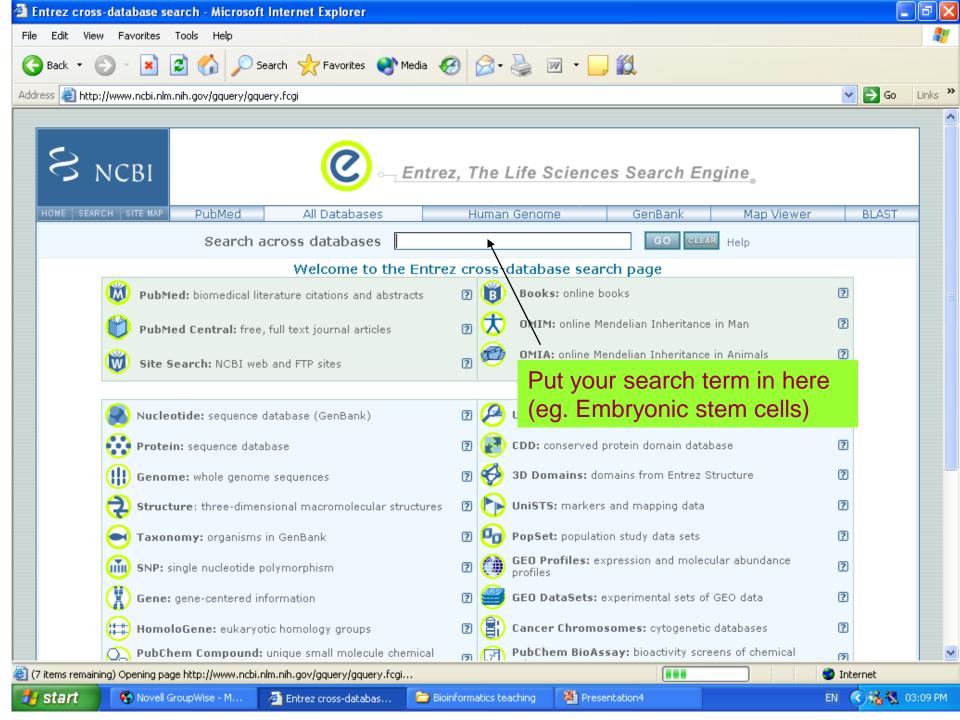
ENTREZ can search across all NCBI databases at the same time

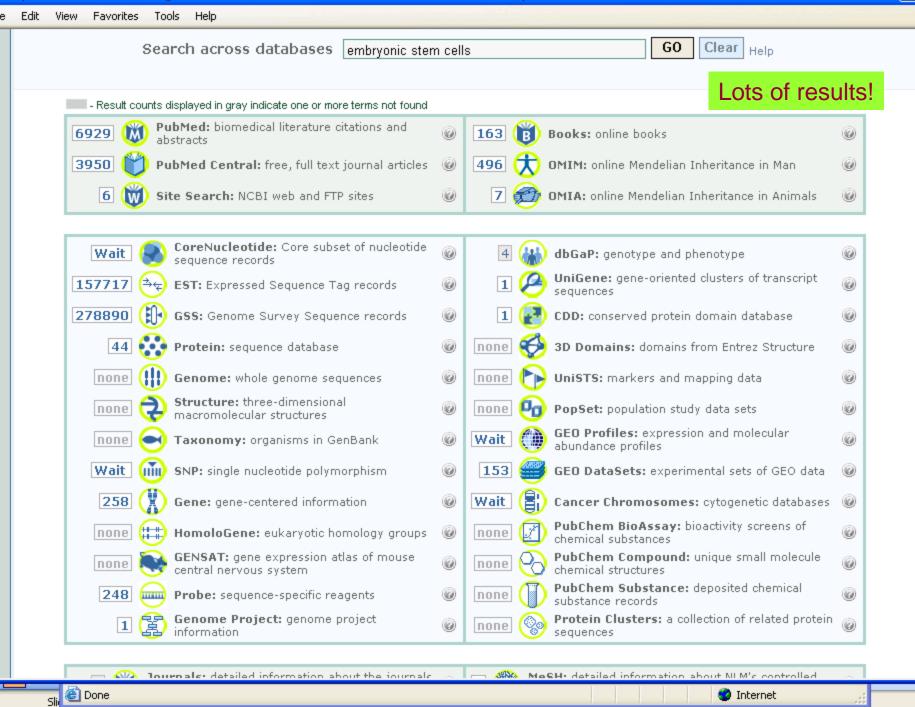


NCBI



ARCH SITE MAR PubMed All Databases GenBank Human Genome Map Viewe Search across databases GO Clear Help Welcome to the Entrez cross-database search page PubMed: biomedical literature citations and abstracts Books: online books B PubMed Central: free, full text journal articles OMIM: online Mendelian Inheritance in Man OMIA: online Mendelian Inheritance in Animals Site Search: NCBI web and FTP sites Nucleotide: Core subset of nucleotide sequence records dbGaP: genotype and phenotype EST: Expressed Sequence Tag records UniGene: gene-oriented clusters of transcript sequences (6) GSS: Genome Survey Sequence records CDD: conserved protein domain database Protein: sequence database 3D Domains: domains from Entrez Structure (6) UniSTS: markers and mapping data Genome: whole genome sequences 6 Structure: three-dimensional macromolecular structures PopSet: population study data sets (6) Taxonomy: organisms in GenBank GEO Profiles: expression and molecular abundance profiles (4) SNP: single nucleotide polymorphism GEO DataSets: experimental sets of GEO data dbVar: Genomic structural variation Cancer Chromosomes: cytogenetic databases (4) Gene: gene-centered information PubChem BioAssay: bioactivity screens of chemical substances (6) SRA: Sequence Read Archive PubChem Compound: unique small molecule chemical structures PubChem Substance: deposited chemical substance records BioSystems: Pathways and systems of interacting molecules









Resources



Welcome to NCBI

The National Center for Biotechnology Information advances science and health by providing access to biomedical and genomic information.

More about the NCBI | Mission | Organization | Research | RSS

Genome Reference Consortium

Formed to improve human and mouse reference assemblies, GRC will fix loci misrepresented in reference assembly, fill remaining gaps, and make alternate representations of complex loci.



11 2 3 4

How To...

- Obtain the full text of an article
- Retrieve all sequences for an organism or taxon
- Find a homolog for a gene in another organism
- Find genes associated with a phenotype or disease
- Design PCR primers and check them for specificity
- Find the function of a gene or gene product
- Determine conserved synteny between the genomes of two organisms

See all ...

Popular Resources

- PubMed
- PubMed Central
- Bookshelf
- BLAST
- Gene
- Nucleotide
- Protein
- · GEO
- Conserved Domains
- Structure
- PubChem

NCBI News

NIH Roadmap

22 Apr 2010

Epigenomics Project data

in GEO database

GEO's Roadmap Epigenomics Project Data Listings page allows ...

March News issue available 09 Apr 2010

Includes My NCBI, E-Utility, and BLAST news.

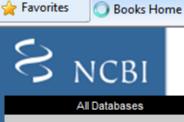
NIH announces Genetic 23 Mar 2010

Testing Registry

NCBI will be responsible for

dayalaning the regist

You can search for biomedical Books **B**ookshelf PubMed Protein Structure PMC Nucleotide Genome Go Clear Preview/Index History Clipboard Details The Bookshelf is a growing collection of biomedical books that can be searched directly by typing a concept into the textbox above and selecting "Go". Try one of these searches: cell cycle control immunodeficiency protein evolution New on the Bookshelf: Comparative Oncology Baba, Alecsandru Ioan; Câtoi, Cornel Bucharest: The Publishing House of the Romanian Academy; 2007 Bethesda (MD): National Library of Medicine (US), NCBI; 2009 Health, United States, 2008 Atlanta (GA): Centers for Disease Control and Prevention; 2008 The National Academies Collection: Reports funded by National Institutes of Health





for

Limits

Familial

Cancer

About Entrez

Books

Overview

Using the books

Information for authors and publishers

What's new 🔊

Project background

FAQ

My NCBI

Privacy Policy

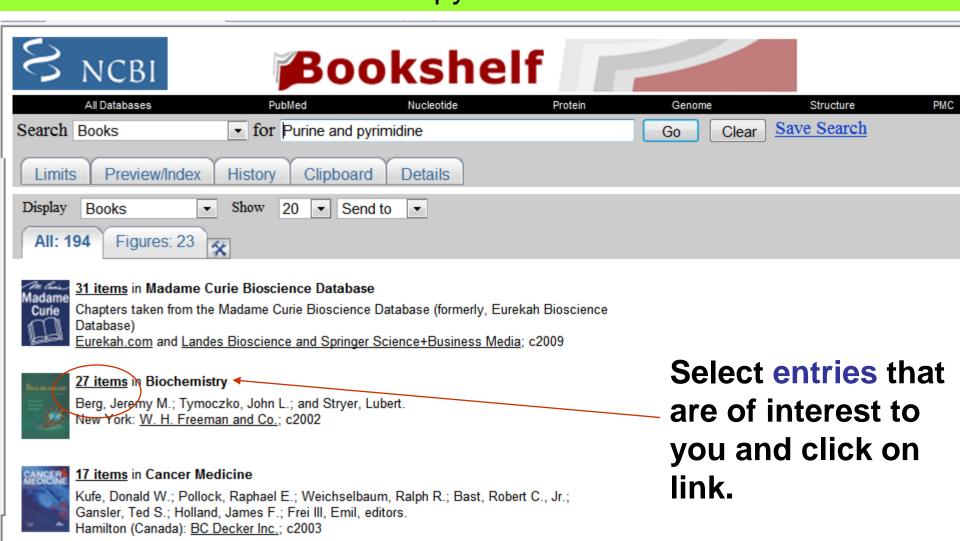
Familial Cancer Syndromes [Internet]

Riegert-Johnson, Douglas L.; Boardman, Lisa A.; Hefferon, Timothy; Spurck, Lauren, editors

Washington (DC): The National Academies Press; c2006-2009

Search: Purine and pyrimidine

Result: You get a number of books containing the words purine and pyrimidine.

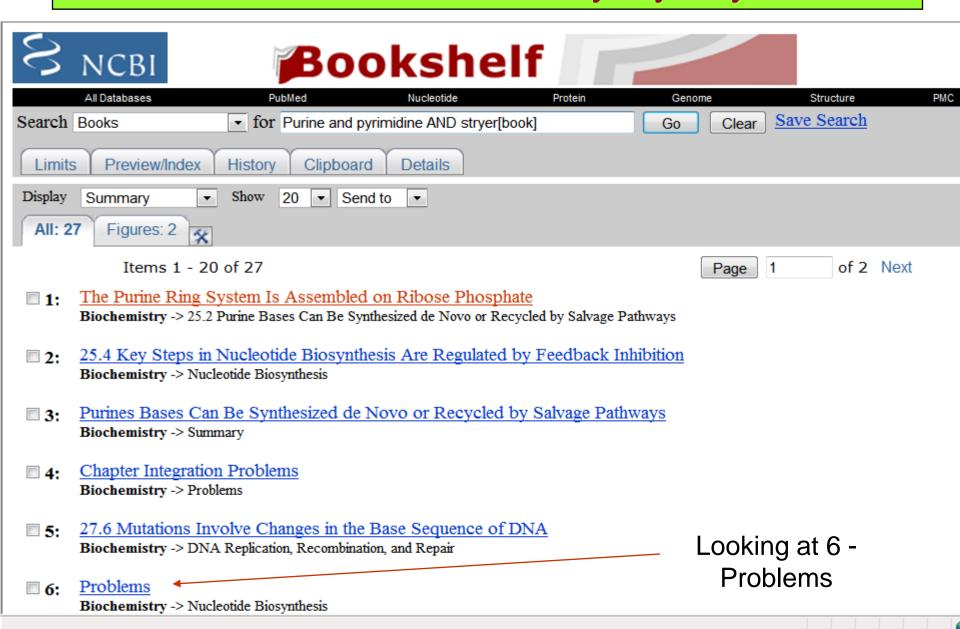


15 items in Introduction to Genetic Analysis

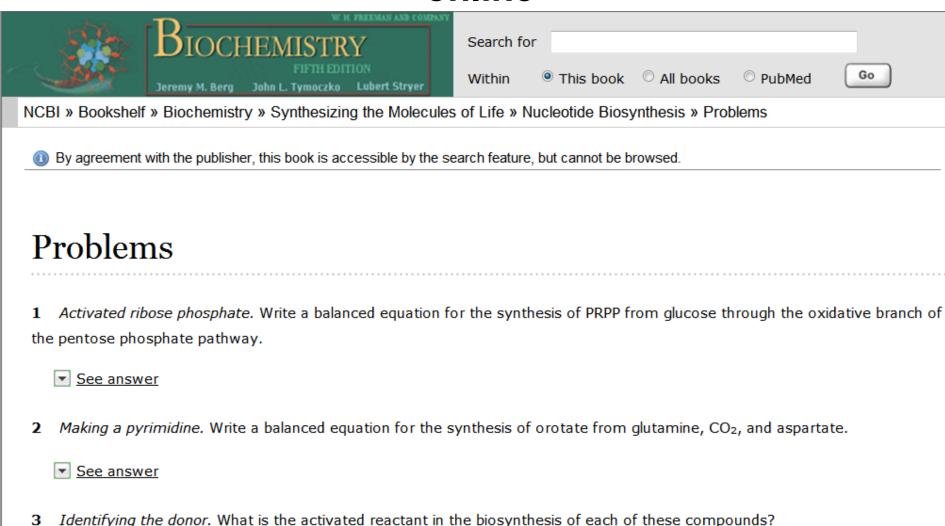
Criffitha Anthony I.E. Millar Jaffray U. Suzuki David T. Lawantin Diabard C. Calbart

Done

By selecting option 2, the search has been limited to a book called "Biochemistry" by Stryer et al



You can test yourself and check your answers online

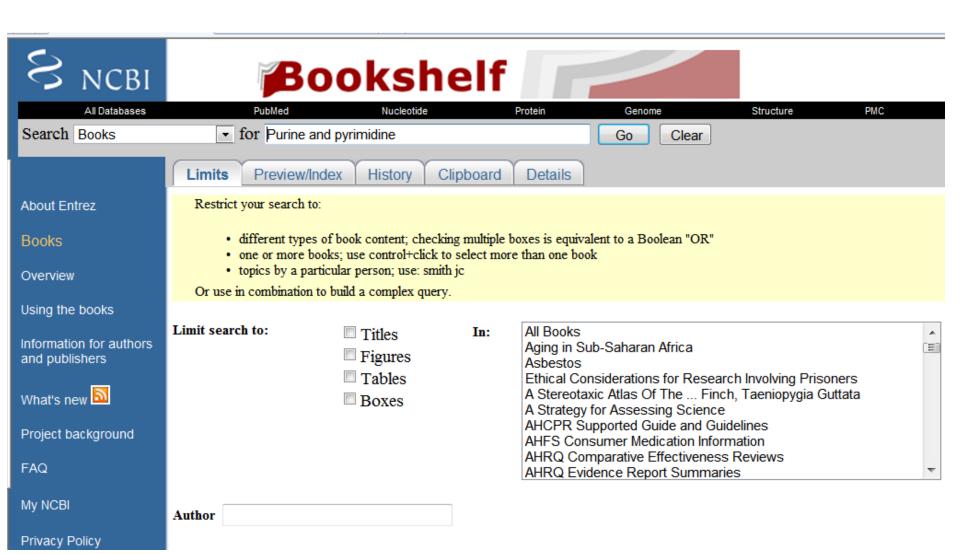


(a) Phosphoribosylamine

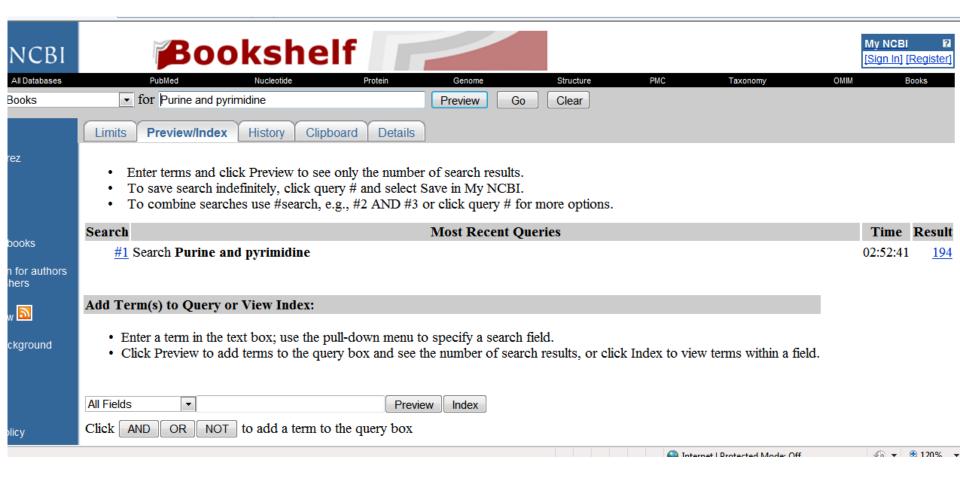
Details, Limits, Preview ...

- 1. Limits: ways to restrict a search to a defined subset of the database.
- 2. Preview/Index: list last searches; combine with new search terms
- History: review, delete, recall and combine past searches (max 100)
- 4. Clipboard: temporarily store results
- 5. Details: review interpretation of the query term

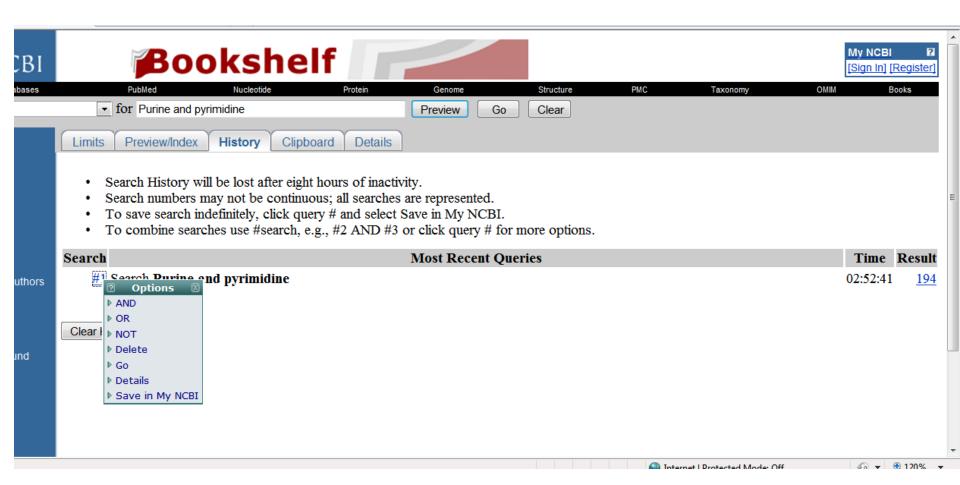
LIMITS



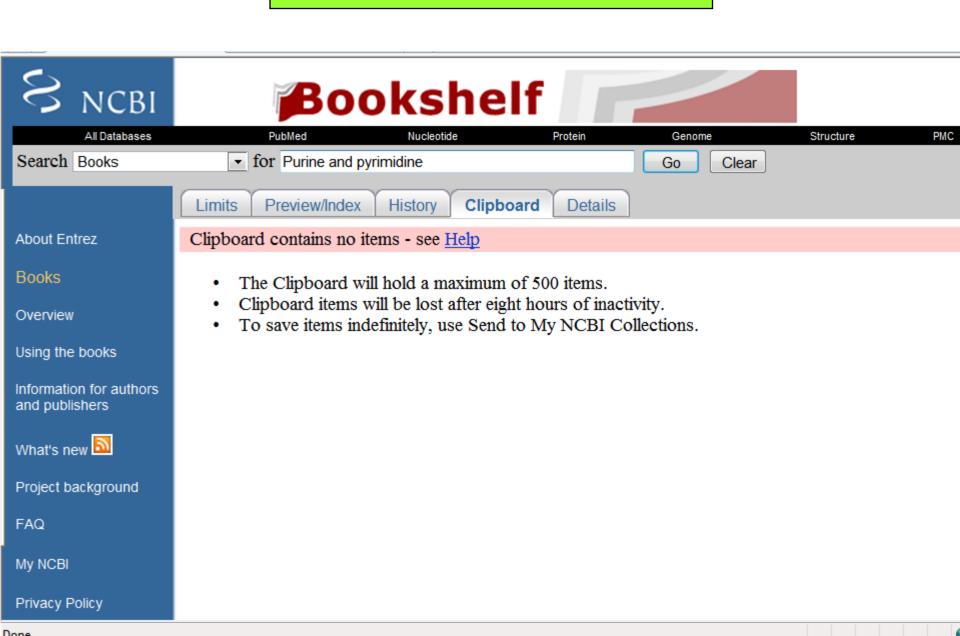
PREVIEW INDEX



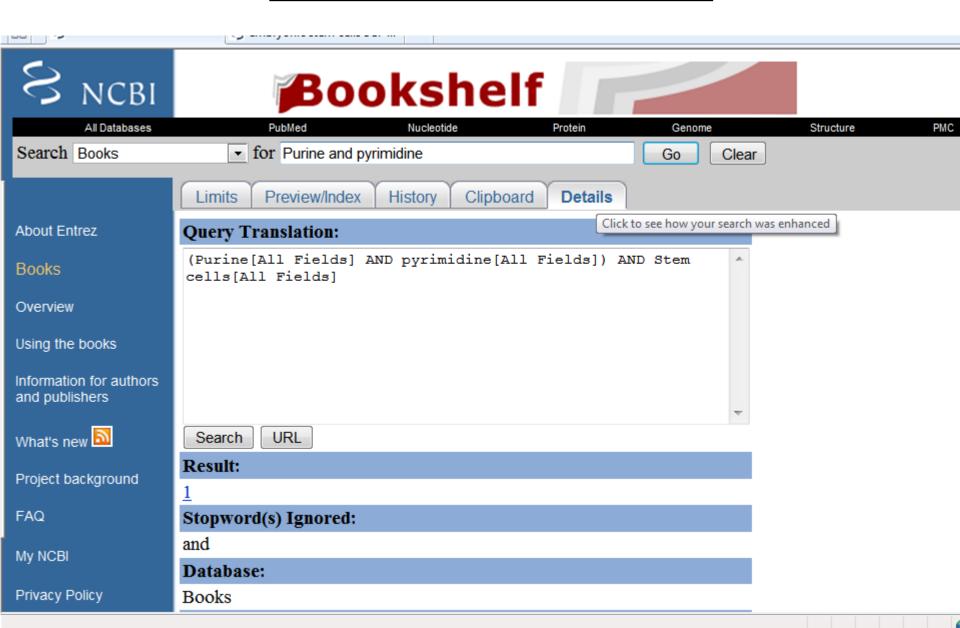
HISTORY



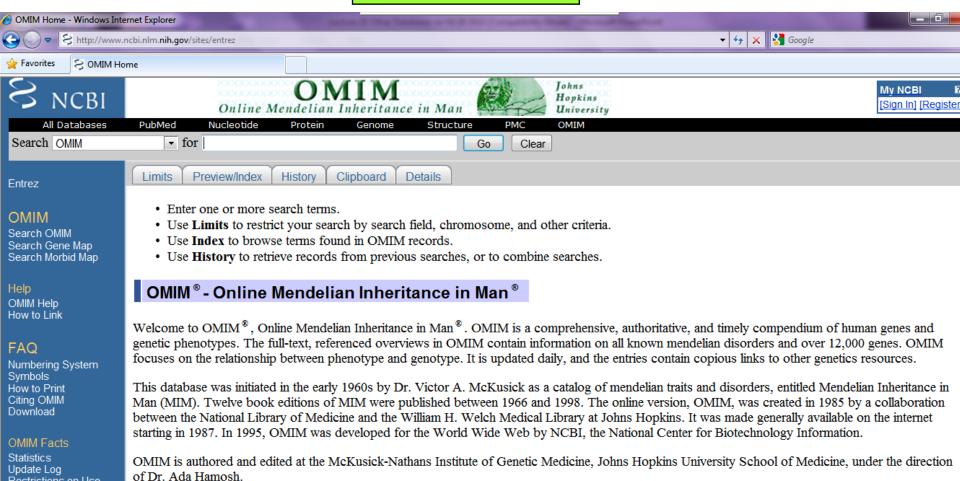
CLIPBOARD



DETAILS



Catalog of human genes and genetic disorders



NLM's Profiles in Science -- The McKusick Papers More...

Restrictions on Use

Allied Resources Genetic Alliance

NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by

OMIM database searching (human genes and genetic disorders)

- OMIM: "online Mendelian inheritance in man".
- Search term: "adenosine deaminase".
 - Found: 47 entries
 - Chose: #102700 (severe combined immunodeficiency)
- NOTE: the # symbol is used to show mutants



OMIM Online Mendelian Inheritance in Man



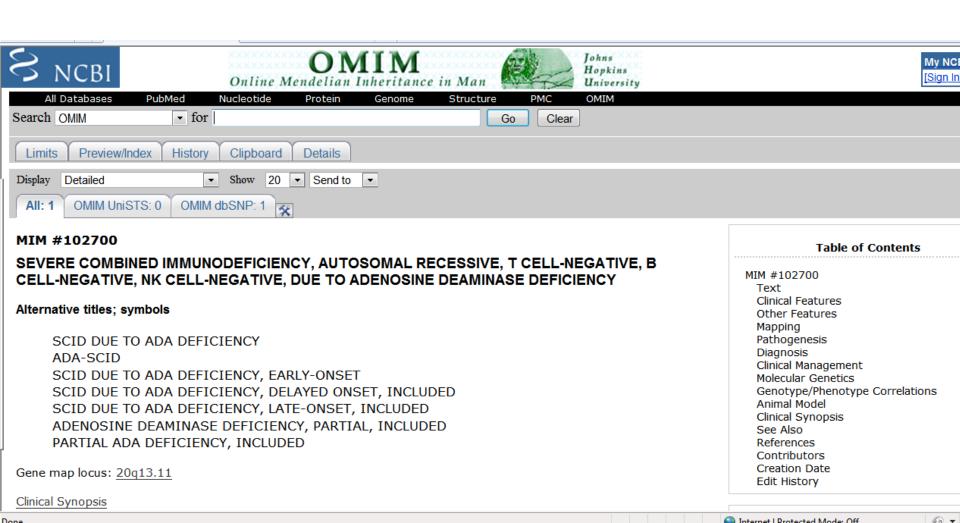
Johns Hopkins University

All Databases PubMed Nucleotide Protein Genome Structure Pl	PMC OMIM	
Search OMIM ✓ for adenosine deaminase Go	Clear Save Search	
Limits Preview/Index History Clipboard Details		
Display Titles ▼ Show 20 ▼ Send to ▼		
All: 47 OMIM UniSTS: 10 OMIM dbSNP: 9		
Items 1 - 20 of 47	Page 1 of 3 Next	
■ 1:*146920. ADENOSINE DEAMINASE, RNA-SPECIFIC; ADAR Gene map locus 1q21.3	MGI, Links	
2:#102700. SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESS -NEGATIVE, B CELL-NEGATIVE, NK CELL-NEGATIVE, DUE TO ADENOSINE DEAMINASE DEFICIENCY SCID DUE TO ADA DEFICIENCY, DELAYED ONSET, INCLUDED Gene map locus 20q13.11	-	
☐ 3: *608958. ADENOSINE DEAMINASE; ADA Gene map locus 20q13.11	MGI, GeneTests, Links	
■ 4:±102770. ADENOSINE MONOPHOSPHATE DEAMINASE 1; AMPD1 MYOADENYLATE DEAMINASE DEFICIENCY, MYOPATHY DUE TO, INCLUDED Gene map locus <u>1p21-p13</u>	MGI, GeneTests, Links	
■ 5.*601218. ADENOSINE DEAMINASE, RNA-SPECIFIC, B1: ADARB1	MGL Links	

D---

The result gives you all the information about the disease in man and includes:

Clinical features; Pathogenesis, Diagnosis; Animal models, Clinical management and references for all the above.



Clinical management

homozygous ADA-deficient fetus, respectively. The diagnoses were confirmed after birth and in abortus tissue.



Clinical Management

Back to Top

Enzyme Replacement Therapy

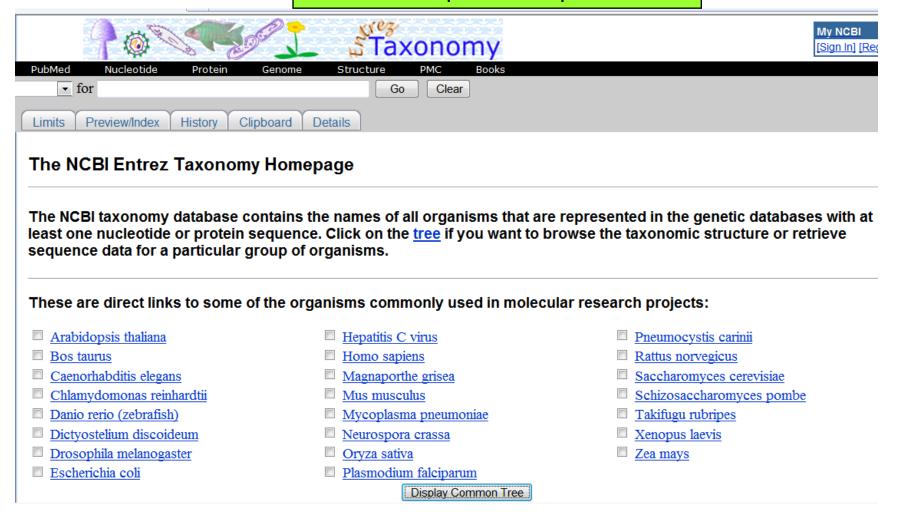
Polmar et al. (1976) reported successful treatment of a child with SCID due to ADA deficiency by 'enzyme replacement therapy' using frozen irradiated red blood cells with normal ADA activity. After treatment, a thymic shadow appeared radiographically, lymphocytic responses were demonstrated in vitro, and there was immunoglobulin synthesis. With infusions at 4-week intervals, the child remained free of infection for 17 months.

Ziegler et al. (1980) reported a patient with SCID due to ADA deficiency who was treated with ADA-positive red cell infusions. Although there was some resolution of interstitial pneumonitis and skeletal abnormalities, there was no evidence of immunologic reconstitution, and the patient died at age 17 months. The authors noted that severe cases of SCID due to ADA deficiency may not respond to exogenous enzyme therapy. Markert et al. (1987) reported 5 ADAdeficient patients who showed no lasting benefit from red blood cell transfusions.

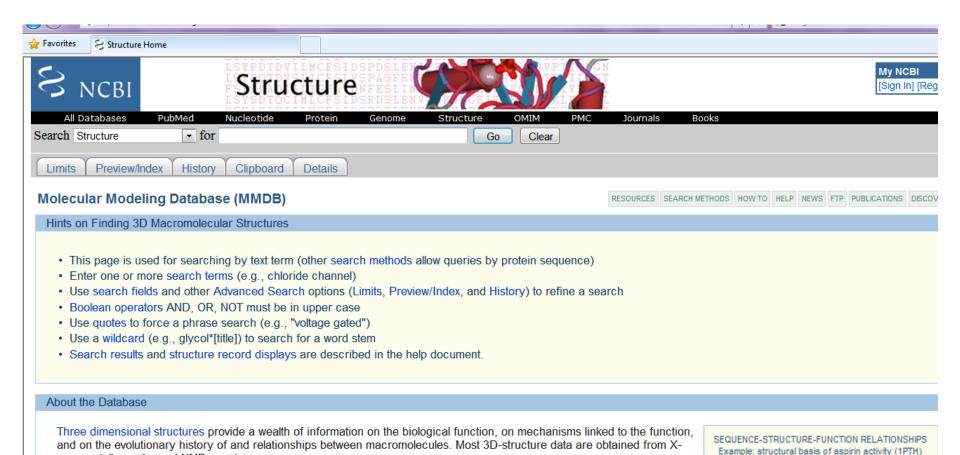
Hershfield et al. (1987) reported successful treatment of 2 SCID ADA-deficient patients with polyethylene glycolmodified bovine intestinal ADA (PEG-ADA). The modified enzyme was rapidly absorbed after intramuscular injection and had a half-life in plasma of 48 to 72 hours. Weekly doses maintained plasma ADA activity at 2 to 3 times the level of red cell ADA in normal subjects, resulting in a decrease in toxic deoxyadenosine nucleotides to less than 0.5% of total adenine nucleotides. The activity of S-adenosylhomocysteine hydrolase, which is inactivated by deoxyadenosine, increased to normal in red cells and nucleated marrow cells. Neither toxic effects nor hypersensitivity reactions were observed. In vitro tests of cellular immune function of each patient showed marked improvement, together with an increase in T lymphocytes. Covalent attachment of polyethylene glycol to ADA blocked access to vulnerable sites on the surface of the protein, inhibiting clearance from the circulation, attack by degrading enzymes, binding of antibodies, and processing by antigen-presenting cells.

Love at al. (1000) reported a child who developed aumptoms of CCID due to ADA deficiency at ago 2 years. Che had

Organisms represented in the genetic database with at least one nucleotide or protein sequence



3D structure database as well as tools for visualization

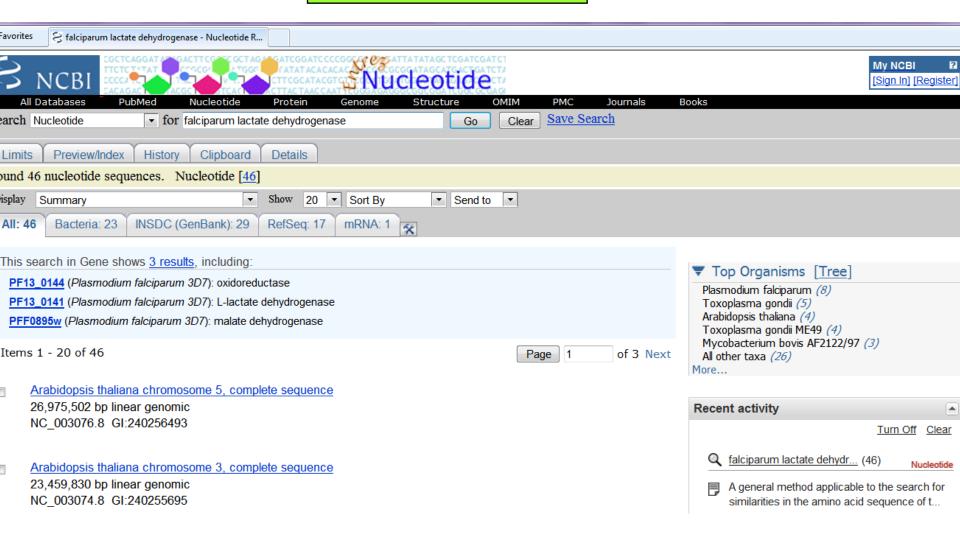


The Molecular Modeling DataBase (MMDB), also known as "Entrez Structure," is a database of experimentally determined structures obtained from the RCSB Protein Data Bank (PDB). MMDB is developed by the Structure Group of the NCBI

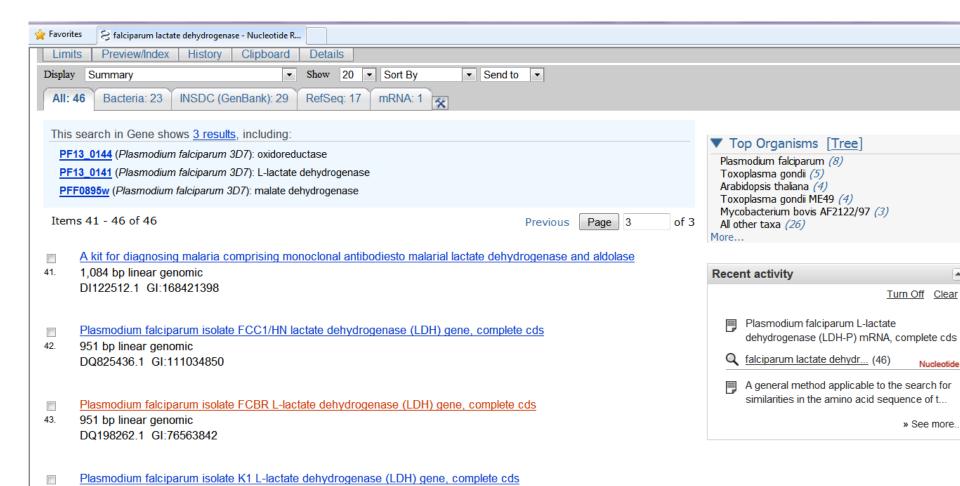
1PTH - Cn3D 4.2

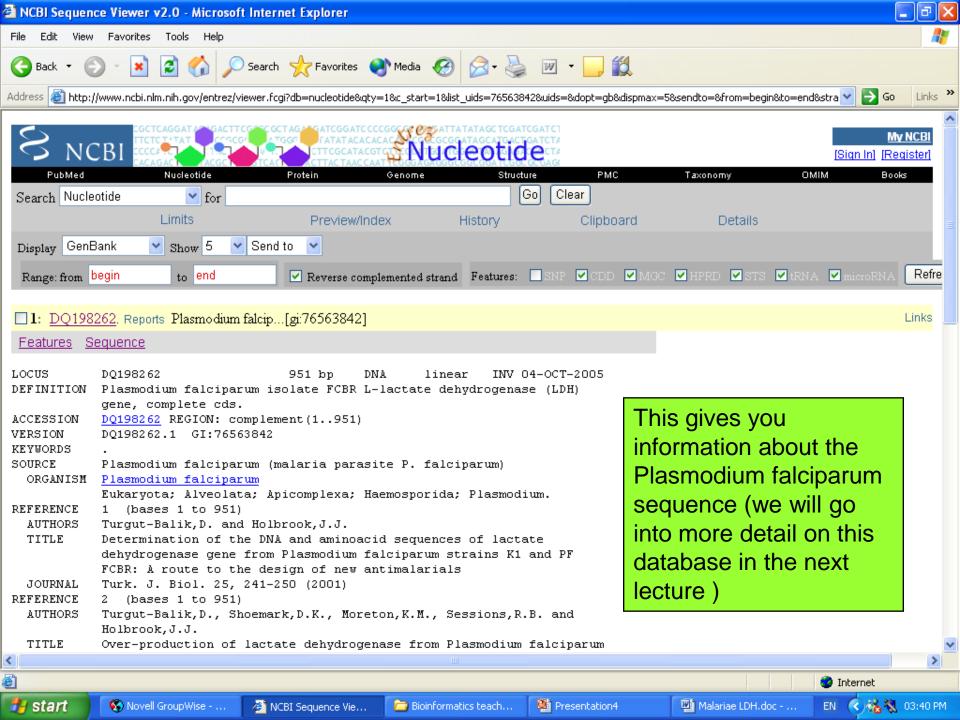
ray crystallography and NMR-spectroscopy.

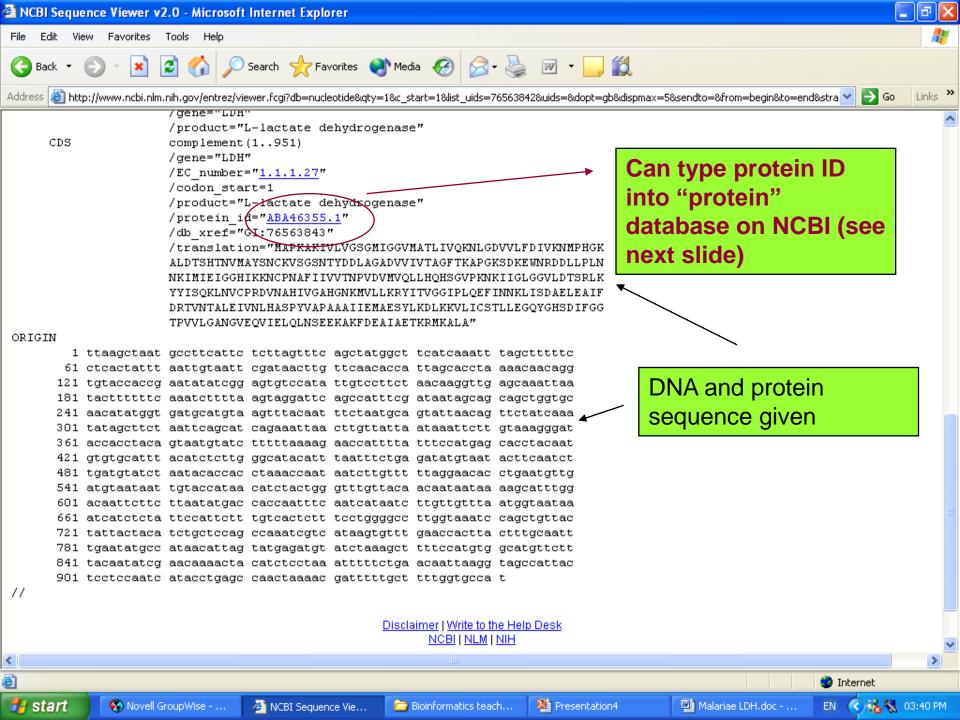
You can search for RNA/DNA sequences on nucleotide



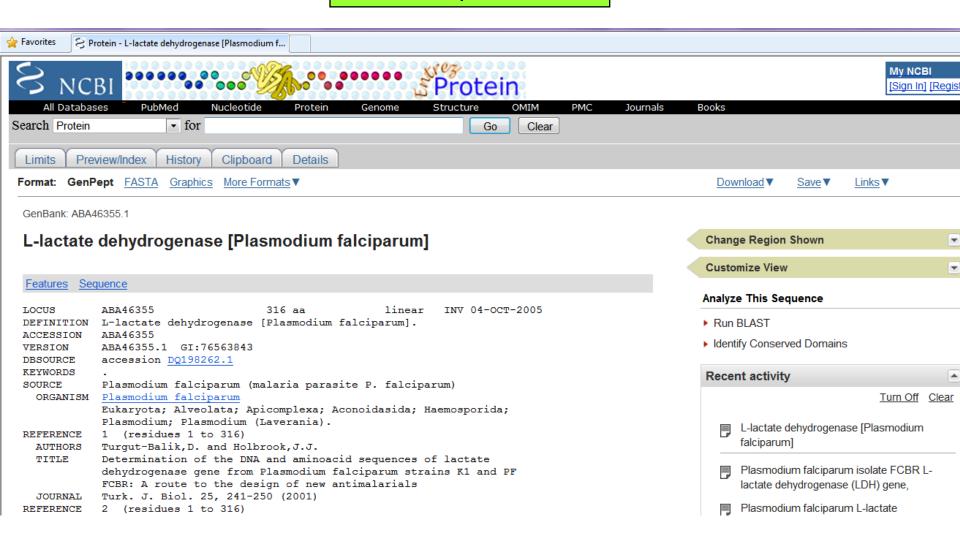
Find entry number 43 and click on the link







Info on the protein



```
Elazig 23169, Turkey
                    Location/Qualifiers
FEATURES
                     1..951
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                    /organism="Plasmodium falciparum"
                     /mol type="genomic DNA"
                                                                   Tells you about the
                     /isolate="FCBR"
                     /db xref="taxon:5833"
                                                                   different regions of
                     <1...>951
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                     /gene="LDH"
                                                                   the protein (see next
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                                                                   lecture for more detail)
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                     DRTVNTALEIVNLHASPYVAPAAAIIEMAESYLKDLKKVLICSTLLEGQYGHSDIFGG
                     TPVVLGANGVEQVIELQLNSEEKAKFDEAIAETKRMKALA"
ORIGIN
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