



---

NCBI Website – Understanding the information on the Locus Link page.

This page provides all the information known about the gene you researched. This will include what is known about function, phenotypes, protein domains, and homologous genes for the gene you are researching. The amount of information available on this page will vary greatly and will depend on how much is known about that particular gene.

**NOTE – Some or most of this information will not be available for every gene.**

This handout will lead you through the information that can be found on this page. It is organized in order from the beginning of the page to the end. **NOTE - Because not all information is available for all genes some sections may not be present in the gene you are researching. In addition, the pages depicted in this example are a composite made from several different genes.**

Most often you will be interested in the function of a gene. To find the function of a gene try the following, in order. You can stop whenever you have the information you feel you need.

- 1 – Look at the summary found in the overview section (pg 3 of this handout)
- 2 – Look at any Pub Med references or OMIM summaries (pg 2 of this handout)
- 3 – Look at the function section for a more in depth description of the different function of this gene, or phenotypes associated with this gene (pg 4 of this handout).

If the above sources of information yield nothing,

- 4 – Look at any homologous genes in other animals, or similar proteins in humans. This can allow you to infer what the function might be (pg 5 of this handout).
- 5 – See if you can piece together the function of the gene from its protein domains (pg 6 of this handout).

## Exon Map

The first thing you will find on the top of the page is an exon map of your gene. The red line represents the entire gene. The red boxes atop the line represent the exons. Note that only a small portion of the gene is made up of exons (coding sequences).

Near the top of the page and throughout there are links provided to other very useful information. NOTE: If one or more of these symbols do not appear, that information is not available for the gene you are researching. A description of what information can be found at each link is below.

## Pub Med

**PUB** or **P** or “(All Pubs)” or “pm”

Lists scientific articles in which this protein or topic is discussed. The abstract (summary) of most papers is available by clicking on the image of a stack of papers to the left of the article.



## OMIM - Online Mendelian Inheritance in Man

**OMIM** or **O**

A description of all diseases or characteristics linked to the protein you are researching.

## AceView

**ACEVIEW**

Shows a depiction of splicing, and alternative splicing, of the gene.

## Map View

**MAP** or “mv”

Links to a chromosome/contig map showing the location of the gene

## Ensembl

e!

Diagram of the assembly of contigs for the gene, and overlaps with known mouse and rat genomes.

## HomoloGene

HOMOL

or H

Homologous genes found in other organisms are listed.

## SNPs.

VAR

or V

A list of all single nucleotide polymorphisms (SNPs) in the gene, whether they are in coding sequence, what affect they have on function.

## Overview

LocusLink Home

GATA4 Index:

Top of Page

Nomenclature

Overview

Function

Relationships

Map

RefSeq

Related Seqs

Links

LocusLink:

Click to Display mRNA-Genomic Alignments (spanning 51058 bps)

PUB OMIM ACEVIEW UNIGENE MAP VAR HOMOL e!

UCSC

*Homo sapiens* Official Gene Symbol and Name (HGNC)

**GATA4: GATA binding protein 4**

LocusID: 2626

Overview ?

**RefSeq Summary:** This gene encodes a member of the GATA family of zinc-finger transcription factors. Members of this family recognize the GATA motif which is present in the promoters of many genes. This protein is thought to regulate genes involved in embryogenesis and in myocardial differentiation and function. Mutations in this gene have been associated with cardiac septal defects.

**Locus Type:** gene with protein product, function known or inferred

**Product:** GATA binding protein 4

**Alias:** GATA-binding protein 4

**Function:** [Submit GeneRIF](#) [\(All Pubs\)](#) ?

**Phenotype:** [Atrial septal defect-2](#)

**Summary** – This section should contain a summary of the protein’s function if known.

**Locus Type** – This section lists the type of locus. The different types and a description are listed below in order of least sure to most sure.

- **Gene model** – A computer program has indicated that there could be a gene here. However, these computer programs do not always accurately detect genes.
- **Hypothetical gene** – A hypothetical gene is supported by evidence that mRNA from this gene is expressed in human cells.
- **Hypothetical protein** – A protein is known to be expressed, but its function is unknown.
- **Gene with protein product**, function known or inferred. In this case the gene has a known protein product. Further, the function of this protein has been identified, or can be inferred through homology with other organisms.

## Function

Click to Display mRNA-Genomic Alignments (spanning 66996 bps)

PUB	OMIM	ACEVIEW	UNIGENE	MAP	VAR	HOMOL	PROV
e!	UCSC						

*Homo sapiens* Official Gene Symbol and Name ([HGNC](#))

**FLT3: fms-related tyrosine kinase 3**  
**LocusID: 2322**

Overview ?

**Locus Type:** gene with protein product, function known or inferred

**Product:** fms-related tyrosine kinase 3

**Alternate Symbols:** FLK2, STK1, CD135

**Function** [Submit GeneRIF](#) [\(All Pubs\)](#) ?

**Phenotype:**

- [Leukemia, acute myeloid](#)
- [Leukemia, acute myeloid, reduced survival in](#)

**EC Number:** [2.7.1.112](#)

**GeneRIF: Gene References into Function:**

- [12239146](#) • duplicated in relapsed acute myeloid leukemia
- [12060771](#) • induces acute promyelocytic leukemia in a mouse model
- [12070009](#) • Analysis of FLT3 length mutations in 1003 patients with acute myeloid leukemia

## Phenotype

Gives links to diseases associated with mutations in the gene

## Gene References into Function

Gives links to descriptions of the different functions of the gene



## Protein Domains

This description can be useful as it lists the different domains found in that protein, along with links to what is known about these domains. By piecing together all the different domains in a protein one can sometimes make a guess as to its function.

<a href="#">LocusLink Home</a> <a href="#">HMGCR Index:</a> <a href="#">Top of Page</a> <a href="#">Nomenclature</a> <a href="#">Overview</a> <a href="#">Function</a> <a href="#">Relationships</a> <a href="#">Map</a> <a href="#">RefSeq</a> <a href="#">Related Seqs</a> <a href="#">Links</a> <a href="#">LocusLink:</a> <a href="#">Collaborators</a> <a href="#">Download</a>	<p>Map Information <a href="#">?</a></p> <p><b>Chromosome:</b> 5 <a href="#">mv</a></p> <p><b>Cytogenetic:</b> 5q13.3-q14 HUGO</p> <p><b>Markers:</b> Chr. 5 <a href="#">D5S2478</a> D5S2478 <a href="#">mv</a></p> <p>Chr. - <a href="#">GDB:177804</a></p> <p>Chr. 5 <a href="#">GDB:181537</a> <a href="#">mv</a></p> <p>NCBI Reference Sequences (RefSeq) <a href="#">?</a></p> <p><b>Category:</b> <b>REVIEWED</b></p> <p><b>mRNA:</b> <a href="#">NM_000859</a></p> <p><b>Protein:</b> <a href="#">NP_000850</a> 3-hydroxy-3-methylglutaryl-Coenzyme A reductase <a href="#">BL</a></p> <p><b>Domains:</b> <a href="#">Hydroxymethylglutaryl-coenzyme A reductase</a> score: 1573</p> <p><a href="#">Patched family. The transmembrane protein Patched is a receptor for the morphogene Sonic Hedgehog. This protein associates with the smoothed protein to transduce hedgehog signals</a> score: 114</p> <p><b>GenBank Source:</b> <a href="#">M11058,M62633</a></p>
---	--

## Related Sequences

This section gives links to similar nucleotide and protein sequences. This section can be useful when looking for proteins with similar functions. By clicking on each individual entry you will receive detailed information about that gene or protein.

<a href="#">LocusLink Home</a> <a href="#">ANC_2H01 Index:</a> <a href="#">Top of Page</a> <a href="#">Nomenclature</a> <a href="#">Overview</a> <a href="#">Map</a> <a href="#">RefSeq</a> <a href="#">Related Seqs</a> <a href="#">Links</a> <a href="#">LocusLink:</a> <a href="#">Collaborators</a> <a href="#">Download</a> <a href="#">FAQ</a> <a href="#">Help</a> <a href="#">Statistics</a> <a href="#">RefSeq:</a> <a href="#">About</a>	<p>Chr. 3 <a href="#">SHGC-77563</a> <a href="#">mv</a></p> <p>NCBI Reference Sequences (RefSeq) <a href="#">?</a></p> <p><b>Category:</b> <b>PROVISIONAL</b></p> <p><b>mRNA:</b> <a href="#">NM_016331</a></p> <p><b>Protein:</b> <a href="#">NP_057415</a> zinc finger protein ANC_2H01 <a href="#">BL</a></p> <p><b>GenBank Source:</b> <a href="#">AF003924</a></p> <p><b>Category:</b> <b>NCBI Genome Annotation</b></p> <p><b>Genomic Contig:</b> <a href="#">NT_005612</a> <a href="#">gb sv mv ev mm</a></p> <p><b>Haplotype:</b> reference</p> <p><b>Annotation for this locus:</b></p> <p><b>Evidence:</b> supported by alignment with mRNA</p> <p><b>mRNA:</b> <a href="#">NM_016331</a></p> <p><b>Protein:</b> <a href="#">NP_057415</a> <a href="#">BL</a></p> <p>Related Sequences <a href="#">?</a></p> <table border="1"> <thead> <tr> <th>Nucleotide</th> <th>Type</th> <th>Protein</th> <th></th> </tr> </thead> <tbody> <tr> <td><a href="#">AB097862</a></td> <td>m</td> <td><a href="#">BAC77610</a></td> <td><a href="#">BL</a></td> </tr> <tr> <td><a href="#">AF003924</a></td> <td>m</td> <td><a href="#">AAF21240</a></td> <td><a href="#">BL</a></td> </tr> <tr> <td><a href="#">BC020500</a></td> <td>m</td> <td><a href="#">AAH20500</a></td> <td><a href="#">BL</a></td> </tr> <tr> <td><a href="#">BC026181</a></td> <td>m</td> <td><a href="#">AAH26181</a></td> <td><a href="#">BL</a></td> </tr> </tbody> </table> <p>Additional Links <a href="#">?</a></p> <ul style="list-style-type: none"> <li>UniGene: <a href="#">Hs.22879</a></li> </ul>	Nucleotide	Type	Protein		<a href="#">AB097862</a>	m	<a href="#">BAC77610</a>	<a href="#">BL</a>	<a href="#">AF003924</a>	m	<a href="#">AAF21240</a>	<a href="#">BL</a>	<a href="#">BC020500</a>	m	<a href="#">AAH20500</a>	<a href="#">BL</a>	<a href="#">BC026181</a>	m	<a href="#">AAH26181</a>	<a href="#">BL</a>
Nucleotide	Type	Protein																			
<a href="#">AB097862</a>	m	<a href="#">BAC77610</a>	<a href="#">BL</a>																		
<a href="#">AF003924</a>	m	<a href="#">AAF21240</a>	<a href="#">BL</a>																		
<a href="#">BC020500</a>	m	<a href="#">AAH20500</a>	<a href="#">BL</a>																		
<a href="#">BC026181</a>	m	<a href="#">AAH26181</a>	<a href="#">BL</a>																		