Module 4: ZFIN Resources

Aims

- Use the ZFIN Single-Box Search to find information on
  - human disease models
  - expression phenotypes
  - mutation details

- Two categories of Antibodies: Curated antibodies in ZFIN and user-contributed antibodies in the Community Antibody Wiki

- An introduction to the Alliance of Genome Resources

- A reminder of where to report sequence assembly problems

OVERVIEW:

The zebrafish model organism database **ZFIN** (https://zfin.org/) provides curated data on zebrafish genes, including gene expression, phenotypes, gene function, mutants, transgenic lines, zebrafish models of human disease, and orthology. This data can be searched, browsed and downloaded. This Module will focus on finding information on zebrafish **human disease models**, **expression phenotypes**, and **mutation details,** with an emphasis on the Single-box faceted search (https://zfin.org/search?q=).

ZFIN continues to provide a platform for researchers to contribute their antibody data and experimental protocols, in the **Antibody and Protocol Wikis.** Users should search the Antibody Wiki in addition to ZFIN to find information on antibodies.

ZFIN is now part of the **Alliance of Genome Resources**, (https://www.alliancegenome.org/) a consortium of 6 model organism databases (MODs) and the Gene Ontology (GO) database. The goal of the **Alliance** is to provide users a central location to access and compare human and model organism data. The Alliance site currently provides data on genes, alleles, gene ontology, orthology, and human disease, as well as a genome browser (JBrowse), with links to gene expression at the MODs and GEO. New features are continually being added to the Alliance.

ZFIN is part of the **GRC Consortium** (https://www.ncbi.nlm.nih.gov/grc), and zebrafish sequence assembly problems can be reported at the GRC website, or by e-mail to ZFIN (zfinadmn@zfin.org).

LIST OF ALL EXAMPLES:

Human Disease examples:

Find all curated zebrafish human disease models.

Which diseases are associated with the human ortholog of ***mecp2***?

What genes are associated with **Cornelia de Lange syndrome**, and are there zebrafish genetic models?

What genes are associated with **hepatocellular carcinoma**? What type of zebrafish models exist?

What are the phenotypes of the "**AB + MO3-nipblb**" disease model fish?

Expression Phenotype examples:

Find all phenotypes with a misexpressed gene.

Which phenotypes have a perturbed ***pitx2*** expression?

Which fish have gene expression phenotypes in **Rohon-Beard neurons**?

Mutation Detail examples:

What mutants are available for the ***fgf8a*** gene and what are their details?

What are the mutation details for the **ti282a** mutant?

Antibody example:

Use the "Ab-S58 Wiki Page" link on the ZFIN **Ab-S58** antibody page to find user comments on the antibody.

Alliance examples:

Use the Alliance link on the ZFIN ***sox10*** gene to navigate to the Alliance zebrafish ***sox10*** page.

Use the Alliance link on the ZFIN **Cornelia de Lange syndrome** disease page to find a **mouse allele** that is associated with the syndrome.

Human Disease Overview:

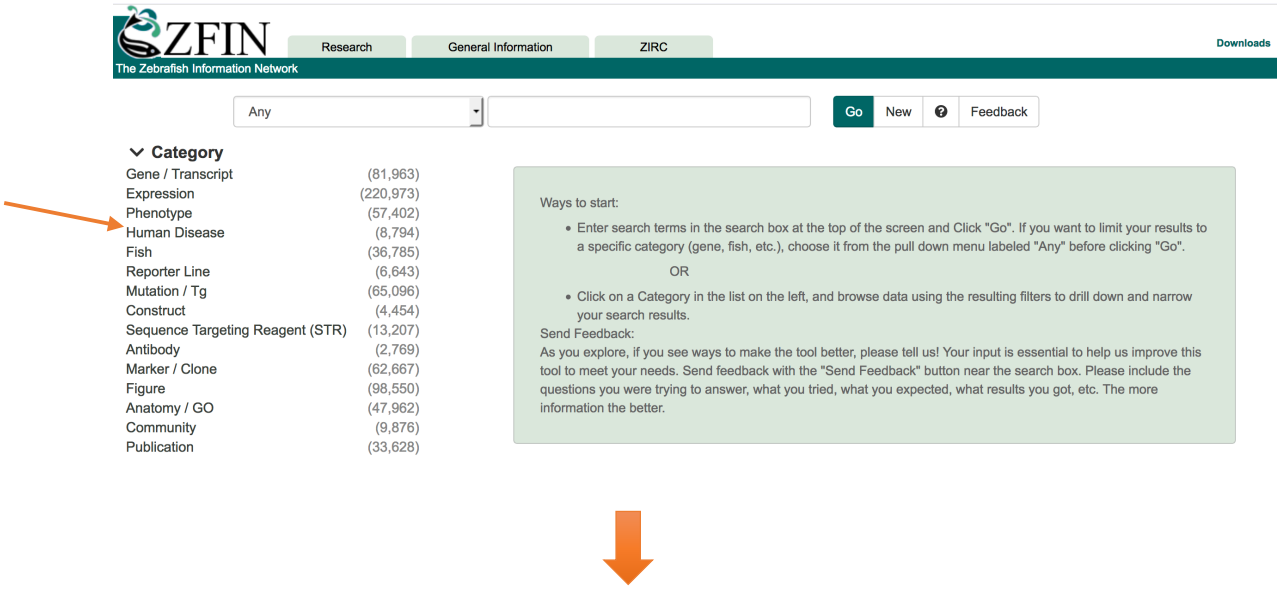
- ZFIN curators curate zebrafish models of human disease from publications, and link publications to diseases.
- Disease models can include mutations, knockdown reagents, and experimental conditions.
- ZFIN human disease terms are from the Disease Ontology. ZFIN Disease pages include a list of human genes associated with that disease (via OMIM), and zebrafish orthologs of the human genes. ZFIN Gene pages include a Disease section.

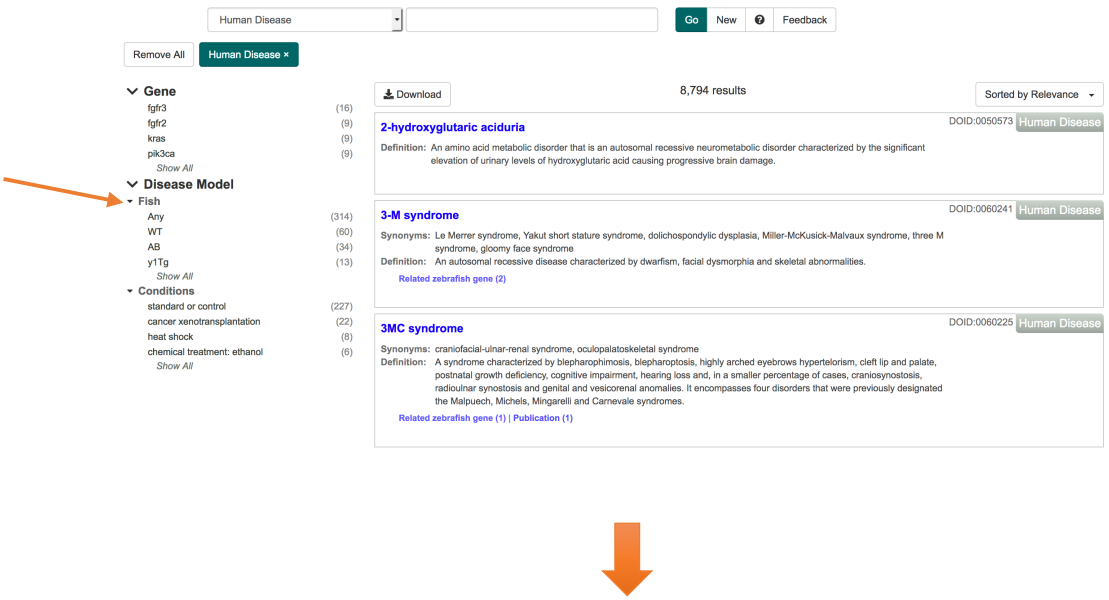
Disease Statistics:

- Over 1700 publications have been associated with a disease in ZFIN.
- Zebrafish models for 315 human diseases have been curated.

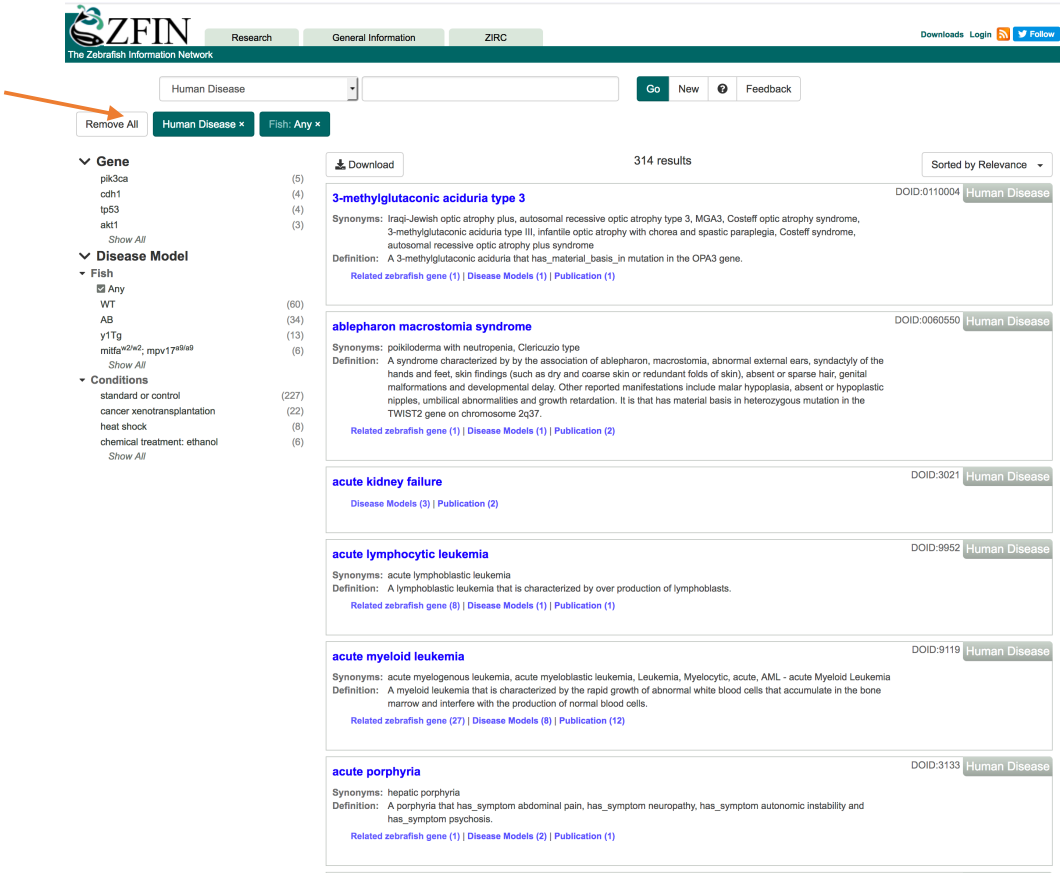
Disease Example 1
Find all ZFIN-curated zebrafish disease models.

1. On the ZFIN home page (zfin.org), Click on “Go” next to the Single Box search. https://zfin.org/search?q=
2. Select the “Human Disease” category
3. In the “Disease Model” facet, “Fish” sub-facet, select “Any”.





Results

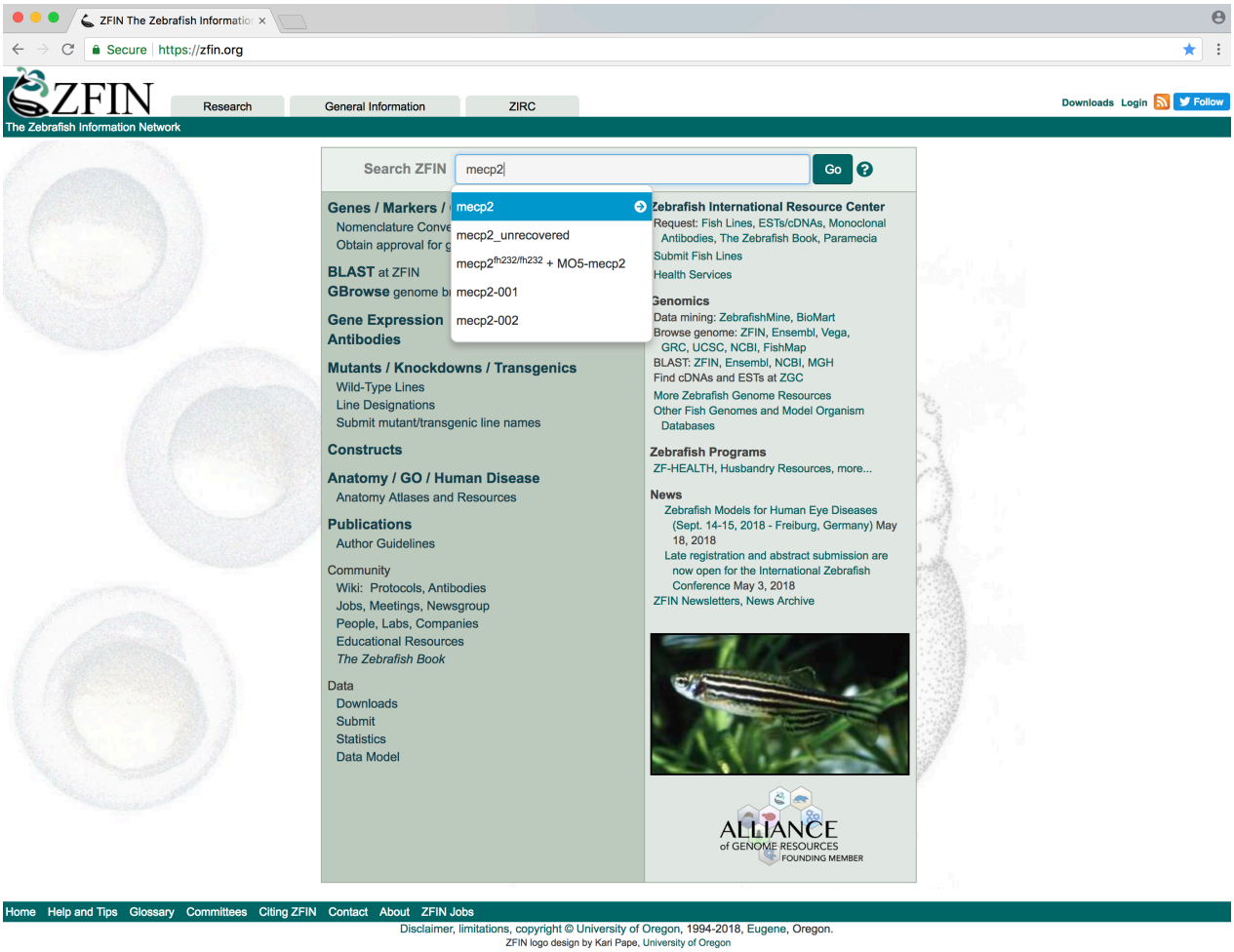


Tip: Single-box Search Help can be accessed by clicking on the nearby "?" icon: https://wiki.zfin.org/display/general/ZFIN+Single+Box+Search+Help

Disease Example 2
Which diseases are associated with the human ortholog of *mecp2*?

1. On the ZFIN home page, type “mecp2” into the Single Box Search.

2. Select “mecp2” from the search results to go to the *mecp2* gene page.
    a. **Hint**: you can click on the arrow next to “mecp2” in the autocomplete options list to go directly to the *mecp2* gene page.



3. Scroll to the “DISEASE ASSOCIATED WITH *mecp2* HUMAN ORTHOLOG” section

ZFIN ID: ZDB-GENE-030131-7190

Your Input Welcome

Gene Name: ***methyl CpG binding protein 2***
Gene Symbol: ***mecp2***
Sequence Ontology ID : SO:0000704
Previous Names: *wu:fk96a04, zgc:111857*
Location: Chr: 8 Mapping Details/Browsers
Nomenclature History

GENE EXPRESSION

All Expression Data:	6 figures from 6 publications
Directly Submitted Expression Data:	1 figure (1 image) from Thisse *et al.*, 2004 [MGC:111857]
Wild-type Stages, Structures:	Zygote:1-cell (0.0h-0.75h) to Adult (90d-730d, breeding adult) brain, brain neuron, central nervous system, midbrain (all 8)
Curated Microarray Expression:	GEO (1)

MUTATIONS AND SEQUENCE TARGETING REAGENTS

Allele	Type	Localization	Consequence	Mutagen	Suppliers
fh232	Point Mutation	Unknown	Premature Stop	ENU	Zebrafish International Resource Center (ZIRC) (order this)
la026687Tg	Transgenic Insertion	Unknown	Unknown	DNA	
mecp2_unrecovered	Point Mutation	Unknown	Unknown	ENU	
sa21196	Point Mutation	Unknown	Premature Stop	ENU	Zebrafish International Resource Center (ZIRC) (order this) European Zebrafish Resource Center (EZRC) (order this)

Targeting reagents: CRISPR1-mecp2 (1), MO1-mecp2 (1), MO2-mecp2 (1), MO3-mecp2 (1), MO4-mecp2 (1) (all 6)

PHENOTYPE

Data:	19 figures from 5 publications
Observed in:	anatomical structure, apoptotic process, astrocyte, brain astrocyte differentiation (all 29)

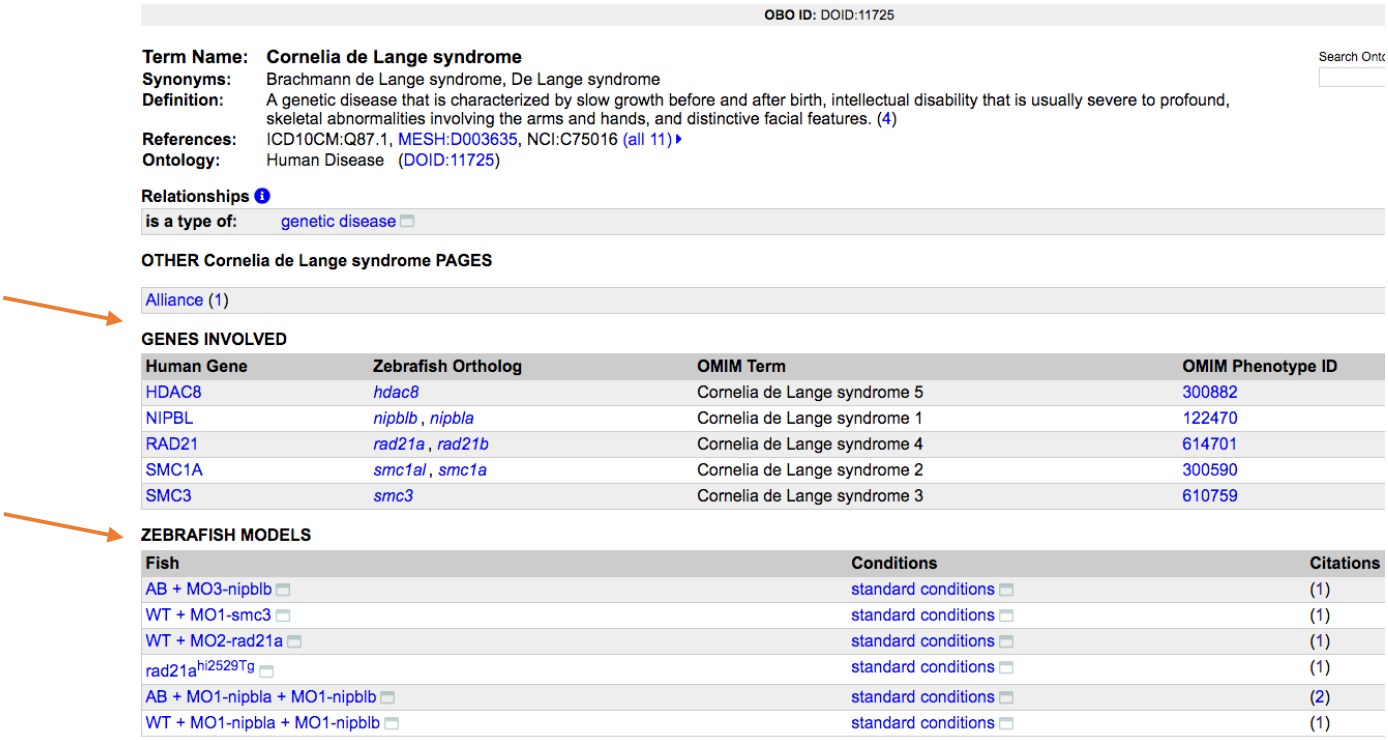
DISEASE ASSOCIATED WITH *mecp2* HUMAN ORTHOLOG

Disease Ontology Term	OMIM Term	OMIM Phenotype ID
Rett syndrome	Rett syndrome	312750
	Rett syndrome, atypical	312750
	Rett syndrome, preserved speech variant	312750
syndromic X-linked intellectual disability Lubs type	Mental retardation, X-linked syndromic, Lubs type	300260
X-linked intellectual disability-psychosis-macroorchidism syndrome	Mental retardation, X-linked, syndromic 13	300055
	{Autism susceptibility, X-linked 3}	300496
	Encephalopathy, neonatal severe	300673

Show first 5 records

Disease Example 3
What genes are associated with Cornelia de Lange syndrome, and are there zebrafish genetic models?

1. Search for "Cornelia de Lange syndrome" using the Single Box Search.
2. Select term from results list and navigate to the ZFIN "Cornelia de Lange syndrome" disease page.
3. Scroll to the "GENES INVOLVED" and "ZEBRAFISH MODELS" sections.



OBO ID: DOID:11725

Term Name: **Cornelia de Lange syndrome**
Synonyms: Brachmann de Lange syndrome, De Lange syndrome
Definition: A genetic disease that is characterized by slow growth before and after birth, intellectual disability that is usually severe to profound, skeletal abnormalities involving the arms and hands, and distinctive facial features. (4)
References: ICD10CM:Q87.1, MESH:D003635, NCI:C75016 (all 11)
Ontology: Human Disease (DOID:11725)

Relationships
is a type of: genetic disease

OTHER Cornelia de Lange syndrome PAGES

Alliance (1)

GENES INVOLVED

Human Gene	Zebrafish Ortholog	OMIM Term	OMIM Phenotype ID
HDAC8	*hdac8*	Cornelia de Lange syndrome 5	300882
NIPBL	*nipblb*, *nipbla*	Cornelia de Lange syndrome 1	122470
RAD21	*rad21a*, *rad21b*	Cornelia de Lange syndrome 4	614701
SMC1A	*smc1al*, *smc1a*	Cornelia de Lange syndrome 2	300590
SMC3	*smc3*	Cornelia de Lange syndrome 3	610759

ZEBRAFISH MODELS

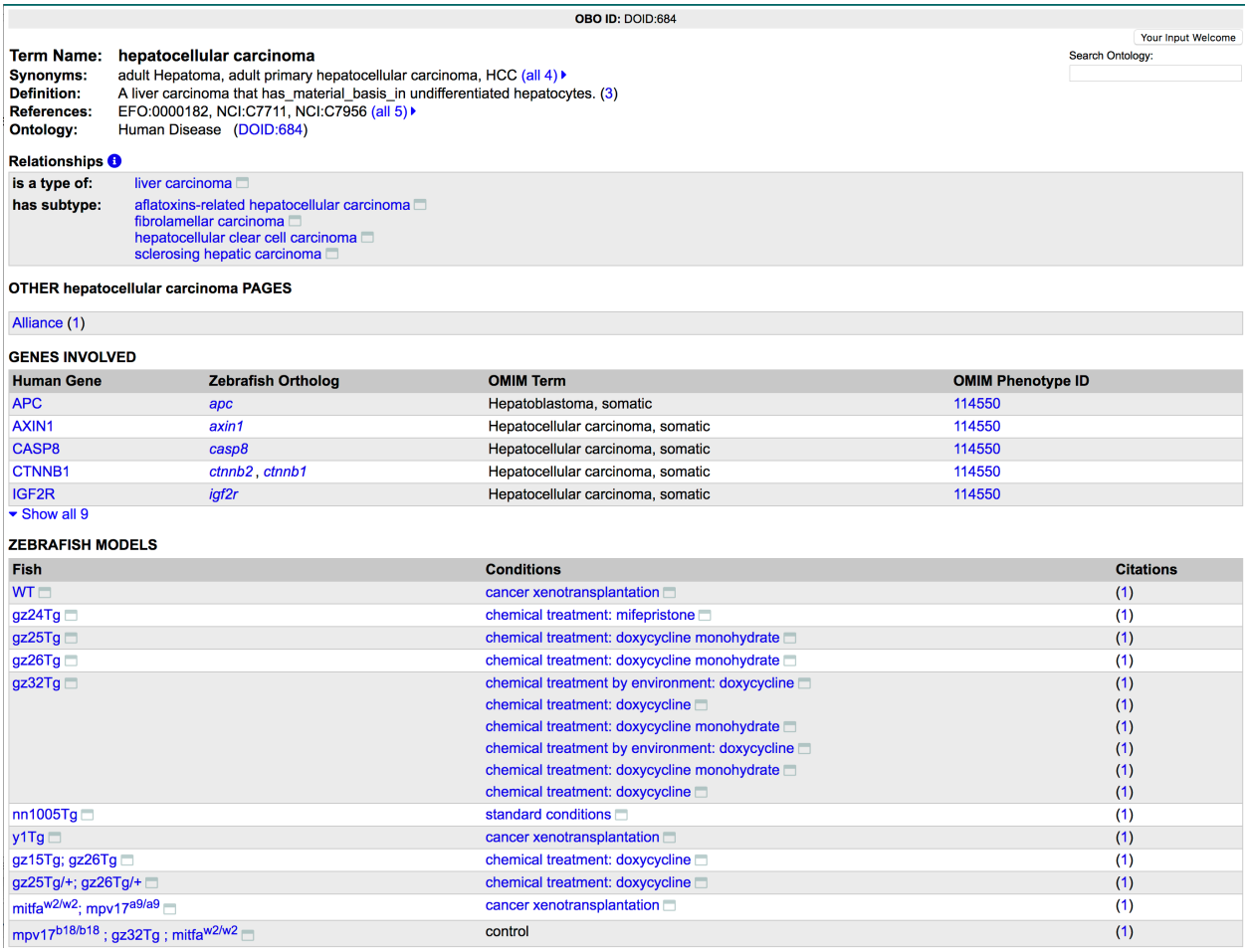
Fish	Conditions	Citations
AB + MO3-nipblb	standard conditions	(1)
WT + MO1-smc3	standard conditions	(1)
WT + MO2-rad21a	standard conditions	(1)
rad21a^{hi2529Tg}	standard conditions	(1)
AB + MO1-nipbla + MO1-nipblb	standard conditions	(2)
WT + MO1-nipbla + MO1-nipblb	standard conditions	(1)

Note: Zebrafish mutant and morphant disease models (such as "AB+ MO3-nipblb" or "rad21a^{hi2529Tg}") are curated from the literature by ZFIN curators, based on author statements in publications.

Disease Example 4

What genes are associated with hepatocellular carcinoma? What type of zebrafish models exist?

1. Search for “hepatocellular carcinoma” using the Single Box Search.
2. Select term from results and navigate to the ZFIN disease page for “hepatocellular carcinoma”.
3. Scroll to the “ZEBRAFISH MODELS” section.



OBO ID: DOID:684

Your Input Welcome

Search Ontology:

Term Name: hepatocellular carcinoma
Synonyms: adult Hepatoma, adult primary hepatocellular carcinoma, HCC (all 4)
Definition: A liver carcinoma that has_material_basis_in undifferentiated hepatocytes. (3)
References: EFO:0000182, NCI:C7711, NCI:C7956 (all 5)
Ontology: Human Disease (DOID:684)

Relationships

is a type of:	liver carcinoma
has subtype:	aflatoxins-related hepatocellular carcinoma fibrolamellar carcinoma hepatocellular clear cell carcinoma sclerosing hepatic carcinoma

OTHER hepatocellular carcinoma PAGES

Alliance (1)

GENES INVOLVED

Human Gene	Zebrafish Ortholog	OMIM Term	OMIM Phenotype ID
APC	*apc*	Hepatoblastoma, somatic	114550
AXIN1	*axin1*	Hepatocellular carcinoma, somatic	114550
CASP8	*casp8*	Hepatocellular carcinoma, somatic	114550
CTNNB1	*ctnnb2*, *ctnnb1*	Hepatocellular carcinoma, somatic	114550
IGF2R	*igf2r*	Hepatocellular carcinoma, somatic	114550

Show all 9

ZEBRAFISH MODELS

Fish	Conditions	Citations
WT	cancer xenotransplantation	(1)
gz24Tg	chemical treatment: mifepristone	(1)
gz25Tg	chemical treatment: doxycycline monohydrate	(1)
gz26Tg	chemical treatment: doxycycline monohydrate	(1)
gz32Tg	chemical treatment by environment: doxycycline	(1)
	chemical treatment: doxycycline	(1)
	chemical treatment: doxycycline monohydrate	(1)
	chemical treatment by environment: doxycycline	(1)
	chemical treatment: doxycycline monohydrate	(1)
	chemical treatment: doxycycline	(1)
nn1005Tg	standard conditions	(1)
y1Tg	cancer xenotransplantation	(1)
gz15Tg; gz26Tg	chemical treatment: doxycycline	(1)
gz25Tg/+; gz26Tg/+	chemical treatment: doxycycline	(1)
$mitfa^{w2/w2}$; $mpv17^{a9/a9}$	cancer xenotransplantation	(1)
$mpv17^{b18/b18}$; gz32Tg ; $mitfa^{w2/w2}$	control	(1)

Note:
Some disease models are a result of chemical treatments or cancer xenotransplantation

Tip:
Click on the icon next to the Fish line designation to see the full name and components of the fish.

Disease Example 5

What are the phenotypes of the "**AB + MO3-nipblb**" fish used to model Cornelia de Lange syndrome?

1. Search for "AB + MO3-nipblb" using the Single-Box Search (or navigate from the Cornelia de Lange syndrome disease page, ZEBRAFISH MODELS section, from Disease Example 3)
2. Navigate to the ZFIN "AB+ MO3-nipblb" FISH page.
3. Scroll to the "PHENOTYPE" section

ZFIN ID: ZDB-FISH-150901-4291

Fish name: AB + MO3-nipblb
Genotype: *AB*
Targeting Reagent: MO3-nipblb

HUMAN DISEASE MODELED by AB + MO3-nipblb

Human Disease	Conditions	Citations
Cornelia de Lange syndrome	standard conditions	Pistocchi *et al.*, 2013

GENE EXPRESSION
Gene expression in AB + MO3-nipblb

RNA expression

Expressed Gene	Structure	Conditions	Figures
axin2	hindbrain	standard conditions	Fig. 4 from Pistocchi *et al.*, 2013
ccnd1	whole organism	standard conditions	Fig. 6 from Fazio *et al.*, 2016 Fig. 5 from Pistocchi *et al.*, 2013
wnt1	hindbrain	standard conditions	Fig. 4 from Pistocchi *et al.*, 2013

Protein expression

Antibody	Antigen Genes	Structure	Conditions	Figures
Ab1-ccnd1	*ccnd1*	whole organism	standard conditions	Fig. 6 from Fazio *et al.*, 2016

Reporter gene expression No data available

PHENOTYPE
Phenotype in AB + MO3-nipblb

Phenotype	Conditions	Figures
apoptotic process increased occurrence, abnormal	standard conditions	Fig. 3 from Pistocchi *et al.*, 2013
brain decreased size, abnormal	standard conditions	Fig. 2 from Pistocchi *et al.*, 2013
cell proliferation disrupted, abnormal	standard conditions	Fig. 4 from Fazio *et al.*, 2016
central nervous system development disrupted, abnormal	standard conditions	Fig. 5 from Pistocchi *et al.*, 2013
eye decreased size, abnormal	standard conditions	Fig. 2 from Pistocchi *et al.*, 2013
neural tube apoptotic, abnormal	standard conditions	Fig. 3 from Pistocchi *et al.*, 2013
neural tube apoptotic process increased occurrence, abnormal	standard conditions	Fig. 4 from Fazio *et al.*, 2016
post-vent region curved, abnormal	standard conditions	Fig. 2 from Pistocchi *et al.*, 2013
post-vent region decreased length, abnormal	standard conditions	Fig. 2 from Pistocchi *et al.*, 2013
regulation of canonical Wnt signaling pathway disrupted, abnormal	standard conditions	Fig. 5 from Pistocchi *et al.*, 2013
whole organism *ccnd1* expression decreased amount, abnormal	standard conditions	Fig. 6 from Fazio *et al.*, 2016

▲ Show first 5 phenotypes

CITATIONS (3)

Note the "Expression Phenotype" in the Phenotype list: "whole organism *ccnd1* expression decreased amount, abnormal".

Extra exercise: Navigate to the scn1laa$^{sa1674/sa1674}$(TL) fish page. Which disease does this fish model? Go to the PHENOTYPE section. Note the Phenotypes with the "**ameliorated**" tag. Which conditions ameliorate the locomotory behavior and nerve impulse transmission phenotypes?

Expression Phenotypes Overview:

- Gene expression patterns in mutants frequently differ from patterns in wild-type animals. These differences provide information on genetic interactions and pathways.

- ZFIN curates differences between perturbed and wild-type gene expression patterns using a limited number of terms (“increased amount”, “decreased amount”, “absent”, “mislocalised”, “spatial pattern”, “increased distribution” and “decreased distribution”) and the tags “abnormal” and “ameliorated”.

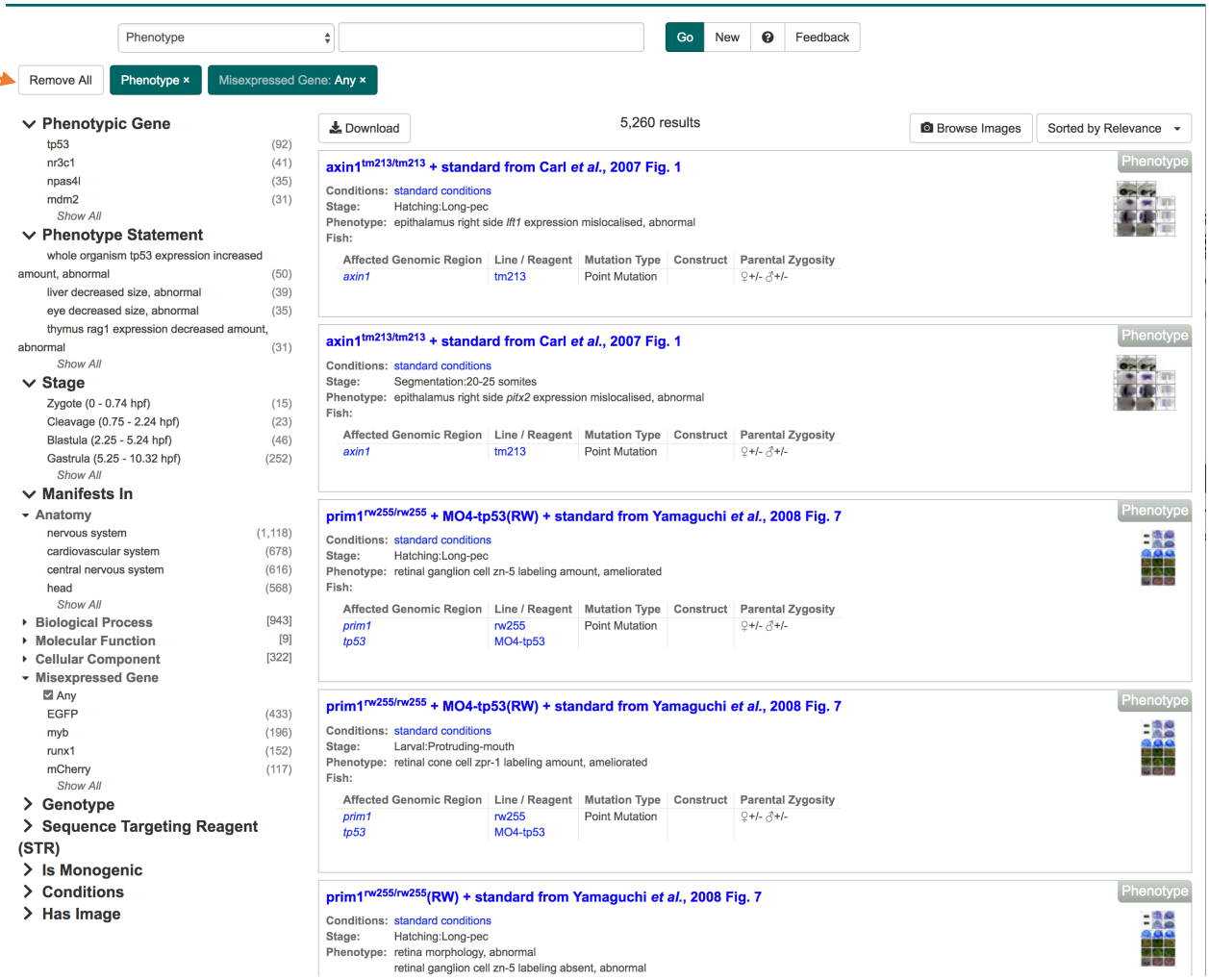
- Expression Phenotypes can be explored using the “Misexpressed gene” facet in five Single-box search categories: Gene/Transcript, Phenotype, Fish, Mutation/Tg and Figure.

Expression Phenotype Statistics

- 912 publications have been curated for expression phenotypes
- 1879 misexpressed genes and over 11000 expression phenotypes have been recorded.

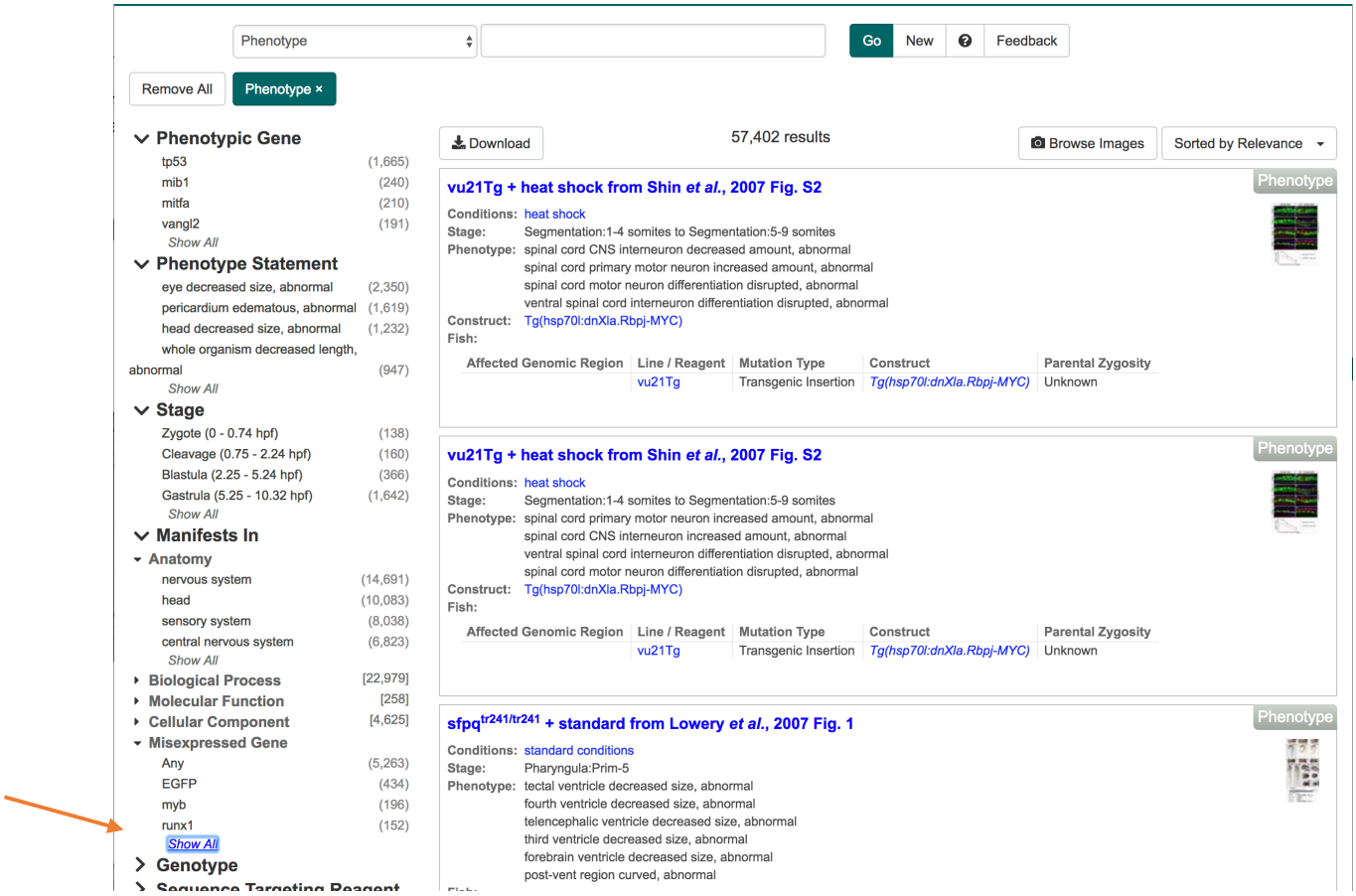
Expression Phenotype Example 1.
Find all phenotypes involving a misexpressed gene.

1. Go to the Single Box Search (https://zfin.org/search?q=)
2. Select the Phenotype Category.
3. Go to the "Misexpressed gene" subfacet and select "Any".

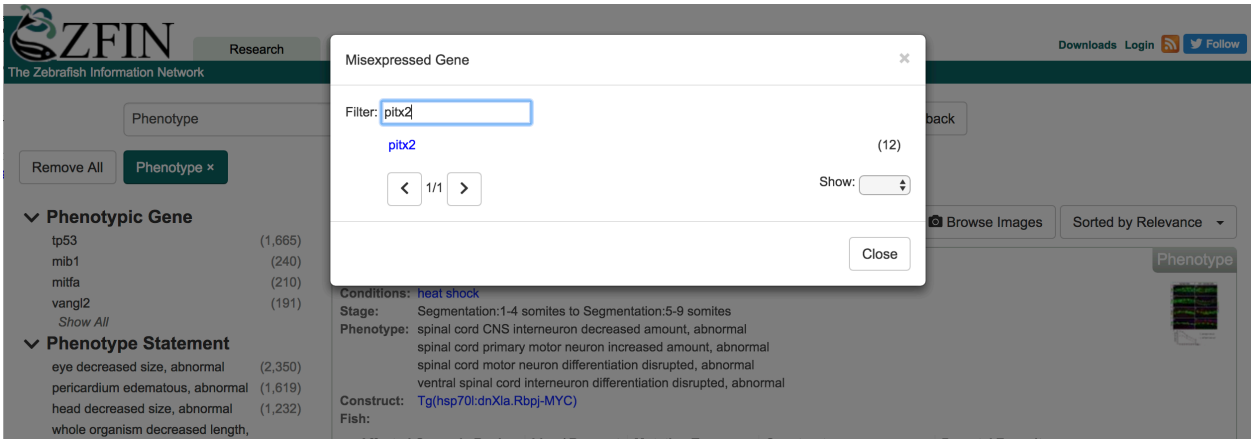


Expression Phenotype Example 2.
Which phenotypes have a perturbed *pitx2* gene expression?

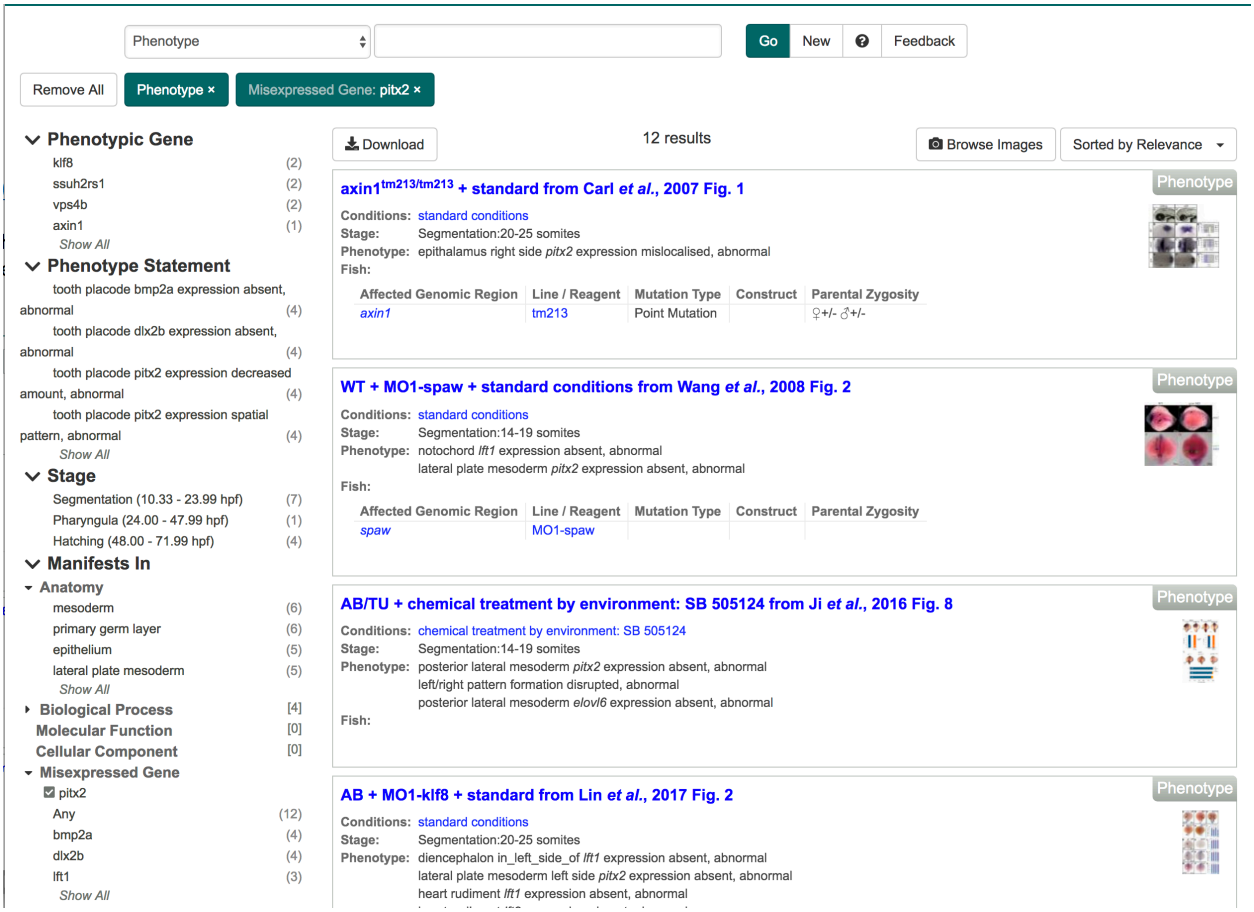
1. Go to the Single Box Search (https://zfin.org/search?q=)
2. Select the Phenotype Category.
3. Go to the “Misexpressed gene” subfacet and click on “Show All”.



4. Type “pitx2” in the “Misexpressed Gene” popup

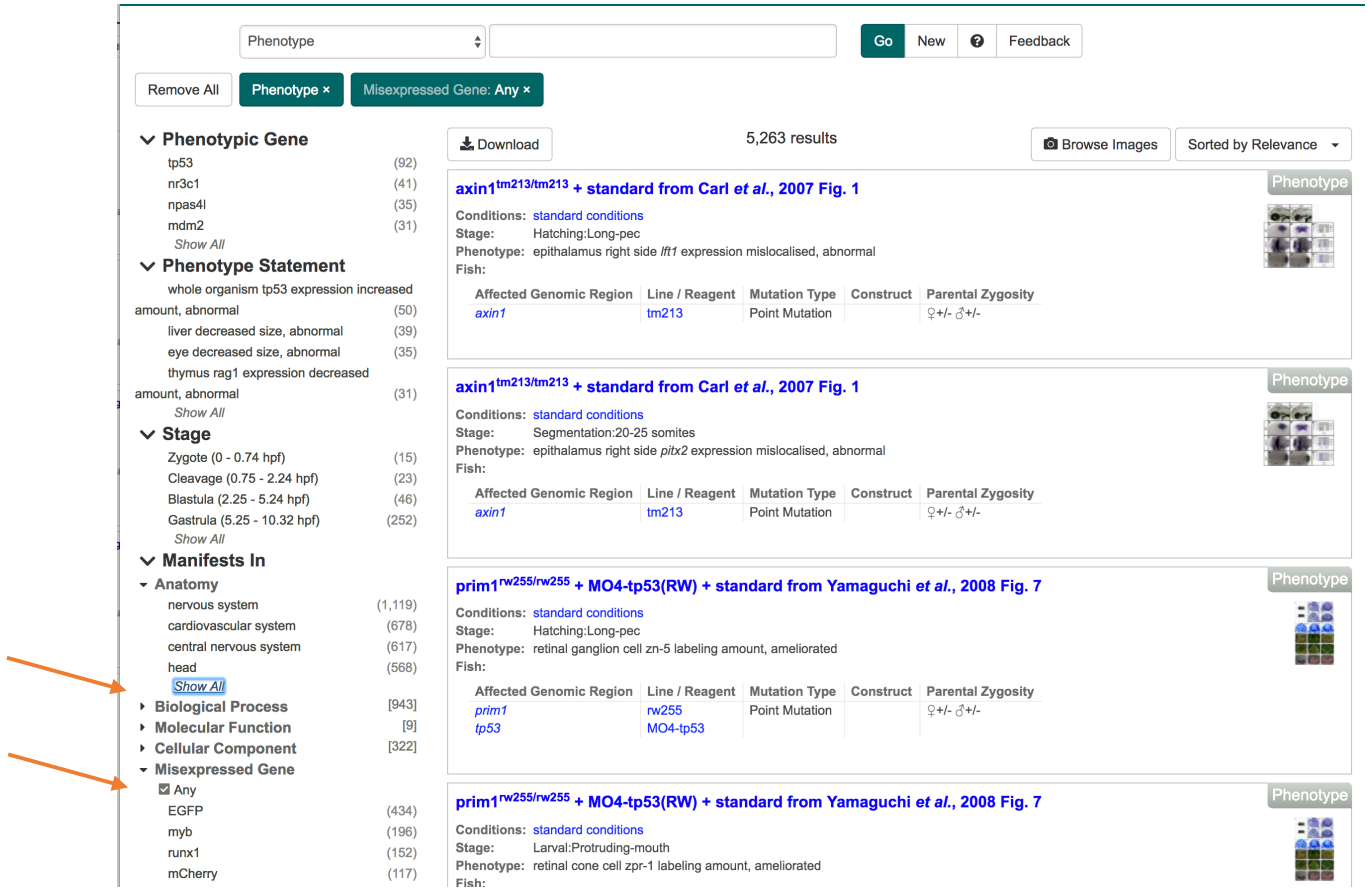


Results:

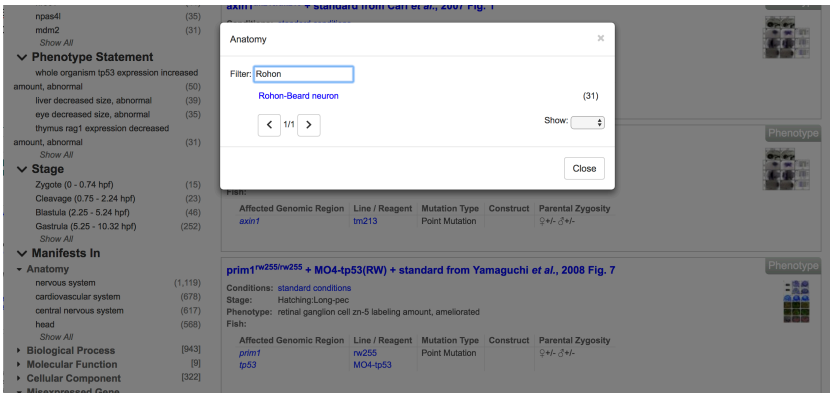

Phenotype
Go
New
Feedback
Remove All
Phenotype ×
Misexpressed Gene: pitx2 ×
Phenotypic Gene
klf8 (2)
ssuh2rs1 (2)
vps4b (2)
axin1 (1)
Show All
Phenotype Statement
tooth placode bmp2a expression absent, abnormal (4)
tooth placode dlx2b expression absent, abnormal (4)
tooth placode pitx2 expression decreased amount, abnormal (4)
tooth placode pitx2 expression spatial pattern, abnormal (4)
Show All
Stage
Segmentation (10.33 - 23.99 hpf) (7)
Pharyngula (24.00 - 47.99 hpf) (1)
Hatching (48.00 - 71.99 hpf) (4)
Manifests In
Anatomy
mesoderm (6)
primary germ layer (6)
epithelium (5)
lateral plate mesoderm (5)
Show All
Biological Process [4]
Molecular Function [0]
Cellular Component [0]
Misexpressed Gene
pitx2
Any (12)
bmp2a (4)
dlx2b (4)
lft1 (3)
Show All
Download
12 results
Browse Images
Sorted by Relevance
axin1tm213/tm213 + standard from Carl et al., 2007 Fig. 1
Phenotype
Conditions: standard conditions
Stage: Segmentation:20-25 somites
Phenotype: epithalamus right side pitx2 expression mislocalised, abnormal
Fish:
Affected Genomic Region | Line / Reagent | Mutation Type | Construct | Parental Zygosity
axin1 | tm213 | Point Mutation | | ♀+/- ♂+/-
WT + MO1-spaw + standard conditions from Wang et al., 2008 Fig. 2
Phenotype
Conditions: standard conditions
Stage: Segmentation:14-19 somites
Phenotype: notochord lft1 expression absent, abnormal
lateral plate mesoderm pitx2 expression absent, abnormal
Fish:
Affected Genomic Region | Line / Reagent | Mutation Type | Construct | Parental Zygosity
spaw | MO1-spaw
AB/TU + chemical treatment by environment: SB 505124 from Ji et al., 2016 Fig. 8
Phenotype
Conditions: chemical treatment by environment: SB 505124
Stage: Segmentation:14-19 somites
Phenotype: posterior lateral mesoderm pitx2 expression absent, abnormal
left/right pattern formation disrupted, abnormal
posterior lateral mesoderm elovl6 expression absent, abnormal
Fish:
AB + MO1-klf8 + standard from Lin et al., 2017 Fig. 2
Phenotype
Conditions: standard conditions
Stage: Segmentation:20-25 somites
Phenotype: diencephalon in_left_side_of lft1 expression absent, abnormal
lateral plate mesoderm left side pitx2 expression absent, abnormal
heart rudiment lft1 expression absent, abnormal

Expression Phenotype Example 3.
Which fish have gene expression phenotypes in Rohon-Beard neurons?

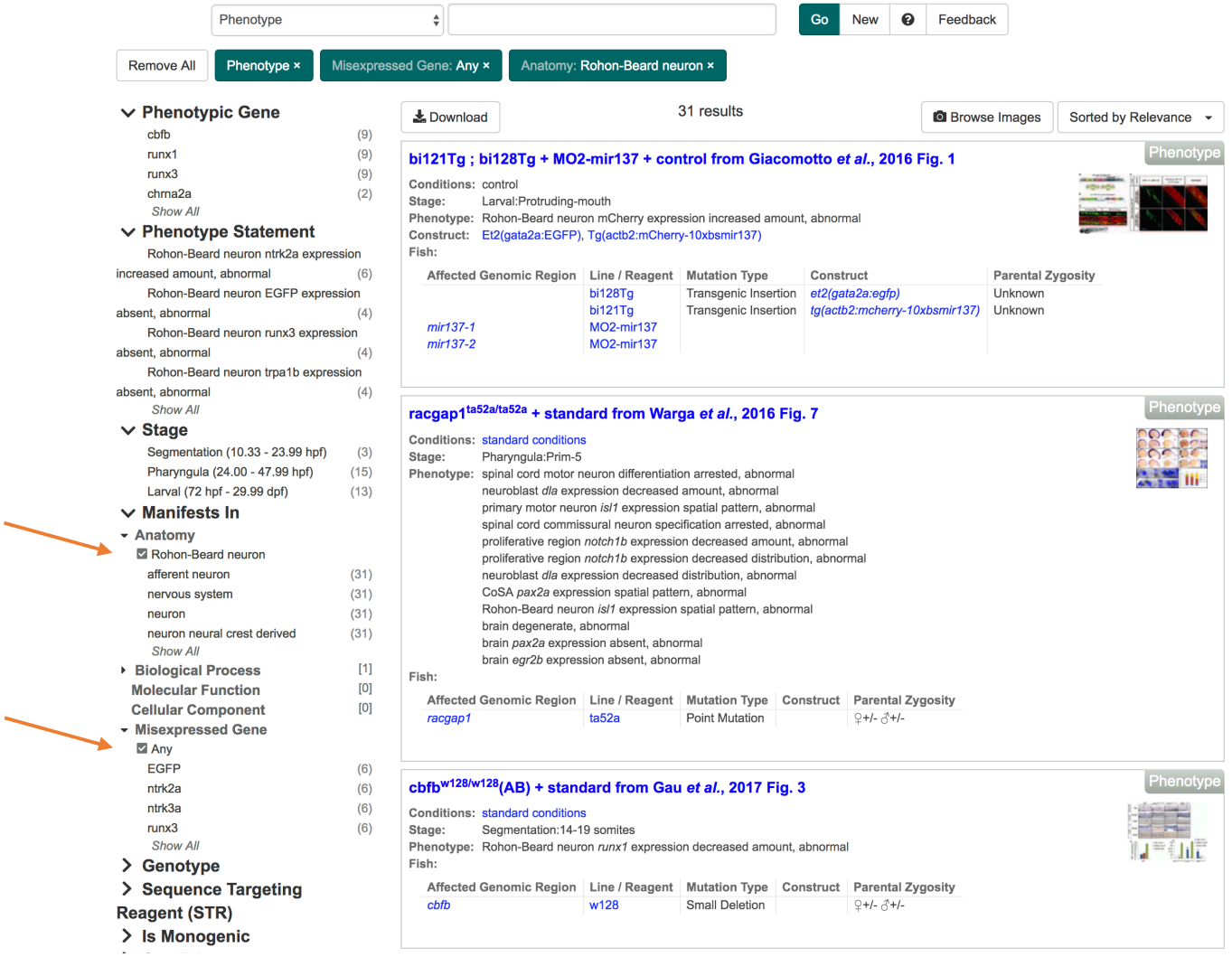
1. Go to the Single Box Search (https://zfin.org/search?q=)
2. Select the Phenotype Category.
3. Go to the “Misexpressed gene” subfacet and select “Any”.
4. Go to the “Manifests in” subfacet and click on “Show All”



5. In the resulting Anatomy popup, type “Rohon” and select “Rohon-Beard neuron” from the list.



Results:


Phenotype
Go
New
Feedback
Remove All
Phenotype ×
Misexpressed Gene: Any ×
Anatomy: Rohon-Beard neuron ×
Phenotypic Gene
cbfb (9)
runx1 (9)
runx3 (9)
chrna2a (2)
Show All
Phenotype Statement
Rohon-Beard neuron ntrk2a expression increased amount, abnormal (6)
Rohon-Beard neuron EGFP expression absent, abnormal (4)
Rohon-Beard neuron runx3 expression absent, abnormal (4)
Rohon-Beard neuron trpa1b expression absent, abnormal (4)
Show All
Stage
Segmentation (10.33 - 23.99 hpf) (3)
Pharyngula (24.00 - 47.99 hpf) (15)
Larval (72 hpf - 29.99 dpf) (13)
Manifests In
Anatomy
Rohon-Beard neuron
afferent neuron (31)
nervous system (31)
neuron (31)
neuron neural crest derived (31)
Show All
Biological Process [1]
Molecular Function [0]
Cellular Component [0]
Misexpressed Gene
Any
EGFP (6)
ntrk2a (6)
ntrk3a (6)
runx3 (6)
Show All
Genotype
Sequence Targeting Reagent (STR)
Is Monogenic
Download
31 results
Browse Images
Sorted by Relevance
Phenotype
bi121Tg ; bi128Tg + MO2-mir137 + control from Giacomotto et al., 2016 Fig. 1
Conditions: control
Stage: Larval:Protruding-mouth
Phenotype: Rohon-Beard neuron mCherry expression increased amount, abnormal
Construct: Et2(gata2a:EGFP), Tg(actb2:mCherry-10xbsmir137)
Fish:
Affected Genomic Region | Line / Reagent | Mutation Type | Construct | Parental Zygosity
bi128Tg | Transgenic Insertion | et2(gata2a:egfp) | Unknown
bi121Tg | Transgenic Insertion | tg(actb2:mcherry-10xbsmir137) | Unknown
mir137-1 | MO2-mir137
mir137-2 | MO2-mir137
Phenotype
racgap1ta52a/ta52a + standard from Warga et al., 2016 Fig. 7
Conditions: standard conditions
Stage: Pharyngula:Prim-5
Phenotype: spinal cord motor neuron differentiation arrested, abnormal
neuroblast dla expression decreased amount, abnormal
primary motor neuron isl1 expression spatial pattern, abnormal
spinal cord commissural neuron specification arrested, abnormal
proliferative region notch1b expression decreased amount, abnormal
proliferative region notch1b expression decreased distribution, abnormal
neuroblast dla expression decreased distribution, abnormal
CoSA pax2a expression spatial pattern, abnormal
Rohon-Beard neuron isl1 expression spatial pattern, abnormal
brain degenerate, abnormal
brain pax2a expression absent, abnormal
brain egr2b expression absent, abnormal
Fish:
Affected Genomic Region | Line / Reagent | Mutation Type | Construct | Parental Zygosity
racgap1 | ta52a | Point Mutation | ♀+/- ♂+/-
Phenotype
cbfbw128/w128(AB) + standard from Gau et al., 2017 Fig. 3
Conditions: standard conditions
Stage: Segmentation:14-19 somites
Phenotype: Rohon-Beard neuron runx1 expression decreased amount, abnormal
Fish:
Affected Genomic Region | Line / Reagent | Mutation Type | Construct | Parental Zygosity
cbfb | w128 | Small Deletion | ♀+/- ♂+/-

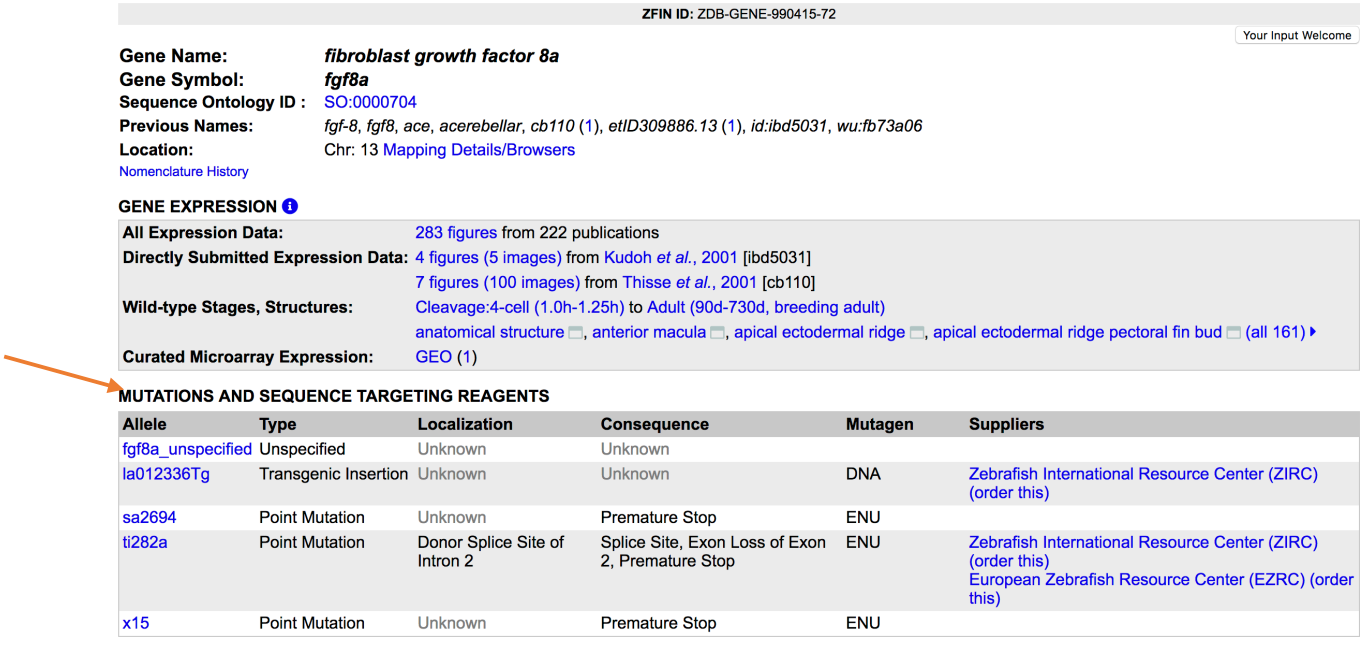
Mutation Details Overview:

- Mutation details information comes from data loads and publications.
- Mutation details are captured as reported by authors or by data provider (e.g. Sanger, Burgess Lab etc.)
- Details from publications are captured as stated by authors and information can be incomplete (e.g. C499T)
- Details captured include changes at the DNA/cDNA level, and transcript and protein consequences

Mutation Details Example 1.

What mutants are available for the *fgf8a* gene and what are their details?

1. Type fgf8a into the Single Box Search, and select the *fgf8a* gene from the results.
2. On the *fgf8a* gene page, scroll down to the "MUTATIONS AND SEQUENCE TARGETING REAGENTS" section



ZFIN ID: ZDB-GENE-990415-72

Your Input Welcome

Gene Name: ***fibroblast growth factor 8a***
Gene Symbol: ***fgf8a***
Sequence Ontology ID : SO:0000704
Previous Names: *fgf-8*, *fgf8*, *ace*, *acerebellar*, *cb110* (1), *etID309886.13* (1), *id:ibd5031*, *wu:fb73a06*
Location: Chr: 13 Mapping Details/Browsers
Nomenclature History

GENE EXPRESSION

All Expression Data:	283 figures from 222 publications
Directly Submitted Expression Data:	4 figures (5 images) from Kudoh *et al.*, 2001 [ibd5031]
	7 figures (100 images) from Thisse *et al.*, 2001 [cb110]
Wild-type Stages, Structures:	Cleavage:4-cell (1.0h-1.25h) to Adult (90d-730d, breeding adult)
	anatomical structure, anterior macula, apical ectodermal ridge, apical ectodermal ridge pectoral fin bud (all 161)
Curated Microarray Expression:	GEO (1)

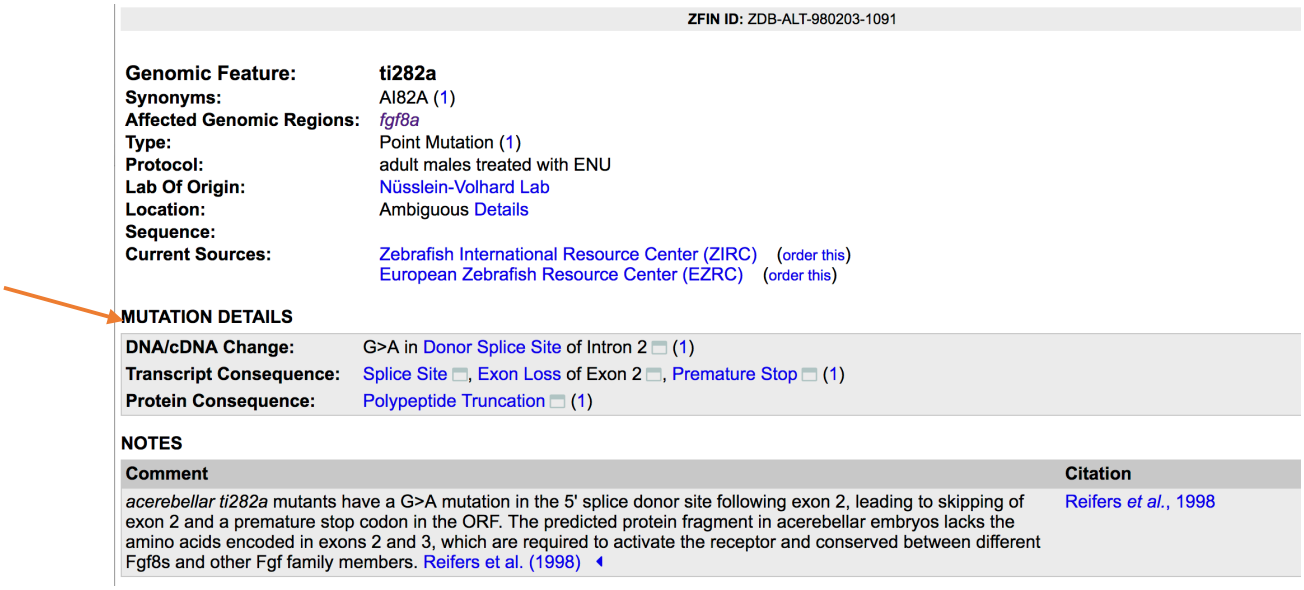
MUTATIONS AND SEQUENCE TARGETING REAGENTS

Allele	Type	Localization	Consequence	Mutagen	Suppliers
fgf8a_unspecified	Unspecified	Unknown	Unknown		
la012336Tg	Transgenic Insertion	Unknown	Unknown	DNA	Zebrafish International Resource Center (ZIRC) (order this)
sa2694	Point Mutation	Unknown	Premature Stop	ENU	
ti282a	Point Mutation	Donor Splice Site of Intron 2	Splice Site, Exon Loss of Exon 2, Premature Stop	ENU	Zebrafish International Resource Center (ZIRC) (order this) European Zebrafish Resource Center (EZRC) (order this)
x15	Point Mutation	Unknown	Premature Stop	ENU	

Mutation Details Example 2.

What are the mutation details for the ti282a mutant?

1. From the fgf8a gene page "MUTATIONS AND SEQUENCE TARGETING REAGENTS" section, click on "ti282a".
2. Go to the "MUTATION DETAILS" section of the ti282a page.



ZFIN ID: ZDB-ALT-980203-1091

Genomic Feature:	**ti282a**
Synonyms:	AI82A (1)
Affected Genomic Regions:	*fgf8a*
Type:	Point Mutation (1)
Protocol:	adult males treated with ENU
Lab Of Origin:	Nüsslein-Volhard Lab
Location:	Ambiguous Details
Sequence:	
Current Sources:	Zebrafish International Resource Center (ZIRC) (order this) European Zebrafish Resource Center (EZRC) (order this)

MUTATION DETAILS

DNA/cDNA Change:	G>A in Donor Splice Site of Intron 2 (1)
Transcript Consequence:	Splice Site, Exon Loss of Exon 2, Premature Stop (1)
Protein Consequence:	Polypeptide Truncation (1)

NOTES

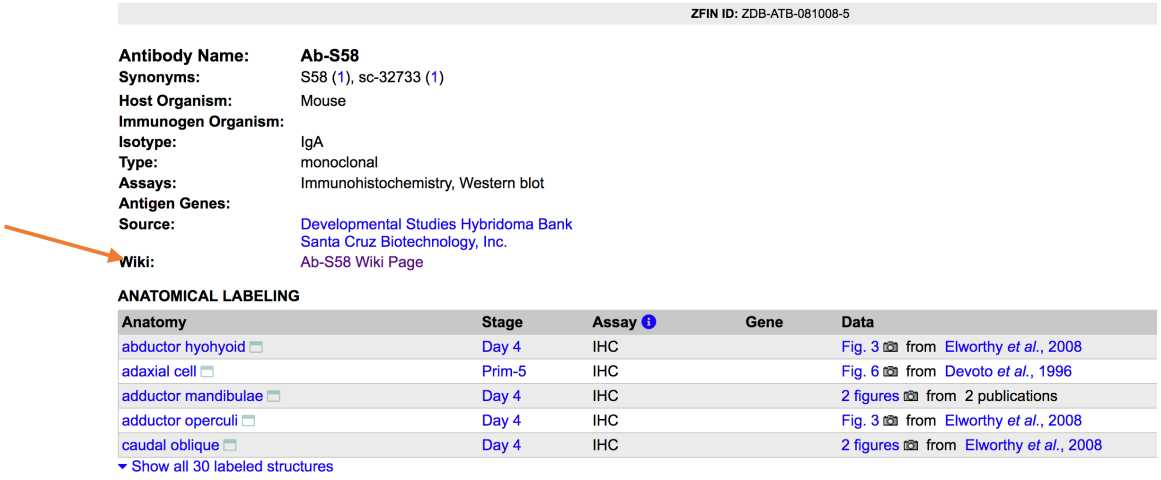
Comment	Citation
acerebellar ti282a mutants have a G>A mutation in the 5' splice donor site following exon 2, leading to skipping of exon 2 and a premature stop codon in the ORF. The predicted protein fragment in acerebellar embryos lacks the amino acids encoded in exons 2 and 3, which are required to activate the receptor and conserved between different Fgf8s and other Fgf family members. Reifers et al. (1998)	Reifers *et al.*, 1998

The Antibody and Protocol Wikis

ZFIN continues to provide a platform for researchers to contribute their antibody data and experimental protocols, in the **Antibody and Protocol Wikis** (https://wiki.zfin.org/)**,** linked from the ZFIN home page. Users can add antibodies and experimental protocols to the Wiki, and can comment on existing antibodies and protocols. The community-created antibodies in the Wiki are not in the ZFIN database, but can be searched in the Wiki (https://wiki.zfin.org/display/AB). ZFIN-curated antibodies are also represented in the Antibody wiki, and linked to/from the ZFIN antibody page.

Antibody Wiki Example

Use the "Ab-S58 Wiki Page" link on the ZFIN **Ab-S58** antibody page to find user comments on the antibody. Does the antibody work with paraformaldehyde-fixed tissue?



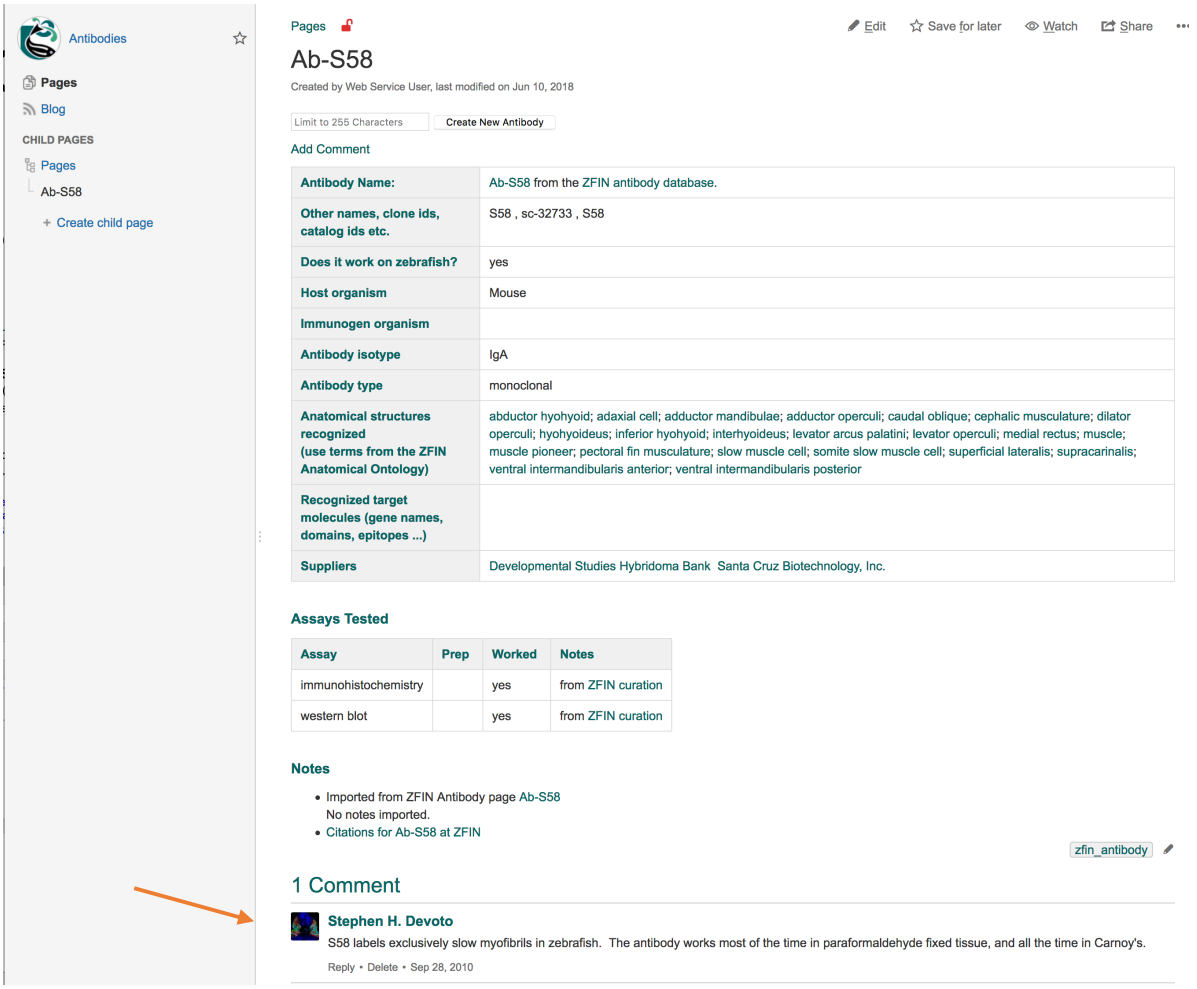
ZFIN ID: ZDB-ATB-081008-5

Antibody Name: **Ab-S58**
Synonyms: S58 (1), sc-32733 (1)
Host Organism: Mouse
Immunogen Organism:
Isotype: IgA
Type: monoclonal
Assays: Immunohistochemistry, Western blot
Antigen Genes:
Source: Developmental Studies Hybridoma Bank
Santa Cruz Biotechnology, Inc.
Wiki: Ab-S58 Wiki Page

ANATOMICAL LABELING

Anatomy	Stage	Assay	Gene	Data
abductor hyohyoid	Day 4	IHC		Fig. 3 from Elworthy *et al.*, 2008
adaxial cell	Prim-5	IHC		Fig. 6 from Devoto *et al.*, 1996
adductor mandibulae	Day 4	IHC		2 figures from 2 publications
adductor operculi	Day 4	IHC		Fig. 3 from Elworthy *et al.*, 2008
caudal oblique	Day 4	IHC		2 figures from Elworthy *et al.*, 2008

Show all 30 labeled structures



Extra exercise: Can you find an aco2 antibody in ZFIN? In the Community Antibody Wiki?

To search the Antibody Wiki:

- https://wiki.zfin.org/display/AB

To search the ZFIN database:

- **ZFIN Antibody Search**: https://zfin.org/action/antibody/search
- **Single-box Antibody Category Search**: https://zfin.org/search?q=&fq=category%3A%22Antibody%22&category=Antibody

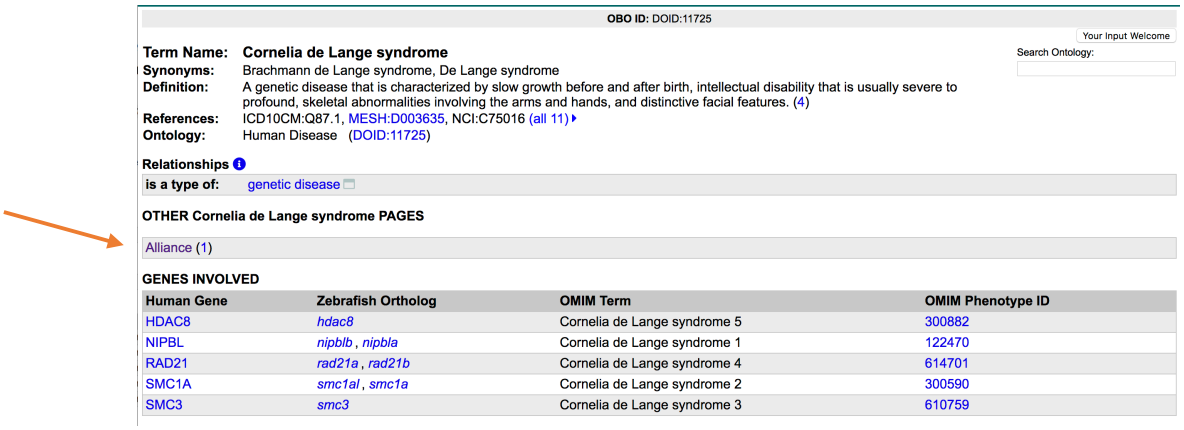
Note: The ZFIN Antibody Search and Single-Box Search currently do **NOT** search the Antibody Wiki.

Alliance Overview:

ZFIN is now part of the **Alliance of Genome Resources**, (https://www.alliancegenome.org/) a consortium of 6 model organism databases (MODs) (MGI, RGD, SGD, WormBase, FlyBase, ZFIN) and the Gene Ontology (GO) database. The goal of the **Alliance** is to provide users a central location to easily access and compare human and model organism data. Current data includes genes, alleles, GO, Orthology, Human Disease and JBrowse, with links to Expression data at the MODs and GEO. New data continues to be added.

Alliance Example 1.
Use the Alliance link on the ZFIN **Cornelia de Lange syndrome** disease page to find a **mouse allele** that is associated with the syndrome.

1. Navigate to the ZFIN Cornelia de Lange syndrome disease page
2. Scroll down to the "OTHER Cornelia de Lange syndrome PAGES" section.
3. Click on "Alliance".



OBO ID: DOID:11725

Your Input Welcome

Term Name: **Cornelia de Lange syndrome**
Search Ontology:
Synonyms: Brachmann de Lange syndrome, De Lange syndrome
Definition: A genetic disease that is characterized by slow growth before and after birth, intellectual disability that is usually severe to profound, skeletal abnormalities involving the arms and hands, and distinctive facial features. (4)
References: ICD10CM:Q87.1, MESH:D003635, NCI:C75016 (all 11)
Ontology: Human Disease (DOID:11725)

Relationships
is a type of: genetic disease

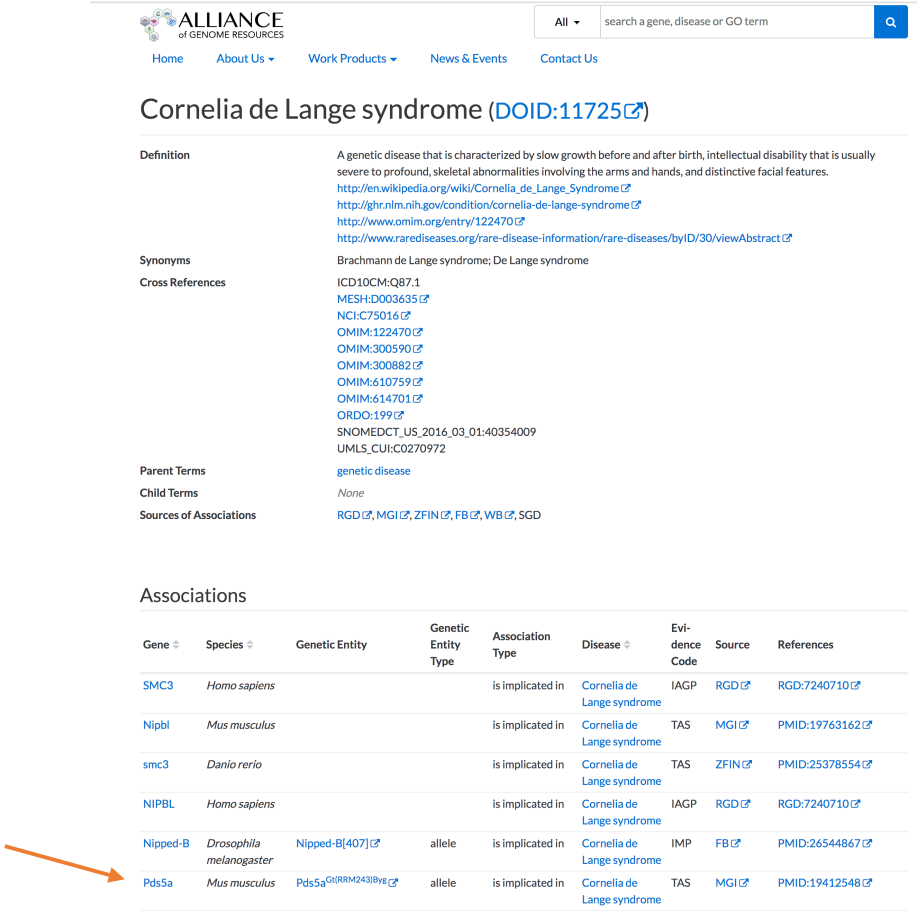
OTHER Cornelia de Lange syndrome PAGES

Alliance (1)

GENES INVOLVED

Human Gene	Zebrafish Ortholog	OMIM Term	OMIM Phenotype ID
HDAC8	*hdac8*	Cornelia de Lange syndrome 5	300882
NIPBL	*nipblb* , *nipbla*	Cornelia de Lange syndrome 1	122470
RAD21	*rad21a* , *rad21b*	Cornelia de Lange syndrome 4	614701
SMC1A	*smc1al* , *smc1a*	Cornelia de Lange syndrome 2	300590
SMC3	*smc3*	Cornelia de Lange syndrome 3	610759

4. On the Alliance disease page, scroll down to the "Associations" section, and find the mouse Pds5a$^{Gt(RRM243)Byg}$ allele



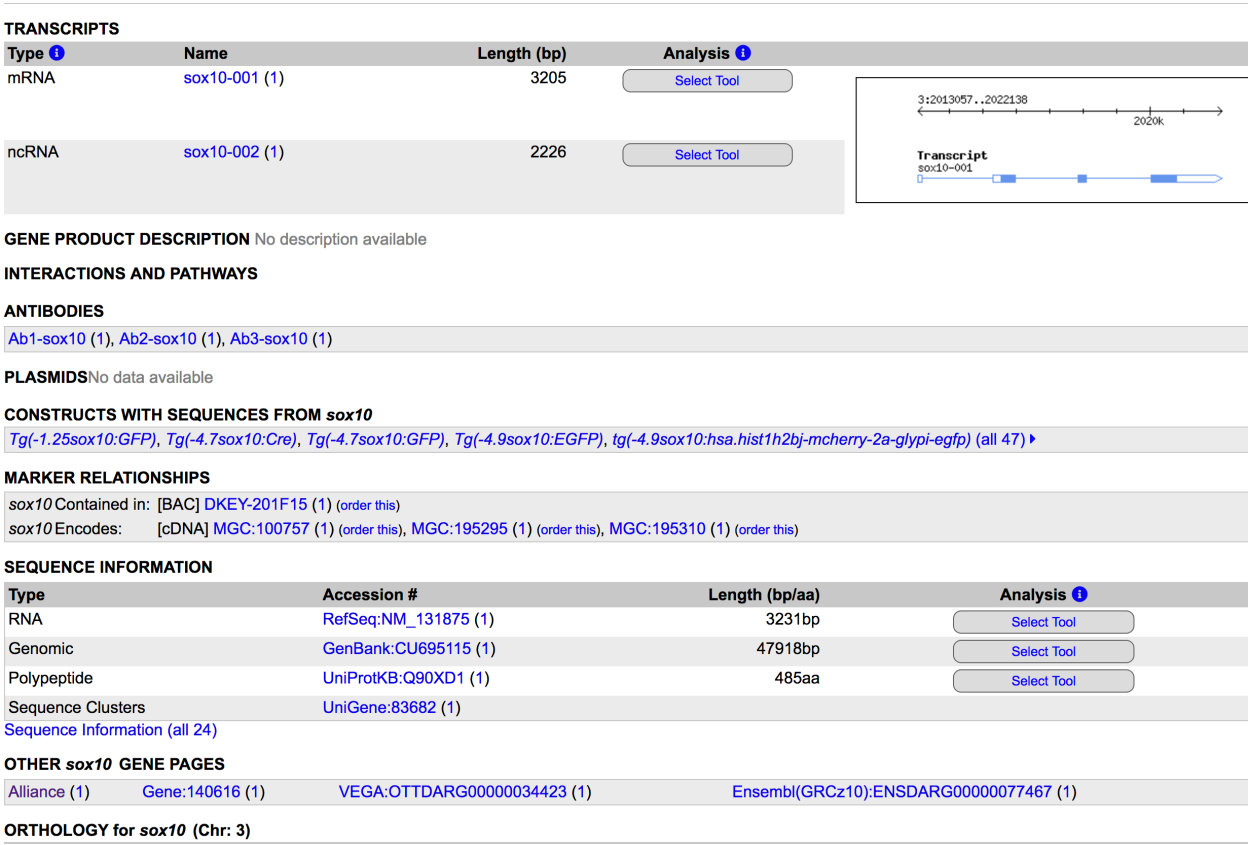
Click on the Pds5a$^{Gt(RRM243)Byg}$ allele symbol to navigate to the MGI Pds5a$^{Gt(RRM243)Byg}$ page.

http://www.informatics.jax.org/allele/MGI:3847257

Alliance Example 2

Use the Alliance link on the ZFIN ***sox10*** gene page to explore GO and Orthology on the Alliance zebrafish ***sox10*** gene page.

1. Navigate to the ZFIN ***sox10*** gene page
2. Scroll down to the "OTHER *sox10* gene pages" section.



TRANSCRIPTS

Type	Name	Length (bp)	Analysis
mRNA	sox10-001 (1)	3205	Select Tool
ncRNA	sox10-002 (1)	2226	Select Tool

GENE PRODUCT DESCRIPTION No description available

INTERACTIONS AND PATHWAYS

ANTIBODIES

Ab1-sox10 (1), Ab2-sox10 (1), Ab3-sox10 (1)

PLASMIDS No data available

CONSTRUCTS WITH SEQUENCES FROM *sox10*

Tg(-1.25sox10:GFP), *Tg(-4.7sox10:Cre)*, *Tg(-4.7sox10:GFP)*, *Tg(-4.9sox10:EGFP)*, *tg(-4.9sox10:hsa.hist1h2bj-mcherry-2a-glypi-egfp)* (all 47)

MARKER RELATIONSHIPS

sox10 Contained in: [BAC] DKEY-201F15 (1) (order this)
sox10 Encodes: [cDNA] MGC:100757 (1) (order this), MGC:195295 (1) (order this), MGC:195310 (1) (order this)

SEQUENCE INFORMATION

Type	Accession #	Length (bp/aa)	Analysis
RNA	RefSeq:NM_131875 (1)	3231bp	Select Tool
Genomic	GenBank:CU695115 (1)	47918bp	Select Tool
Polypeptide	UniProtKB:Q90XD1 (1)	485aa	Select Tool
Sequence Clusters	UniGene:83682 (1)		

Sequence Information (all 24)

OTHER *sox10* GENE PAGES

Alliance (1) Gene:140616 (1) VEGA:OTTDARG00000034423 (1) Ensembl(GRCz10):ENSDARG00000077467 (1)

ORTHOLOGY for *sox10* (Chr: 3)

3. Click on "Alliance"

4. On the Alliance *sox10* gene page, explore the **Function - GO Annotations** ribbon and **Orthology** sections. Note the multiple sources for Orthology calls, and the ability to filter Orthology by Stringency.

ALLIANCE of GENOME RESOURCES

All | search a gene, disease or GO term

Home | About Us | Work Products | News & Events | Contact Us

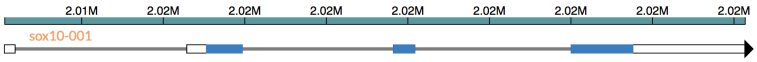
sox10

Symbol	sox10
Name	SRY (sex determining region Y)-box 10
Synonyms	zgc:100757, gos, sox10b, cls, colourless, golas, colorless
Biotype	gene
Description	*Not Available*
Genomic Resources	ENSEMBL:ENSDARG00000077467 NCBI_Gene:140616 UniProtKB:A0PJQ1 UniProtKB:Q6DBW6 UniProtKB:Q90XD1
Additional Information	Literature

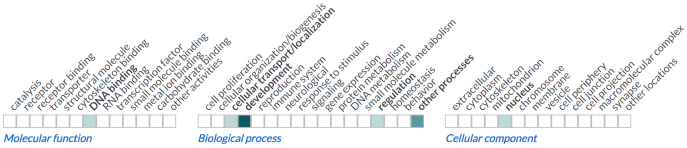
Species *Danio rerio*
Source ZFIN:ZDB-GENE-011207-1

Sequence Feature Viewer

Genome location Chr3:2013057...2022135 (9.08 kb)
Assembly version GRCv10



Function – GO Annotations



Danio rerio | See All sox10 Annotations | Show associations

Orthology

Focus gene sox10 - (Species: Danio rerio)
Gene tree PANTHER:PTHR10270

Stringency: Stringent filter (default) | Moderate filter | No filter / Show all

Show additional filters | Reset filters

Species	Gene symbol	Count	Best	Best reverse	Ensembl Compara	HGNC	Hieranoid	InParanoid	OMA	OrthoFinder	OrthoInspector	PANTHER	PhylomeDB	Roundup	TreeFam	ZFIN
Homo sapiens	SOX10	10 of 11	Yes	Yes	☑	☑	☑	☑	☑	☑	☑	☑	☑	☐	☑	
Mus musculus	Sox10	10 of 11	Yes	Yes	☑	☑	☑	☑	☑	☑	☑	☑	☑	☐	☑	
Rattus norvegicus	Sox10	8 of 9	Yes	Yes	☑	☑	☑	☑	☑	☑	☑	☑		☐		
Drosophila melanogaster	Sox100B	6 of 11	Yes	Yes	☑	☐	☑	☐	☑	☑	☑	☐	☑	☐	☐	

Learn about how orthologs are identified

Disease Associations