

GEPHE SUMMARY

ABCG2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~ABCG2~#gephebase-summary-title)	Gephebase Gene	GP00000015	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Physiology~#gephebase-summary-title)		Trait Category	
Milk yield (https://www.gephebase.org/search-criteria?/and+Trait=~Milk yield~#gephebase-summary-title)		Trait	
Bos bovis		Trait State in Taxon A	
Bos bovis		Trait State in Taxon B	
Data not curated		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Bos taurus~#gephebase-summary-title)		Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Bos taurus~#gephebase-summary-title)	
cattle		cattle	
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus		Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	
species		species	
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; Bovinae; Bos		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; Bovinae; Bos	
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)		Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)	
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)		9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Abcg2		Generic Gene Name	
MXR; ABCP; BCRP; MXR1; ABC15; Bcrp1; Al428558; Abcp		Q7TMS5 (http://www.uniprot.org/uniprot/Q7TMS5)	
10090.ENSMUSP00000031822 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031822)		AFH35515 (https://www.ncbi.nlm.nih.gov/nuccore/AFH35515)	
Belongs to the ABC transporter superfamily. ABCG family. Eye pigment precursor importer (TC 3.A.1.204) subfamily.		UniProtKB Mus musculus GenebankID or UniProtKB	
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524) GO:0042626 : ATPase activity, coupled to transmembrane movement of substances (https://www.ebi.ac.uk/QuickGO/term/GO:0042626) GO:0008092 : cytoskeletal protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008092)		GO - Molecular Function	

GO:0015238 : drug transmembrane transporter activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0015238>)
GO:0042802 : identical protein binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042802>)
GO:0046983 : protein dimerization activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046983>)
GO:0042803 : protein homodimerization activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
GO:0008559 : xenobiotic transmembrane transporting ATPase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008559>)

GO - Biological Process

GO:0055085 : transmembrane transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0055085>)
GO:0033344 : cholesterol efflux (<https://www.ebi.ac.uk/QuickGO/term/GO:0033344>)
GO:0046618 : drug export (<https://www.ebi.ac.uk/QuickGO/term/GO:0046618>)
GO:0015893 : drug transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015893>)
GO:0060136 : embryonic process involved in female pregnancy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060136>)
GO:0019389 : glucuronoside metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019389>)
GO:1904479 : negative regulation of intestinal absorption
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904479>)
GO:0046415 : urate metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046415>)
GO:0015747 : urate transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015747>)

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:0016324 : apical plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016324>)
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)
GO:0031966 : mitochondrial membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031966>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	Aberration Type
Nonsynonymous	SNP Coding Change
Tyr581Ser	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Identification of a missense mutation in the bovine ABCG2 gene with a major effect on the QTL on chromosome 6 affecting milk yield and composition in Holstein cattle. (2005) (https://pubmed.ncbi.nlm.nih.gov/15998908)	Main Reference
Cohen-Zinder M; Seroussi E; Larkin DM; Loor JJ; Everts-van der Wind A; Lee JH; Drackley JK; Band MR; Hernandez AG; Shani M; Lewin HA; Weller JL; Ron M	Authors
We previously localized a quantitative trait locus (QTL) on chromosome 6 affecting milk fat and protein concentration to a 4-cM confidence interval, centered on the microsatellite BM143. We characterized the genes and sequence variation in this region and identified common haplotypes spanning five polymorphic sites in the genes IBSP, SPP1, PKD2, and ABCG2 for two sires heterozygous for this QTL. Expression of SPP1 and ABCG2 in the bovine mammary gland increased from parturition through lactation. SPP1 and all the coding exons of ABCG2 and PKD2 were sequenced for these two sires. The single nucleotide change capable of encoding a substitution of tyrosine-581 to serine (Y581S) in the ABCG2 transporter was the only polymorphism corresponding to the segregation status of all 3 heterozygous and 15 homozygous sires for the QTL in the Israeli and U.S. Holstein populations. The allele substitution fixed effects on the genetic evaluations of 335 Israeli sires were -341 kg milk, +0.16% fat, and +0.13% protein (F-value = 200). No other polymorphism gave significant effect for fat and protein concentration in models that also included Y581S. The allele substitution effects on the genetic evaluations of 670 cows, daughters of two heterozygous sires, were -226 kg milk, 0.09% fat, and 0.08% protein (F-value = 394), with partial dominance towards the 581S homozygotes. We therefore propose that Y581S in ABCG2 is the causative site for this QTL.	Abstract
Genetic support for a quantitative trait nucleotide in the ABCG2 gene affecting milk composition of dairy cattle. (2007) (https://pubmed.ncbi.nlm.nih.gov/17584938)	Additional References

2 (Growth Hormone Receptor, Prolactin receptor) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Milk yield/and+groupHaplotypes=true#gephebase-summary-title>)

No matches found.

EXTERNAL LINKS

COMMENTS