GEPHE SUMMARY

Gephebase Gene
Insulin-like growth factor 2 (IGF2) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Insulin-like growth factor 2 (IGF2)^#gephebase-summary-title)

GepheID
GP00000502

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait #1

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Muscular mass (https://www.gephebase.org/search-criteria?/and+Trait=^Muscular mass^#gephebase-summary-title)

Trait State in Taxon A
Sus scrofa

Trait State in Taxon B
Sus scrofa

Trait #2

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Fat deposition (https://www.gephebase.org/search-criteria?/and+Trait=^Fat deposition^#gephebase-summary-title)

Trait State in Taxon A
-

Trait State in Taxon B
-

Trait #3

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Heart size (https://www.gephebase.org/search-criteria?/and+Trait=^Heart size^#gephebase-summary-title)

Trait State in Taxon A
-

Trait State in Taxon B
-

Ancestral State
Data not curated

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Sus scrofa^#gephebase-summary-title)

Common Name
pig

Synonyms
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus

Taxon B

Latin Name
Sus scrofa domesticus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Sus scrofa domesticus^#gephebase-summary-title)

Common Name
domestic pig

Synonyms
Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig

Rank
subspecies

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus; Sus scrofa

Parent

Sus () - (Rank: genus)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)

NCBI Taxonomy ID

9823
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)

is Taxon A an Infraspecies?

No

Parent

Sus scrofa (pig) - (Rank: species)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)

NCBI Taxonomy ID

9825
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Igf2

Synonyms

Mpr; M6pr; Peg2; Igf-2; Igf-II; AL033362

String

10090.ENSMUSP00000114076
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=
10090.ENSMUSP00000114076)

Sequence Similarities

Belongs to the insulin family.

GO - Molecular Function

GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)
GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)
GO:0005158 : insulin receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0005158)
GO:0005159 : insulin-like growth factor receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0005159)
GO:0005178 : integrin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005178)
GO:0043539 : protein serine/threonine kinase activator activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0043539)
GO:0048018 : receptor ligand activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0048018)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0050731 : positive regulation of peptidyl-tyrosine phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050731)
GO:0051897 : positive regulation of protein kinase B signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0051897)
GO:0001701 : in utero embryonic development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001701)
GO:0043410 : positive regulation of MAPK cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0043410)
GO:0009887 : animal organ morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0009887)
GO:0001649 : osteoblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001649)
GO:0042493 : response to drug (https://www.ebi.ac.uk/QuickGO/term/GO:0042493)
GO:0014070 : response to organic cyclic compound
(https://www.ebi.ac.uk/QuickGO/term/GO:0014070)
GO:0035094 : response to nicotine (https://www.ebi.ac.uk/QuickGO/term/GO:0035094)
GO:0045840 : positive regulation of mitotic nuclear division
(https://www.ebi.ac.uk/QuickGO/term/GO:0045840)
GO:0051781 : positive regulation of cell division
(https://www.ebi.ac.uk/QuickGO/term/GO:0051781)
GO:0001934 : positive regulation of protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001934)
GO:0001892 : embryonic placenta development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001892)
GO:0040018 : positive regulation of multicellular organism growth
(https://www.ebi.ac.uk/QuickGO/term/GO:0040018)
GO:1905564 : positive regulation of vascular endothelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:1905564)
GO:0006006 : glucose metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006006)
GO:0007613 : memory (https://www.ebi.ac.uk/QuickGO/term/GO:0007613)
GO:0045766 : positive regulation of angiogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0045766)
GO:0031017 : exocrine pancreas development
(https://www.ebi.ac.uk/QuickGO/term/GO:0031017)
GO:0042104 : positive regulation of activated T cell proliferation

UniProtKB Mus musculus

P09535 (http://www.uniprot.org/uniprot/P09535)

GenebankID or UniProtKB

ABW24500 (https://www.ncbi.nlm.nih.gov/nuccore/ABW24500)

(https://www.ebi.ac.uk/QuickGO/term/GO:0042104)
GO:0043536 : positive regulation of blood vessel endothelial cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0043536)
GO:0045725 : positive regulation of glycogen biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045725)
GO:0090031 : positive regulation of steroid hormone biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0090031)
GO:0060669 : embryonic placenta morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060669)
GO:0038028 : insulin receptor signaling pathway via phosphatidylinositol 3-kinase
(https://www.ebi.ac.uk/QuickGO/term/GO:0038028)
GO:0045953 : negative regulation of natural killer cell mediated cytotoxicity
(https://www.ebi.ac.uk/QuickGO/term/GO:0045953)
GO:0043085 : positive regulation of catalytic activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0043085)
GO:2000467 : positive regulation of glycogen (starch) synthase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:2000467)
GO:0046628 : positive regulation of insulin receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0046628)
GO:0031056 : regulation of histone modification
(https://www.ebi.ac.uk/QuickGO/term/GO:0031056)
GO:0051147 : regulation of muscle cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0051147)
GO:0051146 : striated muscle cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0051146)

GO - Cellular Component

GO:0062023 : collagen-containing extracellular matrix
(https://www.ebi.ac.uk/QuickGO/term/GO:0062023)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

1bp change

Experimental Evidence

Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)

Main Reference

A regulatory mutation in IGF2 causes a major QTL effect on muscle growth in the pig. (2003) (https://pubmed.ncbi.nlm.nih.gov/14574411)

Authors

Van Laere AS; Nguyen M; Braunschweig M; Nezer C; Collette C; Moreau L; Archibald AL; Haley CS; Buys N; Tally M; Andersson G; Georges M; Andersson L

Abstract

Most traits and disorders have a multifactorial background indicating that they are controlled by environmental factors as well as an unknown number of quantitative trait loci (QTLs). The identification of mutations underlying QTLs is a challenge because each locus explains only a fraction of the phenotypic variation. A paternally expressed QTL affecting muscle growth, fat deposition and size of the heart in pigs maps to the IGF2 (insulin-like growth factor 2) region. Here we show that this QTL is caused by a nucleotide substitution in intron 3 of IGF2. The mutation occurs in an evolutionarily conserved CpG island that is hypomethylated in skeletal muscle. The mutation abrogates in vitro interaction with a nuclear factor, probably a repressor, and pigs inheriting the mutation from their sire have a threefold increase in IGF2 messenger RNA expression in postnatal muscle. Our study establishes a causal relationship between a single-base-pair substitution in a non-coding region and a QTL effect. The result supports the long-held view that regulatory mutations are important for controlling phenotypic variation.

Additional References

RELATED GEPHE

Related Genes

1 (Myostatin (MSTN = GDF8)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9823^/and+Trait=Muscular mass/or+Taxon ID=^9823^/and+Trait=Fat deposition/or+Taxon ID=^9823^/and+Trait=Heart size/or+Taxon ID=^9825^/and+Trait=Muscular mass/or+Taxon ID=^9825^/and+Trait=Fat deposition/or+Taxon ID=^9825^/and+Trait=Heart size/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS