David Landsman, Ph.D.

Chief, Computational Biology Branch

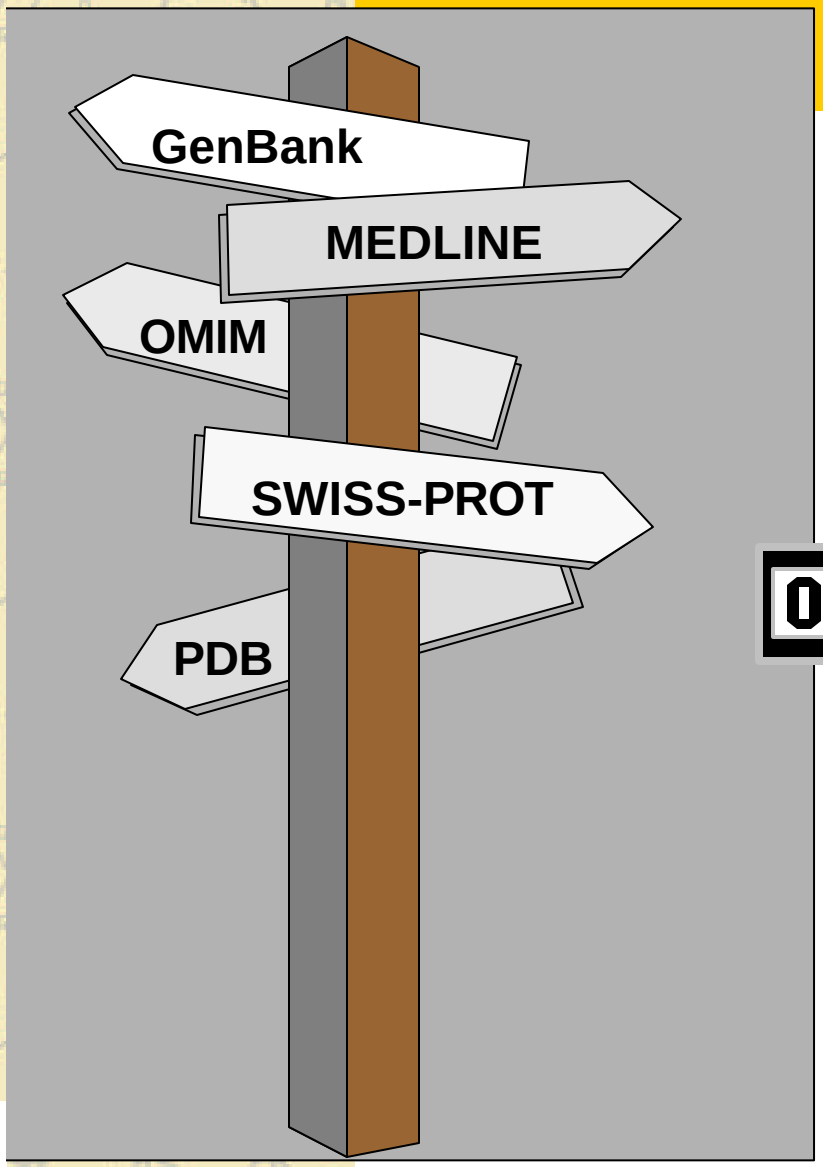
National Center for Biotechnology Information

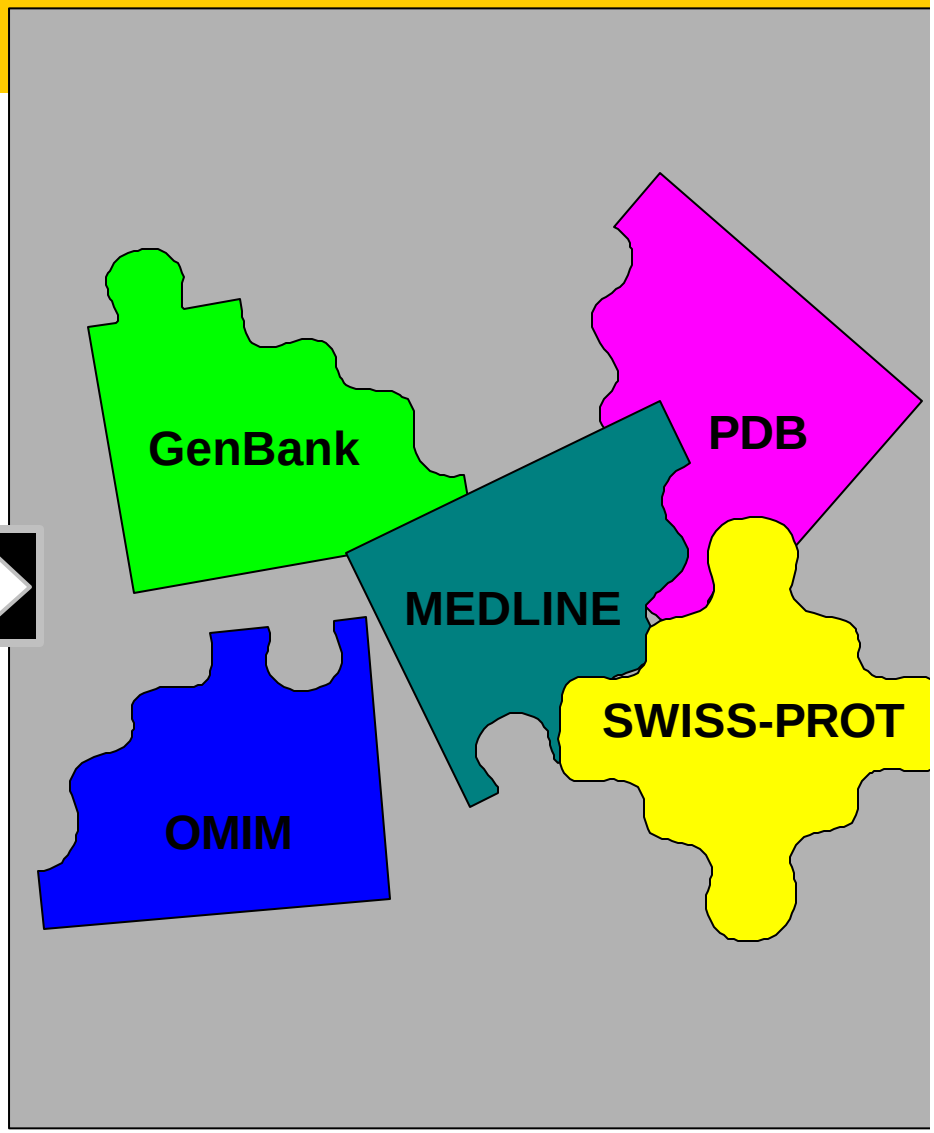
NCBI, NLM, NIH

Links between a Multitude of Data Types at the NCBI

Towards a Comprehensive Genome Information View

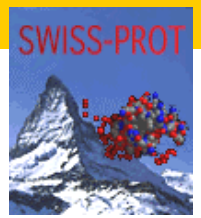
Information puzzles end abruptly?



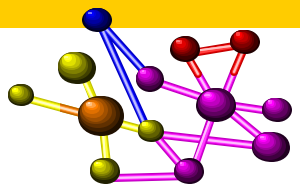


Vertical querying

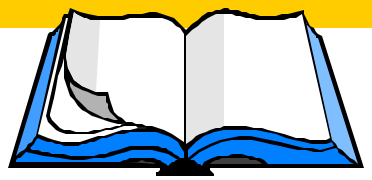
GenBank



SWISS-PROT



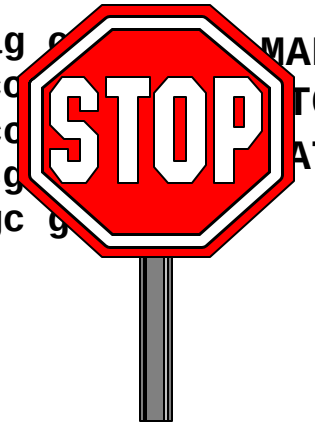
PDB



MEDLINE

query

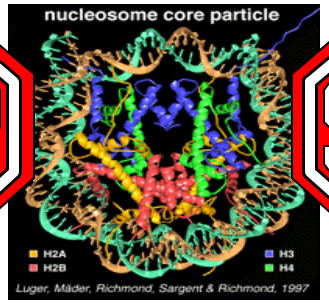
```
tg gcc cga acc aag c
ct gct cgt aag tcc
gt ggg aaa gcc ccc
aa cag ctg gcc acg
cc gcc agg aaa agc g
```



query

MARTKQTARK
TGGKAPRKQ
ATKAARKSA

query



query

Nature 1997 Sep 18;389(6648):251-260

Crystal structure of the nucleosome core particle at 2.8 A resolution.

Luger K, Mader AW, Richmond RK, Sargent DF, Richmond TJ

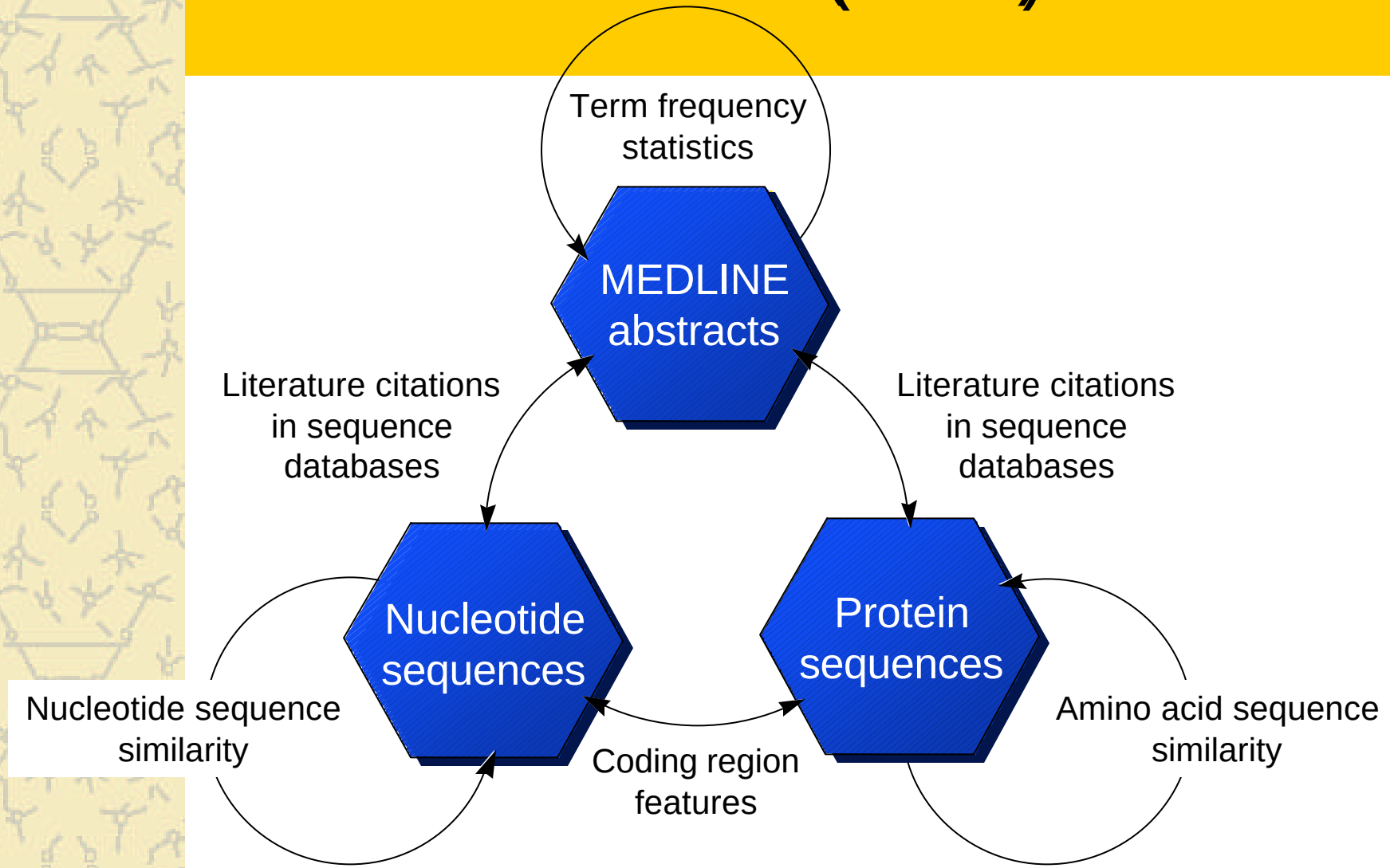
Entrez and its databases

horizontal or lateral querying

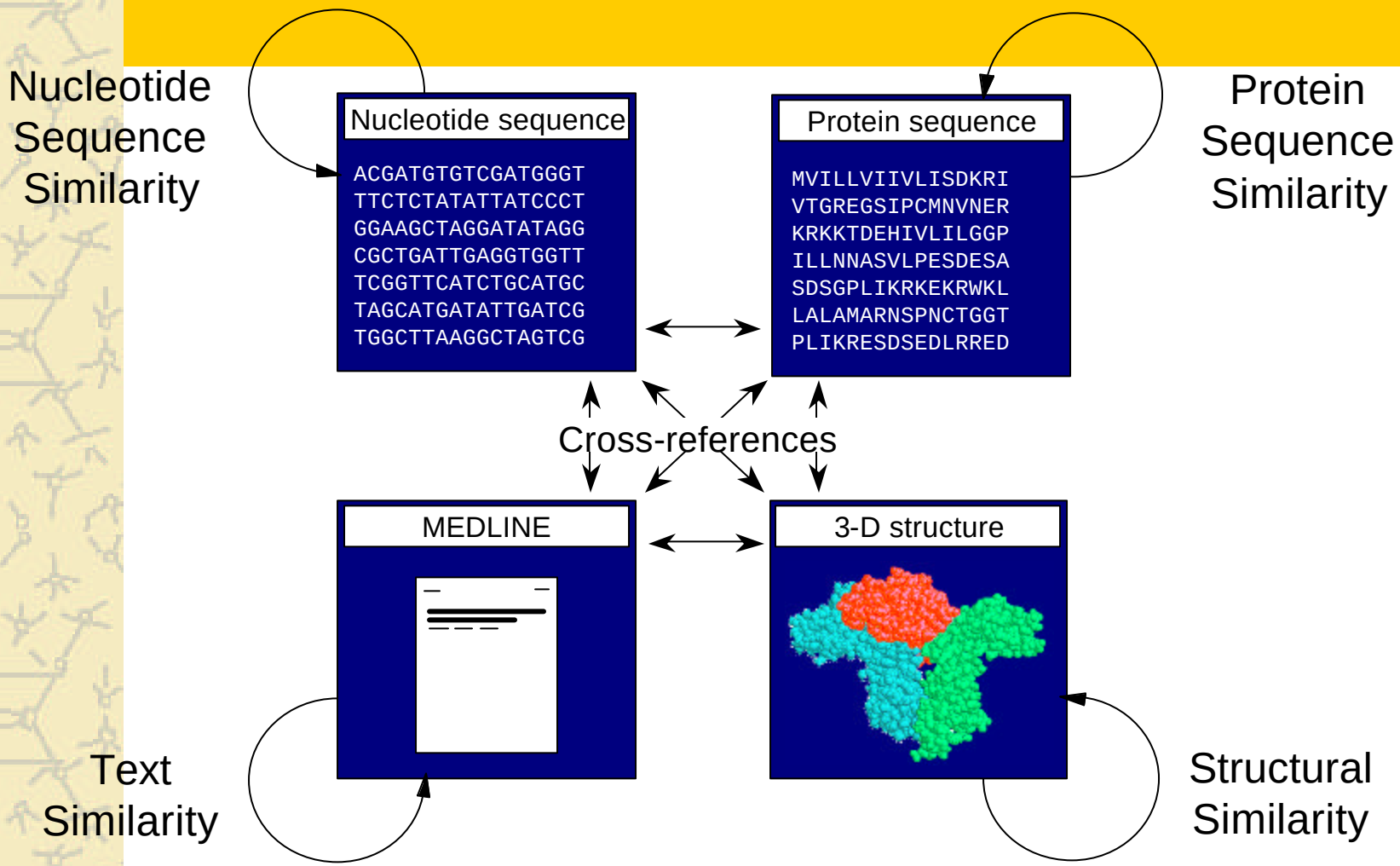
vertical querying

horizontal or lateral querying

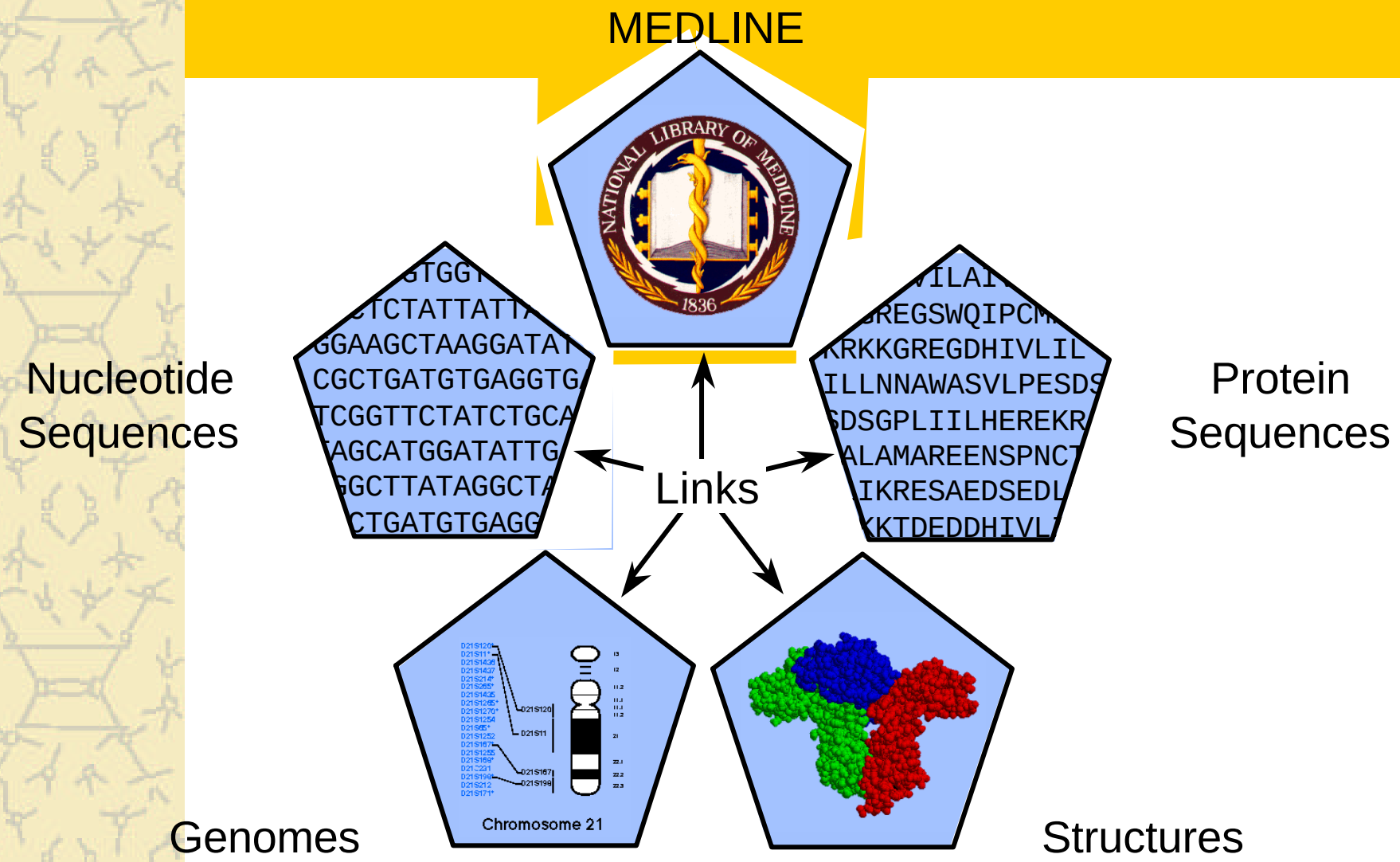
Entrez (1992)


Term frequency statistics
MEDLINE abstracts
Literature citations in sequence databases
Literature citations in sequence databases
Nucleotide sequences
Protein sequences
Nucleotide sequence similarity
Coding region features
Amino acid sequence similarity

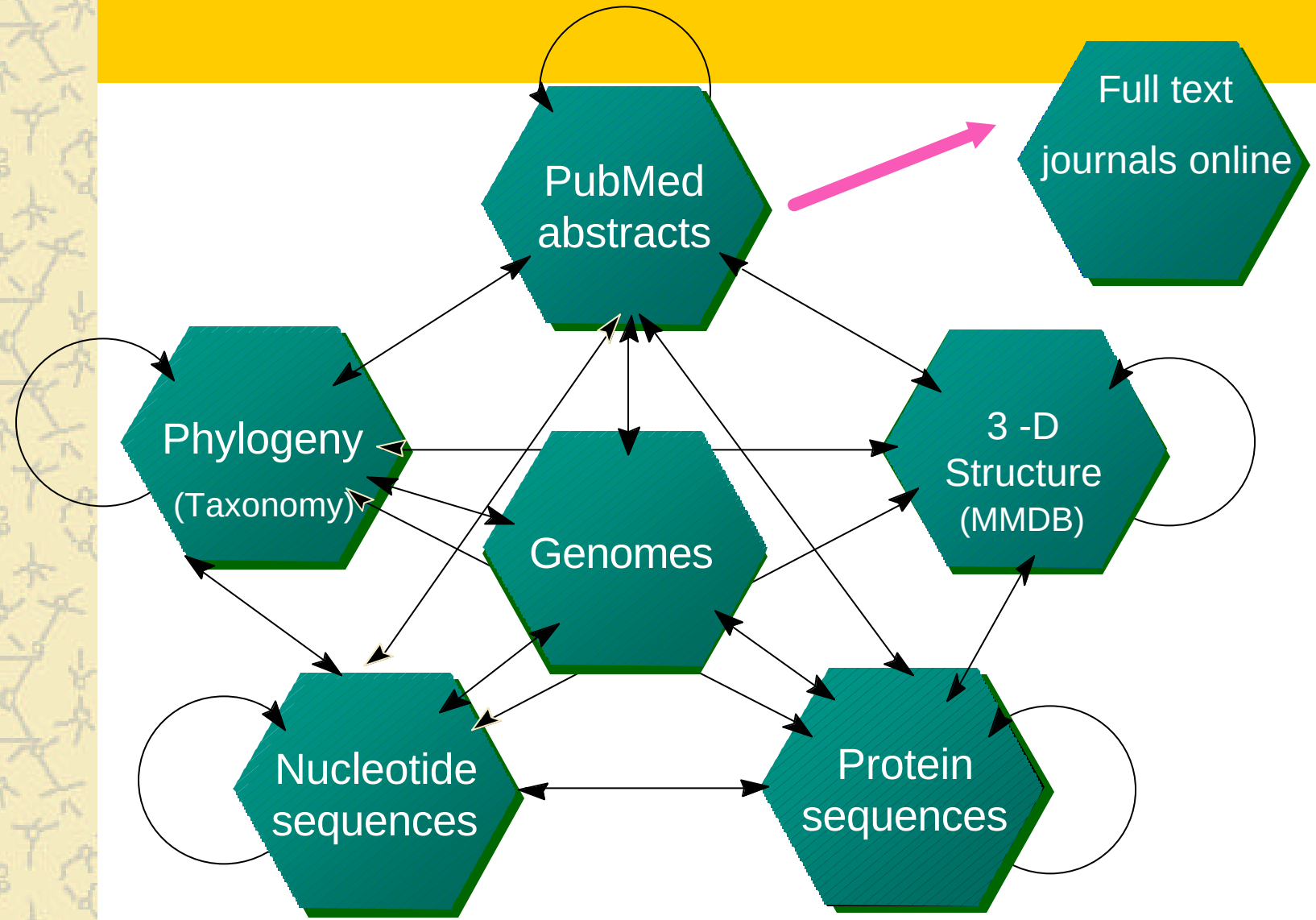
Entrez (1994)


Nucleotide Sequence Similarity
Nucleotide sequence
ACGATGTGTCGATGGGT
TTCTCTATATTATCCCT
GGAAGCTAGGATATAGG
CGCTGATTGAGGTGGTT
TCGGTTCATCTGCATGC
TAGCATGATATTGATCG
TGGCTTAAGGCTAGTCG
Protein Sequence Similarity
Protein sequence
MVILLVIIVLISDKRI
VTGREGSIPCMNVNER
KRKKTDEHIVLILGGP
ILLNNASVLPESDESA
SDSGPLIKRKEKRWKL
LALAMARNSPNCTGGT
PLIKRESDSEDLRRED
Cross-references
MEDLINE
3-D structure
Text Similarity
Structural Similarity

Entrez: Genotype to Phenotype (1996)



Entrez Increases Discovery Space 1998



Objectives

- start with a syndrome search in OMIM
- identify the gene for the syndrome
- locate the gene on the genome
  - other genes nearby
- information about the gene in LocusLink
- links to sequence and structure information
- view structurally similar proteins

OMIM - Online Mendelian Inheritance in Man - Netscape

File Edit View Go Communicator Help

Bookmarks Location: http://www.ncbi.nlm.nih.gov:80/entrez/query.fcgi?db=OMIM What's Related

NCBI

OMIM
Online Mendelian Inheritance in Man

Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM

Search OMIM for Autoimmune lymphoproliferative syndrome (ALPS)

Limits Preview/Index History Clipboard

Entrez

OMIM
Search OMIM
Search Gene Map
Search Morbid Map

Help
OMIM Help
How to Link

FAQ
Numbering System
Symbols
How to Print
Citing OMIM
Download

OMIM Facts
Statistics
Update Log
Restrictions on Use

Allied Resources
Genetic Alliance
Databases
HGMD
Locus-Specific
Model Organisms
MitoMap
Phenotype
Davis Human/Mouse

- Enter one or more search terms.
- Use **Limits** to restrict your search by search field, chromosome, and other criteria.
- Use **Index** to browse terms found in OMIM records.
- Use **History** to retrieve records from previous searches, or to combine searches.

OMIM™ - Online Mendelian Inheritance in Man™

NEW OMIM is now incorporated into NCBI's Entrez system and can be queried using the same approach as the other Entrez databases such as PubMed and GenBank. The previous OMIM pages are still available here.

Welcome to OMIM, Online Mendelian Inheritance in Man. This database is a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues at Johns Hopkins and elsewhere, and developed for the World Wide Web by NCBI, the National Center for Biotechnology Information. The database contains textual information and references. It also contains copious links to MEDLINE and sequence records in the Entrez system, and links to additional related resources at NCBI and elsewhere.

You can do a search by entering one or more terms in the text box above. Advanced search options are accessible via the Limits, Preview/Index, History, and Clipboard options in the grey bar beneath the text box. The OMIM help document provides additional information and examples of basic and advanced searches.

The links to the left provide further technical information, searching options, frequently asked questions (FAQ), and information on allied resources. To return to this page, click on the OMIM link in the black

Document: Done

OMIM - AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME; ALPS - Netscape

File Edit View Go Communicator Help

Bookmarks Location: http://www.ncbi.nlm.nih.gov:80/entrez/dispomim.cgi?id=601859 What's Related

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OMIM
Online Mendelian Inheritance in Man

Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM

Search OMIM for Go Clear

Limits Preview/Index History Clipboard

MIM #601859
Text
Animal Model
References
Contributors
Creation Date
Edit History

Display Detailed Save Text Add to Clipboard

#601859 Related Entries, PubMed, Genome

AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME; ALPS

Alternative titles; symbols

CANALE-SMITH SYNDROME
AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE IA, INCLUDED; ALPS1A, INCLUDED
AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE IB, INCLUDED; ALPS1B, INCLUDED

TEXT

A number sign (#) is used with this entry because autoimmune lymphoproliferative syndrome (ALPS) is associated with mutations in the FAS gene (TNFRSF6, or CD95; 134637) that result in defective apoptosis. The disorder can also result from mutations in the FAS ligand (FASL) gene (TNFSF6, or CD95L; 134638). These 2 forms of the disease have been referred to as ALPS1A and ALPS1B, respectively. Type II ALPS (603909) is caused by mutation in the caspase-10 gene (CASP10; 601762).

Canale and Smith (1967) described a childhood syndrome of autoimmunity (hemolytic anemia and thrombocytopenia with massive lymphadenopathy and splenomegaly). These clinical features are common to most reported cases of autoimmune lymphoproliferative syndrome (Fisher et al., 1995; Rieux-Laucat et al., 1995). Vaishnaw et al. (1997) urged the use of the eponym 'Canale-Smith syndrome'

Document: Done

OMIM - AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE II - Netscape

File Edit View Go Communicator Help

Bookmarks Location: http://www.ncbi.nlm.nih.gov:80/entrez/dispomim.cgi?id=603909 What's Related

NCBI

OMIM
Online Mendelian Inheritance in Man

Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM

Search OMIM for Go Clear

Limits Preview/Index History Clipboard

MIM #603909
Text
References
Contributors
Creation Date
Edit History

Display Detailed Save Text Add to Clipboard

#603909 Related Entries, PubMed

AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE II

Alternative titles; symbols

ALPS2

TEXT

A number sign (#) is used with this entry because of evidence that type II autoimmune lymphoproliferative syndrome (ALPS) results from mutation in the gene encoding caspase-10 (CASP10; 601762).

Programmed cell death (apoptosis) of activated lymphocytes is critical to immune homeostasis. The cell surface protein FAS (TNFRSF6, or CD95; 134637) and its ligand, FASL (TNFSF6, or CD95L; 134638), play a pivotal role in regulating lymphocyte apoptosis, and defective expression of either FAS or FASL results in marked over-accumulation of mature lymphocytes and autoimmune disease in mice. Defective lymphocyte apoptosis caused by mutations in the FAS or FASL genes can result in a severe autoimmune lymphoproliferative syndrome in humans (see 601859). To define the clinical, genetic, and immunologic spectrum of ALPS, Sneller et al. (1997) studied 9 patients and their families. Individual patients were followed for 3 months to 6 years. ALPS was identified in 9 unrelated children (1 per family) as manifested by moderate to massive splenomegaly and lymphadenopathy, hypergammaglobulinemia, autoimmunity, B-cell lymphocytosis, and the expansion of an unusual population of CD4/CD8-deficient T cells that express the alpha/beta T-cell receptor (see 186880). Hemolytic anemia was the most frequent form of autoimmune disease, occurring in 6 patients with or without idiopathic thrombocytopenic purpura. All

Document: Done

OMIM - CASPASE 10, APOPTOSIS-RELATED CYSTEINE PROTEASE; CASP10 - Netscape

File Edit View Go Communicator Help

Bookmarks Location: http://www.ncbi.nlm.nih.gov:80/entrez/dispomim.cgi?id=601762 What's Related

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Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM

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Limits Preview/Index History Clipboard

MIM *601762
Text
Allelic Variants
View List
References
Contributors
Creation Date
Edit History

Gene map

LocusLink
PubMed
HUGO
RefSeq
GenBank
UniGene

LinkOut
HGMD

Display Detailed Save Text Add to Clipboard

***601762** Related Entries, PubMed, Protein, Nucleotide, LinkOut

CASPASE 10, APOPTOSIS-RELATED CYSTEINE PROTEASE; CASP10

Alternative titles; symbols

MCH4
CASPASE 10, ISOFORM B, INCLUDED; CASP10B, INCLUDED
FADD-LIKE ICE 2, INCLUDED; FLICE2, INCLUDED

Gene map locus 2q33-q34

TEXT

A cascade of protease reactions is believed to be responsible for the apoptotic changes observed in mammalian cells undergoing programmed cell death. This cascade involves many members of the aspartate-specific cysteine proteases of the ICE/CED-3 (see 147678) family.

Fernandes-Alnemri et al. (1996) used degenerate PCR to identify a new member of the ICE/CED-3 protease family. They subsequently cloned this gene, termed MCH4, from a human Jurkat T-cell cDNA library. Sequence analysis revealed that MCH4 encodes a 479-amino acid polypeptide. They found that MCH4 is most closely related to MCH5 (601763) and that MCH4 and MCH5 contain the active site pentapeptide QACQG instead of the QACRG present in all other known members of the family. Furthermore, the authors found that the sequences of MCH4 and MCH5 contain Fas-associating protein with death domain (FADD; 602457) like domains, suggesting possible interaction with FADD

Document: Done

MIM Gene map - Netscape

File Edit View Go Communicator Help

Bookmarks Location: http://www.ncbi.nlm.nih.gov/htbin-post/Omim/getmap?l601762 What's Related

NCBI **OMIM** *Online Mendelian Inheritance in Man* Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM

The OMIM Gene map presents the cytogenetic map location of disease genes and other expressed genes described in OMIM. See the OMIM Morbid Map for a list of disease genes organized by disease. For more refined maps of genes and DNA segments, use NCBI Entrez Map Viewer.

Search for: Find Find Next (from the current location)

- Enter gene symbol, chromosomal location, or disorder keyword to search for, e.g. "CYP1", "5", "1pter", "Xq", or "alzheimer".
- You must capitalize X and Y to search for those chromosomes.

2q33-q34, CASP10 to 2q33-q37, SCYA20

<<Move Up Move Down>>

Location	Symbol(s)	St	Title	OMIM	Method	Links
2q33-q34	CASP10, MCH4	P	Caspase 10, apoptosis-related cysteine protease	601762	Psh, REc, A	Map Viewer
2q33-q34	CD28	C	CD28 antigen (Tp44)	186760	A, REn	Map Viewer Mouse 1(Cd28)
2q33-q34	CHRND, ACHRD	C	Cholinergic receptor, nicotinic, delta polypeptide	100720	H, REa, A, LD, RE	Map Viewer Mouse 1(Acrd)
	Comments: linked to ldh-1 in mouse					
2q33-q34	CHRNG, ACHRG	C	Cholinergic receptor, nicotinic, gamma polypeptide	100730	H, REa, LD, RE	Map Viewer Mouse 1(Acrg)
	Disorder: Myasthenia gravis, neonatal transient (2) **Comments:** tightly linked to CHRND by RE					
2q33-q34	FZD5	L	Polarity gene 'frizzled,' Drosophila, homolog of	601723	H	Map Viewer Mouse 1(Mfz5)
2q33-q34	IGFBP2	C	Insulin-like growth factor-binding protein-2, 36kD	146731	REa, A	Map Viewer Mouse 1(Igfbp2)

Entrez Map View - Netscape

File Edit View Go Communicator Help

Bookmarks Location: v/cgi-bin/Entrez/maps.cgi?ORG=hum&CHR=2&maps=loc,morbid,gene&query=CASP10&VERBOSE=ON&ZOOM=10 What's Related

NCBI Entrez Genome

PubMed Entrez BLAST OMIM Taxonomy Structure

Search for On chromosome(s) Find

Map Viewer Help
Human Maps Help
FTP
Chr. 2 Resource

Region Shown:
2q32.3
2q35 Go

out zoom in

Ideogram

Homo sapiens Map View

Chromosome:
1 [2] 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

Query: CASP10 [clear]

Master: Genes On Cytogenetic Map Display settings

Total Genes On Chromosome: **763**
Region Displayed: **2q32.3-2q35**
Genes Labeled: **20** Total Genes in Region: **369**

Genes_seq Morbid Genes_cyto

symbol	location	
COL5A2	2q14-q32	collagen, type V, alpha 2
EVX2	2q31-q32	even-skipped homeo box 2 (homolog of I
FPN1	2q32	ferroportin 1; iron regulated gene 1
KLF7	2q32	Kruppel-like factor 7 (ubiquitous)
HOXD11	2q31-q37	homeo box D11
HOXD9	2q31-q37	homeo box D9
RPE	2q32-q33.3	ribulose-5-phosphate-3-epimerase
AOX1	2q33	aldehyde oxidase 1
FZD7	2q33	frizzled (Drosophila) homolog 7
DAZAP2	2q33-q34	DAZ associated protein 2
CASP10	2q33-q34	caspase 10, apoptosis-related cysteine p
ERBB4	2q33.3-q34	v-erb-a avian erythroblastic leukemia vira

Entrez Map View - Netscape

File Edit View Go Communicator Help

Bookmarks Location: c[176M%3A186M],morbid[11369.50%3A12809.50],gene[11369.50%3A12809.50]&GOTO=11923.90ISCN&zoomin=8 What's Related

Map Viewer Help
Human Maps Help
FTP
Chr. 2 Resource

Region Shown:
2q33.1
2q33.2 Go

out zoom in

Ideogram

Query: CASP10 [clear]

Master: Genes On Cytogenetic Map **Display settings**

Total Genes On Chromosome: **763**
Region Displayed: **2q33.1-2q33.2**
Genes Labeled: **20** Total Genes in Region: **310**

Genes_seq Morbid Genes_cyto

GCL GCG FAP DPP4 BAZ2B PKP4 TANK TBR1 POH1 DKFZP434D156 CLK1 NAB1 CASP10 CFLAR CD28 KLF7 RPE IGFBP5 IGFBP2 TNP1

ITGA6 GNT1 LSFC NIDDM1 PMS1 IDDM12 ALS2 ACHRG NDUFS1 PDC CAE1 CRYG4 LI2 CCP FLNMS CTX

symbol	location	
RP26	2q31-q33	retinitis pigmentosa 26 (autosomal reces
HOXD4	2q31-q37	homeo box D4
HOXD3	2q31-q37	homeo box D3
HOXD1	2q31-q37	homeo box D1
FTHL3	2q32-q33	ferritin, heavy polypeptide-like 3
GLS	2q32-q34	glutaminase
NAB1	2q32.3-q33	NGFI-A binding protein 1 (ERG1 binding
CD28	2q33	CD28 antigen (Tp44)
ORC2L	2q33	origin recognition complex, subunit 2 (yea
PTHR2	2q33	parathyroid hormone receptor 2
ADAM23	2q33	a disintegrin and metalloproteinase dom
MYL1	2q33-q34	myosin, light polypeptide 1, alkali; skelet
CASP10	2q33-q34	caspase 10, apoptosis-related cysteine p
FZD5	2q33-q34	frizzled (Drosophila) homolog 5
CRYGD	2q33-q35	crystallin, gamma D
PNKD	2q33-q35	paroxysmal nonkinesiogenic dyskinesia
PI7	2q33-q35	protease inhibitor 7 (protease nexin I)
PCC	2q33-q35	Cataract, polymorphic congenital, autosc
CRYGC	2q33-q35	crystallin, gamma C
CYP27A1	2q33-qter	cytochrome P450, subfamily XXVIIA (ste

Document: Done

LocusLink Report - Netscape

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Bookmarks Location: http://www.ncbi.nlm.nih.gov/LocusLink/LocRpt.cgi?l=843 What's Related

NCBI LocusLink

PubMed Entrez BLAST OMIM Taxonomy Structure

Search LocusLink Display Brief Organism: All

Query: Go Clear

LocusLink Home
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NCBI Genome Guides
- Fruit Fly
- Human
- Mouse
- Rat
- Zebrafish

RefSeq
- About
- Download
- FAQ
- Statistics

Related Resources
- BDGP
- CGAP
- FlyBase
- GDB

A B C D E F G H I J K L M N O P Q R S T U V W X Y Z

PUB UG GDB

Homo sapiens Official Gene Symbol and Name (HGNC)

CASP10: caspase 10, apoptosis-related cysteine protease

Locus Information — Submit GeneRIF for CASP10

LocusID:	843
Type:	gene with protein product, function known or inferred
Alternate Symbols:	MCH4 FLICE2
Product:	caspase 10, apoptosis-related cysteine protease
UniGene:	Hs.5353
OMIM:	601762

Map Information

NEW Entrez Map Viewer: View Genomic Context

Chromosome:	2		
Cytogenetic:	2q33-q34	RefSeq	
Markers(ePCR):	Chr. 2	U60519	U60519
	Chr. 2	H80712	H80712
	Chr. 2	STSG40075	

Document: Done

LocusLink Report - Netscape

File Edit View Go Communicator Help

Bookmarks Location: http://www.ncbi.nlm.nih.gov/LocusLink/LocRpt.cgi?l=843 What's Related

Resources
- BDGP
- CGAP
- FlyBase
- GDB
- GeneMap'99
- HomoloGene
- Map Viewer
- --fruit fly
- --human
- MGD
- Nomenclature
- OMIM
- RATMAP
- RGD
- UniGene
- ZFIN

Cytogenetic:	2q33-q34	RefSeq	
Markers(ePCR):	Chr. 2	U60519	U60519
	Chr. 2	H80712	H80712
	Chr. 2	STSG40075	

Key: Link to Map Viewer

Homo sapiens CASP10 Reference Sequence (RefSeq)

Status: PROVISIONAL

Nucleotide: NM_001230

Protein: NP_001221 caspase 10, apoptosis-related cysteine protease

GenBank Source: U60519

GenBank Sequences

Nucleotide	Type	Protein
U60519	m	AAC50644
U86214	m	AAB46789

Key: Related Structures

Additional Web Resources

GeneCard for CASP10

To Top

Questions or Comments?

Write to the NCBI Service Desk

Disclaimer Privacy statement

Document: Done

NCBI Sequence Viewer - Netscape

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NCBI

Entrez Protein

PubMed | Nucleotide | Protein | Genome | Structure | Popset

Search Protein for Go Clear

Limits Index History Clipboard

Display Default View as HTML Save Add to Clipboard Hide Brief and LinkBar

1: GI = "1498324" [GenPept] apoptotic cysteine protease... BLink, PubMed, Related Sequences, Nucleotide, Taxonomy, OMIM, Link

```
LOCUS       AAC50644      479 aa                    PRI       21-AUG-1996
DEFINITION  apoptotic cysteine protease proMch4.
ACCESSION   AAC50644
PID         g1498324
VERSION     AAC50644.1  GI:1498324
DBSOURCE    locus HSU60519 accession U60519.1
KEYWORDS    .
SOURCE      human.
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 479)
  AUTHORS   Fernandes-Alnemri,T.F., Armstrong,R.C., Krebs,J., Srinivasula,S.M.,
            Wang,L., Bullrich,F., Fritz,L.C., Trapani,J.A., Tomaselli,K.J.,
            Litwack,G. and Alnemri,E.S.
  TITLE     In vitro activation of CPP32 and Mch3 by Mch4, a novel human
            apoptotic cysteine protease containing two FADD-like domains
  JOURNAL   Proc. Natl. Acad. Sci. U.S.A. 93 (15), 7464-7469 (1996)
  MEDLINE   96353838
REFERENCE   2  (residues 1 to 479)
  AUTHORS   Alnemri,E.S.
  TITLE     Direct Submission
  JOURNAL   Submitted (11-JUN-1996) Pharmacology, Thomas Jefferson University,
            Jefferson Cancer Institute, 233, S. Tenth Street, Philadelphia, PA
            19107, USA
COMMENT     Method: conceptual translation.
FEATURES             Location/Qualifiers
     source          1..479
```

Document: Done

gi|1498324 view - Netscape

File Edit View Go Communicator Help

PubMed Nucleotide Protein Genome Structure Taxonomy Help

Query: gi|1498324 apoptotic cysteine protease proMch4
Lineage: Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo

Best hits | Common Tree | Taxonomy Report | 3D structures | CDD-Search | GI list

166 BLAST hits to 23 unique species Sort by taxonomy proximity

0 Archaea | 0 Bacteria | 159 Metazoa | 0 Fungi | 0 Plants | 0 Viruses | 0 Other Eukaryotae

Keep only | Cut-Off 100 | Select | Reset

479 aa

SCORE	P	GI	ORGANISM
2417	27	4731239	caspase-10/d [Homo sapiens]
2161	27	1835779	Fas-associated death domain protein interleukin-1b-converting en
1155	27	4731237	caspase-10/c [Homo sapiens]
907	18	7619910	caspase-10 [Xenopus laevis]
768	27	2440071	MACH-alpha-1 [Homo sapiens]
766	27	1457959	FADD-homologous ICE/CED-3-like protease [Homo sapiens]
765	27	4583149	caspase 8 [Homo sapiens]
757	27	4379029	MACH-alpha-2 [Homo sapiens]
757	27	2429162	apoptotic caspase Mch5-beta [Homo sapiens]
747	22	3193167	caspase-8 [Mus musculus]
746	22	4138211	caspase-8 [Mus musculus]
734	22	9454380	caspase-8 [Rattus norvegicus]
716	27	1401352	apoptotic cysteine protease Mch5 isoform alpha [Homo sapiens]
631	16	8778118	caspase-8 [Danio rerio]
610	22	2462593	flice [Mus musculus]
578	18	7619906	caspase-8 [Xenopus laevis]
488	27	3928274	MACH-alpha-3 [Homo sapiens]
426	22	1245144	MCH3/SCA-2 [Mesocricetus auratus]
425	22	2094814	caspase-7 [Mus musculus]
425	22	1894917	Lice2 cysteine protease [Mus musculus]
424	22	1945546	Mch3 [Mus musculus]
423	27	1125073	ICE-LAP3 [Homo sapiens]
422	27	1894913	Lice2 beta cysteine protease [Homo sapiens]

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Query: gi|1498324 apoptotic cysteine protease proMch4
Lineage: Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo

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structures

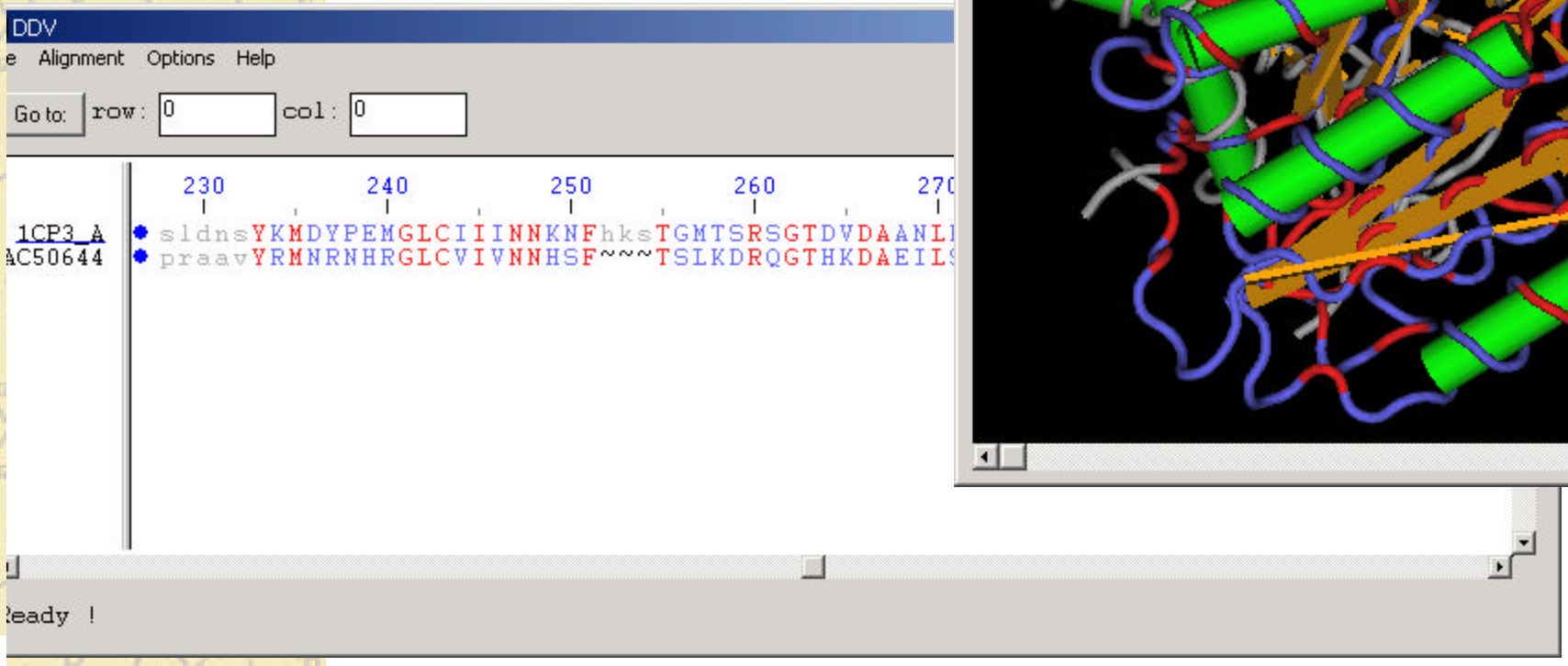
479 aa

SCORE	GI	ORGANISM
323	2780971	Chain A, Crystal Structure Of The Complex Of Apopain With The Tetra
195	2392490	Chain A, Crystal Structure Of The Complex Of Apopain With The Tetra
154	2392491	Chain B, Crystal Structure Of The Complex Of Apopain With The Tetra
151	9257002	Chain B, The 2.8 Angstrom Crystal Structure Of Caspase-3 (Apopain C
146	2914146	Chain A, Crystal Structure Of Inhibited Interleukin-1beta Convertir
146	1127258	Chain A, Interleukin 1-Beta Converting Enzyme (Ice) (E.C.3.4.22.36)
105	4139474	Fadd Death Effector Domain, F25y Mutant, Nmr Minimized Average Stru

Document: Done

Structure of apopain (which has sequence similarity to caspase-10)

Sequence alignment of apopain and caspase-10



ALPS case study

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1: *Cell* 1999 Jul 9;98(1):47-58 Related Articles, Books, OMIM, LinkOut

Inherited human Caspase 10 mutations underlie defective lymphocyte and dendritic cell apoptosis in autoimmune lymphoproliferative syndrome type II.

Wang J, Zheng L, Lobito A, Chan FK, Dale J, Sneller M, Yao X, Puck JM, Straus SE, Lenardo MJ

Laboratory of Immunology, National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, Maryland 20892, USA.

Caspases are cysteine proteases that mediate programmed cell death in phylogenetically diverse multicellular organisms. We report here two kindreds with autoimmune lymphoproliferative syndrome (ALPS) type II, characterized by abnormal lymphocyte and dendritic cell homeostasis and immune regulatory defects, that harbor independent missense mutations in Caspase 10. These encode amino acid substitutions that decrease caspase activity and interfere with death receptor-induced apoptosis, particularly that stimulated by Fas ligand and TRAIL. These results provide evidence that inherited nonlethal caspase abnormalities cause pleiotropic apoptosis defects underlying autoimmunity in ALPS type II.

PMID: 10412980, UI: 99339325

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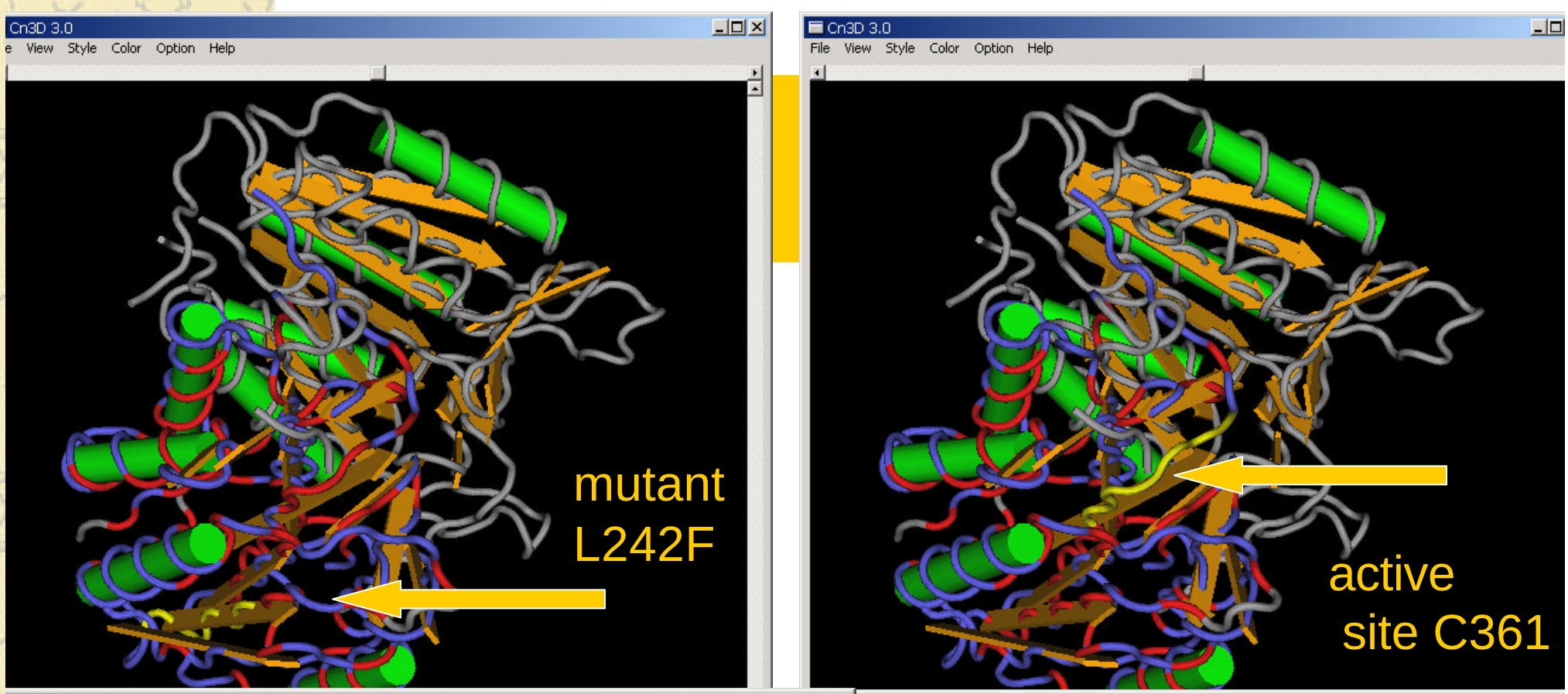
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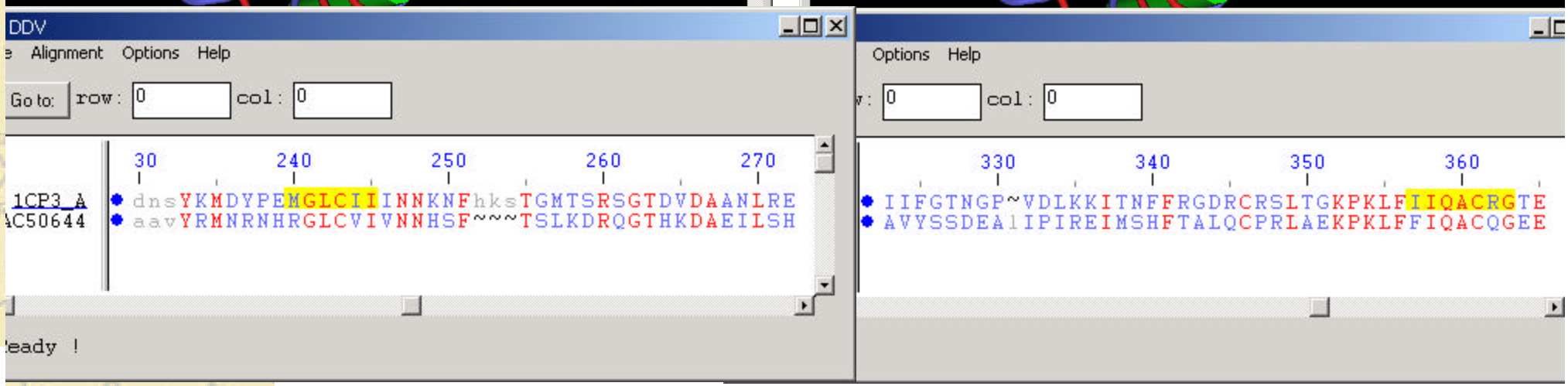
site C361


DDV
Alignment Options Help
Go to: row: 0 col: 0
30 240 250 260 270
1CP3_A dnsYKMDYPEMGLCIIINNKNFhksTGMTSRSGTDVDAANLRE
AC50644 aavYRMNRNHRGLCVIVNNHSF~~~TSLKDRQGTHKDAEILSH
Options Help
col: 0
330 340 350 360
IIFGTNGP~VDLKKITNFFRGDRCRSLTGKPKLFIIQACRGTE
AVYSSDEAIIPIREIMSHFTALQCPRLAEKPKLFFIQACQGEE
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- RGD
- UniGene
- ZFIN

Cytogenetic:	2q33-q34	RefSeq	
Markers(ePCR):	Chr. 2	U60519	U60519
	Chr. 2	H80712	H80712
	Chr. 2	STSG40075	

Key: Link to Map Viewer

Homo sapiens CASP10 Reference Sequence (RefSeq)

Status: PROVISIONAL

Nucleotide: NM_001230

Protein: NP_001221 caspase 10, apoptosis-related cysteine protease

GenBank Source: U60519

GenBank Sequences

Nucleotide	Type	Protein
U60519	m	AAC50644
U86214	m	AAB46730

Key: Related Structures

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