

GEPHE SUMMARY

	Gephebase Gene	GephelD
Callipyge (CLPG1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Callipyge (CLPG1)^#gephebase-summary-title)	GP00000168	
Published	Entry Status	Main curator

PHENOTYPIC CHANGE

	Trait Category
Morphology, Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^/and+Trait Category=^Physiology^#gephebase-summary-title)	Trait
Muscular mass (double-muscling : paternal effect) (https://www.gephebase.org/search-criteria?/and+Trait=^Muscular mass (double-muscling : paternal effect)^#gephebase-summary-title)	Trait State in Taxon A
Ovis aries	Trait State in Taxon B
Ovis aries; Callipyge	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	Taxon B	Latin Name
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)		Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)	
sheep	Common Name	sheep	Common Name
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Synonyms	Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis	Lineage
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	Parent	Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	Parent
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	NCBI Taxonomy ID	9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	NCBI Taxonomy ID
-	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
No		Ovis aries; Callipyge in Rambouillet breed	Taxon B Description

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Ovis aries
DIO3		Q6DN07 (http://www.uniprot.org/uniprot/Q6DN07)
5DIII; DIOIII	Synonyms	AY656759 (https://www.ncbi.nlm.nih.gov/nuccore/AY656759)
-	String	
Belongs to the iodothyronine deiodinase family.	Sequence Similarities	
GO:0004800 : thyroxine 5'-deiodinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004800)	GO - Molecular Function	
GO:0042446 : hormone biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042446)	GO - Biological Process	

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)

GO:0010008 : endosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=%22No%22#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=%22Cis-regulatory%22#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=%22SNP%22#gephebase-summary-title>)

Molecular Details of the Mutation

1bp change in the regulatory region of the DLK1-GTL2 imprinted gene cluster

Experimental Evidence

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=%22Linkage+Mapping%22#gephebase-summary-title>)

Main Reference

The callipyge mutation enhances bidirectional long-range DLK1-GTL2 intergenic transcription in cis. (2006) (<https://pubmed.ncbi.nlm.nih.gov/16690740>)

Authors

Takeda H; Caument F; Smit M; Hiard S; Tordoir X; Cockett N; Georges M; Charlier C

Abstract

The callipyge mutation (CLPG) is an A to G transition that affects a muscle-specific long-range control element located in the middle of the 90-kb DLK1-GTL2 intergenic (IG) region. It causes ectopic expression of a 327-kb cluster of imprinted genes in skeletal muscle, resulting in the callipyge muscular hypertrophy and its non-Mendelian inheritance pattern known as polar overdominance. We herein demonstrate that the CLPG mutation alters the muscular epigenotype of the DLK1-GTL2 IG region in cis, including hypomethylation, acquisition of novel DNase-I hypersensitive sites, and, most strikingly, strongly enhanced bidirectional, long-range IG transcription. The callipyge phenotype thus emerges as a unique model to study the functional significance of IG transcription, which recently has proven to be a widespread, yet elusive, feature of the mammalian genome.

Additional References

Identification of the single base change causing the callipyge muscle hypertrophy phenotype, the only known example of polar overdominance in mammals. (2002) (<https://pubmed.ncbi.nlm.nih.gov/12368241>)

Callipyge mutation affects gene expression in cis: a potential role for chromatin structure. (2006) (<https://pubmed.ncbi.nlm.nih.gov/16415109>)

Mosaicism of Solid Gold supports the causality of a noncoding A-to-G transition in the determinism of the callipyge phenotype. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12586730>)

The callipyge locus: evidence for the trans interaction of reciprocally imprinted genes. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12711215>)

Regulation of DLK1 by the maternally expressed miR-379/miR-544 cluster may underlie callipyge polar overdominance inheritance. (2015) (<https://pubmed.ncbi.nlm.nih.gov/26487685>)

RELATED GEPHE

Related Genes

1 (Myostatin (MSTN = GDF8)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=%229940%22+and+Trait=Muscular+mass+and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Possible miRNA-mediated effect of the Callipyge mutation proposed in mouse ; <https://omia.org/OMIA001354/9940/>