1	0	1	<i>ZNF518 B</i>	synonymous _variant	Othe r	XM_0039 85514.5	XM_003985514.5:c.2034A> T	p.Ile678 =	0.52173 9	11,12	23	99
	A/T																	

Table S3. Numbers of variants identified by whole genome sequencing in the feline case.

Filter / Variant type	Case genotype	
	Homozygous*	Heterozygous
Pass	101	169
3' UTR	34	36
5' UTR	5	11
Frameshift	2	6
In-frame deletion	1	2
In-frame insertion	0	1
Missense	18	60
Non-coding exon	9	7
Splice donor	0	0
Splice acceptor	2	0
Splice region	3	3
Synonymous	22	43
PASS,VQSRTrancheSNP99.00to99.90	5	0
VQSRTrancheINDEL99.00to99.90	3	3
VQSRTrancheINDEL99.90to100.00	2	11
VQSRTrancheSNP99.00to99.90	61	85
VQSRTrancheSNP99.90to100.00	5	18
Total variants	172	286

*One additional cat in the dataset could also be carriers for any variant.

Table S4. A list of candidate genes evaluated in this study.

Ehlers-Danlos syndromes in humans		Congenital scoliosis in humans		Hereditary connective tissue disorders overlapping phenotype genes in addition to genes associated with Ehlers-Danlos syndromes in humans		Kinked tail in cats	
<i>COL5A1</i>	XM_023242950.1	<i>DLL3</i>	XM_003997806.5	<i>ADAMTS2</i>	XM_023254116.1	<i>HES7</i>	XM_003996191.5
<i>COL5A2</i>	XM_003990962.4	<i>MESP2</i>	XM_019833746.2	<i>B3GALT6</i>	XM_011279777.2	<i>T</i>	XM_003986708.3
<i>COL1A1</i>	XM_003996699.4	<i>LFNG</i>	XM_023246563.1	<i>CHST14</i>	XM_003987275.4		
<i>COL3A1</i>	XM_003990961.3	<i>HES7</i>	XM_003996191.4	<i>COL1A1</i>	XM_003996699.4		
<i>COL1A2</i>	XM_003982764.5	<i>NOTCH2</i>	XM_011285094.3	<i>COL1A2</i>	XM_003982764.5		
<i>ADAMTS2</i>	XM_023254116.1	<i>NOTCH3</i>	XM_023246608.1	<i>COL3A1</i>	XM_003990961.3		
<i>PLOD1</i>	XM_003989499.3	<i>SLC35A3</i>	XM_003990360.4	<i>COL5A1</i>	XM_023242950.1		
<i>FKBP14</i>	XM_003982902.4	<i>T (TBXT)</i>	XM_003986708.3	<i>COL5A2</i>	XM_003990962.4		
<i>TNXB</i>	XM_023254711.1	<i>TBX6</i>	XM_011290514.3	<i>DSE</i>	XM_019831105.2		
<i>COL12A1</i>	XM_019830870.1	<i>FBN1</i>	XM_023255387.1	<i>FKBP14</i>	XM_003982902.4		
<i>CHST14</i>	XM_003987275.4	<i>SHH</i>	XM_023242357.1	<i>PLOD1</i>	XM_003989499.3		
<i>DSE</i>	XM_019831105.2	<i>SOX9</i>	XM_023243815.1	<i>TNXB</i>	XM_023254711.1		
<i>B4GALT7</i>	XM_023254211.1	<i>FLNB</i>	XM_019823423.2	<i>COL12A1</i>	XM_019830870.1		
<i>B3GALT6</i>	XM_011279777.2	<i>PTK7</i>	XM_003986191.4	<i>C1R</i>	XM_019834454.2		
<i>SLC39A13 (ZIP13)</i>	XM_023239820.1	<i>HSPG2</i>	XM_023258277.1	<i>C1S</i>	XM_006933467.4		
<i>ZNF469</i>	XM_023245050.1	<i>KIAA1217</i>	XM_023256446.1	<i>SLC39A13</i>	XM_023239820.1		
<i>PRDM5</i>	XM_023252840.1	<i>DCC</i>	XM_019815281.2	<i>ZNF469</i>	XM_023245050.1		
<i>C1R</i>	XM_019834454.2	<i>DVL3</i>	XM_023260347.1	<i>PRDM5</i>	XM_023252840.1		
<i>C1S</i>	XM_006933467.4	<i>FGFR3</i>	XM_023253489.1	<i>LZTS1</i>	XM_003984723.4		
<i>AEBP1 (ACLP)</i>	XM_023250110.1	<i>RECQL4</i>	XM_019823278.2	<i>FLNA</i>	XM_023249411.1		
		<i>ROBO3</i>	XM_019811488.2	<i>COL4A1</i>	XM_023253172.1		
		<i>WNT7A</i>	XM_011280231.3	<i>COL6A1</i>	XM_011285711.3		
		<i>CHRNA3</i>	XM_003991260.4	<i>COL6A2</i>	XM_023238595.1		
		<i>FGFR1</i>	XM_011281522.3	<i>COL6A6</i>	XM_011286221.3		
				<i>ELN</i>	XM_019820582.2		
				<i>FBLN5</i>	XM_019833518.1		
				<i>FBN1</i>	XM_023255387.1		
				<i>LOX</i>	XM_023254614.1		
				<i>MYH11</i>	XM_023246707.1		
				<i>RYR1</i>	XM_023245508.1		
				<i>SELENON (SEPN1)</i>	XM_023258321.1		
				<i>SGCB</i>	XM_023253126.1		
				<i>myosin-7 (MYH7)</i>	XM_006932746.4		
				<i>TTN</i>	XM_023259509.1		
				<i>SMAD2</i>	XM_023241966.1		
				<i>SMAD3</i>	XM_023255153.1		

<i>COL2A1</i>	XM_023256993.1
<i>COL11A1</i>	XM_023258940.1
<i>COL11A2</i>	XM_003985999.5
<i>COL9A1</i>	XM_003986294.4
<i>COL9A2</i>	XM_023258522.1
<i>COL9A3</i>	XM_023251026.1
<i>MED12</i>	XM_004000604.5
<i>FLNB</i>	XM_019823423.2
<i>LOC105261197 (CANT1)</i>	XM_019818343.2
<i>SLC2A10</i>	XM_006929697.2
<i>ABCC6</i>	XM_011290585.3
<i>GGCX</i>	XM_011281186.3
<i>ENPP1</i>	XM_023254376.1
<i>AEBP1</i>	XM_023250110.1
<i>SKI</i>	XM_023257965.1
<i>TGFB2</i>	XM_003999507.5
<i>TGFB3</i>	XM_003987851.3
<i>TGFBR1</i>	XM_023242499.1
<i>TGFBR2</i>	XM_023260647.1

Table S5. Primer sequences used for amplifying cDNA of the feline *COL6A1* gene.

Primer Name	Primer sequence (5'-3')	Product size (bp)	Annealing temperature (°C)
COL6A1-1F	CCTTCGAGAGCAGAGACAGC	580	58
COL6A1-1R	GGGTGCCCATCAGTCACTAC		
COL6A1-2F	CCCTGGTGGACAAGGTCAAG	657	58
COL6A1-2R	GACCTGGCTTCCCACGTTC		
COL6A1-3F	GCCAGACCATTGACACCATC	688	60
COL6A1-3R	CCTTCTCGTCCCTGGTCAC		
COL6A1-4F	GCCCTGGGGAAAGAGGAC	741	60
COL6A1-4R	GCTCCTGCATCTGGTTGTG		
COL6A1-5F	CATCGACAGGCTGAGCAGAG	682	60
COL6A1-5R	GTGGGAGGGGTTCGTCTTG		
COL6A1-6F	ATCAAGGACATGTTTGGCTTC	905	58
COL6A1-6R	TAGAAACATCCAGCTCATCAGG		
COL6A1-6F-internal*	CTGCAGAACTACACCGTGCTG		
COL6A1-6R-internal*	AGAGCAGCAGCCTCTTCTTG		

*These were used as internal sequencing primers.