

Champs référentiels 1.1.1, 1.1.2 et 1.1.3

Chapitre 1, domaine 1 : Documentation
**Sources d'informations en
ligne**

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Faculté de Sciences Pharmaceutiques

12 juin 2007



L'information en ligne

Moteurs et métamoteurs

Les sites « généralistes »

- Moteurs
 - Par mots clés (google, yahoo,...)
 - Spécialisés (picsearch, cnet, imdb,...)
 - « Alternatifs » (quintura...)
- Métamoteurs
 - En ligne (ixquick, aftervote, kartoo, sputtr...)
 - À installer localement : copernic

Les portails spécialisés dans le domaine de la santé

- Le CiSMef : <http://www.cismef.org/>
 - Catalogue et index des sites médicaux francophones
- Le portail Santé : <http://www.sante.fr/>
 - Liens vers les agences sanitaires
 - Ministère, AFSSAPS, HAS, INVS, ...
- L'UMVF : <http://www.umvf.org/>
 - Université médicale virtuelle francophone

Les bases de données spécifiques d'une thématique

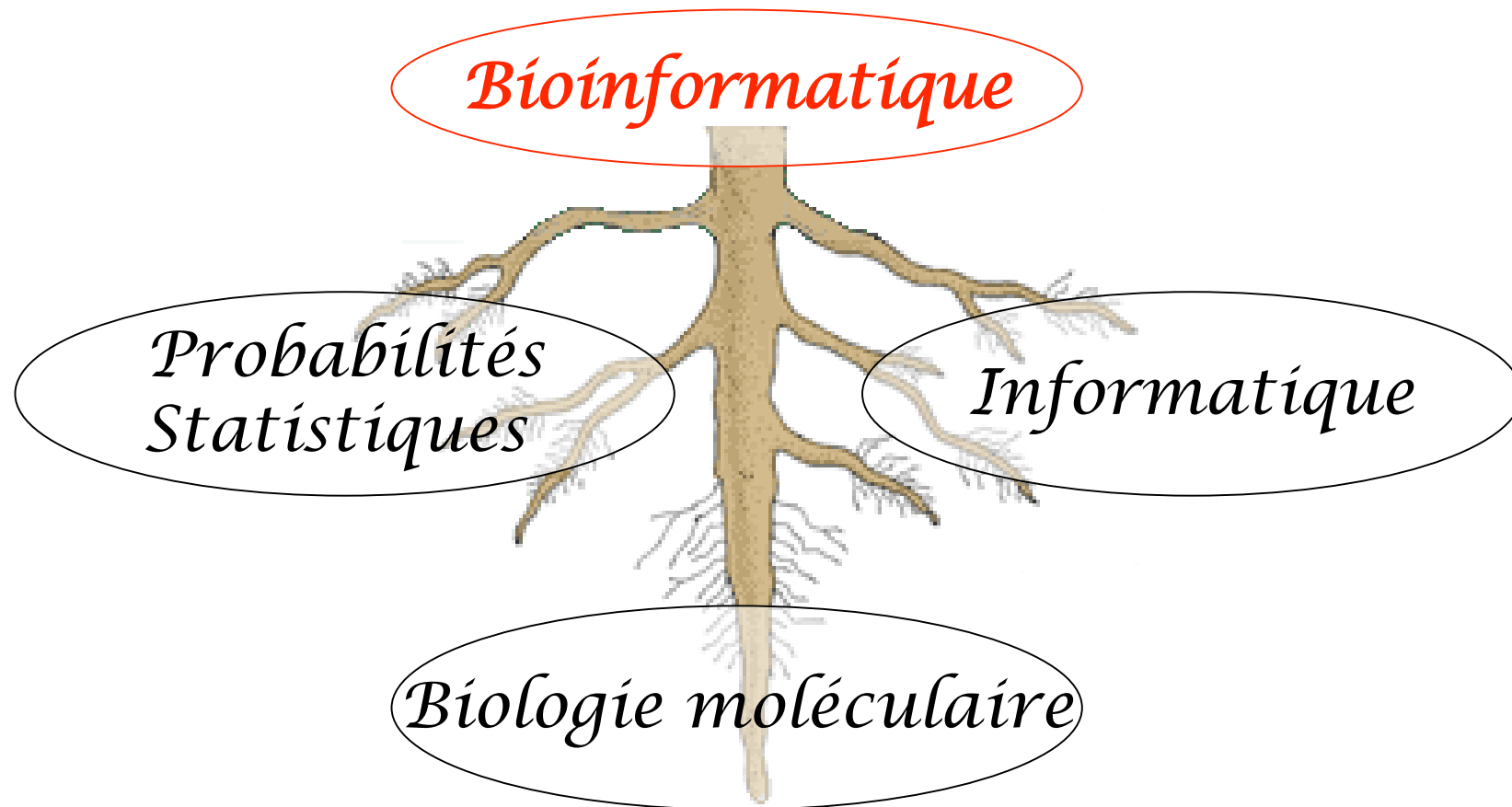
- Bases de médicaments
 - Vidal : <http://use.evidal.net/> (codes sur l'intranet Paris Descartes)
 - Thériaque : <http://www.theriaque.org/>
 - Biam : <http://www.biam2.org/>
 - ⚠ Dernière mise à jour en 2001 (reprise par Vidal)
- Chemfinder : <http://chemfinder.com/>
 - Données physicochimiques
- Toxnet : <http://toxnet.nlm.nih.gov/>
 - Données toxicologiques
- Bcb : <http://www.resip.fr/>
 - Aide à la prescription (banque Claude Bernard, intégrée dans les logiciels de cabinets médicaux et d'officines)

Classifications dans le domaine de la santé

- MeSH (Medical Subject Headings)
 - Mots clés
- MedDRA (Medical Dictionary for Regulatory Activities)
 - Classification d'effets indésirables, pharmacovigilance
- ATC (Anatomical Therapeutic Chemical)
 - Classification de médicaments
- SNOMED (Systematized Nomenclature of Medicine)
 - Nomenclature médicale

Initiation à la fouille de données en bioinformatique

Qu'est-ce que la bioinformatique ?



Plan

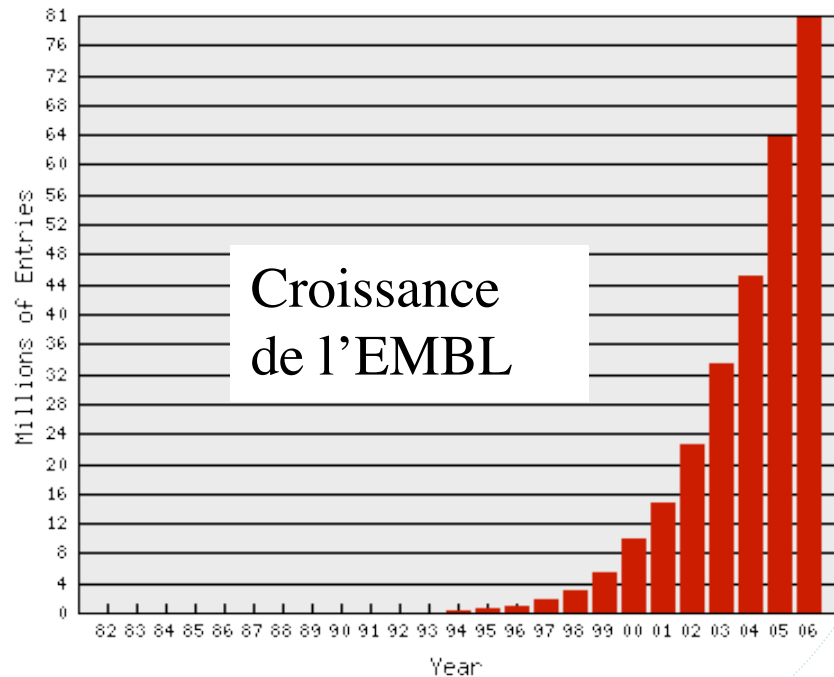
- Introduction aux bases de données
- Données disponibles
 - Bases nucléiques généralistes
 - Bases protéiques généralistes
 - Bases spécialisées
- Recherche d'information
 - L'interface SRS
 - L'interface Entrez
 - Interfaces spécifiques

À l'origine : un besoin de classification

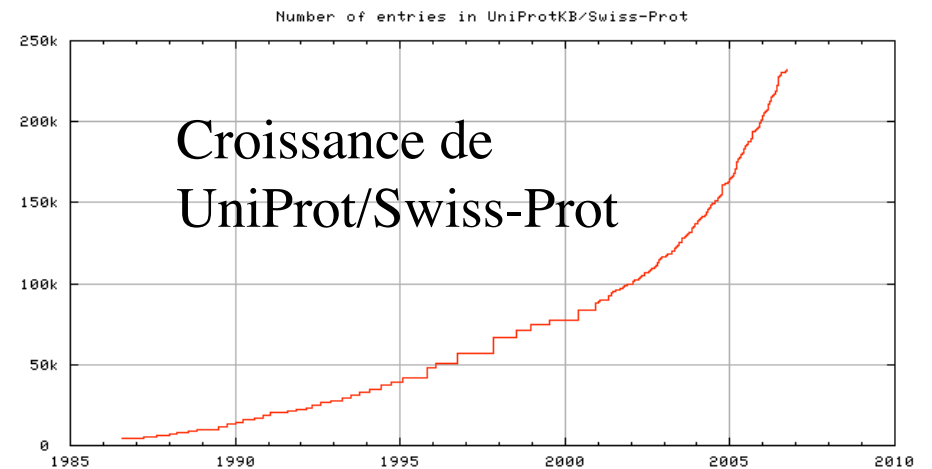
Scientifique	Date	Nombre de plantes répertoriées	
Théophraste	4 th siècle av. J.C.	500	
C. Bauhin	17 th siècle	6000	
C. Von Linné	début 18 th siècle	18000	Invention de la taxonomie
G. Cuvier	fin of 18 th siècle	50000	

L'informatique devient nécessaire : croissance exponentielle de données

81 millions séquences de nucléotides



232000 séquences de protéines

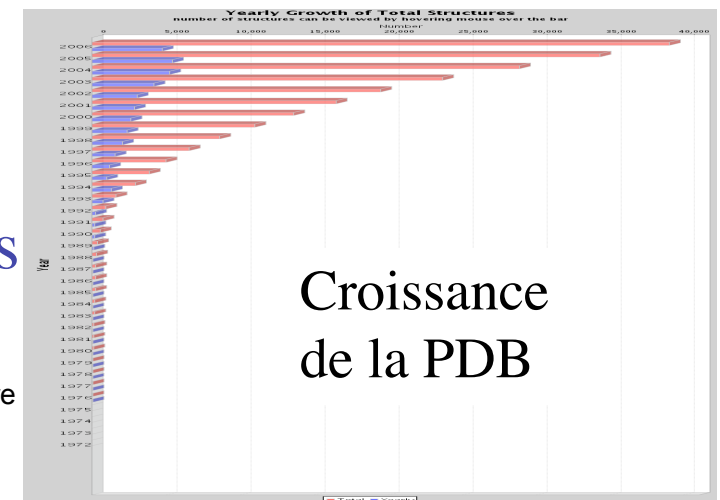


14 millions citations
bibliographiques (PubMed)

I. Nicolis
Faculté de Pharmacie
Université Paris Descartes

36000 structures
de protéines

C2i Métiers de la Santé - Année universitaire
2006/2007 Tous droits réservés.



Quelles données sont disponibles?

- Littérature
- Séquences de nucléotides
- Séquences de protéines
- Structures de protéines
- Motifs fonctionnels de protéines
- Voies métaboliques
- Maladies héréditaires
- Génomes

...

Plan

- Introduction aux bases de données
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Bases de données nucléiques

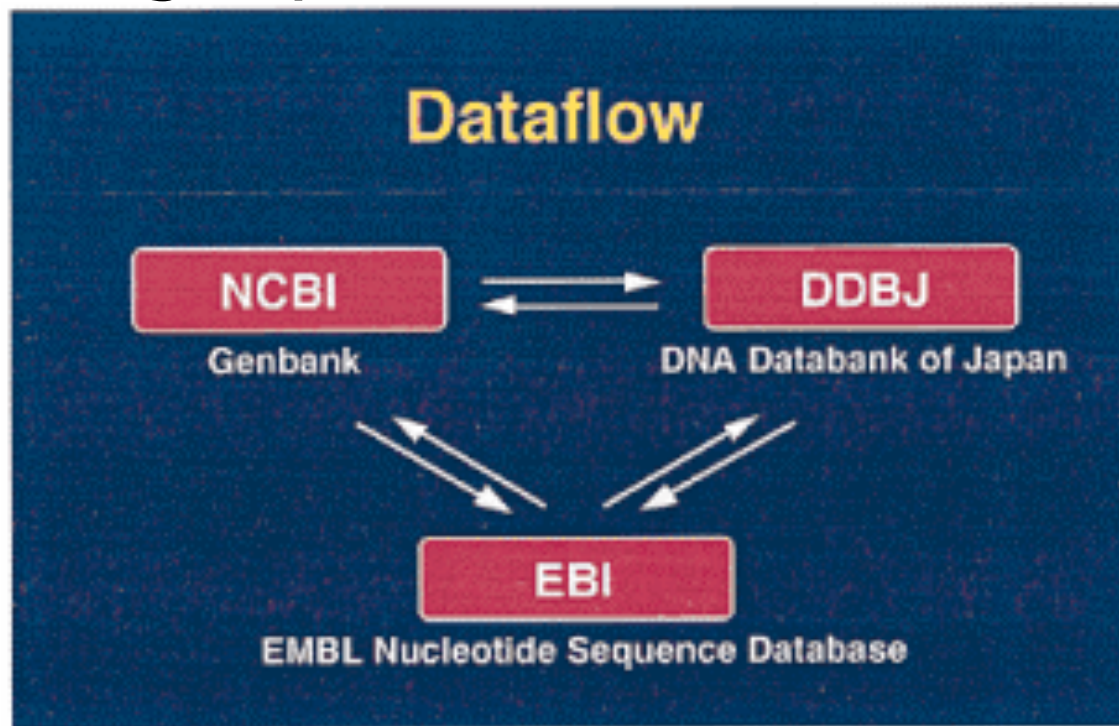
- USA: GSDB (1979) , GenBank (1986)
- Europe: EMBL (1980)
- Japan: DDBJ (1984)

⇒ 1990: collaboration

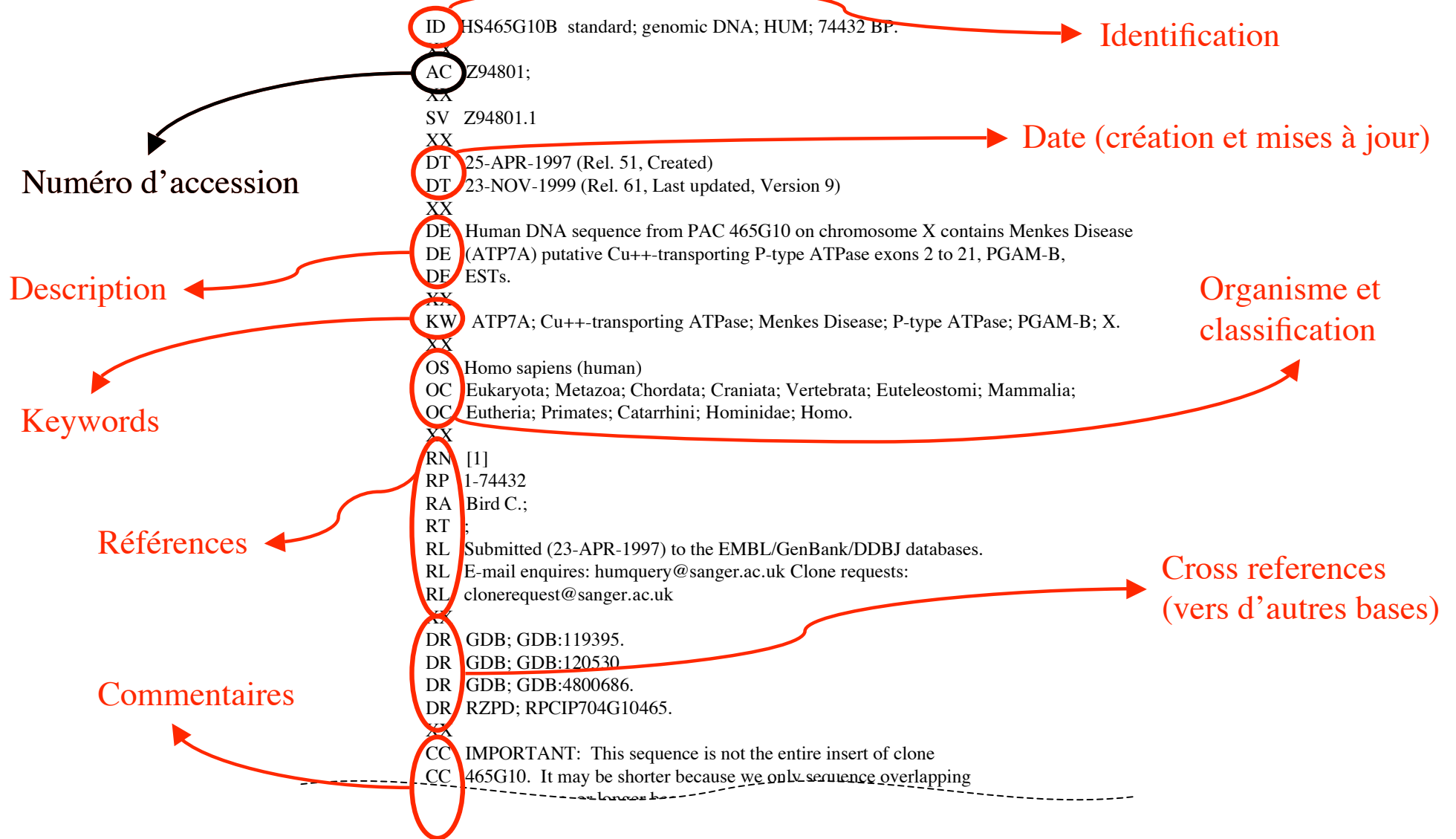
- Création de l'International Nucleotide Sequence Databases (INSD)

International Nucleotide Sequence Databases (INSD)

- Format commun
- Échange quotidien de données



Le format EMBL (I)



Le format EMBL (II)

Features

Séquence

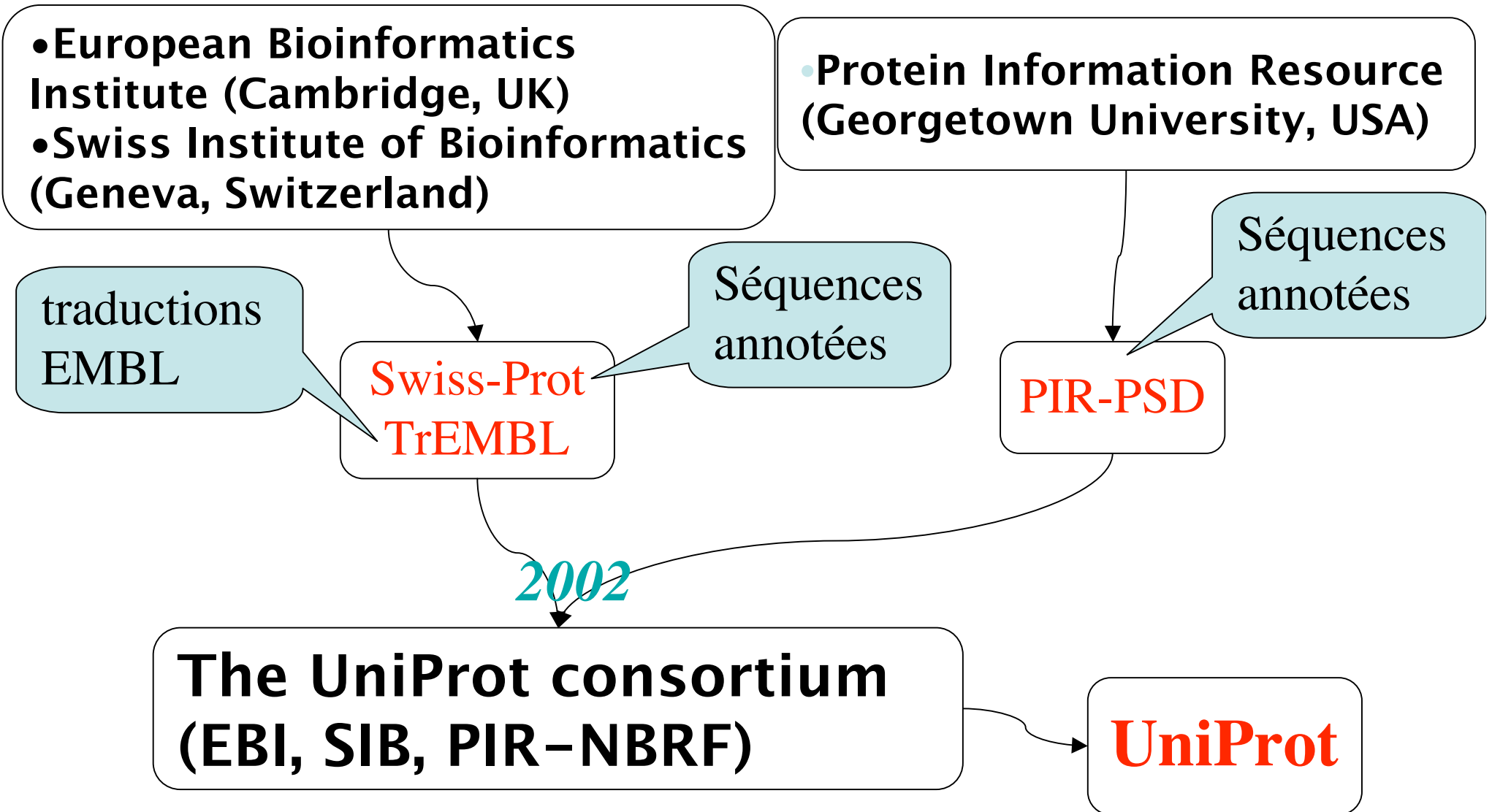
```
##
##
## Key      Location/Qualifiers
##
FH
FT source      1..74432
FT             /chromosome="X"
FT             /db_xref="taxon:9606"
FT             /db_xref="RZPD:RPCIP704G10465"
FT             /mol_type="genomic DNA"
FT             /organism="Homo sapiens"
FT             /map="X"
##-----
FT             /db_xref="GeneW:869"
FT             /db_xref="UniProt/Swiss-Prot:Q04656"
FT             /note="similar to L06133"
FT             /product="Menkes Disease (ATP7A)"
FT             /protein_id="CAB08162.2"
FT             /translation="MDPSMGVNSVTISVEGMTCSVCVWTIEQQIGKVNQVHHIKVSLEE
##-----
FT             /note="13 copies of 2 mer TTT % conserved"
FT repeat_region 9389..9454
##-----
XX
SQ Sequence 74432 BP; 24092 A; 14166 C; 14263 G; 21911 T; 0 other;
tatactaaaa atacaaaaat tagtggggcg ttgtggcggg cacctatagt cccagtact   60
caaaaactc aggcaggaga actgcttga cccaggagat ggaggctca caaacccag
gtacgagcc gatcagagc gggcg ggcaggcgc
acgagatgct ggctatgagt ttgacatctg cctcaccctca gtgcagaaga gagtgatccg   74400
gaccctctgg acagtgctag atgccattga tc                               74432
//
```

Les données
brutes sont
mises en forme
par une
interface web

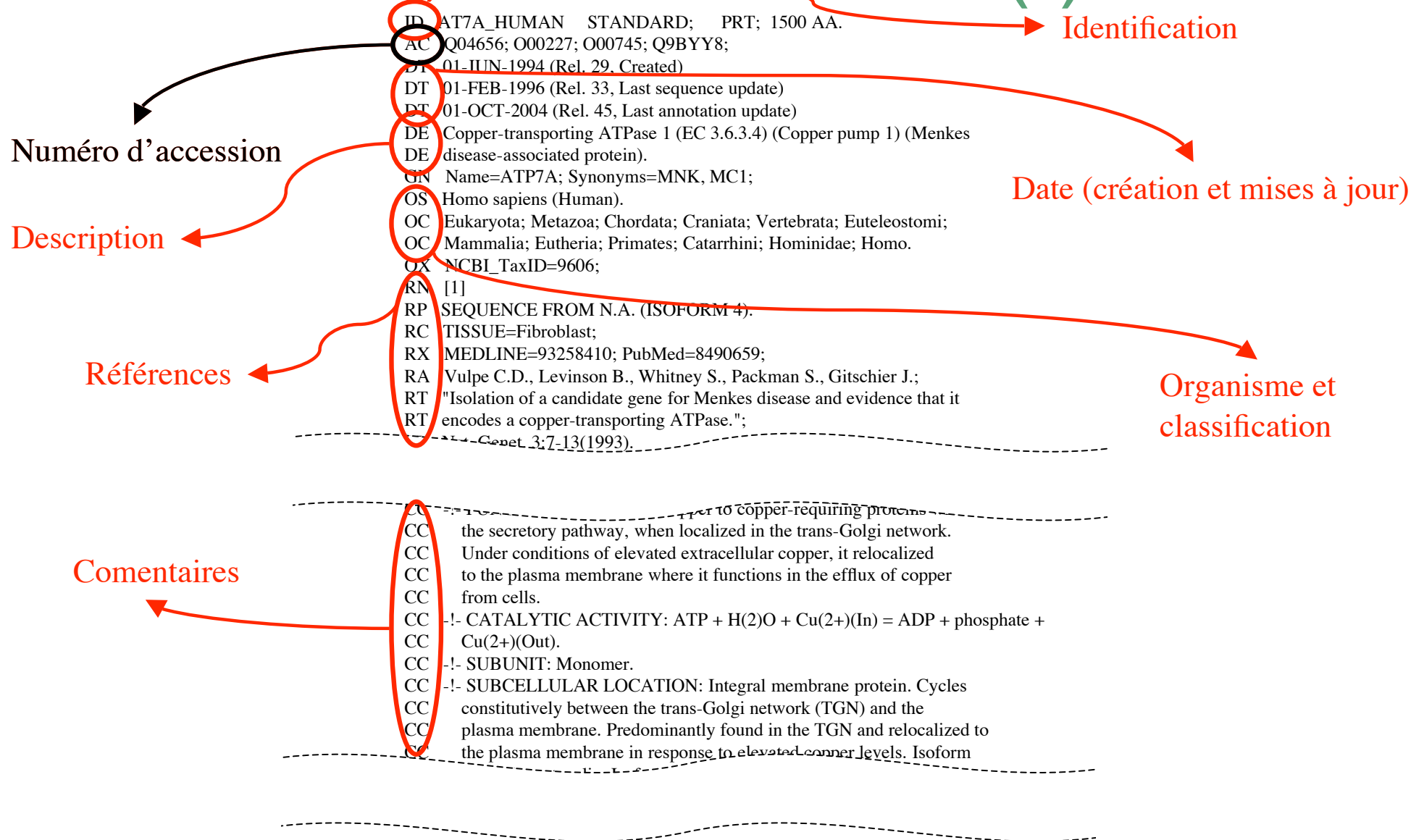
Plan

- Introduction aux bases de données
- **Données disponibles**
 - Bases nucléiques généralistes
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Bases de séquences protéiques



Le format UniProt (I)



Le format UniProt (II)

```
DR EMBL; Z94753; CAB08160.1; -.
DR EMBL; AY011418; AAG47452.1; -.
DR PIR; S36149; S36149.
DR PDB; 1AW0; NMR; @=375-446.
DR PDB; 1KVI; NMR; A=1-79.
DR PDB; 1KVJ; NMR; A=1-79.
DR PDB; 1Q8L; NMR; A=163-246.
DR PDB; 2AW0; NMR; @=375-446.
DR Genew; HGNC:869; ATP7A.
DR MIM; 300011; -.

```

Cross references
(vers d'autres bases)

Keywords

```
KW Metal-binding; Phosphorylation; Polymorphism;
KW Repeat; Transmembrane.

```

```
FT DOMAIN 1 653 Cytoplasmic (Potential).
FT TRANSMEM 654 675 Potential.
FT DOMAIN 676 714 Extracellular (Potential).

```

Les données
brutes sont
mises en forme
par une
interface web

Features

```
FT /FTid=VAR_010007.
FT VARIANT 1362 1362 A -> V (in MD).
FT /FTid=VAR_010008.

```

```
FT TURN
FT HELIX 428 438
FT TURN 439 439
FT STRAND 441 444

```

Séquence

```
SQ SEQUENCE 1500 AA; 163334 MW; B58B24EABAD21627 CRC64;
MTRSMGVNSV TISVEGMTCN SCVWTHIEQOIGKINCSYLUK VSLLEKNATI IYDPKLQTPK
DDTSRNSPKL GLLDRIVNTS KASINSLSD KRSLNSVVTS EPDKHSLLVG DFREDDDTAL

```

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Exemples de bases spécialisées

- [PDB](#): structures 3D de protéines (X-rays ou NMR)
- [Prosite](#): motifs fonctionnels protéiques
- [Blocks](#): domaines protéiques conservés
- [Genatlas](#): carte du génome humain
- [OMIM](#): maladies héréditaires
- [Entrez Genome](#): cartes de génomes
- [Genes and Disease](#): maladies héréditaires
- [Medline](#): littérature
- [Enzyme](#): activité catalytique et cofacteurs
- [Witt](#), [Kegg](#), [PathDB](#): voies métaboliques

Plan

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Comment rechercher l'information?

- Deux portails généraux
 - SRS (EBI, UK et miroirs partout)
 - Entrez (NCBI, USA)
- Plusieurs interfaces spécifiques
 - Expasy (Suisse)
 - Infobiogen (France) (*fermé depuis juillet*)
 - GenomeNet (Japon)
 - PDB (USA)
 - CATH et SCOP (UK)

L'interface SRS

- Un portail général
 - ca. 300 bases de données
 - ca. 150 outils d'analyse
- Site principal :
 - <http://http://srs.ebi.ac.uk/>
- Liste de miroirs:
 - <http://downloads.lionbio.co.uk/publicsrs.html>
 - exemple: <http://srs.pasteur.fr/> (Institut Pasteur, France)
- Attention :
 - Tout n'est pas disponible dans tous les sites miroirs
 - Tous les miroirs n'ont pas la dernière version de l'interface

Une requête simple SRS

The screenshot displays the SRS web interface. At the top, there is a navigation bar with buttons for 'Quick Searches', 'Select Databanks', 'Query Form', 'Tools', 'Results', 'Projects', 'Custom Views', and 'Information'. A 'Help Center' link with a question mark icon is also present. Below the navigation bar, the 'SRS' logo is visible. A search box contains the text 'menkes', and a 'Quick Search' button is next to it. A 'Reset' button is located to the left of the search box. The main content area is divided into several sections:

- Search Options:** Contains instructions for selecting databanks and entering search terms. It includes buttons for 'Standard Query Form' and 'Extended Query Form'. A 'Browse Entries' button is also present.
- Available Databanks:** A list of databanks with expand/collapse controls and a 'Show databanks tooltips' checkbox. The 'Sequence libraries' section is expanded, showing a grid of checkboxes for various databases. 'UNIPROT_SWISSPROT' is checked. Other databases include EMBL, EMBLRELEASE, EMBLNEW, EMBLCON, EMBLWGS, EMBLTPA, UNIPROT, UNIPROT_TREMBL, UNIPROT_VARSPLIC, REFSEQ, REFSEQRELEASE, REFSEQNEW, REFSEQPROTEIN, REFSEQPROTEINRELEASE, REFSEQPROTEINNEW, and WORMPEP.
- Tips:** A section with a blue header containing a tip to bookmark a link to return to the project.

At the bottom of the interface, there is a footer with the text: 'SRS Release 7.1.3.1 Copyright © 1997-2003 LION bioscience AG. All Rights Reserved. Terms of Use Feedback'.

Liste de réponses et leurs détails

Help Center ?

Quick Searches Select Databanks Query Form Tools Results Projects Custom Views Information

SRS

Reset

Query "[libs={uniprot_swissprot pdb}-ALLTEXT:menkes*]" found 21 entries

Apply Options to:

selected results only
 unselected results only

Result Options

Show tools relevant to these results: **Tools**

Link to related information: **Link**

Save results: **Save**

Display Options

View results using: * Complete entries *

Show 30 results per page

Printer friendly view

Apply Display Options

- UNIPROT_SWISSPROT:AHM5_ARATH
- UNIPROT_SWISSPROT:AT7A_CRIGR
- UNIPROT_SWISSPROT:AT7A_HUMAN
- UNIPROT_SWISSPROT:AT7A_MOUSE
- UNIPROT_SWISSPROT:AT7A_RAT
- UNIPROT_SWISSPROT:AT7B_HUMAN
- UNIPROT_SWISSPROT:ATOX_HUMAN
- UNIPROT_SWISSPROT:ATPE_SYN7
- UNIPROT_SWISSPROT:ATSY_SYN7
- UNIPROT_SWISSPROT:YSY1_SYN7
- UNIPROT_SWISSPROT:YSY2_SYN7
- PDB:1AW0
- PDB:1FE0
- PDB:1FE4
- PDB:1FEE
- PDB:1KVI
- PDB:1KVJ
- PDB:1Q8L
- PDB:1S60
- PDB:1S6U
- PDB:2AW0

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 Université Paris Descartes

C2i Métiers de la
 2006/2007

Help Center ?

Quick Searches Select Databanks Query Form Tools Results Projects Custom Views Information

SRS

Reset

Query "([UNIPROT_SWISSPROT-ID:AT7A_HUMAN] | [UNIPROT_SWISSPROT-ID:AT7B_HUMAN])" found 2 entries

Apply Options to:

selected results only
 unselected results only

Result Options

Show tools relevant to these results: **Tools**

Link to related information: **Link**

Save results: **Save**

Display Options

- * Names only *
- * Complete entries *
- SeqSimpleView
- SangerUniProtFastaSeqs
- SangerFastaSeqs
- SVFastaSeqs
- PrimAccFastaSeqs
- FastaSeqs
- SwissView

Show 30 results per page

Printer friendly view

Apply Display Options

UNIPROT_SWISSPROT:AT7A_HUMAN

ID AT7A_HUMAN STANDARD; PRT; 1500 AA.
 AC Q04656; O00227; O00745; Q9BY8;
 DT 01-JUN-1994 (Rel. 29, Created)
 DT 01-FEB-1996 (Rel. 33, Last sequence update)
 DT 01-OCT-2004 (Rel. 45, Last annotation update)
 DE Copper-transporting ATPase 1 (EC 3.6.3.4) (Copper pump 1) (Menkes disease-associated protein).
 GN Name=ATP7A; Synonyms=MNK, MCL;
 OS Homo sapiens (Human).
 OC Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 OX NCBI_TaxID=9606;
 RN [1]
 RP SEQUENCE FROM N.A. (ISOFORM 4).
 RC TISSUE=Fibroblast;
 RX MEDLINE=93258410; PubMed=8490659;
 RA Vulpe C.D., Levinson B., Whitney S., Packman S., Gitschier J.;
 RT "Isolation of a candidate gene for Menkes disease and evidence that it encodes a copper-transporting ATPase."
 RL Nat. Genet. 3:7-13(1993).
 RN [2]
 RP ERRATUM.
 RA Vulpe C.D., Levinson B., Whitney S., Packman S., Gitschier J.;
 RL Nat. Genet. 3:273-273(1993).
 RN [3]
 RP SEQUENCE FROM N.A. (ISOFORM 4).
 RX MEDLINE=95331777; PubMed=7607665;
 RA Tuemer Z., Vural B., Toennesen T., Chelly J., Monaco A.P., Horn N.;
 RT "Characterization of the exon structure of the Menkes disease gene using vectorette PCR."
 RL Genomics 26:437-442(1995).
 RN [4]
 RP SEQUENCE OF 1-1447 FROM N.A. (ISOFORM 4).
 RX MEDLINE=96039257; PubMed=7490081;
 RA Dierick H.A., Ambrosini L., Spencer J., Glover T.W., Mercer J.F.B.;
 RT "Molecular structure of the Menkes disease gene (ATP7A)."
 RL Genomics 28:462-469(1995).
 RN [5]
 RP SEQUENCE OF 1-626 FROM N.A. (ISOFORM 4).
 RC TISSUE=Kidney;
 RX MEDLINE=93258397; PubMed=8490646.

Une requête SRS avancée

Help Center ?

Quick Searches | Select Databanks | Query Form | Tools | Results | Projects | Custom Views | Information

SRS

Reset search UNIPROT_SWISSPROT

Search Options	Fields you can search	Your search terms	Create a view
Combine search terms with: & (AND)	AllText		
Use wildcards <input checked="" type="checkbox"/>	ID		<input checked="" type="checkbox"/>
Get results of type: Entry	Accession Number		<input checked="" type="checkbox"/>
	Primary Accession Number		<input type="checkbox"/>
	Description	menkes	<input checked="" type="checkbox"/>
	GeneName		<input type="checkbox"/>
	Keywords		<input checked="" type="checkbox"/>
	Entry Creation Date	select 1 Jan YYYY 1 Jan YYYY	<input type="checkbox"/>
	LastSequenceUpdate	>= <=	<input type="checkbox"/>
	LastAnnotationUpdate	>= <=	<input type="checkbox"/>
	Organism Name	homo sapiens	<input type="checkbox"/>
	Taxon		<input type="checkbox"/>
	NCBI TaxId	>= <=	<input type="checkbox"/>
	Organelle		<input type="checkbox"/>
	ProteinID		<input type="checkbox"/>
	Checksum		<input type="checkbox"/>
	DbName	aarhus/ghent-2dpage agd anu-2dpage compluyeast-2dpage dictybase echobase eco2dbase	<input type="checkbox"/>
	<input checked="" type="radio"/> or		
	<input type="radio"/> and		
	DBxref		<input type="checkbox"/>
	Sequence Length	>= <=	<input checked="" type="checkbox"/>

Sortie sélective avec mise en forme

Help Center ?

Quick Searches Select Databanks Query Form Tools Results Projects Custom Views Information

SRS

Reset

Query "[uniprot_swissprot-Description:menkes*] & (((uniprot_swissprot-Organism:homo*) & [uniprot_swissprot-Organism:sapiens*]) | [uniprot_swissprot-Organism:homo sapiens*])" found 1 entries

UNIPROT_SWISSPROT	ID	AccNumber	Description	Keywords	SeqLength
UNIPROT_SWISSPROT:AT7A_HUMAN	AT7A_HUMAN	Q04656 O00227 O00745 Q9BY8	Copper-transporting ATPase 1 (EC 3.6.3.4) (Copper pump 1) (Menkes disease-associated protein).	3D-structure; Alternative splicing; ATP-binding; Copper; Copper transport; Disease mutation; Golgi stack; Hydrolase; Magnesium; Metal-binding; Multigene family; Phosphorylation; Polymorphism; Repeat; Transmembrane.	1500

Apply Options to:
 selected results only
 unselected results only

Result Options
Show tools relevant to these

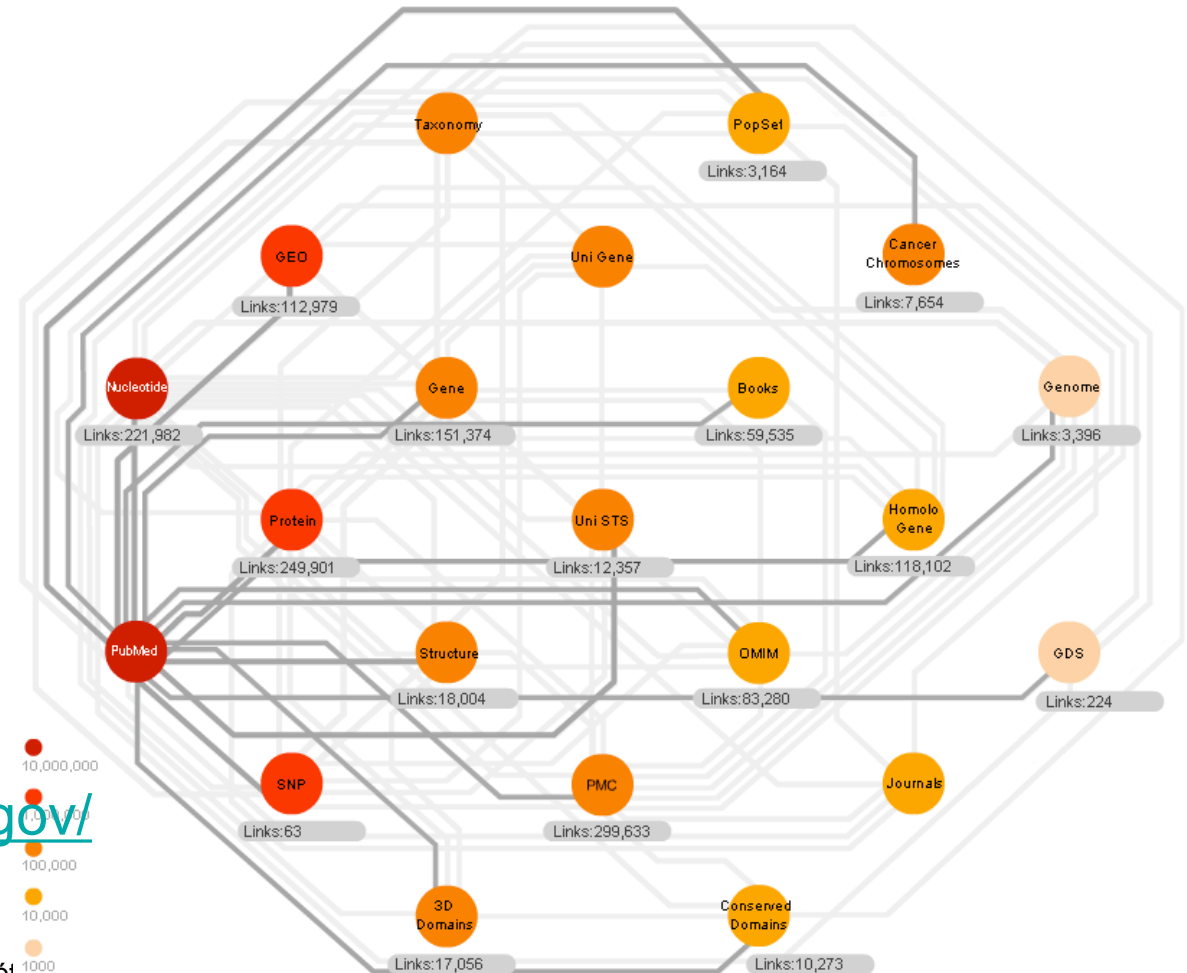
Lien vers l'enregistrement complet

Plan

- Introduction aux bases de données
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L'interface Entrez

- Consultation de bases de données interconnectées
 - Littérature
 - Structures
 - Séquences
 - Génomes
 - Maladies
 - Taxonomie
 - ...



- Mis en place par la NLM
 - <http://www.ncbi.nlm.nih.gov/>

Une recherche simple avec Entrez

- Choix de la base et introduction des mots clés

The screenshot shows the NCBI Entrez search interface. At the top, the NCBI logo and the text "National Center for Biotechnology Information" are visible, along with "National Library of Medicine" and "National Institutes of Health". Below this is a navigation bar with links to "PubMed", "Databases", "BLAST", "OMIM", "Books", "TaxBrowser", and "Structure". The "Databases" link is highlighted with a red arrow. Below the navigation bar is a search form with a dropdown menu for database selection, currently set to "Protein". A red arrow points to the "Protein" option in the dropdown. To the right of the dropdown is a search input field containing the text "menkes", with another red arrow pointing to it. A "Go" button is located to the right of the search input field. Below the search form, there is a "Hot Spots" section with various links and a "Genome Association" section with a link to the WGA resource page.

Liste de résultats liés aux autres bases

The screenshot shows the NCBI Entrez Protein search interface. The search query is 'menkes' and the results are displayed in a list format. The first four results are visible, each with a checkbox, a link to the accession number, a 'Reports' link, a title, and a GenBank accession number. The results are:

- 1: [Q04656](#) Reports Copper-transporting ATPase 1 (Copper pump 1) (Menkes disease-associated protein) gi|1351993|sp|Q04656|ATP7A_HUMAN[1351993] BLink, Conserved Domains, Links
- 2: [NP_000043](#) Reports ATPase, Cu++ transporting, alpha polypeptide [Homo sapiens] gi|53986563|ref|NP_000043.2|[53986563] BLink, Conserved Domains, Links
- 3: [Q64430](#) Reports Copper-transporting ATPase 1 (Copper pump 1) (Menkes disease-associated protein homolog) gi|12644462|sp|Q64430|ATP7A_MOUSE[12644462] BLink, Conserved Domains, Links
- 4: [2GA7A](#) Reports Chain A, Solution Structure Of The Copper(I) Form Of The Third Metal- Binding Domain Of Atp7a Protein (Menkes Disease Protein) gi|112490719|pdb|2GA7A[112490719] BLink, Conserved Domains, Links

The interface also shows a search bar with 'Protein' selected, a 'Go' button, and a 'Save Search' link. The results are sorted by Relevance and displayed in Summary view. The page number is 1 of 5.

Le détail d'une fiche de protéine ou de gène est le même que pour SRS

Lien vers la base génomique

NCBI Entrez Gene

All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy Books

Search Gene for [Go] [Clear]

Limits Preview/Index History Clipboard Details

Display Full Report Show 5 Send to

All: 1 Current Only: 1 Genes Genomes: 1 SNP GeneView: 1

1: ATP7A ATPase, Cu++ transporting, alpha polypeptide (Menkes syndrome) [*Homo sapiens*] updated 18-Sep-2006

GeneID: 538

Entrez Gene Home

Summary

Official Symbol	ATP7A
Official Full Name	ATPase, Cu++ transporting, alpha polypeptide (Menkes syndrome)
Primary source	HGNC:869
Locus tag	RP3-465G10.1
See related	HPRD:02054 ; MIM:300011
Gene type	protein coding
RefSeq status	Validated
Organism	Homo sapiens
Lineage	<i>Eukaryota</i> ; <i>Metazoa</i> ; <i>Chordata</i> ; <i>Craniata</i> ; <i>Vertebrata</i> ; <i>Euteleostomi</i> ; <i>Mammalia</i> ; <i>Eutheria</i> ; <i>Eurarchontoglires</i> ; <i>Primates</i> ; <i>Haplorrhini</i> ; <i>Catarrhini</i> ; <i>Hominidae</i> ; <i>Homo</i>
Also known as	MK; MNK; OHS

Table Of Contents

- Summary
- Genomic regions, transcripts, and products
- Genomic context
- Bibliography
- Interactions
- General gene information
- General protein information
- Reference Sequences
- Related Sequences
- Additional Links

Links

- Books
- Conserved Domains
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- Evidence Viewer

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

NC_000023.9

[77152855] [77192095]

NM_000052.2 5' 3' NP_000043

■ - coding region ■ - untranslated region

Lien vers la carte des chromosomes

The screenshot displays the NCBI Map Viewer interface for the *Homo sapiens* Build 36.2. The main content shows a genomic map of a region on chromosome 22, specifically between 77,035K and 77,210K bp. The map includes tracks for various UniGene clusters (Bt, Gga, Hn, Rn, Ssc, Hs) and a gene model for *ATP7A*. The *ATP7A* gene is highlighted in a pink box, indicating its location within the region. Other genes shown include *COX7B*, *LOC642277*, and *PGAM4*. The interface also features a search bar, navigation buttons (Zoom in/out), and a chromosome ideogram on the left side.

NCBI
NCBI Map Viewer

PubMed Entrez BLAST OMIM Taxonomy Structure

Search Find Find in This View Advanced Search

Human genome overview page (Build 36.2)
Human genome overview page (Build 35.1)
Map Viewer Home

Map Viewer Help
Human Maps Help
FTP
Data As Table View
Maps & Options
Compress Map
Region Shown:
77,035K
77,210K Go

out
zoom
in

You are here:
Ideogram
Xp22.3
Xp22.2
Xp22.1
Xp21
Xp11.3
Xp11.2
Xp11.1
Xq13
Xq21
Xq22
Xq23
Xq24
Xq25
Xq26
Xq27
Xq28

default
master

Homo sapiens Build 36.2 (Current)
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 MT
Query: 538[gene_id] [clear]

Master Map: Genes On Sequence
Region Displayed: 77,035K-77,210K bp

Summary of Maps
Download/View Sequence/Evidence

Symbol
DKFZp564K142 + sv pi
COX7B + OMIM HGNC sv pi
LOC642277 + sv pi
PGAM4 + OMIM HGNC sv pi
ATP7A + OMIM HGNC sv pi

Lien vers la base des maladies héréditaires

NCBI
MIM #309400
Description
Clinical Features
Biochemical Features
Other Features
Cytogenetics
Mapping
Molecular Genetics
Diagnosis
Clinical Management
Population Genetics
Animal Model
See Also
References
Contributors
Creation Date
Edit History

Entrez Gene
Nomenclature
R RefSeq
C GenBank
P Protein
U UniGene

LinkOut
CCR

OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University

All Databases PubMed Nucleotide Protein Genome Structure PMC
Search OMIM for Go Clear
Limits Preview/Index History Clipboard Details
Display Detailed Show 20 Send to
All: 1 OMIM dbSNP: 0 OMIM UniSTS: 1

#309400
MENKES DISEASE

Alternative titles; symbols
MK; MNK
MENKEA SYNDROME
KINKY HAIR DISEASE
STEELY HAIR DISEASE
COPPER TRANSPORT DISEASE

Gene map locus [Xq12-q13](#)

TEXT

DESCRIPTION

A number sign (#) is used with this entry because of evidence that Menkes disease is caused by mutation in the g Cu(2+)-transporting ATPase, alpha polypeptide ([300011](#)). The occipital horn syndrome ([304150](#)) is caused by m

CLINICAL FEATURES

In a family of English-Irish descent living in New York, [Menkes et al. \(1962\)](#) described an X-linked recessive dis retardation in growth, peculiar hair, and focal cerebral and cerebellar degeneration. Severe neurologic impairer birth and progressed rapidly to decerebration. Five males were affected but the gene could by inference be identi to grow brought the affected infants to medical attention at the age of a few weeks and death occurred in the first

Lien vers la base de taxonomie

NCBI Taxonomy Browser

Entrez PubMed Nucleotide Protein Genome Structure PMC Taxonomy Books

Search for as complete name lock

Display levels using filter:

Homo sapiens

Taxonomy ID: 9606
 Genbank common name: **human**
 Rank: species
 Genetic code: [Translation table 1 \(Standard\)](#)
 Mitochondrial genetic code: [Translation table 2 \(Vertebrate Mitochondrial\)](#)
 Other names:
 common name: **man**

[Lineage\(full \)](#)
[cellular organisms](#); [Eukaryota](#); [Fungi/Metazoa group](#); [Metazoa](#); [Eumetazoa](#); [Bilateria](#);
[Coelomata](#); [Deuterostomia](#); [Chordata](#); [Craniata](#); [Vertebrata](#); [Gnathostomata](#);
[Teleostomi](#); [Euteleostomi](#); [Sarcopterygii](#); [Tetrapoda](#); [Amniota](#); [Mammalia](#); [Theria](#);
[Eutheria](#); [Euarchontoglires](#); [Primates](#); [Haplorrhini](#); [Simiiformes](#); [Catarrhini](#);
[Hominoidea](#); [Hominidae](#); [Homo/Pan/Gorilla group](#); [Homo](#)

Entrez records		
Database name	Subtree links	Direct links
Nucleotide	11,510,645	11,509,310
Protein	311,857	311,856
Structure	8,490	8,490
Genome Sequences	51	51
Genome Projects	1	1
Popset	20,799	20,799
SNP	12,702,095	12,702,095
3D Domains	31,862	31,862
Domains	25	25
GEO Datasets	2,899	2,899
GEO Expressions	81,920	81,920
UniGene	86,804	86,804
UniSTS	322,092	322,092
PubMed Central	3,094	3,094
Gene	38,685	38,685
HomoloGene	19,180	19,180
Taxonomy	2	1

Genome Information

[See the NCBI Genome homepage](#)
[Go to NCBI genomic BLAST page for Homo sapiens](#)

Genome view: 24 chromosomes																								
Names	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	X	Y

[See the Mitochondrion Genome](#)
[See the TRACE Assembly](#)

Plan

- Introduction aux bases de données
- Données disponibles
 - Bases nucléiques généralistes
 - Bases protéiques généralistes
 - Bases spécialisées
- Recherche d'information
 - L'interface SRS
 - L'interface Entrez
 - Interfaces spécifiques




Données structurales : PDB



















The screenshot displays the RCSB PDB SRS interface. At the top, there is a navigation bar with buttons for 'Quick Searches', 'Select Databanks', 'Query Form', 'Tools', 'Results', 'Projects', 'Custom Views', and 'Information'. Below this, the 'SRS' section includes a 'Reset' button, a search input field containing 'menkes', and a 'Quick Search' button. The 'Search Options' sidebar on the left provides instructions for selecting databanks and search terms, with buttons for 'Standard Query Form' and 'Extended Query Form'. The 'Available Databanks' section lists various databases under 'Sequence libraries' and 'Protein Sequence libraries (old names)'. The 'Search the Archive' section features a search input field with 'menkes', radio buttons for 'PDB ID', 'Authors', and 'Full Text Search', and checkboxes for 'match exact word' and 'remove similar sequences'. The 'News' section includes a 'Complete News Newsletter' link and a news item dated '21-Sep-2004' about the 'RCSB PDB Art of Science Exhibit'. The 'PDB Mirror' section on the right lists various mirror sites. Red annotations with circles highlight the search input field and the 'Search the Archive' section, with text indicating 'Recherche via l'interface SRS' and 'Recherche via l'interface PDB'.

Une liste de structures NMR et cristallographie de rayons-X

Your query found 10 structures in the current PDB release and you have selected 0 structures so far. You can select specific structures by clicking on the checkbox next to their id. If you do not select any structures, certain options will default to all structures. To examine an individual structure select the Explore link!

Pull down to select option:

KEY:  = Download compressed (GNU zipped) PDB file  = View PDB file  = Structure viewing options

<input type="checkbox"/> 1AW0	   Deposited: 08-Oct-1997 Exp. Method: NMR, 20 Structures	{ EXPLORE }
Title	Fourth Metal-Binding Domain Of The Menkes Copper-Transporting ATPase, NMR, 20 Structures	
Classification	Hydrolase	
Compound	Mol_Id: 1; Molecule: Menkes Copper-Transporting ATPase; Chain: Null; Fragment: Fourth Metal-Binding Domain; Ec: 3.6.1.36; Engineered: Yes; Other_Details: Reduced Apo State	
<input type="checkbox"/> 1FEO	   Deposited: 20-Jul-2000 Exp. Method: X-ray Diffraction Resolution: 1.75 Å	{ EXPLORE }
Title	Crystal Structure Of Cadmium-Hah1	
Classification	Metal Transport	
Compound	Mol_Id: 1; Molecule: Copper Transport Protein Atox1; Chain: A, B; Engineered: Yes	
<input checked="" type="checkbox"/> 1FE4	   Deposited: 20-Jul-2000 Exp. Method: X-ray Diffraction Resolution: 1.75 Å	{ EXPLORE }
Title	Crystal Structure Of Mercury-Hah1	
Classification	Metal Transport	
Compound	Mol_Id: 1; Molecule: Copper Transport Protein Atox1; Chain: A, B; Engineered: Yes	
<input type="checkbox"/> 1FEE	   Deposited: 21-Jul-2000 Exp. Method: X-ray Diffraction Resolution: 1.80 Å	{ EXPLORE }
Title	Crystal Structure Of Copper-Hah1	
Classification	Transport Protein	
Compound	Mol_Id: 1; Molecule: Copper Transport Protein Atox1; Chain: A, B; Synonym: Copper Chaperone, Metal Transport Protein Atx1; Engineered: Yes	
<input type="checkbox"/> 1KVI	   Deposited: 26-Jan-2002 Exp. Method: NMR, 10 Structures	{ EXPLORE }
Title	Solution Structure Of The Reduced Form Of The First Heavy Metal Binding Motif Of The Menkes Protein	
Classification	Hydrolase	
Compound	Mol_Id: 1; Molecule: Copper-Transporting ATPase 1; Chain: A; Fragment: Residues 1-79; Synonym: Menkes Disease-Associated Protein; Ec: 3.6.3.4; Engineered: Yes	
<input type="checkbox"/> 1KVI	   Deposited: 26-Jan-2002 Exp. Method: NMR, 10 Structures	{ EXPLORE }

L'interface web PDB pour explorer la structure



Structure Explorer - 1FE4



Try the Structure Explorer page for [1FE4](#) from the new, reengineered RCSB PDB Web site!

< [Print] >
← 🔍 ? 🏠 📧 💡

Summary Information

Summary Information

[View Structure](#)

[Download/Display File](#)

[Structural Neighbors](#)

[Geometry](#)

[Other Sources](#)

[Sequence Details](#)

Explore

[SearchLite](#) [SearchFields](#)

Title: Crystal Structure Of Mercury-Hah1

Compound: **Mol_Id:** 1; **Molecule:** Copper Transport Protein Atox1; **Chain:** A, B; **Engineered:** Yes

Authors: A. K. Wernimont, D. L. Huffman, A. L. Lamb, T. V. O'Halloran, A. C. Rosenzweig

Exp. Method: X-ray Diffraction

Classification: Metal Transport

Source: Homo sapiens

Primary Citation: **Wernimont, A. K., Huffman, D. L., Lamb, A. L., O'Halloran, T. V., Rosenzweig, A. C.:** Structural Basis for Metal Ion Transfer by the Copper Chaperone for the Menkes and Wilson Disease Proteins *Nat.Struct.Biol.* 7 pp. 766 (2000)

Deposition Date: 20-Jul-2000 *Release Date:* 24-Jan-2001

Resolution [Å]: 1.75 *R-Value:* 0.204

Space Group: P 6₅

Unit Cell: dim [Å]: a 79.95 b 79.95 c 55.61
angles [°]: alpha 90.00 beta 90.00 gamma 120.00

Polymer Chains: A, B *Residues:* 136

Atoms: 1187

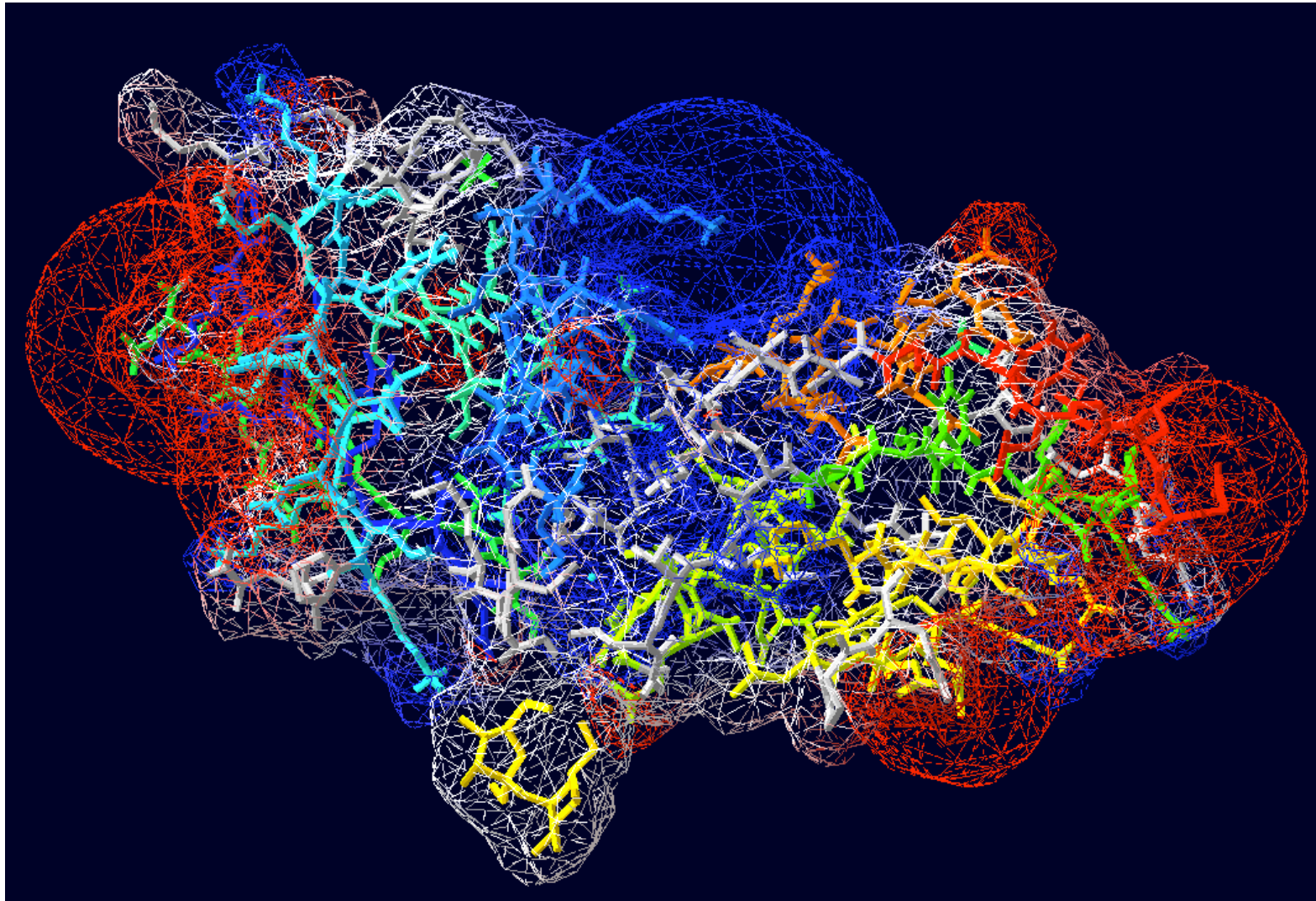
Chemical Components: ("HET" groups)

ID <i>(needs Rasmol)</i>	Name	Formula	Retrieve All PDB IDs Containing
HG	MERCURY (II) ION	Hg	HG
IUM	URANYL (VI) ION	O ₂ U	IUM
SO4	SULFATE ION	2(O ₄ S)	SO4
SUC	SUCROSE	C ₁₂ H ₂₂ O ₁₁	SUC

CATH: [Structural Classification](#)

SCOP: [Structural Classification](#)

Coordonnées 3D disponibles



Outils d'analyse géométrique



Structure Explorer - 1FE4

Title Crystal Structure Of Mercury-Hah1
Classification Metal Transport
Compound Mol_Id: 1; Molecule: Copper Transport Protein Atox1; Chain: A, B; Engineered: Yes
Exp. Method X-ray Diffraction

💡 Try the Structure Explorer page for **1FE4** from the new, reengineered RCSB PDB Web site!

⏪ 📄 ⏩

[Summary Information](#)

[View Structure](#)

[Download/Display File](#)

[Structural Neighbors](#)

[Geometry](#)

[Other Sources](#)

[Sequence Details](#)

Explore

[SearchLite](#) [SearchFields](#)

Table Format:

- [Dihedral Angles](#)
- [Common Bond Angles](#)
- [Bond Lengths](#)

Graphical:

- [Ramachandran Plot](#)
- [Fold Deviation Score](#) (requires Rasmol)

Links:

- [WHAT CHECK Structure Analysis](#) (*PDBREPORT*)
- [Structure Summary](#) (*PROCHECK*)
- [Motif Summary](#) (*PROMOTIF*)
- [Identification of Protein Pockets & Cavities](#) (*castP*)

Geometric values are compared to reference data (high resolution value is assigned to each residue as a multiple of the standard de



Title Crystal Structure Of Mercury-Hah1
Classification Metal Transport
Compound Mol_Id: 1; Molecule: Copper Transport Protein Atox1; Chain: A, B; Engineered: Yes
Exp. Method X-ray Diffraction

Structure Explorer - 1FE4

💡 Try the Structure Explorer page for **1FE4** from the new, reengineered RCSB PDB Web site!

⏪ 📄 ⏩
Ramachandran Plot

[Summary Information](#)

[View Structure](#)

[Download/Display File](#)

[Structural Neighbors](#)

[Geometry](#)

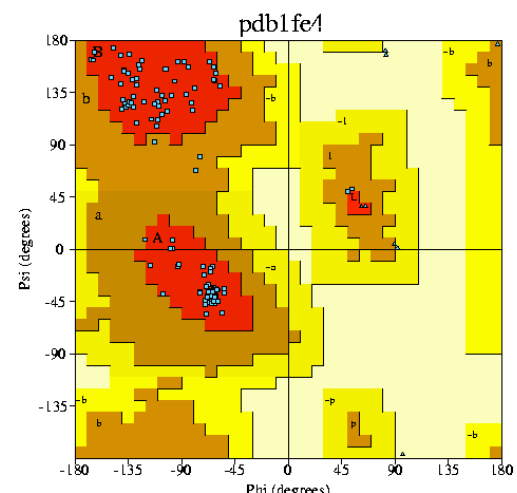
[Other Sources](#)

[Sequence Details](#)

Explore

[SearchLite](#) [SearchFields](#)

• Ramachandran plot (courtesy of PDBsum):



pdb1fe1

Psi (degrees)

Phi (degrees)

• [PROCHECK summary for 1FE4](#) (courtesy of PDBsum):

- Interactive Ramachandran Plot (courtesy of Steve Millener, former Chino and Java)

Liens vers des bases de classification structurale



Structure Explorer - 1FE4

Title Crystal Structure Of Mercury-Hahl
Classification Metal Transport
Compound Mol_Id: 1; Molecule: Copper Transport Protein Atox1; Chain: A, B; Engineered: Yes
Exp. Method X-ray Diffraction



[Try](#) the Structure Explorer page for [1FE4](#) from the new, reengineered RCSB PDB Web site!

Structural Neighbors

- [CATH](#) **Class, Architecture, Topology and Homologous superfamily** - a hierarchical classification of protein domain structures [\[top\]](#)
University College London ([UCL](#))
Features: Complete PDB, fold classification by domain, links to other information
Reference: Orengo, Michie, Jones, Jones, Swindells and Thornton (1997) *Structure* **5(8)** 1093-1108
- [CE](#) **Combinatorial Extension of the optimal path** [\[top\]](#)
Research Collaboratory for Structural Bioinformatics ([RCSB](#))
Features: Complete PDB and representative structure comparison, structure alignments, structure superposition tool
Reference: Shindyalov and Bourne (1998) *Protein Engineering* **11(9)** 739-747
- [FSSP](#) **Fold classification based on Structure-Structure alignment of Proteins** [\[top\]](#)
European Bioinformatics Institute ([EBI](#))
Features: Complete PDB, fold tree, domain dictionary, sequence neighbors, structure superposition
Reference: Holm and Sander (1998) *Nucl. Acids Res.* **26** 316-319
- [SCOP](#) **Structural Classification Of Proteins** [\[top\]](#)
MRC Laboratory of Molecular Biology and Centre for Protein Engineering
Features: Augmented manual classification, class, fold, superfamily and family classification
Reference: Murzin, Brenner, Hubbard and Chothia (1995) *J. Mol. Biol.* **247** 536-540
- [VAST](#) **Vector Alignment Search Tool** [\[top\]](#)
National Center for Biotechnology Information ([NCBI](#))
Features: Complete PDB and representative structure comparison, structure alignments, structure superposition tool
Reference: Gibrat, Madej and Bryant (1996) *Current Opinion in Structural Biology* **6** 377-385

CATH : classification structurale

CATH DHS Gene3D Impala FTP Internal

Home > Top View



CATH PDB code search

Search results for **1fe4**

Found 2 domains and 0 preliminary data entries.

[1fe4 \[PDBsum\]](#)

CATH Classification Entries

Domain	CATH code	Length	Image
1fe4A0	3.30.70.100	68	
1fe4B0	3.30.70.100	68	

CATH
Protein Structure Classification

CATH DHS Gene3D Impala FTP Internal

Home > Top > Class3 > 30 > 70 > 100 > 5 > 2 > 1 > 1fe4A0 View this page as XML

Search

PDB Code
 CATH Code
 General Text


Goto

[SSAP Server](#)
[GRATH Server](#)
[DHS](#)
[Gene3D](#)

Navigation

[Home](#)
[Top of hierarchy](#)

CATH Domain 1fe4A0



1fe4A0

Classification

- **Class** 3
- Alpha Beta
- **Architecture** 3.30
- 2-Layer Sandwich
- **Topology** 3.30.70
- Alpha-Beta Plaits
- **Homologous Superfamily** 3.30.70.100
- HYDROLASE(ACTING ON ACID ANHYDRIDES)
- **Sequence Family (S35)** 3.30.70.100.5
- METAL TRANSPORT
- **Non-identical (S95)** 3.30.70.100.5.2
- METAL TRANSPORT
- **Identical (S100)** 3.30.70.100.5.2.1
- METAL TRANSPORT

PDB Information

PDB Code	1fe4
PDB Header	Copper transport protein atox1. Chain: a, b. Engineered: yes
PDB Source	Homo sapiens. Human. Organ: liver. Expressed in: escherichia coli.

Domain Information

Domain Sequence	MPKHEFSVDMTCGGCAEAVSRVLNKLGGVKYDIDLPNKK VCIESEHSMDTLLATLKKTKTVSYLGL
------------------------	--

CATH considers structural domains as semi-independent folding units. It is quite common that a structural domain is made up of more than one sequence segment (*i.e.* non-sequential stretch of peptide). The table below provides information for each of the segments that make up the structural domain.

Segment Number	Segment Length	PDB Start	PDB Stop
1	68	1	68

Exemple : La base Enzyme

EMBL-EBI European Bioinformatics Institute

Quick Search Library Page Query Form Tools Results Projects

Reset Entry 1 of 4 from Query 3 Next Entry

Entry Information
Entry from: [LENZYME](#)

Entry Options
Link to related information: [Link](#)
Save entry: [Save](#)
View: [Printer Friendly](#)

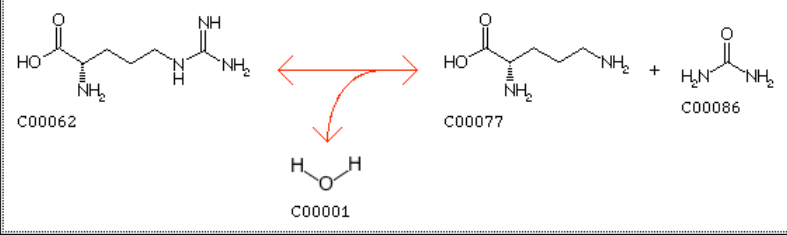
LENZYME:3.5.3.1

ECNumber	EC 3.5.3.1 Enzyme
Source of data	Ligand database
Name	arginase; arginine amidinase; canavanase
Class	Hydrolases acting on carbon-nitrogen bonds
Systematic name	L-arginine amidinohydrolase
Reaction	Arginine + H2O = L-ornithine + urea
Substrate	L-arginine [CPD:C00062] H2O [CPD:C00001]
Product	L-ornithine [CPD:C00077] urea [CPD:C00086]
Comment	Also hydrolyses alpha-N-substituted guanidines and carbonyl groups of amino groups
References	1 Bach, S.J. and Killip, J.D. Study of the kinetic properties of arginase from sheep liver. <i>Biochim. Biophys. Acta</i> 1973; 34: 148-152. 2 Cabello, J., Basilio, C. and Prats, J. Purification, crystallization and X-ray crystallographic study of erythrocyte- and liver arginase from sheep. <i>Biochim. Biophys. Acta</i> 1973; 34: 148-152. 3 Dumitru, I.F. Study of L-arginase from sheep erythrocytes. Purification, crystallization and X-ray crystallographic study. <i>Vitamin. Enzymol.</i> 27 (1973) 1-10. 4 Greenberg, D.M. Arginase, in: <i>Enzymes</i> , 3rd ed., Vol. 6, Academic Press, New York, 1971, pp. 1-10.

KEGG ENZYME: 3.5.3.1

Entry	EC 3.5.3.1	Enzyme
Name	arginase; arginine amidinase; canavanase; L-arginase; arginine transamidinase	
Class	Hydrolases Acting on carbon-nitrogen bonds, other than peptide bonds In linear amidines	
Synonym	L-arginine amidinohydrolase	
Reaction (IUBMB)	L-arginine + H2O = L-ornithine + urea [RN:R00551]	

KEGG REACTION: R00551 [Help](#)

Entry	R00551	Reaction
Name	L-Arginine amidinohydrolase	
Definition	L-Arginine + H2O <=> L-Ornithine + Urea	
Equation	C00062 + C00001 <=> C00077 + C00086	
Chemical		
RPair	RP: A00066 C00062_C00077 main RP: A00979 C00062_C00086 main	
Pathway	PATH: rn00220 Urea cycle and metabolism of amino groups PATH: rn00330 Arginine and proline metabolism	
Enzyme	3.5.3.1	
Ortholog	KO: K01476 arginase	
LinkDB	All DBs	

7788(LOC3977)

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