## HC70AL Summer 2014

## **An Introduction to Bioinformatics**

by Kelli Henry

August 14, 2014



DNA Genetic Code of Life



Entire Genetic Code of a Bacteria



**DNA** Fingerprinting



Cloning: Ethical Issues and Future Consequences



Plants of Tomorrow

# A New Comparative Genomics Field Has Emerged Allowing the Comparison of Entire Genomes!



### Comparison of Mammalian Genomes Attempts To Determine "What Makes a Man, a Man and a Mouse a Mouse"

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There is <1% Difference Between Human & Chimpanzee DNAs!



### DNA Genetic Code of Life



entire Genetic Code of a Bacteria



**DNA** Fingerprinting



Cloning: Ethical Issues and Future Consequences



### Plants of Tomorrow

## SNPS Can Be Used To Trace Human Populations & Origins

### **GENETIC PROSPECTING**



#### 1. African Cradle

Most paleoanthropologists and geneticists agree that modern humans arose some 200,000 years ago in Africa. The earliest modern human fossils were found in Omo Kibish, Ethiopia. Sites in Israel hold the earliest evidence of modern humans outside Africa, but that group went no farther, dying out about 90,000 years ago.

#### 2. Out of Africa

Genetic data show that a small group of modern humans left Africa for good 70,000 to 50,000 years ago and eventually replaced all earlier types of humans, such as Neandertals. All non-Africans are the descendants of these travelers, who may have migrated around the top of the Red Sea or across its narrow southern opening.

#### 3. The First Australians

Discoveries at two ancient sites—artifacts from Malakumanja and fossils from Lake Mungo indicated that modern humans followed a coastal route along southern Asia and reached Australia nearly 50,000 years ago. Their descendants, Australian Aborigines, remained genetically isolated on that island continent until recently.



#### 4. Early Europeans

Paleoanthropologists long thought that the peopling of Europe followed a route from North Africa through y's the Levant. But genetic data show that the DNA of today's western Eurasians resembles that of people in India. It's possible that an inland migration from Asia seeded Europe between 40,000 and 30,000 years ago.

#### 5. Populating Asia

Around 40,000 years ago, humans pushed into Central Asia and arrived on the grassy steppes north of the Himalaya. At the same time, they traveled through Southeast Asia and China, eventually reaching Japan and Siberia. Genetic clues indicate that humans in northern Asia eventually migrated to the Americas.

#### 6. Into the New World

Exactly when the first people arrived in the Americas is still hotly debated. Genetic evidence suggests it was between 20,000 and 15,000 years ago, when sea levels were low and land connected Siberia to Alaska. Ice sheets would have covered the interior of North America, forcing the new arrivals to travel down the west coast

# The Shaping of Modern Human Immune Systems by Multiregional Admixture with Archaic Humans

www.sciencemag.org SCIENCE VOL 334 7 OCTOBER 2011

Comparing 40,000 Year-Old Fossil Genomes to Our Genome Reveals Ancient "Matings" Between Diffferent Human Ancestor Lineages!!



We Have Neanderthal & Denisovan Genes in Our Chromosomes

It's All in the DNA!

Nature Reviews | Genetics September, 2011

### What is **BLAST**?

**Basic Local Alignment Search Tool (BLAST)** 

### What does **BLAST** do?

A family of programs that allows you to input a query sequence and compare it to DNA or protein sequences in a database.

### What Are the Different BLAST Programs?





NCBI/ BLAST Home

BLAST finds regions of similarity between biological sequences. more...

Saved Strategies

Help

The Basic Local Alignment Search Tool (BLAST) finds regions of local similarity between sequences. The program compares nucleotide or protein sequences to sequence databases and calculates the statistical significance of matches. BLAST can be used to infer functional and evolutionary relationships between sequences as well as help identify members of gene families.

### BLAST Assembled RefSeg Genomes

Choose a species genome to search, or list all genomic BLAST databases.

- Human
- Mouse
- Rat
- Cow
- Pig

- Dog Rabbit Chimp
  - Guinea pig
  - Sheep

Fruit fly
Honey bee

- Chicken
- Zebrafish
- Clawed frog

- Arabidopsis
- Rice
- Yeast
- Neurospora crassa
- Microbes

### Basic BLAST

Choose a BLAST program to run.

nucleotide blast	Search a <b>nucleotide</b> database using a <b>nucleotide</b> query <i>Algorithms:</i> blastn, megablast, discontiguous megablast
protein blast	Search <b>protein</b> database using a <b>protein</b> query <i>Algorithms:</i> blastp, psi-blast, phi-blast, delta-blast
<u>blastx</u>	Search protein database using a translated nucleotide query
<u>tblastn</u>	Search translated nucleotide database using a protein query
tblastx	Search translated nucleotide database using a translated nuc

Note: The Arabidopsis Information Resources (TAIR) (http://arabidopsis.org/) has a **BLAST** program specific to Arabidopsis

leotide query

### http://blast.st-va.ncbi.nlm.nih.gov/Blast.cgi

**Obtaining Sequences From the UCLA Sequencing Facility** 

GENOSEO UCLA GENOTYPING & SEQUENCING	WebS	eq
<u>Help</u>	Welcome to WebSeq	
Sequencing & Genotyping	Username:	goldberg_r
<u>core nome Page</u>	Password:	•••••
		Login

### http://www.genetics.ucla.edu/webseq/

# Selecting Sequence to Download



Sequencing

R2R Lookup

<u>Taqman</u>

Genotyping

Summary

Password

Help

Logout

Core Home Page

# WebSeq

#### Sequencing files for Robert B. Goldberg R2R Signup (List) Filter files: (only files with this string in their name will be displayed) R2R Signup (Plate) Select None (Select Range) (?) Download selected Select All Full Service Signup Sorted by date - click on column header to sort by that column. Only the first 1000 files are displayed; to see the full list, filter for "." Full Service Lookup Files older than 90 days will not be available. Select Filename Size Date Tagman Signup (384) 140545GoldR KH1658 H10.ab1 338999 31 Jul 2014 Tagman Signup (96) 140545GoldR KH1659 A12.ab1 320996 31 Jul 2014 Click to LightCycler 480 Signup 140328GoldR\_KH1657\_G04.ab1 325580 18 Jul 2014 download 140258GoldR\_KH1655\_D03.ab1 337431 17 Jul 2014 your 315046 17 Jul 2014 140258GoldR KH1656 E03.ab1 140034GoldR\_KH1651\_A12.ab1 337078 03 Jul 2014 sequence 140034GoldR\_KH1652\_B12.ab1 324386 03 Jul 2014 140034GoldR KH1653 C12.ab1 338397 03 Jul 2014 319160 140034GoldR KH1654 D12.ab1 03 Jul 2014 Sequencing & Genotyping 139943GoldR\_KH1647\_A12.ab1 337773 27 Jun 2014 139943GoldR\_KH1648\_B12.ab1 324774 27 Jun 2014 139943GoldR KH1649 C12.ab1 338079 27 Jun 2014 335365 27 Jun 2014 139943GoldR\_KH1650\_D12.ab1 25 Jun 2014 139887GoldR\_KH1637\_A11.ab1 336985 139887GoldR KH1638 B11.ab1 332496 25 Jun 2014 139887GoldR\_KH1639\_C11.ab1 336603 25 Jun 2014 139887GoldR\_KH1640\_D11.ab1 326932 25 Jun 2014 139887GoldR\_KH1641\_E11.ab1 337227 25 Jun 2014 334507 25 Jun 2014 139887GoldR\_KH1642\_F11.ab1 139887GoldR\_KH1643\_G11.ab1 337335 25 Jun 2014 139887GoldR\_KH1644\_H11.ab1 322988 25 Jun 2014 139887GoldR KH1645 A12.ab1 337273 25 Jun 2014 139887GoldR KH1646 B12.ab1 313747 25 Jun 2014

Download selected Select All Select None (Select Range ) (?)

Please consult detailed help for assistance using WebSeq. To contact us regarding WebSeq, please email webseq@genetics.ucla.edu

Copyright (C) 2002-2014 UCLA Human Genetics Last modified 07 August 2014 00:34:54.

# **Reading the DNA Sequence**



MAC - http://mac.softpedia.com/get/Math-Scientific/FinchTV.shtml

PC - http://www.softpedia.com/get/Science-CAD/FinchTV.shtml

### 4Peaks

MAC - http://www.mekentosj.com/science/4peaks

### What Gene Are You Working With?

Task: Use the DNA sequence you've obtained from sequencing with your gene-specific salk primer to identify your gene

Tools: The Arabidopsis Information Resources (TAIR) (http://arabidopsis.org/)

### **Procedure:**

- 1. Go to the TAIR BLAST page (http://arabidopsis.org/Blast/index.jsp)
- 2. Select BLASTN
- 3. For Dataset, Select "TAIR10 Genes"
- 4. Paste your sequence
- 5. Run BLAST

### **Results/Question:**

1. What is the gene that you're working with?

### How to Perform a BLAST Search for Arabidopsis

Browse       Tools       Portals       Download       Submit       News       ABRC Stocks         onne > Tools > BLAST         ALR BLAST 2.2.8         sease note that this form uses NCBI BLAST2.2.8, and NOT WU-BLAST2.0.         Blast         BLAST <sup>114</sup> program         Datasets:         Input:         TAREID Genes (+Introns, +UTRS) (DNA)         Input:         Pase sequence         (Att g01030)         Upload a file         Results Return Options         Output Title         email:         Input:         Optional, will be added to output for your use         Results Return Options         Output Title         Results Format:         In your web browser:         In your web browser:         Plase be pastient. Replies directly to your browser may take a minute or two. Selecting an e-mail reply allows you to immediately request more searches.	tair	Home Help	Contact Abo	ut Us Login/Regi	ster						
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### Anatomy of a BLAST Result -- Part I BLAST query on TAIR10 Genes (+introns, +UTRs) (DNA) sequences

Query performed by the The Arabidopsis Information Resource (TAIR) for full BLAST options and parameters, refer to the NCBIBLAST Documentation

Your comments and suggestions are requested: Send a Message to TAIR

BLASTN 2.2.24 [Aug-08-2010]

Reference: Altschul, Stephen F., Thomas L. Madden, Alejandro A. Schäffer, Jinghui Zhang, Zheng Zhang, Webb Miller, and David J. Lipman (1997), "Gapped BLAST and PSI-BLAST: a new generation of protein database search programs", Nucleic Acids Res. 25:3389-3402. Query= user-submitted sequence (3396 letters) Database: TAIR10 Genes (+introns, +UTRs) (DNA) 41,671 sequences; 93,974,285 total letters Searching.....done Arabidopsis Gene Identification (AGI) Gene description E value number Score  $\mathbf{E}$ Sequences producing significant alignments: (bits) Value Top hit AT4G12750.1 Symbols: | Homeodomain-like transcriptional ... 1193 0.0 Symbols: atnudt17, NUDT17 | nudix hydrolase h... AT2G01670.1 48 0.001 AT2G35020.1 Symbols: GlcNAc1pUT2 | N-acetylglucosamine-1-... 44 0.017 42 0.066 AT2G38370.1 Symbols: | Plant protein of unknown function... AT2G25800.1 40 0.26 Symbols: | Protein of unknown function (DUF8... AT5G13370.1 Symbols: | Auxin-responsive GH3 family prote... 38 1.0 AT5G44230.1 Symbols: | Pentatricopeptide repeat (PPR) su... 38 1.0 38 1.0 AT1G12560.1 Symbols: ATEXPA7, EXP7, ATEXP7, ATHEXP ALPHA ... AT1G43830.1 Symbols: | transposable element gene | chrl:... 38 1.0 36 4.1 AT5G27395.2 Symbols: | Mitochondrial inner membrane tran... 36 4.1 AT5G23510.2 Symbols: | unknown protein; LOCATED IN: cell... AT5G27395.1 Symbols: Mitochondrial inner membrane tran... 36 4.1 36 4.1 AT5G23510.1 | Symbols: | unknown protein; BEST Arabidopsis... 36 AT5G20350.1 Symbols: TIP1 | Ankyrin repeat family protein... 4.1

### Anatomy of a BLAST Result -- Part II



**Genes Have a Unique Position in the Genome!** 

### Task: Where is your gene located in the genome?

### Tools: The Arabidopsis Information Resources (TAIR) (http://www.arabidopsis.org)

### **Procedure:**

- 1. Go to the Locus Detail page for your gene
- 2. Under Map Links, select Sequence Viewer
- 2. Hover your mouse over your highlighted AGI number. A Locus box appears.
- 3. Select nucleotide seq view at the bottom of the box.

### **Results/Question:**

- 1. What chromosome is your gene in?
- 2. What other genes/markers are next to your gene?
- 3. What is the exact position of your gene in the genome?



### 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, 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 week from the latest published research literature and community data submissions. 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.

TAIR is located at Phoenix Bioinformatics and funded by subscriptions.

Full access to TAIR requires a subscription. ABRC catalog, stock and ordering pages hosted at TAIR will remain freely accessible. Please see our subscription page for further details.

- 🔊 Subscribe to news feed
- 🕒 Follow our Twitter feed
- Join our Facebook group

### **Breaking News**

#### TAIR is going to ICAR2014. [July 23, 2014] We're looking forward to seeing

you in Vancouver. Bring your questions to our workshop (M, 7/28, 4:30-5:30 pm) or our booth.

### ABRC discontinuing distribution of ESTs.

[July 21, 2014] ABRC will no longer be distributing ESTs after July 31, 2014. Individual stock detail

### **Gene Search Results**

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### **Locus Detail Page**

## Click to get DNA sequence (including exons, introns and UTRs)

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### **SeqViewer**



Click view your gene sequence on the chromosome

### **Nucleotide Sequence Viewer**



Note that Arabidopsis has 5 chromosomes.

### **Where Do Your Primers Anneal?**

Task: Where do your gene-specific salk primers anneal relative to your gene?

Tools: The Arabidopsis Information Resources (TAIR) (http://arabidopsis.org/)

**Procedure:** 

- 1. Go to the TAIR BLAST page (http://arabidopsis.org/Blast/index.jsp)
- 2. Select BLASTN
- 3. For Dataset, select "TAIR10 Genes"
- 4. Paste your primer sequence
- 5. Run BLAST

Note: If there are no BLAST results, try selecting a different dataset, such as Upstream Sequences.

### **Results/Question:**

1. Where do your gene-specific salk forward and reverse primers anneal relative to your gene?

Task: Where is the T-DNA inserted relative to your gene?

Tools: The Arabidopsis Information Resources (TAIR) (http://arabidopsis.org/)

**Procedure:** 

- 1. Go to the TAIR BLAST page (http://arabidopsis.org/Blast/index.jsp)
- 2. Select BLASTN
- 3. For Dataset, select "TAIR10 Genes"
- 4. Paste your sequence
- 5. Run BLAST

Note: If there are no BLAST results, try selecting a different dataset, such as Upstream Sequences.

### **Results/Question:**

- 1. At what position in your gene is the T-DNA inserted?
- 2. Is the T-DNA inserted in an exon? Intron? UTR? Upstream sequence?
- 3. What is the orientation of the T-DNA relative to your gene?

# **SALK T-DNA Lines** How Were T-DNA Lines Created?



# **SALK T-DNA Lines** How Were T-DNA Lines Created?



Locus Detail	etair "	ome Help Contact A	bout Us Login/Regis	ter		Gene	\$ Search
	Search Bro	wse Tools	Portals	Download	Submit	News	ABRC Stocks
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	Associated Loci 0	AT4G12750								
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	Mutation Site	gene								
	Description 0	escription PCR was performed on Arabidopsis thaliana lines each of which contains one or more TDNA insertion elements. The resultant fragment for each line was directly sequenced to determine the genomic sequence at the site of insertion. Details of the protocols used can be found at http://signal.salk.edu/tdna protocols.html								
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Go	SALK_101462_CT954850_FN262742_GK-168G10-013368_FLAG_55380 Name:SALK_052476.31.35.X
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List Genes In Range	Sequence ruler - click here to open 10 kb sequence window
See this region in	Click to view SegViewer

Nucleotide view



### **PubMed - Endless Resources**

### http://www.ncbi.nlm.nih.gov/pubmed

SNCBI Resources 🖂	How To 🖂		Sign in to NCBI
Pub Med.gov	PubMed 🔷		Search
US National Library of Medicine National Institutes of Health		Advanced	Help



### PubMed

PubMed comprises more than 24 million citations for biomedical literature from MEDLINE, life science journals, and online books. Citations may include links to full-text content from PubMed Central and publisher web sites.

# PubMed Commons

Featured comment - Aug 10 Recasting the net: R Pazoki suggests searching additional eQTL databases to ID other potential gene associations. <u>1.usa.gov/V0GK70</u>

Using PubMed	PubMed Tools	More Resources
PubMed Quick Start Guide	PubMed Mobile	MeSH Database
Full Text Articles	Single Citation Matcher	Journals in NCBI Databases
PubMed FAQs	Batch Citation Matcher	Clinical Trials
PubMed Tutorials	Clinical Queries	E-Utilities (API)
New and Noteworthy	Topic-Specific Queries	LinkOut

Ex. Author search: Goldberg RB, Harada JJ Keyword: Arabidopsis T-DNA Mutagenesis, etc... Note: If you are off campus, use VPN to access journal articles.

### Where Is Your Gene Expressed?

Task: When during seed development and it which part of the seed is your gene transcribed?

### Tools: Seed Gene Network (http://seedgenenetwork.net/arabidopsis)

**Procedure:** 

- 1. Select Analyze GeneChip Data
- 2. Select Browse
- 3. Enter your AGI Locus ID
- 4. Click Submit
- 5. Click on the Probe Set identifier

### **Results/Question:**

1. When during seed development and it which part of the seed is your gene transcribed?

### http://seedgenenetwork.net/arabidopsis



Click on the name of *Arabidopsis* seed tissues/compartment listed below to find the detail information about GeneChip experiments of *Arabidopsis* seed tissues/compartments. The information includes:

### **Analyze GeneChip Data**



### **Browse Arabidopsis mRNAs Profiling Database**



### **Results for Search by Gene ID**



		Probe Set	Project	Gene Model	Functional Category	Description
Click to view your gene's mRNA profile	1	254778_at	Arabidopsis ATH1 Array	AT4G12750	Transcription	DNA binding / sequence-specific DNA binding / transcription factor
during seed						

development

### **Gene Expression Profile – Part I**



- Click "Download GeneChip Data" on the top to get the signal intensities of all GeneChip experiments for this probe set.
- Also, you can view where the probe set aligns in the genome and 454 ESTs that map to corresponding gene using "Genome Browser".



### **Gene Expression Profile – Part II**



#### GeneChip Experiments (Organized by Stage and Tissue/Compartment)

Abbreviation: A, absent; INS, inconsistent detection call between biological replicas; M, marginal; P, present

Abbreviation of Stage and Tissue/Compartment:

Stage: PGLOB - Pre-Globular Stage; GLOB - Globular Stage; HRT - Heart Stage; LCOT - Linear Cotyledon Stage; MG - Maturation Green Stage

Tissue: CZE - Chalazal Endosperm; CZSC - Chalazal Seed Coat; EP - Embryo Proper; GSC - General Seed Coat; MCE - Micropylar Endosperm; PEN - Peripheral Endosperm; S - Suspensor; WS - Whole Seed Signal intensities (relative mRNA prevalences) and signal detection calls (P, A, or M) were generated using MAS 5.0 algorithm. For comparative purposes, GeneChip data were scaled globally to a target intensity of 500 for all probe sets on the chip using MAS 5.0 default parameters. Each probe set was manually assigned a consensus detection calls of P, A, or M, respectively. By contrast, probe sets with different, or discordant, detection calls for the two biological replicates (e.g., P and A; P and M) were assigned a consensus detection call of Insufficient (INS). Consensus detection calls are used to shown in the plot.