

GEPHE SUMMARY

<p>Kit (type III receptor protein-tyrosine kinase) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit (type III receptor protein-tyrosine kinase)^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00002229</p> <p>Martin</p> <p>Entry Status</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)</p> <p>Coloration (coat ; white-spotting) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat ; white-spotting)^#gephebase-summary-title)</p> <p>WT camels</p> <p>Camels with white-spotting</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)</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>Camelus dromedarius (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Camelus dromedarius^#gephebase-summary-title)</p> <p>Arabian camel</p> <p>Arabian camel; camel; dromedaries; dromedary; dromedary camel; one-humped camel; Camelus dromedarius Linnaeus, 1758</p> <p>species</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; Cetartiodactyla; Tylopoda; Camelidae; Camelus</p> <p>Camelus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9836)</p> <p>9838 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9838)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Infrasppecies?</p>	<p>Taxon B</p> <p>Camelus dromedarius (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Camelus dromedarius^#gephebase-summary-title)</p> <p>Arabian camel</p> <p>Arabian camel; camel; dromedaries; dromedary; dromedary camel; one-humped camel; Camelus dromedarius Linnaeus, 1758</p> <p>species</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; Cetartiodactyla; Tylopoda; Camelidae; Camelus</p> <p>Camelus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9836)</p> <p>9838 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9838)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Infrasppecies?</p>
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GENOTYPIC CHANGE

<p>Kit</p> <p>W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfscow3; SI</p> <p>10090.ENSMUSP00000005815 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P05532 (http://www.uniprot.org/uniprot/P05532)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
<p>GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)</p> <p>GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)</p> <p>GO:0042803 : protein homodimerization activity</p>		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)
GO:0002020 : protease binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0002020>)
GO:0004714 : transmembrane receptor protein tyrosine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004714>)
GO:0004713 : protein tyrosine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004713>)
GO:0019955 : cytokine binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019955>)
GO:0005020 : stem cell factor receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005020>)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)
GO:0035234 : ectopic germ cell programmed cell death
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035234>)
GO:0035162 : embryonic hemopoiesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035162>)
GO:0008584 : male gonad development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008584>)
GO:0001541 : ovarian follicle development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001541>)
GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
GO:0043406 : positive regulation of MAP kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043406>)
GO:0010628 : positive regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:0043410 : positive regulation of MAPK cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043410>)
GO:0007283 : spermatogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0007283>)
GO:0008360 : regulation of cell shape
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008360>)
GO:0048070 : regulation of developmental pigmentation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048070>)
GO:0006468 : protein phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006468>)
GO:0060326 : cell chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060326>)
GO:0006935 : chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0006935>)
GO:0048565 : digestive tract development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048565>)
GO:0006954 : inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006954>)
GO:0019221 : cytokine-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019221>)
GO:0048863 : stem cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048863>)
GO:0048066 : developmental pigmentation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048066>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0009968 : negative regulation of signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009968>)
GO:0046777 : protein autophosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)
GO:0030218 : erythrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030218>)
GO:0018108 : peptidyl-tyrosine phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018108>)
GO:0097067 : cellular response to thyroid hormone stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097067>)
GO:1904349 : positive regulation of small intestine smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904349>)
GO:0000187 : activation of MAPK activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000187>)
GO:0046427 : positive regulation of JAK-STAT cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046427>)
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042531>)
GO:0030335 : positive regulation of cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030335>)
GO:0046686 : response to cadmium ion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046686>)
GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)

GO:0031532 : actin cytoskeleton reorganization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031532>)
GO:0002371 : dendritic cell cytokine production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002371>)
GO:0050910 : detection of mechanical stimulus involved in sensory perception of sound
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050910>)
GO:0050673 : epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050673>)
GO:0038162 : erythropoietin-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038162>)
GO:0038093 : Fc receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038093>)
GO:0007281 : germ cell development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007281>)
GO:0008354 : germ cell migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0008354>)
GO:0006687 : glycosphingolipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006687>)
GO:0035701 : hematopoietic stem cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035701>)
GO:0030097 : hemopoiesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0030097>)
GO:0002327 : immature B cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002327>)
GO:0038109 : Kit signaling pathway (<https://www.ebi.ac.uk/QuickGO/term/GO:0038109>)
GO:0030032 : lamellipodium assembly
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030032>)
GO:0002320 : lymphoid progenitor cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002320>)
GO:0002551 : mast cell chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0002551>)
GO:0032762 : mast cell cytokine production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032762>)
GO:0043303 : mast cell degranulation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043303>)
GO:0060374 : mast cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060374>)
GO:0035855 : megakaryocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035855>)
GO:0097326 : melanocyte adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0097326>)
GO:0097324 : melanocyte migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0097324>)
GO:0002573 : myeloid leukocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002573>)
GO:0002318 : myeloid progenitor cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002318>)
GO:0043069 : negative regulation of programmed cell death
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043069>)
GO:1904343 : positive regulation of colon smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904343>)
GO:0051091 : positive regulation of DNA-binding transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051091>)
GO:0048170 : positive regulation of long-term neuronal synaptic plasticity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048170>)
GO:0045747 : positive regulation of Notch signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045747>)
GO:0031274 : positive regulation of pseudopodium assembly
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031274>)
GO:0120072 : positive regulation of pyloric antrum smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0120072>)
GO:1905065 : positive regulation of vascular smooth muscle cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1905065>)
GO:1904251 : regulation of bile acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904251>)
GO:0009314 : response to radiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0009314>)
GO:0048103 : somatic stem cell division
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048103>)
GO:0035019 : somatic stem cell population maintenance
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035019>)
GO:0007286 : spermatid development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007286>)
GO:0030217 : T cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030217>)
GO:0043586 : tongue development (<https://www.ebi.ac.uk/QuickGO/term/GO:0043586>)
GO:0008542 : visual learning (<https://www.ebi.ac.uk/QuickGO/term/GO:0008542>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)
GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)
GO:0009986 : cell surface (<https://www.ebi.ac.uk/QuickGO/term/GO:0009986>)

GO:0009898 : cytoplasmic side of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009898>)
GO:0009897 : external side of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009897>)
GO:0001669 : acrosomal vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0001669>)
GO:0005911 : cell-cell junction (<https://www.ebi.ac.uk/QuickGO/term/GO:0005911>)
GO:0042629 : mast cell granule (<https://www.ebi.ac.uk/QuickGO/term/GO:0042629>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title))

Aberration Type

Deletion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Deletion^#gephebase-summary-title))

Deletion Size

1-9 bp

Molecular Details of the Mutation

c.1842delG p.M614IfsX5

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene^#gephebase-summary-title))

Main Reference

A Frameshift Mutation in KIT is Associated with White Spotting in the Arabian Camel. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28282952>)

Authors

Holl H; Isaza R; Mohamoud Y; Ahmed A; Almathen F; Youcef C; Gaouar S; Antczak DF; Brooks S

Abstract

While the typical Arabian camel is characterized by a single colored coat, there are rare populations with white spotting patterns. White spotting coat patterns are found in virtually all domesticated species, but are rare in wild species. Theories suggest that white spotting is linked to the domestication process, and is occasionally associated with health disorders. Though mutations have been found in a diverse array of species, fewer than 30 genes have been associated with spotting patterns, thus providing a key set of candidate genes for the Arabian camel. We obtained 26 spotted camels and 24 solid controls for candidate gene analysis. One spotted and eight solid camels were whole genome sequenced as part of a separate project. The spotted camel was heterozygous for a frameshift deletion in KIT (c.1842delG, named KITW1 for White spotting 1), whereas all other camels were wild-type (KIT+/KIT+). No additional mutations unique to the spotted camel were detected in the EDNRB, EDN3, SOX10, KITLG, PDGFRA, MITF, and PAX3 candidate white spotting genes. Sanger sequencing of the study population identified an additional five KITW1/KIT+ spotted camels. The frameshift results in a premature stop codon five amino acids downstream, thus terminating KIT at the tyrosine kinase domain. An additional 13 spotted camels tested KIT+/KIT+, but due to phenotypic differences when compared to the KITW1/KIT+ camels, they likely represent an independent mutation. Our study suggests that there are at least two causes of white spotting in the Arabian camel, the newly described KITW1 allele and an uncharacterized mutation.

Additional References

RELATED GEPHE

Related Genes

1 (Agouti (ASIP)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9838^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9838^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage ; <https://omia.org/OMIA001737/9838/>