Helwig

Introductional thoughts

Entrez

DNA / Gene

Genor

Protein:

Phenotype

Summary

Databases in bioinformatics

NCBI resources & GenBank

Till Helge Helwig

Proseminar "Genome Bioinformatics" (Dr. Kay Nieselt)

22.05.2007

Introductional thoughts

2 Entrez

3 DNA / Genes

Genome

6 Proteins

6 Phenotypes

7 Summary

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Introductional thoughts

Entrez

DNA / Gene

Genor

Profess

Phenotype

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Introductional thoughts

- many research groups all over the world
- huge amount of data generated daily
- interpretation impossible without comparing to other data
- collecting all data in a central repository is essential

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Entrez

DNA / Gen

Genor

Protein

Phenotype

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Introductional thoughts

- many research groups all over the world
- huge amount of data generated daily
- interpretation impossible without comparing to other data
- collecting all data in a central repository is essential
- 1988: creation of the NCBI

Introductional thoughts

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What is the NCBI?

- National Center for Biotechnology Information
- project of the National Library of Medicine (NLM)
- thus belongs to the National Institute of Health (NIH)
- primarily a project to build information systems for molecular biology
- today a huge collection of databases and tools is available on the website http://www.ncbi.nlm.nih.gov

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How to access a huge database?

- many different databases
- huge amounts of data
- lots of very different queries
- results readable and usable for everybody

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How to access a huge database?

- many different databases
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- results readable and usable for everybody
- Entrez The Life Sciences Search Engine

The main component of the NCBI resources

Entrez

- integrated database retrieval system
- links all NCBI resources together
- provides access to more than 91 million DNA and protein sequences
- the whole NCBI website is searchable, too
- accessible via webbrowser or using the "E-Utilities"

Collecting information from everywhere

 search results are extended with links to biomedical literature using PubMed and PubMed Central

- 16.5 million citations
- 750 000 fulltext-articles, including whole articles from well known science journals
- LinkOut provides links to external projects and resources
- most query results also contain related data from other NCBI databases and direct links to tools

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 using "My NCBI" you can customize the behaviour of Entrez

- query results can be shown directly, sent by email or provided as RSS feed
- the output is possible in many different formats including:
 - FastA format
 - XML documents
 - GenBank flat file
 - ..

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DNA / Con

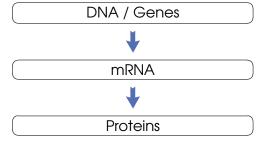
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Central dogma of biology



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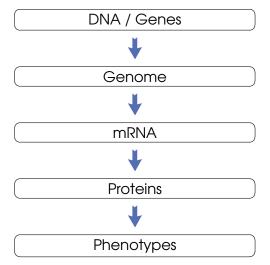
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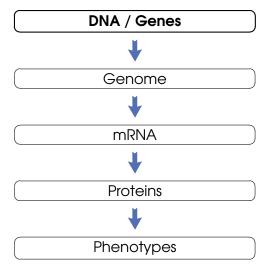
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Protein

Phenotype

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GenBank General information

- fastest growing and therefore largest public database of nucleotide sequences
- currently about 61 million sequences from more than 240 000 organism recorded
- collaboration with DDBJ and EMBL
- each entry contains:
 - unique accession id, e.g. AC202656, which is shared in DDBJ and EMBL
 - description & scientific name
 - biologically relevant sections (mutation sites, ...)
 - taxonomy information & literature references



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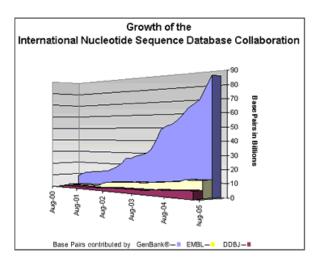
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GenBank (2)



Source: http://www.ncbi.nlm.nih.gov/Genbank/index.html



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Summary

GenBank (3)

Organization of the records

- entries are stored in divisions
- first divisions were build from taxonomic relations: Bacteria (BAC), Viruses (VRL), Primates (PRI) and Rodents (ROD)
- later other divisions were added describing the used sequencing strategy:
 Expressed sequence tag (EST), Genome survey (GSS), High throughput genomic (HTG), ...
- today there are 18 divisions

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Попотурс

Gene databases Entrez Gene

- gene-specific information
- focus is set on completely sequenced genomes
- actively researched genomes are included as well
- database is build by information accumulated by the NCBI staff and international collaborations
- all entries are linked to other resources

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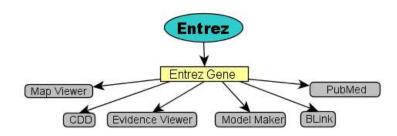
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Gene databases (2)

Entrez Gene - The infrastructure



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Summary

Gene databases (3)

UniGene & ProtEST

UniGene

- contains clusters of sequences from GenBank describing one unique gene
- attempts to handle the redundancy of the GenBank entries for selected organisms
- 87 000 clusters for human sequences in the release from 2006

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Summary

Gene databases (3)

UniGene & ProtEST

UniGene

- contains clusters of sequences from GenBank describing one unique gene
- attempts to handle the redundancy of the GenBank entries for selected organisms
- 87 000 clusters for human sequences in the release from 2006

ProtEST

 provides pre-computed protein alignments and translations for UniGene entries



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Gene databases (4)

HomoloGene & dbMHC

HomoloGene

- collection of homologs among the genes of 18 completely sequenced eukaryotic genomes
- finding homologs is difficult and a typical problem of bioinformatics
- records are linked to information from OMIM and COGs

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Gene databases (4)

HomoloGene & dbMHC

HomoloGene

- collection of homologs among the genes of 18 completely sequenced eukaryotic genomes
- finding homologs is difficult and a typical problem of bioinformatics
- records are linked to information from OMIM and COGs

dbMHC

- MHC is of high interest to researchers, because it encodes the HLA
- information on variations in the relevant genes are collected all over the world
- integrated databases for several other medical relevant topics



Genome

Proteins

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Gene databases (5)

dbSNP & RefSeq

dbSNP

- records about single nucleotide polymorphisms (SNP)
- 12 million entries about the human genome, 22 million about other organisms
- SNPs are displayed graphically

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Gene databases (5)

dbSNP

- records about single nucleotide polymorphisms (SNP)
- 12 million entries about the human genome, 22 million about other organisms
- SNPs are displayed graphically

RefSeq

- information about DNA, RNA and proteins of major research organisms
- attempt to build non-redundant data sets from the huge amount of information in the other databases
- currently about 4.1 million sequences represent about 3700 organisms



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Gene tools

ORF, Splign & Spidey

- Open reading frame finder (ORF)
 - takes sequence or GenBank accession number
 - searches all possible reading frames of a specified length

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Summary

Gene tools

ORF, Splign & Spidey

Open reading frame finder (ORF)

- takes sequence or GenBank accession number
- searches all possible reading frames of a specified length

• Splign

 aligns cDNA with genomic DNA or spliced sequences (Needleman-Wunsch + BLAST)

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Summary

Gene tools ORF, Splign & Spidey

Open reading frame finder (ORF)

- takes sequence or GenBank accession number
- searches all possible reading frames of a specified length

• Splign

 aligns cDNA with genomic DNA or spliced sequences (Needleman-Wunsch + BLAST)

Spidey

 creates alignments of eukaryotic mRNA with single genomic sequences using a splice-site model

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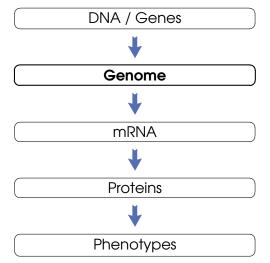
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Genome databases

- 370 complete microbial genomic sequences
- 2 500 viral sequences
- 1 050 reference sequences for eukaryotic organelles
- 20 genomes from higher organisms
- results are linked to resources for graphical views of sequences
- relevant COGs are included in the results as well as pre-calculated neighbours for microbial genomes

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Genome databases (2)

Trace Archives & Genome Project

The Trace Archives

- 1.3 billion traces (raw sequence data from sequencing projects) stored currently
- 860 organisms represented

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Genome databases (2)

Trace Archives & Genome Project

• The Trace Archives

- 1.3 billion traces (raw sequence data from sequencing projects) stored currently
- 860 organisms represented

Genome Project

- records about status, progress and results of sequencing projects
- keeps track even of projects that have not yet produced results
- many biological facts are added to each description

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Summary

Genome tools Map Viewer

Map Viewer

- maps for every zoom factor are available from single genes to complete chromosomes
- genetic and physical markers can be shown along the sequence
- taxonomic list shows organisms for which maps are available
- accessed by many other resources to display query results (e.g. Entrez Gene)

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Special resources

- resources for medical purposes
 - databases containing information on specific topics like influenza or cancer
 - opportunity for quick check on pathogenicity of an organism

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Special resources

- resources for medical purposes
 - databases containing information on specific topics like influenza or cancer
 - opportunity for quick check on pathogenicity of an organism
- resources for organization of databases
 - Clusters of Orthologous Groups (COGs) combine information from completely sequenced prokaryotic protein sequences
 - similar database available for eukaryotic proteins (KOGs)

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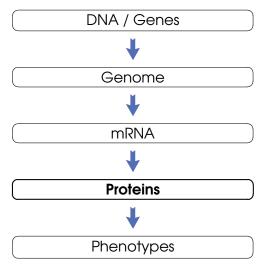
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Genor

Proteins

Phenotype

Summary

Protein resources

• BLAST Link (BLink)

produces pre-calculated alignments of protein sequences

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Proteins

Phenotype

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Protein resources

BLAST Link (BLink)

- produces pre-calculated alignments of protein sequences
- Open Mass-Spectometry Search Algorithm
 - OMSSA helps to identify spectra taken from tandem mass spectometry
 - similiar to BLAST as it calculates an "Expect-Value"

Proteins

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Protein resources

BLAST Link (BLink)

produces pre-calculated alignments of protein sequences

Open Mass-Spectometry Search Algorithm

- OMSSA helps to identify spectra taken from tandem mass spectometry
- similiar to BLAST as it calculates an "Expect-Value"

Molecular Modeling Database (MMDB)

- The molecular modeling database is built from entries of the Protein Database (PDB)
- contains information of experimentally found 3D structures in biomolecules

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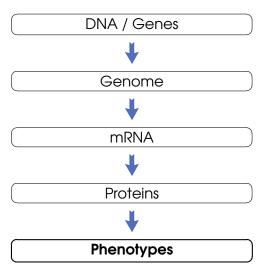
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Phenotypes

Summary

Phenotype databases

- Online Mendelian Inheritance in Man (OMIM)
 - list of genes and genetic disorders
 - connection to disease phenotypes
 - 17 000 entries

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Phenotypes

Summary

Phenotype databases

- Online Mendelian Inheritance in Man (OMIM)
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- Online Mendelian Inheritance in Animals (OMIA)
 - same as OMIM but with information taken from animals

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Phenotypes

Summary

Phenotype databases

- Online Mendelian Inheritance in Man (OMIM)
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- Online Mendelian Inheritance in Animals (OMIA)
 - same as OMIM but with information taken from animals
- GENSAT
 - atlas of gene expressions in the central nervous system of the mouse

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Phenotypes

Summary

Phenotype databases (2)

- Gene Expression Omnibus (GEO)
 - repository for high-throughput data
 - results of microarray experiments, serial analysis of gene expressions (SAGE) experiments, mass spectometry peptide profiling, ...
 - about 3 billion measurements are recorded

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Summary

Summary

- databases, tools and other resources for every imaginable kind of information
- effective search through all these with Entrez
- heavy linkage between the resources
- visit the homepage and take a look around yourselves: http://www.ncbi.nlm.nih.gov

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Summary

Summary

- databases, tools and other resources for every imaginable kind of information
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Thank you for listening.