

GEPHE SUMMARY

ABCA1 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^ABCA1^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^ABCA1^#gephebase-summary-title</a> )	Gephebase Gene	GP00001682	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology, Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^/and+Trait+Category=^Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^/and+Trait+Category=^Physiology^#gephebase-summary-title</a> )	Trait
Coloration (feathers) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+feathers^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+feathers^#gephebase-summary-title</a> )	Trait State in Taxon A
Gallus gallus	Trait State in Taxon B
Wisconsin hypoalpha mutant (WHAM) chicken; white	

Trait #2	Trait Category
Morphology, Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^/and+Trait+Category=^Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^/and+Trait+Category=^Physiology^#gephebase-summary-title</a> )	Trait
Cholesterol metabolism ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Cholesterol+metabolism^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Cholesterol+metabolism^#gephebase-summary-title</a> )	Trait State in Taxon A
Gallus gallus	Trait State in Taxon B
Wisconsin hypoalpha mutant (WHAM) chicken; >90% reduction in plasma HDL due to hypercatabolism by the kidney of lipid-poor apoA-I	

Taxon A	Ancestral State
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title</a> )	Taxonomic Status

Taxon A	Latin Name
Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus^#gephebase-summary-title</a> )	Gallus gallus
chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus	Lineage
Gallus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030</a> )	Parent
9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031</a> )	NCBI Taxonomy ID
is Taxon A an Intraspecies?	
No	

Taxon B	Latin Name
Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus^#gephebase-summary-title</a> )	Gallus gallus
chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus	Lineage
Gallus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030</a> )	Parent
9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031</a> )	NCBI Taxonomy ID
is Taxon B an Intraspecies?	
Yes	
Wisconsin hypoalpha mutant (WHAM) chicken	Taxon B Description

GENOTYPIC CHANGE

<p>Abca1</p> <p>Abc1; ABC-1</p> <p>10090.ENSMUSP00000030010 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000030010">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000030010</a>)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>P41233 (<a href="http://www.uniprot.org/uniprot/P41233">http://www.uniprot.org/uniprot/P41233</a>)</p> <p>0</p> <p>Belongs to the ABC transporter superfamily. ABCA family.</p> <p>GO:0005524 : ATP binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005524">https://www.ebi.ac.uk/QuickGO/term/GO:0005524</a>)            GO:0042626 : ATPase activity, coupled to transmembrane movement of substances (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042626">https://www.ebi.ac.uk/QuickGO/term/GO:0042626</a>)            GO:0090554 : phosphatidylcholine-translocating ATPase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0090554">https://www.ebi.ac.uk/QuickGO/term/GO:0090554</a>)            GO:0017127 : cholesterol transporter activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0017127">https://www.ebi.ac.uk/QuickGO/term/GO:0017127</a>)            GO:0005319 : lipid transporter activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005319">https://www.ebi.ac.uk/QuickGO/term/GO:0005319</a>)            GO:0051117 : ATPase binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0051117">https://www.ebi.ac.uk/QuickGO/term/GO:0051117</a>)            GO:0005102 : signaling receptor binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005102">https://www.ebi.ac.uk/QuickGO/term/GO:0005102</a>)            GO:0016887 : ATPase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0016887">https://www.ebi.ac.uk/QuickGO/term/GO:0016887</a>)            GO:0019905 : syntaxin binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0019905">https://www.ebi.ac.uk/QuickGO/term/GO:0019905</a>)            GO:0034186 : apolipoprotein A-I binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0034186">https://www.ebi.ac.uk/QuickGO/term/GO:0034186</a>)            GO:0034185 : apolipoprotein binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0034185">https://www.ebi.ac.uk/QuickGO/term/GO:0034185</a>)            GO:0008035 : high-density lipoprotein particle binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008035">https://www.ebi.ac.uk/QuickGO/term/GO:0008035</a>)            GO:0008509 : anion transmembrane transporter activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008509">https://www.ebi.ac.uk/QuickGO/term/GO:0008509</a>)            GO:0034188 : apolipoprotein A-I receptor activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0034188">https://www.ebi.ac.uk/QuickGO/term/GO:0034188</a>)            GO:0090556 : phosphatidylserine-translocating ATPase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0090556">https://www.ebi.ac.uk/QuickGO/term/GO:0090556</a>)            GO:0005548 : phospholipid transporter activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005548">https://www.ebi.ac.uk/QuickGO/term/GO:0005548</a>)            GO:0031267 : small GTPase binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031267">https://www.ebi.ac.uk/QuickGO/term/GO:0031267</a>)</p> <p>GO:0045332 : phospholipid translocation (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0045332">https://www.ebi.ac.uk/QuickGO/term/GO:0045332</a>)            GO:0033344 : cholesterol efflux (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0033344">https://www.ebi.ac.uk/QuickGO/term/GO:0033344</a>)            GO:0042632 : cholesterol homeostasis (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042632">https://www.ebi.ac.uk/QuickGO/term/GO:0042632</a>)            GO:0008203 : cholesterol metabolic process (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008203">https://www.ebi.ac.uk/QuickGO/term/GO:0008203</a>)            GO:0007186 : G protein-coupled receptor signaling pathway (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007186">https://www.ebi.ac.uk/QuickGO/term/GO:0007186</a>)            GO:0034380 : high-density lipoprotein particle assembly (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0034380">https://www.ebi.ac.uk/QuickGO/term/GO:0034380</a>)            GO:0042158 : lipoprotein biosynthetic process (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042158">https://www.ebi.ac.uk/QuickGO/term/GO:0042158</a>)            GO:0042157 : lipoprotein metabolic process (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042157">https://www.ebi.ac.uk/QuickGO/term/GO:0042157</a>)            GO:0033700 : phospholipid efflux (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0033700">https://www.ebi.ac.uk/QuickGO/term/GO:0033700</a>)            GO:0010875 : positive regulation of cholesterol efflux (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0010875">https://www.ebi.ac.uk/QuickGO/term/GO:0010875</a>)            GO:0032489 : regulation of Cdc42 protein signal transduction (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0032489">https://www.ebi.ac.uk/QuickGO/term/GO:0032489</a>)            GO:0043691 : reverse cholesterol transport (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043691">https://www.ebi.ac.uk/QuickGO/term/GO:0043691</a>)            GO:0042493 : response to drug (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042493">https://www.ebi.ac.uk/QuickGO/term/GO:0042493</a>)            GO:0071222 : cellular response to lipopolysaccharide (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0071222">https://www.ebi.ac.uk/QuickGO/term/GO:0071222</a>)            GO:0071300 : cellular response to retinoic acid (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0071300">https://www.ebi.ac.uk/QuickGO/term/GO:0071300</a>)            GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007189">https://www.ebi.ac.uk/QuickGO/term/GO:0007189</a>)            GO:0006911 : phagocytosis, engulfment (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0006911">https://www.ebi.ac.uk/QuickGO/term/GO:0006911</a>)            GO:0007584 : response to nutrient (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007584">https://www.ebi.ac.uk/QuickGO/term/GO:0007584</a>)            GO:0007040 : lysosome organization</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
---	---	---	--

(<https://www.ebi.ac.uk/QuickGO/term/GO:0007040>)  
 GO:0071397 : cellular response to cholesterol  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0071397>)  
 GO:0030301 : cholesterol transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0030301>)  
 GO:0006869 : lipid transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0006869>)  
 GO:0015914 : phospholipid transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015914>)  
 GO:0071404 : cellular response to low-density lipoprotein particle stimulus  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0071404>)  
 GO:0016197 : endosomal transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0016197>)  
 GO:0050702 : interleukin-1 beta secretion  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0050702>)  
 GO:0032367 : intracellular cholesterol transport  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0032367>)  
 GO:0002790 : peptide secretion (<https://www.ebi.ac.uk/QuickGO/term/GO:0002790>)  
 GO:0055091 : phospholipid homeostasis  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0055091>)  
 GO:0060155 : platelet dense granule organization  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060155>)  
 GO:0006497 : protein lipidation (<https://www.ebi.ac.uk/QuickGO/term/GO:0006497>)  
 GO:0034616 : response to laminar fluid shear stress  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0034616>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)  
 GO:0045121 : membrane raft (<https://www.ebi.ac.uk/QuickGO/term/GO:0045121>)  
 GO:0005887 : integral component of plasma membrane  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)  
 GO:0005794 : Golgi apparatus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005794>)  
 GO:0043231 : intracellular membrane-bounded organelle  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)  
 GO:0009986 : cell surface (<https://www.ebi.ac.uk/QuickGO/term/GO:0009986>)  
 GO:0030139 : endocytic vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0030139>)  
 GO:0048471 : perinuclear region of cytoplasm  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)  
 GO:0009897 : external side of plasma membrane  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0009897>)  
 GO:0045335 : phagocytic vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0045335>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~SNP^#gephebase-summary-title))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

amino acid substitution at a residue that is conserved in the ABCA1 gene between human; mouse; chicken; and Takifugu rubripes. Generation of a mouse ABCA1 protein with the mutation disrupts protein function.

Experimental Evidence

Linkage Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Linkage+Mapping^#gephebase-summary-title))

	Taxon A	Taxon B	Position
Codon	AAA	GAA	265
Amino-acid	Glu	Lys	89

Main Reference

Identification and functional analysis of a naturally occurring E89K mutation in the ABCA1 gene of the WHAM chicken. (2002) (<https://pubmed.ncbi.nlm.nih.gov/12364545>)

Authors

Attie AD; Hamon Y; Brooks-Wilson AR; Gray-Keller MP; MacDonald ML; Rigot V; Tebon A; Zhang LH; Mulligan JD; Singaraja RR; Bitgood JJ; Cook ME; Kastelein JJ; Chimini G; Hayden MR

Abstract

The Wisconsin hypoalpha mutant (WHAM) chicken has a >90% reduction in plasma HDL due to hypercatabolism by the kidney of lipid-poor apoA-I. The WHAM chickens have a recessive white skin phenotype caused by a single-gene mutation that maps to the chicken Z-chromosome. This corresponds to human 9q31.1, a chromosomal segment that contains the ATP-binding cassette protein-1 (ABCA1) gene, which is mutated in Tangier Disease and familial hypoalphalipoproteinemia. Complete sequencing of the WHAM ABCA1 cDNA identified a missense mutation near the N-terminus of the protein (E89K). The substitution of this evolutionary conserved glutamate residue for lysine in the mouse ABCA1 transporter leads to complete loss of function, resulting principally from defective intracellular trafficking and very little ABCA1 reaching the plasma membrane. The WHAM chicken is a naturally occurring animal model for Tangier Disease.

Additional References

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLC01B3, SOX10, tyrosinase (TYR),

No matches found.

## EXTERNAL LINKS

## COMMENTS