Human genomics: vestiges of Eden or skeletons in the closet?

Dennis R. Venema

Trinity Western University

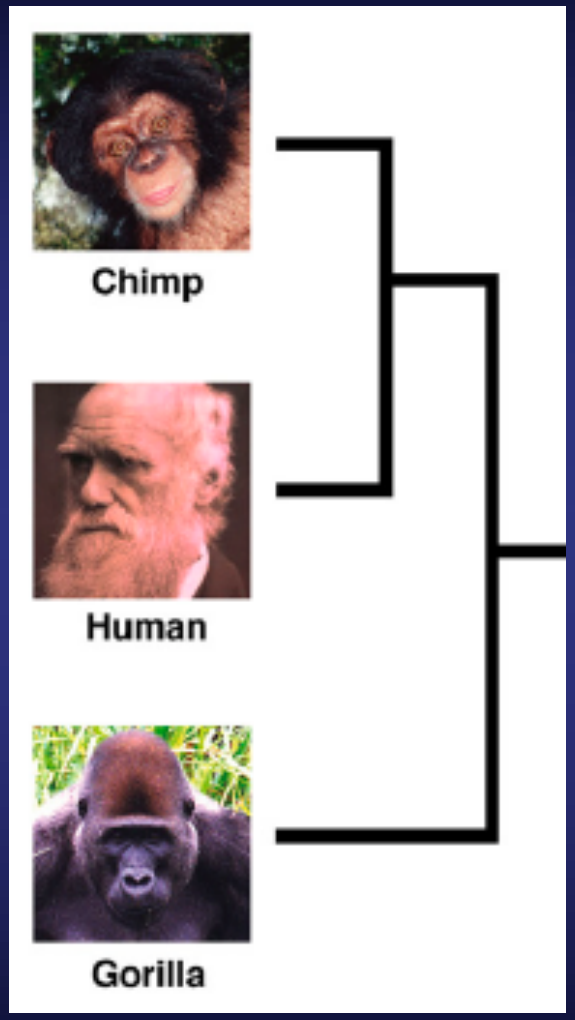
ASA Meeting
August 2, 2009
Baylor University

Talk outline:

Evidence for human : chimp common ancestry

Evidence for ancestral hominid population size

Thoughts on Biblical scientific concordism



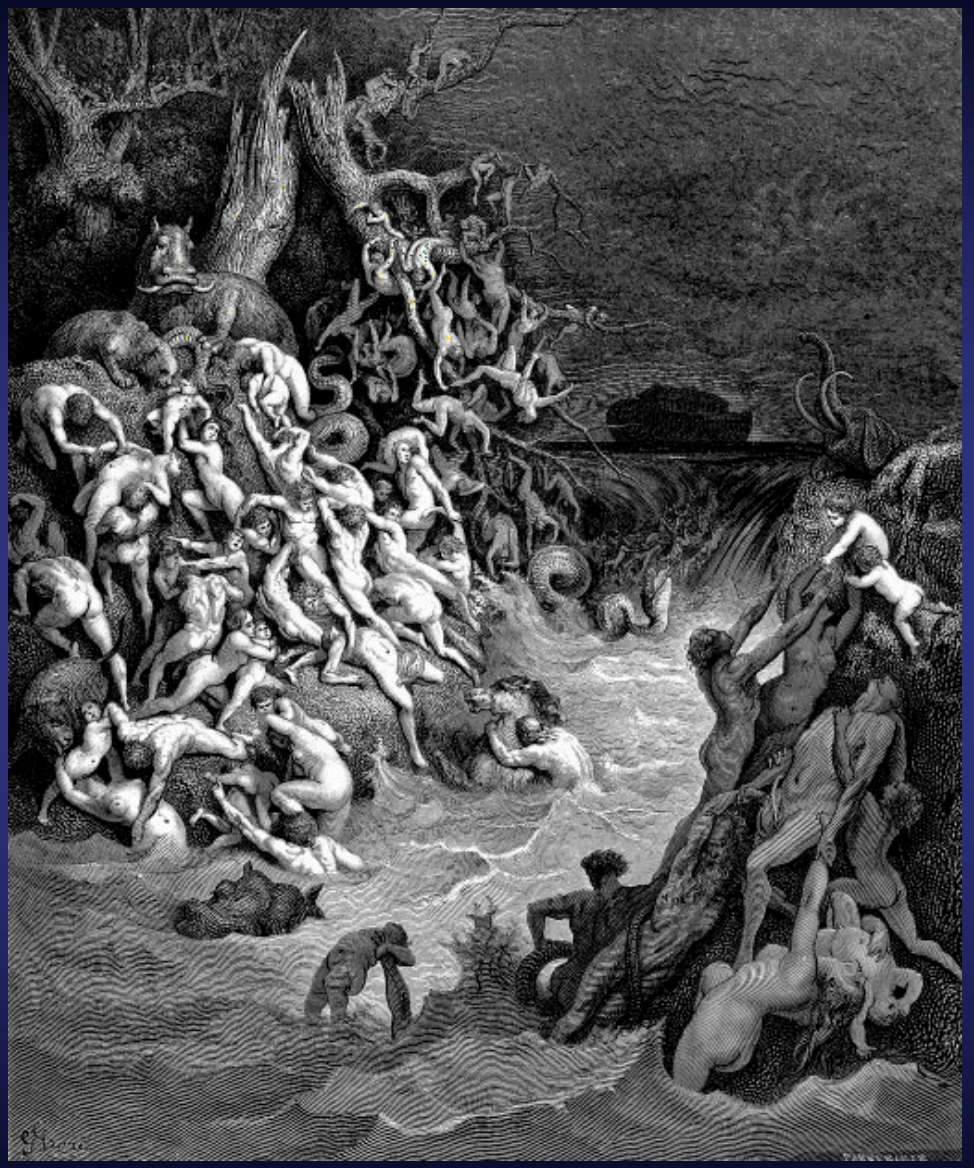
BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

Talk outline:

Evidence for human : chimp common ancestry

Evidence for ancestral hominid population size

Thoughts on Biblical scientific concordism



Gustave Dore: *The Deluge*
(Dore Illustrated Bible, 1865)

Talk outline:

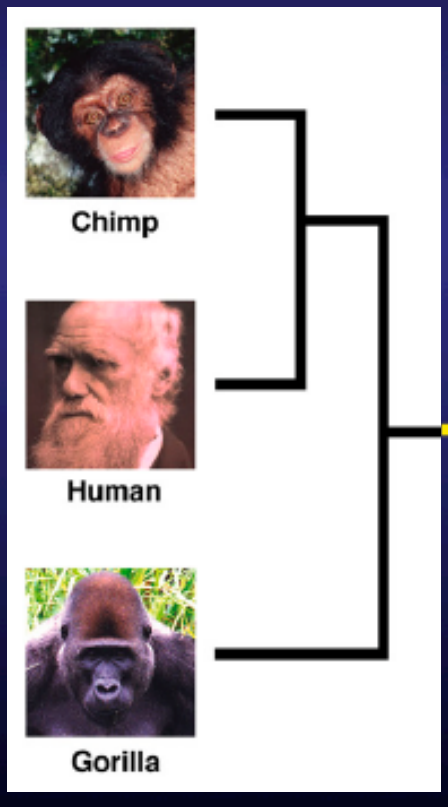
Evidence for human : chimp common ancestry

Evidence for ancestral hominid population size

Thoughts on Biblical scientific concordism

Titian: *The Fall of Man* (c. 1570)

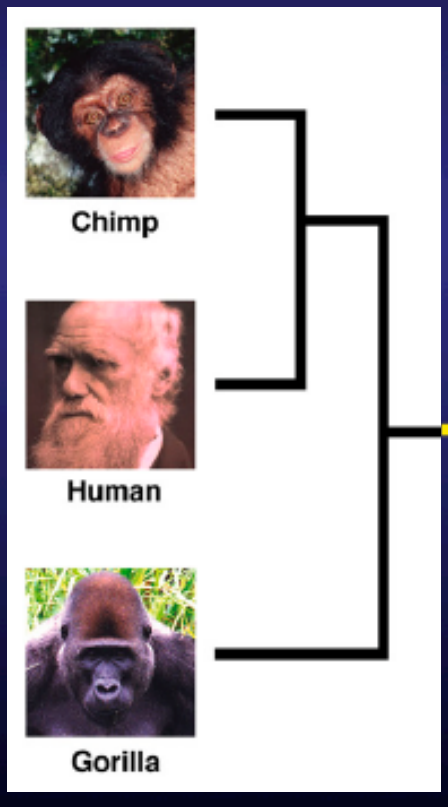
Evidence for human :
chimp common ancestry



BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

- Homology
- Redundancy
- Synteny
- Pseudogeny
- Parsimony

Evidence for human : chimp common ancestry



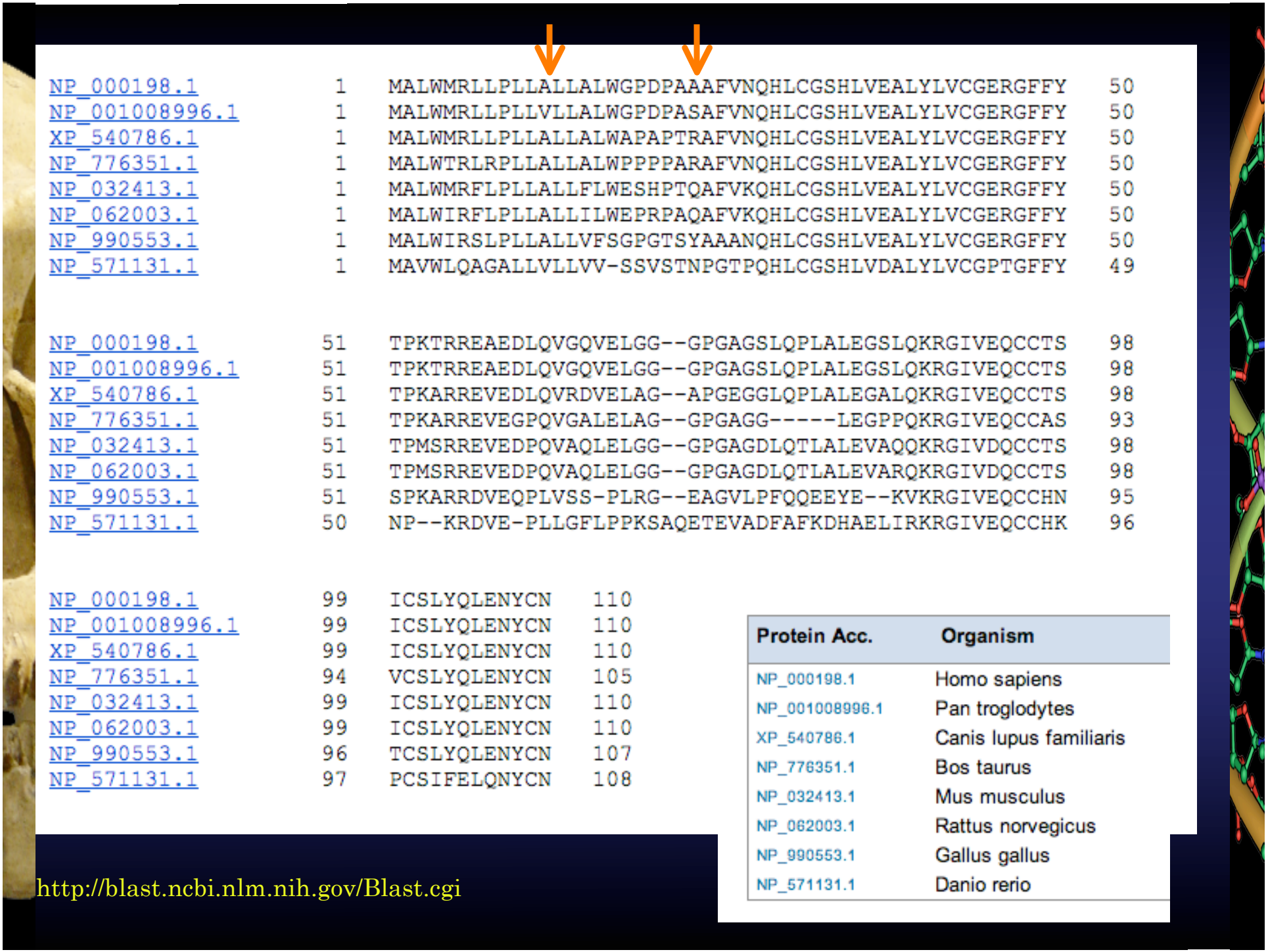
BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

- Homology
- Redundancy
- Synteny
- Pseudogeny
- Parsimony

```
NP_000198.1           1   MALWMRLLPLLALLALWGPDPAAAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_001008996.1        1   MALWMRLLPLLVLLALWGPDPASAFVNQHLCGSHLVEALYLVCGERGFFY   50
XP_540786.1           1   MALWMRLLPLLALLALWAPAPTRAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_776351.1           1   MALWTRLRPLLALLALWPPPPARAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_032413.1           1   MALWMRFLPLLALLFLWESHPTQAFVKQHLCGSHLVEALYLVCGERGFFY   50
NP_062003.1           1   MALWIRFLPLLALLILWEPRPAQAFVKQHLCGSHLVEALYLVCGERGFFY   50
NP_990553.1           1   MALWIRSLPLLALLVFSGPGTSYAAANQHLCGSHLVEALYLVCGERGFFY   50
NP_571131.1           1   MAVWLQAGALLVLLVV-SSVSTNPGTPQHLCGSHLVDALYLVCGPTGFFY   49

NP_000198.1          51   TPKTRREAEDLQVGQVELGG--GPGAGSLQPLALEGSLQKRGIVEQCCTS   98
NP_001008996.1       51   TPKTRREAEDLQVGQVELGG--GPGAGSLQPLALEGSLQKRGIVEQCCTS   98
XP_540786.1          51   TPKARREVEDLQVRDVELAG--APGEGGLQPLALEGALQKRGIVEQCCTS   98
NP_776351.1          51   TPKARREVEGPQVGALELAG--GPGAGG-----LEGPPQKRGIVEQCCAS   93
NP_032413.1          51   TPMSRREVEDPQVAQLELGG--GPGAGDLQTLALEVAQQKRGIVDQCCTS   98
NP_062003.1          51   TPMSRREVEDPQVAQLELGG--GPGAGDLQTLALEVARQKRGIVDQCCTS   98
NP_990553.1          51   SPKARRDVEQPLVSS-PLRG--EAGVLPFQQEEYE--KVKRGIVEQCCHN   95
NP_571131.1          50   NP--KRDVE-PLLGFLPPKSAQETEVADFAFKDHAELIRKRGIVEQCCHK   96

NP_000198.1          99   ICSLYQLENYCN   110
NP_001008996.1       99   ICSLYQLENYCN   110
XP_540786.1          99   ICSLYQLENYCN   110
NP_776351.1          94   VCSLYQLENYCN   105
NP_032413.1          99   ICSLYQLENYCN   110
NP_062003.1          99   ICSLYQLENYCN   110
NP_990553.1          96   TCSLYQLENYCN   107
NP_571131.1          97   PCSIFELQNYCN   108
```

Protein Acc.	Organism
NP_000198.1	Homo sapiens
NP_001008996.1	Pan troglodytes
XP_540786.1	Canis lupus familiaris
NP_776351.1	Bos taurus
NP_032413.1	Mus musculus
NP_062003.1	Rattus norvegicus
NP_990553.1	Gallus gallus
NP_571131.1	Danio rerio

http://blast.ncbi.nlm.nih.gov/Blast.cgi



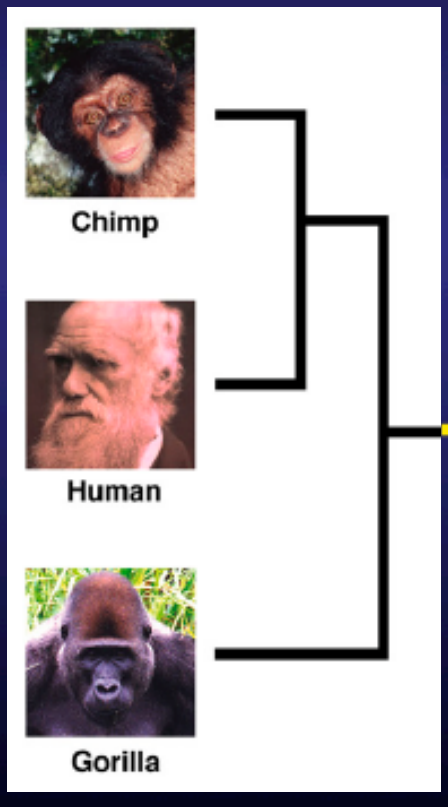
```
NP_000198.1            1   MALWMRLLPLLALLALWGPDPAAAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_001008996.1         1   MALWMRLLPLLVLLALWGPDPASAFVNQHLCGSHLVEALYLVCGERGFFY   50
XP_540786.1            1   MALWMRLLPLLALLALWAPAPTRAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_776351.1            1   MALWTRLRPLLALLALWPPPPARAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_032413.1            1   MALWMRFLPLLALLFLWESHPTQAFVKQHLCGSHLVEALYLVCGERGFFY   50
NP_062003.1            1   MALWIRFLPLLALLILWEPRPAQAFVKQHLCGSHLVEALYLVCGERGFFY   50
NP_990553.1            1   MALWIRSLPLLALLVFSGPGTSYAAANQHLCGSHLVEALYLVCGERGFFY   50
NP_571131.1            1   MAVWLQAGALLVLLVV-SSVSTNPGTPQHLCGSHLVDALYLVCGPTGFFY   49

NP_000198.1           51   TPKTRREAEDLQVGQVELGG--GPGAGSLQPLALEGSLQKRGIVEQCCTS   98
NP_001008996.1        51   TPKTRREAEDLQVGQVELGG--GPGAGSLQPLALEGSLQKRGIVEQCCTS   98
XP_540786.1           51   TPKARREVEDLQVRDVELAG--APGEGGLQPLALEGALQKRGIVEQCCTS   98
NP_776351.1           51   TPKARREVEGPQVGALELAG--GPGAGG-----LEGPPQKRGIVEQCCAS   93
NP_032413.1           51   TPMSRREVEDPQVAQLELGG--GPGAGDLQTLALEVAQQKRGIVDQCCTS   98
NP_062003.1           51   TPMSRREVEDPQVAQLELGG--GPGAGDLQTLALEVARQKRGIVDQCCTS   98
NP_990553.1           51   SPKARRDVEQPLVSS-PLRG--EAGVLPFQQEEYE--KVKRGIVEQCCHN   95
NP_571131.1           50   NP--KRDVE-PLLGFLPPKSAQETEVADFAFKDHAELIRKRGIVEQCCHK   96

NP_000198.1           99   ICSLYQLENYCN   110
NP_001008996.1        99   ICSLYQLENYCN   110
XP_540786.1           99   ICSLYQLENYCN   110
NP_776351.1           94   VCSLYQLENYCN   105
NP_032413.1           99   ICSLYQLENYCN   110
NP_062003.1           99   ICSLYQLENYCN   110
NP_990553.1           96   TCSLYQLENYCN   107
NP_571131.1           97   PCSIFELQNYCN   108
```

Protein Acc.	Organism
NP_000198.1	Homo sapiens
NP_001008996.1	Pan troglodytes
XP_540786.1	Canis lupus familiaris
NP_776351.1	Bos taurus
NP_032413.1	Mus musculus
NP_062003.1	Rattus norvegicus
NP_990553.1	Gallus gallus
NP_571131.1	Danio rerio

http://blast.ncbi.nlm.nih.gov/Blast.cgi

"Obviously, *function* constrains the sequence, giving merely the appearance of common descent when in fact this is an example of common design."

Evidence for human :
chimp common ancestry



BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

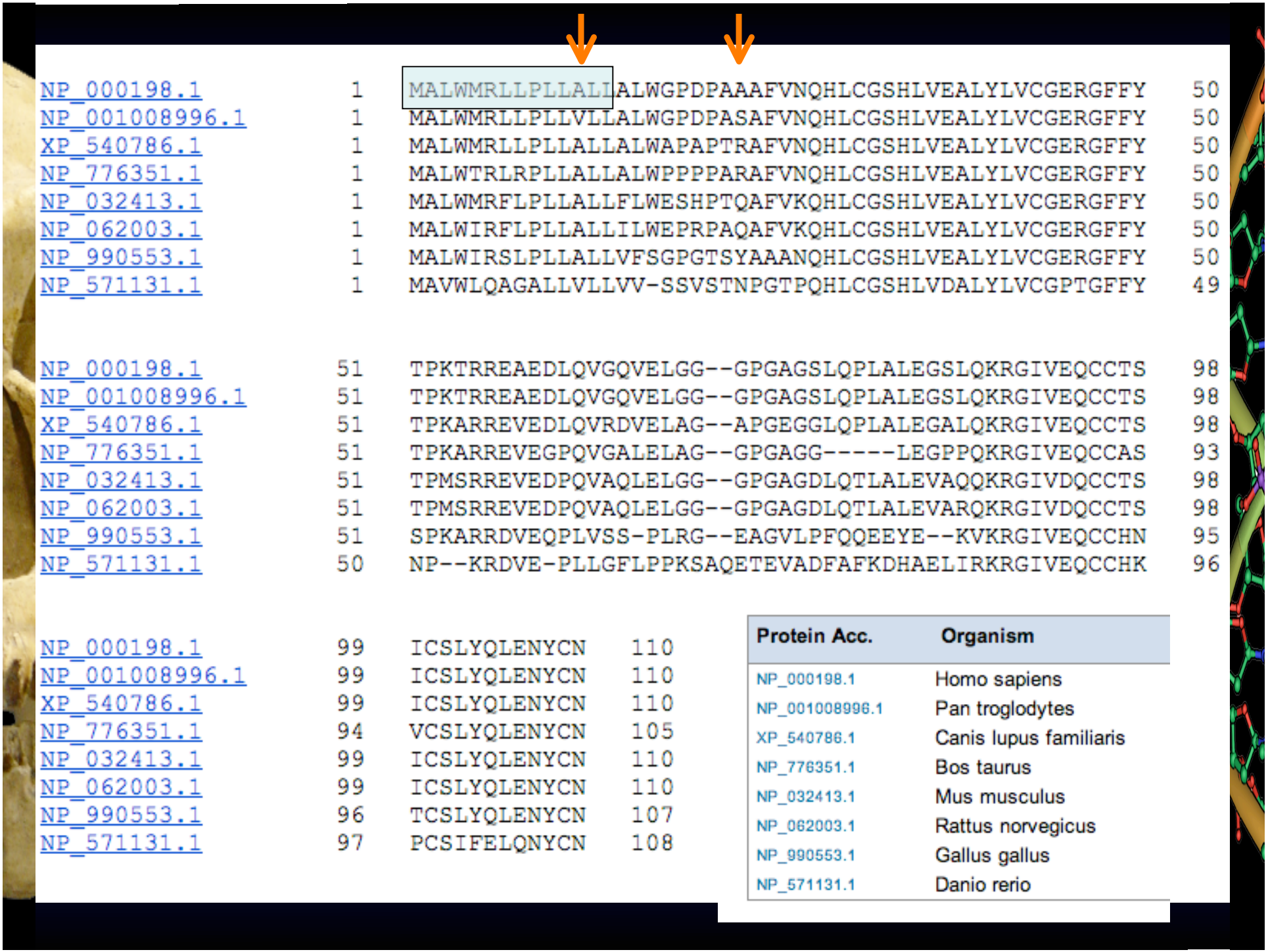
Homology

Redundancy

Synteny

Pseudogeny

Parsimony



```
NP_000198.1          1   MALWMRLLPLLALLALWGPDPAAAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_001008996.1       1   MALWMRLLPLLVLLALWGPDPASAFVNQHLCGSHLVEALYLVCGERGFFY   50
XP_540786.1          1   MALWMRLLPLLALLALWAPAPTRAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_776351.1          1   MALWTRLRPLLALLALWPPPPARAFVNQHLCGSHLVEALYLVCGERGFFY   50
NP_032413.1          1   MALWMRFLPLLALLFLWESHPTQAFVKQHLCGSHLVEALYLVCGERGFFY   50
NP_062003.1          1   MALWIRFLPLLALLILWEPRPAQAFVKQHLCGSHLVEALYLVCGERGFFY   50
NP_990553.1          1   MALWIRSLPLLALLVFSGPGTSYAAANQHLCGSHLVEALYLVCGERGFFY   50
NP_571131.1          1   MAVWLQAGALLVLLVV-SSVSTNPGTPQHLCGSHLVDALYLVCGPTGFFY   49

NP_000198.1         51   TPKTRREAEDLQVGQVELGG--GPGAGSLQPLALEGSLQKRGIVEQCCTS   98
NP_001008996.1      51   TPKTRREAEDLQVGQVELGG--GPGAGSLQPLALEGSLQKRGIVEQCCTS   98
XP_540786.1         51   TPKARREVEDLQVRDVELAG--APGEGGLQPLALEGALQKRGIVEQCCTS   98
NP_776351.1         51   TPKARREVEGPQVGALELAG--GPGAGG-----LEGPPQKRGIVEQCCAS   93
NP_032413.1         51   TPMSRREVEDPQVAQLELGG--GPGAGDLQTLALEVAQQKRGIVDQCCTS   98
NP_062003.1         51   TPMSRREVEDPQVAQLELGG--GPGAGDLQTLALEVARQKRGIVDQCCTS   98
NP_990553.1         51   SPKARRDVEQPLVSS-PLRG--EAGVLPFQQEEYE--KVKRGIVEQCCHN   95
NP_571131.1         50   NP--KRDVE-PLLGFLPPKSAQETEVADFAFKDHAELIRKRGIVEQCCHK   96

NP_000198.1         99   ICSLYQLENYCN   110
NP_001008996.1      99   ICSLYQLENYCN   110
XP_540786.1         99   ICSLYQLENYCN   110
NP_776351.1         94   VCSLYQLENYCN   105
NP_032413.1         99   ICSLYQLENYCN   110
NP_062003.1         99   ICSLYQLENYCN   110
NP_990553.1         96   TCSLYQLENYCN   107
NP_571131.1         97   PCSIFELQNYCN   108
```

Protein Acc.	Organism
NP_000198.1	Homo sapiens
NP_001008996.1	Pan troglodytes
XP_540786.1	Canis lupus familiaris
NP_776351.1	Bos taurus
NP_032413.1	Mus musculus
NP_062003.1	Rattus norvegicus
NP_990553.1	Gallus gallus
NP_571131.1	Danio rerio

Redundancy

```
HS atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg
   Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Ala Leu Leu

PT atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
   Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Val Leu Leu
```

Observed: 41/42 = 97.6% identity

Redundancy

```
HS atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg
   Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Ala Leu Leu

PT atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
   Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Val Leu Leu

DS atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
         a   a           a   a   a   a   a   a       a   a
         g   c           g   g   c   g   c   c       c   c
         t   t           t   t   t   t   t   t       t   t
           tta         aga tta tta     tta tta     tta tta
           ttg         agg ttg ttg     ttg ttg     ttg ttg
```

Observed: 41/42 = 97.6% identity

Function-constrained, max divergence (excluding position 12): 22/42 = 52.3% identity

Redundancy

```
HS  atg  gcc  ctg  tgg  atg  cgc  ctc  ctg  ccc  ctg  ctg  gcg  ctg  ctg
    Met  Ala  Leu  Trp  Met  Arg  Leu  Leu  Pro  Leu  Leu  Ala  Leu  Leu

PT  atg  gcc  ctg  tgg  atg  cgc  ctc  ctg  ccc  ctg  ctg  gtg  ctg  ctg
    Met  Ala  Leu  Trp  Met  Arg  Leu  Leu  Pro  Leu  Leu  Val  Leu  Leu

DS  atg  gcc  ctg  tgg  atg  cgc  ctc  ctg  ccc  ctg  ctg  gtg  ctg  ctg
          a    a              a    a    a    a    a    a         a    a
          g    c              g    g    c    g    c    c         c    c
          t    t              t    t    t    t    t    t         t    t
               tta            aga  tta  tta       tta  tta       tta  tta
               ttg            agg  ttg  ttg       ttg  ttg       ttg  ttg
```

Observed: 41/42 = 97.6% identity

Function-constrained, max divergence (excluding position 12): 22/42 = 52.3% identity

Redundancy

```
HS  atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg
    Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Ala Leu Leu

PT  atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
    Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Val Leu Leu

DS  atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
          a   a           a   a   a   a   a   a       a   a
          g   c           g   g   c   g   c   c       c   c
          t   t           t   t   t   t   t   t       t   t
            tta           aga tta tta     tta tta     tta tta
            ttg           agg ttg ttg     ttg ttg     ttg ttg
```

Observed: 41/42 = 97.6% identity

Function-constrained, max divergence (excluding position 12): 22/42 = 52.3% identity

Possible codon combinations (excl. position 12): $4^2 \times 6^8 = 26{,}873{,}856$

Redundancy

```
HS  atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg
    Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Ala Leu Leu

PT  atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
    Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Val Leu Leu

DS  atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg
          a   a         a   a   a   a   a   a       a   a
          g   c         g   g   c   g   c   c       c   c
          t   t         t   t   t   t   t   t       t   t
            tta         aga tta tta     tta tta     tta tta
            ttg         agg ttg ttg     ttg ttg     ttg ttg
```

Of the > 26 million possible combinations that preserve the sequence at the amino acid level, ***the observed pattern is the one most consistent with common descent.***

```
HS   atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg gcc ctc tgg gga cct gac
PT   atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg gcc ctc tgg gga cct gac
GG   atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg gcc ctc tgg gga cct gac
     Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Ala Leu Leu Ala Leu Trp Gly Pro Asp
      1   4   6   1   1   6   6   6   4   6   6  Val  6   6   4   6   1   4   4   2

HS   cca gcc gca gcc ttt gtg aac caa cac ctg tgc ggc tca cac ctg gtg gaa gct ctc tac
PT   cca gcc tcg gcc ttt gtg aac caa cac ctg tgc ggc tcc cac ctg gtg gaa gct ctc tac
GG   cca gcc gcg gcc ttt gtg aac caa cac ctg tgc ggc tcc cac ctg gtg gaa gct ctc tac
     Pro Ala Ala Ala Phe Val Asn Gln His Leu Cys Gly Ser His Leu Val Glu Ala Leu Tyr
      4   4  Ser  4   2   4   2   2   2   6   2   4   6   2   6   4   2   4   6   2
```

```
HS   atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg gcc ctc tgg gga cct gac
PT   atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gtg ctg ctg gcc ctc tgg gga cct gac
GG   atg gcc ctg tgg atg cgc ctc ctg ccc ctg ctg gcg ctg ctg gcc ctc tgg gga cct gac
     Met Ala Leu Trp Met Arg Leu Leu Pro Leu Leu Ala Leu Leu Ala Leu Trp Gly Pro Asp
      1   4   6   1   1   6   6   6   4   6   6  Val  6   6   4   6   1   4   4   2

HS   cca gcc gca gcc ttt gtg aac caa cac ctg tgc ggc tca cac ctg gtg gaa gct ctc tac
PT   cca gcc tcg gcc ttt gtg aac caa cac ctg tgc ggc tcc cac ctg gtg gaa gct ctc tac
GG   cca gcc gcg gcc ttt gtg aac caa cac ctg tgc ggc tcc cac ctg gtg gaa gct ctc tac
     Pro Ala Ala Ala Phe Val Asn Gln His Leu Cys Gly Ser His Leu Val Glu Ala Leu Tyr
      4   4  Ser  4   2   4   2   2   2   6   2   4   6   2   6   4   2   4   6   2
```

Possible codon combinations (excluding positions 12, 23 and 33):

$$1^4 \times 2^9 \times 4^{12} \times 6^{12} = 18.69 \times 10^{18}$$

Of the > 18 *quintillion possible combinations* that maintain the conserved sequence at the amino acid level, ***the observed pattern is the one most consistent with common descent.***

Note that this is only one tiny fragment in a genome with > 98% nucleotide identity with the chimpanzee genome over 2,700,000,000 DNA base pairs.

Reference	Sample Type	Sample Size	Percent Nucleotide Mismatches
Britten 2002	Random sample	0.78 Mb	1.2-1.69%
Ebersberger et al. 2002	Random sample	1.9 Mb	1.24%
Liu et al. 2003	Chromosome 7	4.97 Mb	1.13%
Wildman et al. 2003	Gene exons	90 kb	0.87%
International Chimpanzee Chromosome 22 Consortium 2004	Finished chromosomal sequence	33.3 Mb	1.44%
Nielsen et al. 2005	Gene exons	18.5 Mb	0.6%
Chimpanzee Sequencing and Analysis Consortium 2005	Rough draft genome sequence	2700 Mb	1.23%

Table 1. Summary of Human/Chimpanzee Genome Similarity Estimates.

Occas. Papers of the BSG No. 7, pp. 1-18
www.bryancore.org/bsg/

The Chimpanzee Genome and the Problem of Biological Similarity

TODD CHARLES WOOD[1]

Note that this is only one tiny fragment in a genome with > 98% nucleotide identity with the chimpanzee genome over 2,700,000,000 DNA base pairs.

Reference	Sample Type	Sample Size	Percent Nucleotide Mismatches
Britten 2002	Random sample	0.78 Mb	1.2-1.69%
Ebersberger et al. 2002	Random sample	1.9 Mb	1.24%
Liu et al. 2003	Chromosome 7	4.97 Mb	1.13%
Wildman et al. 2003	Gene exons	90 kb	0.87%
International Chimpanzee Chromosome 22 Consortium 2004	Finished chromosomal sequence	33.3 Mb	1.44%
Nielsen et al. 2005	Gene exons	18.5 Mb	0.6%
Chimpanzee Sequencing and Analysis Consortium 2005	Rough draft genome sequence	2700 Mb	1.23%

Table 1. Summary of Human/Chimpanzee Genome Similarity Estimates.

Occas. Papers of the BSG No. 7, pp. 1-18
www.bryancore.org/bsg/

The Chimpanzee Genome and the Problem of Biological Similarity

TODD CHARLES WOOD[1]

Note that this is only one tiny fragment in a genome with > 98% nucleotide identity with the chimpanzee genome over 2,700,000,000 DNA base pairs.

Reference	Sample Type	Sample Size	Percent Nucleotide Mismatches
Britten 2002	Random sample	0.78 Mb	1.2-1.69%
Ebersberger et al. 2002	Random sample	1.9 Mb	1.24%
Liu et al. 2003	Chromosome 7	4.97 Mb	1.13%
Wildman et al. 2003	Gene exons	90 kb	0.87%
International Chimpanzee Chromosome 22 Consortium 2004	Finished chromosomal sequence	33.3 Mb	1.44%
Nielsen et al. 2005	Gene exons	18.5 Mb	0.6%
Chimpanzee Sequencing and Analysis Consortium 2005	Rough draft genome sequence	2700 Mb	1.23%

Table 1. Summary of Human/Chimpanzee Genome Similarity Estimates.

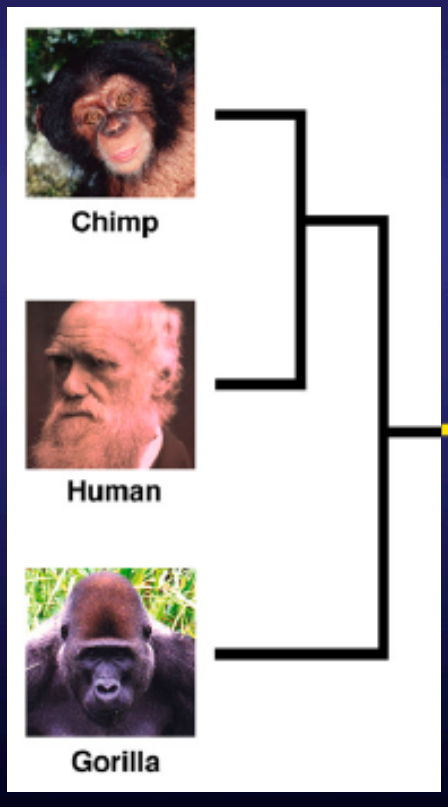
Samples taken from gene exons are even more identical

Occas. Papers of the BSG No. 7, pp. 1-18
www.ryancore.org/bsg/

The Chimpanzee Genome and the Problem of Biological Similarity

TODD CHARLES WOOD[1]

Evidence for human : chimp common ancestry



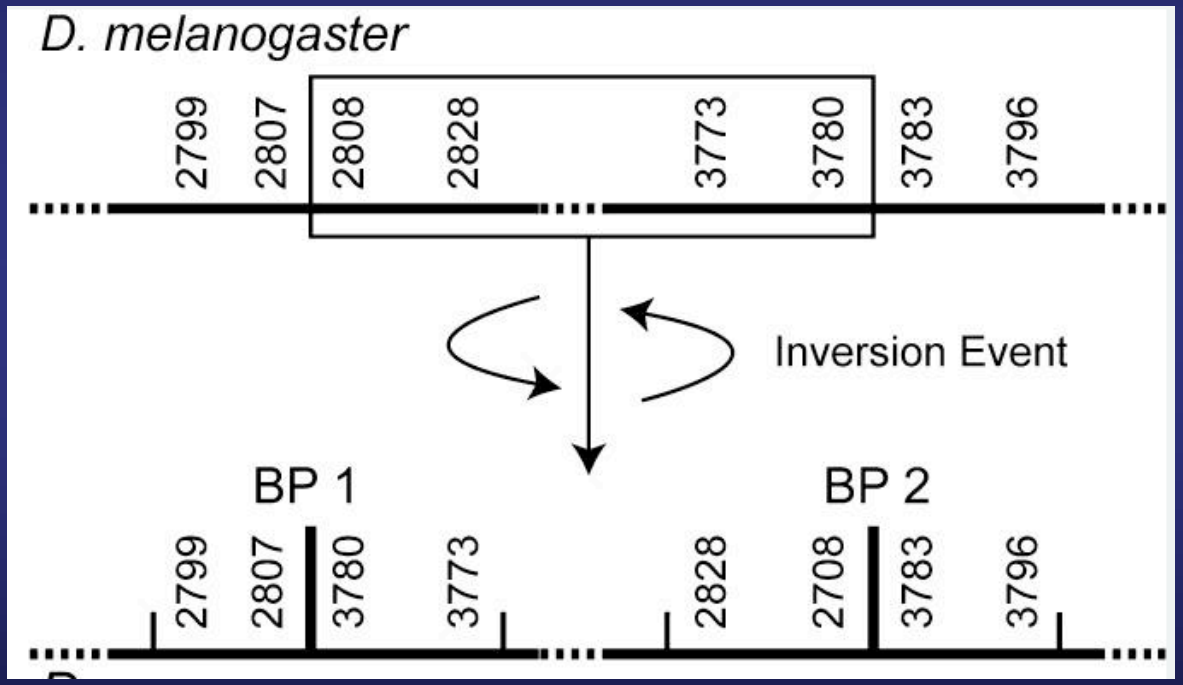
BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

- Homology
- Redundancy
- Synteny
- Pseudogeny
- Parsimony

Chromosomal mutations (within species):

Inversions
Translocations

The evidence for a chromosomal rearrangement is *synteny:* the retention of numerous genes in the spatial arrangement

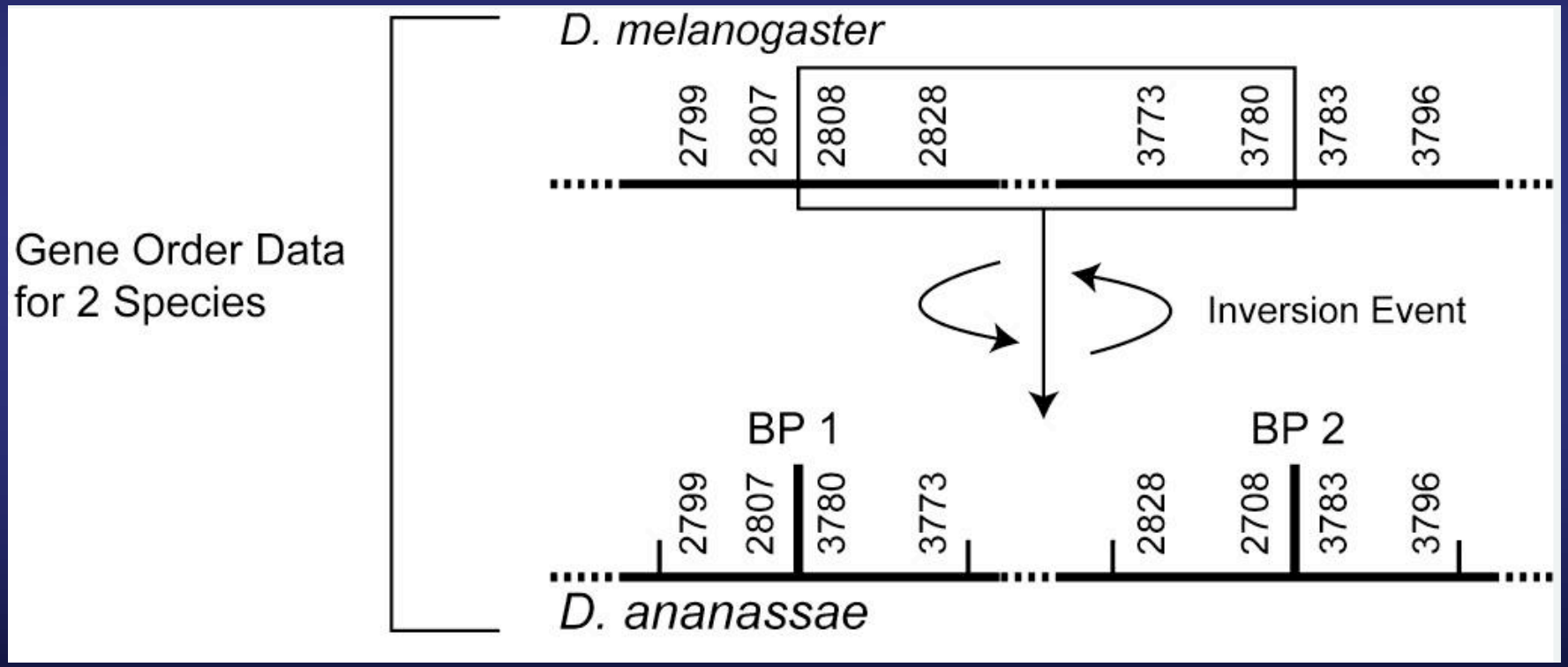


Bhutkar, A. et al. Genetics 2008;179:1657-1680

Chromosomal mutations (within species):

Inversions
Translocations

The evidence for a chromosomal rearrangement is *synteny:* the retention of numerous genes in the spatial arrangement

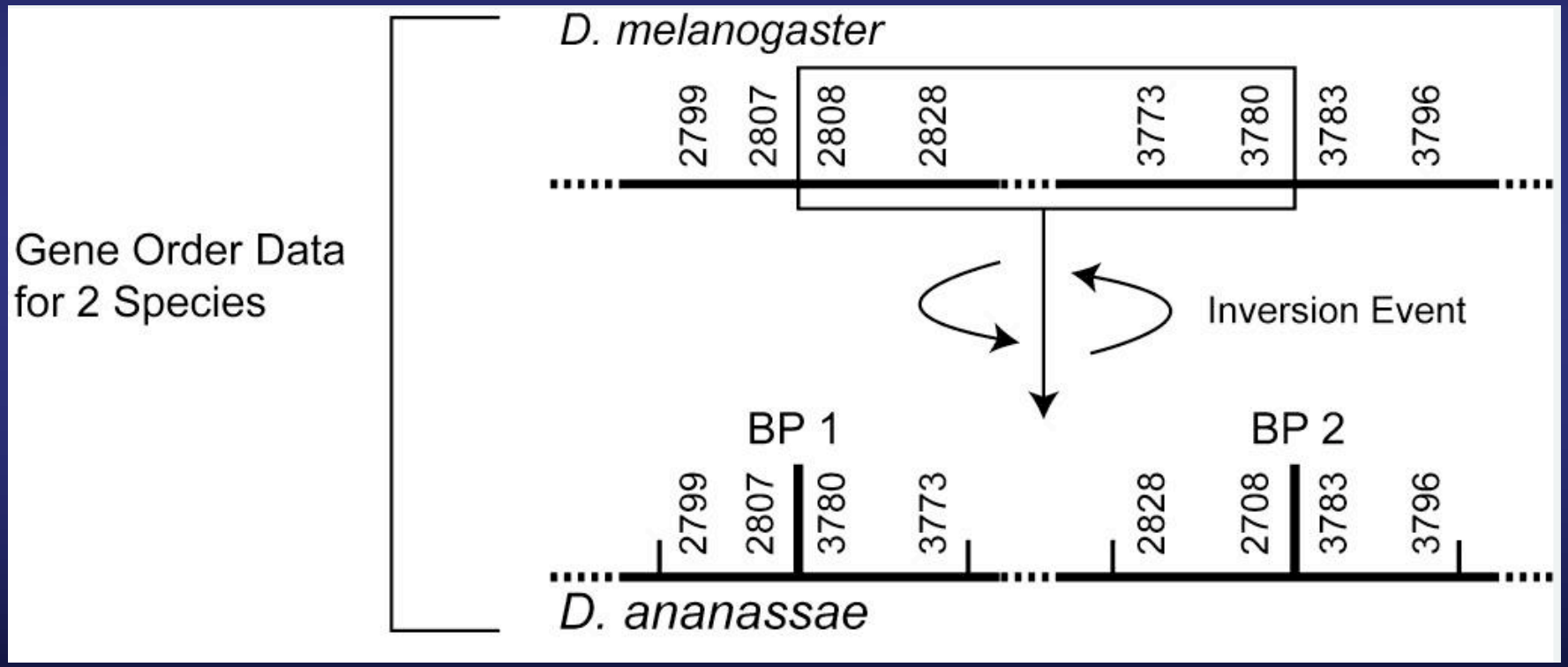


Bhutkar, A. et al. Genetics 2008;179:1657-1680

Chromosomal mutations (within species):

Inversions
Translocations

The evidence for a chromosomal rearrangement is *synteny:* the retention of numerous genes in the spatial arrangement



Bhutkar, A. et al. Genetics 2008;179:1657-1680

These events can also be traced between closely related species.

Drosophilid species as a model for chromosome change over time

Genetics. 2008 July; 179(3): 1657–1680.
doi: 10.1534/genetics.107.086108.
PMCID: PMC2475759

Chromosomal Rearrangement Inferred From Comparisons of 12 Drosophila Genomes

Arjun Bhutkar,[*†1] Stephen W. Schaeffer,[‡] Susan M. Russo,[*] Mu Xu,[‡] Temple F. Smith,[†] and William M. Gelbart[*]

[*]Department of Molecular and Cellular Biology, Harvard University, Cambridge, Massachusetts 02138, [†]Bioinformatics Program, Boston University, Boston, Massachusetts 02215 and [‡]Department of Biology and Institute of Molecular Evolutionary Genetics, Pennsylvania State University, University Park, Pennsylvania 16802-5301

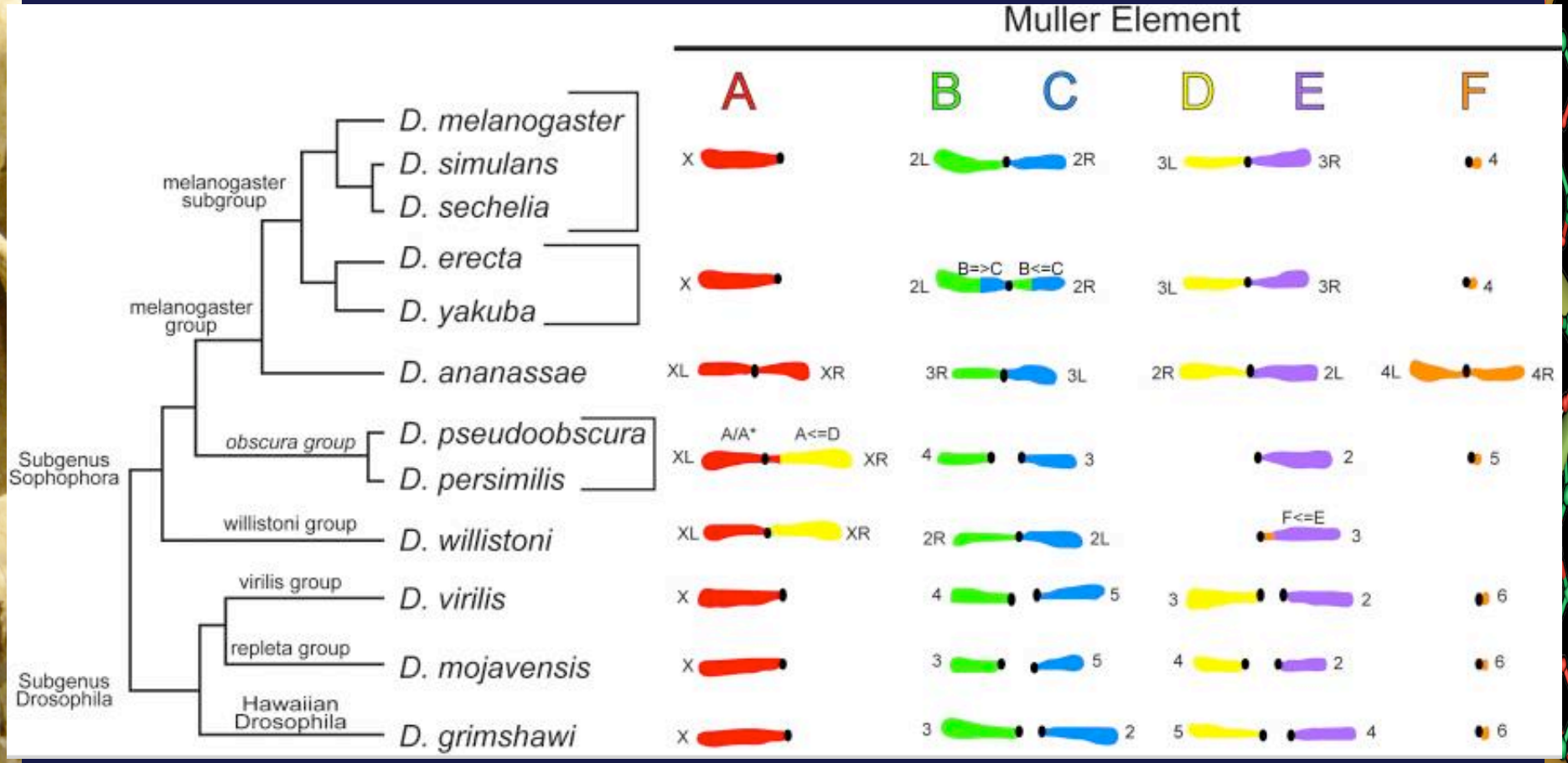
[1]*Corresponding author:* Gelbart Lab, Department of Molecular and Cellular Biology, Harvard University, 16 Divinity Ave., Room 4059, Cambridge, MA 02138. E-mail: arjunb@morgan.harvard.edu

Genetics. 2008 July; 179(3): 1601–1655.
doi: 10.1534/genetics.107.086074.
PMCID: PMC2475758

Polytene Chromosomal Maps of 11 Drosophila Species: The Order of Genomic Scaffolds Inferred From Genetic and Physical Maps

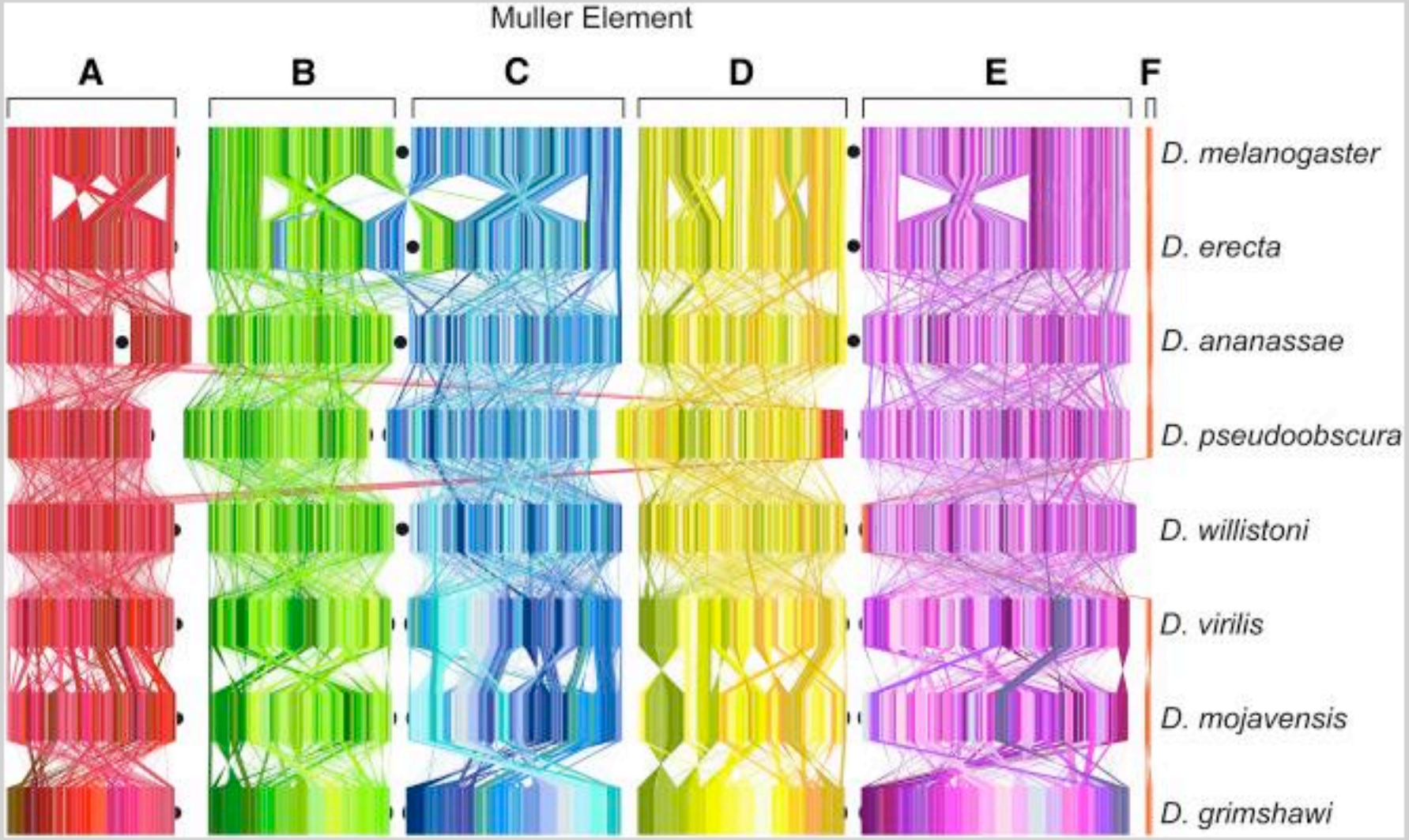
Stephen W. Schaeffer,[*1] Arjun Bhutkar,[†‡] Bryant F. McAllister,[§] Muneo Matsuda,[¶] Luciano M. Matzkin,[&] Patrick M. O'Grady,[**] Claudia Rohde,[††] Vera L. S. Valente,[‡‡] Montserrat Aguadé,[§§] Wyatt W. Anderson,[¶¶] Kevin Edwards,[**] Ana C. L. Garcia,[‡‡] Josh Goodman,[&&] James Hartigan,[***] Eiko Kataoka,[¶] Richard T. Lapoint,[†††] Elena R

Drosophilid phylogeny and chromosomal synteny



Schaeffer, S. W. et al. Genetics 2008;179:1601-1655

Synteny relative in 8 Drosophilid species

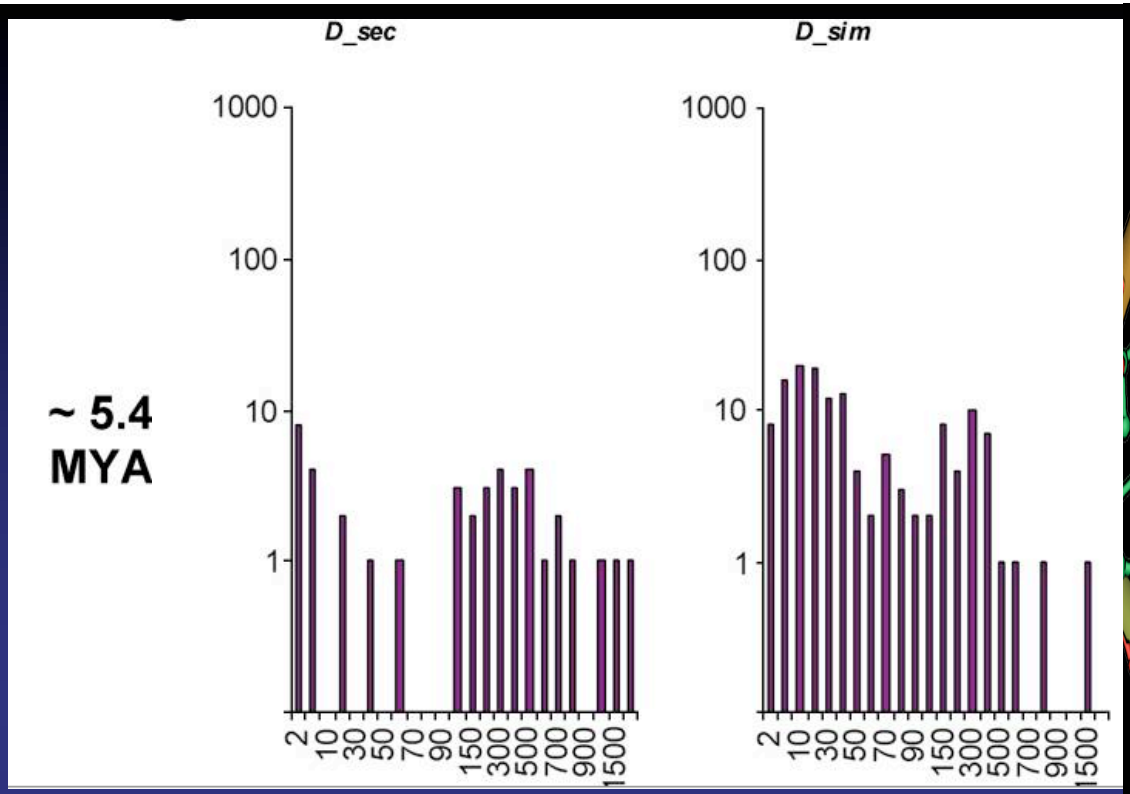


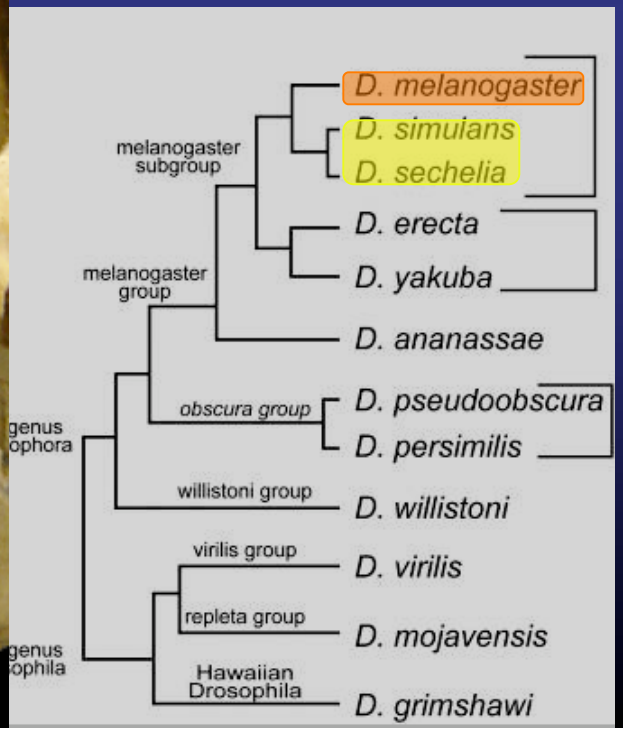
Schaeffer, S. W. et al. Genetics 2008;179:1601-1655

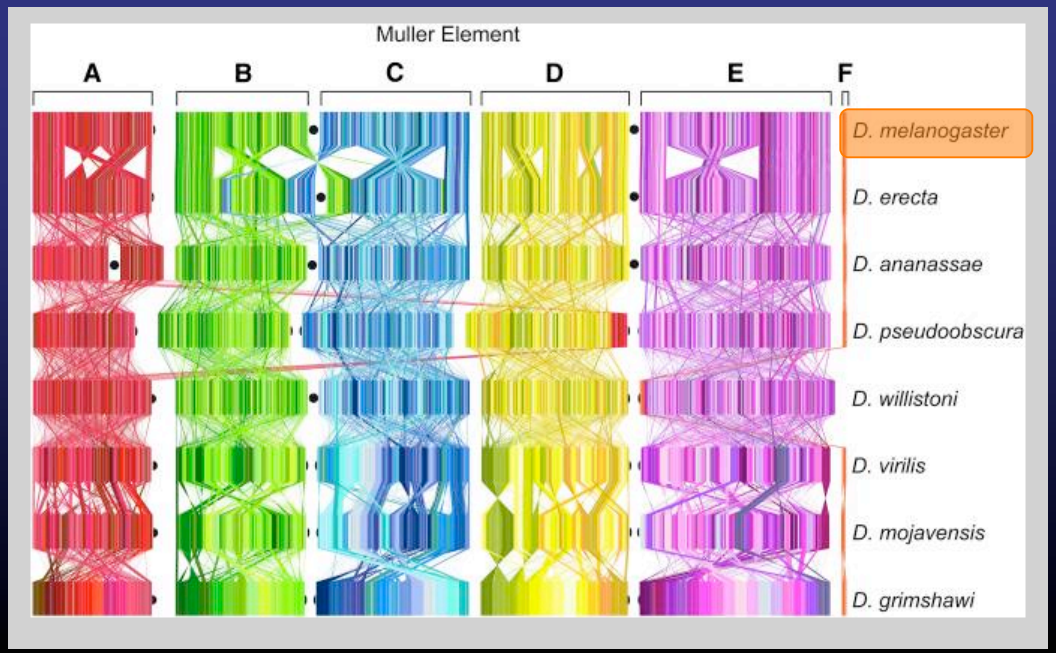
Synteny block size as a function of time since divergence

Bhutkar, A. et al. Genetics 2008; 179:1657-1680

Schaeffer, S. W. et al. Genetics 2008; 179:1601-1655



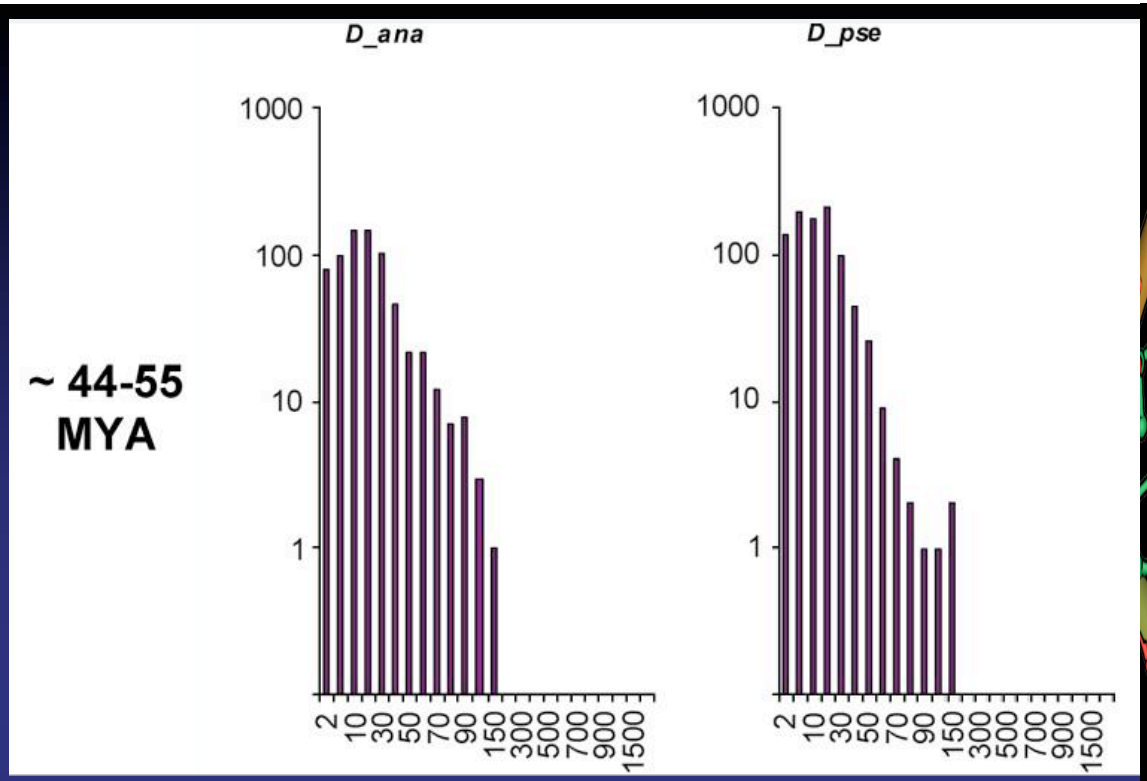


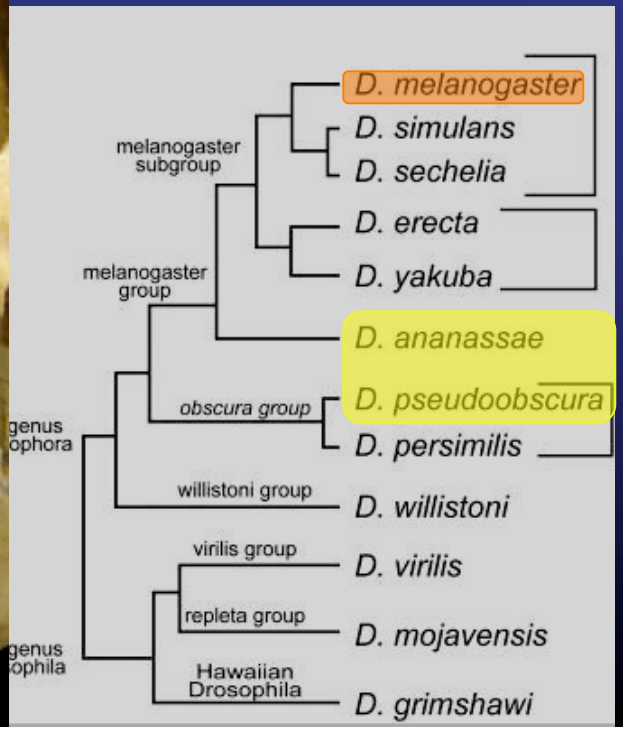


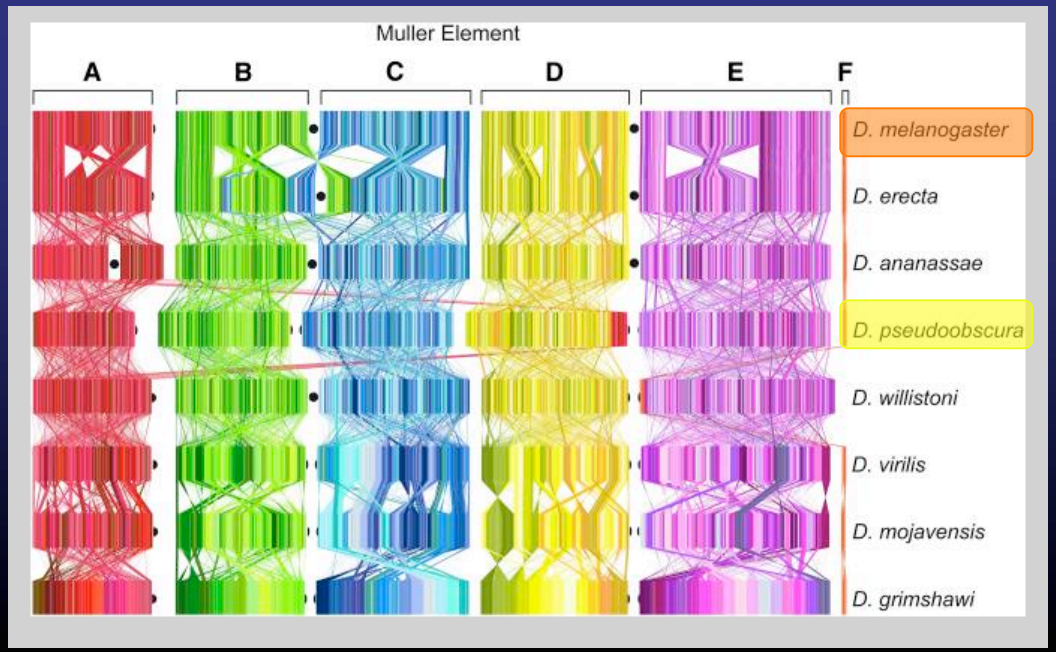
Synteny block size as a function of time since divergence

Bhutkar, A. et al. Genetics 2008; 179:1657-1680

Schaeffer, S. W. et al. Genetics 2008; 179:1601-1655

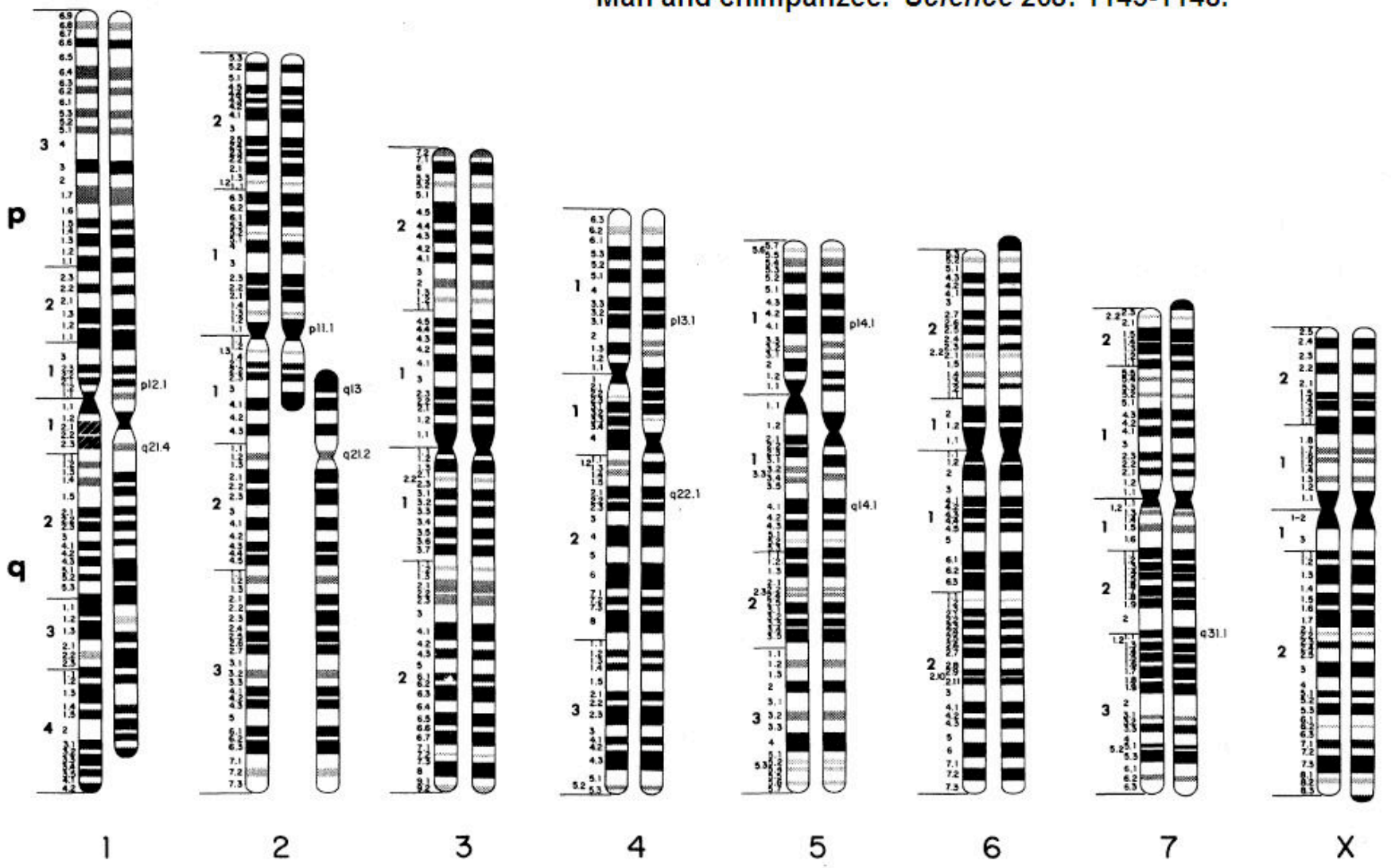




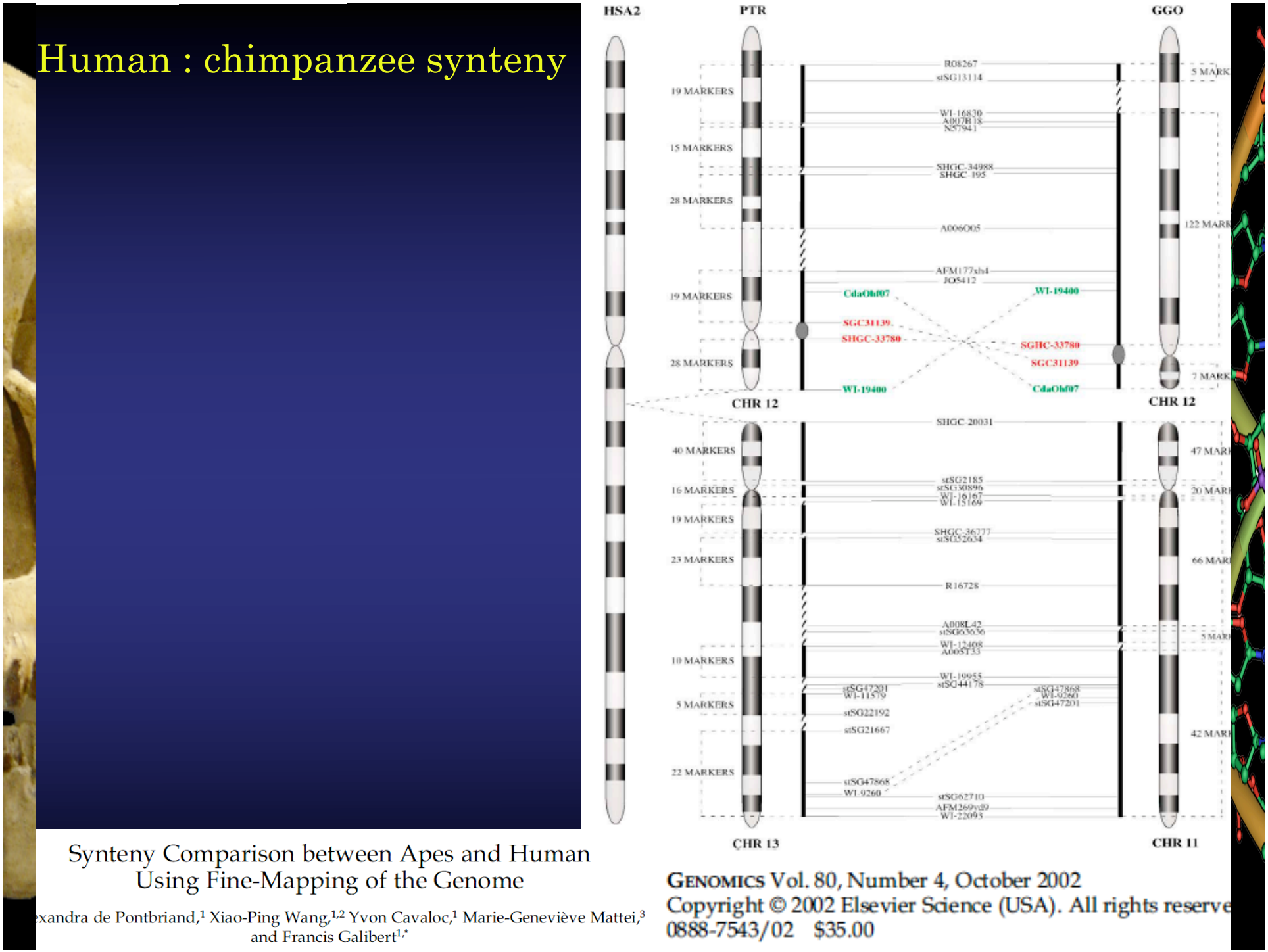


Human : chimpanzee synteny

Yunis, J.J., Sawyer, J.R., and Dunham, K. 1980. The striking resemblance of high-resolution G-banded chromosomes of Man and chimpanzee. *Science* 208: 1145-1148.



Human : chimpanzee synteny



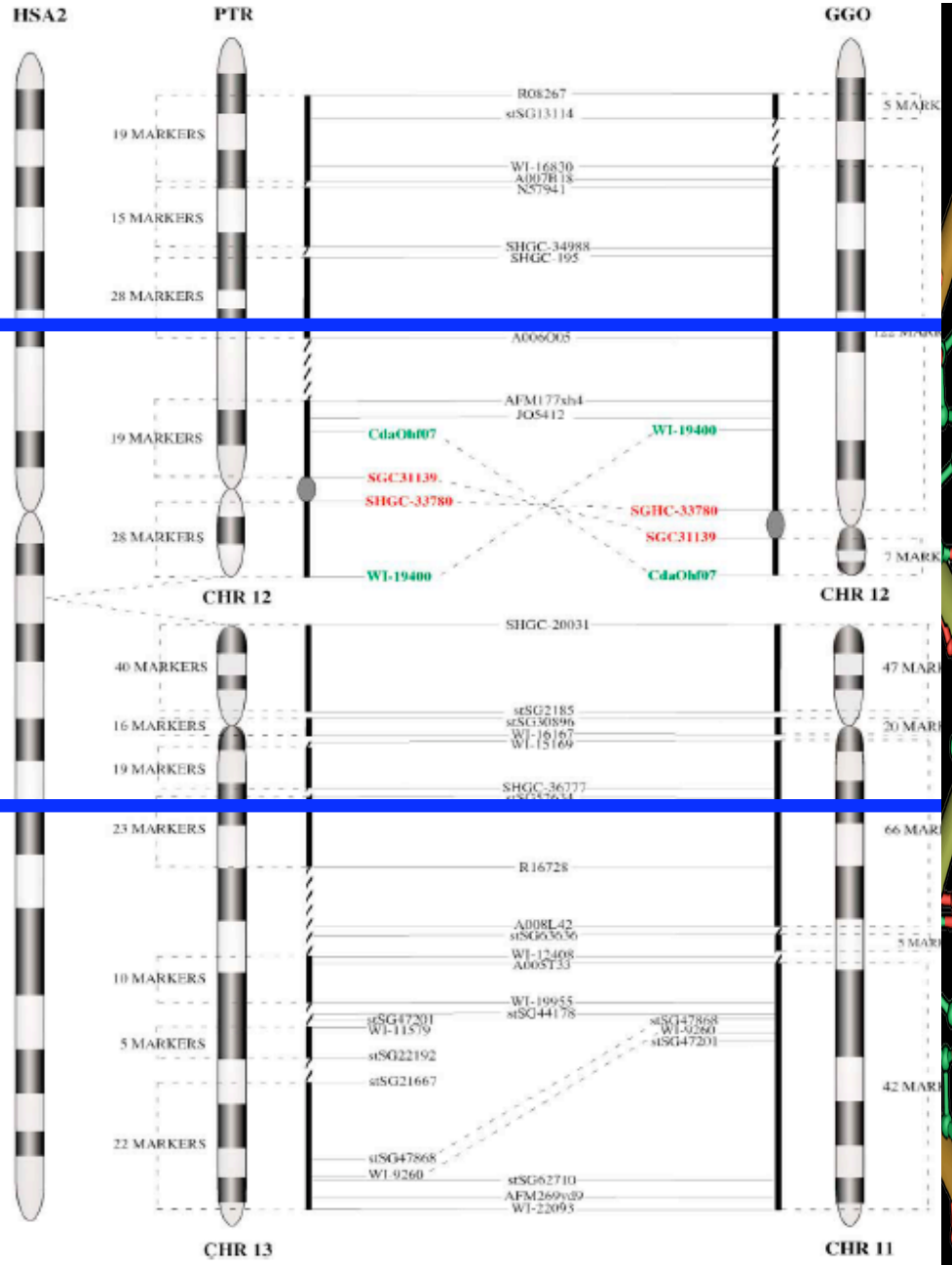
Synteny Comparison between Apes and Human Using Fine-Mapping of the Genome

xandra de Pontbriand,[1] Xiao-Ping Wang,[1,2] Yvon Cavaloc,[1] Marie-Geneviève Mattei,[3] and Francis Galibert[1,*]

GENOMICS Vol. 80, Number 4, October 2002

0888-7543/02 $35.00

Human : chimpanzee synteny



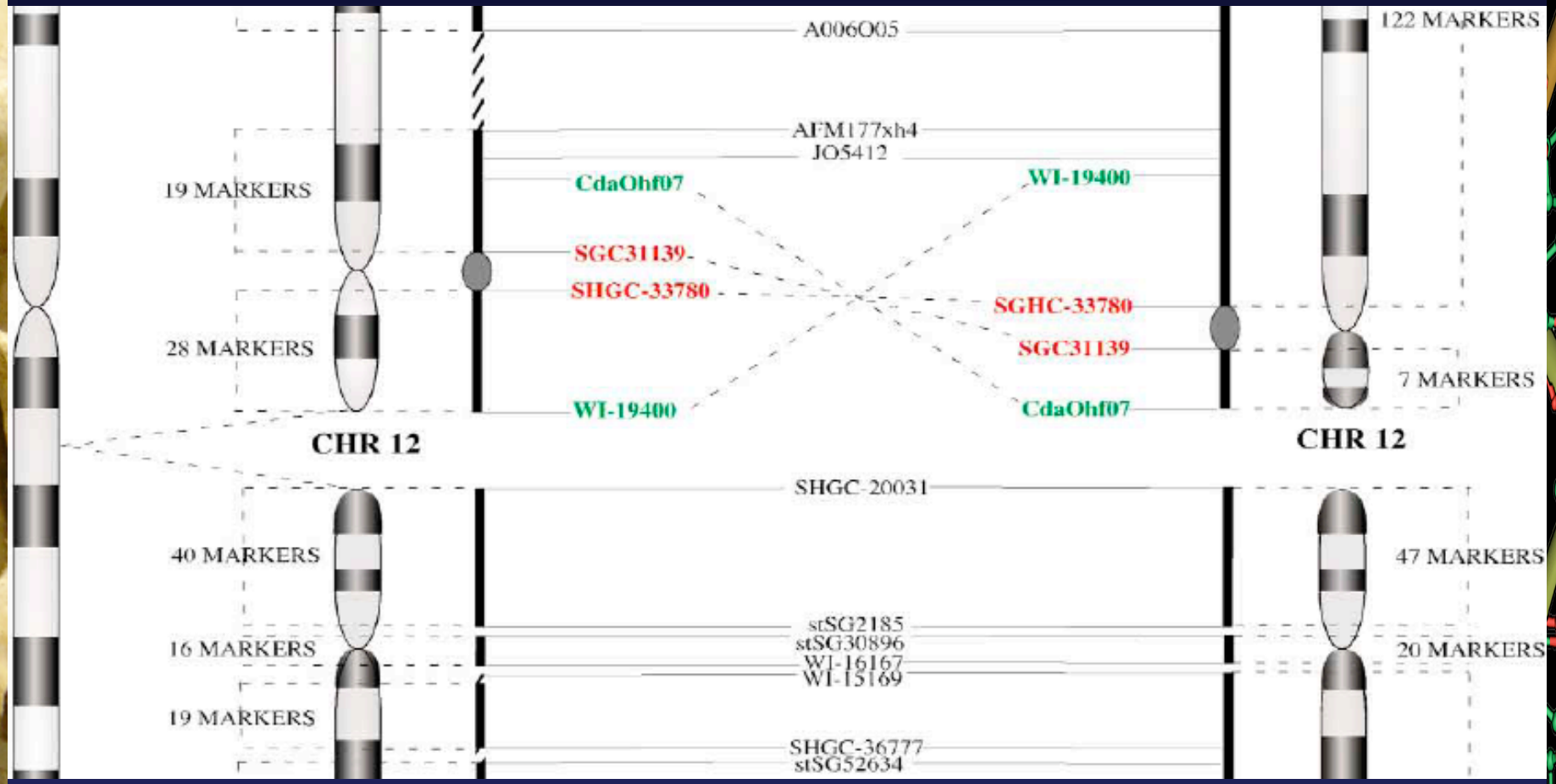
Synteny Comparison between Apes and Human Using Fine-Mapping of the Genome

xandra de Pontbriand,[1] Xiao-Ping Wang,[1,2] Yvon Cavaloc,[1] Marie-Geneviève Mattei,[3] and Francis Galibert[1,*]

GENOMICS Vol. 80, Number 4, October 2002

0888-7543/02 $35.00

Human : chimpanzee synteny



Synteny Comparison between Apes and Human Using Fine-Mapping of the Genome

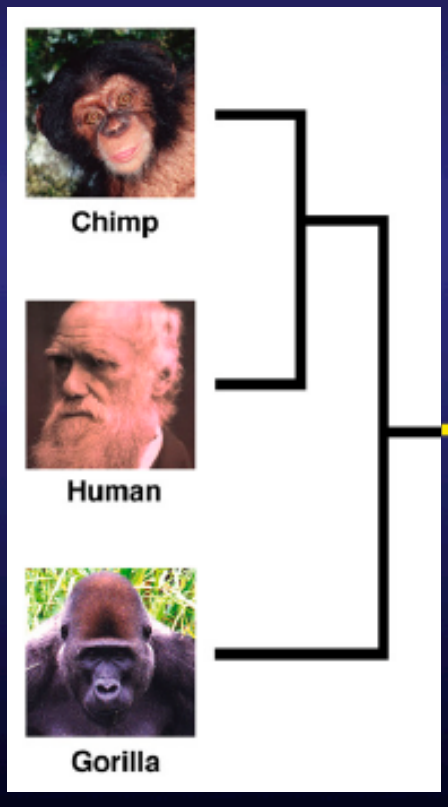
xandra de Pontbriand,[1] Xiao-Ping Wang,[1,2] Yvon Cavaloc,[1] Marie-Geneviève Mattei,[3] and Francis Galibert[1,*]

GENOMICS Vol. 80, Number 4, October 2002

0888-7543/02 $35.00

"Surely synteny reflects a higher-order genomic organization required for function?"

Evidence for human :
chimp common ancestry



BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

Homology

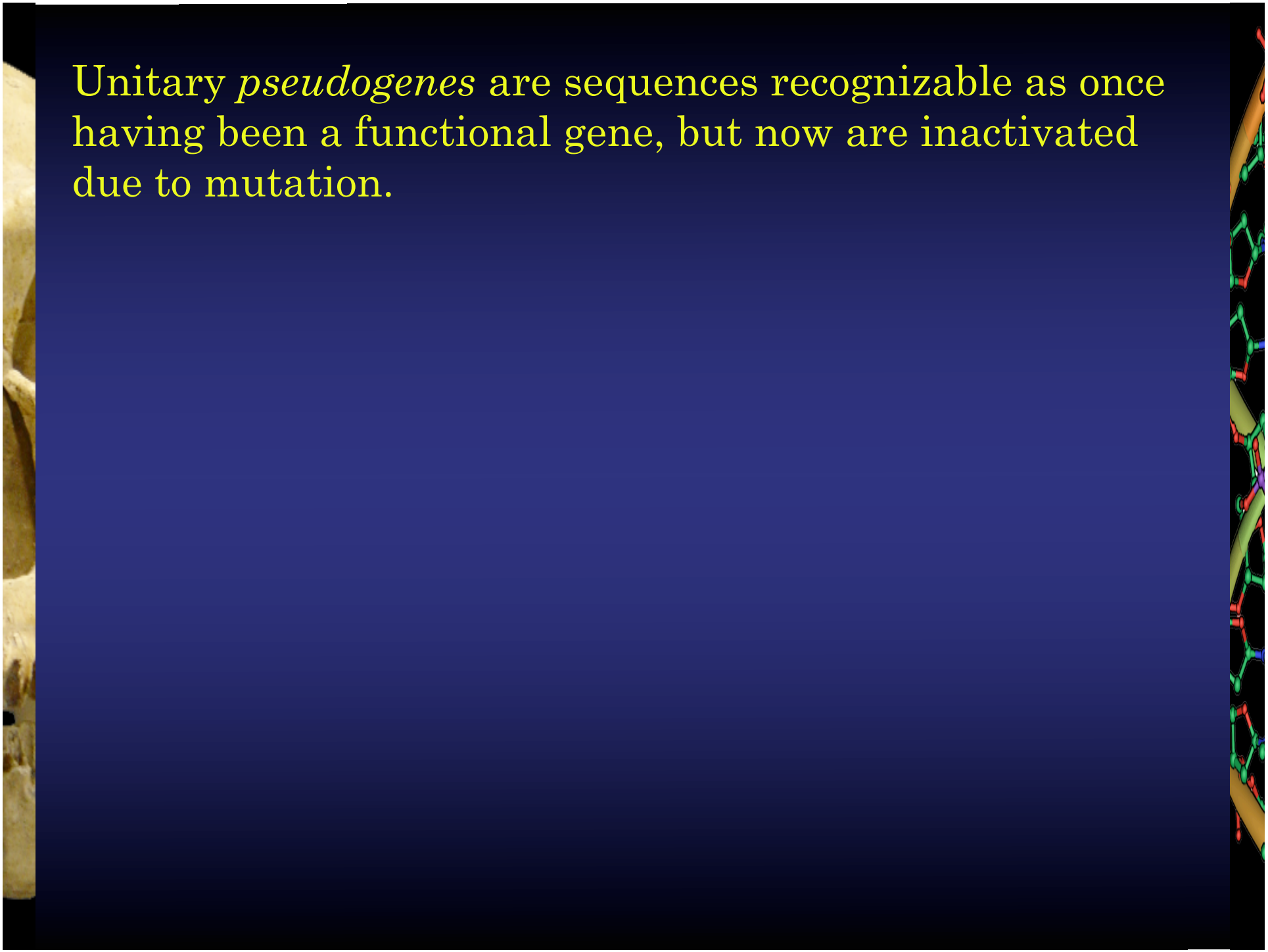
Redundancy

Synteny

Pseudogeny

Parsimony

Unitary *pseudogenes* are sequences recognizable as once having been a functional gene, but now are inactivated due to mutation.



Human specific loss of olfactory receptor genes

Yoav Gilad*,†,‡, Orna Man‡, Svante Pääbo*, and Doron Lancet‡

+ Author Affiliations

Edited by Henry C. Harpending, University of Utah, Salt Lake City, UT, and approved January 3, 2003 (received for review September 20, 2002)

Published online before print February 28, 2003, doi: 10.1073/pnas.0535697100

PNAS March 18, 2003 vol. 100 no. 6 3324–3327

The primate olfactory receptor subgenome reveals numerous pseudogenes shared between humans and great apes (with identical inactivating mutations), as well as human-specific pseudogenized loci.

These pseudogenes are retained in syntenic blocks between genomes.

Human specific loss of olfactory receptor genes

Yoav Gilad*,†,‡, Orna Man‡, Svante Pääbo*, and Doron Lancet‡

+ Author Affiliations

Edited by Henry C. Harpending, University of Utah, Salt Lake City, UT, and approved January 3, 2003 (received for review September 20, 2002)

« Previous | Next Article »
Table of Contents

This Article

Published online before print February 28, 2003, doi: 10.1073/pnas.0535697100

PNAS March 18, 2003 vol. 100 no. 6 3324–3327

Abstract *Free*
Figures Only
» Full Text

Table 1

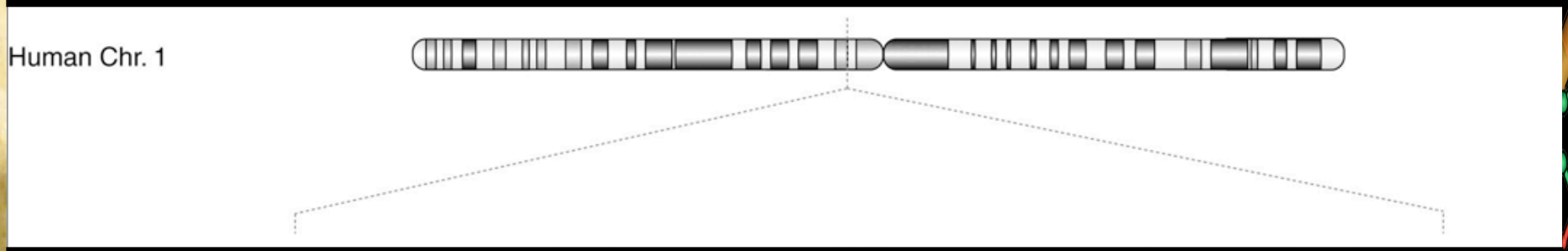
OR genes analyzed

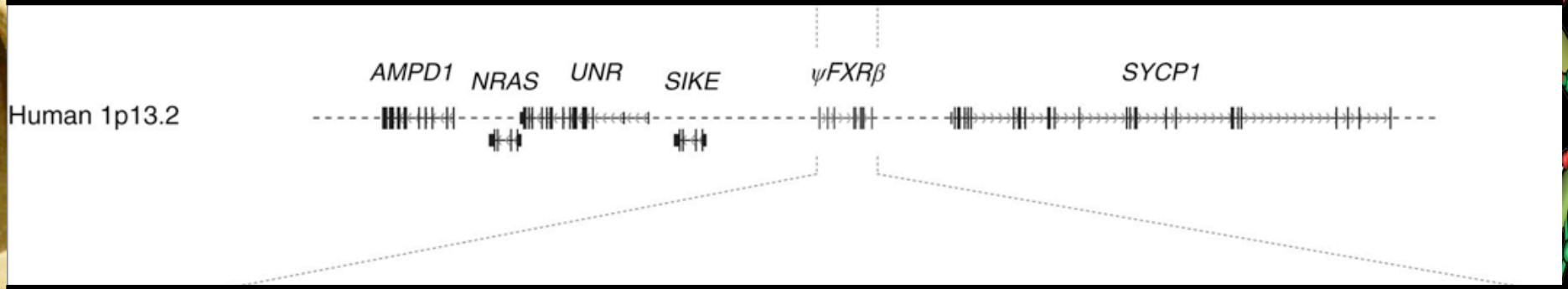
Locus	Human	Chimp	Gorilla	Orang	Rhesus
10A3		824-stop			
10A5					396-sto
10AA1p	302-del	302-de	302-del		
10J5					238-sto
10T1p	391-stop				
11H7p	691-stop				
11i1p	274-stop	274-stop	274-stop		ND
11K1p	176-stop	176-stop	176-stop		176-sto
12D1p	557-del			235-ins	
13C3				180-stop	
13C6p	235-del	744-del			
13D1				105-stop	
13E1p	182-del	182-del			
13H1				300-del	404-del
1C1					
1J2					317-sto
1L3					ND
1S1			96-del		
2Ai1p	528-del	528-del		203-del	
2J3				530-ins	
2L8			444-stop	245-ins	
2Q1p	505-del			507-del	518-del
2T7p	77-del				
4A13p	174-stop		406-stop		
4A4				309-del	
4E1p					
4F15			ND	328-del	
4G3p	178-del	178-del	178-del	178-del	178-del

Analysis of Nuclear Receptor Pseudogenes in Vertebrates: How the Silent Tell Their Stories

Zhengdong D. Zhang, Philip Cayting,* George Weinstock,† and Mark Gerstein*‡§*

Mol. Biol. Evol. 25(1):131–143. 2008
doi:10.1093/molbev/msm251
Advance Access publication December 7, 2007

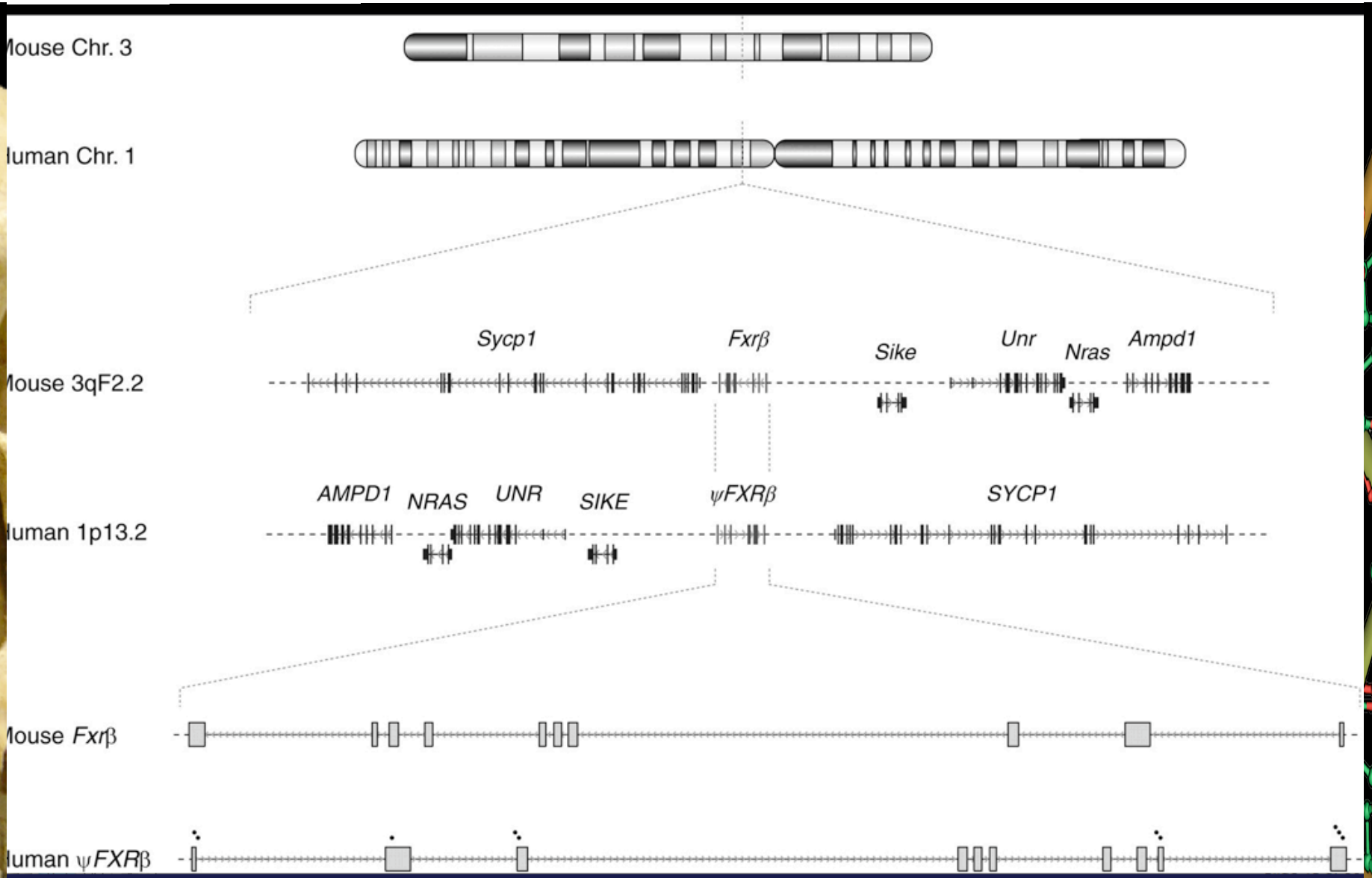




Analysis of Nuclear Receptor Pseudogenes in Vertebrates: How the Silent Tell Their Stories

Zhengdong D. Zhang, Philip Cayting,* George Weinstock,† and Mark Gerstein*‡§*

Mol. Biol. Evol. 25(1):131–143. 2008
doi:10.1093/molbev/msm251
Advance Access publication December 7, 2007



Analysis of Nuclear Receptor Pseudogenes in Vertebrates: How the Silent Tell Their Stories

Zhengdong D. Zhang, Philip Cayting,* George Weinstock,† and Mark Gerstein*‡§*

Mol. Biol. Evol. 25(1):131–143. 2008
doi:10.1093/molbev/msm251
Advance Access publication December 7, 2007

Analysis of Nuclear Receptor Pseudogenes in Vertebrates: How the Silent Tell Their Stories

Zhengdong D. Zhang, Philip Cayting,* George Weinstock,† and Mark Gerstein*‡§*

Mol. Biol. Evol. 25(1):131–143. 2008
doi:10.1093/molbev/msm251
Advance Access publication December 7, 2007

Farnesoid receptor Beta

```
HS      atg gca aat act tat gtc act gca tgt gat agg tat tgt ctt gct gaa cca gt. cag tgc
PT      atg gca aat act tat gtc act gca tgt gat agg tat tgt ctt gct gaa cca gt. cag tgc
CLF     atg gca aat act tat gtc act aca tct gat ggg tac tgt ctt gct gaa cca gtg cag tac
MM      atg gca aat acc tat gtt gct acg tct gat ggg tac tac ctc gct gaa ccc aca cag tat
```

HS = *Homo sapiens* (human)
PT = *Pan troglodytes* (chimpanzee)
CLF = *Canis lupus familiaris* (dog)
MM = *Mus musculus* (mouse)

RESEARCH ARTICLE

OPEN ACCESS

Loss of Egg Yolk Genes in Mammals and the Origin of Lactation and Placentation

Citation: Brawand D, Wahli W, Kaessmann H (2008) Loss of Egg Yolk Genes in Mammals and the Origin of Lactation and Placentation. PLoS Biol 6(3): e63. doi:10.1371/journal.pbio.0060063

Vitellogenin loci persist as pseudogenes in non-prototherian mammals:

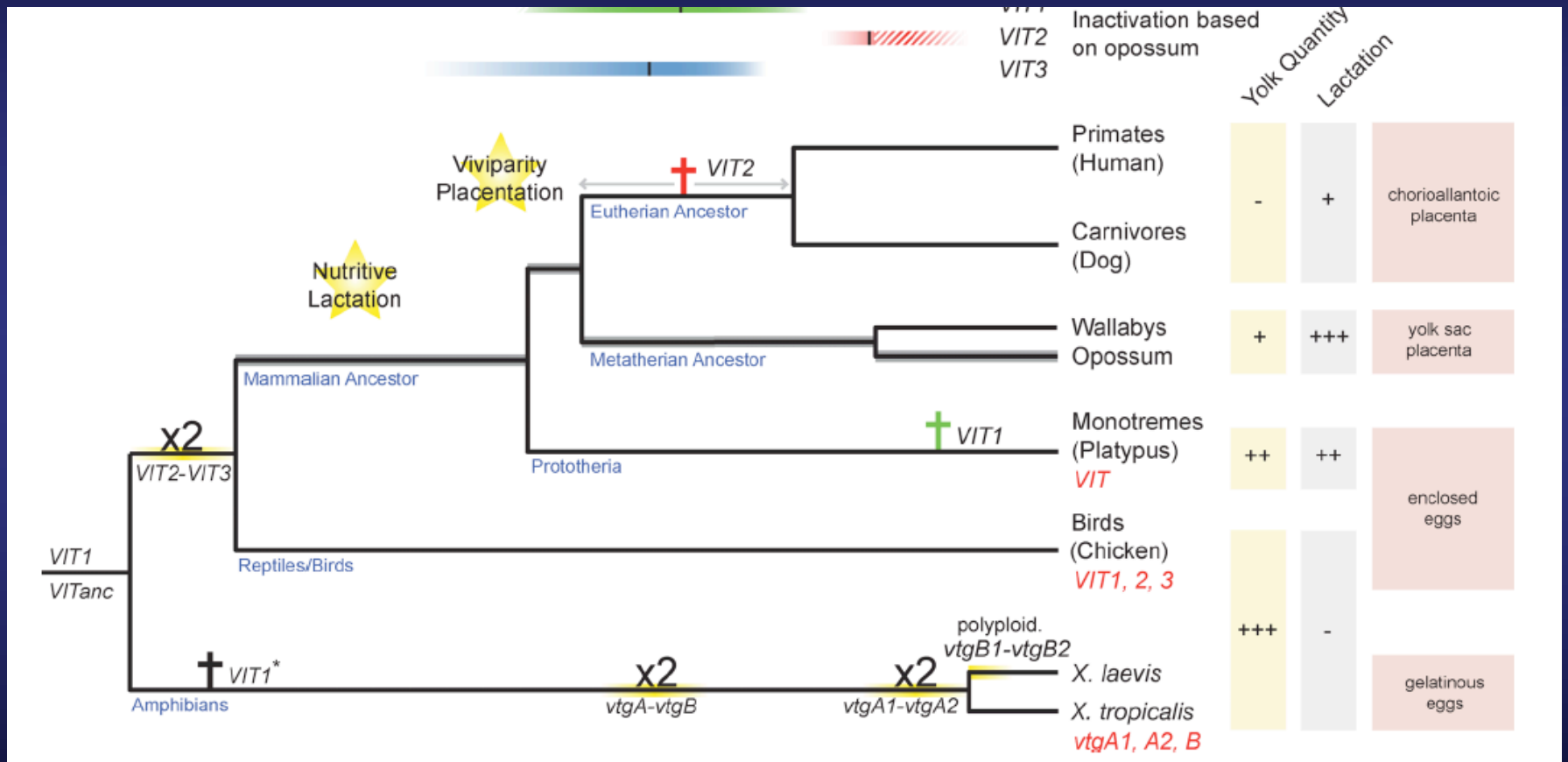


Figure 1. *VIT* Gene Evolution in Tetrapods

The topology and divergence times of the tree are based on previous studies [19,24,25,41]. Latin crosses indicate *VIT* inactivation events in eutherians and monotremes. Inactivation estimates (including approximated 95% prediction intervals) based on opossum *VIT* sequences are indicated by colored bars at the top (see also Figure 3). Duplications ("x2") are indicated. *VITanc* is the likely ancestor of both the amphibian *vtgA1/vtgA2* and *VIT2/VIT3* genes

The *vitellogenin (VIT1)* pseudogene in humans is found in a region syntenic with the chicken genome

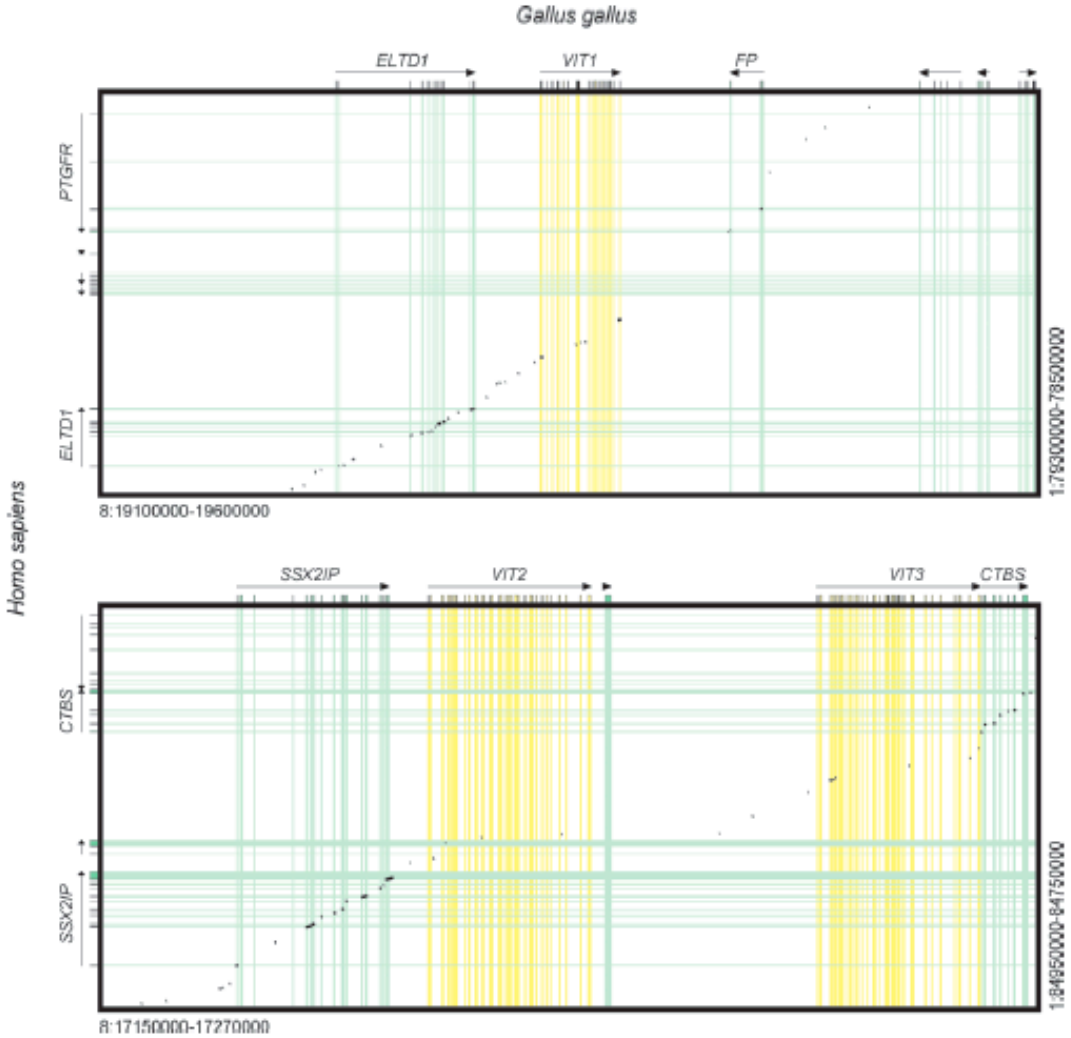


Figure 2. Genome Alignment (Dot Plot Representing SIM Alignments) of Human/Chicken Syntenic Regions *VIT 1-VIT3* Regions

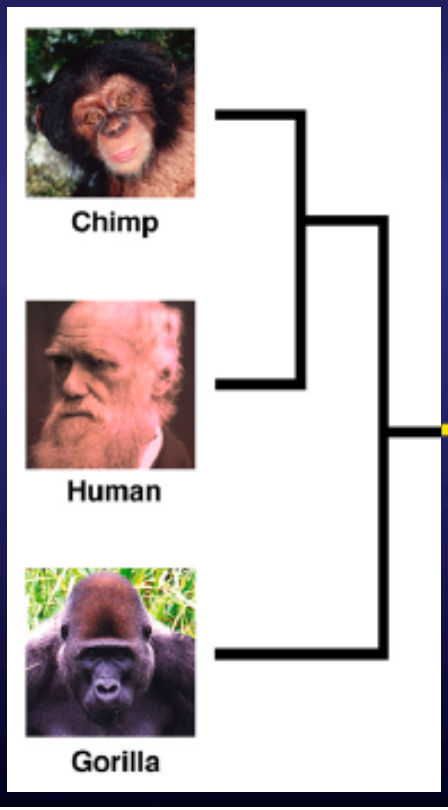
The chain with the best cumulative score is shown. Alignment of flanking genes confirms the synteny of the aligned regions. The combined alignments of *VIT1* coding sequences showed significantly higher alignment scores than the genomic background (introns and intergenic regions) in the chain, as assessed by a Mann-Whitney *U* test ($p < 0.05$). Thus, we can statistically exclude that detected *VIT1* remnants from humans represent spurious sequence matches. The coding sequence matches for *VIT2/3* may be too short to provide statistical significance or partially spurious.

doi:10.1371/journal.pbio.0060063.g002

"Perhaps pseudogenes have an as yet undiscovered function?"

"The identical inactivation mutations could indicate multiple independent inactivations in a non-random manner."

Evidence for human : chimp common ancestry



BMC Biology 2007, 5:7 doi:10.1186/1741-7007-5-7

Homology

Redundancy

Synteny

Pseudogeny

Parsimony

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology		
Redundancy		
Synteny		
Pseudogeny		

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	
Redundancy		
Synteny		
Pseudogeny		

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	
Redundancy	As predicted	
Synteny		
Pseudogeny		

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	
Redundancy	As predicted	
Synteny	Observed synteny consistent with recent divergence	
Pseudogeny		

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	
Redundancy	As predicted	
Synteny	Observed synteny consistent with recent divergence	
Pseudogeny	As predicted	

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	Designer constrained by function, *appearance* of common descent required
Redundancy	As predicted	
Synteny	Observed synteny consistent with recent divergence	
Pseudogeny	As predicted	

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	Designer constrained by function, *appearance* of common descent required
Redundancy	As predicted	Designer constrained by function, *appearance* of common descent required
Synteny	Observed synteny consistent with recent divergence	
Pseudogeny	As predicted	

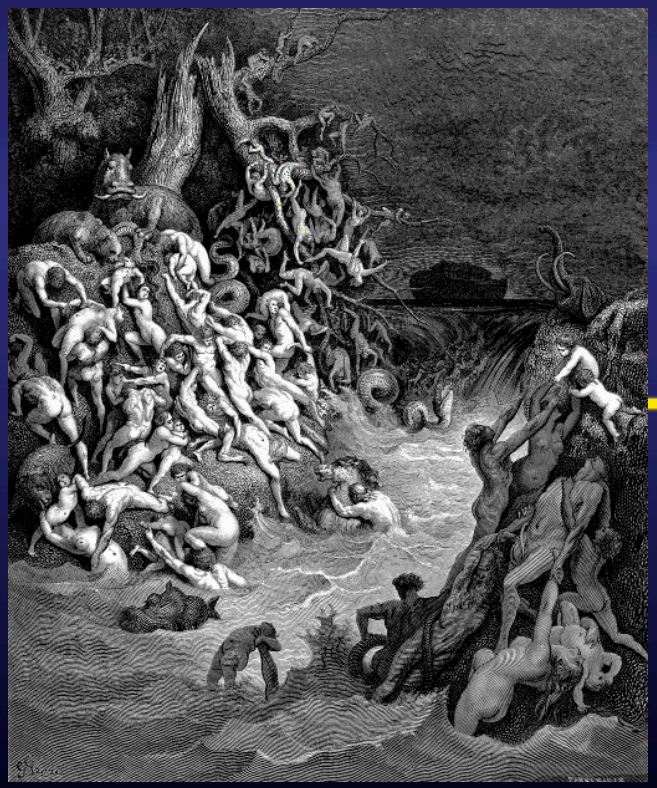
Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	Designer constrained by function, *appearance* of common descent required
Redundancy	As predicted	Designer constrained by function, *appearance* of common descent required
Synteny	Observed synteny consistent with recent divergence	Designer constrained function, *appearance* of common descent required
Pseudogeny	As predicted	

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Homology	As predicted	Designer constrained by function, *appearance* of common descent required
Redundancy	As predicted	Designer constrained by function, *appearance* of common descent required
Synteny	Observed synteny consistent with recent divergence	Designer constrained function, *appearance* of common descent required
Pseudogeny	As predicted	Designer constrained by function, *appearance* of common descent required

Evidence for ancestral hominid population size



Gustave Dore: *The Deluge*
(Dore Illustrated Bible, 1865)

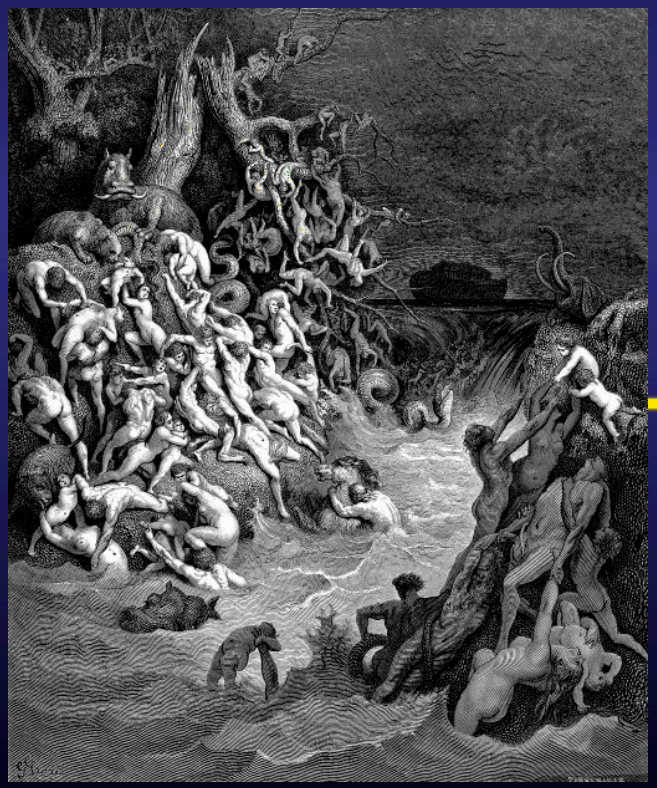
Species trees vs. gene trees

Trans-specific polymorphisms

Polymorphism / heterozygosity - based estimates

Linkage disequillibrium estimates

Evidence for ancestral hominid population size



Gustave Dore: *The Deluge*
(Dore Illustrated Bible, 1865)

Species trees vs. gene trees

Trans-specific polymorphisms

Polymorphism / heterozygosity - based estimates

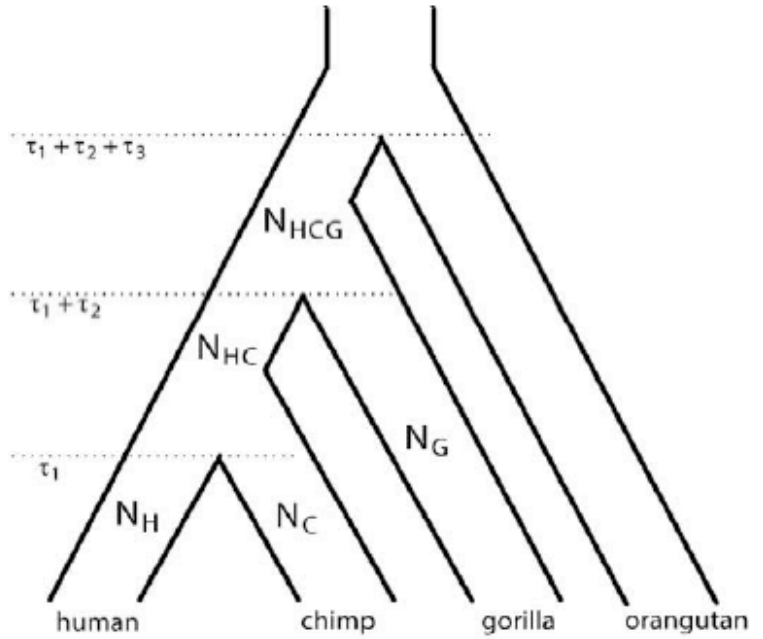
Linkage disequillibrium estimates

OPEN ACCESS Freely available online PLoS GENETICS

Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model

Asger Hobolth[1*], Ole F. Christensen[2], Thomas Mailund[2,3], Mikkel H. Schierup[2]

PLoS Genetics February 2007 | Volume 3 | Issue 2 | e7



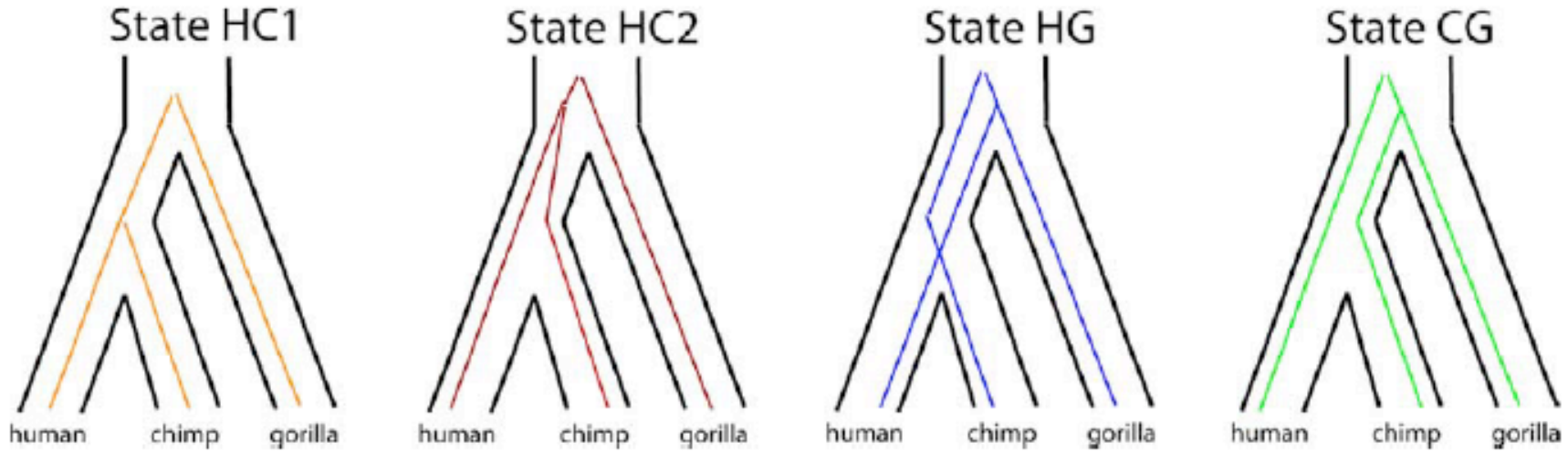


Figure 1. Genetic and Species Relationships May Differ

OPEN ACCESS Freely available online PLoS GENETICS

Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model

Asger Hobolth[1*], Ole F. Christensen[2], Thomas Mailund[2,3], Mikkel H. Schierup[2]

PLoS Genetics February 2007 | Volume 3 | Issue 2 | e7

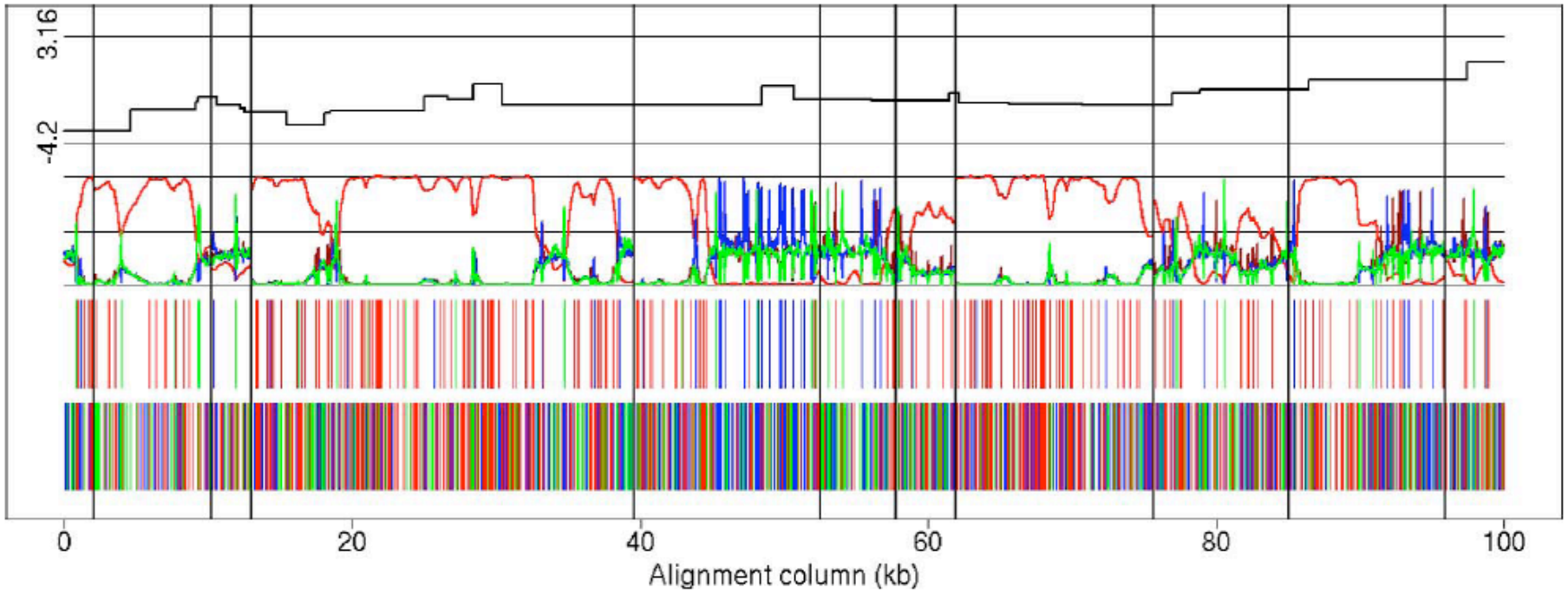
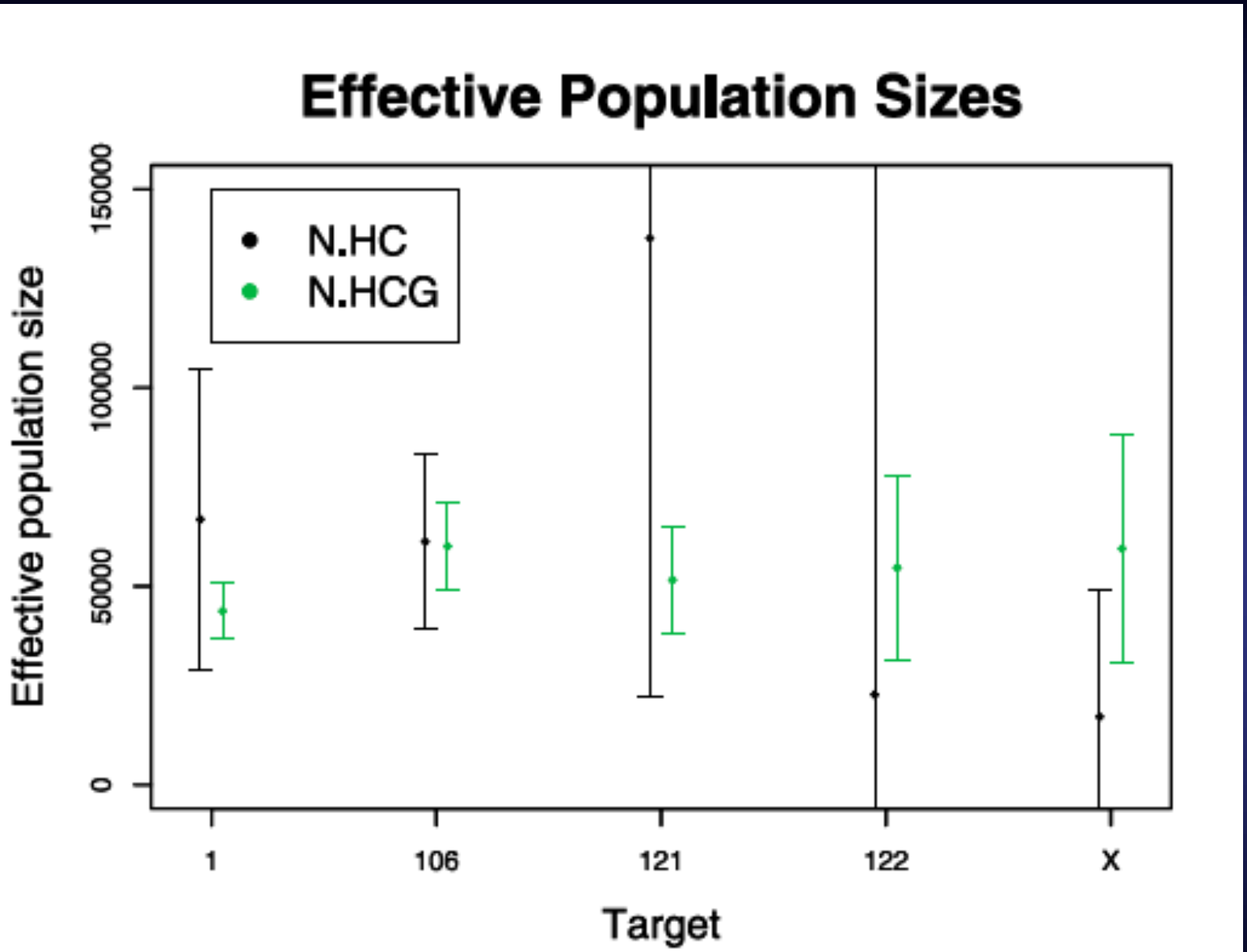
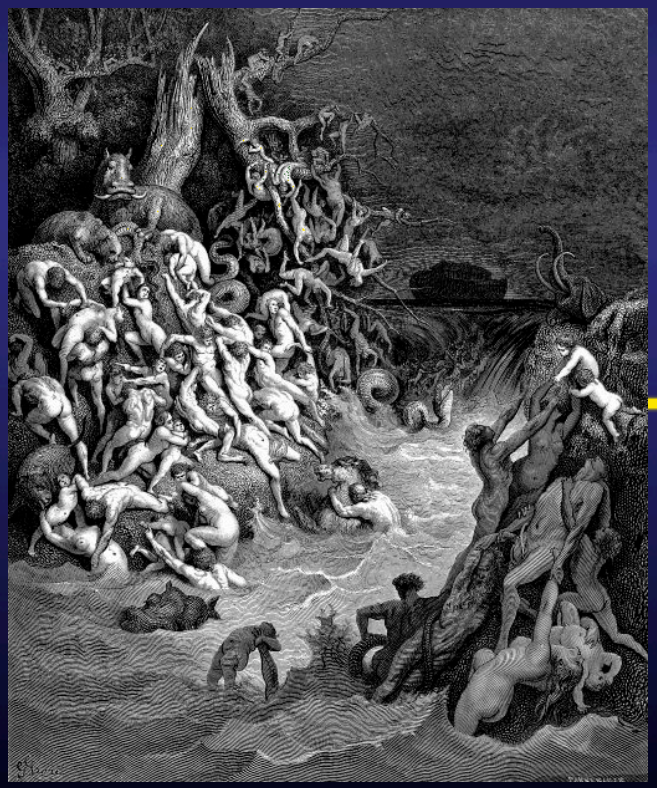


Figure 3. Inferred Genealogies from Real Data

(From bottom to top) Analysis of the first 100 kb from Target 1.
(Bottom) Site information without outgroup: sites shared by HC in red, by HG in blue, and by CG in green (compare first, third, and last columns in Table 1).
(Second from bottom) Site information with outgroup: sites strongly supporting states HC1 or HC2 in red, HG in blue, and CG in green (compare second, third, and last columns in Table 1).
(Third from bottom) Posterior probabilities: Coloring as in second from bottom, except that state HC2 is dark red.
(Top) Fine-scale recombination rate estimates (log scale). The vertical lines mark subdivisions of the multiple alignment due to more than 50-base-pair deletions in one species (see "Data" in Materials and Methods).
doi:10.1371/journal.pgen.0030007.g003


Effective Population Sizes
Effective population size
150000
100000
50000
0
N.HC
N.HCG
1
106
121
122
X
Target

Evidence for ancestral hominid population size



Gustave Dore: *The Deluge*
(Dore Illustrated Bible, 1865)

Species trees vs. gene trees

Trans-specific polymorphisms

Polymorphism / heterozygosity - based estimates

Linkage disequillibrium estimates

Recent human effective population size estimated from linkage disequilibrium

Albert Tenesa,[1,2,3] Pau Navarro,[3] Ben J. Hayes,[4] David L. Duffy,[5] Geraldine M. Clarke,[6] Mike E. Goddard,[4,7] and Peter M. Visscher[3,5,8]

520 Genome Research
www.genome.org

17:520–526

Table 1. N_e estimates from the nonlinear model (Method 1)

	Population							
	CEU		YRI		JPT		HCB	
Chromosome	*n*	N_e	*n*	N_e	*n*	N_e	*n*	N_e
1	1,350,336	2824	1,602,447	6197	1,155,365	2549	1,170,837	2694
2	1,770,347	3197	1,958,708	6675	1,382,444	3200	1,404,237	3294
3	1,318,345	3085	1,300,983	6897	936,628	2534	949,450	2743
4	1,052,614	3232	1,086,591	6351	772,233	3226	790,459	3312
5	1,074,385	3116	974,226	6932	795,412	2554	803,800	2762
6	1,323,089	3031	1,359,998	6745	1,101,243	2678	1,114,544	2854
7	946,353	2196	816,514	6745	600,416	2739	613,743	2897
8	2,292,984	3029	2,531,320	7004	2,036,130	2650	2,061,705	2802
9	1,779,571	3048	1,928,452	7019	1,564,337	2678	1,572,613	2830
10	919,144	3227	1,044,438	6868	770,465	3036	782,584	2860
11	789,664	3027	771,232	6181	613,884	2520	614,225	2669
12	987,085	3157	993,895	7007	733,950	2812	740,215	2919
13	660,098	2031	795,844	4806	547,950	1928	554,326	2041
14	521,596	2232	495,815	4719	390,544	1983	397,645	2134
15	436,105	1933	465,532	4143	346,811	1830	354,237	1878
16	417,032	3017	419,655	7334	324,468	2825	324,315	2935
17	411,015	2935	387,225	6384	307,220	2843	312,735	2752
18	1,223,437	2409	1,459,903	5771	1,008,782	2315	1,029,745	2365
19	270,281	3126	259,975	7416	208,095	2656	218,568	2608
20	393,509	3338	355,554	6864	270,347	2534	275,690	2795
21	656,672	1485	748,128	3855	663,630	1440	672,430	1574
22	619,770	1459	669,718	3246	587,244	1217	584,544	1081
X	924,274	3613	1,117,100	9421	738,824	3141	788,186	3458
Mean	**962,509**	**2772**	**1,023,620**	**6286**	**776,366**	**2517**	**788,297**	**2620**
SD	**513,402**	**598**	**581,811**	**1357**	**447,337**	**524**	**452,066**	**557**

(*n*) Number of marker pairs; (N_e) effective population size. Intermarker distance was in the range of 5 kb to100 kb for all SNP pairs.

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Population genetics		

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Population genetics	Complex speciation, large ancestral population	

Two competing hypotheses:

	Common descent	**Non-evolutionary Designer**
Population genetics	Complex speciation, large ancestral population	Designer constrained by function, *appearance* of common descent required

Thoughts on Biblical scientific concordism

Titian: *The Fall of Man* (c. 1570)

Thoughts on Biblical scientific concordism

Titian: *The Fall of Man* (c. 1570)

"Ratcheting concordism"

Thoughts on Biblical scientific concordism

Titian: *The Fall of Man* (c. 1570)

"Ratcheting concordism"

vs.

ANE phenomenological perspective

Thoughts on Biblical scientific concordism

Titian: *The Fall of Man* (c. 1570)

"Ratcheting concordism"

vs.

ANE phenomenological perspective

Higher view of Scripture?

Titian: *The Fall of Man* (c. 1570)

Questions?