

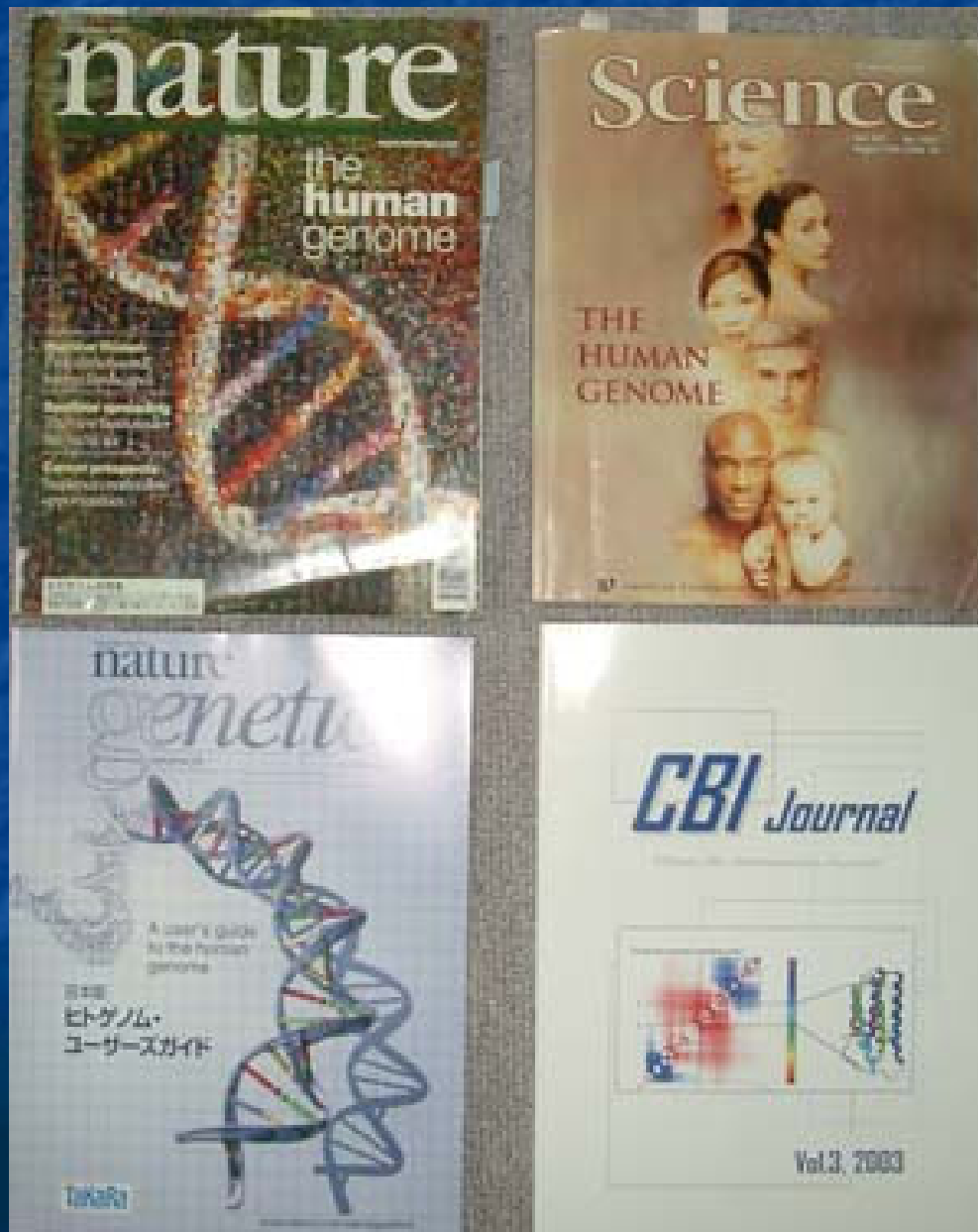
ヒトゲノム情報へ アクセスするための ブラウザについて

東京医科歯科大学

生命情報学

田中義智

はじめに



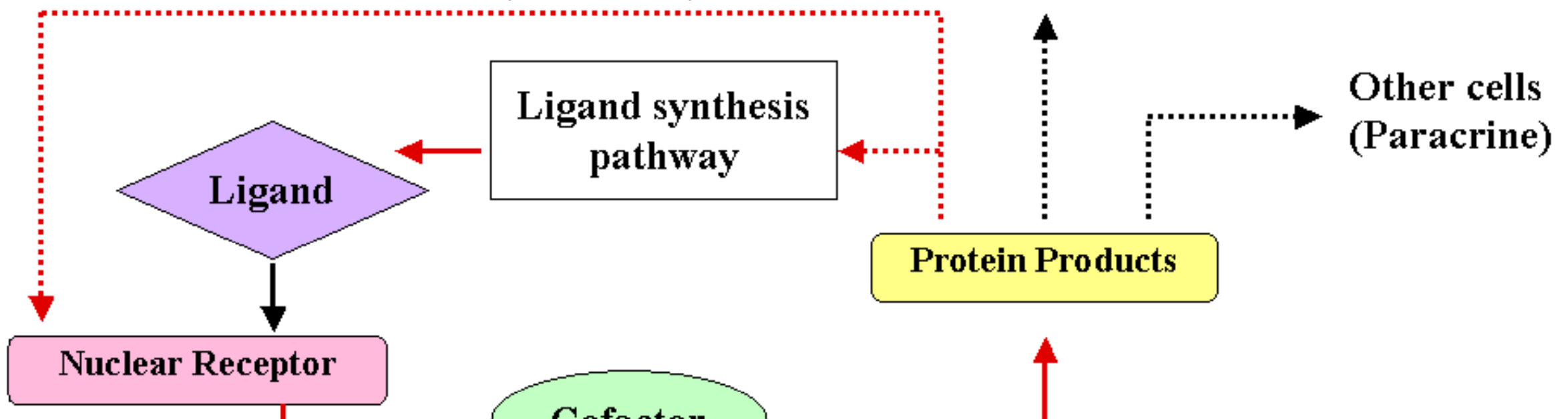
2003年4月14日にヒトゲノム完全解読宣言がなされました。(ゲノムの遺伝子領域とユークロマチンの99%が99.99%の精度で解読された。)これによりゲノム情報を用いたさまざまな研究が可能となった。

本チュートリアルでは、ヒトゲノム情報を利用するための代表的なブラウザとその使用法を紹介する

修士課程での研究

Feedback (Autocrine)

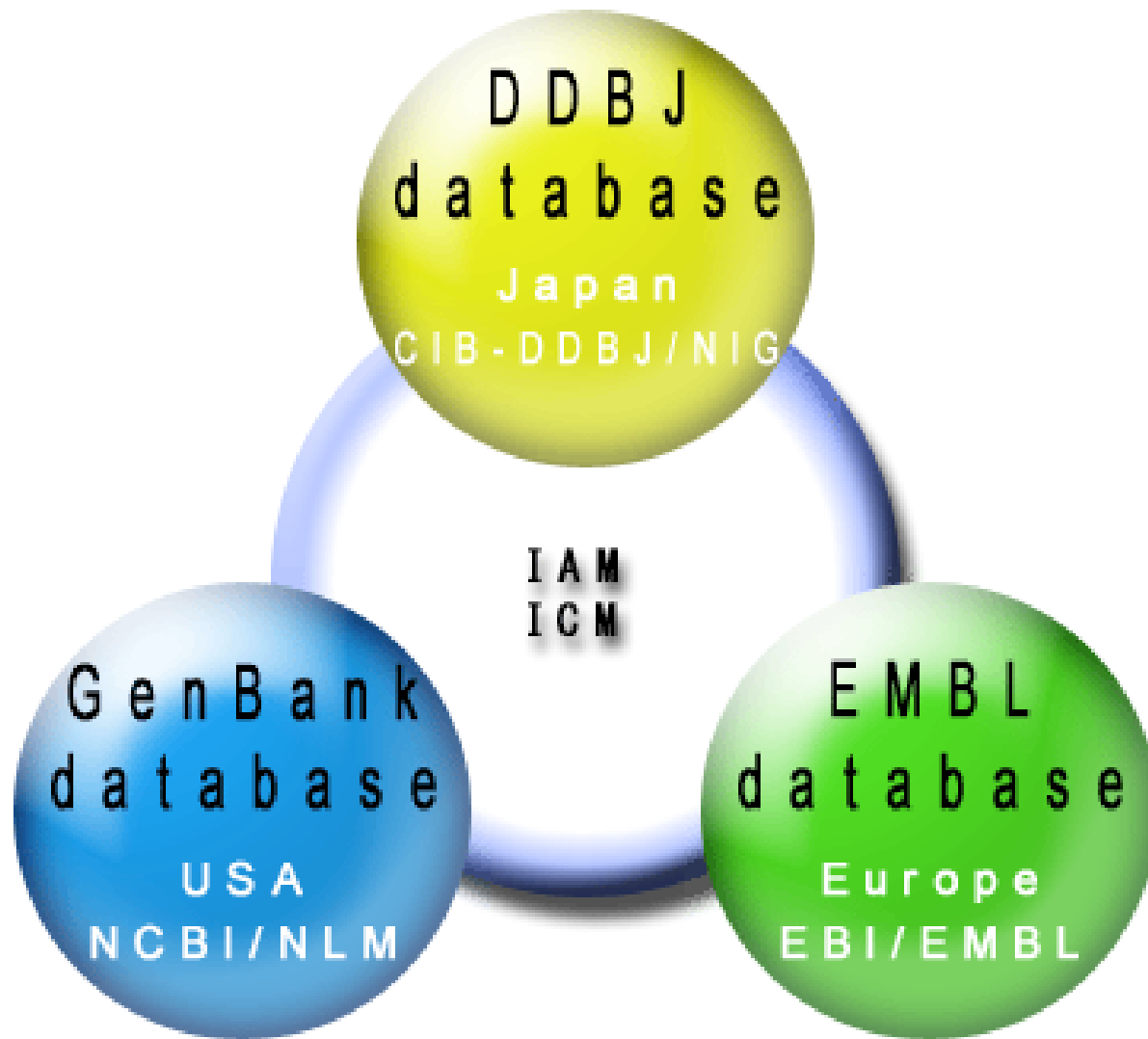
Other functions



チュートリアルの内容

- 遺伝子の総合的な情報を調べる
- 遺伝子の位置と近傍の遺伝子を調べる
- 塩基配列の収集
 - 遺伝子内部、上流、下流
- SNPsを調べる

国際塩基配列データベース



- エントリ数は異なるが基本的に同じ塩基配列データ
- 塩基配列に与えられたアクセッション番号は3者間で共通となる
- 数は多いが重複が多く、品質もまちまち

配列のアノテーション

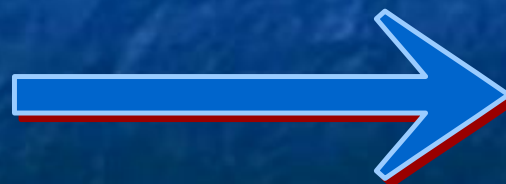
- ヒトゲノム配列はNCBIで組み立てた配列を基準配列としている

その後のアノテーション (annotation, 注釈付け) はサイトにより異なる



NCBIのRefSeq計画

- 分子生物学のセントラルドグマを構成する分子 (DNA, mRNA, タンパク質) についての**基準**となる配列を提供
 - GenBankに重複登録されているデータを簡素化 (non-redundant)
 - mRNA : NM_*
 - タンパク質 : NP_*
 - ゲノム配列 : NT_*



重複の減少
品質保証



代表的なブラウザ

- NCBI

- <http://www.ncbi.nlm.nih.gov/>

- UCSC

- <http://genome.ucsc.edu/>

- Ensemble

- <http://www.ensembl.org/>

遺伝子の情報

- NCBIのEntrez Geneを使用
 - ゲノム完全配列によるRefSeqとしてアノテーションされた遺伝子とLocusLinkからの情報を統合
 - LocusLink上の遺伝子に関する情報はEntrez Geneに移されている
 - LocusIDとGeneIDは互換性がある



National Center for Biotechnology Information

National Library of Medicine

National Institutes of Health

PubMed

Entrez

BLAST

OMIM

Books

TaxBrowser

Structure

Search

Entrez



homo abca1

Go

Entrezを選択

homo abca1と入力

Goをクリック

ots

Site

Guide to NCBI resources

About NCBI

An introduction for researchers, educators and the public

GenBank

Sequence submission support and software

Literature databases

PubMed, OMIM, Books, and PubMed Central

Established in 1988 as a national resource for molecular biology information, NCBI creates public databases, conducts research in computational biology, develops software tools for analyzing genome data, and disseminates biomedical information - all for the better understanding of molecular processes affecting human health and disease. [More...](#)

The Genetic Landscape of Diabetes

Over 17 million Americans have diabetes. Explore the genes discovered thus far with "The Genetic Landscape of Diabetes" as your guide.

- ▶ Clusters of orthologous groups
- ▶ Coffee Break, Genes & Disease, NCBI Handbook
- ▶ Electronic PCR
- ▶ Entrez Home
- ▶ Entrez Tools
- ▶ Gene expression omnibus (GEO)
- ▶ Human genome resources

Search across databases

homo abca1

GO

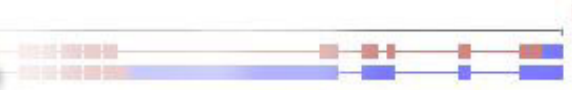
CLEAR

Help

- | | |
|--|--|
| <p>none PubMed: biomedical literature citations and abstracts ?</p> <p>1 PubMed Central: free, full text journal articles ?</p> | <p>none Books: online books ?</p> <p>none OMIM: Online Mendelian Inheritance in Man ?</p> <p>222 Site Search: NCBI web and FTP sites ?</p> |
| <p>58 Nucleotide: sequence database (GenBank) ?</p> <p>40 Protein: sequence database ?</p> <p>1 Genome: whole genome sequences ?</p> <p>5444 Structure: three-dimensional macromolecular structures ?</p> <p>none Taxonomy: organisms in GenBank ?</p> <p>696 SNP: single nucleotide polymorphism ?</p> <p>13 Gene: gene-centered information ?</p> <p>13 Eukaryotic: eukaryotic homology groups ?</p> | <p>1 UniGene: gene-oriented clusters of transcript sequences ?</p> <p>45 CDD: conserved protein domain database ?</p> <p>21878 3D Domains: domains from Entrez Structure ?</p> <p>19 UniSTS: markers and mapping data ?</p> <p>1 PopSet: population study data sets ?</p> <p>289 GEO Profiles: expression and molecular abundance profiles ?</p> <p>none GEO DataSets: experimental sets of GEO data ?</p> <p>none Cancer Chromosomes: cytogenetic databases ?</p> |
| <p>1 Journals: information about the journals indexed in PubMed and other Entrez databases ?</p> | <p>5 MeSH: detailed information about NLM's controlled vocabulary ?</p> |

クリック

- Result counts displayed in gray indicate one or more terms not found



Search for current records only

[Limits](#) [Preview/Index](#) [History](#) [Clipboard](#) [Details](#)

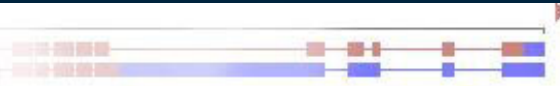
Display Show:

Items 1-13 of 13

One page.

- 1: [ABCA1](#)** Links
 ATP-binding cassette, sub-family A (ABC1), member 1 [*Homo sapiens*]
Other Aliases: クリック CERP, HDLDT1, TGD
Other Designations: cassette 1; ATP-binding cassette, sub-family A member 1; cholesterol efflux regulatory protein; high density lipoprotein deficiency, Tangier type, 1
Chromosome: 9; **Location:** 9q31.1
GeneID: 19
- 2: [APOA1](#)** Links
 apolipoprotein A-I [*Homo sapiens*]
Chromosome: 11; **Location:** 11q23-q24
GeneID: 335
- 3: [ABCA7](#)** Links
 ATP-binding cassette, sub-family A (ABC1), member 7 [*Homo sapiens*]
Other Aliases: HGNC:37, ABCA-SSN, ABCX
Other Designations: ATP-binding cassette, sub-family A, member 7; autoantigen SS-N; macrophage ABC transporter
Chromosome: 19; **Location:** 19p13.3
GeneID: 10347
- 4: [ABCA5](#)** Links
 ATP-binding cassette, sub-family A (ABC1), member 5 [*Homo sapiens*]
Other Aliases: HGNC:35, ABC13, EST90625
Other Designations: ATP-binding cassette A5; ATP-binding cassette, sub-family A , member 5
Chromosome: 17; **Location:** 17q24.3
GeneID: 23461

- Entrez
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- Gene**
- Search
- Gene Help
- FAQ**
- FTP site**
- Related sites**
- Entrez Genome
- Genomic Biology
- HomoloGene
- LocusLink
- Map Viewer
- OMIM
- RefSeq
- UniGene
- Feedback**
- Help Desk
- Corrections
- About GeneRIFs
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- Map Viewer



Entrez PubMed Nucleotide Protein Genome Structure PMC Taxonomy Books OMIM

Search for current records only

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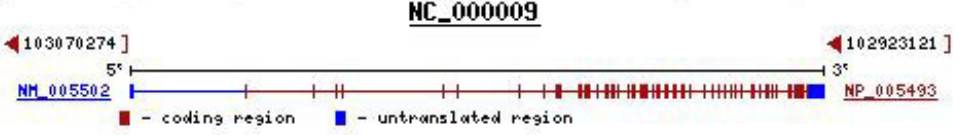
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- [Genomic Biology](#)
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- [Map Viewer](#)

Show:

1: ABCA1 ATP-binding cassette, sub-family A (ABC1), member 1 [*Homo sapiens*] [Links](#)

GeneID: 19 Locus tag: [HGNC:29](#); [MIM: 600046](#) updated 12-Aug-2004

Transcripts and products: (shown on reverse complement genome) [RefSeq below](#)



Genomic context: chromosome: 9; Maps: 9q31.1



Gene type: protein coding
Gene name: ABCA1
Gene description: ATP-binding cassette, sub-family A (ABC1), member 1
RefSeq status: Reviewed
Organism: *Homo sapiens*

Lineage: *Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo*
Gene aliases: TGD; ABC1; CERP; HDLDT1

Summary: The membrane-associated protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intracellular membranes. ABC genes are divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This protein is a member of the ABC1 subfamily. Members of the ABC1 subfamily comprise the only major ABC subfamily found exclusively in multicellular eukaryotes. With cholesterol as its substrate, this protein functions as a cholesterol efflux pump in the cellular lipid removal pathway. Mutations in this gene have been associated with Tangier's disease and familial high-density lipoprotein deficiency.

Bibliography: Gene References into Function (GeneRIF): [Submit](#) [help](#)

[PubMed](#) links

GeneRIFs:

1. Two polymorphisms were associated with plasma levels of ApoA1, 1 in the promoter (C-564T) and 1 in the coding (R1587K) regions, whereas only 1 polymorphism (R219K) was associated with the risk of myocardial infarction.
2. ABCA1 is not required for a positive feedback pathway for stimulation of potentially anti-atherogenic apoE secretion by alpha-helix-containing molecules including apoA-I and apoE
3. Both mutations prevent normal trafficking of ABCA1, thereby explaining their inability to mediate apoA1-dependent lipid efflux
4. A two step model of cholesterol efflux is suggested that can explain the functional interactions of ABCA1 with apoA-I and other cholesterol acceptors, based on formation of a tight complex between ABCA1 and its ligands

遺伝子の位置と近傍遺伝子を調べる

- TNF α を例にNCBIのMap Viewerを使用
- <http://www.ncbi.nlm.nih.gov/>

The screenshot shows the NCBI homepage with a search bar containing 'Gene' and 'tnfa'. The search results for 'tnfa' are displayed, including a 'What does NCBI do?' section and a 'Hot Spots' section. The 'Hot Spots' section lists various resources, with 'Map Viewer' highlighted in a red box. A yellow callout box points to the 'Map Viewer' link with the text 'Map Viewer をクリック'.

NCBI National Center for Biotechnology Information
National Library of Medicine National Institutes of Health

PubMed Entrez BLAST OMIM Books TaxBrowser Structure

Search Gene for tnfa Go

SITE MAP
Guide to NCBI resources

About NCBI
An introduction for researchers, educators and the public

GenBank
Sequence submission support and software

Literature databases
PubMed, OMIM, Books, and PubMed Central

Molecular databases
Sequences, structures, and taxonomy

Genomics
The human genome, whole genomes, and related resources

What does NCBI do?
Established in 1988 as a national resource for molecular biology information, NCBI creates public databases, conducts research in computational biology, develops software tools for analyzing genome data, and disseminates biomedical information - all for the better understanding of molecular processes affecting human health and disease. [More...](#)

Hot Spots

- ▶ Clusters of orthologous groups
- ▶ Coffee Break, Genes & Disease, NCBI Handbook
- ▶ Electronic PCR
- ▶ Entrez Home
- ▶ Entrez Tools
- ▶ Gene expression omnibus (GEO)
- ▶ Human genome resources
- ▶ LocusLink
- ▶ Malaria genetics & genomics
- ▶ **Map Viewer**
- ▶ dbMHC
- ▶ Mouse genome resources

Cancer Chromosomes
Find cytogenetic, clinical, and reference information in the new Cancer Chromosomes database. Cancer Chromosomes, part of the Entrez retrieval system, integrates information from the NCI Mitelman Database of Chromosome Aberrations in Cancer, the NCI Recurrent Aberrations in Cancer database, and the NCI/NCBI SKY/M-FISH & CGH Database.

Entrez Gene

Map Viewer をクリック

PubMed Central

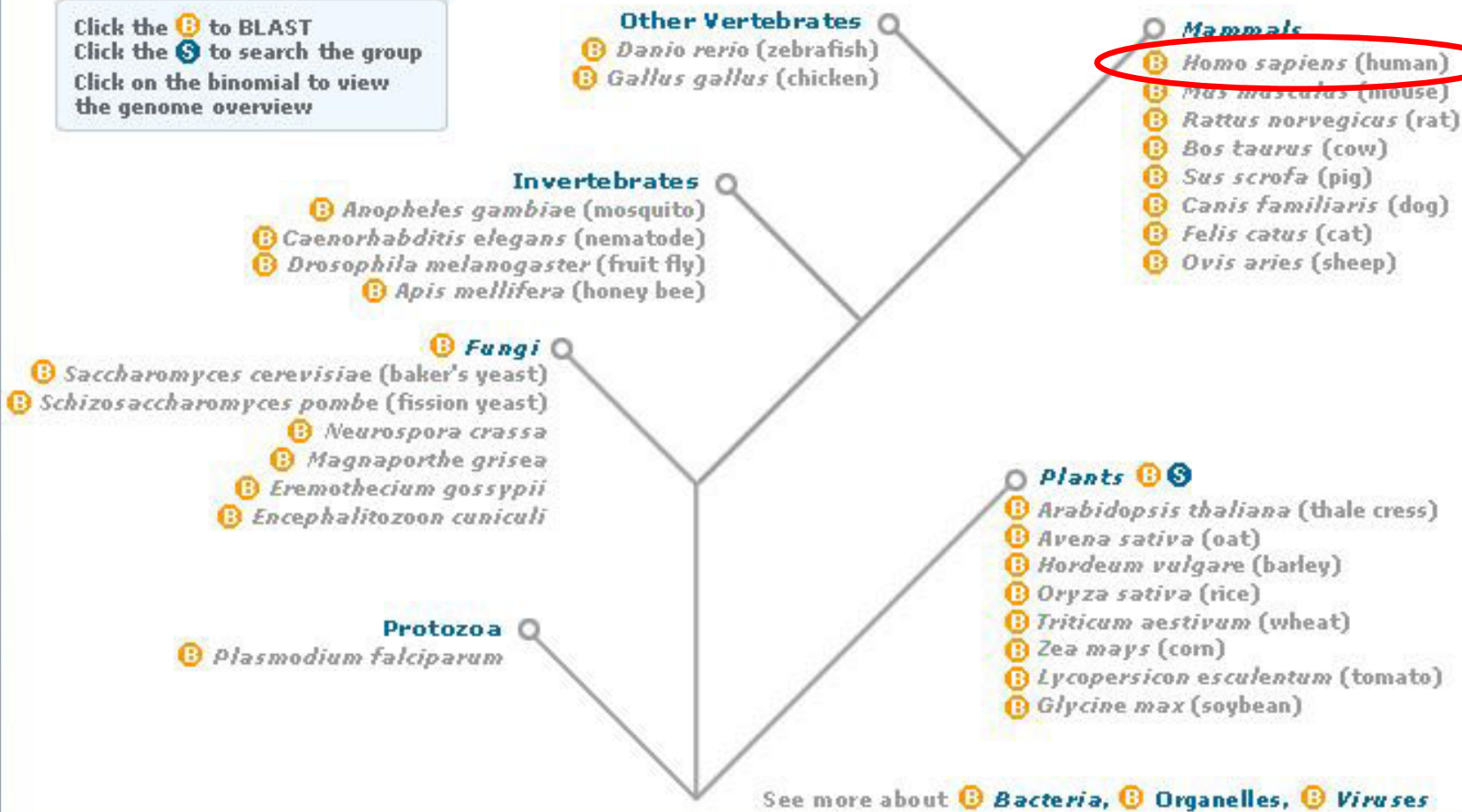


Search for

New! - An advanced search capability is now available for all organisms in the Map Viewer. This feature allows users to restrict the search to specific chromosomes or to search for objects with specific attributes. For instance, you can now search for all genes that are associated with

Homo sapiens (human) をクリック

Click the **B** to BLAST
Click the **S** to search the group
Click on the binomial to view the genome overview



The Map Viewer supports search and display of genomic information by chromosomal position. Regions of interest can be retrieved by text queries (e.g. gene or marker name) or by sequence alignment (BLAST). View results at the whole genome level, and select what to display in more detail. Multiple options exist to configure your display, download data, navigate to related data, and analyze supporting information using the tools provided. [More...](#)

PubMed

Nucleotide

Protein

Genome

Structure

PopSet

Search for

tnfa

on chromosome(s)

assembly

All

Find

Show related entries

Help

FTP

tnfaと入力

Findボタンを押す

MapViewer Home

Map Viewer

Help

Human Maps Help

Mouse Maps Help

NCBI Handbook

Related Resources

Human Genome

Guide

Mouse Genomic

Biology

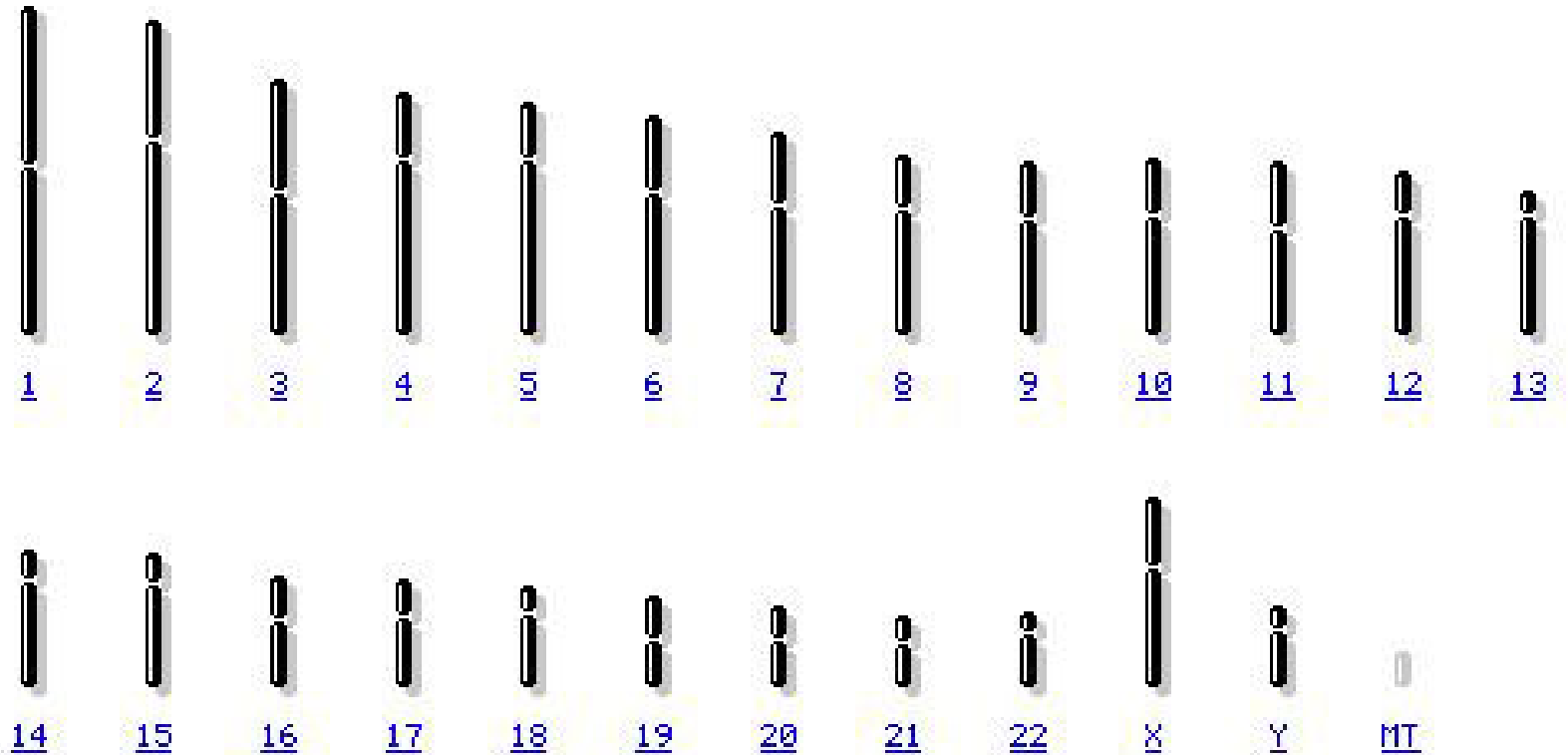
LocusLink

Gene

OMIM

UniGene

Homo_sapiens genome view
build 34 version 3 statistics



[Search for](#)

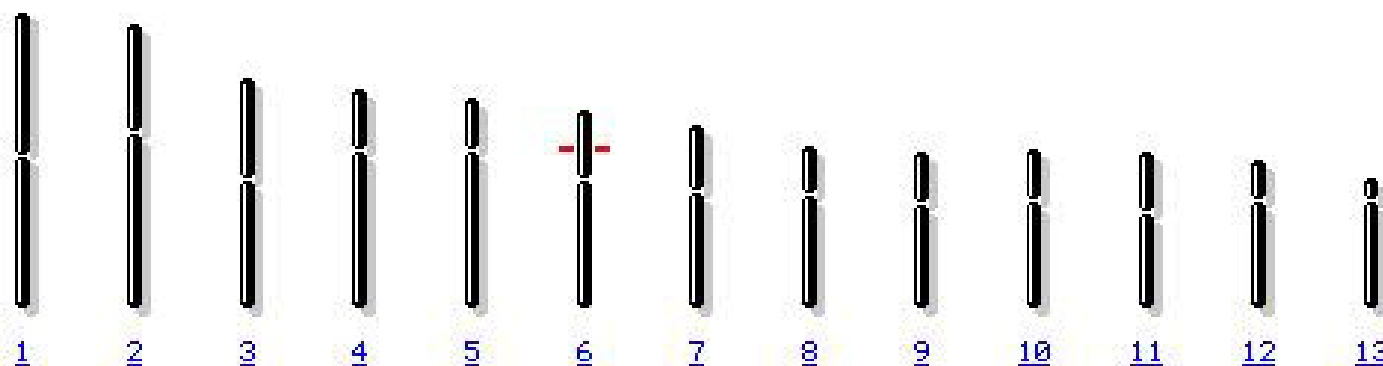
[on chromosome\(s\)](#)

 Show related entries

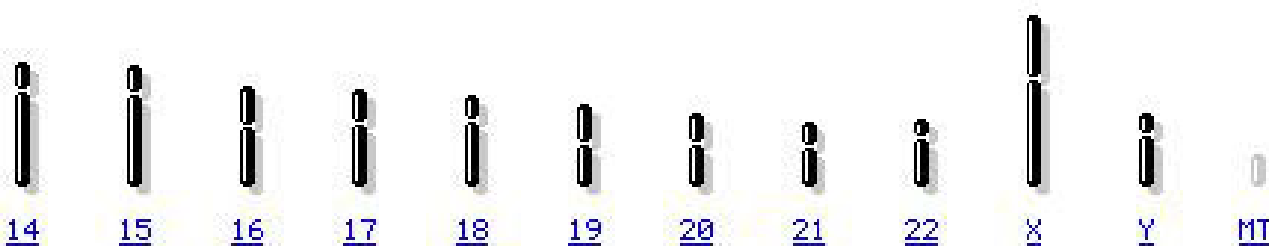
[Help](#)
[FTP](#)
[MapView](#)

Homo_sapiens genome view

build 34 version 3 statistics



Hits:



Hits:

Search results for query "tnfa": 2 hits

| Chr | Match | Map element | Type | Maps |
|-----|---|-------------|------|--|
| 6 | all matches | | | |
| 6 | Malaria, cerebral, susceptibility to; Septic shock, susceptibility... | TNF | MIM | Pheno Morbid |
| 6 | tumor necrosis factor (TNF superfamily, member 2) | TNF | Gene | Genes_seq Genes_cyto |

クリック

[Search](#)[Find](#)[Find in This View](#)[Advanced Search](#)[Map Viewer Home](#)[Map Viewer Help](#)
[Human Maps Help](#)
[FTP](#)[Data As Table View](#)**Maps & Options** [Compress Map](#)

Region Shown:

[Go](#) [out](#)
 [zoom](#)
 [in](#)

You are here:

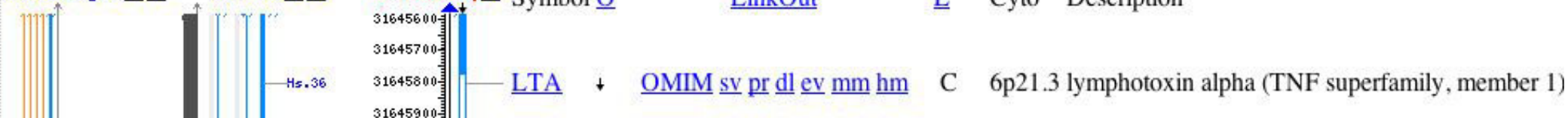
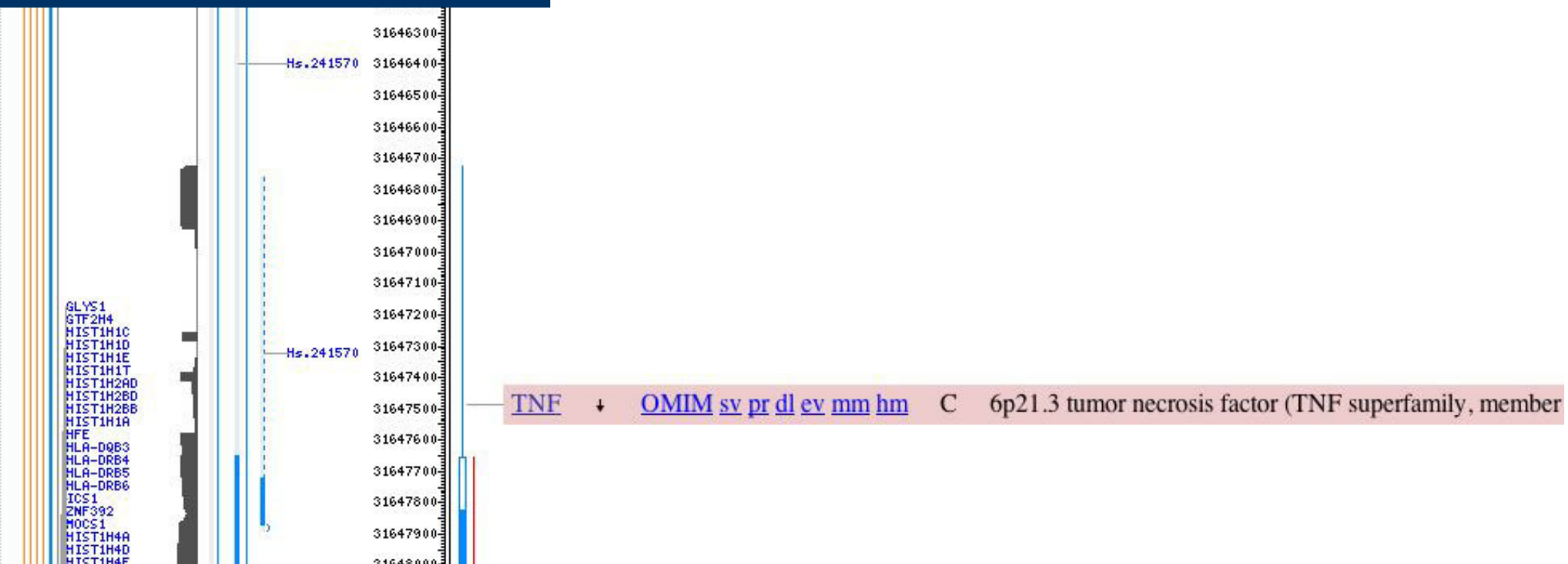
Ideogram [default](#)
 [master](#)***Homo sapiens* build 34.3**[BLAST The Human Genome](#)Chromosome: [1](#) [2](#) [3](#) [4](#) [5](#) [**6**] [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#)Query: tnfa [\[clear\]](#)

Master Map: Genes On Sequence

[Summary of Maps](#)[Maps & Options](#)Region Displayed: 31,645K-31,652K bp [Download/View Sequence/Evidence](#)[Genes_cyto](#) [HsUniG](#) [Genes_seq](#) Symbol [O](#)[LinkOut](#)[E](#)

Cyto

Description

**クリックしてズームアウト**



Map Viewer Home

Homo sapiens build 34.3

BLAST The Human Genome

Map Viewer Help Human Maps Help FTP Data As Table 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: tnfa [clear]

Master Map: Genes On Sequence

Summary

Maps & Options

Region Displayed: 30,794K-32,503K bp

Download/View Sequence/Evidence

クリック

Maps & Options

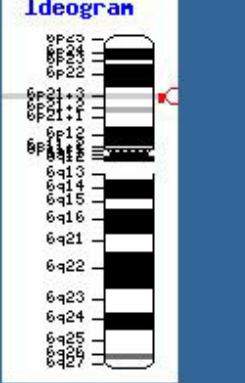
Compress Map

Region Shown:

30,794K 32,503K Go

out zoom in

You are here:



default master

| Gen... | HsUniG | Genes_seq | Symbol | LinkOut | OMIM | sv | pr | dl | ev | mm | hm | C | Chromosome | Description |
|--------|--------|-----------|--------------------------|--|------|----|----|----|----|----|----|---|------------|---|
| | | | FLOT1 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | flotillin 1 |
| | | | DDR1 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | discoidin domain receptor family, member 1 |
| | | | VARS2L | sv pr dl ev mm hm | | | | | | | | C | 6p21 | valyl-tRNA synthetase 2-like |
| | | | PSORSIC1 | sv pr dl ev mm | | | | | | | | C | 6p21.3 | psoriasis susceptibility 1 candidate 1 |
| | | | C6orf18 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | chromosome 6 open reading frame 18 |
| | | | MICA | OMIM sv pr dl ev mm | | | | | | | | C | 6p21.3 | MHC class I polypeptide-related sequence A |
| | | | MICB | OMIM sv pr dl ev mm | | | | | | | | C | 6p21.3 | MHC class I polypeptide-related sequence B |
| | | | BAT1 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | HLA-B associated transcript 1 |
| | | | NFKBIL1 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | nuclear factor of kappa light polypeptide gene enhancer 1 |
| | | | TNF | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | tumor necrosis factor (TNF superfamily, member 1) |
| | | | BAT2 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | HLA-B associated transcript 2 |
| | | | BAT3 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | HLA-B associated transcript 3 |
| | | | BAT5 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | HLA-B associated transcript 5 |
| | | | LY6G6D | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | lymphocyte antigen 6 complex, locus G6D |
| | | | MSH5 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | mutS homolog 5 (E. coli) |
| | | | C6orf27 | sv pr dl ev mm hm | | | | | | | | C | 6p21.31 | chromosome 6 open reading frame 27 |
| | | | VARS2 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | valyl-tRNA synthetase 2 |
| | | | C6orf29 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | chromosome 6 open reading frame 29 |
| | | | BAT8 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.31 | HLA-B associated transcript 8 |
| | | | C2 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | complement component 2 |
| | | | SKIV2L | OMIM sv pr dl ev mm | | | | | | | | C | 6p21 | superkiller viralicidic activity 2-like (S. cerevisiae) |
| | | | C4B | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | complement component 4B |
| | | | TNXB | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | tenascin XB |
| | | | CREBL1 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | cAMP responsive element binding protein-like 1 |
| | | | PPT2 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | palmitoyl-protein thioesterase 2 |
| | | | AGPAT1 | OMIM sv pr dl ev mm hm | | | | | | | | C | 6p21.3 | 1-acylglycerol-3-phosphate O-acyltransferase 1 (liver) |

Genes On Sequence

All Sequence Maps

next

Region Displayed: 30,794K-32,503K bp

[Download/View Sequence/Evidence](#)[Download Data](#)

Total Genes On Chromosome: 1247 [7 not localized]

Genes in Region: 87

| start | stop | Symbol | Orientation | LinkOut | E | Cyto | Description |
|----------|----------|-----------------------------|-------------|--|---|---------|--|
| 30794311 | 30799353 | OK/SW-cl.56 | + | sv pr dl ev mm hm | C | 6p21.32 | beta 5-tubulin |
| 30801665 | 30816615 | FLOT1 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | flotillin 1 |
| 30817139 | 30818486 | IER3 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | immediate early response 3 |
| 30886824 | 30904616 | LOC401247 | - | sv pr dl ev mm | I | 6p21.32 | hypothetical gene supported by AK098012 |
| 30958112 | 30974186 | DDR1 | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | discoidin domain receptor family, member 1 |
| 30982253 | 30988128 | LOC399704 | + | sv pr dl ev mm hm | I | 6p21.32 | hypothetical gene supported by NM_001517 |
| 30988230 | 31000472 | VARS2L | + | sv pr dl ev mm hm | C | 6p21 | valyl-tRNA synthetase 2-like |
| 31005384 | 31010332 | LOC389376 | - | sv pr dl ev mm | I | 6p21.32 | similar to GSGL541 |
| 31025803 | 31028242 | DPCR1 | + | OMIM sv pr dl ev mm hm | C | 6p21.32 | diffuse panbronchiolitis critical region 1 |
| 31057739 | 31063917 | C6orf205 | + | sv pr dl ev mm hm | C | 6p21.32 | chromosome 6 open reading frame 205 |
| 31185246 | 31186578 | C6orf15 | - | sv pr dl ev mm hm | C | 6p21.3 | chromosome 6 open reading frame 15 |
| 31188869 | 31214104 | PSORS1C1 | + | sv pr dl ev mm | C | 6p21.3 | psoriasis susceptibility 1 candidate 1 |
| 31189606 | 31194452 | CDSN | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | corneodesmosin |
| 31211578 | 31213385 | PSORS1C2 | - | sv pr dl ev mm | C | 6p21.3 | psoriasis susceptibility 1 candidate 2 |
| 31216476 | 31231822 | C6orf18 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | chromosome 6 open reading frame 18 |
| 31233529 | 31236784 | TCF19 | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | transcription factor 19 (SC1) |
| 31238369 | 31240302 | POU5F1 | - | OMIM sv pr dl ev mm hm | C | 6p21.31 | POU domain, class 5, transcription factor 1 |
| 31271803 | 31278005 | LOC253018 | + | sv pr dl ev mm | C | 6p21.32 | hypothetical protein LOC253018 |
| 31341277 | 31344599 | HLA-C | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | major histocompatibility complex, class I, C |
| 31426059 | 31429136 | HLA-B | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | major histocompatibility complex, class I, B |
| 31475571 | 31487290 | MICA | + | OMIM sv pr dl ev mm | C | 6p21.3 | MHC class I polypeptide-related sequence A |
| 31535334 | 31537950 | HCP5 | + | OMIM sv pr dl ev mm | C | 6p21.3 | HLA complex P5 |
| 31570311 | 31583243 | MICB | + | OMIM sv pr dl ev mm | C | 6p21.3 | MHC class I polypeptide-related sequence B |
| 31591101 | 31592427 | PIIP9 | - | sv dl ev mm | C | 6p21.1 | peptidylprolyl isomerase (cyclophilin) pseudogene 9 |
| 31601078 | 31602347 | LOC401250 | + | sv pr dl ev mm | I | 6p21.31 | similar to coiled-coil domain 1 protein precursor |
| 31602338 | 31614244 | BAT1 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | HLA-B associated transcript 1 |
| 31616558 | 31618942 | ATP6V1G2 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | ATPase, H+ transporting, lysosomal 13kDa, V1 subunit G isoform 2 |
| 31619686 | 31630919 | NFKBIL1 | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | nuclear factor of kappa light polypeptide gene enhancer in B-cells inhibitor |
| 31644397 | 31646402 | LTA | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | lymphotoxin alpha (TNF superfamily, member 1) |
| 31647654 | 31650416 | TNF | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | tumor necrosis factor (TNF superfamily, member 2) |
| 31652639 | 31654506 | LTB | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | lymphotoxin beta (TNF superfamily, member 3) |
| 31659719 | 31660757 | LST1 | + | OMIM sv pr dl ev mm | C | 6p21.3 | leukocyte specific transcript 1 |
| 31660976 | 31665045 | NCR3 | - | sv pr dl ev mm hm | C | 6p21.3 | natural cytotoxicity triggering receptor 3 |
| 31687317 | 31689082 | AIF1 | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | allograft inflammatory factor 1 |
| 31692874 | 31709815 | BAT2 | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | HLA-B associated transcript 2 |
| 31711095 | 31724722 | BAT3 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | HLA-B associated transcript 3 |
| 31727945 | 31730261 | APOM | + | OMIM sv pr dl ev mm hm | C | 6p21.31 | apolipoprotein M |
| 31730349 | 31732785 | C6orf47 | - | sv pr dl ev mm | C | 6p21.3 | chromosome 6 open reading frame 47 |
| 31734142 | 31737447 | BAT4 | - | OMIM sv pr dl ev mm hm | C | 6p21.3 | HLA-B associated transcript 4 |
| 31737941 | 31742120 | CSNK2B | + | OMIM sv pr dl ev mm hm | C | 6p21.3 | casein kinase 2, beta polypeptide |
| 31742433 | 31744341 | LY6G5B | + | sv pr dl ev mm hm | C | 6p21.3 | lymphocyte antigen 6 complex, locus G5B |

クリック

#Homo sapiens Genome (build 34.3)

#Chromosome: 6

#####

#Map: genes

#Region: 30794477.12..32503592.88

| #start | stop | Symbol | 0 | LinkOut | | pr | dl | ev | mm | hm | Cyto | Description | | |
|----------|----------|-------------|---|---------|------|----|----|----|----|----|------|--------------|---|--|
| 30794311 | 30799353 | OK/SW-cl.56 | | + | | sv | pr | dl | ev | mm | hm | C | 6p21.32 beta 5-tubulin | |
| 30801665 | 30816615 | FLOT1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | flotillin 1 | |
| 30817139 | 30818486 | IER3 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | immediate early response 3 | |
| 30886824 | 30904616 | LOC401247 | | - | | sv | pr | dl | ev | mm | I | 6p21.32 | hypothetical gene supported by AK098012 | |
| 30958112 | 30974186 | DDR1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | discoilin domain receptor family, member 1 | |
| 30982253 | 30988128 | LOC399704 | | + | | sv | pr | dl | ev | mm | hm | I | 6p21.32 | hypothetical gene supported by NM_001517 |
| 30988230 | 31000472 | VAR52L | + | | sv | pr | dl | ev | mm | hm | C | 6p21 | valyl-tRNA synthetase 2-like | |
| 31005384 | 31010332 | LOC389376 | | - | | sv | pr | dl | ev | mm | I | 6p21.32 | similar to GSGL541 | |
| 31025803 | 31028242 | DPCR1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.32 | diffuse panbronchiolitis critical region 1 | |
| 31057739 | 31063917 | C6orf205 | | + | | sv | pr | dl | ev | mm | hm | C | 6p21.32 | chromosome 6 open reading frame 205 |
| 31105246 | 31106570 | C6orf15 | - | | sv | pr | dl | ev | mm | hm | C | 6p21.3 | chromosome 6 open reading frame 15 | |
| 31188869 | 31214104 | PSORS1C1 | | + | | sv | pr | dl | ev | mm | C | 6p21.3 | psoriasis susceptibility 1 candidate 1 | |
| 31189606 | 31194452 | CDSN | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | corneodesmosin | |
| 31211578 | 31213385 | PSORS1C2 | | - | | sv | pr | dl | ev | mm | C | 6p21.3 | psoriasis susceptibility 1 candidate 2 | |
| 31216476 | 31231822 | C6orf18 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | chromosome 6 open reading frame 18 | |
| 31233529 | 31236784 | TCF19 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | transcription factor 19 (TCF19) | |
| 31238369 | 31240302 | POU5F1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.31 | POU domain, class 5, transcription factor 1 | |
| 31271803 | 31278005 | LOC253018 | | + | | sv | pr | dl | ev | mm | C | 6p21.32 | hypothetical protein LOC253018 | |
| 31341277 | 31344599 | HLA-C | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | major histocompatibility complex, class I, C | |
| 31426059 | 31429136 | HLA-B | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | major histocompatibility complex, class I, B | |
| 31475571 | 31487290 | MICA | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | MHC class I polypeptide-related sequence A | |
| 31535334 | 31537950 | HCP5 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA complex P5 | |
| 31570311 | 31583243 | MICB | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | MHC class I polypeptide-related sequence B | |
| 31591101 | 31592427 | PP1P9 | - | | sv | | dl | ev | mm | hm | C | 6p21.1 | peptidylprolyl isomerase (cyclophilin) pseudogene 9 | |
| 31601078 | 31602347 | LOC401250 | | + | | sv | pr | dl | ev | mm | I | 6p21.31 | similar to coiled-coil domain 1 protein precursor | |
| 31602338 | 31614244 | BAT1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA-B associated transcript 1 | |
| 31616558 | 31618942 | ATP6V1G2 | | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | ATPase, H+ transporting, lysosomal 13kDa, V1 subunit G isoform 2 |
| 31619686 | 31630919 | NFKBIL1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | nuclear factor of kappa light polypeptide gene enhancer in B-cells inhibitor-like 1 | |
| 31644397 | 31646402 | LTA | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphotoxin alpha (TNF superfamily, member 1) | |
| 31647654 | 31650416 | TNF | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | tumor necrosis factor (TNF superfamily, member 2) | |
| 31652639 | 31654506 | LTB | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphotoxin beta (TNF superfamily, member 3) | |
| 31659719 | 31660757 | LST1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | leukocyte specific transcript 1 | |
| 31660976 | 31665045 | NCR3 | - | | sv | pr | dl | ev | mm | hm | C | 6p21.3 | natural cytotoxicity triggering receptor 3 | |
| 31687317 | 31689082 | A1F1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | allograft inflammatory factor 1 | |
| 31692874 | 31709815 | BAT2 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA-B associated transcript 2 | |
| 31711095 | 31724722 | BAT3 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA-B associated transcript 3 | |
| 31727945 | 31730261 | APOM | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.31 | apolipoprotein M | |
| 31730349 | 31732705 | C6orf47 | - | | sv | pr | dl | ev | mm | hm | C | 6p21.3 | chromosome 6 open reading frame 47 | |
| 31734142 | 31737447 | BAT4 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA-B associated transcript 4 | |
| 31737941 | 31742120 | CSNK2B | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | casein kinase 2, beta polypeptide | |
| 31742433 | 31744341 | LY6G5B | + | | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphocyte antigen 6 complex, locus G5B | |
| 31748739 | 31756092 | LY6G5C | - | | sv | pr | dl | ev | mm | hm | C | 6p21.31 | lymphocyte antigen 6 complex, locus G5C | |
| 31759016 | 31775375 | BAT5 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA-B associated transcript 5 | |
| 31778962 | 31789975 | LY6G6D | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphocyte antigen 6 complex, locus G6D | |
| 31784397 | 31785873 | LY6G6E | - | | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphocyte antigen 6 complex, locus G6E | |
| 31790705 | 31793790 | LY6G6C | - | | sv | pr | dl | ev | mm | hm | C | 6p21.31 | lymphocyte antigen 6 complex, locus G6C | |
| 31795438 | 31797128 | C6orf25 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.31 | chromosome 6 open reading frame 25 | |
| 31799094 | 31802316 | DDAH2 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | dimethylarginine dimethylaminohydrolase 2 | |
| 31802640 | 31800590 | CLIC1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p22.1-p21.2 | chloride intracellular channel 1 | |
| 31812050 | 31836905 | MSH5 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | mutS homolog 5 (E. coli) | |
| 31835109 | 31836883 | LOC401251 | | + | | sv | pr | dl | ev | mm | hm | E | 6p21.31 | similar to NG23 |
| 31837793 | 31848836 | C6orf27 | - | | sv | pr | dl | ev | mm | hm | C | 6p21.31 | chromosome 6 open reading frame 27 | |
| 31849575 | 31867792 | VAR52 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | valyl-tRNA synthetase 2 | |
| 31869454 | 31879025 | LSM2 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | LSM2 homolog, U6 small nuclear RNA associated (S. cerevisiae) | |
| 31881604 | 31887094 | HSPA1L | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | heat shock 70kDa protein 1-like | |
| 31887624 | 31890006 | HSPA1A | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | heat shock 70kDa protein 1A | |
| 31899816 | 31902327 | HSPA1B | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | heat shock 70kDa protein 1B | |
| 31906905 | 31911831 | C6orf48 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | chromosome 6 open reading frame 48 | |

| | A | B | C | D | E | F | G | H | I | J | K | L | M | N |
|----|----------|----------|-------------|---|---------|----|----|----|----|----|----|---|---------|---|
| 1 | #start | stop | Symbol | O | LinkOut | | | | | | | E | Cyto | Description |
| 2 | 30794311 | 30799353 | OK/SW-cl.56 | + | | sv | pr | dl | ev | mm | hm | C | 6p21.32 | beta 5-tubulin |
| 3 | 30801665 | 30816615 | FLOT1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | flotillin 1 |
| 4 | 30817139 | 30818486 | IER3 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | immediate early response 3 |
| 5 | 30886824 | 30904616 | LOC401247 | - | | sv | pr | dl | ev | mm | | I | 6p21.32 | hypothetical gene supported by AK098012 |
| 6 | 30958112 | 30974186 | DDR1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | discoidin domain receptor family, member 1 |
| 7 | 30982253 | 30988128 | LOC399704 | + | | sv | pr | dl | ev | mm | hm | I | 6p21.32 | hypothetical gene supported by NM_001517 |
| 8 | 30988230 | 31000472 | VARS2L | + | | sv | pr | dl | ev | mm | hm | C | 6p21 | valyl-tRNA synthetase 2-like |
| 9 | 31005384 | 31010332 | LOC389376 | - | | sv | pr | dl | ev | mm | | I | 6p21.32 | similar to GSGL541 |
| 10 | 31025803 | 31028242 | DPCR1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.32 | diffuse panbronchiolitis critical region 1 |
| 11 | 31057739 | 31063917 | C6orf205 | + | | sv | pr | dl | ev | mm | hm | C | 6p21.32 | chromosome 6 open reading frame 205 |
| 12 | 31185246 | 31186578 | C6orf15 | - | | sv | pr | dl | ev | mm | hm | C | 6p21.3 | chromosome 6 open reading frame 15 |
| 13 | 31188869 | 31214104 | PSORS1C1 | + | | sv | pr | dl | ev | mm | | C | 6p21.3 | psoriasis susceptibility 1 candidate 1 |
| 14 | 31189606 | 31194452 | CDSN | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | corneodesmosin |
| 15 | 31211578 | 31213385 | PSORS1C2 | - | | sv | pr | dl | ev | mm | | C | 6p21.3 | psoriasis susceptibility 1 candidate 2 |
| 16 | 31216476 | 31231822 | C6orf18 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | chromosome 6 open reading frame 18 |
| 17 | 31233529 | 31236784 | TCF19 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | transcription factor 19 (SC1) |
| 18 | 31238369 | 31240302 | POU5F1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.31 | POU domain, class 5, transcription factor 1 |
| 19 | 31271803 | 31278005 | LOC253018 | + | | sv | pr | dl | ev | mm | | C | 6p21.32 | hypothetical protein LOC253018 |
| 20 | 31341277 | 31344599 | HLA-C | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | major histocompatibility complex, class I, C |
| 21 | 31426059 | 31429136 | HLA-B | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | major histocompatibility complex, class I, B |
| 22 | 31475571 | 31487290 | MICA | + | OMIM | sv | pr | dl | ev | mm | | C | 6p21.3 | MHC class I polypeptide-related sequence A |
| 23 | 31535334 | 31537950 | HCP5 | + | OMIM | sv | pr | dl | ev | mm | | C | 6p21.3 | HLA complex P5 |
| 24 | 31570311 | 31583243 | MICB | + | OMIM | sv | pr | dl | ev | mm | | C | 6p21.3 | MHC class I polypeptide-related sequence B |
| 25 | 31591101 | 31592427 | PIIP9 | - | | sv | | dl | ev | mm | | C | 6p21.1 | peptidylprolyl isomerase (cyclophilin) pseudogene 9 |
| 26 | 31601078 | 31602347 | LOC401250 | + | | sv | pr | dl | ev | mm | | I | 6p21.31 | similar to coiled-coil domain 1 protein precursor |
| 27 | 31602338 | 31614244 | BAT1 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | HLA-B associated transcript 1 |
| 28 | 31616558 | 31618942 | ATP6V1G2 | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | ATPase, H+ transporting, lysosomal 13kDa, V1 subunit G isoform 2 |
| 29 | 31619686 | 31630919 | NFKBIL1 | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | nuclear factor of kappa light polypeptide gene enhancer in B-cells inhibitor-like 1 |
| 30 | 31644397 | 31646402 | LTA | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphotoxin alpha (TNF superfamily, member 1) |
| 31 | 31647654 | 31650416 | TNF | + | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | tumor necrosis factor (TNF superfamily, member 2) |
| 32 | 31652639 | 31654506 | LTB | - | OMIM | sv | pr | dl | ev | mm | hm | C | 6p21.3 | lymphotoxin beta (TNF superfamily, member 3) |

核内受容体について

細胞質

核

リガンド(脂溶性)

核内受容体

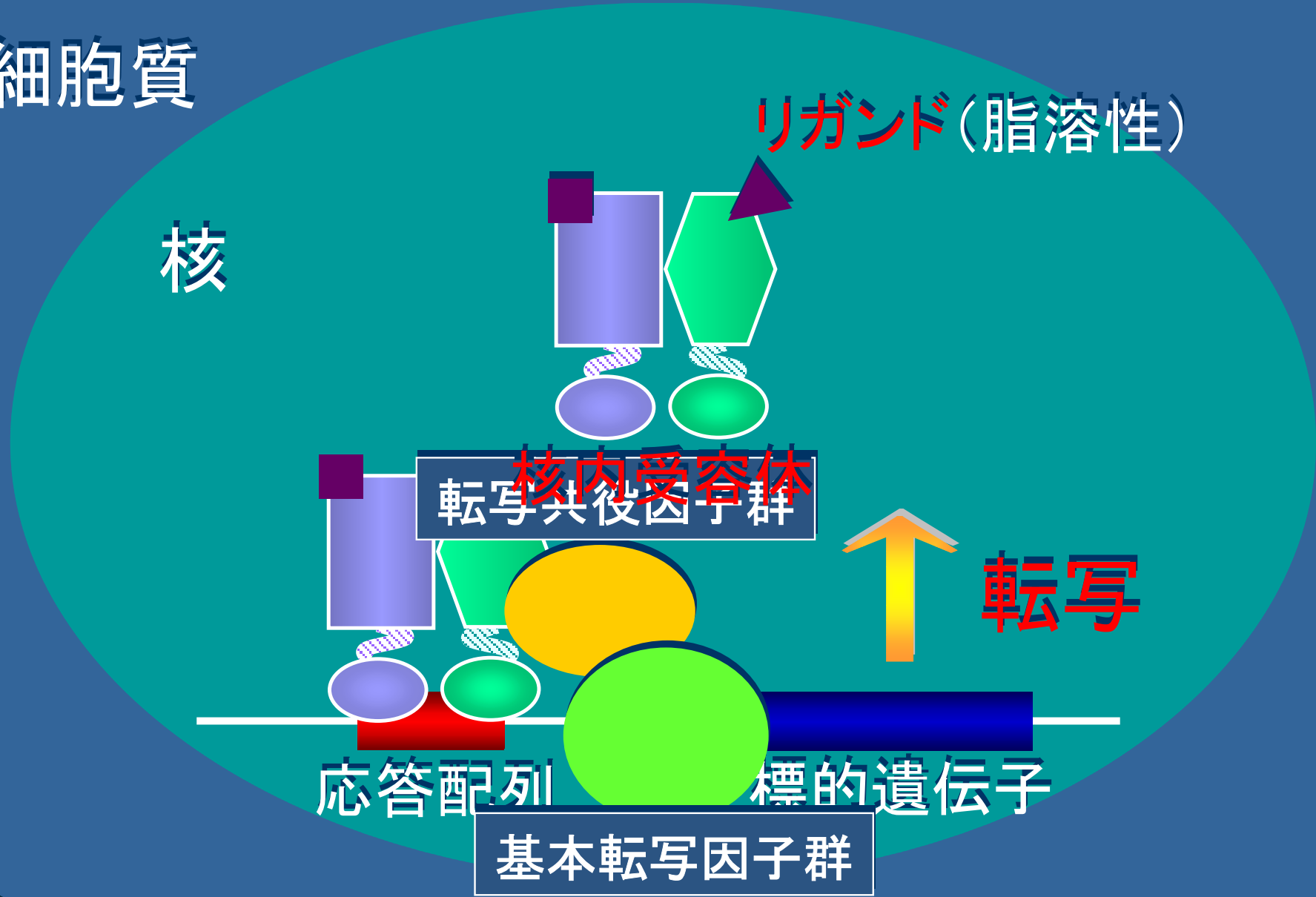
転写共役因子群

転写

応答配列

標的遺伝子

基本転写因子群



配列の収集

- 遺伝子の配列および上流、下流配列
- UCSCのゲノムブラウザを使用
 - <http://genome.ucsc.edu/>
 - Human hg17 assembly (May 2004 : based on NCBI Build 35)
 - 他にhg16(July 2003,based on NCBI Build 34), hg15(April 2003,based on NCBI Build 33)が選択可能
 - ABCA1遺伝子を例に説明

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Sorter

Blat

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About the UCSC Genome Bioinformatics Site

This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also shows the reference sequence for 13 species and provides a portal to the ENCODE project.

We encourage you to explore these sequences with our tools. The Genome Browser zooms and scrolls over chromosomes and tracks from annotators worldwide. The Gene Sorter shows expression, homology and other information on groups of genes that can be used to quickly map your sequence to the genome. The Table Browser provides convenient access to the underlying database.

News

10 August 2004 - *Anopheles gambiae* Genome Browser Released

The UCSC Genome Bioinformatics Group has released a Genome Browser and Blat server for the *Anopheles gambiae* v. 2.0 (Feb. 2003). This assembly -- UCSC version anoGam1 -- was produced by the International Anopheles Genome Project and the UCSC Genome Bioinformatics Group.

The MOZ2 assembly is a 10x whole genome shotgun assembly. The assembled portion of the genome is about 278 Mbp. The genome is composed of 13,494 unique scaffolds, the largest scaffold being 23.1 Mbp. Approximately 85% of the sequence has been assigned to chromosomes. Chromosomes chr2L, chr2R, chr3L, chr3R, and chrX are represented by 13, 49, 42, 28, and 10 large scaffolds respectively. No scaffolds are assigned to the Y chromosome. The unassigned scaffolds, concatenated together in arbitrary order, can be found in the artificial unknown chromosome.

For more information about the initial *A. gambiae* assembly, see Holt et al. (2002), [The Genome Sequence of the Malaria Mosquito *Anopheles gambiae*](#), *Science* 298:129-149.

Downloads of the anoGam1 data and annotations may be obtained from the UCSC Genome Browser [FTP server](#) or [Download Center](#). The annotation tracks were generated by UCSC and collaborators worldwide.

Many thanks to the International Anopheles Genome Project and Ensembl for providing the sequence of this genome. The genome browser and Blat server are maintained by Angie Hinrichs, Galt Barber, Donna Karolchik, and sysadmins Paul Tatarisky and Jorge Garcia. See the [Credits](#) page for a list of organizations and individuals who contributed to this release.

29 July 2004 - Zv3 Zebrafish Genome Browser Available: The Zv3 Zebrafish genome assembly is now available on the UCSC Genome Browser and Blat server. [Read more.](#)

クリック

Human Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#).
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| | | | | |
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| genome | assembly | position | image width | |
| Human | May 2004 | abca1 | 620 | Submit |

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abca1と入力

About the Human hg17 assembly

The May 2004 human reference sequence is based on NCBI Build 35 and was produced by the International Human Genome Sequencing Consortium.

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, or a cytological band, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.

| Request: | Genome Browser Response: |
|---------------------|--|
| chr7 | Displays all of chromosome 7 |
| 20p13 | Displays region for band p13 on chr 20 |
| chr3:1-1000000 | Displays first million bases of chr 3, counting from p arm telomere |
| D16S3046 | Displays region around STS marker D16S3046 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well. |
| RH18061;RH80175 | Displays region between STS markers RH18061;RH80175. Includes 100,000 bases on each side as well. |
| AA205474 | Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17 |
| AC008101 | Displays region of clone with GenBank accession AC008101 |
| AF083811 | Displays region of mRNA with GenBank accession number AF083811 |
| PRNP | Displays region of genome with HUGO identifier PRNP |
| NM_017414 | Displays the region of genome with RefSeq identifier NM_017414 |
| NP_059110 | Displays the region of genome with protein accession number NP_059110 |
| pseudogene mRNA | Lists transcribed pseudogenes, but not cDNAs |
| homeobox caudal | Lists mRNAs for caudal homeobox genes |
| zinc finger | Lists many zinc finger mRNAs |
| kruppel zinc finger | Lists only kruppel-like zinc fingers |
| huntington | Lists candidate genes associated with Huntington's disease |
| zahler | Lists mRNAs deposited by scientist named Zahler |
| Evans,J.E. | Lists mRNAs deposited by co-author J.E. Evans |

Use this last format for author queries. Although GenBank requires the search format *Evans JE*, internally it uses the format *Evans,J.E.*.

Known Genes

[ABCA1 at chr9:104726262-104770073](#) - (AF258627) ATP binding cassette transporter 1 (Fragment).
[ABCA1 at chr9:104622849-104769967](#) - (AF285167) ATP-binding cassette, sub-family A (ABC1), member 1
[ABCA1 at chr9:104697401-104769991](#) - (AK024328) Hypothetical protein FLJ14266.
[ABCA10 at chr17:64655743-64736391](#) - (AL832004) Hypothetical protein DKFZp451N043.
[ABCA10 at chr17:64655771-64752582](#) - (AY247065) ATP-binding cassette sub-family A member 10.
[ABCA10 at chr17:64655745-64659881](#) - (BC031026) Hypothetical protein (Fragment).
[ABCA10 at chr17:64656439-64664651](#) - (BC051320) Similar to ATP-binding cassette, sub-family A (ABC1), member 8 (Fragment).
[ABCA12 at chr2:215622806-215739890](#) - (AF418105) ATP-binding cassette, sub-family A (ABC1), member 12
[ABCA12 at chr2:215622534-215637694](#) - (AL080207) Hypothetical protein (Fragment).
[ABCA12 at chr2:215622851-215722316](#) - (AY033486) ATP-binding cassette transporter family A member 12.
[ABCA12 at chr2:215621772-215828657](#) - (AY219711) ABCA12 transporter subfamily A.
[ABCA13 at chr7:48098270-48462457](#) - (AF501281) ATP binding cassette transporter A13 (Fragment).
[ABCA13 at chr7:48090363-48093582](#) - (AK091195) Hypothetical protein FLJ33876.
[ABCA13 at chr7:48337073-48464303](#) - (AK091270) Hypothetical protein FLJ33951.
[ABCA13 at chr7:48015103-48089853](#) - (AK126220) CDNA FLJ44232 fis, clone THYMU3006811, weakly similar to Homo sapiens ATP-binding cas
[ABCA13 at chr7:48271920-48464353](#) - (AK128516) CDNA FLJ46673 fis, clone TRACH3009061, weakly similar to ATP-binding cassette, sub-fa
[ABCA13 at chr7:48015103-48464352](#) - (AY204751) ABC A13.

RefSeq Genes

クリック

[ABCA1 at chr9:104622837-104769991](#) - (NM_005502) ATP-binding cassette, sub-family A member 1
[ABCA10 at chr17:64655743-64752582](#) - (NM_080282) ATP-binding cassette, sub-family A, member 10
[ABCA12 at chr2:215621772-215722316](#) - (NM_015657) ATP-binding cassette, sub-family A, member 12
[ABCA12 at chr2:215621772-215828657](#) - (NM_173076) ATP-binding cassette, sub-family A, member 12
[ABCA13 at chr7:48015103-48464352](#) - (NM_152701) ATP binding cassette gene, sub-family A (ABC1),

Human Aligned mRNA Search Results

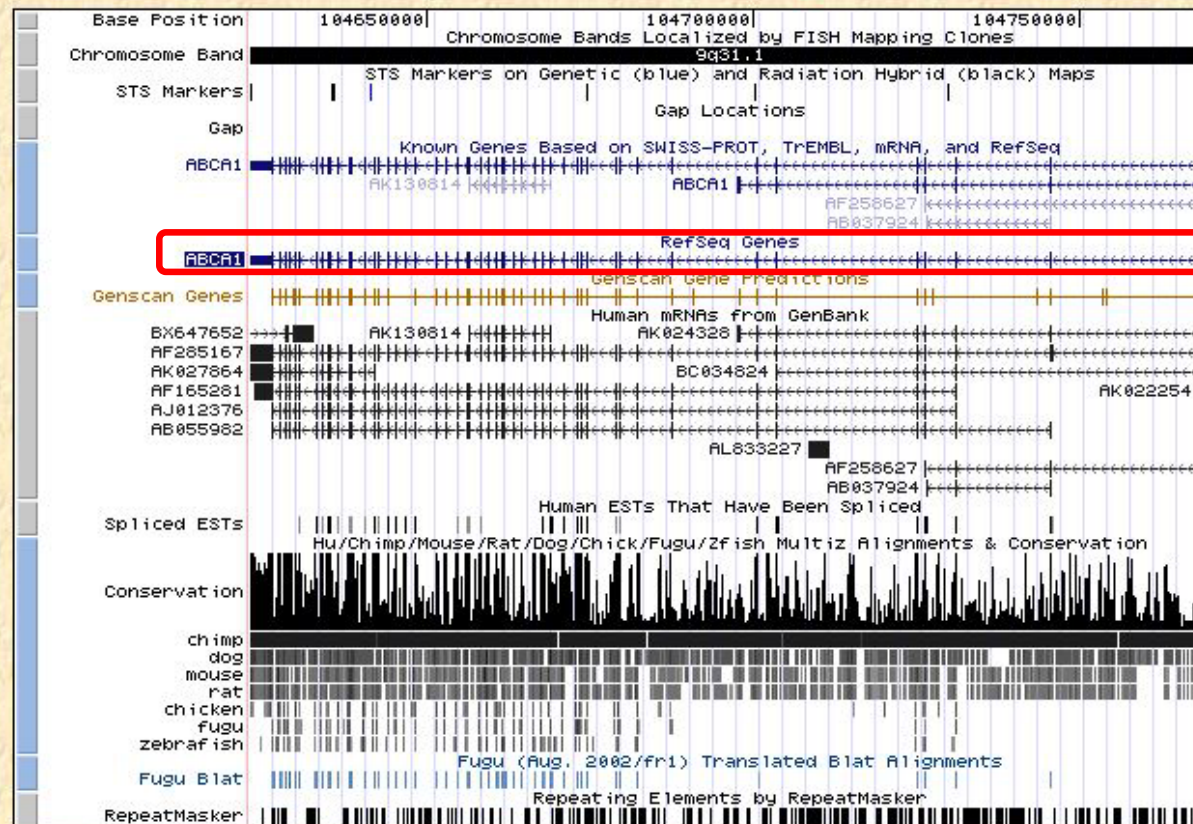
[BC051320](#) - Homo sapiens ATP-binding cassette, sub-family A (ABC1), member 10, mRNA (cDNA clone IMAGE:6249210), partial cds.
[BC031026](#) - Homo sapiens ATP-binding cassette, sub-family A (ABC1), member 10, mRNA (cDNA clone IMAGE:4712782), partial cds.
[AY219711](#) - Homo sapiens ABCA12 transporter subfamily A mRNA, complete cds.
[AB055982](#) - Homo sapiens mRNA for ABCA1, complete cds.
[AY247065](#) - Homo sapiens ATP-binding cassette sub-family A member 10 (ABCA10) mRNA, complete cds.
[AY033486](#) - Homo sapiens ATP-binding cassette transporter family A member 12 (ABCA12) mRNA, complete cds.
[AF501281](#) - Homo sapiens ATP binding cassette transporter A13 (ABCA13) mRNA, partial cds.
[AF258627](#) - Homo sapiens ATP binding cassette transporter 1 (ABCA1) mRNA, partial cds.
[AF285167](#) - Homo sapiens ATP-binding cassette transporter 1 (ABCA1) mRNA, complete cds.
[AK131360](#) - Homo sapiens cDNA FLJ16398 fis, clone TRACH3000420, weakly similar to Homo sapiens ATP-binding cassette transporter fam
[AK123578](#) - Homo sapiens cDNA FLJ41584 fis, clone CTONG2020445, highly similar to Homo sapiens ATP-binding cassette transporter fam

Non-Human Aligned mRNA Search Results

[AF420436](#) - Mus musculus, Abca12
[AY032590](#) - Arabidopsis thaliana, ATP-binding cassette transporter AtABCA1
[AB109217](#) - Arabidopsis thaliana, AtABCA1
[AY243472](#) - Mus musculus, ATP-binding cassette transporter sub-family A
[AY243471](#) - Mus musculus, ATP-binding cassette transporter sub-family A
[AY243470](#) - Mus musculus, ATP-binding cassette transporter sub-family A
[AY208182](#) - Rattus norvegicus, ATP-binding cassette 1
[AY160971](#) - Mus musculus, ATP-binding cassette transporter
[AF362377](#) - Gallus gallus, ATP-binding cassette transporter 1

UCSC Genome Browser on Human May 2004 Assembly

move <<<< << < > >> >>>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x
 position chr9:104,622,837-104,769,991 size 147,155 bp. image width: 620 jump



クリック

move start Click on a feature for details. Click on base position to zoom in move end
 < 2.0 > around cursor. Click on left mini-buttons for track-specific options < 2.0 >

reset all hide all Chromosome Guidelines Labels: left center refresh

Use drop down controls below and press refresh to alter tracks displayed.
 Tracks with lots of items will automatically be displayed in more compact modes.

Mapping and Sequencing Tracks

| | | | | |
|----------------------------------|---------------------------------|-----------------------------|-----------------------------|-------------------------------|
| Base Position | Chromosome Band | STS Markers | FISH Clones | Recomb Rate |
| full ▾ | dense ▾ | dense ▾ | hide ▾ | hide ▾ |
| Map Contigs | Assembly | Gap | Coverage | BAC End Pairs |
| hide ▾ | hide ▾ | dense ▾ | hide ▾ | hide ▾ |
| Fosmid End Pairs | GC Percent | | | |
| hide ▾ | hide ▾ | | | |

Genes and Gene Prediction Tracks

RefSeq Gene

RefSeq Gene ABCA1

RefSeq: [NM_005502](#) Status: **Reviewed**

CDS: 3' complete

OMIM: [600046](#)

LocusLink: [19](#)

Entrez Gene: [19](#)

PubMed on Gene: [ABCA1](#)

PubMed on Product: [ATP-binding cassette sub-family A member 1](#)

GeneLynx [ABCA1](#)

GeneCards: [ABCA1](#)

AceView: [ABCA1](#)

Stanford SOURCE: [NM_005502](#)

Summary of ABCA1

The membrane-associated protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intracellular membranes. ABC genes are divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This protein is a member of the ABC1 subfamily. Members of the ABC1 subfamily comprise the only major ABC subfamily found exclusively in multicellular eukaryotes. With cholesterol as its substrate, this protein functions as a cholesterol efflux pump in the cellular lipid removal pathway. Mutations in this gene have been associated with Tangier's disease and familial high-density lipoprotein deficiency.

mRNA/Genomic Alignments

| SIZE | IDENTITY | CHROMOSOME | STRAND | START | END | QUERY | START | END | TOTAL |
|-----------------------|------------------------|-------------------|-------------------|---------------------------|---------------------------|---------------------------|-------------------|-----------------------|-----------------------|
| 10412 | 100.0% | 9 | - | 104622838 | 104769991 | NM_005502 | 1 | 10412 | 10412 |

Position: [chr9:104622838-104769991](#)

Band: 9q31.1

Genomic Size: 147154

Strand: -

Links to sequence:

[クリック](#)

- [Predicted Protein](#)
- [mRNA Sequence](#) may be different from the genomic sequence.
- [Genomic Sequence](#) from assembly

Genomic Sequence Near Gene

Get Genomic Sequence Near Gene

Note: if you would prefer to get DNA for more than one feature of this track at a time, try the [Table Browser](#): perform an Advanced Query and select FASTA as the output format.

Sequence Retrieval Region Options:

Promoter/Upstream by bases

5' UTR Exons

CDS Exons

3' UTR Exons

Introns

Downstream by bases

One FASTA record per gene.

One FASTA record per region (exon, intron, etc.) with extra bases upstream (5') and extra downstream (3')

Split UTR and CDS parts of an exon into separate FASTA records

Note: if a feature is close to the beginning or end of a chromosome and upstream/downstream bases are added, they may be truncated in order to avoid extending past the edge of the chromosome.

Sequence Formatting Options:

Exons in upper case, everything else in lower case.

CDS in upper case, UTR in lower case.

All upper case.

All lower case.

Mask repeats: to lower case to N

チェックを入れて、10000と入力

クリック

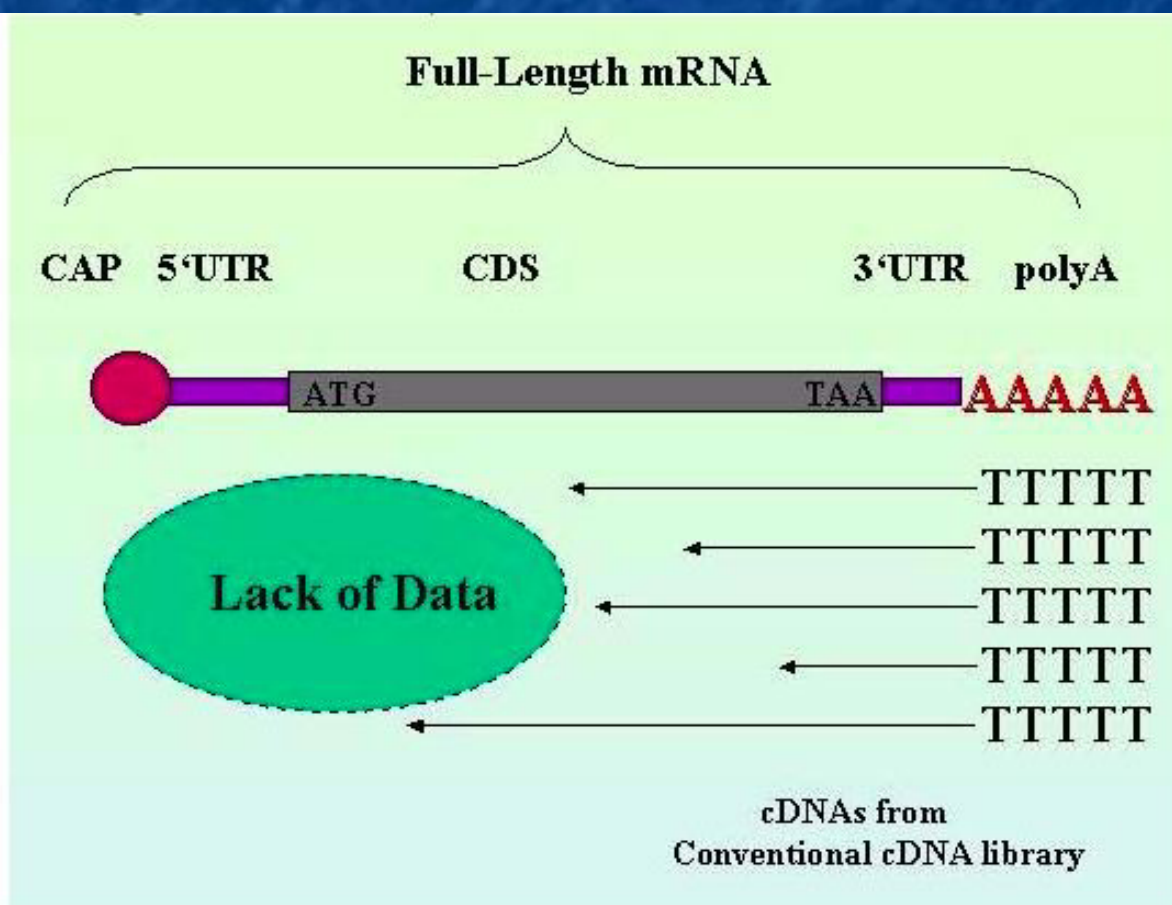
プロモータ配列収集法 2

- DBTSS (DataBase of Transcriptional Start Sites)
 - Release 3.0 (September, 2003)
 - Based by UCSC hg13
 - 先ほどと同じ、*ABCA1*遺伝子を例に説明
 - <http://dbtss.hgc.jp/index.html>

DBTSSの特徴

- mRNA5'端に存在するキャップ構造を特異的に選別する**オリゴキャッピング法**を用いて、完全長cDNAライブラリを構築
- この完全長cDNAライブラリとヒトゲノム配列とを比較し、ゲノム上への転写開始点マッピングをし、公開している
- 最近ではヒトとマウスのプロモータ配列の比較が出来る
- dbSNPデータベースをもとにしたSNPのマッピング

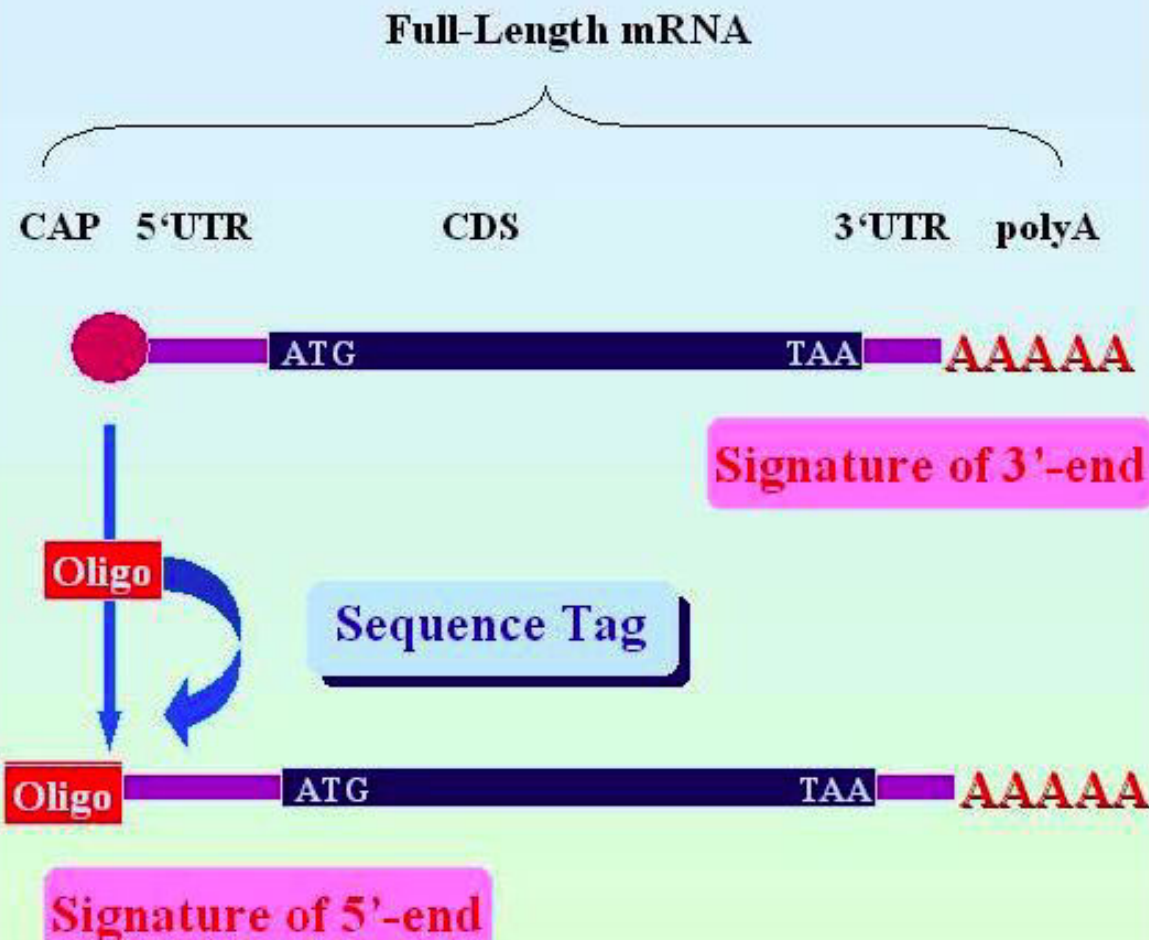
従来のcDNA作成技術の欠点



- 5'末端が欠失していることが多い
 - 逆転写酵素が途中で鋳型mRNAから脱落
 - 5'末端が加水分解により失われてしまったmRNAが鋳型として用いられた

オリゴキャッピング法

Introduction of Sequence Tag



- 5'末端のキャップ構造を合成オリゴで置換する
 - 完全な5'末端を持つ mRNAを選別
 - 完全長cDNAライブラリを構築可能

Database Search

Keyword Search

Species:

Category:

RefSeq ID (NM_) (e.g. 123)

Keyword:

クリック

(May, 2003)

(Jan 10, 2003)

(May, 2003)

(May, 2003)

Example:

Release 3.0 (September, 2003)

Based by UCSC hg13

We recommend to use the Internet Explorer 5.0 or later for visiting our database.

Mirror site [Poland](#) [Germany](#)

Database

ANSFAC links will be implemented shortly !

DBTSS: Database of Transcriptional Start Sites

Current version is based by UCSC hg13

ABSTRACT

About this Database

Based on 400,225 and 580,209 human and mouse [full-length cDNA sequences](#), DBTSS contains exact information of the genomic positions of the transcriptional start sites and the adjacent promoters for 8,793 and 6,875 human and mouse genes, respectively. Of these, 3,324 could be paired as mutually homologous genes between human and mouse and their promoters could be compared with each other. Using this information, DBTSS enables users to investigate what kind of sequence elements are contained in the promoters of their genes of interest and which of them are conserved between human and mouse. Also, users can search for promoters containing putative binding sites of particular transcription factors (TFs). For this, complex queries are supported on recorded features, so that users can optimize the search conditions by specifying arbitrary combinations and cut-offs for each TF and whether or not the sequence conservation of their binding sites between human and mouse should be considered. DBTSS should serve as a unique database which meets the practical requirements of experimental biologists and should lay firm foundation for deciphering the transcriptional network of human genes.

English | Japanese

Database Search

Keyword Search

Species:

Category:

RefSeq ID (NM_) (e.g., 123)

Keyword:

(May, 2003)

(Jan 10, 2003)

(May, 2003)

(May, 2003)

Comparative Analysis

H.sapiens vs M.musculus

Example:
NM_000993 vs NM_053257

(May, 2003)

SNP Search

<RefSNP ID>

rs

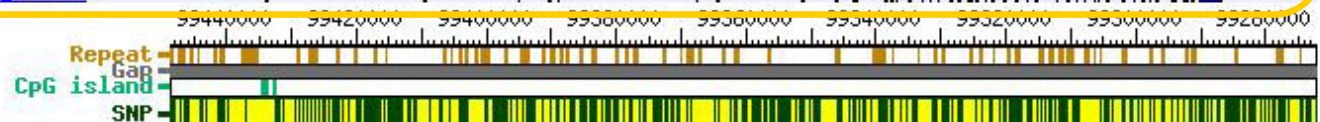
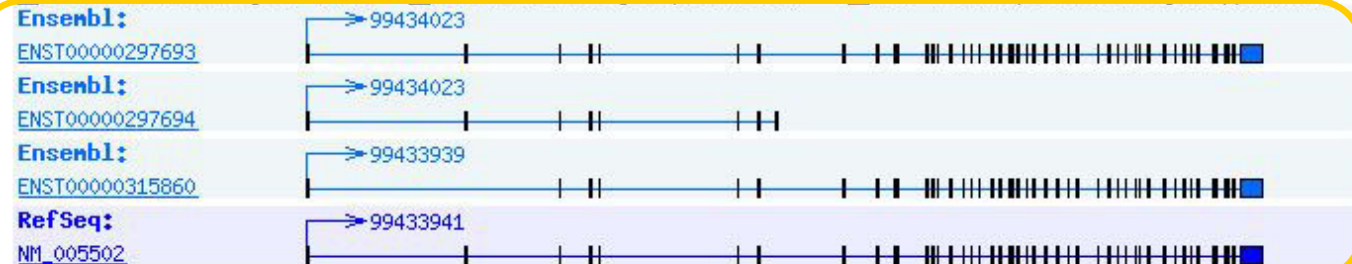
Gene Structure

Chromosome: chr9 Strand: Minus Sequence:

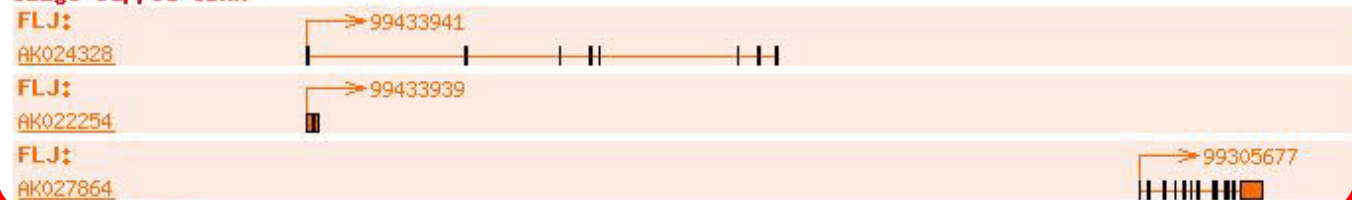


Ensembl, RefSeq

■ : Untranslated Region (UTR) ■ : Protein Coding Sequence (CDS) ■ : 5' end Sequence of Oligo-capped cDNA

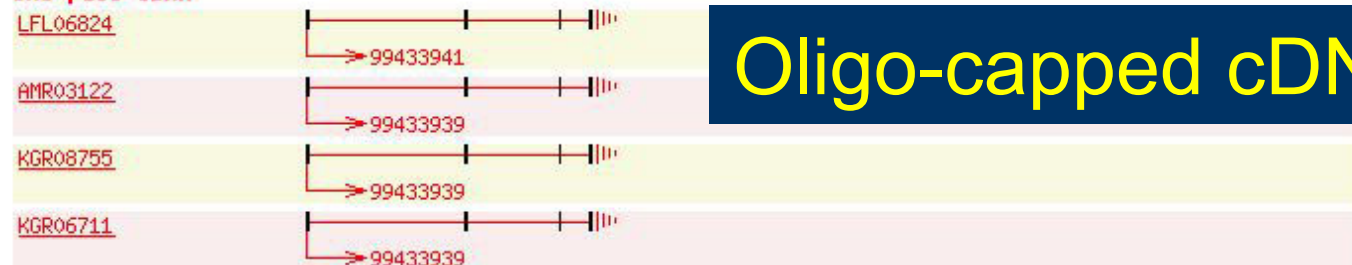


Oligo-capped cDNA



Oligo-capped cDNA

One-pass cDNA



Species: H.sapiens Chromosome: chr9 Range: 99433886-99433992 Strand: -

Excision of promoter sequence

upstream: bp (1-10000)
downstream: bp (1-10000)
designated as 0:

上流1000, 下流100と入力

submitをクリック

Promoter Sequence

| | |
|----------------------|------------------------------|
| Species | <i>H. sapiens</i> |
| Chrom, Range, Strand | chr9, 99433841-99434941, (-) |

agct : UTR agct : ORF A : Selected TSS

>upstream:1000, downstream:100, designated as 0:most upstream Oligo-capped cDNA

```
cctactcttgcccttttttttggccctccagtgttttgggtagttttgct
cccctacagccaaaggcaaacagataagttggaggctctggagtggctaca
taattttacacgactgcaattctctggctgcacttcacaaatgtatacaa
actaaatacaagtcctgtgtttttatcacaggaggctgatcaatataat
gaaattaaaagggggctgggtccatattgttctgtgtttttgtttgtttgt
ttctttttttgtttttgtggcctccttctctcaatttatgaagagaagc
agtaagatgttcctctcgggtcctctgagggacctggggagctcaggctg
ggaatctccaaggcagtaggtcgcctatcaaaaatcaaagtcaggtttg
tggggggaaaacaaaagcagcccattaccagaggactgtccgccttccc
ctcaccagcctaggcctttgaaaggaaacaaaagacaagacaaaatga
ttggcgtcctgagggagattcagcctagagctctctctcccccaatccct
ccctccggctgaggaaactaacaaggaaaaaaattgcggaagcagg
atntagaggaagcaaattccactgggtgcccttggctgccgggaacgtgga
ctagagagtctgcggcgcagccccgagcccagcgttcccgcgcgtctta
ggccggcggggcccggggcgggggaaggggacgcagaccgaggaccctaaga
cacctgtgtaccctccacccccacccccacccactcccccaactccct
agatgtgtcgtgggcggctgaacgtcgcccgtttaaggggcggggcccgg
ctccacgtgctttctgctgagtgaactacataaacagaggccggga
acggggcggggaggaggagagcacaggctttgaccgatagtaacctctg
cgctcgggtgcagccgaatctataaaaggaaactagtcggcaaaaacccc
Gtaattgcgagcgagagtgagtggggccgggaccgcagagccgagccga
cccttctctccgggctgcggcagggcagggcggggagctccgcgcacca
a
```

FASTA形式で目的の配列が表示される

SNPsとは

- DNA多型のなかの1つ
- 1塩基多型 (SNPs : **S**ingle **N**ucleotide **P**olymorphisms)
 - ゲノム内のある1塩基が個体によって異なり、ヒト集団内に1%以上で存在するもの
 - ヒトでは**1~2kpb**に**1個**の割合で存在
- なぜSNPsが注目を集める？
 - 遺伝マーカーとして有用 → 疾患関連遺伝子探索
 - ゲノム中に安定して存在
 - 検出が簡便であるので、高速に大量のSNPタイピングが可能

SNPsを調べる

NCBI
National Center for Biotechnology Information
National Library of Medicine National Institutes of Health

PubMed Entrez BLAST OMIM Books TaxBrowser Structure

Search **SNP** for **homo abca1** **Go**

SNPを選択 / **homo abca1と入力**

Goをクリック

resources

About NCBI
An introduction for researchers, educators and the public

GenBank
Sequence submission support and software

Literature databases
PubMed OMIM

Established in 1988 as a national resource for molecular biology information, NCBI creates public databases, conducts research in computational biology, develops software tools for analyzing genome data, and disseminates biomedical information - all for the better understanding of molecular processes affecting human health and disease. [More...](#)

HIV-1 Protein Interaction Database

HIV/AIDS researchers can now access a database of known interactions of HIV-1 proteins with human hosts. The database offers a comprehensive list of these interactions with links to sequence data, and genes. [Read more...](#)

Hot Spots

- ▶ Clusters of orthologous groups
- ▶ Coffee Break, Genes & Disease, NCBI Handbook
- ▶ Electronic PCR
- ▶ Entrez Home
- ▶ Entrez Tools
- ▶ Gene expression omnibus (GEO)
- ▶ Human genome resources

NCBIのトップページ

NCBIのdbSNPを使用



PubMed Nucleotide Protein Genome Structure Popset Taxonomy SNP

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Display Show: Sort Send to

Items 1-20 of 696 Page of 35 Next

1: [rs2853581](#) [*Homo sapiens*] Links

ACCAGAGGAGATGCTTTCTTAACA [A / G] AAATAGTATCTTATCAAACATCCA

Not on mRNA No 3D No OMIM

Gene Viewをクリック

2: [rs2853582](#) [*Homo sapiens*] Links

GCTGGTTCATTAGT

Not on mRNA No 3D No OMIM

RIKENSNPRC, YUSUKE

3: [rs2853578](#) [*Homo sapiens*] Links

GTCTGGGGGGGCTTCGCCTACTTGC [A / G] GGATGTGGTGGAGCAGGCAATCAT

Not on mRNA No 3D No OMIM

4: [rs2853577](#) [*Homo sapiens*] Links

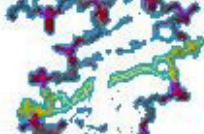
GGACTTGCATTTCTGGAAGTGTTTC [C / T] AAAAGAGAAACACCGGAACATGCT

Not on mRNA No 3D No OMIM

5: [rs2853574](#) [*Homo sapiens*] Links

TCGAGCTGACCCCTTTGAGGACATG [C / T] GGTACGTCTGGGGGGGCTTCGCCT

Not on mRNA No 3D No OMIM



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- Advanced
- Mouse Strains **NEW**

HAPLOTYPE

- Specifications
- Sample HapSet
- Sample Individual

SNP linked to Gene (geneID:19)

SNP are linked from gene [ABCA1](#) via the following methods:

- [Contig Annotation](#)
- [GenBank\(mrna\) Mapping](#)

all rs# to Batch Query all rs# to file.

Gene Model (mRNA alignment) information from genome sequence

| Total gene model (contig mRNA transcript): | | | | | 1 |
|--|---------------------------|---------------------------|------------------|--------------|-----------------|
| Contig | mrna | protein | mrna orientation | transcript | snp list |
| NT_008470 | NM_005502 | NP_005493 | reverse | minus strand | currently shown |

in gene region cSNP has frequency double hit haplotype tagged

| gene model | Contig | mrna | protein | mrna orientation | transcript | snp count |
|---------------------------|---------------------------|---------------------------|---------------------------|------------------|--------------|------------|
| (contig mRNA transcript): | NT_008470 | NM_005502 | NP_005493 | reverse | minus strand | 23, coding |



Snp In Gene Model Legend:

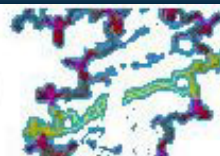
- █ - Region: exon
- ▬ - Region: intron
- | - snp: coding
- | - snp: synonymous
- | - snp: nonsynonymous
- | - snp: untranslated
- | - snp: intron
- | - snp: splice-site
- | - snp: coding: synonymy unknown

| Contig position | dbSNP rs# cluster id | Heterozygosity | Validation | 3D | OMIM | Function |
|-----------------|----------------------------|----------------|------------|-----|------|-----------------------------------|
| 9211750 | rs12346609 | N.D. | | Yes | | synonymous |
| 9219868 | rs13306077 | N.D. | | Yes | | contig reference C Gly [G] 3 2061 |
| 9222317 | rs13306075 | N.D. | | | | synonymous T Thr [T] 3 1792 |
| 9223623 | rs1883024 | N.D. | | | | contig reference C Thr [T] 3 1792 |
| 9225897 | rs1997618 | N.D. | | | | synonymous A Gln [Q] 3 1678 |
| 9230233 | rs2066716 | 0.231 | | H | | contig reference G Gln [Q] 3 1678 |
| | | 0.231 | | H | | nonsynonymous C Pro [P] 2 1648 |
| | | | | | | contig reference T Leu [L] 2 1648 |
| | | | | | | nonsynonymous G Arg [R] 2 1587 |
| | | | | | | contig reference A Lys [K] 2 1587 |
| | | | | | | nonsynonymous C Thr [T] 2 1555 |
| | | | | | | contig reference T Ile [I] 2 1555 |
| | | | | | | synonymous A Thr [T] 3 1427 |
| | | | | | | contig reference G Thr [T] 3 1427 |

クリック

遺伝子全体の概要が示される

SNPs個々の情報



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- Method
- Population
- Detail
- Class
- Publication
- Chromosome Report
- Locus Information
- STS Markers
- Free Form Search
- Simple
- Advanced
- Mouse Strains **NEW**

Reference SNP(refSNP) Cluster Report: rs1883024

| | |
|--|---|
| refSNP ID: rs1883024 | Allele |
| Organism: human (<i>Homo sapiens</i>) | Variation Class: SNP: single nucleotide polymorphism |
| Molecule Type: Genomic | Alleles: C/T |
| Created in build: 92 | Ancestral Allele: Not available |
| Last updated in build: 105 | |

SNP Details are categorized in the following sections:

[Submission](#) [Fasta](#) [Resource](#) [GeneView](#) [Map](#) [Variation](#) [Validation](#)

Submitter records for this RefSNP Cluster

The submission **ss2762065** has the longest flanking sequence of all cluster members and was used to instantiate sequence for **rs1883024** during BLAST analysis for the current build.

| NCBI Assay ID | Handle Submitter ID | Validation Status | Entry Date | Update Date | Build Added | Molecule Type | Sequence Orientation | Observed Alleles | Freq Warning | Ancestral Allele | Success Rate |
|---------------------------|--|-------------------|------------|-------------|-------------|---------------|----------------------|------------------|--------------|------------------|--------------|
| ss2762065 | TSC-CSHL TSC0897350 | | 01/02/01 | 10/10/03 | 92 | Genomic | forward | C/T | | | unknown |
| ss4040182 | SC_JCM AX060719.1_5265 | | 09/26/01 | 10/10/03 | 105 | Genomic | forward | T/C | | | unknown |

Fasta sequence (Legend)

>gn|dbSNP|rs1883024|allelePos=502|totalLen=589|taxid=9606|snpclass=1|alleles="C/T"|mol=Genomic|build=105

```

CAGgtaacct tgactaagtt acttaaccta tctaaacctc agtttctcca tctgtgaaat
ggagacagta atcatagcta tttccaaact gttgtgagaa tccaatgagt taaaggtata
agtcctcac cacagecct gccacatag tcagtgatca ctaTGTCTG AACACTGTAA
TTACTTCGCC ATATTCTCTG ATCATAGTGT TTTGCCTGG TATGTGACTA GAATTCTTT
CTGAGGTTTA TGGGCATGGT TGGTGGGTAT GCACCTGCCT GCAGGAGCCC GGTTTGGGGG
CATTACCTTG TACCTGGTAT GTTTTCTTTC AGGTGTGGTT CAATAACAAG GGCTGGCATG
CAATCAGCTC TTTCTGAAT GTCATCAACA ATGCCATTCT CCGGGCCAAC CTGCAAAAAGG
GAGAGAACCC TAGCCAT...
AGCAGCTCTC AGAGGTG...
GTAAGTGTGG CTGTGT...
GAGAAAAATG TGAATCTCAT TGTAGGG
    
```

この位置がC or T

Nucleic Acid Research Database Collection

Nucleic Acids Research

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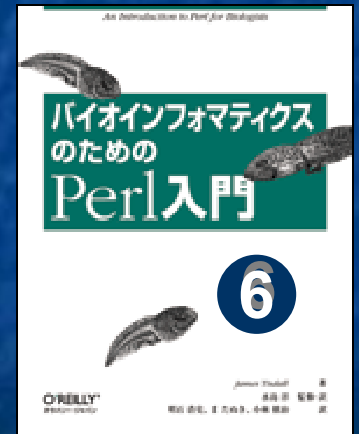
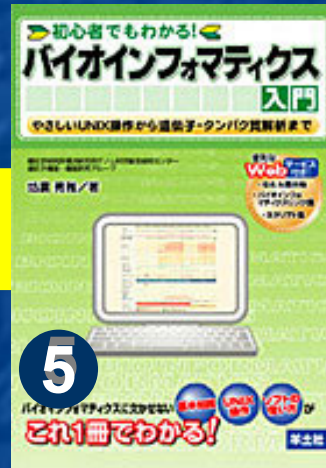
- 毎年始めに出版している
- 同じデータベースが毎年掲載されるわけではないので、過去数年分を参照する必要あり
- すべて無料でダウンロードできる
- http://nar.oupjournals.org/content/vol32/suppl_1/

参考文献



基本

応用



参考文献

- ① 「できるバイオインフォマティクス」
広川貴次、美宅成樹 著
申川書店
- ② 「今日からできるバイオデータベースとウェブツールの手とり足とり活用法」
中村保一、磯合敦、石川淳 編集
羊土社
- ③ 「東京大学バイオインフォマティクス集中講義」
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- ⑤ 「初心者でもわかる！バイオインフォマティクス入門 やさしいUNIX操作から遺伝子・タンパク質解析まで」
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- ⑦ 「バイオ研究が10倍はかどるMacOSX活用マニュアル セットアップから超簡単データ解析まで」
中村保一、磯合敦、荻島創一 著
羊土社

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ヒトゲノム情報にアクセスする代表的なブラウザの基本的な使い方をいくつか紹介した。本チュートリアルをきっかけとして、ヒトゲノム情報を最大限利用できるよう、各ブラウザの発展的な使用法を身につけてもらいたい