

## GEPHE SUMMARY

| Gephebase Gene                                                                                                                                                                                                                                                           |            | GepheID      |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------|--------------|
| Myostatin (MSTN = GDF8) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase~Myostatin+(MSTN+=GDF8)*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene</a><br>Gephebase~Myostatin (MSTN = GDF8)*#gephebase-summary-title) | GP00000697 | Main curator |
| Entry Status                                                                                                                                                                                                                                                             | Martin     |              |
| Published                                                                                                                                                                                                                                                                |            |              |

## PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                               |  |                                                                                                                                                                                                                                                                                                                                            |  |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
|                                                                                                                                                                                                                                                                                                                               |  | Trait Category                                                                                                                                                                                                                                                                                                                             |  |
| Morphology, Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^/and+Trait+Category=^Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^/and+Trait+Category=^Physiology^#gephebase-summary-title</a> )                |  |                                                                                                                                                                                                                                                                                                                                            |  |
|                                                                                                                                                                                                                                                                                                                               |  | Trait                                                                                                                                                                                                                                                                                                                                      |  |
| Racing performance ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Racing+performance^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Racing+performance^#gephebase-summary-title</a> )                                                                                      |  |                                                                                                                                                                                                                                                                                                                                            |  |
|                                                                                                                                                                                                                                                                                                                               |  | Trait State in Taxon A                                                                                                                                                                                                                                                                                                                     |  |
| Canis familiaris - Whippet                                                                                                                                                                                                                                                                                                    |  |                                                                                                                                                                                                                                                                                                                                            |  |
|                                                                                                                                                                                                                                                                                                                               |  | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                     |  |
| Canis familiaris - Whippet with muscular hypertrophy                                                                                                                                                                                                                                                                          |  |                                                                                                                                                                                                                                                                                                                                            |  |
|                                                                                                                                                                                                                                                                                                                               |  | Ancestral State                                                                                                                                                                                                                                                                                                                            |  |
| Taxon A                                                                                                                                                                                                                                                                                                                       |  |                                                                                                                                                                                                                                                                                                                                            |  |
|                                                                                                                                                                                                                                                                                                                               |  | Taxonomic Status                                                                                                                                                                                                                                                                                                                           |  |
| 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> )                                                                                  |  |                                                                                                                                                                                                                                                                                                                                            |  |
| Taxon A                                                                                                                                                                                                                                                                                                                       |  | Taxon B                                                                                                                                                                                                                                                                                                                                    |  |
| Latin Name                                                                                                                                                                                                                                                                                                                    |  | Latin Name                                                                                                                                                                                                                                                                                                                                 |  |
| Canis lupus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Canis+lupus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Canis+lupus^#gephebase-summary-title</a> )                                                                                 |  | Canis lupus familiaris ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Canis+lupus+familiaris^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Canis+lupus+familiaris^#gephebase-summary-title</a> )                                                             |  |
| Common Name                                                                                                                                                                                                                                                                                                                   |  | Common Name                                                                                                                                                                                                                                                                                                                                |  |
| gray wolf                                                                                                                                                                                                                                                                                                                     |  | dog                                                                                                                                                                                                                                                                                                                                        |  |
| Synonyms                                                                                                                                                                                                                                                                                                                      |  | Synonyms                                                                                                                                                                                                                                                                                                                                   |  |
| gray wolf; grey wolf; Canis lupus Linnaeus, 1758                                                                                                                                                                                                                                                                              |  | Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758                                                                                                                                                                                                         |  |
| Rank                                                                                                                                                                                                                                                                                                                          |  | Rank                                                                                                                                                                                                                                                                                                                                       |  |
| species                                                                                                                                                                                                                                                                                                                       |  | subspecies                                                                                                                                                                                                                                                                                                                                 |  |
| Lineage                                                                                                                                                                                                                                                                                                                       |  | Lineage                                                                                                                                                                                                                                                                                                                                    |  |
| cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis |  | cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus |  |
| Parent                                                                                                                                                                                                                                                                                                                        |  | Parent                                                                                                                                                                                                                                                                                                                                     |  |
| Canis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611</a> )                                                                                                                                                  |  | Canis lupus (gray wolf) - (Rank: species) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a> )                                                                                                                                              |  |
| NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                              |  | NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                                           |  |
| 9612                                                                                                                                                                                                                                                                                                                          |  | 9615                                                                                                                                                                                                                                                                                                                                       |  |
| ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a> )                                                                                                                                                                           |  | ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a> )                                                                                                                                                                                        |  |
| is Taxon A an Intraspecies?                                                                                                                                                                                                                                                                                                   |  | is Taxon B an Intraspecies?                                                                                                                                                                                                                                                                                                                |  |
| Yes                                                                                                                                                                                                                                                                                                                           |  |                                                                                                                                                                                                                                                                                                                                            |  |
| Taxon A Description                                                                                                                                                                                                                                                                                                           |  | Taxon B Description                                                                                                                                                                                                                                                                                                                        |  |
| Canis familiaris - Whippet                                                                                                                                                                                                                                                                                                    |  | Canis familiaris - Whippet                                                                                                                                                                                                                                                                                                                 |  |

## GENOTYPIC CHANGE

|                                                                                                                                                                                                                                    |                         |                                                                                                                        |                         |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------|------------------------------------------------------------------------------------------------------------------------|-------------------------|
| MSTN                                                                                                                                                                                                                               | Generic Gene Name       | O14793 ( <a href="http://www.uniprot.org/uniprot/O14793">http://www.uniprot.org/uniprot/O14793</a> )                   | UniProtKB Homo sapiens  |
| GDF8; MSLHP                                                                                                                                                                                                                        | Synonyms                | AAR14343 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AAR14343">https://www.ncbi.nlm.nih.gov/nuccore/AAR14343</a> ) | GenebankID or UniProtKB |
| 9606.ENSPP00000260950<br>( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000260950">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000260950</a> ) | String                  |                                                                                                                        |                         |
|                                                                                                                                                                                                                                    | Sequence Similarities   |                                                                                                                        |                         |
| Belongs to the TGF-beta family.                                                                                                                                                                                                    |                         |                                                                                                                        |                         |
| GO:0042802 : identical protein binding<br>( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042802">https://www.ebi.ac.uk/QuickGO/term/GO:0042802</a> )                                                                            | GO - Molecular Function |                                                                                                                        |                         |
| GO:0042803 : protein homodimerization activity<br>( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042803">https://www.ebi.ac.uk/QuickGO/term/GO:0042803</a> )                                                                    |                         |                                                                                                                        |                         |

|                                                                                                                                                                                                                                        |                                   |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------|
| GO:0008201 : heparin binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008201">https://www.ebi.ac.uk/QuickGO/term/GO:0008201</a> )                                                                                             |                                   |
| GO:0005125 : cytokine activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005125">https://www.ebi.ac.uk/QuickGO/term/GO:0005125</a> )                                                                                           |                                   |
| GO:0008083 : growth factor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008083">https://www.ebi.ac.uk/QuickGO/term/GO:0008083</a> )                                                                                      |                                   |
| GO:0005102 : signaling receptor binding                                                                                                                                                                                                |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005102">https://www.ebi.ac.uk/QuickGO/term/GO:0005102</a> )                                                                                                                          |                                   |
| GO:0005160 : transforming growth factor beta receptor binding                                                                                                                                                                          |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005160">https://www.ebi.ac.uk/QuickGO/term/GO:0005160</a> )                                                                                                                          |                                   |
| GO - Biological Process                                                                                                                                                                                                                |                                   |
| GO:0045893 : positive regulation of transcription, DNA-templated                                                                                                                                                                       |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0045893">https://www.ebi.ac.uk/QuickGO/term/GO:0045893</a> )                                                                                                                          |                                   |
| GO:0048468 : cell development ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048468">https://www.ebi.ac.uk/QuickGO/term/GO:0048468</a> )                                                                                            |                                   |
| GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation                                                                                                                                                    |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0010862">https://www.ebi.ac.uk/QuickGO/term/GO:0010862</a> )                                                                                                                          |                                   |
| GO:0042981 : regulation of apoptotic process                                                                                                                                                                                           |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042981">https://www.ebi.ac.uk/QuickGO/term/GO:0042981</a> )                                                                                                                          |                                   |
| GO:0043408 : regulation of MAPK cascade                                                                                                                                                                                                |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043408">https://www.ebi.ac.uk/QuickGO/term/GO:0043408</a> )                                                                                                                          |                                   |
| GO:0060395 : SMAD protein signal transduction                                                                                                                                                                                          |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0060395">https://www.ebi.ac.uk/QuickGO/term/GO:0060395</a> )                                                                                                                          |                                   |
| GO:0043627 : response to estrogen ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043627">https://www.ebi.ac.uk/QuickGO/term/GO:0043627</a> )                                                                                        |                                   |
| GO:0046716 : muscle cell cellular homeostasis                                                                                                                                                                                          |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046716">https://www.ebi.ac.uk/QuickGO/term/GO:0046716</a> )                                                                                                                          |                                   |
| GO:0045471 : response to ethanol ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0045471">https://www.ebi.ac.uk/QuickGO/term/GO:0045471</a> )                                                                                         |                                   |
| GO:0033574 : response to testosterone                                                                                                                                                                                                  |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0033574">https://www.ebi.ac.uk/QuickGO/term/GO:0033574</a> )                                                                                                                          |                                   |
| GO:0007179 : transforming growth factor beta receptor signaling pathway                                                                                                                                                                |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007179">https://www.ebi.ac.uk/QuickGO/term/GO:0007179</a> )                                                                                                                          |                                   |
| GO:0009408 : response to heat ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0009408">https://www.ebi.ac.uk/QuickGO/term/GO:0009408</a> )                                                                                            |                                   |
| GO:0071549 : cellular response to dexamethasone stimulus                                                                                                                                                                               |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0071549">https://www.ebi.ac.uk/QuickGO/term/GO:0071549</a> )                                                                                                                          |                                   |
| GO:0007517 : muscle organ development                                                                                                                                                                                                  |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007517">https://www.ebi.ac.uk/QuickGO/term/GO:0007517</a> )                                                                                                                          |                                   |
| GO:0014839 : myoblast migration involved in skeletal muscle regeneration                                                                                                                                                               |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0014839">https://www.ebi.ac.uk/QuickGO/term/GO:0014839</a> )                                                                                                                          |                                   |
| GO:0046627 : negative regulation of insulin receptor signaling pathway                                                                                                                                                                 |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046627">https://www.ebi.ac.uk/QuickGO/term/GO:0046627</a> )                                                                                                                          |                                   |
| GO:0033673 : negative regulation of kinase activity                                                                                                                                                                                    |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0033673">https://www.ebi.ac.uk/QuickGO/term/GO:0033673</a> )                                                                                                                          |                                   |
| GO:0014741 : negative regulation of muscle hypertrophy                                                                                                                                                                                 |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0014741">https://www.ebi.ac.uk/QuickGO/term/GO:0014741</a> )                                                                                                                          |                                   |
| GO:0045662 : negative regulation of myoblast differentiation                                                                                                                                                                           |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0045662">https://www.ebi.ac.uk/QuickGO/term/GO:0045662</a> )                                                                                                                          |                                   |
| GO:2000818 : negative regulation of myoblast proliferation                                                                                                                                                                             |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:2000818">https://www.ebi.ac.uk/QuickGO/term/GO:2000818</a> )                                                                                                                          |                                   |
| GO:0051898 : negative regulation of protein kinase B signaling                                                                                                                                                                         |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0051898">https://www.ebi.ac.uk/QuickGO/term/GO:0051898</a> )                                                                                                                          |                                   |
| GO:1902725 : negative regulation of satellite cell differentiation                                                                                                                                                                     |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:1902725">https://www.ebi.ac.uk/QuickGO/term/GO:1902725</a> )                                                                                                                          |                                   |
| GO:1902723 : negative regulation of skeletal muscle satellite cell proliferation                                                                                                                                                       |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:1902723">https://www.ebi.ac.uk/QuickGO/term/GO:1902723</a> )                                                                                                                          |                                   |
| GO:0048632 : negative regulation of skeletal muscle tissue growth                                                                                                                                                                      |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048632">https://www.ebi.ac.uk/QuickGO/term/GO:0048632</a> )                                                                                                                          |                                   |
| GO:0022602 : ovulation cycle process                                                                                                                                                                                                   |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0022602">https://www.ebi.ac.uk/QuickGO/term/GO:0022602</a> )                                                                                                                          |                                   |
| GO:0010592 : positive regulation of lamellipodium assembly                                                                                                                                                                             |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0010592">https://www.ebi.ac.uk/QuickGO/term/GO:0010592</a> )                                                                                                                          |                                   |
| GO:0010759 : positive regulation of macrophage chemotaxis                                                                                                                                                                              |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0010759">https://www.ebi.ac.uk/QuickGO/term/GO:0010759</a> )                                                                                                                          |                                   |
| GO:0051602 : response to electrical stimulus                                                                                                                                                                                           |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0051602">https://www.ebi.ac.uk/QuickGO/term/GO:0051602</a> )                                                                                                                          |                                   |
| GO:0009629 : response to gravity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0009629">https://www.ebi.ac.uk/QuickGO/term/GO:0009629</a> )                                                                                         |                                   |
| GO:0014850 : response to muscle activity                                                                                                                                                                                               |                                   |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0014850">https://www.ebi.ac.uk/QuickGO/term/GO:0014850</a> )                                                                                                                          |                                   |
| GO:0014732 : skeletal muscle atrophy ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0014732">https://www.ebi.ac.uk/QuickGO/term/GO:0014732</a> )                                                                                     |                                   |
| GO - Cellular Component                                                                                                                                                                                                                |                                   |
| GO:0005737 : cytoplasm ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005737">https://www.ebi.ac.uk/QuickGO/term/GO:0005737</a> )                                                                                                   |                                   |
| GO:0005615 : extracellular space ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005615">https://www.ebi.ac.uk/QuickGO/term/GO:0005615</a> )                                                                                         |                                   |
|                                                                                                                                                                                                                                        | Presumptive Null                  |
| Yes ( <a +yes+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive+Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+Yes+"#gephebase-summary-title</a> )              |                                   |
|                                                                                                                                                                                                                                        | Molecular Type                    |
| Coding ( <a +coding+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular+Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding+"#gephebase-summary-title</a> )         |                                   |
|                                                                                                                                                                                                                                        | Aberration Type                   |
| Deletion ( <a +deletion+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration+Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+Deletion+"#gephebase-summary-title</a> ) |                                   |
|                                                                                                                                                                                                                                        | Deletion Size                     |
| 1-9 bp                                                                                                                                                                                                                                 |                                   |
|                                                                                                                                                                                                                                        | Molecular Details of the Mutation |
| 2bp deletion in the third exon c.939_940delTG p.C313*                                                                                                                                                                                  |                                   |
|                                                                                                                                                                                                                                        | Experimental Evidence             |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |                       |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------|
| Candidate Gene ( <a href="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</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Main Reference        |
| A mutation in the myostatin gene increases muscle mass and enhances racing performance in heterozygote dogs. (2007) ( <a href="https://pubmed.ncbi.nlm.nih.gov/17530926">https://pubmed.ncbi.nlm.nih.gov/17530926</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Authors               |
| Mosher DS; Quignon P; Bustamante CD; Sutter NB; Mellersh CS; Parker HG; Ostrander EA                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | Abstract              |
| Double muscling is a trait previously described in several mammalian species including cattle and sheep and is caused by mutations in the myostatin (MSTN) gene (previously referred to as GDF8). Here we describe a new mutation in MSTN found in the whippet dog breed that results in a double-muscled phenotype known as the “bully” whippet. Individuals with this phenotype carry two copies of a two-base-pair deletion in the third exon of MSTN leading to a premature stop codon at amino acid 313. Individuals carrying only one copy of the mutation are, on average, more muscular than wild-type individuals ( $p = 7.43 \times 10^{-6}$ ; Kruskal-Wallis Test) and are significantly faster than individuals carrying the wild-type genotype in competitive racing events (Kendall’s nonparametric measure, $\tau = 0.3619$ ; $p$ approximately 0.00028). These results highlight the utility of performance-enhancing polymorphisms, marking the first time a mutation in MSTN has been quantitatively linked to increased athletic performance. | Additional References |

RELATED GEPHE

|                                                                                                                                                                                                                                                                                                                                                                                                                                    |                    |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------|
| 1 (Myosin heavy chain 9) ( <a href="https://www.gephebase.org/search-criteria?/or+Taxon ID=~9612~/and+Trait=Racing performance/or+Taxon ID=~9615~/and+Trait=Racing performance/and+groupHaplotypes=true#gephebase-summary-title">https://www.gephebase.org/search-criteria?/or+Taxon ID=~9612~/and+Trait=Racing performance/or+Taxon ID=~9615~/and+Trait=Racing performance/and+groupHaplotypes=true#gephebase-summary-title</a> ) | Related Genes      |
| No matches found.                                                                                                                                                                                                                                                                                                                                                                                                                  | Related Haplotypes |

EXTERNAL LINKS

COMMENTS

@Parallelism <https://omia.org/OMIA000683/9615/>