

Bioinformatique

<http://www.luminy.univ-mrs.fr>

- Qu'est ce?

Ce que ce n'est pas...

Définitions!

Exemples

Science / Technologie ?

(créateurs / utilisateurs?)

Bref historique

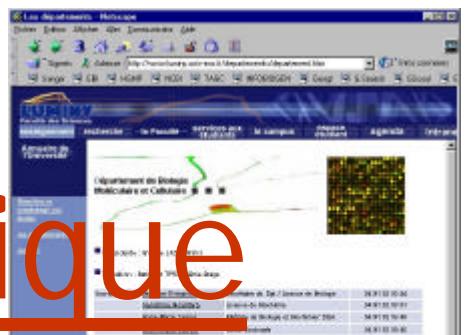
Catégories

- A quoi ça ressemble?

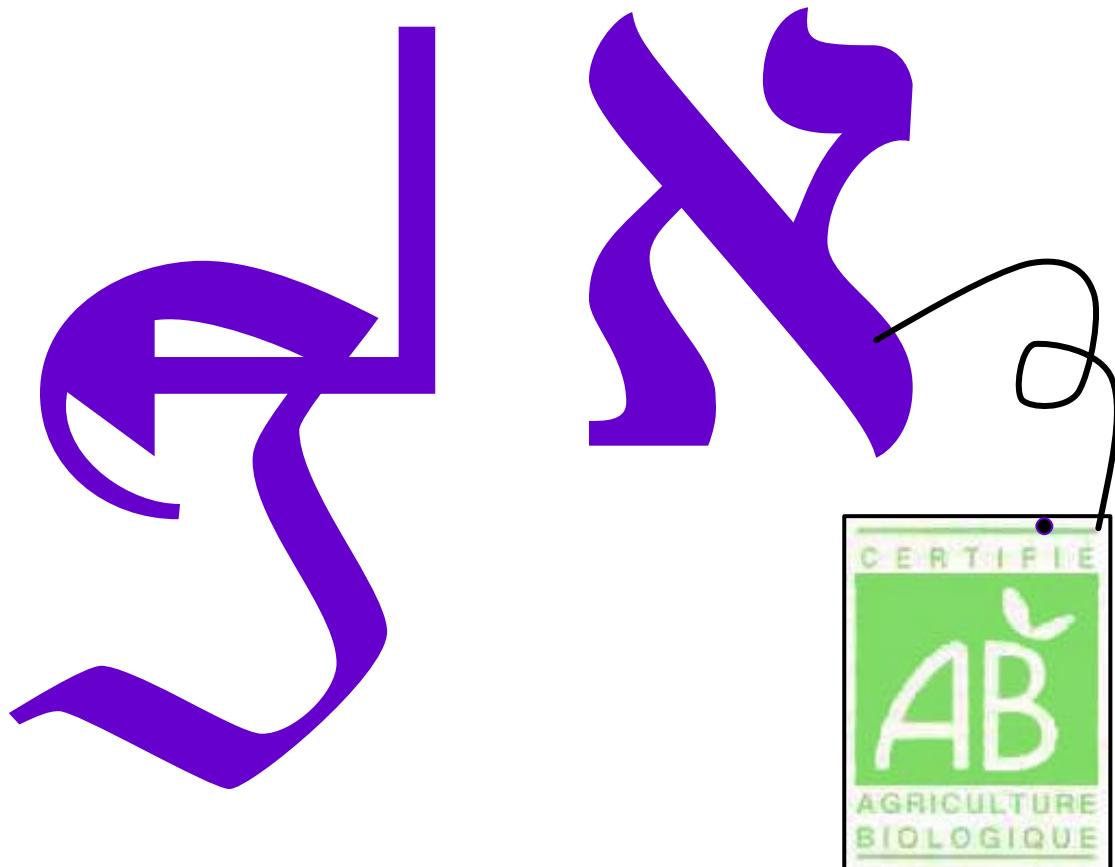
lignes de commande

logiciels interactifs

interfaces Internet



Informatique «Bio»





Enki Bilal (1977) Mémoires d'Outre Espace
éditions *Les humanoïdes Associés*

Bioinformatiques

Bioinfo de labo

support au clonage,
séquençage,
& PCR...

Analyse séquence

identification gènes,
comparaisons de
séquences,
prédiction motifs...

Phylogénie

évolution à
l'échelle
moléculaire...

Structure des protéines

calcul,
visualisation,
prédiction...

Liaison génétique

gènes candidats
de maladies
génétiques...

Génomique fonctionnelle

transcriptome,
protéome,
interactome...

Bref historique

1970 Needlman & Wunsch

Alignement optimal de 2 séquences

1974 Chou & Fasman

Prédiction structure 11 protéines

1978 Dayhoff *et. al.*

Matrice de substitution (PAM)

1980 EMBL data library

Banque internationale de séquence

1981 Smith & Waterman

Similarités de séquences dans les banques

1981 Felsenstein *et. al.*

Arbres phylogénét. par comparaison de séq. (phylip)

1984 Devereux *et. al.*

Logiciel d'analyse de séquence (UW GCG)

1987 Feng & Doolittle

Alignement multiple progressif de séquences

1988 Bairoch *et. al.*

Banque de signatures protéiques (PROSITE)

1988 Pearson & Lipman

Sim. de séq. dans les banques (FASTA)

Bref historique (suite)

1990 Atschul *et. al.*

Sim. de séq. dans les banques (BLAST)

1991 Bowie *et. al.*

Prédiction struct. 111 protéines

1993 Borodovsky *et. al.*

Prédiction gènes génomes bactériens (GeneMark)

1995 Venter *et. al.*

Séquençage 'shotgun' génome Haemophilus 1.8Mb

1997 Sonnhammer *et. al.*

Banque de domaines protéiques (PFam)

1997 Burge *et. al.*

Prédiction gènes génomes eucaryotes (GENSCAN)

1998 Eisen *et. al.*

Analyse des profils d'expression (CLUSTER)

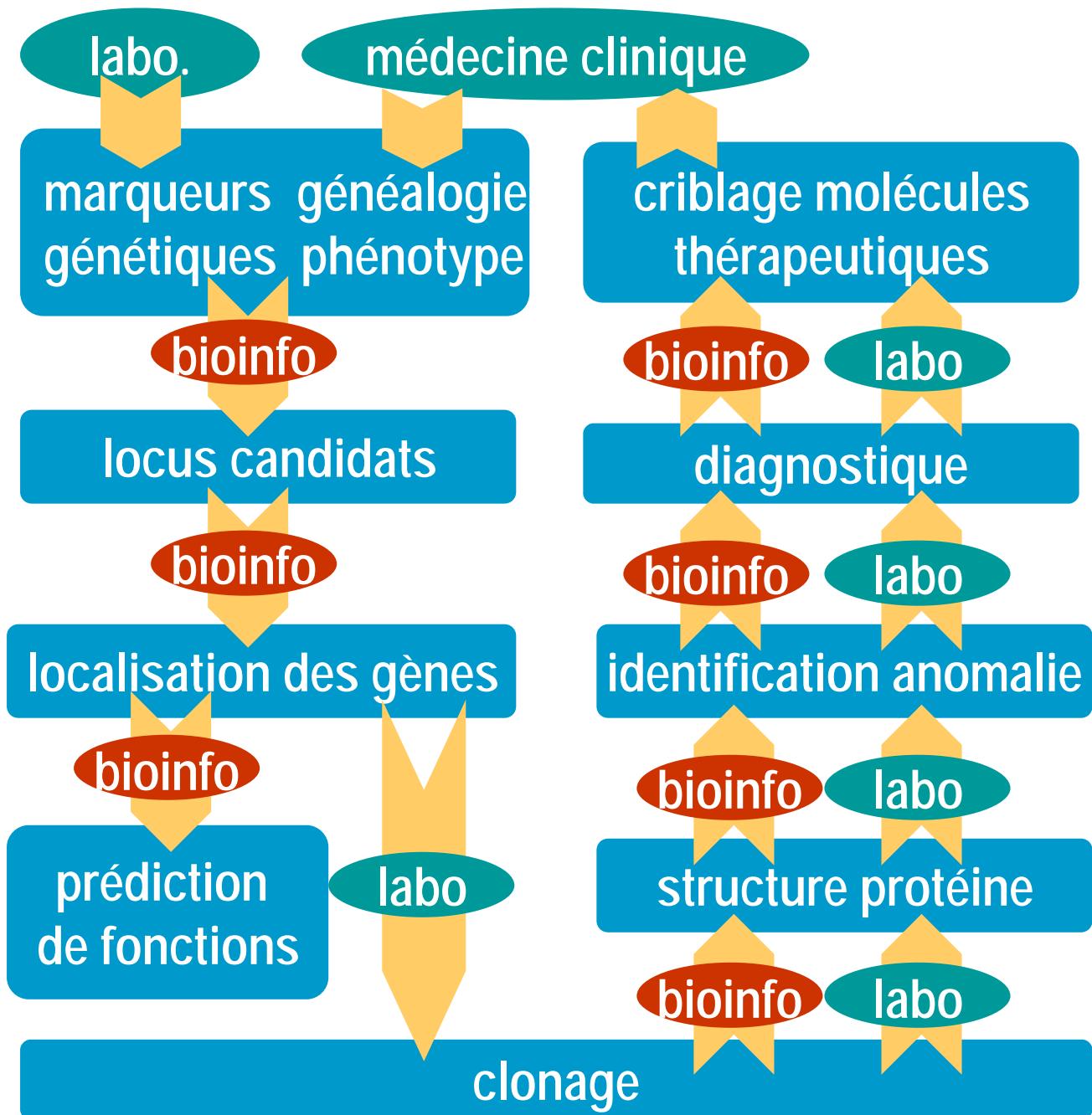
2000 Venter *et. al.*

'Shotgun' génome complet Drosophile 120 Mb

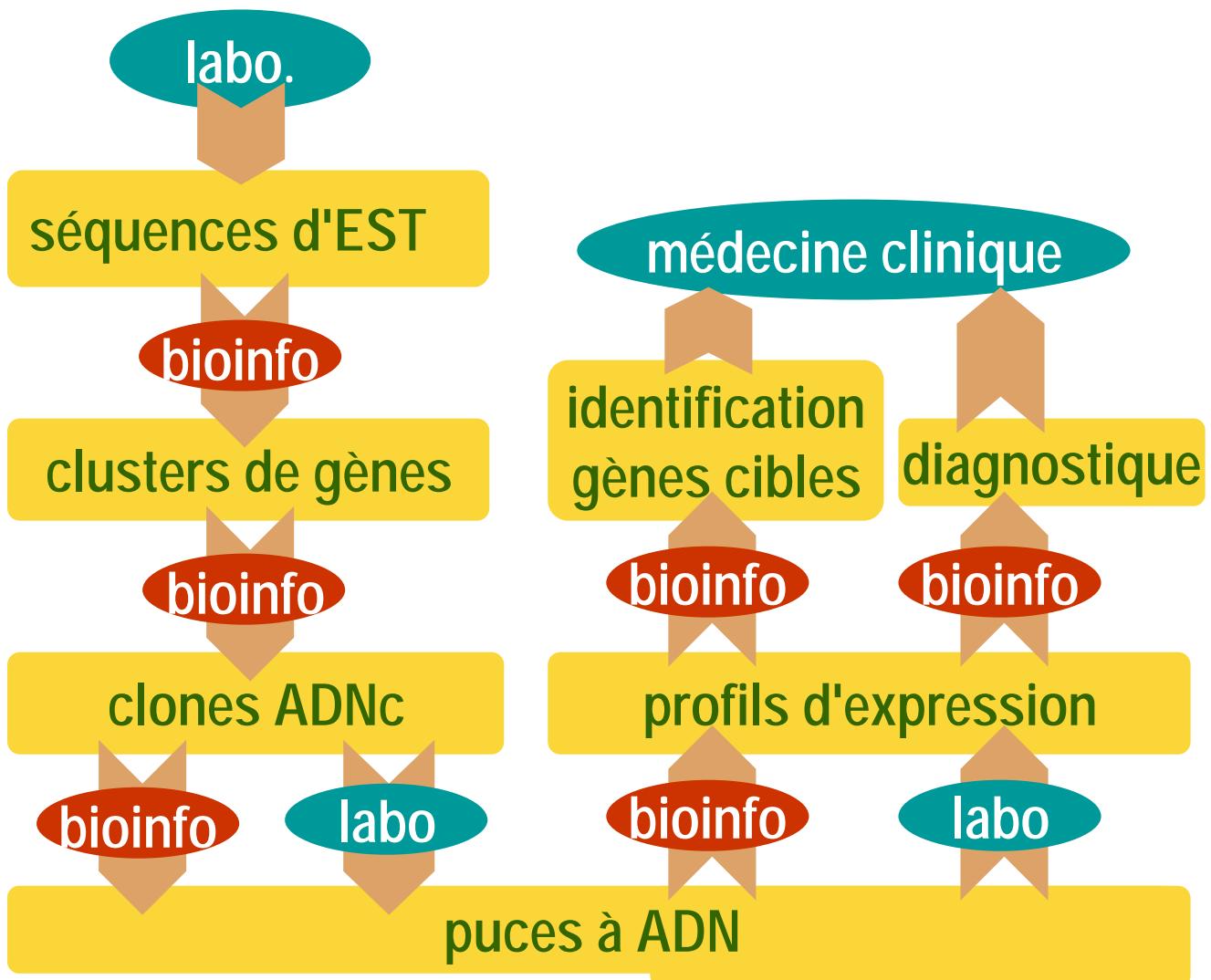
2001 Venter *et. al.*

'Shotgun' génome complet Humain 3000 Mb

Exemple d 'application 1



Exemple d 'application 2



Ligne de commande

```
Unix % genefinder -corrfactor 0.8 -tablenamefile
/packages/genefinder_washu/genefinder/human/humtables.hex -cpenalty 6.35 -penaltyfactor
0.85 -maxintronlength 10500 -minintronlength 70 -penaltycluster 50 -seqfile
pax6.wild.type.seq

Genefinder Version 084, release 960118
Parameters:
-seqfile pax6.wild.type.seq, -genefile "", -debug 0, -syncodon FALSE,
-normalize FALSE, -exoncutoff 3, -genecutoff 0, -atgcutoff 0,
-intron3cutoff 0, -intron5cutoff 0, -orfccutoff 5, -minexonlength 33,
Sequence: hsa1280.embl,.C
Length: 22253, nuc counts (N,A,C,G,T): 0 6645 4848 4692 6068

394 3' sites w/ scores > 0.00
310 5' sites w/ scores > 0.00
232 atg sites w/ scores > 0.00

Sequence: hsa1280.embl,
Length: 22253, nuc counts (N,A,C,G,T): 0 6068 4692 4848 6645

408 3' sites w/ scores > 0.00
429 5' sites w/ scores > 0.00
249 atg sites w/ scores > 0.00

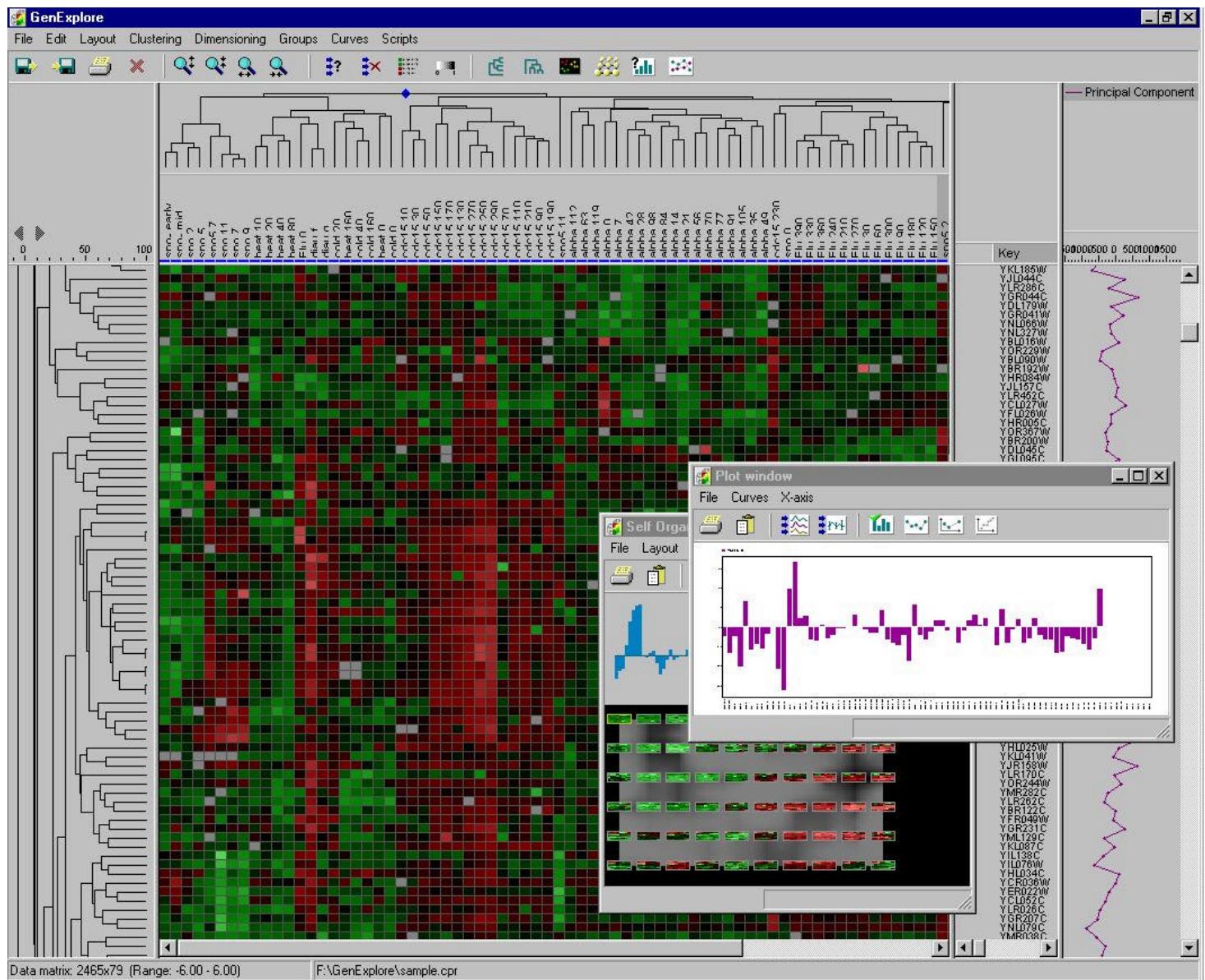
No predicted genes in hsa1280.embl,.C.

ORFs:

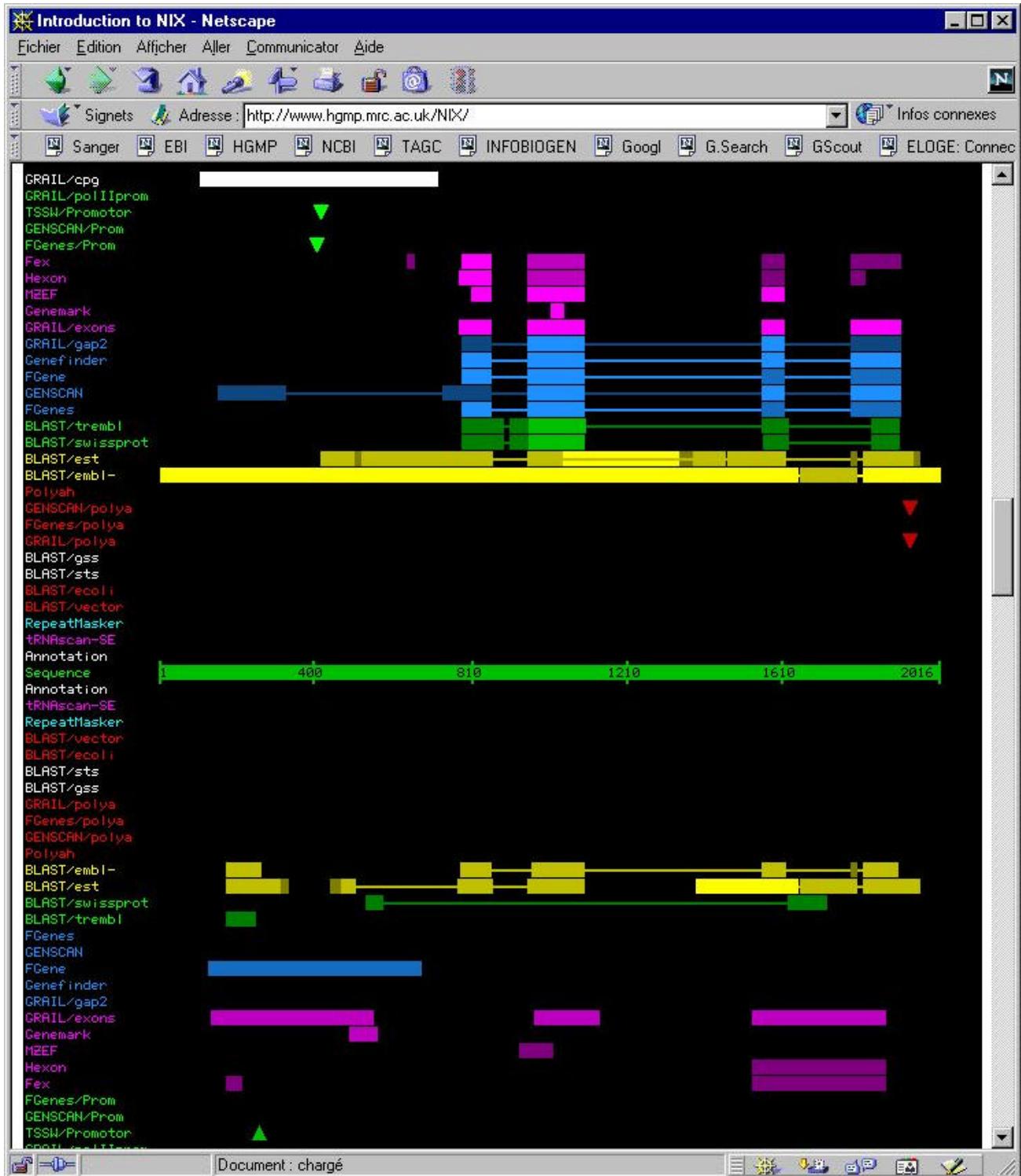
ORF: 17183 16701 483 4.16 5.79 6.00
*TKIISTMSKRPTPRHCYHTEPAMLSPRECLELCLEGQLMSLGCQKARQPFPSCTRGLLQLSSRFFRLYQTVLLCPGNPDLOQSTHIRA
QVHTYRPSAPTSLLFSHSHLLLACDIRVSKVSAYTTEPWAGTGCLTVVEITASEKEEVSQPAAVACTPT*
17144 17109 36 1.12
1 3' 17122 1.62
atg 17114 2.07
2 3' 17100 0.99
atg 17066 1.10
2 5' 17056 2.54
1 3' 17056 0.92
1 5' 17054 1.24
2 3' 17010 1.55
17009 16983 27 2.10
0 3' 16997 0.85
2 3' 16989 0.67
2 3' 16977 0.94
0 3' 16967 1.24
0 3' 16931 1.53
16853 16719 135 5.79
1 3' 16798 1.52
2 5' 16795 1.03
0 5' 16737 2.49

ORF: 5582 4998 585 5.66 4.06 6.00
*QSPPKNLTCGWGGGDCQLTQNFLTATRRVKVKSPWTSSTSVPNHHRTLADLTLSRPQTAGDENPPESGATSGGSCCPRLPNNSKKRG
GVSCAPSSWEPTHREGPVGEALGSALPGLGHRALSPSPRLLGLSGPDSCQGRDHQDLRLLGAEGWTLVSAKFGCHLGVSSVYKWESYGFK
SQAGMGLPTLGGVNR*
5507 5457 51 1.27
2 3' 5445 2.97
5408 5256 153 4.06
0 3' 5405 3.67
1 5' 5363 1.70
0 3' 5324 1.78
```

Logiciels interactifs



Interfaces Internet



Centres de ressources

Infobiogen

The screenshot shows the Infobiogen homepage with a blue header "Centre de Ressources INFOBIOGEN". Below it, a banner says "Le GIS INFOBIOGEN dépend du Centre de Ressources INFOBIOGEN". The main menu includes "Accueil", "Activités Accès au site R&D", "Services SRS PDB GDB GoldenPath SEQWEB Analyse de Séquences", and "Liens DEAMBULUM ORPHANET AGORA". A sidebar on the left lists "Banques de données de biomolécules, génomes, maladies génétiques", "Documentation Informatique - bioinformatique - Médecine", and "Informations sur les Services en Accès Authentifié destinées aux utilisateurs détenteurs d'un compte". Buttons for "Présentation d'INFOBIOGEN", "Education-Enseignement", "Nouveautés", and "Liens sur l'ailleurs..." are also present.

EBI

The screenshot shows the EBI homepage with a purple header "EMBL Outstation European Bioinformatics Institute". Below it, a banner says "The EMBL Outstation - European Bioinformatics Institute (EMBL) (Hinxton, Hinxton, UK) serving the Bioinformatics". The main menu includes "EMBL WebAlign", "Eltis 2001 Workshop", "ENSEMBL", "Information", "Databases", "Tools", "Submissions", "Groups", "Publications", and "Search EBI". A sidebar on the left lists "January 1st 2001 Web-based alignment tool for alignments of alignment databases", "January 16th 2001 The EBI organizes a workshop on ethical, legal, and social implications of Human Genome Project", "January 18th 2001 EMBL 20th anniversary with many improvements", and "January 19th 2001 EMBL 20th anniversary with many improvements". A footer at the bottom right reads "EMBL-European Bioinformatics Institute, Wellcome Trust Genome Campus, Hinxton, Cambridgeshire CB10 1SD, UK".

NCBI

The screenshot shows the NCBI homepage with a blue header "National Center for Biotechnology Information". Below it, a banner says "National Library of Medicine National Institutes of Health". The main menu includes "PubMed", "Entrez", "BLAST", "OMIM", "Taxonomy", and "Structure". A search bar "Search | GenBank" is present. A sidebar on the left lists "About NCBI", "GenBank", "Molecular databases", "Literature databases", "Genomic biology", "Tools", "Research at NCBI", and "Education". A central column features "What does NCBI do?", "OMIM in Entrez", "NCBI in the News", and "NCBI in the News". A sidebar on the right lists "Hot Topics" such as "Cancer genome anatomy project", "Clusters of orthologous groups", "Caffe Break", "Electronic PCR", "Gene expression profiles", "Genes and diseases", "Human genome resources", "Human/mouse homolog maps", "LocusLink", "Matrix genetics & genetics", "ORF Finder", "Reference sequence project", and "Reproducibility".

Expasy

The screenshot shows the ExPASy Molecular Biology Server homepage with a blue header "ExPASy Molecular Biology Server". Below it, a banner says "ExPASy Molecular Biology Server". The main menu includes "Databases", "Tools and Software Packages", "Education and Services", and "Documentation". A sidebar on the left lists "What does NCBI do?", "OMIM in Entrez", "NCBI in the News", and "NCBI in the News". A central column features "Links to other molecular biology databases", "The ExPASy EBI server", "Swiss-2DPAGE", "Swiss-2DPAGE training", "Links to lists of molecular biology resources", and "Links to some other molecular biology servers". A sidebar on the right lists "What's new in ExPASy", "Swiss-2DPAGE", "Swiss-2DPAGE training", "How to create HTML links to ExPASy", "Complete table of available documents", and "Links to some other molecular biology servers".

Plan du cours

1. Introduction
2. Banques de données
3. Matrices de substitutions
4. Alignement de 2 séquences
5. Recherche dans les banques par similarité de séquence
6. Alignements multiples
7. Motifs & signatures protéiques
8. Profils & domaines protéiques

Bases de données

- Modes de constitution

redondantes

Soumission directe:
archives **primaires**

incomplètes

Sélection & expertise:
banques de **référence**

- Catégories de données

base de données moléculaires

banques de connaissances

- Accès

Numéros d'accession

Annotation

Formats

Banques de données moléculaires

Séquences nucléiques

EMBL/GENBANK

dbEST

*UNIGENE

*ENSEMBL

*RefSeq...

Séquences protéiques

TrEMBL

*SWISSPROT

*PIR

Structures protéines

PDB, * CATH, * SCOP...

* banques de référence expertisées (non redondantes)

Banques de connaissances

Fonct/relat-ionnelles

LocusLink

GeneCards

PROSITE, PFam

KEGG...

Ontologies

Taxonomie

Symboles gènes

Enzymes

Tissus...

Organisme centrique

GDB

MGI

FlyBase

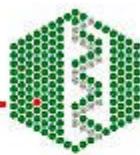
AceDB...

Bibliographiques

MEDLINE

Journaux Online

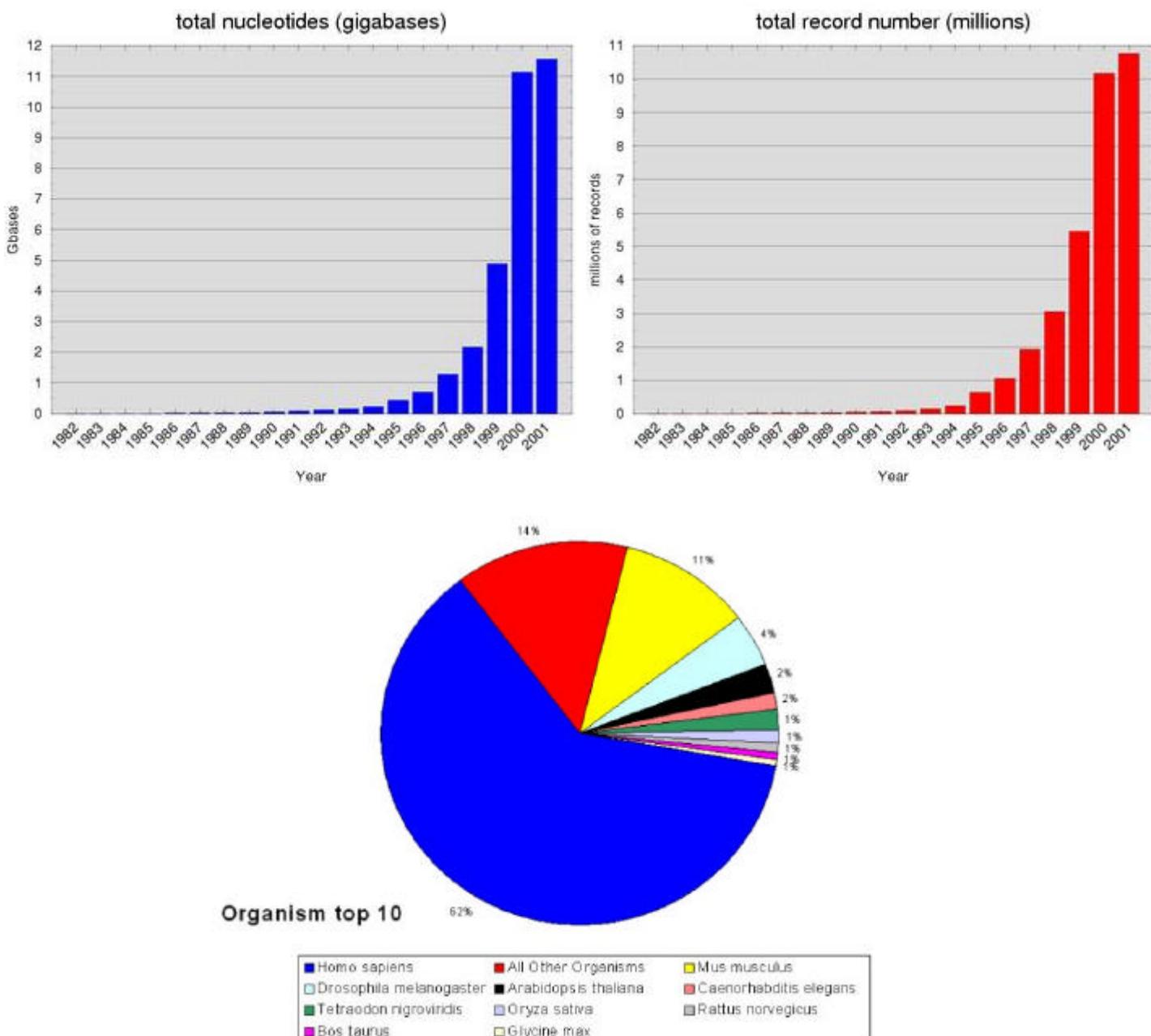
OMIM



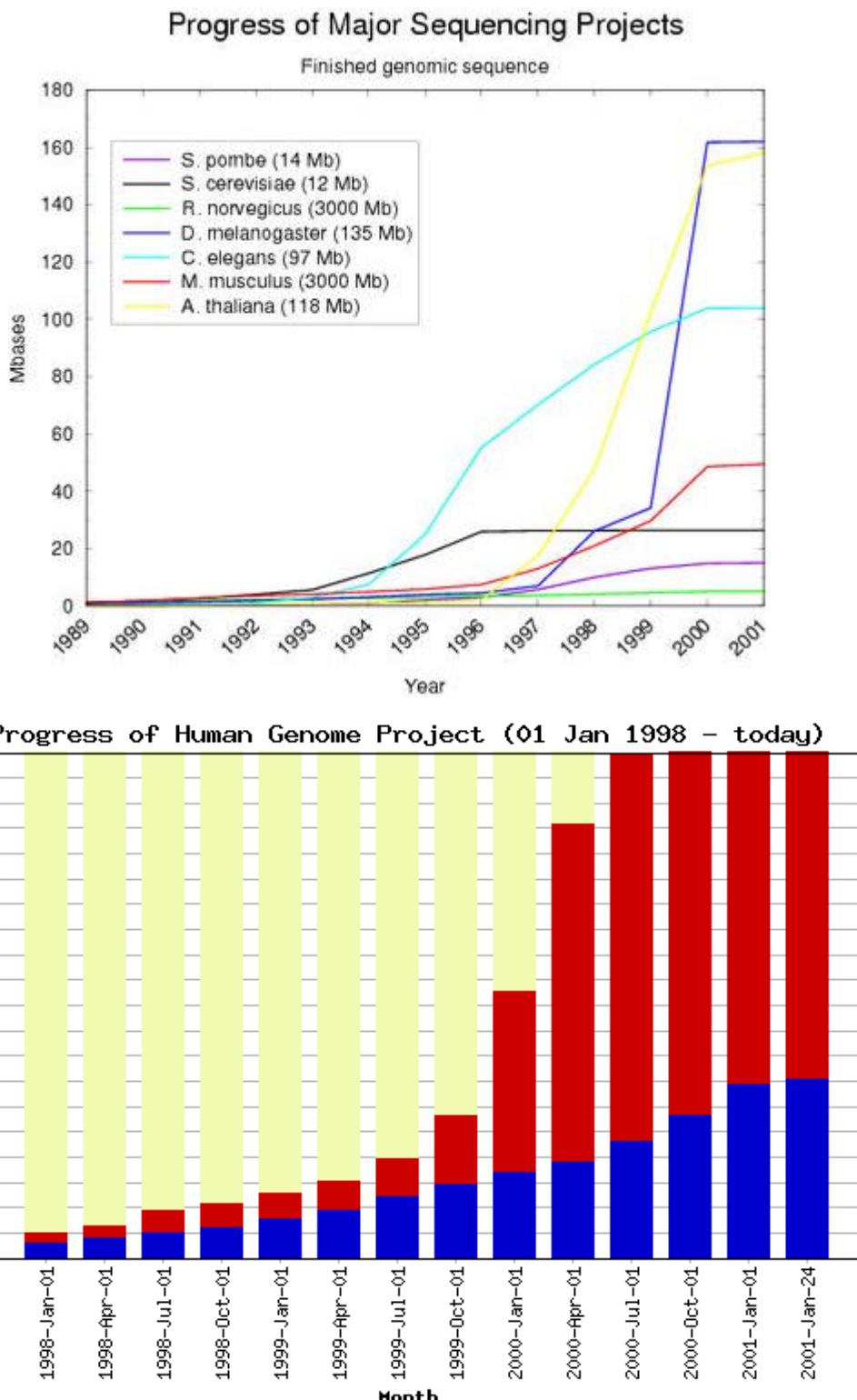
EMBL Nucleotide Sequence Database Statistics

EMBL Database growth

This morning, the EMBL Database contained **11,571,085,945** bases in **10,770,275** records.



EMBL: génomes



Séquence brute

```
GGGTTTATACGGATCTCCCTCTGTTGATAATTATGCCATTAAAGGTTTACCAAGTTCATAAATTAA  
GTAAAAATGAACCCATAAAAACAAAAGAGGTTCATCTACTTTAAGGAATTAAACCAAGGAATTAAAT  
TCATATTAAATAGCCATGGTTCCAGTTTACTGGCAGAGTACAAAACCTAATAGTGAATCCCTCTG  
AGCATTCAAAATCTCAGTGAATGAAGATAATTGACTGAATGGGATGTCATCTAAAAGGCCACCTGA  
CACTCTTATGAGGGAGGCTTATTCAAAGCAAAGATTGTCTTCCAAAATACCCATATGAACCACCC  
AGATTAACATTCACCTCTGAAATGTGGCATCCCAATATCTACTCTGATGGAAATTATGTATTCTATCT  
TGCATGGAGACAATGCTGAAGAACAGGAATGACTTGGTCTCCGGCTAAAAGATTGATACCGTACTTCT  
TAGTGTAAATTCTGCTCAATGAGCCAATCCAGATTCTCAGCAAATGTAGATGCAGCTAAAGCTAC  
CGTAAATATCTATATAAGAGGATTAGAATCATACCCATGGAAGTTAAAAGACTGTCAAAAATCAT  
TGGATGAGTGTTCAGCGGAAGACATAGAATATTTAAAATGTTCCAGTGAATGTTCTACCAGTACCCAG  
TGATGATTATGAAGATGAAGAACATGGAGGATGGCACCTATATCTTAACCTATGATGAGGATGAAGAA  
GAGGATGAAGAGATGGATGATGAGTAGTGCTGATTAACTTAGTTAGTGTAGTTAAACTTTAGT  
GCTTAGATTTAGTGTAACTTAGTTAGTGTAGTTAACTTAGTGTAGTTAGTGTAGTTAACTTTAGT  
GATTAGATTTAGTGTCTAGATTTAATGTTAACTTAGTGTAGTTAGTGTAGTTAACTTTAGT
```

Format FASTA

```
>X62440 African swine fever virus DNA for ubiquitin conjugating enzyme  
GGGTTTATACGGATCTCCCTCTGTTGATAATTATGCCATTAAAGGTTTACCAAGTTCATAAATTAA  
GTAAAAATGAACCCATAAAAACAAAAGAGGTTCATCTACTTTAAGGAATTAAACCAAGGAATTAAAT  
TCATATTAAATAGCCATGGTTCCAGTTTACTGGCAGAGTACAAAACCTAATAGTGAATCCCTCTG  
AGCATTCAAAATCTCAGTGAATGAAGATAATTGACTGAATGGGATGTCATCTAAAAGGCCACCTGA  
CACTCTTATGAGGGAGGCTTATTCAAAGCAAAGATTGTCTTCCAAAATACCCATATGAACCACCC  
AGATTAACATTCACCTCTGAAATGTGGCATCCCAATATCTACTCTGATGGAAATTATGTATTCTATCT  
TGCATGGAGACAATGCTGAAGAACAGGAATGACTTGGTCTCCGGCTAAAAGATTGATACCGTACTTCT  
TAGTGTAAATTCTGCTCAATGAGCCAATCCAGATTCTCAGCAAATGTAGATGCAGCTAAAGCTAC  
CGTAAATATCTATATAAGAGGATTAGAATCATACCCATGGAAGTTAAAAGACTGTCAAAAATCAT  
TGGATGAGTGTTCAGCGGAAGACATAGAATATTTAAAATGTTCCAGTGAATGTTCTACCAGTACCCAG  
TGATGATTATGAAGATGAAGAACATGGAGGATGGCACCTATATCTTAACCTATGATGAGGATGAAGAA  
GAGGATGAAGAGATGGATGATGAGTAGTGCTGATTAACTTAGTGTAGTTAGTGTAGTTAAACTTTAGT  
GCTTAGATTTAGTGTAACTTAGTTAGTGTAGTTAGTGTAGTTAGTGTAGTTAACTTTAGT  
GATTAGATTTAGTGTCTAGATTTAATGTTAACTTAGTGTAGTTAGTGTAGTTAACTTTAGT
```

Fiche EMBL

ID ASFVUCE standard; DNA; VRL; 979 BP.

XX

AC X62440;

XX

SV X62440.1

XX

DT 15-JAN-1992 (Rel. 30, Created)

DT 27-APR-1992 (Rel. 31, Last updated, Version 2)

XX

DE African swine fever virus DNA for ubiquitin conjugating enzyme

XX

KW ubiquitin; ubiquitin-conjugating enzyme.

XX

OS African swine fever virus

OC Viruses; dsDNA viruses, no RNA stage; Asfarviridae;

OC African swine fever-like viruses.

XX

RN [1]

RX MEDLINE; 92155177.

RA Hingamp P.M., Arnold J.E., Mayer R.J., Dixon L.K.;

RT "A Ubiquitin Conjugating Enzyme Encoded by African Swine Fever Virus";

RL EMBO J. 11:361-366(1992).

XX

RN [2]

RP 1-979

RA Dixon L.K.;

RT ;

RL Submitted (04-OCT-1991) to the EMBL/GenBank/DDBJ databases.

RL L.K. Dixon, Institute of Animal Health, Pirbright Laboratory, Ash Road,

RL Pirbright, Woking, Surrey GU24 0NF, UK

XX

DR SWISS-PROT; P25869; UBC_ASFM2.

XX

FH Key Location/Qualifiers

FH

FT source 1..979

FT /db_xref="taxon:10497"

FT /organism="African swine fever virus"

FT /cell_line="pig blood"

FT /clone_lib="lambda EMBL1 Lmw"

FT /clone="Lmw22"

FT /isolate="Malawi LIL20/1"

FT 5' UTR 1..155

FT CDS 156..797

FT /db_xref="SWISS-PROT:P25869"

FT /gene="ASFVUBC"

FT /product="ubiquitin conjugating enzyme"

FT /protein_id="CAA44305.1"

FT /translation="MVSSFLLAEYKNLIVNPSEHFKISVNEDNLTEWDVILKGPPDTLY
EGGLFKAKIVFPPKYPYEPPLRTFTSEMWHPNIYSDGKLCISILHGDNAEEQGMTWS
PAQKIDTVLLSVISLLNEPNPDSPANVDAAKSYRKYLYKEDLESYPMEVKKT
VKKSLDEC
SAEDIEYFKNVNVLPVPSPDDYEDEEMEDGTYILTYDDEDEEEDEEMDE"

FT misc_feature 408..410

FT /note="active site cysteine"

FT 3' UTR 798..979

XX

SQ Sequence 979 BP; 329 A; 151 C; 182 G; 317 T; 0 other;
gggtttata cggatctccc ttctcgtttataattatgc cattaaagg tttaccagtt 60
cataaaatcta gtaaaaatga accccataaa aaacaaaaga ggtcata ctttaagga 120
[...]
aaactttagt gattagatt tagtgcttag atttaatgt ttaaacttta gtgattagat 960
tttagtgctt agatttaa 979

//

Fiche GENBANK

LOCUS ASFVUCE 979 bp DNA VRL 27-APR-1992
 DEFINITION African swine fever virus DNA for ubiquitin conjugating enzyme.
 ACCESSION X62440
 VERSION X62440.1 GI:58649
 KEYWORDS ubiquitin; ubiquitin-conjugating enzyme.
 SOURCE African swine fever virus.
 ORGANISM African swine fever virus
 Viruses; dsDNA viruses, no RNA stage; African swine fever-like viruses.
 REFERENCE 1 (bases 1 to 979)
 AUTHORS Hingamp,P.M., Arnold,J.E., Mayer,R.J. and Dixon,L.K.
 TITLE A ubiquitin conjugating enzyme encoded by African swine fever virus
 JOURNAL EMBO J. 11 (1), 361-366 (1992)
 MEDLINE 92155177
 REFERENCE 2 (bases 1 to 979)
 AUTHORS Dixon,L.K.
 TITLE Direct Submission
 JOURNAL Submitted (04-OCT-1991) L.K. Dixon, Institute of Animal Health,
 Pirbright Laboratory, Ash Road, Pirbright, Woking, Surrey GU24 0NF,
 UK
 FEATURES Location/Qualifiers
 source 1..979
 /organism="African swine fever virus"
 /isolate="Malawi LIL20/1"
 /db_xref="taxon:10497"
 /clone="Lmw22"
 /cell_line="pig blood"
 /clone_lib="lambda EMBL1 Lmw"
 5' UTR 1..155
 gene 156..797
 /gene="ASFVUBC"
 CDS 156..797
 /gene="ASFVUBC"
 /codon_start=1
 /product="ubiquitin conjugating enzyme"
 /protein_id="CAA44305.1"
 /db_xref="GI:58650"
 /db_xref="SWISS-PROT:P25869"
 /translation="MVSSFLLAEYKNLIVNPSEHFKISVNEDNLTEWDVILKGPPDTLYEGGLFKAKIVFPPKYPYEPPRLTFTSEMWHPIYSDGKLCISILHGDNAAEQGMTWSPAQKIDTVLLSVISLLNEPNPDSPANVDAAKSYRKYLYKEDLESYPMEVKKTVKKSLEDCSAEDIEYFKNVPVNLPVPSDDYEDEEMEDGYITYILTYDDEEEEDEEMDDE"
 misc_feature 408..410
 /gene="ASFVUBC"
 /note="active site"
 3' UTR 798..979
 BASE COUNT 329 a 151 c 182 g 317 t
 ORIGIN
 1 gggttttata cgatctccc ttctcggttataattatgc cattaaagggtttaccagtt
 61 cataaaattta gtaaaaatga accccataaaa aaacaaaaga gttcatcta cttaagga
 [...]
 901 aaacttttagt gattagattt tagtgcttag atttaatgt taaaactta gtgatttagat
 961 tttagtgctt agatttaa
 //

Fiche SWI SSPROT

ID UBC_ASFM2 STANDARD; PRT; 213 AA.
AC P25869;
DT 01-MAY-1992 (Rel. 22, Created)
DT 01-MAY-1992 (Rel. 22, Last sequence update)
DT 01-AUG-1992 (Rel. 23, Last annotation update)
DE UBIQUITIN-CONJUGATING ENZYME E2-21 KDA (EC 6.3.2.19)
DE (UBIQUITIN-PROTEIN LIGASE) (UBIQUITIN CARRIER PROTEIN).
GN ASFV-UBC.
OS African swine fever virus (isolate Malawi Lil 20/1) (ASFV).
OC Viruses; dsDNA viruses, no RNA stage; Asfarviridae;
OC African swine fever-like viruses.
OX NCBI_TaxID=10500;
RN [1]
RP SEQUENCE FROM N.A.
RX MEDLINE=92155177; PubMed=1310934;
RA Hingamp P.M., Arnold J.E., Mayer R.J., Dixon L.K.;
RT "A ubiquitin conjugating enzyme encoded by African swine fever
virus.";
RL EMBO J. 11:361-366(1992).
CC -!- FUNCTION: CATALYZES THE COVALENT ATTACHMENT OF UBIQUITIN TO
CC OTHER PROTEINS.
CC -!- CATALYTIC ACTIVITY: ATP + UBIQUITIN + PROTEIN LYSINE = AMP +
CC PYROPHOSPHATE + PROTEIN N-UBIQUITYLLYSINE.
CC -!- PATHWAY: SECOND STEP IN UBIQUITIN CONJUGATION.
CC -!- MISCELLANEOUS: A CYSTEINE RESIDUE IS REQUIRED FOR
CC UBIQUITIN-THIOLESTER FORMATION.
CC -!- SIMILARITY: BELONGS TO THE UBIQUITIN-CONJUGATING ENZYME FAMILY.
DR EMBL; X62440; CAA44305.1; -.
DR EMBL; X71982; CAA50851.1; -.
DR PIR; S19158; S19158.
DR HSSP; Q02159; 2UCZ.
DR InterPro; IPR000608; -.
DR Pfam; PF00179; UQ_con; 1.
DR PROSITE; PS00183; UBIQUITIN_CONJUGAT_1; 1.
DR PROSITE; PS50127; UBIQUITIN_CONJUGAT_2; 1.
KW Ubiquitin conjugation; Ligase.
FT BINDING 85 85 UBIQUITIN (BY SIMILARITY).
FT DOMAIN 183 213 ASP/GLU-RICH (BASIC).
SQ SEQUENCE 213 AA; 24468 MW; F9671BC7385D6DCE CRC64;
MVSSFLLAEY KNLIVNPSEH FKISVNEDNL TEWDVILKGP PDTLYEGGLF KAKIVFPPKY
PYEPPRLTFT SEMWHPNIYS DGKLCISILH GDNAEEQGMT WSPAQKIDTV LLSVISLLNE
PNPDSPANVD AAKSYRKYLY KEDLESYPME VKKTVKKSLD ECSAEDIEYF KNVPVNLPV
PSDDYEDEEM EDGTYILTYD DEDEEEDEEM DDE

//

Abstracts PubMed

The screenshot shows the Entrez-PubMed interface in a Netscape browser window. The title bar reads "Entrez-PubMed - Netscape". The menu bar includes "Fichier", "Edition", "Afficher", "Aller", "Communicator", and "Aide". The toolbar has icons for various functions like search, history, and file operations. The address bar shows the URL: "Adresse : .gov:80/entrez/query.fcgi?cmd=Retrieve&db=PubMed&list_uids=10812478&dopt=Abstract". The bookmarks bar includes links to Sanger, EBI, HGMP, NCBI, TAGC, INFOBIOGEN, Google, G.Search, GScout, and ELOGE: Connect. The main content area features the NCBI logo and the National Library of Medicine logo. The PubMed search bar has "Search [PubMed] for" and buttons for "Go" and "Clear". Below the search bar are buttons for "Limits", "Preview/Index", "History", and "Clipboard". A dropdown menu shows "Display" selected, with "Abstract" highlighted. Other options include "Save", "Text", "Order", and "Add to Clipboard". The search results list the first article: "1: Bioinformatics 2000 Jan;16(1):65-75". A blue button below the title says "Full text article at bioinformatics.oupjournals.org". The article title is "A brief history of genome research and bioinformatics in France." by "Danchin A". The author's affiliation is "Regulation of Gene Expression, Institut Pasteur, Paris, France." and the email is "adanchin@pasteur.fr". The text discusses the slow development of in silico genomics in France due to political reasons, mentioning the Groupement de Recherche sur les Génomes (GREG) and the Groupement de Recherche 1029 (GDR 1029) of the Centre National de la Recherche Scientifique (CNRS). It highlights the creation of dynamics that will place France among developed nations supporting Large-Scale Biology. Publication types listed are "Historical article". The PMID is 10812478. At the bottom, there is another "Display" button with "Abstract" selected.

Journaux 'Online'



OMIM

OMIM - CREUTZFELDT-JAKOB DISEASE; CJD - Netscape

Fichier Edition Afficher Ailler Communicator Aide

Signets Adresser : http://www.ncbi.nlm.nih.gov:80/entrez/dispmim.cgi?id=123400 Infos connexes

Sanger EBI HGMP NCBI TAGC INFOBIOGEN Google G.Search GScout ELOGE Conn

NCBI

MIM #123400

Description Clinical Features Biochemical Features Inheritance Population Genetics Animal Model See Also References Contributors Creation Date Edit History

MINI-MIM Clinical Synopsis Gene map

LinkOut CCR

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

Display Detailed Save Text Add to Clipboard

123400 Related Entries, PubMed, LinkOut

CREUTZFELDT-JAKOB DISEASE; CJD

Gene map locus [20pter-p12](#)

TEXT

DESCRIPTION

A number sign (#) is used with this entry because of evidence that the disorder is caused by mutations in the prion protein gene (PRNP; [176640](#)).

[Johnson and Gibbs \(1998\)](#) provided a comprehensive review of Creutzfeldt-Jakob disease and related transmissible spongiform encephalopathies.

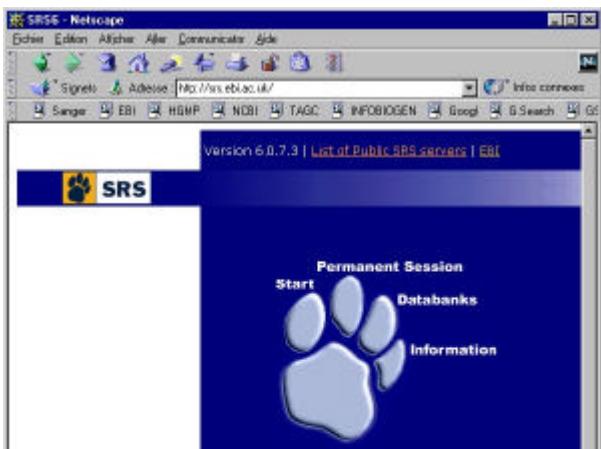
CLINICAL FEATURES

[Jakob et al. \(1950\)](#) gave a follow-up on the first reported family, the Backer kindred. Three generations may have been affected, with male-to-male transmission. [Davidson and Rabiner \(1940\)](#) described 3 affected sibs. [Friede and DeJong \(1964\)](#) and later [May et al. \(1968\)](#) described affected father and 3 daughters. Onset was between 38 and 45 years. The illness lasted only 10 months to 2 years. The disorder began with forgetfulness and nervousness and progressed with jerky, trembling movements of the hands, loss of facial expression, and unsteady gait. Pathologic findings included severe status spongiosus, diffuse nerve cell degeneration and some glial proliferation. Creutzfeldt-Jakob disease may be a mixed category.

[Rosenthal et al. \(1976\)](#) reported a remarkable kindred in which the proband had clinically typical Creutzfeldt-Jakob disease, but neuropathologic studies showed encephalopathy. A first cousin had chronic spongiform dementia without spongy changes at autopsy. Both had PAS-positive, eosinophilic plaques throughout the brain. The pedigree indicated neurologic disease with or without subacute or chronic dementia in 16 members of the kindred. A general, genetically determined susceptibility to neurologic disease was postulated.

In a Chilean family, [Cartier et al. \(1985\)](#) described a brother and sister and possibly a

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Recherche par l'annotation

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Recherche par l'annotation

The screenshot displays a dual-panel interface for a biological database search, likely SRS.

Left Panel (Query Form):

- Toolbar:** Includes icons for Signets, Adresses, Infos connexes, Sanger, EBI, HGMP, NCBI, TAGC, INFOBIOGEN, Google, G.Search, GSoult, and ELOGE: Connec.
- Menu Bar:** Fichier, Edition, Afficher, Aler, Communicator, Aide.
- Address Bar:** Adresse : <http://srs.ebi.ac.uk/srs6bin/cgi-bin/wgetz>
- Navigation Buttons:** TOP PAGE, QUERY (highlighted), RESULTS, SESSIONS, VIEWS, DATABANKS, HELP.
- Search Input:** search SWISSPROT SPTRREMBL.
- Info Field:** about field AllText
- Submit Query Buttons:** Reset, Submit Query.
- Search Criteria:** Description: ubiquitin, Organism: Homo sapiens, GeneName: Ubc, Keywords: [empty].
- Configuration Options:** append wildcards to words (checked), combine searches with AND, Number of entries to display per page (30), Extended query form, Configure your own SRS Quick Search_.

Right Panel (Query Result):

- Toolbar:** Includes icons for Signets, Adresses, Infos connexes, Sanger, EBI, HGMP, NCBI, TAGC, INFOBIOGEN, Google, G.Search, GSoult, and ELOGE: Connec.
- Menu Bar:** Fichier, Edition, Afficher, Aler, Communicator, Aide.
- Address Bar:** Adresse : <http://srs.ebi.ac.uk/srs6bin/cgi-bin/wgetz>
- Navigation Buttons:** TOP PAGE, QUERY, RESULTS (highlighted), SESSIONS, VIEWS, DATABANKS, HELP.
- Search Summary:** query "[libs-(swissprot sptrreml)-Description: ubiquitin*] & [libs-Organism: Homo* & sapiens*] & [libs-GeneName: Ubc*]" found 10 entries
- Table of Results:** Shows 10 entries from the SWISSPROT SPTRREMBL database.

SWISSPROT SPTRREMBL	Accession	Description	SeqLength
SWISSPROT:UBC1_HUMAN	P50550	UBIQUITIN-CONJUGATING ENZYME E2-18 KDA (EC 6.3.2.19) (UBIQUITIN-PROTEIN LIGASE) (UBIQUITIN CARRIER PROTEIN) (P18).	158
SWISSPROT:UBS5A_HUMAN	P51668	UBIQUITIN-CONJUGATING ENZYME E2-17 KDA 1 (EC 6.3.2.19) (UBIQUITIN-PROTEIN LIGASE) (UBIQUITIN CARRIER PROTEIN) (E2(17)K8 1).	147
SWISSPROT:UBS5B_HUMAN	P51669	UBIQUITIN-CONJUGATING ENZYME E2-17 KDA 2 (EC 6.3.2.19) (UBIQUITIN-PROTEIN LIGASE) (UBIQUITIN CARRIER PROTEIN) (E2(17)K8 2).	147
SWISSPROT:UBSC_HUMAN	P47986	UBIQUITIN-CONJUGATING ENZYME E2-17 KDA 3 (EC 6.3.2.19) (UBIQUITIN-PROTEIN LIGASE) (UBIQUITIN CARRIER PROTEIN) (E2(17)K8 3).	147
SWISSPROT:UBC6_HUMAN	P51965	UBIQUITIN-CONJUGATING ENZYME E2-21 KDA UBC6 (EC 6.3.2.19) (UBIQUITIN-PROTEIN LIGASE) (UBIQUITIN CARRIER PROTEIN).	193
SWISSPROT:UBC7_HUMAN		UBIQUITIN-CONJUGATING	