



"

Biological evolution is the central organizing principle of modern biology...modern chimpanzees, other great apes, and humans are descended from a common ancestor that is now extinct.

"

-U.S. National Academy of Sciences, Science, Evolution, and Creationism (2008)

The history of evolutionary thought with regards to common ancestry dates back to 1698...





...when the English anatomist Edward Tyson acquired the remains of a chimpanzee, and published his astonishment at the anatomical similarity between the chimpanzee and a human.

Pierre-Louis Moreau de Maupertuis, 1745



Maupertuis' *Vénus Physique* is an early attempt at a materialistic explanation of the origin of species, suggesting that natural variation and selection give rise to function.

Immanuel Kant, 1790



Wrote that the "analogy of animal forms" implies a common original type.

Charles Darwin published On the Origin of Species in 1859

Sixty years after his grandfather posed the question of common ancestry.

"

Therefore I should infer from analogy that probably all the organic beings which have ever lived on this earth have descended from some one primordial form, into which life was first breathed.

"

-Darwin, On the Origin of Species, 1859

Since then, biologists have discovered that...

...all life forms share the same fundamental biochemical organization:



It is such similarities that have prompted bioinformaticians to meticulously compare sequences, pseudogenes, and repeated sequences in the genomes of humans and chimpanzees.

Even more recently, the field of bioinformatics has begun to contribute to the search for answers...

bio-in-for-mat-ics
noun pl but singular in constr \bī-ō-in-fər-'ma-tiks\
the collection, classification, storage, and analysis of biochemical and biological information using computers especially as
applied to molecular genetics and genomics

- bio-in-for-mat-ic </

http://www.merriam-webster.com/dictionary/bioinformatics

• In 2002, Ebersberger et al. aligned 1.9 million nucleotides of chimpanzee DNA with the human genome, and found a difference of a mere 1.24%.

• In 2003, Lie et al. compared 4.97 million nucleotides of human DNA from chromosome 7 to the corresponding chimpanzee DNA, and found that the sequences differed by 1.13%.

• The publication of a rough draft of the chimpanzee genome, also in 2003, provided even greater evidence for the similarity between chimpanzees and humans.

The entire sequence differs from the human genome by 86-89% (Tomkins, 2011); this statistic is a matter of live debate, and fluctuates regularly in the literature.

Using bioinformatics, we can examine the arguments for/against common ancestry for ourselves.

*This is an informational talk, I will not draw conclusions about the strength of the data.

The Roadmap

Example: Chromosome 2/PAX3

Entrez/BLAST Tutorial

The Contribution of Bioinformatics

History of Evolutionary Thought

Example: Human Chromosome 2

• The structure of human chromosome 2 resembles the structure one might see if chimpanzee chromosomes 2a and 2b were fused together.

• Scientists accept that there is good evidence that chromosome 2 is composed of two fused chromosomes.

Regardless of the plausibility of a fusion event...

How similar are the genes on human chromosome 2 to those on chimpanzee chromosomes 2a and 2b?

PAX3 is a gene that belongs to the paired-box family of transcription factors, located on human chromosome 2, and chimpanzee chromosome 2b.

• Associated with ear, eye, and facial development.

Entrez BLAST



Entrez BLAST

blastn <u>blastp</u> blas	ttat tblastn tblastx
Enter Query S	BLASTN programs search nucleotide databases using a nucleotide query. more
Enter accession n	number(s), gi(s) FASTA sequence(s) 🕢 <u>Clear</u> Query subrange 🕢
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Or, upload file	Choose File) no file selected
Job Title	
	Enter a descriptive title for your BLAST search 😡
Choose Sean	ch Set
Database	Genome (reference only) 27005 sequences 😡
Exclude	Models (XM/XP) Uncultured/environmental sample sequences
Entrez Query	
Optional	Enter an Entrez query to limit search 😡
Program Sele	ction
Optimize for	Highly similar sequences (megablast)
	O More dissimilar sequences (discontiguous megablast)
	O Somewhat similar sequences (blastn)
	Choose a BLAST algorithm 🛞



Entrez BLAST

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A BLAST comparison of the protein sequences yields...

> ref Length=S	XP 0	01165390.1 UGM PREDICTED: paired box protein Pax-3 isoform	6 [Pan troglodytes]
GENE II	D: 49	59971 PAX3 paired box 3 [Pan troglodytes]	
Score = Identit	= 97 ties	77 bits (2526), Expect = 0.0, Method: Compositional matrix ad = $474/474$ (100%), Positives = $474/474$ (100%), Gaps = $0/474$ (0	just. 8)
Query 1	1	MTTLAGAVPRMMRPGPGQNYPRSGFPLEVSTPLGQGRVNQLGGVFINGRPLPNHIRHKIV MTTLAGAVPRMMRPGPGONYPRSGFPLEVSTPLGOGRVNQLGGVFINGRPLPNHIRHKIV	60
Sbjct 1	1	MTTLAGAVPRMMRPGPGQNYPRSGFPLEVSTPLGQGRVNQLGGVFINGRPLPNHIRHKIV	60
Query 6	61	EMAHHGIRPCVISRQLRVSHGCVSKILCRYQETGSIRPGAIGGSKPKQVTTPDVEKKIEE	120
Sbjet 6	61	EMAHHGIRPCVISRQLRVSHGCVSKILCRYQETGSIRPGAIGGSKPKQVTTPDVEKKIEE	120
Query 1	121	YKRENPGMFSWEIRDKLLKDAVCDRNTVPSVSSISRILRSKFGKGEEEEADLERKEAEES	180
Sbjct 1	121	YKRENPGMFSWEIRDKLLKDAVCDRNIVPSVSSISRILRSKFGKGEEEEADLERKEAEES	180
Query 1	181	EKKAKHSIDGILSERASAPQSDEGSDIDSEPDLPLKRKQRRSRTTFTAEQLEELERAFER	240
Sbjct 1	181	EKKAKHSIDGILSERASAPQSDEGSDIDSEPDLPLKRKQRRSRIFFIAEQLEELERAFER	240
Query 2	241	THYPDIYTREELAQRAKLTEARVQVWFSNRRARWRKQAGANQLMAFNHLIPGGFPPTAMP	300
Sbjct 2	241	THYPDIYIRELLAQRAKLIEARVQVWFSNRRARWRKQAGANQLMAFNHLIPGGFPPIAMP	300
Query 3	301	TLPTYQLSETSYQPTSIPQAVSDPSSTVHRPQPLPPSTVHQSTIPSNPDSSSAYCLPSTR	360
Sbjet 3	301	TLPTYQLSETSYQPTSIPQAVSDPSSTVHRPQPLPPSTVHQSTIPSNPDSSSATCLPSTR	360
Query 3	361	HGFSSYTDSFVPPSGPSNPMNPTIGNGLSPQVMGLLTNHGGVPHQPQTDYALSPLTGGLE	420
Sbjct 3	361	HGFSSYIDSFVPPSGPSNPMNPTIGNGLSPQVMGLLINHGGVPHQPQIDYALSPLIGGLE	420
Query 4	421	PTTTVSASCSQRLDHMKSLDSLPTSQSYCPPTYSTTGYSMDPVTGYQYGQYGQS 474	
Sbjct 4	421	PTTTVSASCSQRLDHMKSLDSLPTSQSYCPPTYSTTGYSMDPVTGYQYGQYGQS PTTTVSASCSQRLDHMKSLDSLPTSQSYCPPTYSTTGYSMDPVTGYQYGQYGQS 474	

The query and the subject sequences are identical.



UCSC's Genome Browser

A tutorial

This site contains the reference sequence and working draft assemblies for a large collection of genomes.



UCSC's Genome Browser



Example: GULOP (pseudogene)

Darwinian scientists claim that much non-coding DNA is functionless "genetic flotsam and jetsam" (i.e., junk) that has accumulated many random mutations.

Pseudogenes, genes that have lost their protein-coding ability, are often called "junk DNA".

"At face value, pseudogenes hardly seem like genomic features that would be designed by a wise engineer....given the relative ease of origin of pseudogenes, their persistence in genomes is understandable in the light of evolutionary reasoning."

-John C. Advise

(From John C. Avise, Inside the Human Genome: A Case for Non-Intelligent Design [Oxford University Press, 2010, pp. 115-6])

Using Genome Browser:

A gene that presumably encoded the L-gulono-y-lactone oxidase enzyme in primates (GULO) exists as a pseudogene (GULOP) in both humans and chimpanzees...

UCSC's Genome Browser





Human position: <u>chr8:13909-146302299</u> size: 146288391 Strand: + Chimp position: <u>chr8:3911-143967758</u> size: 143963848 Chain ID: 6 Score: 11541415440 Approximate Score within browser window: 2569589

A BLAST comparison of the two sequences reveals a high sequence similarity, though the two are not identical:

Query	1189	AACAgtt-ttttgtttgtttgtttgtttgtttgttttgtt	1247
Sbjct	118378	AACA-TTCTTTTTGTTT-TGT-TTTGTTTTGTTTT-TTTTTTTT	118323
Query	1248	GTCTCGCTCTGTCACCCAAGCTGGAGTGCAGTGGCGCGGGTCTCAGCTCACTGCCAGCTCC	1307
Sbjct	118322	GTCTCGCTCTGTCGCCCAGGCTGGAGTGCAGTGGCGCGCGATCTCGGCTCACTGCAAGCTCC	118263
Query	1308	ACCTCCCAGGTTCACACCATTCTCCTCCAGCCTCCCGAGTAGCTGGGACTACAGGTG	1367
Sbjct	118262	GCCTCCCGGGTTCACGCCATTCTCCTGCCTCAGCCTCCCGAGTAGCTGGGACTACAGGCG	118203
Query	1368	CCCGCCACCACCCCAGCTAATTTTTTGTATTTTTAGTAGAGATGGGGTTTCACCGTGTT	1427
Sbjct	118202	CCCGCTACCACGCCCGGCTAATTTTTTGTATTTTTAGTAGAGATGGGGTTTCACCGTGTT	118143
Query	1428	AGCCAGGATGGTCTCGATCTCCTGACCTCGTGATCTGCCTGGCCTCC-AAAGTGC	1486
Sbjct	118142	AGCCAGGATGGTCTCGATCTCCTGACCTCGTGATCCGCCCGC	118083
Query	1487	TGGGATTACAGGTGTGAGCCACTGTGCCCGGCACTC 1522	
Sbjct	118082	TGGGATTACAGGCGTGAGCCACCGCGCCCGGC-CTC 118048	



Alu sequences are mobile genetic elements that appear to insert into the genome at random...

The vast majority of them are found in identical locations in the human and chimp genomes, which strongly suggests they were inserted in the common ancestor of humans and chimpanzees before they diverged.

Using Genome Browser:

AluSq2 is an SINE located in the SPTLC2 gene in humans...







In the new age of technology, bioinformatics has become the battleground of choice for evolutionary biologists...

The field is a collaborative effort among scientists all over the globe, and provides a wealth of information about comparative genomics:

- Blogs
- Articles
- Papers



Yet the plethora of genetic information about common ancestry already available is only just the beginning...



A List of Useful Databases

- NCBI
 - PubMed
 - Entrez
 - BLAST
- Ensembl (European)
- UCSC's Genome Browser

"

Genetic mechanism in all its happenstance has produced the genetic *basis* of humanness. *Genetics describes the process, ordained and upheld by God,* to make the creature that expresses God's 'image and likeness'...

"

-Graeme Finlay (emphasis added)