

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 (zfinadm@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”.

The ZFIN Information Network

Research General Information ZIRC Downloads

Any [Go] New [Feedback]

Category	Count
Gene / Transcript	(81,963)
Expression	(220,973)
Phenotype	(57,402)
Human Disease	(8,794)
Fish	(36,785)
Reporter Line	(6,643)
Mutation / Tg	(65,096)
Construct	(4,454)
Sequence Targeting Reagent (STR)	(13,207)
Antibody	(2,769)
Marker / Clone	(62,667)
Figure	(98,550)
Anatomy / GO	(47,962)
Community	(9,876)
Publication	(33,628)

Ways to start:

- Enter search terms in the search box at the top of the screen and Click "Go". If you want to limit your results to a specific category (gene, fish, etc.), choose it from the pull down menu labeled "Any" before clicking "Go".

OR

- Click on a Category in the list on the left, and browse data using the resulting filters to drill down and narrow your search results.

Send Feedback:
As you explore, if you see ways to make the tool better, please tell us! Your input is essential to help us improve this tool to meet your needs. Send feedback with the "Send Feedback" button near the search box. Please include the questions you were trying to answer, what you tried, what you expected, what results you got, etc. The more information the better.

Human Disease [Go] [New] [Feedback]

Remove All Human Disease x

Gene (16)
tgif3 (9)
tgif2 (9)
kras (9)
pi3ca (9)
Show All

Disease Model

Fish (314)
Any (80)
WT (34)
AB (13)
y1Tg (13)
Show All

Conditions (227)
standard or control (22)
cancer xenotransplantation (8)
heat shock (6)
chemical treatment: ethanol (6)
Show All

Download 8,794 results Sorted by Relevance

2-hydroxyglutaric aciduria (DOI:0050573) Human Disease
Definition: An amino acid metabolic disorder that is an autosomal recessive neurometabolic disorder characterized by the significant elevation of urinary levels of hydroxyglutaric acid causing progressive brain damage.

3-M syndrome (DOI:0060241) Human Disease
Synonyms: Le Merer syndrome, Yakut short stature syndrome, dolichospondylic dysplasia, Miller-McKusick-Malvaux syndrome, three M syndrome, gloomy face syndrome
Definition: An autosomal recessive disease characterized by dwarfism, facial dysmorphia and skeletal abnormalities.
Related zebrafish gene (2)

3MC syndrome (DOI:0060225) Human Disease
Synonyms: craniofacial-urinar-renal syndrome, oculopalatoskeletal syndrome
Definition: A syndrome characterized by blepharophimosis, blepharoptosis, highly arched eyebrows hypertelorism, cleft lip and palate, postnatal growth deficiency, cognitive impairment, hearing loss and, in a smaller percentage of cases, craniosynostosis, radioulnar synostosis and genital and vesicovaginal anomalies. It encompasses four disorders that were previously designated the Malpuech, Michels, Mingarelli and Carnevale syndromes.
Related zebrafish gene (1) | Publication (1)



Results

ZFIN The Zebrafish Information Network [Research] [General Information] [ZIRC] [Downloads] [Login] [Follow]

Human Disease [Go] [New] [Feedback]

Remove All Human Disease x Fish: Any x

Gene (5)
pi3ca (4)
cdh1 (4)
tp53 (4)
akt1 (3)
Show All

Disease Model

Fish (314)
Any (60)
WT (34)
AB (13)
y1Tg (13)
mitfa^{29a2}; mpy17^{91a9} (6)
Show All

Conditions (227)
standard or control (22)
cancer xenotransplantation (8)
heat shock (6)
chemical treatment: ethanol (6)
Show All

Download 314 results Sorted by Relevance

3-methylglutaconic aciduria type 3 (DOI:0110004) Human Disease
Synonyms: Iraqi-Jewish optic atrophy plus, autosomal recessive optic atrophy type 3, MGA3, Costeff optic atrophy syndrome, 3-methylglutaconic aciduria type III, infantile optic atrophy with chorea and spastic paraplegia, Costeff syndrome, autosomal recessive optic atrophy plus syndrome
Definition: A 3-methylglutaconic aciduria that has_material_basis_in mutation in the OPA3 gene.
Related zebrafish gene (1) | Disease Models (1) | Publication (1)

ablepharon macrostomia syndrome (DOI:0060550) Human Disease
Synonyms: poikiloderma with neutropenia, Clericuzio type
Definition: A syndrome characterized by the association of ablepharon, macrostomia, abnormal external ears, syndactyly of the hands and feet, skin findings (such as dry and coarse skin or redundant folds of skin), absent or sparse hair, genital malformations and developmental delay. Other reported manifestations include malar hypoplasia, absent or hypoplastic nipples, umbilical abnormalities and growth retardation. It is that has material basis in heterozygous mutation in the TWIST2 gene on chromosome 2q37.
Related zebrafish gene (1) | Disease Models (1) | Publication (2)

acute kidney failure (DOI:3021) Human Disease
Disease Models (3) | Publication (2)

acute lymphocytic leukemia (DOI:0952) Human Disease
Synonyms: acute lymphoblastic leukemia
Definition: A lymphoblastic leukemia that is characterized by over production of lymphoblasts.
Related zebrafish gene (8) | Disease Models (1) | Publication (1)

acute myeloid leukemia (DOI:9119) Human Disease
Synonyms: acute myelogenous leukemia, acute myeloblastic leukemia, Leukemia, Myelocytic, acute, AML - acute Myeloid Leukemia
Definition: A myeloid leukemia that is characterized by the rapid growth of abnormal white blood cells that accumulate in the bone marrow and interfere with the production of normal blood cells.
Related zebrafish gene (27) | Disease Models (8) | Publication (12)

acute porphyria (DOI:3133) Human Disease
Synonyms: hepatic porphyria
Definition: A porphyria that has_symptom abdominal pain, has_symptom neuropathy, has_symptom autonomic instability and has_symptom psychosis.
Related zebrafish gene (1) | Disease Models (2) | Publication (1)

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.

The screenshot shows the ZFIN website interface. At the top, there is a navigation bar with the ZFIN logo and the text "The Zebrafish Information Network". Below the navigation bar, there are tabs for "Research", "General Information", and "ZIRC". A search bar is located in the center, with the text "mecp2" entered. A dropdown menu is open below the search bar, showing several search results: "mecp2", "mecp2_unrecovered", "mecp2^{fh232/mh232} + MO5-mecp2", "mecp2-001", and "mecp2-002". The "mecp2" result is highlighted in blue. To the right of the search bar, there is a "Go" button. Below the search bar, there is a sidebar with various categories and links, including "Genes / Markers / Nomenclature Conventions", "BLAST at ZFIN", "GBrowse genome browser", "Gene Expression", "Antibodies", "Mutants / Knockdowns / Transgenics", "Constructs", "Anatomy / GO / Human Disease", "Publications", "Community", "Data", and "Downloads". The main content area on the right contains information about the Zebrafish International Resource Center, Genomics, Zebrafish Programs, and News. At the bottom of the page, there is a footer with navigation links and a disclaimer.

Search ZFIN

Genes / Markers / **mecp2** **Zebrafish International Resource Center**
 Nomenclature Conventions: Request: Fish Lines, ESTs/cDNAs, Monoclonal
 Obtain approval for g Antibodies, The Zebrafish Book, Paramecia
BLAST at ZFIN mec2^{fh232/mh232} + MO5-mecp2 Submit Fish Lines
GBrowse genome browser mec2-001 Health Services
Gene Expression mec2-002 **Genomics**
Antibodies Data mining: ZebrafishMine, BioMart
 Browse genome: ZFIN, Ensembl, Vega,
 GRC, UCSC, NCBI, FishMap
Mutants / Knockdowns / Transgenics BLAST: ZFIN, Ensembl, NCBI, MGH
 Wild-Type Lines Find cDNAs and ESTs at ZGC
 Line Designations More Zebrafish Genome Resources
 Submit mutant/transgenic line names Other Fish Genomes and Model Organism
 Databases
Constructs **Zebrafish Programs**
 ZF-HEALTH, Husbandry Resources, more...
Anatomy / GO / Human Disease **News**
 Anatomy Atlases and Resources Zebrafish Models for Human Eye Diseases
 (Sept. 14-15, 2018 - Freiburg, Germany) May
 18, 2018
 Late registration and abstract submission are
 now open for the International Zebrafish
 Conference May 3, 2018
 ZFIN Newsletters, News Archive

Publications
 Author Guidelines

Community
 Wiki: Protocols, Antibodies
 Jobs, Meetings, Newsgroup
 People, Labs, Companies
 Educational Resources
 The Zebrafish Book

Data
 Downloads
 Submit
 Statistics
 Data Model

ALLIANCE
 of GENOME RESOURCES
 FOUNDING MEMBER

Home Help and Tips Glossary Committees Citing ZFIN Contact About ZFIN Jobs
 Disclaimer, limitations, copyright © University of Oregon, 1994-2018, Eugene, Oregon.
 ZFIN logo design by Karl Pape, University of Oregon

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 <input type="checkbox"/>	Rett syndrome	312750
	Rett syndrome, atypical	312750
	Rett syndrome, preserved speech variant	312750
syndromic X-linked intellectual disability Lubs type <input type="checkbox"/>	Mental retardation, X-linked syndromic, Lubs type	300260
X-linked intellectual disability-psychosis-macroorchidism syndrome <input type="checkbox"/>	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** Search Onto

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	smc1a1 , smc1a	Cornelia de Lange syndrome 2	300590
SMC3	smc3	Cornelia de Lange syndrome 3	610759

ZEBRAFISH MODELS

Fish	Conditions	Citations
AB + MO3-nipblb <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(1)
WT + MO1-smc3 <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(1)
WT + MO2-rad21a <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(1)
rad21a^{hi2529Tg} <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(1)
AB + MO1-nipbla + MO1-nipblb <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(2)
WT + MO1-nipbla + MO1-nipblb <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(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

Term Name: hepatocellular carcinoma Search Ontology:

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	<i>apc</i>	Hepatoblastoma, somatic	114550
AXIN1	<i>axin1</i>	Hepatocellular carcinoma, somatic	114550
CASP8	<i>casp8</i>	Hepatocellular carcinoma, somatic	114550
CTNNB1	<i>ctnnb2</i> , <i>ctnnb1</i>	Hepatocellular carcinoma, somatic	114550
IGF2R	<i>igf2r</i>	Hepatocellular carcinoma, somatic	114550

▼ Show all 9

ZEBRAFISH MODELS

Fish	Conditions	Citations
WT <input type="checkbox"/>	cancer xenotransplantation <input type="checkbox"/>	(1)
gz24Tg <input type="checkbox"/>	chemical treatment: mifepristone <input type="checkbox"/>	(1)
gz25Tg <input type="checkbox"/>	chemical treatment: doxycycline monohydrate <input type="checkbox"/>	(1)
gz26Tg <input type="checkbox"/>	chemical treatment: doxycycline monohydrate <input type="checkbox"/>	(1)
gz32Tg <input type="checkbox"/>	chemical treatment by environment: doxycycline <input type="checkbox"/>	(1)
	chemical treatment: doxycycline <input type="checkbox"/>	(1)
	chemical treatment: doxycycline monohydrate <input type="checkbox"/>	(1)
	chemical treatment by environment: doxycycline <input type="checkbox"/>	(1)
	chemical treatment: doxycycline monohydrate <input type="checkbox"/>	(1)
	chemical treatment: doxycycline <input type="checkbox"/>	(1)
nn1005Tg <input type="checkbox"/>	standard conditions <input type="checkbox"/>	(1)
y1Tg <input type="checkbox"/>	cancer xenotransplantation <input type="checkbox"/>	(1)
gz15Tg; gz26Tg <input type="checkbox"/>	chemical treatment: doxycycline <input type="checkbox"/>	(1)
gz25Tg/+; gz26Tg/+ <input type="checkbox"/>	chemical treatment: doxycycline <input type="checkbox"/>	(1)
mitfa ^{w2/w2} ; mpv17 ^{a9/a9} <input type="checkbox"/>	cancer xenotransplantation <input type="checkbox"/>	(1)
mpv17 ^{b18/b18} ; gz32Tg; mitfa ^{w2/w2} <input type="checkbox"/>	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 <input type="checkbox"/>	standard conditions <input type="checkbox"/>	Pistocchi <i>et al.</i>, 2013

GENE EXPRESSION ⓘ
Gene expression in AB + MO3-nipblb

RNA expression

Expressed Gene	Structure	Conditions	Figures
axin2	hindbrain <input type="checkbox"/>	standard conditions <input type="checkbox"/>	Fig. 4 from Pistocchi <i>et al.</i>, 2013
ccnd1	whole organism <input type="checkbox"/>	standard conditions <input type="checkbox"/>	Fig. 6 from Fazio <i>et al.</i>, 2016 Fig. 5 from Pistocchi <i>et al.</i>, 2013
wnt1	hindbrain <input type="checkbox"/>	standard conditions <input type="checkbox"/>	Fig. 4 from Pistocchi <i>et al.</i>, 2013

Protein expression

Antibody	Antigen Genes	Structure	Conditions	Figures
Ab1-ccnd1	ccnd1	whole organism <input type="checkbox"/>	standard conditions <input type="checkbox"/>	Fig. 6 from Fazio <i>et al.</i>, 2016

Reporter gene expression No data available

PHENOTYPE ⓘ
Phenotype in AB + MO3-nipblb

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

Go
New
?
Feedback

Remove All
Phenotype x
Misexpressed Gene: Any x

Phenotypic Gene

tp53 (92)

nr3c1 (41)

npas4l (35)

mdm2 (31)

Show All

Phenotype Statement

whole organism tp53 expression increased amount, abnormal (50)

liver decreased size, abnormal (39)

eye decreased size, abnormal (35)

thymus rag1 expression decreased amount, abnormal (31)

Show All

Stage

Zygote (0 - 0.74 hpf) (15)

Cleavage (0.75 - 2.24 hpf) (23)

Blastula (2.25 - 5.24 hpf) (46)

Gastrula (5.25 - 10.32 hpf) (252)

Show All

Manifests In

Anatomy

nervous system (1,118)

cardiovascular system (678)

central nervous system (616)

head (568)

Show All

Biological Process [943]

Molecular Function [9]

Cellular Component [322]

Misexpressed Gene

Any (433)

EGFP (196)

myb (152)

runx1 (117)

Show All

Genotype

Sequence Targeting Reagent (STR)

Is Monogenic

Conditions

Has Image

Download
5,260 results
Browse Images
Sorted by Relevance

axin1^{tm213/tm213} + standard from Carl et al., 2007 Fig. 1

Conditions: [standard conditions](#)

Stage: Hatching:Long-pec

Phenotype: epithalamus right side *irf1* expression mislocalised, abnormal

Fish:

Affected Genomic Region	Line / Reagent	Mutation Type	Construct	Parental Zygosity
axin1	tm213	Point Mutation		♀+/- ♂+/-

axin1^{tm213/tm213} + standard from Carl et al., 2007 Fig. 1

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		♀+/- ♂+/-

prim1^{rw255/rw255} + MO4-tp53(RW) + standard from Yamaguchi et al., 2008 Fig. 7

Conditions: [standard conditions](#)

Stage: Hatching:Long-pec

Phenotype: retinal ganglion cell zn-5 labeling amount, ameliorated

Fish:

Affected Genomic Region	Line / Reagent	Mutation Type	Construct	Parental Zygosity
prim1	rw255	Point Mutation		♀+/- ♂+/-
tp53	MO4-tp53			

prim1^{rw255/rw255} + MO4-tp53(RW) + standard from Yamaguchi et al., 2008 Fig. 7

Conditions: [standard conditions](#)

Stage: Larval:Protruding-mouth

Phenotype: retinal cone cell *zpr-1* labeling amount, ameliorated

Fish:

Affected Genomic Region	Line / Reagent	Mutation Type	Construct	Parental Zygosity
prim1	rw255	Point Mutation		♀+/- ♂+/-
tp53	MO4-tp53			

prim1^{rw255/rw255}(RW) + standard from Yamaguchi et al., 2008 Fig. 7

Conditions: [standard conditions](#)

Stage: Hatching:Long-pec

Phenotype: retina morphology, abnormal

retinal ganglion cell zn-5 labeling absent, abnormal

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 x
Misexpressed Gene: pitx2 x

Download
12 results
Browse Images
Sorted by Relevance

Phenotypic Gene

- klf8 (2)
- ssuh2rs1 (2)
- vps4b (2)
- axin1 (1)
- [Show All](#)

Phenotype Statement

- tooth placode bmp2a expression absent, abnormal (4)
- tooth placode dix2b 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 (12)
- Any (12)
- bmp2a (4)
- dix2b (4)
- lft1 (3)
- [Show All](#)

Affected Genomic Region	Line / Reagent	Mutation Type	Construct	Parental Zygosity
axin1^{tm213}/tm213 + standard from Carl et al., 2007 Fig. 1				
Conditions: standard conditions				
Stage: Segmentation:20-25 somites				
Phenotype: epithalamus right side <i>pitx2</i> expression mislocalised, abnormal				
Fish: 				
axin1	tm213	Point Mutation		♀+/- ♂+/-
WT + MO1-spaw + standard conditions from Wang et al., 2008 Fig. 2				
Conditions: standard conditions				
Stage: Segmentation:14-19 somites				
Phenotype: notochord <i>lft1</i> expression absent, abnormal				
lateral plate mesoderm <i>pitx2</i> expression absent, abnormal				
Fish: 				
spaw	MO1-spaw			
AB/TU + chemical treatment by environment: SB 505124 from Ji et al., 2016 Fig. 8				
Conditions: chemical treatment by environment: SB 505124				
Stage: Segmentation:14-19 somites				
Phenotype: posterior lateral mesoderm <i>pitx2</i> expression absent, abnormal				
left/right pattern formation disrupted, abnormal				
posterior lateral mesoderm <i>elov16</i> expression absent, abnormal				
Fish: 				
AB + MO1-klf8 + standard from Lin et al., 2017 Fig. 2				
Conditions: standard conditions				
Stage: Segmentation:20-25 somites				
Phenotype: diencephalon in_left_side_of <i>lft1</i> expression absent, abnormal				
lateral plate mesoderm left side <i>pitx2</i> expression absent, abnormal				
heart rudiment <i>lft1</i> expression absent, abnormal				
Fish: 				

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”

The screenshot shows the ZFIN search results interface. At the top, there is a search bar with the text 'Phenotype' and buttons for 'Go', 'New', and 'Feedback'. Below the search bar, there are filters for 'Remove All', 'Phenotype x', and 'Misexpressed Gene: Any x'. The main content area displays 5,263 results, sorted by Relevance. The results are organized into sections: Phenotypic Gene, Phenotype Statement, Stage, and Manifests In. The 'Manifests In' section is expanded to 'Anatomy', and a popup window is open showing a list of anatomical structures with 'Rohon-Beard neuron' selected. The popup also includes a 'Filter' field with the text 'Rohon-Beard neuron' and a 'Show' dropdown menu. The background results show several entries with details such as 'Affected Genomic Region', 'Line / Reagent', 'Mutation Type', 'Construct', and 'Parental Zygosity'.

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

This is a close-up view of the 'Anatomy' popup window. The 'Filter' field contains the text 'Rohon'. Below the filter, a list of anatomical structures is displayed, with 'Rohon-Beard neuron' highlighted. The popup also includes navigation arrows, a 'Show' dropdown menu, and a 'Close' button. The background shows the search results page with the 'Anatomy' section expanded.

Results:

Phenotype

Go
New
Feedback

Remove All
Phenotype x
Misexpressed Gene: Any x
Anatomy: Rohon-Beard neuron x

Download
31 results
Browse Images
Sorted by Relevance

Phenotypic Gene

- cbfb (9)
- runx1 (9)
- runx3 (9)
- chma2a (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 (31)
 - 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 (6)
 - EGFP (6)
 - ntrk2a (6)
 - ntrk3a (6)
 - runx3 (6)
 - [Show All](#)
- Genotype**
- Sequence Targeting**
- Reagent (STR)**
- Is Monogenic**

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			

racgap1^{ta52a/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		♀+/- ♂+/-

cbfb^{w128/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), etlD309886.13 (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
<i>fgf8a_unspecified</i>	Unspecified	Unknown	Unknown		
<i>la012336Tg</i>	Transgenic Insertion	Unknown	Unknown	DNA	Zebrafish International Resource Center (ZIRC) (order this)
<i>sa2694</i>	Point Mutation	Unknown	Premature Stop	ENU	
<i>ti282a</i>	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)
<i>x15</i>	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:	<i>fgf8a</i>
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 <input type="checkbox"/> (1)
Transcript Consequence:	Splice Site <input type="checkbox"/> , Exon Loss of Exon 2 <input type="checkbox"/> , Premature Stop <input type="checkbox"/> (1)
Protein Consequence:	Polypeptide Truncation <input type="checkbox"/> (1)
NOTES	
Comment	Citation
<i>acerebellar ti282a</i> 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 ¹	Gene	Data
abductor hyohyoid <input type="checkbox"/>	Day 4	IHC		Fig. 3  from Elworthy et al., 2008
adaxial cell <input type="checkbox"/>	Prim-5	IHC		Fig. 6  from Devoto et al., 1996
adductor mandibulae <input type="checkbox"/>	Day 4	IHC		2 figures  from 2 publications
adductor operculi <input type="checkbox"/>	Day 4	IHC		Fig. 3  from Elworthy et al., 2008
caudal oblique <input type="checkbox"/>	Day 4	IHC		2 figures  from Elworthy et al., 2008

[▼ Show all 30 labeled structures](#)

Antibodies

Pages

Blog

CHILD PAGES

Pages

Ab-S58

+ Create child page

Pages

Ab-S58

Created by Web Service User, last modified on Jun 10, 2018

Limit to 255 Characters [Create New Antibody](#)

Add Comment

Antibody Name:	Ab-S58 from the ZFIN antibody database.
Other names, clone ids, catalog ids etc.	S58 , sc-32733 , S58
Does it work on zebrafish?	yes
Host organism	Mouse
Immunogen organism	
Antibody isotype	IgA
Antibody type	monoclonal
Anatomical structures recognized (use terms from the ZFIN Anatomical Ontology)	abductor hyohyoid; adaxial cell; adductor mandibulae; adductor operculi; caudal oblique; cephalic musculature; dilator operculi; hyohyoideus; inferior hyohyoid; interhyoideus; levator arcus palatini; levator operculi; medial rectus; muscle; muscle pioneer; pectoral fin musculature; slow muscle cell; somite slow muscle cell; superficial lateralis; supracarinalis; ventral intermandibularis anterior; ventral intermandibularis posterior
Recognized target molecules (gene names, domains, epitopes ...)	
Suppliers	Developmental Studies Hybridoma Bank Santa Cruz Biotechnology, Inc.

Assays Tested

Assay	Prep	Worked	Notes
immunohistochemistry		yes	from ZFIN curation
western blot		yes	from ZFIN curation

Notes

- Imported from ZFIN Antibody page Ab-S58
- No notes imported.
- Citations for Ab-S58 at ZFIN

[zfin_antibody](#)

1 Comment

Stephen H. Devoto
 S58 labels exclusively slow myofibrils in zebrafish. The antibody works most of the time in paraformaldehyde fixed tissue, and all the time in Carnoy's.
Reply • Delete • Sep 28, 2010

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.

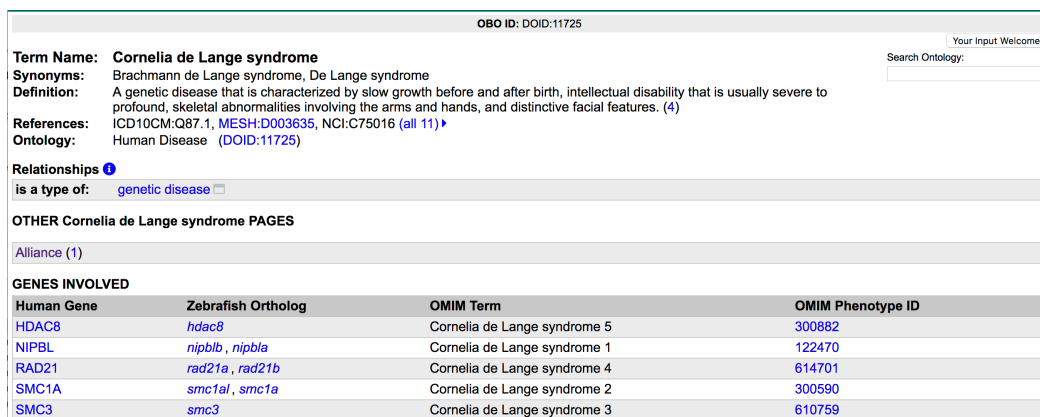
Alliance Overview:

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: DOI:11725

Your Input Welcome
Search Ontology:

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 (DOI: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	smc1a , 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


All ▾ search a gene, disease or GO term

[Home](#) [About Us](#) [Work Products](#) [News & Events](#) [Contact Us](#)

Cornelia de Lange syndrome (DOID:11725)

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.
http://en.wikipedia.org/wiki/Cornelia_de_Lange_Syndrome
<http://ghr.nlm.nih.gov/condition/cornelia-de-lange-syndrome>
<http://www.omim.org/entry/122470>
<http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/30/viewAbstract>

Synonyms Brachmann de Lange syndrome; De Lange syndrome

Cross References ICD10CM:Q87.1
 MESH:D003635
 NCI:C75016
 OMIM:122470
 OMIM:300590
 OMIM:300882
 OMIM:610759
 OMIM:614701
 ORDO:199
 SNOMEDCT_US_2016_03_01:40354009
 UMLS_CUI:C0270972

Parent Terms [genetic disease](#)

Child Terms None

Sources of Associations [RGD](#), [MGI](#), [ZFIN](#), [FB](#), [WB](#), [SGD](#)

Associations

Gene	Species	Genetic Entity	Genetic Entity Type	Association Type	Disease	Evidence Code	Source	References
SMC3	<i>Homo sapiens</i>			is implicated in	Cornelia de Lange syndrome	IAGP	RGD	RGD:7240710
Nipbl	<i>Mus musculus</i>			is implicated in	Cornelia de Lange syndrome	TAS	MGI	PMID:19763162
smc3	<i>Danio rerio</i>			is implicated in	Cornelia de Lange syndrome	TAS	ZFIN	PMID:25378554
NIPBL	<i>Homo sapiens</i>			is implicated in	Cornelia de Lange syndrome	IAGP	RGD	RGD:7240710
Nipped-B	<i>Drosophila melanogaster</i>	Nipped-B[407]	allele	is implicated in	Cornelia de Lange syndrome	IMP	FB	PMID:26544867
Pds5a	<i>Mus musculus</i>	Pds5a^{Gt(RRM243)Byg}	allele	is implicated in	Cornelia de Lange syndrome	TAS	MGI	PMID:19412548

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>

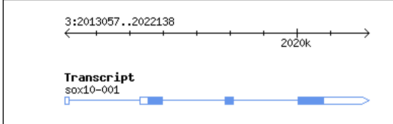
Alliance Example 2

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 \(1\)](#)

PLASMIDSNo 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.



All search a gene, disease or GO term

- [Home](#)
- [About Us](#)
- [Work Products](#)
- [News & Events](#)
- [Contact Us](#)

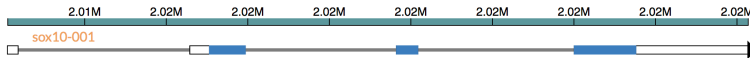
sox10

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:ENSDBG00000077467](#) [NCBI_Gene:140616](#) [UniProtKB:A0PJQ1](#) [UniProtKB:Q6DBW6](#) [UniProtKB:Q90XD1](#)
Additional Information [Literature](#)

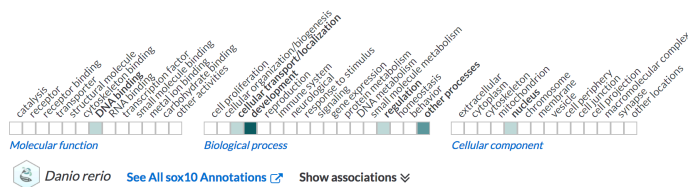
Species *Danio rerio*
Source [ZFIN:ZDB-GENE-011207-1](#)

Sequence Feature Viewer

Genome location [Chr3:2013057..2022135](#) (9.08 kb)
Assembly version GRChv10



Function – GO Annotations



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	HCNC	Higranoid	Infranoid	OMA	OrthoFinder	OrthoInspector	PANTHER	PhyloMeDB	Roundup	TreeFam	ZFIN
<i>Homo sapiens</i>	SOX10	10 of 11	Yes	Yes	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>
<i>Mus musculus</i>	Sox10	10 of 11	Yes	Yes	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>
<i>Rattus norvegicus</i>	Sox10	8 of 9	Yes	Yes	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>
<i>Drosophila melanogaster</i>	Sox100B	6 of 11	Yes	Yes	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>

[Learn about how orthologs are identified](#)

Disease Associations