

GEPHE SUMMARY

Amhr2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] Amhr2 [^] #gephebase-summary-title)	Gephebase Gene	GP00002147	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category= [^] Physiology [^] #gephebase-summary-title)	Trait Category
Sex determination (female vs male) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Sex determination (female vs male)<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=[^]Sex determination (female vs male)[^]#gephebase-summary-title)	Trait
Female (homozygous)	Trait State in Taxon A
Male (heterozygous)	Trait State in Taxon B
Unknown	Ancestral State
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [^] Intraspecific [^] #gephebase-summary-title)	Taxonomic Status

Taxon A #1	
Takifugu rubripes (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Takifugu rubripes<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Takifugu rubripes[^]#gephebase-summary-title)	Latin Name
torafugu	Common Name
Fugu rubripes; Sphaeroides rubripes; Tetraodon rubripes; torafugu; tiger puffer; Takifugu rubripes (Temminck & Schlegel, 1850)	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percomorphaceae; Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradontoidea; Tetraodontidae; Takifugu	Lineage
Takifugu () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31032)	Parent
31033 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31033)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?

Taxon B #1	
Takifugu rubripes (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Takifugu rubripes<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Takifugu rubripes[^]#gephebase-summary-title)	Latin Name
torafugu	Common Name
Fugu rubripes; Sphaeroides rubripes; Tetraodon rubripes; torafugu; tiger puffer; Takifugu rubripes (Temminck & Schlegel, 1850)	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percomorphaceae; Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradontoidea; Tetraodontidae; Takifugu	Lineage
Takifugu () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31032)	Parent
31033 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31033)	NCBI Taxonomy ID
No	is Taxon B an Intraspecies?

Taxon A #2	
Takifugu poecilonotus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Takifugu poecilonotus<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Takifugu poecilonotus[^]#gephebase-summary-title)	Latin Name
finepatterned puffer	Common Name
Fugu poecilonotus; Tetraodon poecilonotus; finepatterned puffer; komonfugu; Takifugu poecilonotus (Temminck & Schlegel, 1850)	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percomorphaceae;	Lineage

Taxon B #2	
Takifugu poecilonotus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Takifugu poecilonotus<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Takifugu poecilonotus[^]#gephebase-summary-title)	Latin Name
finepatterned puffer	Common Name
Fugu poecilonotus; Tetraodon poecilonotus; finepatterned puffer; komonfugu; Takifugu poecilonotus (Temminck & Schlegel, 1850)	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percomorphaceae;	Lineage

Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradoidea; Tetraodontidae;
Takifugu

Parent

Takifugu () - (Rank: genus)
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31032>)
NCBI Taxonomy ID

176188
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=176188>)
is Taxon A an Intraspecies?

No

Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradoidea; Tetraodontidae;
Takifugu

Parent

Takifugu () - (Rank: genus)
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31032>)
NCBI Taxonomy ID

176188
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=176188>)
is Taxon B an Intraspecies?

No

Taxon A #3

Latin Name

Takifugu pardalis
(<https://www.gephebase.org/search-criteria?and+Taxon+and+Synonyms=~Takifugu+pardalis~#gephebase-summary-title>)

Common Name

panther puffer

Synonyms

Fugu pardalis; panther puffer; higanfugu; Takifugu pardalis (Temminck & Schlegel, 1850)

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaeae; Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradoidea; Tetraodontidae; Takifugu

Parent

Takifugu () - (Rank: genus)
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31032>)
NCBI Taxonomy ID

98921
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=98921>)
is Taxon A an Intraspecies?

No

Taxon B #3

Latin Name

Takifugu pardalis
(<https://www.gephebase.org/search-criteria?and+Taxon+and+Synonyms=~Takifugu+pardalis~#gephebase-summary-title>)

Common Name

panther puffer

Synonyms

Fugu pardalis; panther puffer; higanfugu; Takifugu pardalis (Temminck & Schlegel, 1850)

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaeae; Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradoidea; Tetraodontidae; Takifugu

Parent

Takifugu () - (Rank: genus)
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=31032>)
NCBI Taxonomy ID

98921
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=98921>)
is Taxon B an Intraspecies?

No

GENOTYPIC CHANGE

<p>AMHR2</p> <p>AMHR; MR2; MISR2; MISR1</p> <p>9606.ENSPO0000257863 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000257863)</p> <p>Sequence Similarities</p> <p>Belongs to the protein kinase superfamily. TKL Ser/Thr protein kinase family. TGFB receptor subfamily.</p> <p>GO - Molecular Function</p> <p>GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524) GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872) GO:0004674 : protein serine/threonine kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004674) GO:0042562 : hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042562) GO:1990272 : anti-Mullerian hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:1990272) GO:0005026 : transforming growth factor beta receptor activity, type II (https://www.ebi.ac.uk/QuickGO/term/GO:0005026) GO:0005024 : transforming growth factor beta-activated receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005024)</p> <p>GO - Biological Process</p> <p>GO:0008584 : male gonad development (https://www.ebi.ac.uk/QuickGO/term/GO:0008584) GO:0007548 : sex differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0007548) GO:0006468 : protein phosphorylation (https://www.ebi.ac.uk/QuickGO/term/GO:0006468) GO:0030509 : BMP signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0030509) GO:0007179 : transforming growth factor beta receptor signaling pathway</p>	<p>Generic Gene Name</p> <p>Q16671 (http://www.uniprot.org/uniprot/Q16671)</p> <p>Synonyms</p> <p>0</p> <p>String</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0007179>)
 GO:0008585 : female gonad development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0008585>)
 GO:0001568 : blood vessel development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0001568>)
 GO:0071363 : cellular response to growth factor stimulus
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0071363>)
 GO:1990262 : anti-Mullerian hormone signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:1990262>)
 GO:0001880 : Mullerian duct regression
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0001880>)
 GO:0001944 : vasculature development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0001944>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
 GO:0005887 : integral component of plasma membrane
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)
 GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null="No" #gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

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=))

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 Coding Change

Nonsynonymous

Molecular Details of the Mutation

His/Asp384 heterozygous males have reduced Amhr2 activity due to decreased activity of the kinase signaling domain

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=))

	Taxon A	Taxon B	Position
Codon	CAT	GAT	1152
Amino-acid	His	Asp	384

Main Reference

A trans-species missense SNP in Amhr2 is associated with sex determination in the tiger pufferfish, *Takifugu rubripes* (fugu). (2012) (<https://pubmed.ncbi.nlm.nih.gov/22807687/>)

Authors

Kamiya T; Kai W; Tasumi S; Oka A; Matsunaga T; Mizuno N; Fujita M; Suetake H; Suzuki S; Hosoya S; Tohari S; Brenner S; Miyadai T; Venkatesh B; Suzuki Y; Kikuchi K

Abstract

Heterogametic sex chromosomes have evolved independently in various lineages of vertebrates. Such sex chromosome pairs often contain nonrecombining regions, with one of the chromosomes harboring a master sex-determining (SD) gene. It is hypothesized that these sex chromosomes evolved from a pair of autosomes that diverged after acquiring the SD gene. By linkage and association mapping of the SD locus in fugu (*Takifugu rubripes*), we show that a SNP (C/G) in the anti-Müllerian hormone receptor type II (*Amhr2*) gene is the only polymorphism associated with phenotypic sex. This SNP changes an amino acid (His/Asp384) in the kinase domain. While females are homozygous (His/His384), males are heterozygous. Sex in fugu is most likely determined by a combination of the two alleles of *Amhr2*. Consistent with this model, the medaka *hotei* mutant carrying a substitution in the kinase domain of *Amhr2* causes a female phenotype. The association of the *Amhr2* SNP with phenotypic sex is conserved in two other species of *Takifugu* but not in *Tetraodon*. The fugu SD locus shows no sign of recombination suppression between X and Y chromosomes. Thus, fugu sex chromosomes represent an unusual example of proto-sex chromosomes. Such undifferentiated X-Y chromosomes may be more common in vertebrates than previously thought.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Sexual Trait @TransspecificPolymorphism @BalancingSelection ; <https://omia.org/OMIA001728/31033/>

