

GEPHE SUMMARY

|                                                                                                                                                                                                                                                                                                                                                   |                |              |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|--------------|
| Insulin-like growth factor receptor 1 (IGF1R) ( <a +insulin-like+growth+factor+receptor+1+(igf1r)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Insulin-like+growth+factor+receptor+1+(IGF1R)^#gephebase-summary-title</a> ) | Gephebase Gene | GepheID      |
|                                                                                                                                                                                                                                                                                                                                                   | GP00001479     |              |
|                                                                                                                                                                                                                                                                                                                                                   | Prigent        | Main curator |
| Published                                                                                                                                                                                                                                                                                                                                         | Entry Status   |              |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                               |  |                                                                                                                                                                                                                                                                                                                                                                                  |  |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
|                                                                                                                                                                                                                                                                                                                               |  | Trait Category                                                                                                                                                                                                                                                                                                                                                                   |  |
| Morphology ( <a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology^#gephebase-summary-title</a> )                                                                                      |  | Trait                                                                                                                                                                                                                                                                                                                                                                            |  |
| Body size (weight) ( <a +body+size+(weight)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Body+size+(weight)^#gephebase-summary-title</a> )                                                                                |  | Trait State in Taxon A                                                                                                                                                                                                                                                                                                                                                           |  |
| Gray wolf of 38 to 66 kg                                                                                                                                                                                                                                                                                                      |  | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                                                           |  |
| Dog of 4.6 kg on average                                                                                                                                                                                                                                                                                                      |  | Ancestral State                                                                                                                                                                                                                                                                                                                                                                  |  |
| Taxon A                                                                                                                                                                                                                                                                                                                       |  | Taxonomic Status                                                                                                                                                                                                                                                                                                                                                                 |  |
| Domesticated ( <a +domesticated^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">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 +canis+lupus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus^#gephebase-summary-title</a> )                                                                           |  | Homo sapiens ( <a +homo+sapiens^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens^#gephebase-summary-title</a> )                                                                                                                           |  |
| Common Name                                                                                                                                                                                                                                                                                                                   |  | Common Name                                                                                                                                                                                                                                                                                                                                                                      |  |
| gray wolf                                                                                                                                                                                                                                                                                                                     |  | human                                                                                                                                                                                                                                                                                                                                                                            |  |
| Synonyms                                                                                                                                                                                                                                                                                                                      |  | Synonyms                                                                                                                                                                                                                                                                                                                                                                         |  |
| gray wolf; grey wolf; Canis lupus Linnaeus, 1758                                                                                                                                                                                                                                                                              |  | human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens                                                                                                                                                   |  |
| Rank                                                                                                                                                                                                                                                                                                                          |  | Rank                                                                                                                                                                                                                                                                                                                                                                             |  |
| species                                                                                                                                                                                                                                                                                                                       |  | species                                                                                                                                                                                                                                                                                                                                                                          |  |
| 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; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo |  |
| 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> )                                                                                                                                                  |  | Homo () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )                                                                                                                                                                                                      |  |
| NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                              |  | NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                                                                                 |  |
| 9612 ( <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> )                                                                                                                                                                      |  | 9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )                                                                                                                                                                                                                         |  |
| is Taxon A an Intraspecies?                                                                                                                                                                                                                                                                                                   |  | is Taxon B an Intraspecies?                                                                                                                                                                                                                                                                                                                                                      |  |
| No                                                                                                                                                                                                                                                                                                                            |  | No                                                                                                                                                                                                                                                                                                                                                                               |  |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                              |  |                                                                                                                                                         |  |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---------------------------------------------------------------------------------------------------------------------------------------------------------|--|
|                                                                                                                                                                                                                              |  | Generic Gene Name                                                                                                                                       |  |
| IGF1R                                                                                                                                                                                                                        |  | UniProtKB Homo sapiens P08069 ( <a href="http://www.uniprot.org/uniprot/P08069">http://www.uniprot.org/uniprot/P08069</a> )                             |  |
| Synonyms                                                                                                                                                                                                                     |  | GenebankID or UniProtKB XM_545828.5 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/XM_545828.5">https://www.ncbi.nlm.nih.gov/nuccore/XM_545828.5</a> ) |  |
| String                                                                                                                                                                                                                       |  |                                                                                                                                                         |  |
| 9606.ENSPO0000268035 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000268035">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000268035</a> ) |  |                                                                                                                                                         |  |
| Sequence Similarities                                                                                                                                                                                                        |  |                                                                                                                                                         |  |
| Belongs to the protein kinase superfamily. Tyr protein kinase family. Insulin receptor subfamily.                                                                                                                            |  |                                                                                                                                                         |  |
| GO - Molecular Function                                                                                                                                                                                                      |  |                                                                                                                                                         |  |
| GO:0004888 : transmembrane signaling receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004888">https://www.ebi.ac.uk/QuickGO/term/GO:0004888</a> )                                                         |  |                                                                                                                                                         |  |

GO:0005524 : ATP binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005524>)

GO:0042802 : identical protein binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042802>)

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:0043559 : insulin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0043559>)

GO:0043560 : insulin receptor substrate binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043560>)

GO:0005009 : insulin-activated receptor activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005009>)

GO:0043548 : phosphatidylinositol 3-kinase binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043548>)

GO:0005158 : insulin receptor binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005158>)

GO:0005520 : insulin-like growth factor binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005520>)

GO:0031994 : insulin-like growth factor I binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031994>)

GO:0005010 : insulin-like growth factor-activated receptor activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005010>)

#### GO - Biological Process

GO:0006955 : immune response (<https://www.ebi.ac.uk/QuickGO/term/GO:0006955>)

GO:0007165 : signal transduction (<https://www.ebi.ac.uk/QuickGO/term/GO:0007165>)

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:0070374 : positive regulation of ERK1 and ERK2 cascade

(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)

GO:0008284 : positive regulation of cell proliferation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)

GO:0009968 : negative regulation of signal transduction

(<https://www.ebi.ac.uk/QuickGO/term/GO:0009968>)

GO:0008286 : insulin receptor signaling pathway

(<https://www.ebi.ac.uk/QuickGO/term/GO:0008286>)

GO:0048009 : insulin-like growth factor receptor signaling pathway

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048009>)

GO:0046777 : protein autophosphorylation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)

GO:0120162 : positive regulation of cold-induced thermogenesis

(<https://www.ebi.ac.uk/QuickGO/term/GO:0120162>)

GO:0030335 : positive regulation of cell migration

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030335>)

GO:0042593 : glucose homeostasis (<https://www.ebi.ac.uk/QuickGO/term/GO:0042593>)

GO:1904646 : cellular response to amyloid-beta

(<https://www.ebi.ac.uk/QuickGO/term/GO:1904646>)

GO:0014065 : phosphatidylinositol 3-kinase signaling

(<https://www.ebi.ac.uk/QuickGO/term/GO:0014065>)

GO:0048015 : phosphatidylinositol-mediated signaling

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048015>)

GO:0097242 : amyloid-beta clearance

(<https://www.ebi.ac.uk/QuickGO/term/GO:0097242>)

GO:0097062 : dendritic spine maintenance

(<https://www.ebi.ac.uk/QuickGO/term/GO:0097062>)

GO:0051389 : inactivation of MAPKK activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0051389>)

GO:0038083 : peptidyl-tyrosine autophosphorylation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0038083>)

GO:0043243 : positive regulation of protein complex disassembly

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043243>)

GO:0051262 : protein tetramerization (<https://www.ebi.ac.uk/QuickGO/term/GO:0051262>)

GO:0046328 : regulation of JNK cascade

(<https://www.ebi.ac.uk/QuickGO/term/GO:0046328>)

#### 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:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)

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:0030424 : axon (<https://www.ebi.ac.uk/QuickGO/term/GO:0030424>)

GO:0043231 : intracellular membrane-bounded organelle

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)

GO:0005899 : insulin receptor complex

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005899>)

GO:0035867 : alphav-beta3 integrin-IGF-1-IGF1R complex

(<https://www.ebi.ac.uk/QuickGO/term/GO:0035867>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>)

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

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>)

Nonsynonymous

G>A p.R204H in exon 2

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

|            | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon      | -       | -       | -        |
| Amino-acid | -       | -       | -        |

Derived variants at six genes explain nearly half of size reduction in dog breeds. (2013) (<https://pubmed.ncbi.nlm.nih.gov/24026177>)

Rimbault M; Beale HC; Schoenebeck JJ; Hoopes BC; Allen JJ; Kilroy-Glynn P; Wayne RK; Sutter NB; Ostrander EA

Main Reference

Authors

Abstract

Selective breeding of dogs by humans has generated extraordinary diversity in body size. A number of multibreed analyses have been undertaken to identify the genetic basis of this diversity. We analyzed four loci discovered in a previous genome-wide association study that used 60,968 SNPs to identify size-associated genomic intervals, which were too large to assign causative roles to genes. First, we performed fine-mapping to define critical intervals that included the candidate genes GHR, HMGA2, SMAD2, and STC2, identifying five highly associated markers at the four loci. We hypothesize that three of the variants are likely to be causative. We then genotyped each marker, together with previously reported size-associated variants in the IGF1 and IGF1R genes, on a panel of 500 domestic dogs from 93 breeds, and identified the ancestral allele by genotyping the same markers on 30 wild canids. We observed that the derived alleles at all markers correlated with reduced body size, and smaller dogs are more likely to carry derived alleles at multiple markers. However, breeds are not generally fixed at all markers; multiple combinations of genotypes are found within most breeds. Finally, we show that 46%-52.5% of the variance in body size of dog breeds can be explained by seven markers in proximity to exceptional candidate genes. Among breeds with standard weights <41 kg (90 lb), the genotypes accounted for 64.3% of variance in weight. This work advances our understanding of mammalian growth by describing genetic contributions to canine size determination in non-giant dog breeds.

Additional References

RELATED GEPHE

5 (growth hormone receptor (GHR), HMGA2, Insulin-like growth factor 1 (IGF1), SMAD family member 2 (SMAD2), stanniocalcin 2 (STC2)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9612^/and+Trait=Body size/or+Taxon ID=^9606^/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title>)

No matches found.

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS

each variant is potentially causal