

# Using the caBIO Home Page Freestyle Lexical Mine Tool to Find Genes Associated with a Disease

## Page Contents

- [Searching for Genes Associated with a Disease](#)
- [Search Results](#)
- [Finding the Gene Associated with the Evidence](#)
- [Disease Ontologies](#)
- [NCI Thesaurus Disease Ontologies](#)

## Documentation Table of Contents

- [Documentation Main Page](#)
- [Creation of the Cancer Gene Index](#)
- [Data, Metadata, and Annotations](#)
- [Cancer Gene Index Gene-Disease and Gene-Compound XML Documents](#)
- [caBIO APIs](#)
- [Cancer Gene Index Shared Parsed Data and Code](#)
- [caBIO Portlet Templated Searches](#)
- [caBIO Home Page](#)
- [caBIO iPhone Application](#)
- [caBIO Portlet Simple Searches](#)
- [Glossary](#)
- [Credits and Resources](#)



### Need Additional Help?

If you need additional support, please contact the [NCICB Support Group](#).




### To Print the Guide

We recommend you print one wiki page of the guide at a time. To do this, click the printer icon at the top right of the page; then from the browser File menu, choose Print. Printing multiple pages at one time is more complex. For instructions, refer to [Printing multiple pages](#).



### Having Trouble Reading the Text?

Resizing the text for any web page is easy. For information on how to do this in your web browser, refer to this [W3C tutorial](#) .

## Searching for Genes Associated with a Disease

To use the [caBIO Home Page Freestyle Lexical Mine](#) tool to find genes that are associated with a cancer-related disease, begin typing keywords into the search field. Because the tool suggests caBIO terms that match the characters you have entered, it is relatively easy to find a disease term that matches your desired disease concept.

HOME

JAVA DOCS

WELCOME TO CABIO v4.3.1

The cancer Bioinformatics Infrastructure Objects (caBIO) model and architecture is a synthesis of software, vocabulary, and metadata models for cancer research. Each of the caBIO domain objects represents an entity found in biomedical research such as Gene, Chromosome, Nucleic Acid Sequence, SNP, Library, Clone, and Pathway. Given the dynamic nature of this information, the data in caBIO is updated on a monthly basis through a series of ETL (Extract, Transform, and Load) processes.

References:

- caBIO Overview - High-level overview of caBIO
- caBIO GForge site - Contains news, information, documents, defects, feedback, and reports
- caBIO Download site - Contains download packages and release notes
- caBIO 4.3.1 Release Notes - Latest release notes for caBIO
- caBIO 4.3.1 Data Refresh Release Notes - Release notes describing the current data content of caBIO
- caBIO 4.3.1 javadocs - Java API documentation
- caBIO 4.3.1 Tech Guide - Technical Guide

caBIO API 4.3.1, Tag: CABIO\_431\_QA\_20091209, Build Date: 2010-01-20 16:18:54

Search for Biological Entities

Continue

WHAT'S NEW

- Introduce Entrez Genes that are not in Unigene
- Enhancement on the display for the search results for FreestyleLM search

Text Search

Center for Biomedical Informatics and Information Technology

FREESTYLE LEXICAL MINE

ovarian ser

ovarian serous carcinoma

ovarian serous carcinomas

ovarian serous borderline tumors

ovarian serous adenocarcinoma

ovarian serous tumors

ovarian sertoli-leydig cell tumor

ovarian serous papillary cancer (ospc)

ovarian serous cystadenocarcinoma

ovarian serous tumors ( ost )

ovarian serous neoplasms

more options ...

If you would like to search for any disease term that contains a string use the special character `"`. For example, "ovarian serous adenocarcinoma" would only return objects that have attributes containing this *exact* disease term, but `"ovarian"` will retrieve objects with attributes containing any term with the string "ovarian."

After entering a search term (1), click the Submit button (2) to retrieve results. Although you may limit your search by clicking on the `more options ...` link, this is not required (3).

Tip

If you have cannot find an appropriate search term, click the `Contact Us` link for help.

Text Search

Center for Biomedical Informatics and Information Technology

FREESTYLE LEXICAL MINE

"ovarian serous carcinoma" 1

Search 2

more options ... 3


## Search Results

The caBIO Home Page Freestyle Lexical Mine will retrieve objects with attributes that match your search term. These objects are grouped by type, which are shown as tabs at the top of the results page. To view genes that are associated with your disease term of interest, click the Evidence tab at the top of the page (1).

Each row in the Evidence results table is a truncated view of an Evidence object (that is, not all attributes and methods are shown on this page), where columns include:

- the class and identifier (Class/Id),
- evidence of the gene-disease association (Sentence),
- whether the evidence was collected from experiments involving cell lines ( Cellline Status ),
- whether the evidence is negative that is, gene X is not associated with disease or compound Y; Negation Status ),
- the PubMed identifier for the abstract from which the evidence was extracted (Pubmed Id), and
- whether the status of the sentence ( Sentence Status ).

For additional information on the attributes of the Evidence type objects, refer to the section [Data, Metadata, and Annotations](#).



All (29)
Evidence (28)
Disease Ontology (1)

gov.nih.nci.cabio.domain.Evidence

Class/Id	Sentence	Cellline Status	Negation Status	Pubmed Id	Sentence Status
<a href="#">Evidence#613299</a>	CONCLUSIONS: (1) Our data suggest that COX-2 overexpression leads to increased PGE(2), 6-keto-PGF(1 alpha) and TXB(2) biosynthesis, which may be mechanisms underlying the contribution of COX-2 to the development of ovarian serous carcinoma.	no	no	12487927	finished
<a href="#">Evidence#956032</a>	Mutations in BRAF and KRAS characterize the development of low-grade ovarian serous carcinoma.	no	no	12644542	finished
<a href="#">Evidence#724929</a>	Mutations in BRAF and KRAS characterize the development of low-grade ovarian serous carcinoma.	no	no	12644542	finished
<a href="#">Evidence#687051</a>	Expression of c-ABL, c-KIT, and platelet-derived growth factor receptor-beta in ovarian serous carcinoma and normal ovarian surface epithelium.	no	no	12910520	finished
<a href="#">Evidence#1225455</a>	AIMS: It has been suggested that WT-1 is helpful in distinguishing a primary ovarian serous carcinoma (OSC) from a primary uterine serous carcinoma (USC).	no	no	14764054	finished
<a href="#">Evidence#1225468</a>	WT-1 may also be helpful in differentiating poorly differentiated OSC from poorly differentiated ovarian endometrioid carcinoma.	no	no	14764054	finished
<a href="#">Evidence#1041696</a>	We investigated iNOS and COX-2 expression in relation to clinical outcome in 78 International Federation of Gynecology and Obstetrics (FIGO) stage III ovarian serous carcinoma with a low grade of differentiation (G3).	no	no	14984945	finished
<a href="#">Evidence#612284</a>	We investigated iNOS and COX-2 expression in relation to clinical outcome in 78 International Federation of Gynecology and Obstetrics (FIGO) stage III ovarian serous carcinoma with a low grade of differentiation (G3).	no	no	14984945	no_fact
<a href="#">Evidence#747492</a>	Expression of Cox-2, CD34, Bcl-2, and p53 and survival in patients with primary peritoneal serous carcinoma and primary ovarian serous carcinoma.	no	no	15084845	finished
<a href="#">Evidence#654024</a>	The aim of the study was to test the prognostic value of the microvessel density (MVD) within the tumor and the vascular endothelial growth factor (VEGF) expression on clinical response to chemotherapy, on brief disease-free interval, and on cause-specific survival in advanced ovarian serous carcinoma.	no	no	15361189	no_fact
<a href="#">Evidence#971645</a>	In addition, immunohistochemistry using calretinin, cytokeratin 5/6, and WT1 was performed on all cases of mesothelioma, pulmonary adenocarcinoma, ovarian serous carcinoma, and renal cell carcinoma.	no	no	15389250	finished

## Finding the Gene Associated with the Evidence

To discover which gene is associated with each piece of evidence, click on the Class/Id link for the desired object. This will open the full Evidence type object. Scroll over to the right, and click on `getGeneFunctionAssociationCollection` method link (1) to view the Gene Disease Association type object. This object has a role attribute that contains one or more [Role Codes](#) or [Role Details](#) that describe the nature of the gene-disease relationship, as well as a notation that the Cancer Gene Index is the source of these data.



### Tip

If you do not want to spend time navigating through the caBIO object model for candidate gene-disease associations that were found to be false positives to other data by [expert human curators](#), select only Evidence objects where the Sentence Status is finished and the Negation Status is no.



### Note

A single piece of evidence may have multiple Role Codes and Role Details describing the gene-disease association, and the evidence may also describe gene-compound associations. Thus, after clicking the `getGeneFunctionAssociationCollection` link, you may see multiple retrieved objects of type `gov.nih.nci.cabio.domain.GeneDiseaseAssociation` and even multiple object retrieved records of type `gov.nih.nci.cabio.domain.GeneAgentAssociation`.

Click on the `getGene` link (2) to access the related Gene object (bottom panel). This Gene object contains the full name and HUGO Gene Symbol in the `fullName` and `hugoSymbol` columns (3), for example, for the gene associated with the disease of interest and a specific piece of evidence.

To explore additional genes associated with the disease term of interest, navigate back to the evidence page and repeat this process.

Criteria: gov.nih.nci.cabio.domain.Evidence[@id=932369]							
gov.nih.nci.cabio.domain.Evidence							
bigid	cellineStatus	comments	id	sentenceStatus	evidenceCodeCollection	geneFunctionAssociationCollection	interactionCollection
-	no	-	932369-VV-42	no_fact	<a href="#">getEvidenceCodeCollection</a>	<a href="#">getGeneFunctionAssociationCollection</a>	<a href="#">getInteractionCollection</a>

1

Criteria: Evidence[@id=932369]							
gov.nih.nci.cabio.domain.GeneDiseaseAssociation							
bigid	id	role	source	gene	evidenceCollection	diseaseOntology	
-	19658304	Not assigned	Cancer Gene Index	<a href="#">getGene</a>	<a href="#">getEvidenceCollection</a>	<a href="#">getDiseaseOntology</a>	

2

Criteria: GeneDiseaseAssociation[@id=19658304]							
gov.nih.nci.cabio.domain.Gene							
bigid	clusterId	fullName	hugoSymbol	id	symbol	geneFunctionAssociationCollection	
hdt://2500.1.PMEUGUCCL5/77FY7S8DI	193717	Interleukin 10	IL10	9158	IL10	<a href="#">getGeneFunctionAssociationCollection</a>	

3



#### Be Careful

If you find yourself in a part of the object model that you do not understand or if you get confused, stop and navigate your web browser back to the search results page with the Evidence tab.

For your reference, the subset of caBIO classes that are related to the Cancer Gene Index are [shown](#). The full model is available on the [NCI SVN](#), but you must have the [Enterprise Architect](#) modeling tool to view this file.

## Disease Ontologies

As you become comfortable navigating the caBIO object model, you may wish to start using additional method links to discover the pathway/s in which a particular Cancer Gene Index gene is involved, clinical protocol information for a disease or compound, and other related data. It is also possible to view disease ontologies for a given gene-disease concept pair by navigating through the object model to records for objects in the class `Disease Ontology`, `gov.nih.nci.cabio.domain.DiseaseOntology`. This can be helpful if you would like to search for genes that are associated with parent, sister, or child concepts to your disease search term.

You can reach these records from either the Disease Ontology tab of the main search result page or by clicking through the model starting from retrieved evidence type objects. To view related disease concepts from the Disease Ontology tab, click on the tab and select the any hyperlink in the `Class/Id` column. This will reveal a `Disease Ontology` type object. If you would like to view parent or child disease concepts linked with an evidence object, select the `getGeneFunctionAssociationCollection` link on the Evidence object page (1) and then click the `getDiseaseOntology` link in the Gene DiseaseAssociation object record (2). Once you have pulled up a `Disease Ontology` record, you can find parent disease concepts by scrolling to the right and selecting the `getParentDiseaseOntologyRelationshipCollection` link; child disease concepts can be accessed by clicking on the `getChildDiseaseOntologyRelationshipCollection` link.



#### Note

If the disease concept of interest has neither parent nor child concepts, you must search the [NCI Thesaurus](#) using your disease term or EVS ID listed in the EVS ID column of the Disease Ontology object or use the [EVS API](#).


## NCI Thesaurus Disease Ontologies

To view disease ontologies in the NCI Thesaurus, open a new browser tab or window and navigate to the [NCI Thesaurus web page](#), enter in your disease term (2, "ovarian serous adenocarcinoma") or NCI Thesaurus concept code (for example, "C7550", and click the Search button (3). If required, select your exact search term from the list to view the NCI Term page



#### Identifier Tip

The EVS Identifier for a term is also its NCI Thesaurus Concept Code.



1

ovarian serous2

Search?

☐ Exact Match☐ Begins With☒ Contains3

[Home](#) | [View Hierarchy](#) | [Subsets](#) | [Help](#) | [Term Suggestion](#)

Quick Links

## Welcome

Version: July 27, 2009 (09.09c)

NCI Thesaurus (NCIt) provides reference terminology for many NCI and other systems. It covers vocabulary for clinical care, translational and basic research, and public information and administrative activities.





**NCIt features:**

- Stable, unique codes for biomedical concepts;
- Preferred terms, synonyms, definitions, research codes, external source codes, and other information;
- Links to [NCI Metathesaurus](#) and other information sources;
- Over 200,000 cross-links between concepts, providing formal logic-based definition of many concepts;
- Extensive content integrated from NCI and other partners, much available as separate NCIt [subsets](#)
- Updated frequently by a team of subject matter experts.

NCIt is a widely recognized standard for biomedical coding and reference, used by a broad variety of public and private partners both nationally and internationally.

4

[NCI Home](#) | [Contact Us](#) | [Policies](#) | [Accessibility](#) | [FOIA](#)



[NCI Enterprise Vocabulary Services:](#) Terminology resources and services for NCI and the biomedical community.

[NCI Metathesaurus:](#) Comprehensive database of 4,600,000 terms from 70 terminologies.

[NCI BioPortal:](#) View NCI and other terminologies in an integrated ontology environment.

[NCI Terminology Resources:](#) More information on NCI dictionaries and resources.

You may view parent and child terms for any disease term by clicking on the Relationships tab (blue box). For example, "ovarian serous adenocarcinoma" has the children "ovarian serous cystadenocarcinoma" and "ovarian serous papillary adenocarcinoma" and the parent terms "malignant ovarian serous tumor," "ovarian adenocarcinoma," and "serous adenocarcinoma." Alternatively, if you would like to view where your term fits in the entire disease hierarchy, click the red View in Hierarchy button (green box).

## Ovarian Serous Adenocarcinoma (Code C7550)

[Suggest changes to this concept](#)

Terms & Properties

Relationships

Synonym Details

View All

View in Hierarchy

View History

### Terms and Properties

**Definition:** Ovarian serous adenocarcinoma is a serous neoplasm characterized by nuclear atypia, high mitotic activity, stratification, glandular complexity, branching papillary fronds and stromal invasion. --2002

**Preferred Name:** Ovarian Serous Adenocarcinoma

**NCI Thesaurus Code:** C7550

**NCI Metathesaurus CUI:** CL028288 ([see NCI Metathesaurus info](#))

**Synonyms & Abbreviations:** ([see Synonym Details](#))

Ovarian Serous Adenocarcinoma

Ovarian Serous Carcinoma

Serous Adenocarcinoma of Ovary

Serous Adenocarcinoma of the Ovary

Serous Carcinoma of Ovary

Serous Carcinoma of the Ovary

**External Source Codes:**

NCI META CUI

CL028288 ([see NCI Metathesaurus info](#))

**Other Properties:**

Semantic\_Type

Neoplastic Process

**Additional Concept Data:**

**URL to Bookmark:** <http://ncit.nci.nih.gov/ncitbrowser/ConceptReport.jsp?dictionary=NCI%20Thesaurus&code=C7550>