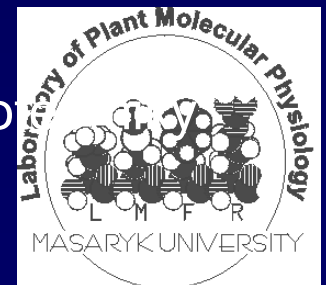


Základy genomiky



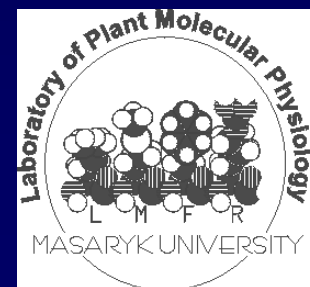
Jan Hejato

Masarykova univerzita, Oddělení funkční genomiky a proteomiky
Laboratoř molekulární fyziologie rostlin



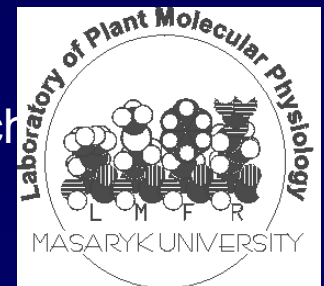
Základy genomiky I.

- Zdrojová literatura ke kapitole I:
 - Plant Functional Genomics, ed. Erich Grotewold, 2003, Humana Press, Totowa, New Jersey
 - Úvod do praktické bioinformatiky, Fatima Cvrčková, 2006, Academia, Praha



Základy genomiky I.

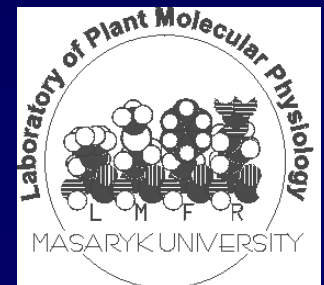
- Role BIOINFORMATIKY v současném pojetí FUNKČNÍ GENOMIKY
- Databáze
 - Spektrum „on-line“ zdrojů
 - PRIMÁRNÍ, SEKUNDÁRNÍ a STRUKTURÁLNÍ databáze
 - GENOMOVÉ zdroje
- Analytické nástroje
 - Vyhledávání homologií
 - Vyhledávání sekvenčních motivů, otevřených čtecích rámců, restrikčních míst....
 - Další www genomové nástroje



Základy genomiky I.

GENOMIKA-co to je?

- V širším pojetí-zkoumá **STRUKTURU** a **FUNKCI** genomů
 - Předpokladem je znalost genomu (sekvencí)-práce s databázemi
- V užším pojetí zkoumá **FUNKCI** jednotlivých genů - **FUNKČNÍ GENOMIKA**
 - používá zejména přístupy **REVERZNÍ GENETIKY**



GENOMIKA-co to je?

role BIOINFORMATIKY ve FUNKČNÍ GENOMICE

Přístupy „klasické“ genetiky



3

:

1

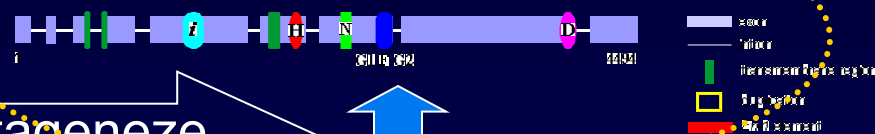


?

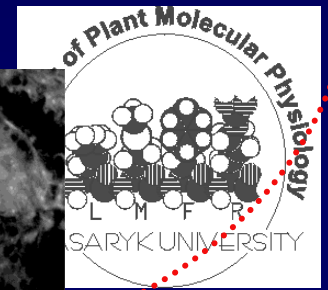
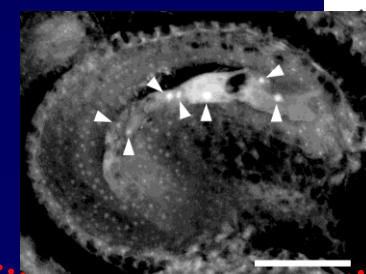
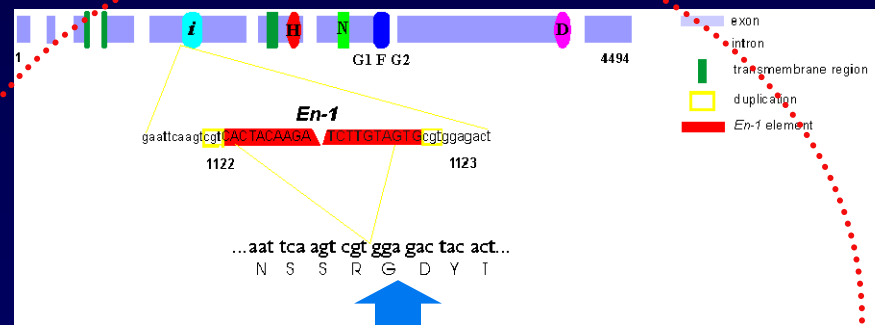
„Reverzně genetický“ přístup

5'TTATATATATATATATAAAAAATAAAATAAA
AGAACAAAAAGAAAATAAAATA...3'

BIOINFORMATIKA

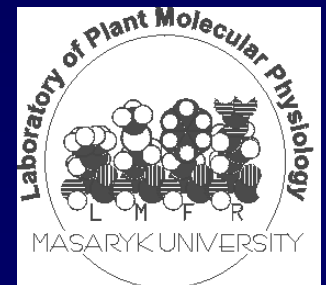


FUNKČNÍ GENOMIKA



Základy genomiky I.

- Role BIOINFORMATIKY v současném pojetí FUNKČNÍ GENOMIKY
- Databáze
 - Spektrum „on-line“ zdrojů



Databáze

Spektrum on-line zdrojů

EMBNet National Nodes

Vienna Biocenter	Austria	http://www.at.embnet.org/
BEN	Belgium	http://www.be.embnet.org/
BioBase	Denmark	http://biobase.dk/
CSC	Finland	http://www.fi.embnet.org/
INFOBIOGEN	France	http://www.infobiogen.fr/
GENIUSnet	Germany	http://genome.dkfz-heidelberg.de/biounit/
IMBB	Greece	http://www.imbb.forth.gr/
HEN	Hungary	http://www.hu.embnet.org/
INCB	Ireland	http://acer.gen.tcd.ie/
INN	Israel	http://dapsas.welzmann.ac.il/bcd/inn.html
IEN-ADR	Italy	http://bio-www.ba.cnr.it:8000/BioWWW/Bio-WWW.htm
CAOS/CAMM	Netherlands	http://www.caos.kun.nl/
Bio	Norway	http://www.no.embnet.org/
IBB	Poland	http://www.ibb.waw.pl/
IGC	Portugal	http://www.igc.gulbenkian.pt/
GeneBee	Russia	http://www.genebee.msu.su/
CNB-CSIC	Spain	http://www.es.embnet.org/
BMC	Sweden	http://www.embnet.se/
SIB	Switzerland	http://www.ch.embnet.org/
SEQNET	UK	http://www.seqnet.dl.ac.uk/

EMBNet Specialist Nodes

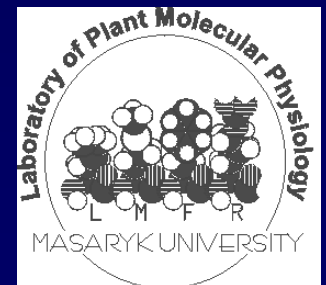
MIPS	Germany	http://www.mips.biochem.mpg.de/
ICGEB	Italy	http://www.icgeb.trieste.it/
Pharmacia Upjohn	Sweden	http://www.pnu.com/
F.Hoffmann-La Roche	Switzerland	http://www.roche.com/
EBI	UK	http://www.ebi.ac.uk/
HGMP-RC	UK	http://www.hgmp.mrc.ac.uk/
Sanger	UK	http://www.sanger.ac.uk/
UMBER	UK	http://www.bioinf.man.ac.uk/dbbrowser

EMBNet Associate Nodes

IBBM	Argentina	http://sol.biol.unlp.edu.ar/embnet
ANGES	Australia	http://www.angis.su.oz.au/
CBI	China	http://www.cbi.pku.edu.cn/
CIGB	Cuba	http://bio.cigb.edu.cu/
CDFD	India	http://salarjung.embnet.org.in/
SANBI	South Africa	http://www.sanbi.ac.za

USA Information Providers

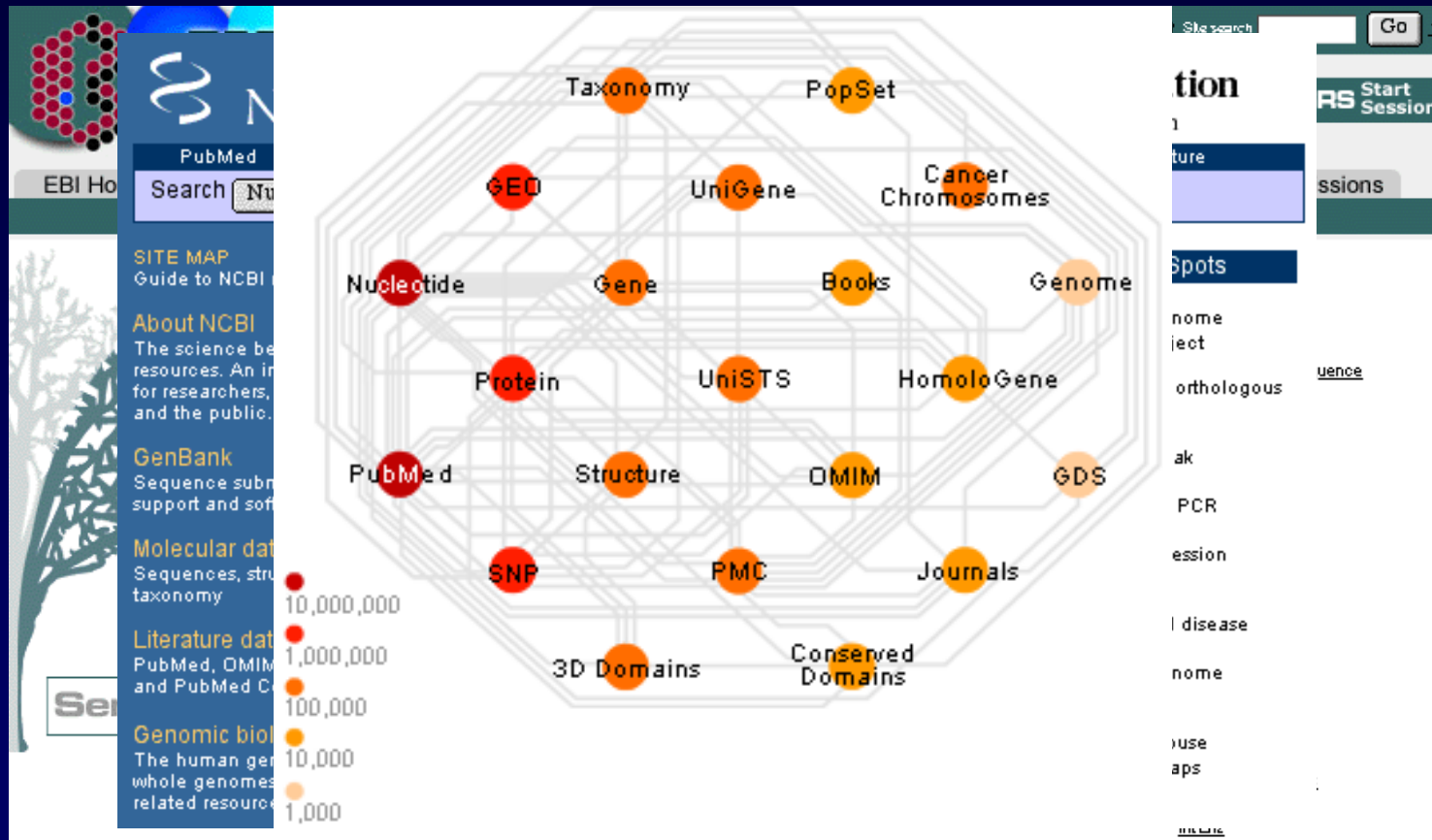
NCBI	USA	http://www.ncbi.nlm.nih.gov/
NLM	USA	http://www.nlm.nih.gov/
NIH	USA	http://www.nih.gov/



Databáze

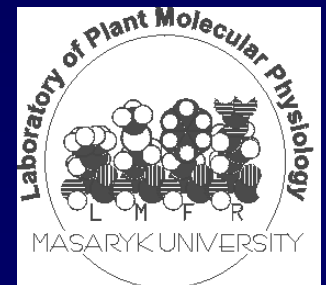
Spektrum on-line zdrojů

- EBI <http://www.ebi.ac.uk/services>
- NCBI <http://www.ncbi.nlm.nih.gov/>



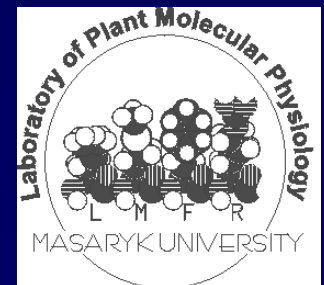
Základy genomiky I.

- Role BIOINFORMATIKY v současném pojetí FUNKČNÍ GENOMIKY
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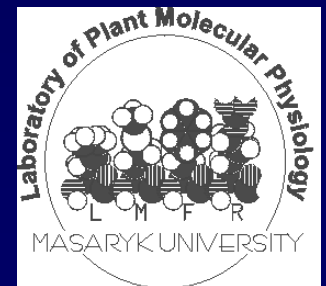
Primární databáze

- zahrnují soubory primárních dat – sekvencí DNA a proteinů
 - Sekvence v databázích tzv. „Velké trojky“:
 - EMBL, <http://www.ebi.ac.uk/embl/>
 - GenBank, <http://www.ncbi.nih.gov/Genbank/GenbankSearch.html>
 - DDBJ, <http://www.ddbj.nig.ac.jp>
 - denně vzájemná výměna a zálohování dat
 - velká datová náročnost (kapacita i software)
 - září 2003 27,2 x 10⁶ záznamů o zhruba 33 x 10⁹ bp



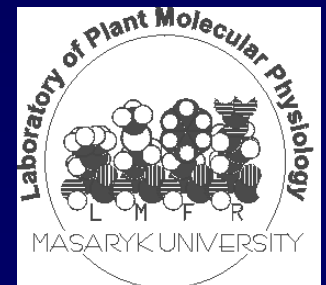
Primární databáze

- zahrnují soubory primárních dat – sekvencí DNA a proteinů
 - Proteinové sekvence:
 - PIR, <http://pir.georgetown.edu/>
 - MIPS, <http://www.mips.biochem.mpg.de>
 - SWISS-PROT, <http://www.expasy.org/sprot/>



Primární databáze

- Typy sekvencí v primárních databázích
 - standardní nukleotidové sekvence získané kvalitním sekvencováním
 - ESTs (Expressed Sequence Tags)
 - HGTS (High Throughput Genome Sequencing)
 - neanotované „surové“ výsledky sekvenačních projektů
 - referenční sekvence anotovaných genomů
 - TPAs (Third Party Annotation)
 - sekvence anotované jinými než původními autory



Primární databáze

- GenBank (NCBI) <http://www.ncbi.nlm.nih.gov/>

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

PubMed Entrez BLAST OMIM Books TaxBrowser Structure

Search for

SITE MAP
Guide to NCBI resources

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The science behind our resources. An introduction for researchers, educators and the public.

GenBank
Sequence submission support and software

Molecular databases
Sequences, structures and taxonomy

Literature databases
PubMed, OMIM, Books and PubMed Central

Genomic biology
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...](#)

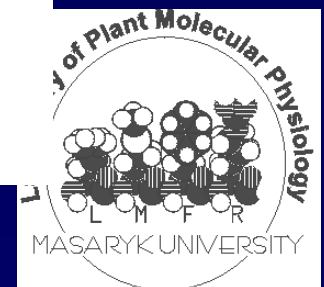
Hot Spots

- ▶ Cancer genome anatomy project
- ▶ Clusters of orthologous groups
- ▶ Coffee Break
- ▶ Electronic PCR
- ▶ Gene expression omnibus
- ▶ Genes and disease
- ▶ Human genome resources
- ▶ Human/mouse homology maps
- ▶ LocusLink

Mouse Genome
Resources: explore tools for manipulating the mouse genome.

Try these: Map Viewer Sequencing Progress Human-Mouse Homology

BLink and get results fast!



Proteinové sekundární databáze

- databáze funkčních nebo strukturálních *motivů* získaných srovnáváním primárních dat (sekvencí)

□ PROSITE, <http://www.expasy.org/prosite/> browser/PRINTS/

The screenshot displays the PROSITE database interface. At the top, there is a navigation bar with links: [ExPASy Home page](#), [Site Map](#), [Search ExPASy](#), [Contact us](#), [Swiss-Prot](#), [PROSITE](#), and [Proteomics tools](#). Below this, there are regional mirrors: [Hosted by: SIR](#), [Switzerland](#), [Mirror sites](#), [Australia](#), [Belgium](#), [Canada](#), [China](#), [Korea](#), [Taiwan](#), [USA](#).

The main content area shows two search results:

- >[PDOC50109](#) [PS50109](#) **HIS_KIN** Histidine kinase domain [profile].
402 - 671 NASHDIRGALAGMKGLIDICRDGVKPGSDVDTTLNQVNVCAKDLVALLNSVLDMSKIRSG
KMQLVEEDFNLEKLLLEDVIDFYHPVAMKKGVDVVLDPHDgavFKPSNVRGDSGRLEKILN
NLVSNVAKFTVD--GHIAVRAWAQrggnsavvlasykpvakfvksmEcknkeesstye
teissirnnanTMEFVPEVDDTGKGIEMEMRKSVPENYVQVREtAQGHQGTGLGLGLVQ
SLVRLMGGHIRITDKAMGeKGTCPQFNVLTT
- >[PDOC50110](#) [PS50110](#) **RESPONSE_REGULATORY** Response regulatory domain [profile].
987 - 1085 RVLVDDNPISRKRVATGKLEKMGVSeVEQCDSGKRALRLVTEGLtgreeggsvdklpFDY
IPMDCQMPMDGYRATRELRkvekSYGVRTPIIAVSGHD-----

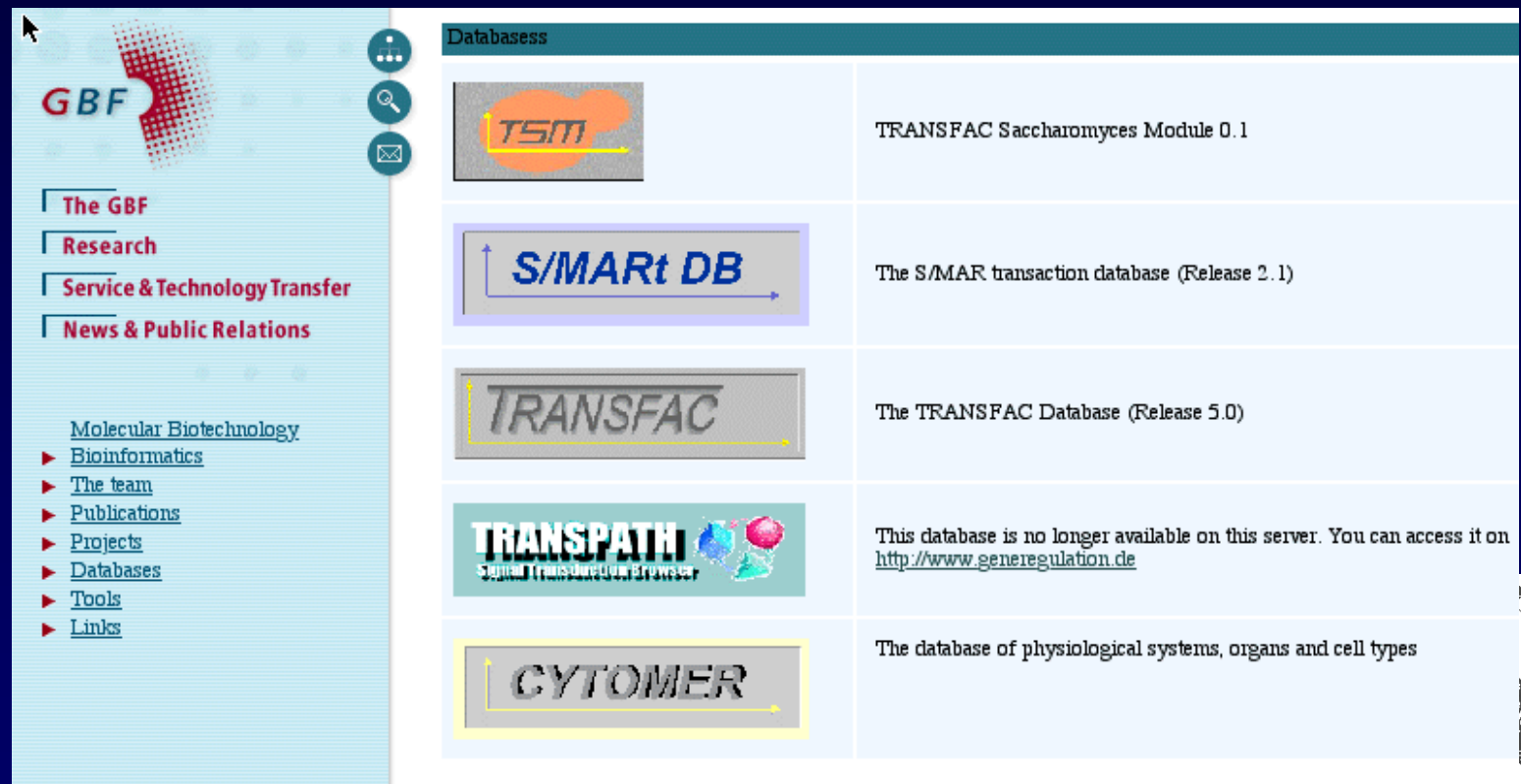
Below the results is a section titled "Graphical summary of hits (java applet)". It contains a graphical representation of a protein sequence with two red vertical cursors indicating a zoom region. The zoomed-in region shows a yellow bar labeled "HIS_KIN" and another yellow bar labeled "RESPONSE_REGULATORY". Below the graphical summary, it states "98 hits with 12 PROSITE entries".

At the bottom of the interface, there is another navigation bar with the same links as the top. Below this, there are buttons for "START THE SCAN" and "RESET". At the very bottom, there is a search input field with a "GO" button and a link: [MULTIPLE AMBIGUOUS \(\)](#) (to test a pattern, see [help](#)).


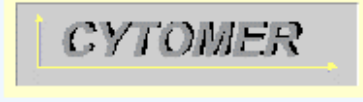


Sekundární databáze DNA

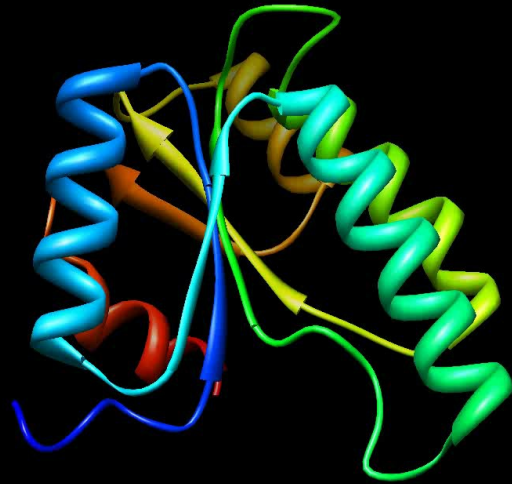
- TRANSFAC <http://www.gene-regulation.com/>



The screenshot shows the GBF website interface. On the left is a navigation menu with the GBF logo and links for 'The GBF', 'Research', 'Service & Technology Transfer', and 'News & Public Relations'. Below these are links for 'Molecular Biotechnology', 'Bioinformatics', 'The team', 'Publications', 'Projects', 'Databases', 'Tools', and 'Links'. The main content area is titled 'Databases' and contains a table with the following entries:

Database Logo	Description
	TRANSFAC Saccharomyces Module 0.1
	The S/MAR transaction database (Release 2.1)
	The TRANSFAC Database (Release 5.0)
	This database is no longer available on this server. You can access it on http://www.generegulation.de
	The database of physiological systems, organs and cell types

Strukturální databáze





Seite 1 von 2

1P5Y

and Mutants

Engineered: Yes; Mutation: Yes



[Download/Display File](#)

[Structural Neighbors](#)

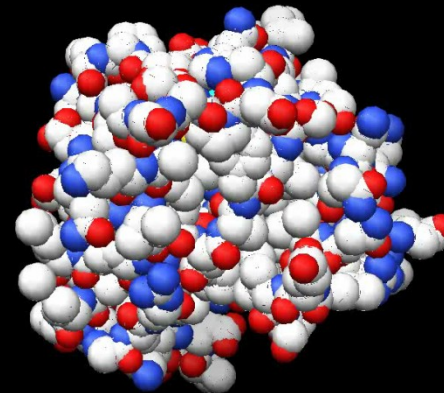
[Geometry](#)

[Other Sources](#)

[Sequence Details](#)

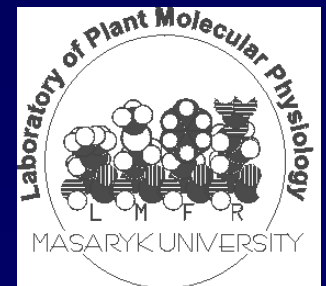
Explore

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



Základy genomiky I.

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 - GENOMOVÉ zdroje



Primární data-genomové zdroje

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

PubMed Entrez BLAST OMIM Books TaxBrowser Structure

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Hot Spots

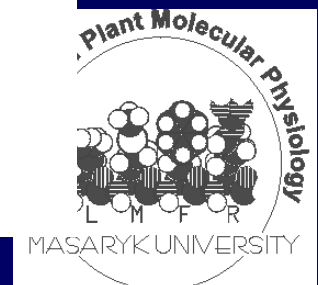
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- ▶ Clusters of orthologous groups
- ▶ Coffee Break
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- ▶ Human genome resources
- ▶ Human/mouse homology maps
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Mouse Genome

Resources: explore tools for manipulating the mouse genome.

Try these: Map Viewer Sequencing Progress Human-Mouse Homology

BLink and get results fast!



Primární data-genomové zdroje

NCBI Entrez Genome

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Help

Search for on chromosome(s) Find

Show linked entries [Help](#) [FTP](#)

Entrez Genomes
MapViewer Home
Prominent organisms
FTP SITE
Related Databases:
TAIR
TIGR
MIPS
KAOS
Sequencing Projects:
SPP Consortium
CSH / WashU
TIGR
Kazusa
ESSA
Genoscope

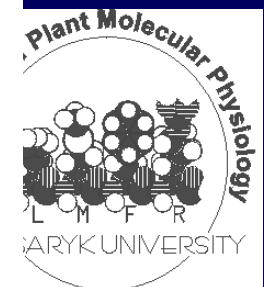
Arabidopsis thaliana genome view [BLAST search Arabidopsis genome](#)

I II III IV V VI VII

Lineage: [Eukaryota](#); [Viridiplantae](#); [Streptophyta](#); [Embryophyta](#); [Tracheophyta](#); [Spermatophyta](#); [Magnoliophyta](#); [eudicotyledons](#); [core eudicots](#); [Rosidae](#); [eurosids II](#); [Brassicales](#); [Brassicaceae](#); [Arabidopsis](#)

Arabidopsis thaliana is a small flowering plant that is widely used as a model organism in plant biology. Arabidopsis is a member of the mustard (Brassicaceae) family, which includes cultivated species such as cabbage and radish. Arabidopsis is not of major agronomic significance, but it offers important advantages for basic research in genetics and molecular biology. Its genome has been sequenced by an international collaboration collectively termed the [Arabidopsis Genome Initiative \(AGI\)](#) ([The Arabidopsis Genome Initiative, 2000, Nature, 408:796-815](#)).

This sequence, map, and annotations are the result of a collaboration between [TIGR](#), [MIPS](#), and [TAIR](#). The non-redundant sequence of the chromosomes (pseudomolecules) and their annotations were provided to NCBI by TIGR on behalf of the collaborators.



Primární data-genomové zdroje

NCBI

PubMed Entrez BLAST OMIM Taxonomy Structure

Search Find in This View

MapViewer Home

Map Viewer Help
Arabidopsis Maps Help
FTP
Data As Table View
Maps & Options
Compress Map
Region Show:
out
200n
in
Clone

ideogram
 master

Arabidopsis thaliana Map View

Chromosome: [I](#) | [II](#) | [III](#) | [IV](#) | [V](#)

Master Map: Gene

Total Genes On Chromosome: 7173
Region Displayed: 0-30M bp [Download/View Sequence/Evidence](#)
Genes Labeled: 20 Total Genes in Region: 7173

Clone	Gene	Accession	Description
At1g05180	YUP8H12.21	TIGR MIPS TAIR NM_100396 NP_172010	auxin-resistance protein AXR1
At1g08210	T23G18.7	TIGR MIPS TAIR NM_100695 NP_563808	expressed protein
At1g11250	T28P6.10	TIGR MIPS TAIR NM_100997 NP_172591	syntaxin-related protein At-SYR1, putative
At1g14670	T5E21.15	TIGR MIPS TAIR NM_101334 NP_172919	endomembrane protein, putative
At1g17790	F2H15.2	TIGR MIPS TAIR NM_101642 NP_564037	expressed protein
At1g21050	T22I11.13	TIGR MIPS TAIR NM_101958 NP_564130	expressed protein
At1g24210	F3I6.14	TIGR MIPS TAIR NM_102267 NP_564212	expressed protein
At1g28370	F3M18.20	TIGR MIPS TAIR NM_102603 NP_174159	ethylene-responsive element binding factor, putative
At1g31885	F5M6.28	TIGR MIPS TAIR NM_102926 NP_174472	major intrinsic protein, putative
At1g35670	F15O4.8	TIGR MIPS TAIR NM_103271 NP_174807	calcium-dependent protein kinase
At1g48160	F21D18.11	TIGR MIPS TAIR NM_103712 NP_175250	signal recognition particle 19 kDa protein subunit, putative
At1g51980	F5F19.4	TIGR MIPS TAIR NM_104079 NP_175610	mitochondrial processing peptidase alpha subunit, putative
At1g55150	T7N22.9	TIGR MIPS TAIR NM_104388 NP_175911	ethylene-responsive RNA helicase, putative
At1g60140	T13D8.4	TIGR MIPS TAIR NM_104705 NP_176221	trehalose-6-phosphate synthase, putative
At1g63750	F24D7.6	TIGR MIPS TAIR NM_105052 NP_176562	putative disease resistance protein
At1g67090	F5A8.1	TIGR MIPS TAIR NM_105379 NP_176880	ribulose-bisphosphate carboxylase small unit, putative
At1g69870	T17F3.10	TIGR MIPS TAIR NM_105655 NP_564979	putative peptide transporter
At1g72970	F3N23.17	TIGR MIPS TAIR NM_105955 NP_565050	expressed protein
At1g75780	T4O12.1	TIGR MIPS TAIR NM_106228 NP_177706	tubulin beta-1 chain
At1g78700	F9K20.26	TIGR MIPS TAIR NM_106517 NP_565187	expressed protein

[Disclaimer](#) | [Write to the Help Desk](#)
[NCBI](#) | [NLM](#) | [NIH](#)

Molecular Physiology
UNIVERSITY

Primární data-genomové zdroje

NCBI Nucleotide

Search for

History has expired due to inactivity.

Show:

I: NC_003070. Arabidopsis thali...[gi:22330780] [Links](#)

Reverse Complement Strand
[View on plus strand](#) [Protein coding genes](#) [Hide Toolbar](#)

Search for gene CDS with gene and mRNA
 Hide sequence

30020639M 20M 10M 1 5'

At1g05200 At1g05190 At1g05180 At1g05170

Legend:
- CDS (pink line)
- RNA (blue line)
- gene (grey line)
- sequence fragment shown (red line)

Sequence:

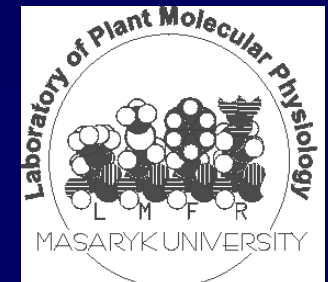
```
1501830 CTTTTTGGT ATCAGTTCAC CGGAGGCGAAA AATCGTCTCT CGCTTGACCT GCGAAGATCC At1g05180  
mRNA-protein id:  
CDS  
M
```

```
1501770 AAGCAGTAAA AAGATCCAGG AGCATGTGG AAGAGAGGCC AACAAATGGA GAACCTAATA At1g05180  
mRNA-protein id:  
CDS  
Q A V K R S R R H V E E E P T M V E P K
```

```
1501710 CCAAGTACGA TCGTCAGCTC AGSTATAACAT ACTCTTTTTT CTAAACTCT ACTTCGACT At1g05180  
mRNA-protein id:  
CDS  
T K Y D R Q L R
```

```
1501650 ACTCTTGTCG GAGGAGAAAT TGATGAGCTA GAATTCGTTG GGTGACTGTA TTAGGATTTG At1g05180  
mRNA-protein id:  
CDS  
I H
```

```
1501590 GGGGAGGTA GGTCAAGCGG CCTTGGARGA AGCGAGTATC TGTTACTCA ATTGTGGCCC At1g05180  
mRNA-protein id:  
CDS  
G E V G Q A A L E E A S I C L L N C G P
```



Primární data-genomové zdroje

- Human Genome Browser <http://genome.ucsc.edu/>

UCSC Genome Bioinformatics

Genome Browser - Family Browser - Blat - Table Browser - FAQ - Help

Genome
Browser

About the UCSC Genome Bioinformatics Site

This site contains the reference sequence for the human and *C. elegans* genomes and working drafts for the mouse, rat, Fugu, *Drosophila*, *C. briggsae*, and SARS genomes. It also contains the CFTR (cystic fibrosis) region in 13 species.

Family
Browser

We encourage you to explore these sequences with our tools. The Genome Browser zooms and scrolls over chromosomes, showing the work of annotators worldwide. The Family Browser shows expression, homology and other information on groups of genes that can be related in many ways. The Table Browser provides convenient access to the underlying database. Blat quickly maps your sequence to the genome.

Blat

Tables

Downloads

News

[News Archives](#) ▶

Release Log

22 Dec. 2003 - Advance Warning of Browser Outage 30 Dec. 2003

Custom
Tracks

Electrical power to the UCSC School of Engineering building will be shut down on 30 December from 7 a.m. to 3 p.m. PST. The Genome Browser and Blat servers will not be available during this time period.

Mirrors

Archives

10 Dec. 2003 - Human/Chimp Alignment Tracks Released

Credits

UCSC has released alignments of the Nov. 2003 chimpanzee draft assembly to the July 2003 human assembly in the Genome Browser. These alignments may be viewed on the [Human July 2003](#) assembly. This release coincides with today's [announcement](#) by the National Human Genome Research Institute (NHGRI) of the first draft assembly of the chimpanzee genome.

Pubs

Cite Us

The set of human/chimpanzee alignments consists of a reciprocal best-in-genome net track and a chimp chain track. These alignments were generated using the blastz program developed at Pennsylvania State University and the programs blat, actChain, chainNet, and netSyntenic developed at UCSC by Jim Kent. Research scientists should find these tracks useful for locating orthologous regions and studying genome rearrangement in the two species.

Licenses

Jobs

For more information about the alignment tracks, refer to the track description pages. The tables may be downloaded from the Genome Browser FTP server's [hg16 database directory](#). The chimp sequence and alignment data are downloadable from the [hg16 human/chimp alignments directory](#).

Contact Us

The chimp sequence used in these alignments was obtained from the 13 Nov. 2003 Arachne assembly. We'd like to thank NHGRI, the Eli & Edythe L. Broad Institute at MIT/Harvard, and Washington University School of Medicine for providing this sequence, and LaDeana Hillier, Washington University School of Medicine, and the Whitehead Institute for their work on the alignments. We'd also like to acknowledge the members of the UCSC team who contributed to the release of these alignments in the Genome Browser: Jim Kent, Kate Rosenbloom, Heather Trumbower, and Donna Karolchik.

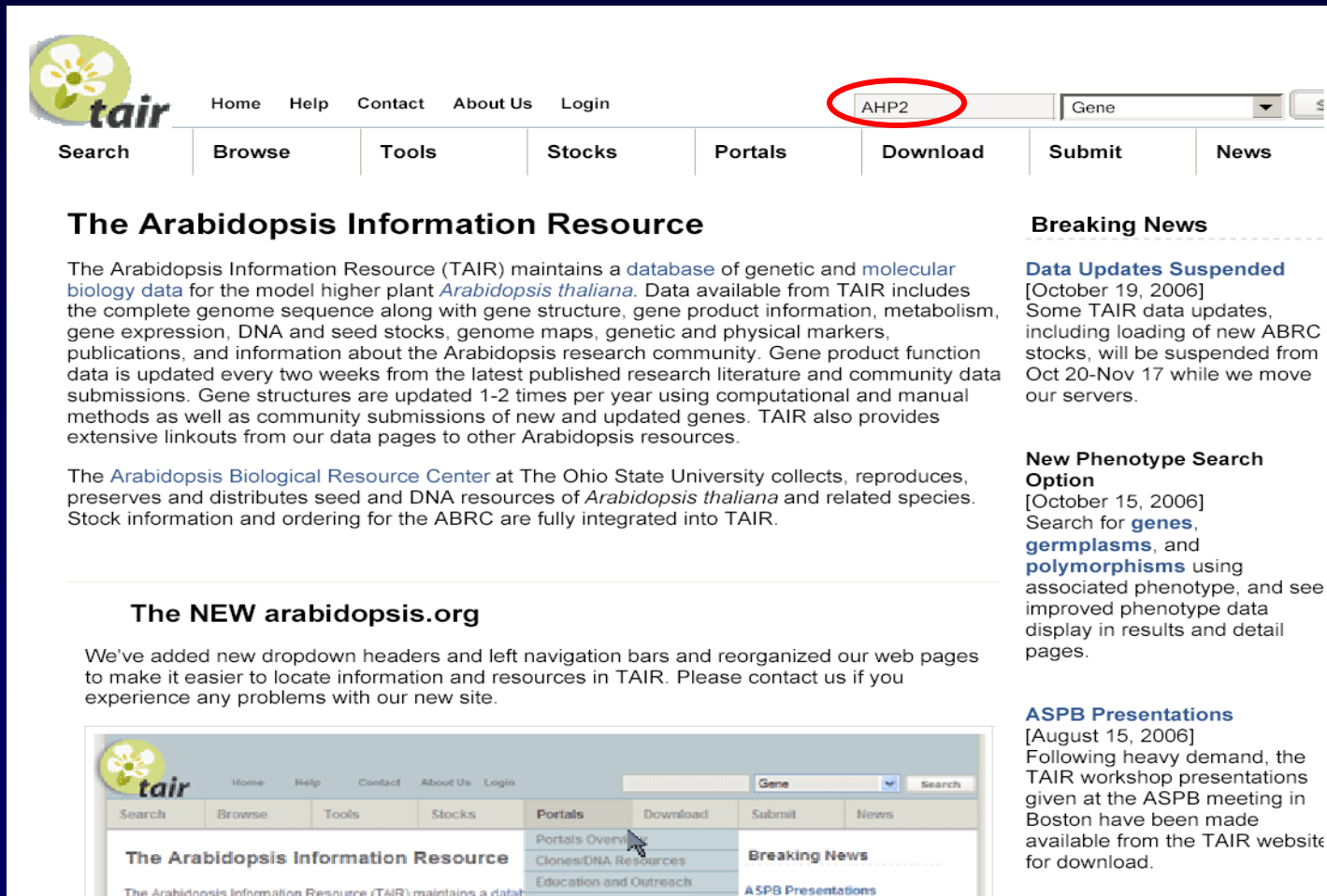
24 Nov. 2003 - Build 32 Mouse Assembly (Oct. 2003) Released: We have released a Genome Browser and Blat server for the latest mouse genome assembly, NCBI Build 32 (UCSC v. mm4). [Read more.](#)

31 Oct. 2003 - V. 3.1 *D. melanogaster* Assembly (Jan. 2003) Released: We have added the *Drosophila melanogaster* (fruitfly) assembly to



Primární data-genomové zdroje

- TAIR, The Arabidopsis Information Resource, <http://www.arabidopsis.org>



The screenshot displays the TAIR website interface. At the top left is the TAIR logo, a green flower icon. To its right are navigation links: Home, Help, Contact, About Us, and Login. A search bar contains the text 'AHP2' and a dropdown menu is set to 'Gene'. Below the search bar is a horizontal navigation menu with buttons for Search, Browse, Tools, Stocks, Portals, Download, Submit, and News. The main content area is divided into two columns. The left column features the heading 'The Arabidopsis Information Resource' followed by a paragraph describing the database and its contents. Below this is another paragraph about the Arabidopsis Biological Resource Center. The right column has a 'Breaking News' section with two news items: 'Data Updates Suspended' and 'New Phenotype Search Option'. At the bottom of the screenshot, there is a section titled 'The NEW arabidopsis.org' with a paragraph about website updates. A smaller version of the website interface is shown at the bottom left, with a mouse cursor pointing to the 'Portals' button. A play button icon is visible on the far left of the slide.

The Arabidopsis Information Resource

The Arabidopsis Information Resource (TAIR) maintains a [database](#) of genetic and [molecular biology data](#) for the model higher plant *Arabidopsis thaliana*. Data available from TAIR includes the complete genome sequence along with gene structure, gene product information, metabolism, gene expression, DNA and seed stocks, genome maps, genetic and physical markers, publications, and information about the Arabidopsis research community. Gene product function data is updated every two weeks from the latest published research literature and community data submissions. Gene structures are updated 1-2 times per year using computational and manual methods as well as community submissions of new and updated genes. TAIR also provides extensive linkouts from our data pages to other Arabidopsis resources.

The [Arabidopsis Biological Resource Center](#) at The Ohio State University collects, reproduces, preserves and distributes seed and DNA resources of *Arabidopsis thaliana* and related species. Stock information and ordering for the ABRC are fully integrated into TAIR.

The NEW arabidopsis.org

We've added new dropdown headers and left navigation bars and reorganized our web pages to make it easier to locate information and resources in TAIR. Please contact us if you experience any problems with our new site.

Breaking News

Data Updates Suspended
[October 19, 2006]
Some TAIR data updates, including loading of new ABRC stocks, will be suspended from Oct 20-Nov 17 while we move our servers.

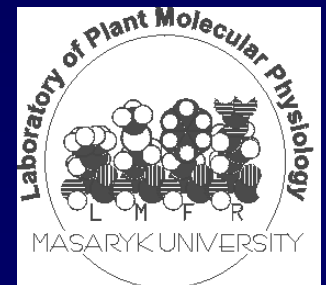
New Phenotype Search Option
[October 15, 2006]
Search for **genes**, **germplasms**, and **polymorphisms** using associated phenotype, and see improved phenotype data display in results and detail pages.

ASPB Presentations
[August 15, 2006]
Following heavy demand, the TAIR workshop presentations given at the ASPB meeting in Boston have been made available from the TAIR website for download.



Základy genomiky I.

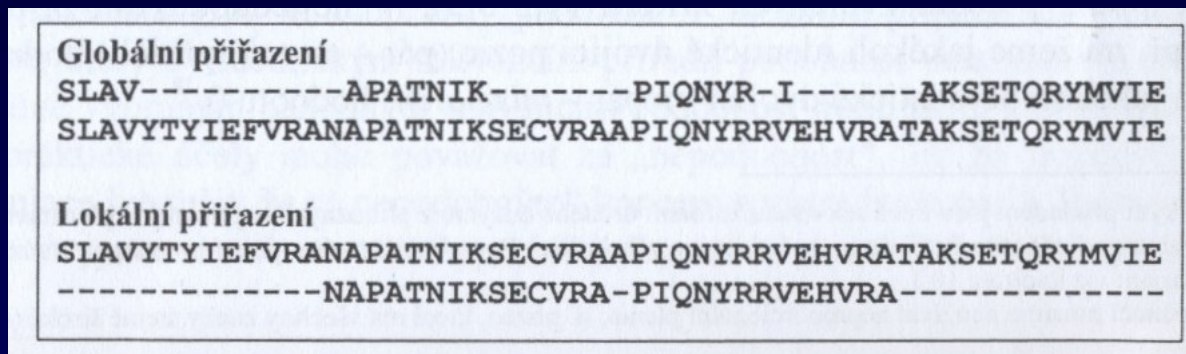
- Role BIOINFORMATIKY v současném pojetí FUNKČNÍ GENOMIKY
- Databáze
 - Spektrum „on-line“ zdrojů
 - PRIMÁRNÍ, SEKUNDÁRNÍ a STRUKTURÁLNÍ databáze
 - GENOMOVÉ zdroje
- Analytické nástroje
 - Vyhledávání homologií



Analytické nástroje

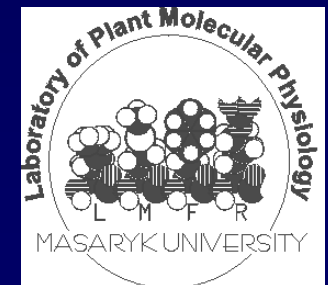
porovnávání dvou sekvencí

□ Globální vs. lokální přiřazení



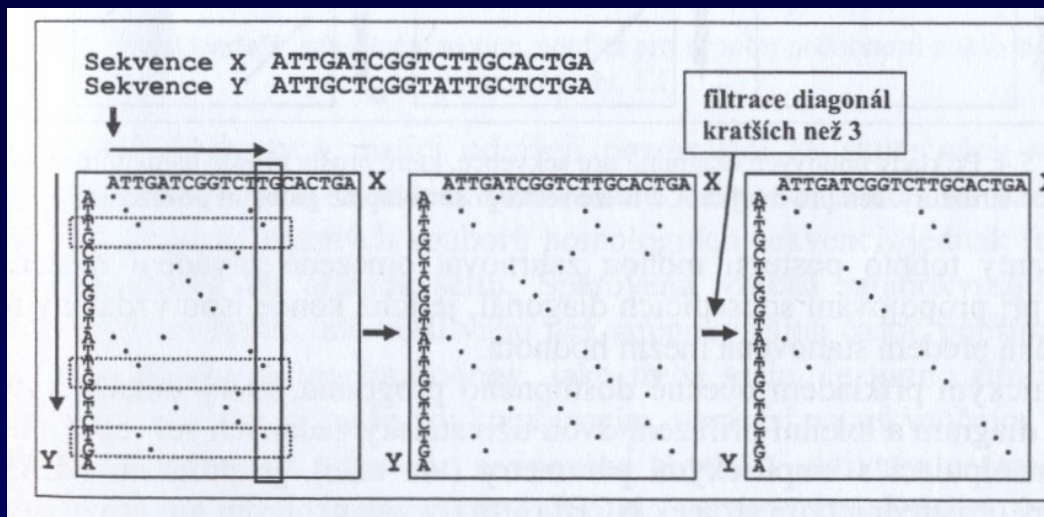
Cvrčková, Úvod do praktické bioinformatiky

- globální přiřazení pouze u sekvencí, které jsou si podobné po celé délce srovnávané sekvence (za cenu vnášení mezer do jedné nebo obou sekvencí)
- globální přiřazení se používá především v případě mnohačetného přiřazování (CLUSTALW, viz dále)
- lokální přiřazení umožní identifikaci a srovnání i v případě porovnávání pouze úseků sekvencí s významnou mírou podobnosti, např. i při záměně pořadí proteinových domén během evoluce



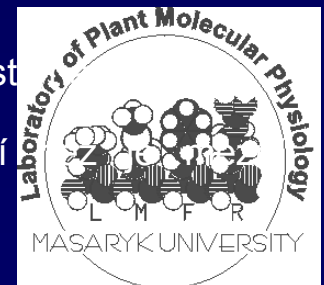
Analytické nástroje porovnávání dvou sekvencí

- Volba správného typu přiřazení pomocí bodového diagramu (dotplot)



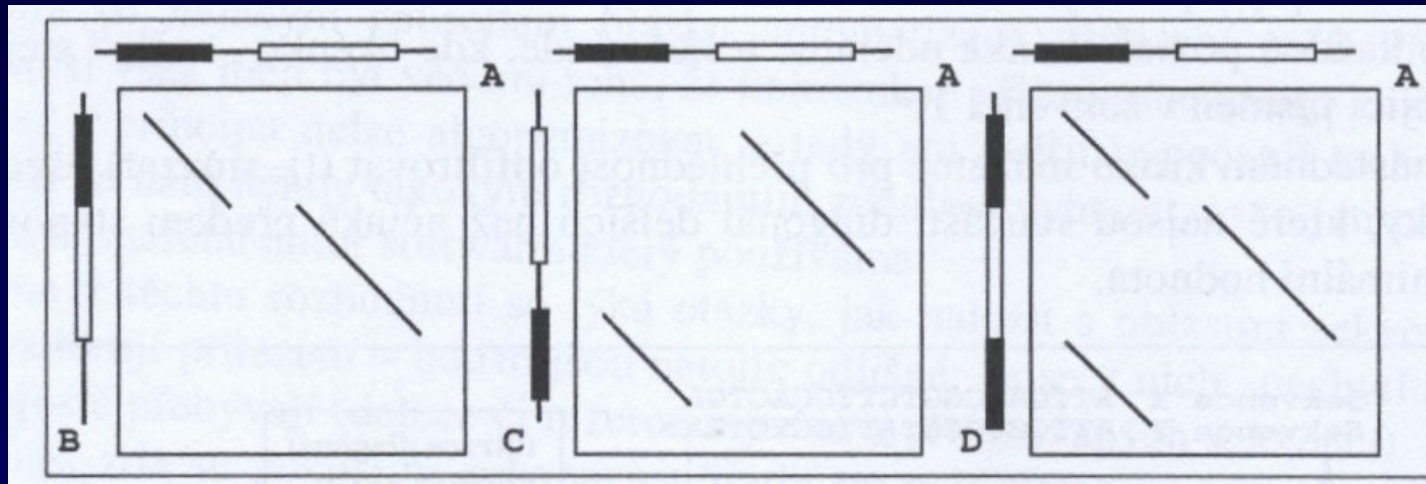
Cvrčková, Úvod do praktické bioinformatiky

- vynesení sekvencí proti sobě
- identifikace shody v okně o dané velikosti
- „odfiltrování“ diagonál o délce menší než daná hodnota (threshold)



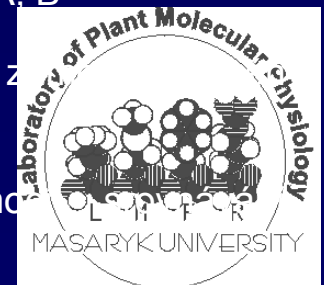
Analytické nástroje porovnávání dvou sekvencí

- příklady srovnání sekvencí pomocí bodového diagramu



Cvrčková, Úvod do praktické bioinformatiky

- globálně lze srovnávat pouze sekvence A, B
- ostatní sekvence prošly během evoluce ztrátou částí a je nutné je porovnávat lokálně
- bodový diagram lze získat pomocí programem BLAST2 (viz dále)



Analytické nástroje

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

NCBI *nucleotide-nucleotide* **BLAST**

Nucleotide Protein Translations Retrieve results for an RID

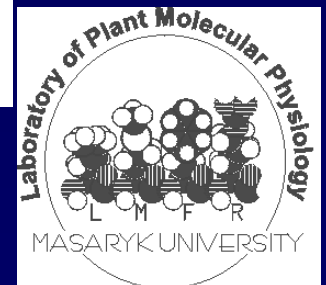
[Search](#)

```
aacaccctgc
acaccatcat cattatcacc atcgttttgg ggcgatggtt tgtggttcca
gcgatattaat
ataattaatt tattccacat gagatatgat atgatatact atgtattttt
tgtttttttt
ttattttgtaa acctttaata taacaagaac tacaaaaaat gaaaa
```

[Set subsequence](#) From: To:

[Choose database](#)

Now: **BLAST!** or



Analytické nástroje

```

 >gi|5016088|ref|NM_001101.2| actin, beta (ACTB), mRNA
      Length = 1793
      Score = 1110 bits (560), Expect = 0.0
      Identities = 965/1100 (87%)
      Strand = Plus / Plus

Query: 156 gtcgacaacggctctggcatgtgcaaggccggatttgcggagacgatgctccccggccc 215
      |||
Sbjct: 101 gtcgacaacggctccggcatgtgcaaggccggcttcgcgggcgacgatgccccggggcc 160

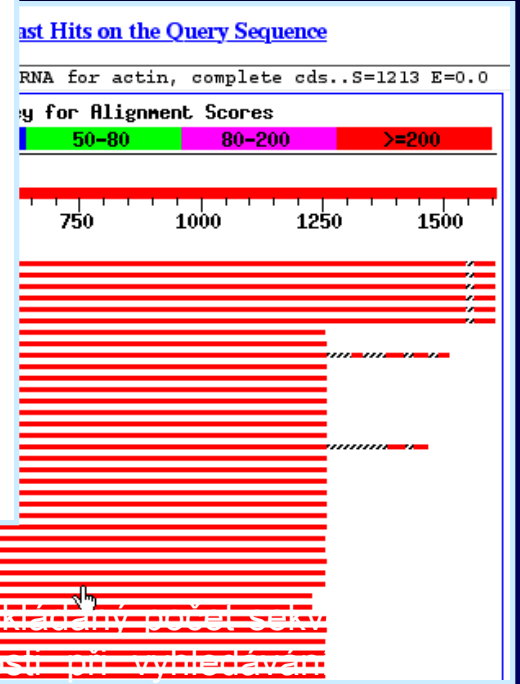
Query: 216 gtcttcccatcgattgtgggacgtcccgcaccagggtgtgatggtcggcatgggcag 275
      |||
Sbjct: 161 gtcttcccctccatcggtggggcgcccaggcaccagggtgtgatggtcggcatgggtcag 220

Query: 276 aaggactcgtacgtgggtgatgaggcgcagagcaagcgtggtatcctcaccctgaagtac 335
      |||
Sbjct: 221 aaggattcctatgtgggcgacgaggccagagcaagagaggcatcctcaccctgaagtac 280

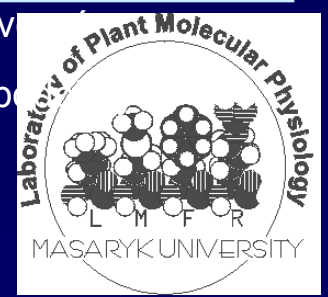
Query: 336 cccattgagcaccggtatcgtgaccaactgggacgatatggagaagatctggcaccacacc 395
      |||
Sbjct: 281 cccatcgagcaccggcatcgtcaccaactgggacgacatggagaaaatctggcaccacacc 340
  
```

E= expectancy value

Expect = 0.0



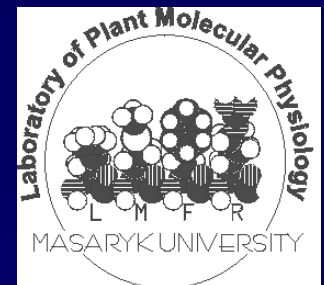
- „expectancy value“ udává předpokládaný počet sekvencí podobnosti při vyhledávání se stejnou délkou v stejné velké databázi složené z náhodných sekvencí
- výsledek udává frakci totožných a u proteinů i pozic, příp. počet vložených mezer



Analytické nástroje

specializované typy BLASTu

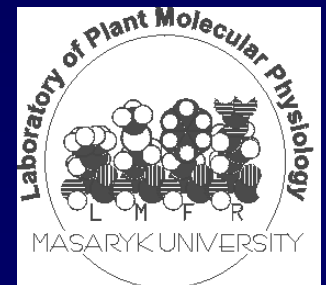
- V současnosti existuje celá řada specializovaných verzí programu BLAST
 - vyhledávání podle zdroje (organismu) sekvencí, např. známých genomů mikroorganismů
 - BLASTP
 - vyhledávání podobnosti k proteinu v databázi proteinových sekvencí
 - BLASTN
 - vyhledávání podobnosti k nukleotidové sekvenci v databázi nukleotidových sekvencí
 - další varianty jako např. MEGABLAST pro identifikaci totožných nebo velice podobných sekvencí (vyhledává dlouhé podobné úseky nukl. sekvencí)
 - BLASTX
 - vyhledávání podobnosti k proteinu v databázi nukleotidových sekvencí přeložených do sekvence aa



Analytické nástroje

specializované typy BLASTu

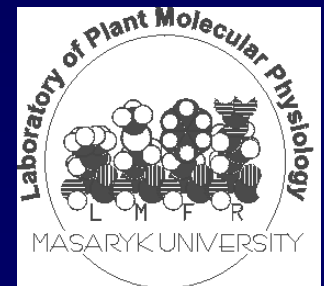
- V současnosti existuje celá řada specializovaných verzí programu BLAST
 - TBLASTN
 - vyhledávání k sekvenci nukleotidů přeložené do sekvence aa v databázi proteinů
 - TBLASTX
 - vyhledávání k sekvenci nukleotidů přeložené do sekvence aa v databázi nukleotidových sekvencí přeložených do sekvence aa



Analytické nástroje

specializované typy BLASTu

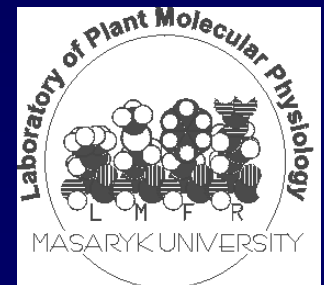
- V současnosti existuje celá řada specializovaných verzí programu BLAST
 - PSI-BLAST (Position-Specific Iterated Blast)
 - Prvním krokem je standardní BLAST, při kterém PSI-BLAST identifikuje skupinu podobných sekvencí s E hodnotou lepší než minimální hodnota (standardně 0,005)
 - PSI-BLAST vytváří pro každé přiřazení tzv. PSSM (position specific substitution matrix)
 - PSSM matice zohledňuje výskyt jedné aminokyseliny ve stejné pozici se zvýšenou frekvencí u sekvencí identifikovaných jako podobné v prvním kole pomocí BLAST, což může znamenat funkční konzervovanost



Analytické nástroje

specializované typy BLASTu

- V současnosti existuje celá řada specializovaných verzí programu BLAST
 - PHI-BLAST (Pattern-Hit Initiated Blast)
 - Určen k identifikaci specifické sekvence, např. motivu (pattern) v sekvenci podobných proteinových sekvencí
 - Sekvenci motivu je třeba vložit pomocí speciálního syntaxu
 - [LVIMF] znamená buď Leu, Val, Ile, Met nebo Phe
 - - je oddělovník (neznačená nic)
 - x(5) znamená 5 jakýchkoliv aminokyselin
 - x(3, 5) znamená 3 až 5 jakýchkoliv aminokyselin

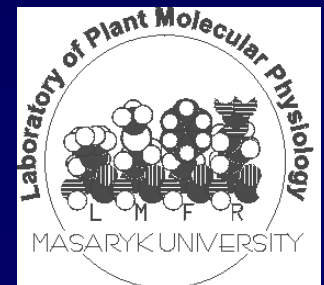


Analytické nástroje

specializované typy BLASTu

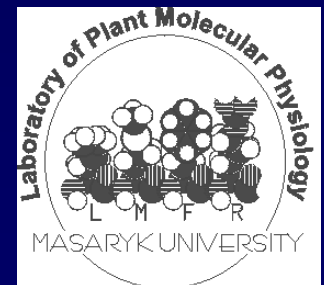
□ Příklad vyhledávání pomocí PHI-BLAST

```
>gi|4758958|ref|NP_004148.1| Human cAMP-dependent protein kinase  
MSHIQIPPGLTELLQGYTVEVLRQQPPDLVEFAVEYFTRLREARAPASVLPAAATPRQSLGHPPPEPGPDR  
VADAKGDSESEDEDLEVPVPSRFNRRVSVCAETYNPDEEEEDTDPRVIHPKTDEQRCRLQEACKDILLF  
KNLDQEQLSQVLDAMFERIVKADEHVIDQGDDGDNFYVIERGTYDILVTKDNQTRSVGQYDNRGS  
TSEGSLWGLDRVTFRRIIVKNNAKKRKMFESFIESVPLLKSLEVSERMKIVDVI GEK  
IYKGERIITQGEKADSFYIIESGEVSI LIRSRTKSNKDGGNQEVEIARCHKGQYFGELALVTNKPRAAS  
AYAVGDVKCLVMDVQAFERLLGPCMDIMKRNI SHYEEQLVKMFGSSVDLGNLGG  
  
[LIVMF] -G-E-x- [GAS] - [LIVM] -x(5,11) -R- [STAQ] -A-x- [LIVMA] -x- [STACV] .
```



Základy genomiky I.

- Role BIOINFORMATIKY v současném pojetí FUNKČNÍ GENOMIKY
- Databáze
 - Spektrum „on-line“ zdrojů
 - PRIMÁRNÍ, SEKUNDÁRNÍ a STRUKTURÁLNÍ databáze
 - GENOMOVÉ zdroje
- Analytické nástroje
 - Vyhledávání homologií
 - Vyhledávání sekvenčních motivů, otevřených čtecích rámců, restrikčních míst....



Analytické nástroje

- <http://workbench.sdsc.edu/>

Biology WorkBench
click here to toggle between menus and buttons
WE Moved! <http://workbench.sdsc.edu/>
Version 3.2

Session Tools Protein Tools **Nucleic Tools** Alignment Tools Structure Tools (Alpha)

beta-glucosidase

GBPLN:804655 *Hordeum vulgare* L. beta-glucosidase (BGQ60) gene, complete cds.
 GBPLN:170248 *Nicotiana tabacum* glucan beta-1,3-glucosidase gene, complete cds.

Select All Deselect All Ndjinn BATCH Add Edit Delete Copy View Download ViewRecords
BL2SEQ BL2SEQX BLASTN BLASTX TBLASTX FASTA FASTX FASTY SSEARCH CLUSTALW
CLUSTALWPROF ALIGN LALIGN LFASTA PATTERNMATCHDB PATTERNMATCH TACG PRIMERS
NASTATS BESTSCOR PFSCAN PRIMERCHECK PRIMERTM SIXFRAME REVCOMP RANDSEQ

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Center for Molecular Physiology and Experimental Research
MASARYK UNIVERSITY

Analytické nástroje

View
View Nucleic Sequence(s)

Format Case

[Download/view all sequences in text format](#)

[\[NEXT\]](#) [\[BOTTOM\]](#)

Nicotiana tabacum glucan beta-1,3-glucosidase gene, complete cds.
GBPLN:170248, 4699 bp

>170248
GAGCTCCCTTGGGGGGCAAGGGCAAAACTTTTTGCTAAATGGAAAAATATTATACCAAGTGTGTTGTAATA
GTTACTCAATTTGAATTAACAAGGGGCAAAATTTGACTATTTTTGCCCTTATATCTTTTTGGTCACAAAAAC
ATAAAATATCCCATCCGAAATTC AAAATGGTCCATTATCGGCAAGTAGCTTTCTTTAAATTATAGTTAGTT
GACAAAACACTATCAAGATATCATTATTATAATAATAACTTCAAAGTCCATCATCTTAGCTGCCTCCTCA
GTAGAGCCGCCAGTAAAAAAGACCGATCAAATAAAAGCCGCCATTTAAAAAATGAATTTTAGGACTCTC
GATTGGCACGTAAGTGCCAAAACCTCTTCCAATACTTTGCTGCAACTTGGGGCTGCTAGGTTCTGAGCTTC
CAGATATGGGATATTTCTAAGTTTATCTCCTAAATTTACATCTCAACTAATATTAAAGAAATTAACAGGTA
CAGCAAATCATAAAAATTTTCTCTAAAGAAGACAATGAATCCGGTTACTGATTCATTTGGCTTTTTAGAG
TCTGCATGCCATATTCAC TAAGGGGTCTTTTGGTAC AAGAAATAATAATAAATTTTCGGGATAGAATTT
GAGATTGCATTTATCTTGTGTTTTAATTTATAAGTATTAGCTAAATTTCAAGATAAATTTTACACTAAAAATAG
TAAAAATCAACTATCACATGTAGAAGGTGGAATGGAATAGCTAATCCCATAGCCACTCACATAGAATATCC
TTATTTATCTCACTATTTTTACCAAATGATCGGTTAGTCTTTCATGAGAATCCAGTATCCTCAATAAATGCA
GTAAGAAGTTAGAAAATTTTCATTAATCAATTCATATAATTTAAAAAATATTAGATATGGAGCACTTAAG
ATACAATAAAGATGTACCGTTAATAATAAAGATAAGATAGAGTTTTAAATAGGAAAAAAAAAACGGTT
CGAGACACTCTTATGGAAGGCCTTGTCTTCAAAGTAGATTCATTCATTGCTCTGGTGC AATAGCAAAA
TGACATCTTACTCTTAAGATACAGCGAGCCACTCTACAATCTTCTATTGTATACTCAAAATGAAAGTTTTA
GAGAACTTTCAATCTCTCACTACTTTTTAAGGGAAATTC AAAATACGACCAATATTTACTTTACTTTAC
TTATAGTTAAATGATATGAATTTTATTTTAAATTTGAATTGAAAAATTTAAATTTACTTTGATTTAATATAA



Analytické nástroje

Regex pattern:

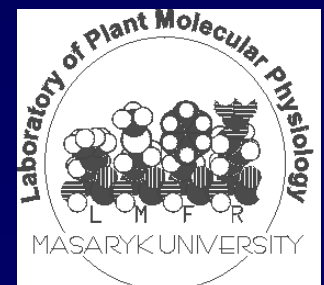
ctt. {1, 32}ctt

0 sequences were searched

1 match was found

Matches are indicated in blue

```
> 170248
GAGCTCCCTTGGGGGGCAGGGCAAACTTTTTGCTAAATGGAAAAATATTATACCAAGTGTITGTAATA
GTTACTCAATTTGAATTAACAAAGGGGCAAAATTTGACTATTTTGCCCTTATATCTTTTGGTCACAAAAAC
ATAAAATATCCCATCCGAAATTC AAATGGTCCATTTATCGGCAAGTAGCTTTCCTTTTAAATATAGTTAGTT
GACAAAACACTATCAAGATATCAITTTATAATAATAAACTTCAAAGTCCATCATCTTTAGCTGCCTCCTCA
GTAGAGCCGCCAGTAAAAAAGACCGATCAAAATAAAGCCGCAATTAATAATGAATTTTAGGACTCTC
GATTGGCACGTAAGTGC AAAACTCTTCCAATACTTTTGGCTGCAACTTGGGGCTGCTAGGTTCTGAGCTTC
CAGATATGGGATATTTCTAAGTTTATCTCCTAATTTACATCTCAACTAATTTAAGAAATFAAACAGGTA
CAGCAATCATAAAATTTTCCCTCTAAAGAAGACAAATGAATCCGGTTACTGTATTCATTGGCCTTTTTAGAG
CTTGCATGCCATATTCCTAAGGGGTCGTTTGGTACAAGAAATAATAATAATAATTTTGGGATAGAATTT
GAGATTGCATTTATCTTGTGTTTAAATATAAGTATTAGCTAATTTTCAAGATAAAATTTTACACTAAAATAG
TAAAATCAACTATCACATGTAGAAGGTGGAATGGAATAGCTAATCCATAGCCACTCACATAGAATATCC
TTATTTATCTCACTATTTTACCAATGATCGGTTAGTCTTTCATGAGAATCCAGTATCCTCAATAAATGCA
GTAAGAAGTTAGAAAAATTTTCAATTAATCAATTCATATAATTTAAAAATATTTAGATATGGAGCACTAAG
ATACAATAAAAGATGTACCCTTAATAATAAAGATAAGATAGAGTTTTTAAATAGGAAAAAAAAAACGGTT
CGAGACACTCTTTATGGAAAGCGTTTGTCTTCAAAGTAGATTCTCATTCATTGCTCTGGTGCAATAGCAAAA
TGACATCTTACTCTTAAGATACAGCGAGCCACTCTACAATCTTCTATTGTATACTCAAAATGAAAGTTTTTA
GAGAACTTTTCAAATCTCTCAACTACTTTTAAAGGGAATTC AAAATACGACCAATATTTATTTACTTACTTAC
TTATAGTTAAATGATATGAATTTTTATTTTAAATTTGAATGAAAAATTTAAATTACTTTGATTTAATATAA
ACAATAGATATCGCTAAGTATTTACCACAAACATGGAGATACTACAGAAGATTTTATTTATTTGTAAACGAT
GATTAAGCAGCTATTCATCTGGTTGTGCAGGATGAAAAGAAAGTAACTAGCTATAATTTCTTTTGTAAAGT
```



Analytické nástroje

Frame 1, 1 stop codon

Nicotiana tabacum glucan beta-1,3-glucosidase gene, complete cds. Tran

>170248 Translated - Frame 1
ELPWGARAKLFAKWKNIIIPSVCSYSI*INKGANLTILPL

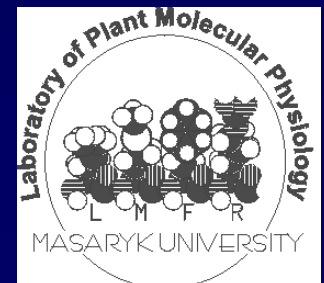
```
      E L P W G A R A K L F A K W K N I I P S
1    gagtcaccttggggggcaagggcaaaaactttttgctaaatggaaaaatattataccaagt 60
      V C N S Y S I * I N K G A N L T I L P L
61   gtttgaatagttactcaatttgaattaacaaaggggcaaatgactatgcttgcctta 120
```

Frame 2, 1 stop codon

Nicotiana tabacum glucan beta-1,3-glucosidase gene, complete cds. Tran

>170248 Translated - Frame 2
SSLGGQGQNFLLNGKILYQVFE LTKGQI*LFCP

```
      S S L G G Q G Q N F L L N G K I L Y Q V
2    agtcaccttggggggcaagggcaaaaactttttgctaaatggaaaaatattataccaagt 61
      F V I V T Q F E L T K G Q I * L F C P
62   tttgtaatagttactcaatttgaattaacaaaggggcaaatgactatgcttgcctta 120
```



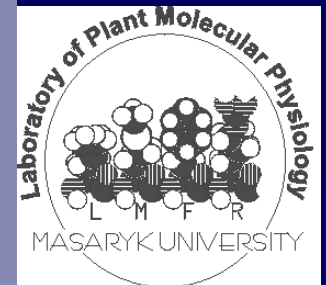
Analytické nástroje

Selected Sequence(s)

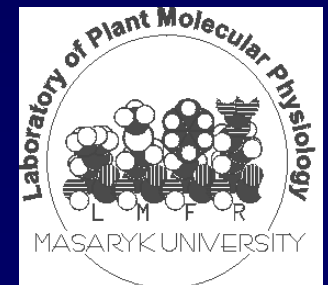
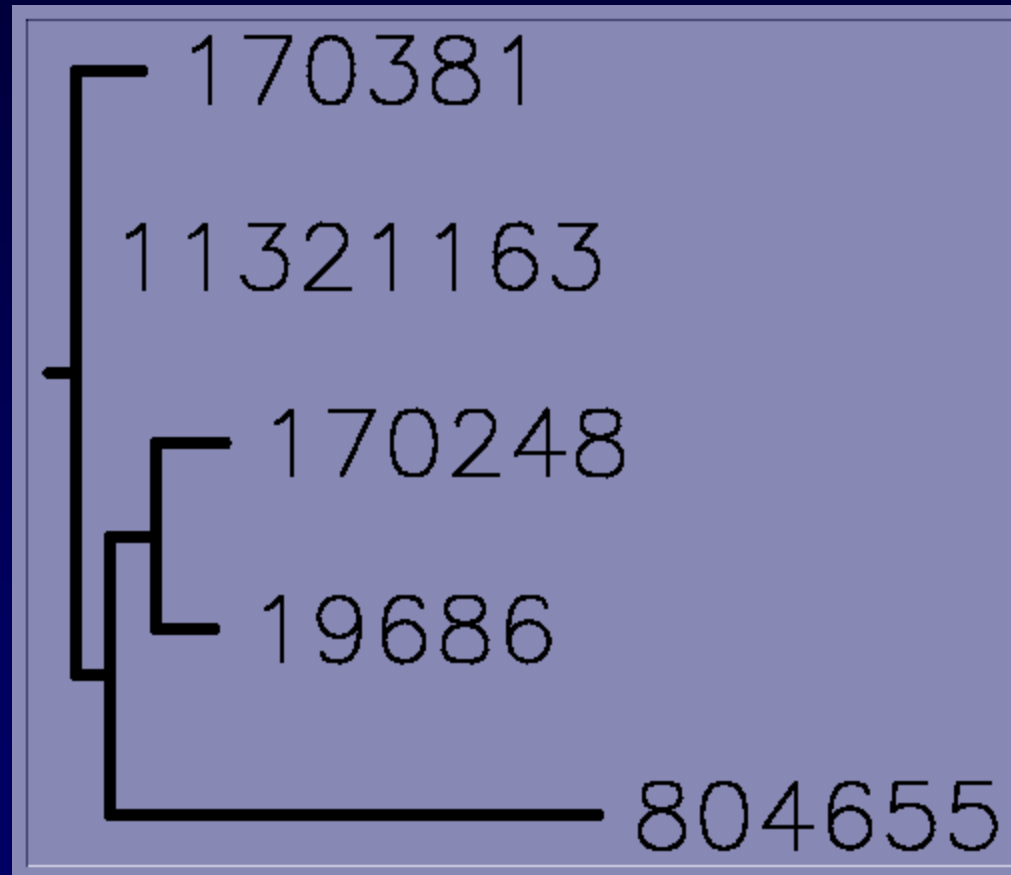
- *Lycopersicon esculentum* beta-1,3-glucanase mRNA, complete cds.,
- *Capsicum annuum* clone GC170 beta-1,3-glucanase-like protein gene.,
- *Nicotiana tabacum* glucan beta-1,3-glucosidase gene, complete cds.,
- *Nicotiana plumbaginifolia* beta-(1,3)-glucanase gene for a vacuolar,
- *Hordeum vulgare* L. beta-glucosidase (BGQ60) gene, complete cds.

[Download a PostScript version of the output](#)

```
.....
2560 GTTTGCTTGGTCTGTGCTTGAAGACTTGGAGTGGAGACTGGGCTAGACTGGGGTTTGGG 804855
      2550      2560      2570      2580      2590      2700
24 ..... AATTGGT. 170381
1 ..... 11321163
2430 ..... CAAGCAATT. 170248
1743 CAGTCAAATCATTGACACAACTGCAAAAAACAAGCCAAAATGGTAAAAAATAAATTC 19686
2620 GATCGTCTATGTGGACTTGAATACTCTGAAGAGGTAGCCGAAAGGACTGAGGCTTTGGTT 804855
      2710      2720      2730      2740      2750      2760
32 ..... ATTAATGCTGCTTCAAGCATTCTTGTGGCA. CCAAGATTGAGATAAG 170381
1 ..... 11321163
2438 ..... AG.ATAATGATTTAGTTTCTAAGCACTAATT.CAATCTTATTGAGC 170248
1803 AGCATGTTTAGAATTCCTATGTGCAAGCCGCACTGACTATTTCTAATTCATATTGAGC 19686
2680 CAAGAAGATGCTCCGGAAGAAGAGGAGCTAGGATCGCAAAACAGCATCGGGAGGATC 804855
      2770      2780      2790      2800      2810      2820
79 AGAGGCTCAA... ATAGGCT... TTCTATGGAATGATCCCAAGCAAGTTGCCATGAG 170381
1 T... ATCGGCT... TTGCTATGGAATGATCCCAAGCAAGTTGCCATGAG 11321163
2484 AGCGGCTCAATCAATAGGCT... TTGCTATGGAATGATCCCAAGCAAGTTGCCAATC 170248
1863 AGCGGCTCAATCAATAGGCT... TTGCTATGGAATGATCCCAAGCAAGTTGCCAATC 19686
2740 AGCTGCTTCAAGCTTCAATAAAAAAGATAATGTAATGCTTTTCTATGCAAGCTGAG
      2830      2840      2850      2860      2870      2880
132 ATTCTGAAGTTATACAGC.. TCTACAAGTCCAGAAAATTAAGAACTGAGGCTTTATGA 170381
45 ATTCGGAAGTTATACAGC.. TCTACAAGTCCAGAAAATTAAGAACTGAGGCTTTATGA 11321163
2540 ATTCTGAAGTTATACAGC.. TCTACAAGTCCAGAAAATTAAGAACTGAGGCTTTATGA 170248
1919 ATTCGGAAGTTATACAGC.. TCTACAAGTCCAGAAAATTAAGAACTGAGGCTTTATGA 19686
2800 ACTTACGGCTCTTGGCTAAAGAGCACTGCGCAATATGCGCAGCGCGCAAAITGGAG 804855
```




Analytické nástroje



Analytické nástroje

- VPCR <http://elanor.sci.muni.cz/LMFR/vpcr/>

SEARCH  [ABOUT](#) [DOWNLOAD](#) [LINKS](#)

VPCR 2.0 (WWW interface) - Please, enter nucleotide primer sequences ([IUB codes](#) allowed for degenerate primers). VPCR 2.0 searches the specified database for matches to the primers. If matches are found within 10000 bases, a PCR simulation model predicts amplification. Calculated PCR products are displayed within a minute.

NOTE: Abilities of VPCR 2.0 are still limited by BLAST capabilities and settings, as well as inability of our current software to deal with more than a couple thousand matches per primer. For example, using primers shorter or roughly equal to our 11-base word size misses most matches. Primers with overrepresented sequences cause problems as well. We are now busy solving most of these problems, please, be patient. If you have a minute, please, let us know what kind of expectations you have for VPCR 2.0 etc. Currently, this address is for testing VPCR 2.0, stable features will be installed on [VPCR 2.0 Homepage](#).

Search using in the database for

Primer 1

Primer 2

Primer 3

Primer 4


Primer 5

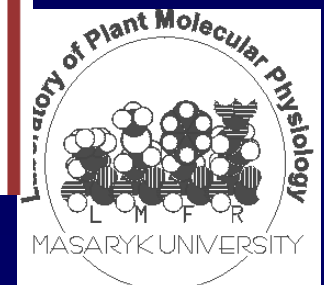
Primer 6

Primer 7

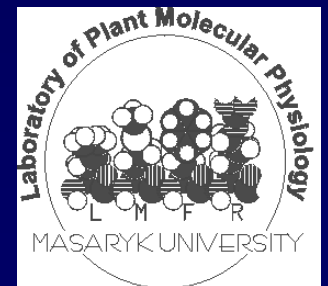
Primer 8

Annealing temperature



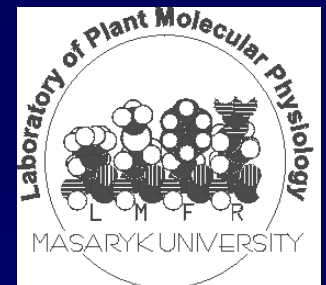


Analytické nástroje



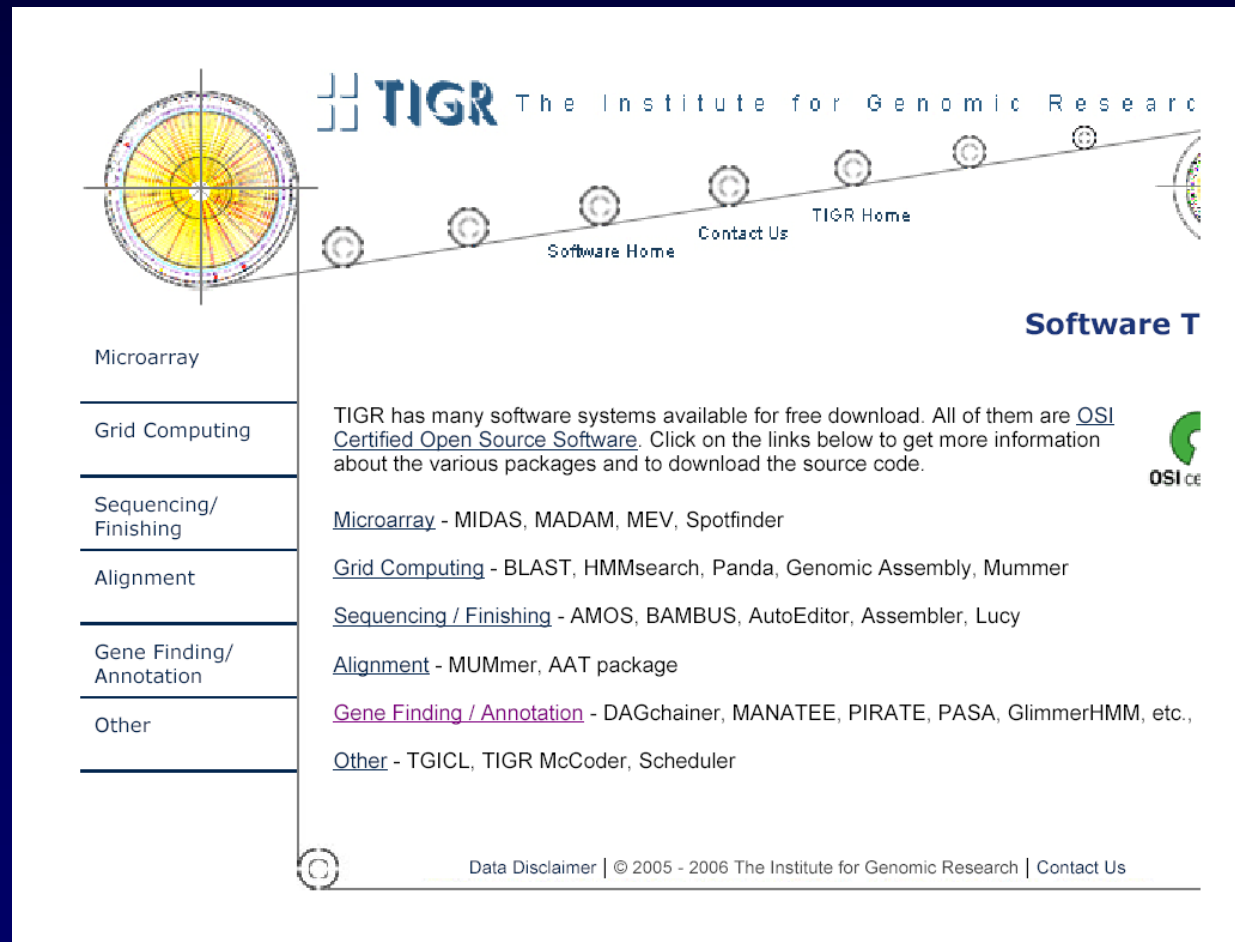
Základy genomiky I.

- Role BIOINFORMATIKY v současném pojetí FUNKČNÍ GENOMIKY
- Databáze
 - Spektrum „on-line“ zdrojů
 - PRIMÁRNÍ, SEKUNDÁRNÍ a STRUKTURÁLNÍ databáze
 - GENOMOVÉ zdroje
- Analytické nástroje
 - Vyhledávání homologií
 - Vyhledávání sekvenčních motivů, otevřených čtecích rámců, restrikčních míst....
 - Další [www](#) genomové nástroje



www analytické nástroje

- TIGR (The Institute for Genomic Research, <http://www.tigr.org/software/>)



The screenshot shows the TIGR website's software page. At the top left is a circular genomic map. To its right is the TIGR logo and the text 'The Institute for Genomic Research'. A navigation bar contains links for 'Software Home', 'Contact Us', and 'TIGR Home'. Below this is a 'Software T' section with a table of categories and their corresponding software tools. A copyright notice is at the bottom left, and a disclaimer is at the bottom center. On the right side, there is an OSI logo and a logo for the Institute of Plant Molecular Physiology at Masaryk University.

TIGR The Institute for Genomic Research

Software Home Contact Us TIGR Home

Software T

Microarray	
Grid Computing	TIGR has many software systems available for free download. All of them are OSI Certified Open Source Software . Click on the links below to get more information about the various packages and to download the source code.
Sequencing/Finishing	Microarray - MIDAS, MADAM, MEV, Spotfinder
Alignment	Grid Computing - BLAST, HMMsearch, Panda, Genomic Assembly, Mummer Sequencing / Finishing - AMOS, BAMBUS, AutoEditor, Assembler, Lucy
Gene Finding/Annotation	Alignment - MUMmer, AAT package Gene Finding / Annotation - DAGchainer, MANATEE, PIRATE, PASA, GlimmerHMM, etc.,
Other	Other - TGICL, TIGR McCoder, Scheduler

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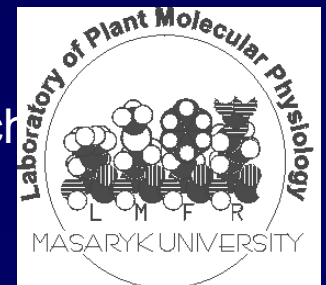
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Institute of Plant Molecular Physiology
MASARYK UNIVERSITY

Základy genomiky I.

shrnutí

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dotazy

