

## GEPHE SUMMARY

<p>HES7 (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+HES7+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+HES7+gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002190</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+gephebase-summary-title</a>)</p> <p>Organ size (tail; short) (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Organ+size+tail+short+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Organ+size+tail+short+gephebase-summary-title</a>)</p> <p>WT cat</p> <p>Short tail - Chinese short-tailed feral cat ; and Japanese Bobtail cat</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Felis catus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+gephebase-summary-title</a>)</p> <p>Common Name</p> <p>domestic cat</p> <p>Synonyms</p> <p>Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Felis</p> <p>Parent</p> <p>Felis () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682</a>)</p> <p>9685 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Felis catus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+gephebase-summary-title</a>)</p> <p>Common Name</p> <p>domestic cat</p> <p>Synonyms</p> <p>Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Felis</p> <p>Parent</p> <p>Felis () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682</a>)</p> <p>9685 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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## GENOTYPIC CHANGE

<p>HES7</p> <p>SCDO4; bHLHb37; BHLHB37</p> <p>9606.ENSP00000446205 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000446205">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000446205</a>)</p> <p>-</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>Q9BYE0 (<a href="http://www.uniprot.org/uniprot/Q9BYE0">http://www.uniprot.org/uniprot/Q9BYE0</a>)</p> <p>()</p> <p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
<p>GO:0046983 : protein dimerization activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046983">https://www.ebi.ac.uk/QuickGO/term/GO:0046983</a>)</p> <p>GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0000977">https://www.ebi.ac.uk/QuickGO/term/GO:0000977</a>)</p> <p>GO:0008134 : transcription factor binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008134">https://www.ebi.ac.uk/QuickGO/term/GO:0008134</a>)</p>		

GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)  
 GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)  
 GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001227>)

GO - Biological Process

GO:0050767 : regulation of neurogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0050767>)  
 GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)  
 GO:0001501 : skeletal system development (<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)  
 GO:0009952 : anterior/posterior pattern specification (<https://www.ebi.ac.uk/QuickGO/term/GO:0009952>)  
 GO:0000122 : negative regulation of transcription by RNA polymerase II (<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)  
 GO:0007219 : Notch signaling pathway (<https://www.ebi.ac.uk/QuickGO/term/GO:0007219>)  
 GO:0001756 : somitogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0001756>)  
 GO:0007498 : mesoderm development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007498>)  
 GO:0036342 : post-anal tail morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0036342>)  
 GO:0048511 : rhythmic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0048511>)

GO - Cellular Component

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)  
 GO:0000790 : nuclear chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000790>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>) Presumptive Null  
 Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title>) Molecular Type  
 SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>) Aberration Type  
 Nonsynonymous SNP Coding Change  
 g.2819475A>G c.5A>G p.V2A Molecular Details of the Mutation  
 Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title>) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Val	Ala	2

Whole genome sequencing in cats, identifies new models for blindness in AIPL1 and somite segmentation in HES7. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27030474>) Main Reference  
 Lyons LA; Creighton EK; Alhaddad H; Beale HC; Grahn RA; Rah H; Maggs DJ; Helps CR; Gandolfi B Authors

The reduced cost and improved efficiency of whole genome sequencing (WGS) is drastically improving the development of cats as biomedical models. Persian cats are models for Leber's congenital amaurosis (LCA), the most severe and earliest onset form of visual impairment in humans. Cats with innocuous breed-defining traits, such as a bobbed tail, can also be models for somite segmentation and vertebral column development. Abstract

The first WGS in cats was conducted on a trio segregating for LCA and the bobbed tail abnormality. Variants were identified using FreeBayes and effects predicted using SnpEff. Variants within a known haplotype block for cat LCA and specific candidate genes for both phenotypes were prioritized by the predicted variant effect on the proteins and concordant segregation within the trio. The efficiency of WGS of a single trio of domestic cats was evaluated.

A stop gain was identified at position c.577C > T in cat AIPL1, a predicted p.Arg193\*. A c.5A > G variant causing a p.V2A was identified in HES7. The variants segregated concordantly in a Persian - Japanese bobtail pedigree. Over 1700 cats from 40 different breeds and populations were genotyped for the AIPL1 variant, defining an allelic frequency in only Persian-related breeds of 1.15%. A sub-set of cats was genotyped for the HES7 variant, supporting the variant as private to the Japanese bobtail breed. Approximately 18 million SNPs were identified for application in cat research. The cat AIPL1 variant would have been considered a high priority variant for evaluation, regardless of a priori knowledge from previous genetic studies.

This study represents the first effort of the 99 Lives Cat Genome Sequencing Initiative to identify disease-causing variants in the domestic cat using WGS. The current cat reference assembly is efficient for gene and variant identification. However, as the feline variant database improves, development of cats as biomedical models for human disease will be more efficient, providing an alternative, large animal model for drug and gene therapy trials. Undiagnosed human patients with early-onset blindness should be screened for this AIPL1 variant. The HES7 variant should further calibrate the somite segmentation clock.

Whole Genome Sequencing Identifies a Missense Mutation in HES7 Associated with Short Tails in Asian Domestic Cats. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27560986>) Additional References

RELATED GEPHE

1 (TBXT) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9685^/and+Trait=Organ size/and+groupHaplotypes=true#gephebase-summary-title>)

Related Genes

No matches found.

## EXTERNAL LINKS

## COMMENTS

<https://omia.org/OMIA001987/9685/>