

GEPHE SUMMARY

Insulin-like growth factor 2 (IGF2) (<a +insulin-like+growth+factor+2+(igf2)+"#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+2+(IGF2)+"#gephebase-summary-title)		Gephebase Gene	GP00000502	GepheID
Published		Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Trait #1	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)	
Muscular mass (<a +muscular+mass+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Muscular+mass+"#gephebase-summary-title)	
Sus scrofa	
Sus scrofa	

Trait #2	Trait Category
Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title)	
Fat deposition (<a +fat+deposition+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Fat+deposition+"#gephebase-summary-title)	
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Trait #3	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)	
Heart size (<a +heart+size+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Heart+size+"#gephebase-summary-title)	
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Data not curated		Ancestral State	
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)		Taxonomic Status	
Sus scrofa		Sus scrofa domesticus	
(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Sus+scrofa+"#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Sus+scrofa+domesticus+"#gephebase-summary-title)	
pig		domestic pig	
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus		Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	
species		subspecies	
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		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		Parent
Sus () - (Rank: genus)		Sus scrofa (pig) - (Rank: species)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9823		9825	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Igf2	P09535 (http://www.uniprot.org/uniprot/P09535)	GenebankID or UniProtKB
Mpr; M6pr; Peg2; Igf-2; Igf-II; AL033362	ABW24500 (https://www.ncbi.nlm.nih.gov/nuccore/ABW24500)	
	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	

(<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

