

GEPHE SUMMARY

	Gephebase Gene		GepheID
Callipyge (CLPG1) (<a +callipyge+(clpg1)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Callipyge+(CLPG1)+"#gephebase-summary-title)		GP00000168	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology, Physiology (<a #gephebase-summary-title"="" +morphology+"="" and+trait+category="+Physiology+" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"/and+Trait+Category="+Physiology+"#gephebase-summary-title)			
	Trait		
Muscular mass (double-muscling ; paternal effect) (<a +muscular+mass+(double-muscling+;+paternal+effect)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">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 (<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)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Ovis aries (<a +ovis+aries+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Ovis+aries+"#gephebase-summary-title)		Ovis aries (<a +ovis+aries+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Ovis+aries+"#gephebase-summary-title)	
	Common Name		Common Name
sheep		sheep	
	Synonyms		Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758		Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	
	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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis		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	
	Parent		Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)		Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)		9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Ovis aries; Callipyge in Rambouillet breed	

GENOTYPIC CHANGE

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

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>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	Aberration Type
1bp change in the regulatory region of the DLK1-GTL2 imprinted gene cluster	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence
The callipyge mutation enhances bidirectional long-range DLK1-GTL2 intergenic transcription in cis. (2006) (https://pubmed.ncbi.nlm.nih.gov/16690740)	Main Reference
Takeda H; Caiment F; Smit M; Hiard S; Tordoir X; Cockett N; Georges M; Charlier C	Authors

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.

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)	Additional References
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

1 (Myostatin (MSTN = GDF8)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9940~/and+Trait=Muscular mass/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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