

GEPHE SUMMARY

FOX L2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^FOX L2^#gephebase-summary-title)		Gephebase Gene	GP00000352	GepheID
Published		Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait
Horns absence (https://www.gephebase.org/search-criteria?/and+Trait=^Horns absence^#gephebase-summary-title)	Trait State in Taxon A
Capra aegagrus hircus	Trait State in Taxon B
Capra aegagrus hircus	

Trait #2	Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Physiology^#gephebase-summary-title)	Trait
Somatic sex change (https://www.gephebase.org/search-criteria?/and+Trait=^Somatic sex change^#gephebase-summary-title)	Trait State in Taxon A
-	Trait State in Taxon B
-	

Taxon A		Taxon B	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title)			
Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)	goat	Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)	goat
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species		Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra	
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)	9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)	Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)	9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
Yes		Yes	
Capra aegagrus hircus		Capra aegagrus hircus	

GENOTYPIC CHANGE

FOXL2	Generic Gene Name	Q8MIP2 (http://www.uniprot.org/uniprot/Q8MIP2)	UniProtKB Capra hircus
	Synonyms		GenebankID or UniProtKB
	String	AY112725 (https://www.ncbi.nlm.nih.gov/nucleotide/AY112725)	
	Sequence Similarities		
-			
-			
-			
GO - Molecular Function			
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)			
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)			
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)			
GO:0043028 : cysteine-type endopeptidase regulator activity involved in apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043028)			
GO - Biological Process			
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)			
GO:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)			
GO:0001541 : ovarian follicle development (https://www.ebi.ac.uk/QuickGO/term/GO:0001541)			
GO:0008210 : estrogen metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0008210)			
GO:0019101 : female somatic sex determination (https://www.ebi.ac.uk/QuickGO/term/GO:0019101)			
GO:0006309 : apoptotic DNA fragmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0006309)			
GO:0048048 : embryonic eye morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0048048)			
GO:0002074 : extraocular skeletal muscle development (https://www.ebi.ac.uk/QuickGO/term/GO:0002074)			
GO:0060014 : granulosa cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0060014)			
GO:0043065 : positive regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043065)			
GO:0043280 : positive regulation of cysteine-type endopeptidase activity involved in apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043280)			
GO - Cellular Component			
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)			
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
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)			Aberration Type
10-100 kb			Deletion Size
11.7 kbp deletion of mainly repetitive sequences ; Alters transcription of two flanking genes; but FOXL2 in particular is proposed as the main determinant of the phenotype			Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)			Experimental Evidence
A 11.7-kb deletion triggers intersexuality and polledness in goats. (2001) (https://pubmed.ncbi.nlm.nih.gov/11726932)			Main Reference
Pailhoux E; Vigier B; Chaffaux S; Servel N; Taourit S; Furet JP; Fellous M; Grosclaude F; Cribiu EP; Cotinot C; Vaiman D			Authors
Mammalian sex determination is governed by the presence of the sex determining region Y gene (SRY) on the Y chromosome. Familial cases of SRY-negative XX sex reversal are rare in humans, often hampering the discovery of new sex-determining genes. The mouse model is also insufficient to correctly apprehend the sex-determination cascade, as the human pathway is much more sensitive to gene dosage. Other species might therefore be considered in this respect. In goats, the polled intersex syndrome (PIS) mutation associates polledness and intersexuality. The sex reversal affects exclusively the XX individuals in a recessive manner, whereas the absence of horns is dominant in both sexes. The syndrome is caused by an autosomal gene located at chromosome band 1q43 (ref. 9), shown to be homologous to human chromosome band 3q23 (ref. 10). Through a positional cloning approach, we demonstrate that the mutation underlying PIS is the deletion of a critical 11.7-kb DNA element containing mainly repetitive sequences. This deletion affects the transcription of at least two genes: PISRT1, encoding a 1.5-kb mRNA devoid of open reading frame (ORF), and FOXL2, recently shown to be responsible for blepharophimosis ptosis epicanthus inversus syndrome (BPES) in humans. These two genes are located 20 and 200 kb telomeric from the deletion, respectively.			Abstract
R-spondin1 and FOXL2 act into two distinct cellular types during goat ovarian differentiation. (2008) (https://pubmed.ncbi.nlm.nih.gov/18384673) FOXL2 is a female sex-determining gene in the goat. (2014) (https://pubmed.ncbi.nlm.nih.gov/24485832)			Additional References

RELATED GEPHE

No matches found.	Related Genes
-------------------	---------------

No matches found.

EXTERNAL LINKS

COMMENTS

@Pleiotropy @SexualTrait <https://omia.org/OMIA000483/9925/>