

GEPHE SUMMARY

ESR1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="ESR1"#gephebase-summary-title)	Gephebase Gene	GP00002440	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Behavior (https://www.gephebase.org/search-criteria?/and+Trait Category="Behavior"#gephebase-summary-title)	Trait Category		
Aggression behavior (<a aggression"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Aggression behavior"#gephebase-summary-title)	Trait		
White throated sparrow - tan-striped (TS) morph - less aggressive - homozygous for the standard arrangement ZAL2	Trait State in Taxon A		
White throated sparrow - white-striped morphs - more aggressive - both sexes are heterozygous and carry one copy of ZAL2m (inverted region) and one copy of ZAL2	Trait State in Taxon B		
	Ancestral State		
Taxon A	Taxonomic Status		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Intraspecific"#gephebase-summary-title)			
	Taxon A	Taxon B	
Zonotrichia albicollis (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Zonotrichia albicollis"#gephebase-summary-title)	Latin Name	Zonotrichia albicollis (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Zonotrichia albicollis"#gephebase-summary-title)	Latin Name
white-throated sparrow	Common Name	white-throated sparrow	Common Name
white-throated sparrow; Zonotrichia albicollis (Gmelin, 1789)	Synonyms	white-throated sparrow; Zonotrichia albicollis (Gmelin, 1789)	Synonyms
species	Rank	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; Passeriformes; Passerellidae; Zonotrichia	Lineage	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; Passeriformes; Passerellidae; Zonotrichia	Lineage
Zonotrichia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 44387)	Parent	Zonotrichia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 44387)	Parent
44394 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 44394)	NCBI Taxonomy ID	44394 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 44394)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Esr1	Generic Gene Name	P19785 (http://www.uniprot.org/uniprot/P19785)	UniProtKB Mus musculus
ESR; ER; ERa; Estr; Estra; Nr3a1; ERalpha; ER-alpha; Esr	Synonyms	0	GenebankID or UniProtKB
10090.ENSMUSP00000070070 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000070070)	String		
Belongs to the nuclear hormone receptor family. NR3 subfamily.	Sequence Similarities		
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)	GO - Molecular Function		
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)			

GO:0043565 : sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043565>)
GO:0008134 : transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008134>)
GO:0008270 : zinc ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008270>)
GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0044877 : protein-containing complex binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044877>)
GO:0051117 : ATPase binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0051117>)
GO:0008013 : beta-catenin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008013>)
GO:0003682 : chromatin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003682>)
GO:0019899 : enzyme binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019899>)
GO:0004879 : nuclear receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004879>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0005496 : steroid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005496>)
GO:0019901 : protein kinase binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019901>)
GO:1990837 : sequence-specific double-stranded DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990837>)
GO:0003707 : steroid hormone receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003707>)
GO:0001222 : transcription corepressor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001222>)
GO:1990841 : promoter-specific chromatin binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990841>)
GO:0001223 : transcription coactivator binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001223>)
GO:0001221 : transcription cofactor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001221>)
GO:0042562 : hormone binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0042562>)
GO:0017025 : TBP-class protein binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0017025>)
GO:0034056 : estrogen response element binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0034056>)
GO:0030284 : nuclear estrogen receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030284>)
GO:0030331 : nuclear estrogen receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030331>)
GO:0036312 : phosphatidylinositol 3-kinase regulatory subunit binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0036312>)
GO:0001093 : TFIIIB-class transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001093>)
GO:0031798 : type 1 metabotropic glutamate receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031798>)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0006357 : regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)
GO:0006355 : regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006355>)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0010629 : negative regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010629>)
GO:1901215 : negative regulation of neuron death
(<https://www.ebi.ac.uk/QuickGO/term/GO:1901215>)
GO:0090209 : negative regulation of triglyceride metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090209>)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)
GO:0051000 : positive regulation of nitric-oxide synthase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051000>)
GO:0008584 : male gonad development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008584>)
GO:0060749 : mammary gland alveolus development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060749>)
GO:0007204 : positive regulation of cytosolic calcium ion concentration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007204>)
GO:0050679 : positive regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050679>)
GO:0048662 : negative regulation of smooth muscle cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048662>)
GO:0043627 : response to estrogen (<https://www.ebi.ac.uk/QuickGO/term/GO:0043627>)
GO:0032355 : response to estradiol (<https://www.ebi.ac.uk/QuickGO/term/GO:0032355>)

GO:0048863 : stem cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048863>)
GO:0043433 : negative regulation of DNA-binding transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043433>)
GO:0007200 : phospholipase C-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007200>)
GO:0006366 : transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006366>)
GO:0010863 : positive regulation of phospholipase C activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010863>)
GO:0060527 : prostate epithelial cord arborization involved in prostate glandular acinus morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060527>)
GO:0060523 : prostate epithelial cord elongation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060523>)
GO:0060687 : regulation of branching involved in prostate gland morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060687>)
GO:0043523 : regulation of neuron apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043523>)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045429>)
GO:0048146 : positive regulation of fibroblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048146>)
GO:0051091 : positive regulation of DNA-binding transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051091>)
GO:0071392 : cellular response to estradiol stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071392>)
GO:0050727 : regulation of inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050727>)
GO:0060068 : vagina development (<https://www.ebi.ac.uk/QuickGO/term/GO:0060068>)
GO:0045899 : positive regulation of RNA polymerase II transcriptional preinitiation complex assembly (<https://www.ebi.ac.uk/QuickGO/term/GO:0045899>)
GO:0043124 : negative regulation of I-kappaB kinase/NF-kappaB signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043124>)
GO:0060065 : uterus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0060065>)
GO:0048144 : fibroblast proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048144>)
GO:0008209 : androgen metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008209>)
GO:0001547 : antral ovarian follicle growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001547>)
GO:0071391 : cellular response to estrogen stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071391>)
GO:0002064 : epithelial cell development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002064>)
GO:0060750 : epithelial cell proliferation involved in mammary gland duct elongation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060750>)
GO:0030520 : intracellular estrogen receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030520>)
GO:0030518 : intracellular steroid hormone receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030518>)
GO:0060745 : mammary gland branching involved in pregnancy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060745>)
GO:0046325 : negative regulation of glucose import
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046325>)
GO:1903799 : negative regulation of miRNA maturation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1903799>)
GO:0045839 : negative regulation of mitotic nuclear division
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045839>)
GO:0045742 : positive regulation of epidermal growth factor receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045742>)
GO:0071168 : protein localization to chromatin
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071168>)
GO:1904035 : regulation of epithelial cell apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904035>)
GO:0034121 : regulation of toll-like receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0034121>)
GO:0060011 : Sertoli cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060011>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0005794 : Golgi apparatus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005794>)
GO:0005667 : transcription factor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005667>)
GO:0005739 : mitochondrion (<https://www.ebi.ac.uk/QuickGO/term/GO:0005739>)
GO:0032991 : protein-containing complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032991>)
GO:0043005 : neuron projection (<https://www.ebi.ac.uk/QuickGO/term/GO:0043005>)

GO:0030315 : T-tubule (<https://www.ebi.ac.uk/QuickGO/term/GO:0030315>)
 GO:0048471 : perinuclear region of cytoplasm
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)
 GO:0000785 : chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000785>)
 GO:0043204 : perikaryon (<https://www.ebi.ac.uk/QuickGO/term/GO:0043204>)
 GO:0043195 : terminal bouton (<https://www.ebi.ac.uk/QuickGO/term/GO:0043195>)
 GO:0000791 : euchromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000791>)
 GO:0097550 : transcription preinitiation complex
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0097550>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

Increase in expression of the gene in the white-striped morph. Differential allelic expression in heterozygotes.

Experimental Evidence

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

Main Reference

A supergene-linked estrogen receptor drives alternative phenotypes in a polymorphic songbird. (2020) (<https://pubmed.ncbi.nlm.nih.gov/32817554>)

Authors

Merritt JR; Grogan KE; Zinzow-Kramer WM; Sun D; Ortlund EA; Yi SV; Maney DL

Abstract

Behavioral evolution relies on genetic changes, yet few behaviors can be traced to specific genetic sequences in vertebrates. Here we provide experimental evidence showing that differentiation of a single gene has contributed to the evolution of divergent behavioral phenotypes in the white-throated sparrow, a common backyard songbird. In this species, a series of chromosomal inversions has formed a supergene that segregates with an aggressive phenotype. The supergene has captured ESR1, the gene that encodes estrogen receptor $ER\pm$; as a result, this gene is accumulating changes that now distinguish the supergene allele from the standard allele. Our results show that in birds of the more aggressive phenotype, $ER\pm$ knockdown caused a phenotypic change to that of the less aggressive phenotype. We next showed that in a free-living population, aggression is predicted by allelic imbalance favoring the supergene allele. Finally, we identified cis-regulatory features, both genetic and epigenetic, that explain the allelic imbalance. This work provides a rare illustration of how genotypic divergence has led to behavioral phenotypic divergence in a vertebrate.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@SuperGene @Inversion @Epimutation - cis-regulation that is attributable to both genetic and epigenetic factors