

|                                                                                                                                             |                |              |
|---------------------------------------------------------------------------------------------------------------------------------------------|----------------|--------------|
|                                                                                                                                             | Gephebase Gene | GepheID      |
| Melanophilin (MLPH) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> | GP00000644     |              |
| Gephebase="Melanophilin (MLPH)"#gephebase-summary-title)                                                                                    |                | Main curator |
|                                                                                                                                             | Entry Status   | Martin       |
| Published                                                                                                                                   |                |              |

|                                                                                                                                                                                             |                        |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------|
|                                                                                                                                                                                             | Trait Category         |
| Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology">#https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology</a> )           |                        |
|                                                                                                                                                                                             | Trait                  |
| Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat">#https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat</a> )            |                        |
|                                                                                                                                                                                             | Trait State in Taxon A |
| Felis catus                                                                                                                                                                                 |                        |
|                                                                                                                                                                                             | Trait State in Taxon B |
| Felis catus - "dilute"                                                                                                                                                                      |                        |
|                                                                                                                                                                                             | Ancestral State        |
| Taxon A                                                                                                                                                                                     |                        |
|                                                                                                                                                                                             | Taxonomic Status       |
| Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated">#https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated</a> ) |                        |

| GENOTYPIC CHANGE                                                                                                                                                                                                                            |                         |                                                                                                                      |  |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------|----------------------------------------------------------------------------------------------------------------------|--|
|                                                                                                                                                                                                                                             | Generic Gene Name       | UniProtKB Mus musculus                                                                                               |  |
| Mlph                                                                                                                                                                                                                                        |                         | Q91V27 ( <a href="http://www.uniprot.org/uniprot/Q91V27">http://www.uniprot.org/uniprot/Q91V27</a> )                 |  |
|                                                                                                                                                                                                                                             | Synonyms                | GenebankID or UniProtKB                                                                                              |  |
| In; l1Rk3; Slac-2a; AW228792; D1Wsu84e; l(1)-3Rk; 2210418F23Rik; 5031433I09Rik; Ln; Slac2a                                                                                                                                                  |                         | ABF06442 ( <a href="https://www.ncbi.nlm.nih.gov/nucore/ABF06442">https://www.ncbi.nlm.nih.gov/nucore/ABF06442</a> ) |  |
|                                                                                                                                                                                                                                             | String                  |                                                                                                                      |  |
| 10090.ENSMUSP00000027528<br>( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000027528">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000027528</a> ) |                         |                                                                                                                      |  |
|                                                                                                                                                                                                                                             | Sequence Similarities   |                                                                                                                      |  |
| -                                                                                                                                                                                                                                           |                         |                                                                                                                      |  |
|                                                                                                                                                                                                                                             | GO - Molecular Function |                                                                                                                      |  |
| GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )                                                                                                |                         |                                                                                                                      |  |
| GO:0017137 : Rab GTPase binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0017137">https://www.ebi.ac.uk/QuickGO/term/GO:0017137</a> )                                                                                               |                         |                                                                                                                      |  |
| GO:0003779 : actin binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003779">https://www.ebi.ac.uk/QuickGO/term/GO:0003779</a> )                                                                                                    |                         |                                                                                                                      |  |

GO:0030674 : protein binding, bridging  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030674>)  
GO:0051010 : microtubule plus-end binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051010>)  
GO:0017022 : myosin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0017022>)  
GO:0031489 : myosin V binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0031489>)  
GO - Biological Process  
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)  
GO:0030318 : melanocyte differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)  
GO:0032400 : melanosome localization  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032400>)  
GO:0006605 : protein targeting (<https://www.ebi.ac.uk/QuickGO/term/GO:0006605>)  
GO - Cellular Component

GO:0015629 : actin cytoskeleton (<https://www.ebi.ac.uk/QuickGO/term/GO:0015629>)  
GO:0030425 : dendrite (<https://www.ebi.ac.uk/QuickGO/term/GO:0030425>)  
GO:0048471 : perinuclear region of cytoplasm  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)  
GO:0005815 : microtubule organizing center  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005815>)  
GO:0030864 : cortical actin cytoskeleton  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030864>)  
GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)  
GO:0001725 : stress fiber (<https://www.ebi.ac.uk/QuickGO/term/GO:0001725>)  
GO:0016461 : unconventional myosin complex  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016461>)

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|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Presumptive Null                  |
| Yes ( <a href="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</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Molecular Type                    |
| Coding ( <a href="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</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Aberration Type                   |
| Deletion ( <a href="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</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | Deletion Size                     |
| 1-9 bp                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Molecular Details of the Mutation |
| 1bp deletion resulting in frameshift c.83delT                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |                                   |
| Linkage Mapping ( <a href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Experimental Evidence             |
| A homozygous single-base deletion in MLPH causes the dilute coat color phenotype in the domestic cat. (2006) ( <a href="https://pubmed.ncbi.nlm.nih.gov/16860533">https://pubmed.ncbi.nlm.nih.gov/16860533</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | Main Reference                    |
| Ishida Y; David VA; Eizirik E; SchÃ¶ffer AA; Neelam BA; Roelke ME; Hannah SS; O'Brien SJ; Menotti-Raymond M                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Authors                           |
| Three proteins have been described in humans and mice as being essential for even distribution, transport, and translocation of pigment granules, with defects in these molecules giving rise to lighter skin/coat color. The dilute phenotype in domestic cats affects both eumelanin and phaeomelanin pigment pathways; for example, black pigmentation combined with dilute appears gray and orange pigments appear cream. The dilute pigmentation segregates as a fully penetrant, autosomal recessive trait. We conducted classical linkage mapping with microsatellites in a large multigeneration pedigree of domestic cats and detected tight linkage for dilute on cat chromosome C1 (theta=0.08, LOD=10.81). Fine-mapping identified a genomic region exhibiting conserved synteny to human chromosome 2, which included one of the three dilute candidate genes, melanophilin (MLPH). Sequence analysis in dilute cats identified a single base pair deletion in exon 2 of MLPH transcripts that introduces a stop codon 11 amino acids downstream, resulting in the truncation of the bulk of the MLPH protein. The occurrence of this homozygous variant in 97 unrelated dilute cats representing 26 cat breeds and random-bred cats, along with 89 unrelated wild-type cats representing 29 breeds and random-bred cats, supports the finding that dilute is caused by this single mutation in MLPH (p<0.00001). Single-nucleotide polymorphism analyses in dilute individuals identified a single haplotype in dilute cats, suggesting that a single mutation event in MLPH gave rise to dilute in domestic cats. | Abstract                          |
|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Additional References             |

RELATED GEPHE

|                                                                                                                                                                                                                                                                                                                                                                                                                             |                    |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------|
|                                                                                                                                                                                                                                                                                                                                                                                                                             | Related Genes      |
| 6 (Agouti, Kit (type III receptor protein-tyrosine kinase), MC1R, Taqpep, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) ( <a href="https://www.gephebase.org/search-criteria?/or+Taxon ID=~9685~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title">https://www.gephebase.org/search-criteria?/or+Taxon ID=~9685~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title</a> ) | Related Haplotypes |
| No matches found.                                                                                                                                                                                                                                                                                                                                                                                                           |                    |

EXTERNAL LINKS

COMMENTS

@Parallelism <https://omia.org/OMIA000031/9685/>

