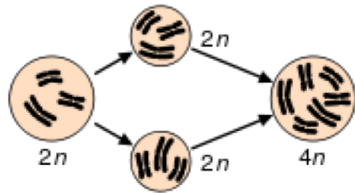
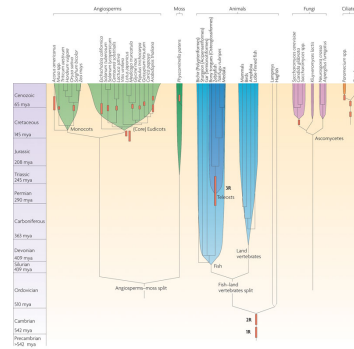


Genome duplications (polyploidy) / ancient genome duplications (paleopolyploidy)/ ancient genome hybridization (allo-paleopolyploidy)



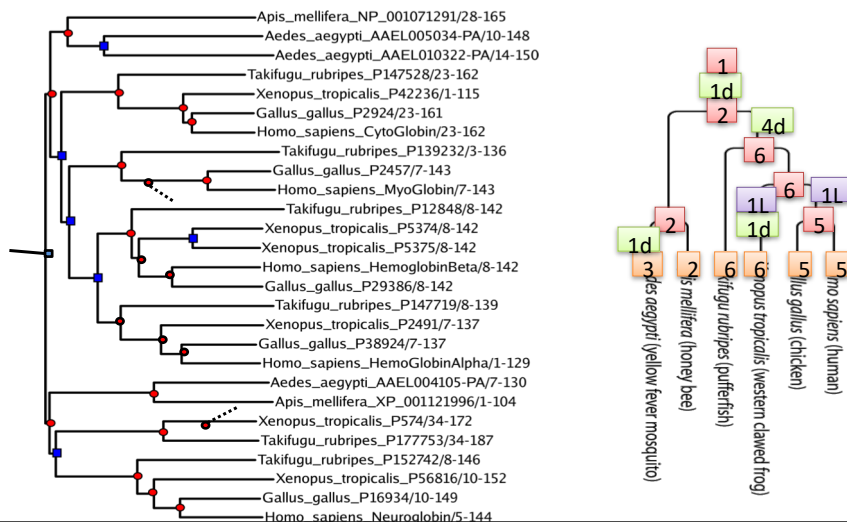
Mechanism? e.g. a diploid cell undergoes failed meiosis, producing diploid gametes, which self-fertilize to produce a tetraploid zygote.

How to detect paleoploidy?

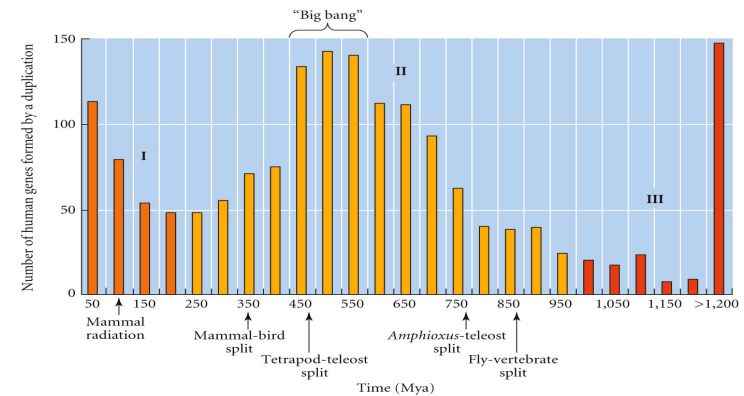


Non-sequitur? How this lecture fits in the course.

- Something to expect in gene trees.
 - If I see two paralogs in human my first guess is that they duplicated at base of vertebrates: provides scenario/search image
- *We want to detective What* has happened, this is generally difficult: here we are clear what happened because of synteny
- Some fringe / *in extremis* discussion of paralogy & orthology
- Much more well defined exposition of
 - Again importance of gene loss: provides nice example of expansion contraction / biphasic model of genome evolution
 - Changes in Speed of evolution



Phylogenetic timing of duplicates hints are massive duplications at the base of vertebrates



EVOLUTION 2e, Figure 20.19

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Paramecium genome duplications

Vol 444 | 9 November 2006 | doi:10.1038/nature05230

nature

ARTICLES

Global trends of whole-genome duplications revealed by the ciliate *Paramecium tetraurelia*

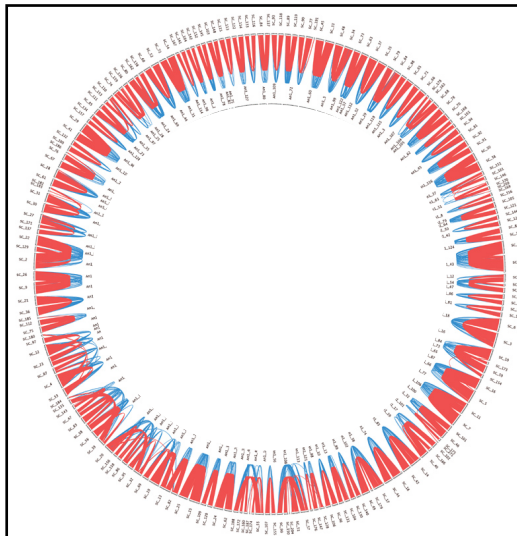
Jean-Marc Aury¹*, Olivier Jaillon¹*, Laurent Duret², Benjamin Noel³, Claire Jubin⁴, Betina M. Porcel¹, Béatrice Séguens¹, Vincent Daubin², Véronique Anthouard¹, Nathalie Aïach¹, Olivier Arnaiz², Alain Billaut¹, Janine Beisson², Isabelle Blanc¹, Khaled Bouhouche¹, Francisco Cámara³, Sandra Duharcourt¹, Roderic Guigo³, Delphine Gogendeau¹, Michael Katinka¹, Anne-Marie Keller², Roland Kissmehl¹, Catherine Klotz², France Koll¹, Anne Le Mouél¹, Gersende Lepère¹, Sophie Malinsky⁴, Mariusz Nowacki⁴, Jacek K. Nowak⁴, Helmut Plattner⁶, Julie Poulain¹, Françoise Ruiz¹, Vincent Serrano¹, Marek Zagulski¹, Philippe Dessen⁸, Mireille Bétermier^{3,4}, Jean Weissenbach¹, Claude Scarpelli¹, Vincent Schächter¹, Linda Sperling¹, Eric Meyer¹, Jean Cohen¹ & Patrick Wincker¹



Comparison of two scaffolds originating from a common ancestor at the recent WGD



Whole genome (all scaffolds) can be aligned to itself



Saccharomyces cerevisiae

Molecular evidence for an ancient duplication of the entire yeast genome

Kenneth H. Wolfe & Denis C. Shields

Department of Genetics, University of Dublin, Trinity College, Dublin 2, Ireland

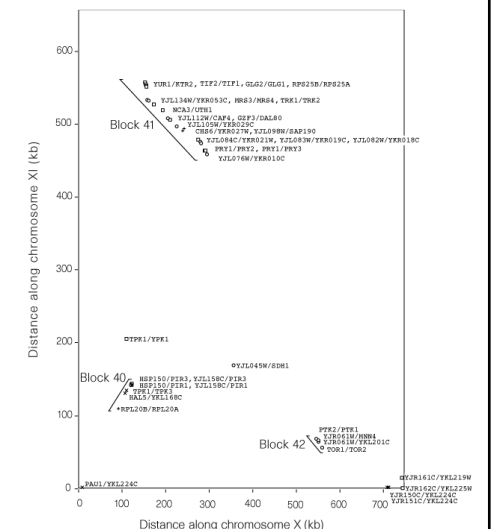
Gene duplication is an important source of evolutionary novelty^{1,2}. Most duplications are of just a single gene, but Ohn³ proposed that whole-genome duplication (polyploidy) is an important evolutionary mechanism. Many duplicate genes have been found in *Saccharomyces cerevisiae*, and these often seem to be phenotypically redundant^{4,5}. Here we show that the arrangement of duplicated genes in the *S. cerevisiae* genome is consistent with Ohn's hypothesis. We propose a model in which this species is a degenerate tetraploid resulting from a whole-genome duplication that occurred after the divergence of *Saccharomyces* from *Kluyveromyces*. Only a small fraction of the genes were subsequently retained in duplicate (most were deleted), and gene order was rearranged by many reciprocal translocations between chromosomes. Protein pairs derived from this duplication event make up 13% of all yeast proteins, and include pairs of transcription factors, protein kinases, mRNAs, cyclins and pheromones. Tetraploidy may have facilitated the evolution of anaerobic fermentation in *Saccharomyces*.

We searched systematically for duplicated regions^{6,7} in the complete yeast genome⁸ by using BLAST⁹ amino-acid sequence similarity searches of all yeast proteins against one another, and plotted the results on dot matrices. Duplicate regions are visible as a diagonal series of 'bars' with conserved gene orientation. In the example shown in Fig. 1, three separate diagonals indicate three distinct regional duplications between chromosomes X and XI. Within each region the homologues are interspersed with genes that are not now duplicated. We propose that this is the result of random deletion of individual duplicated genes from one or other chromosome subsequent to the initial duplication of the whole region.

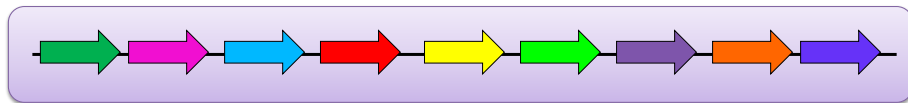
In the whole genome, 35 duplicate regions were identified

First Published Online 12 SEP 1997

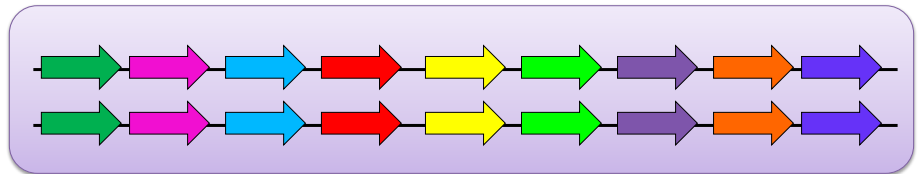
NATURE GENETICS 17(1) JUNE 1997



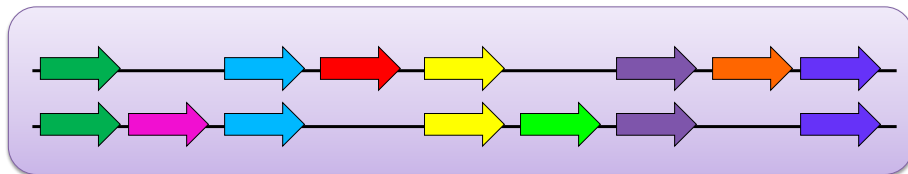
Just before genome duplication



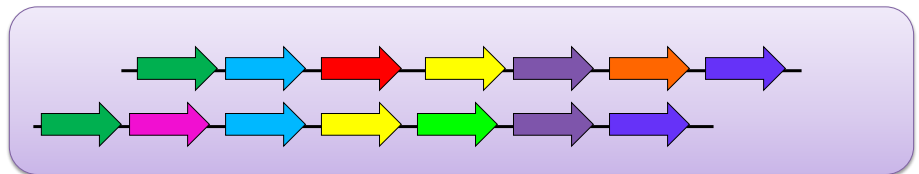
Just after genome duplication

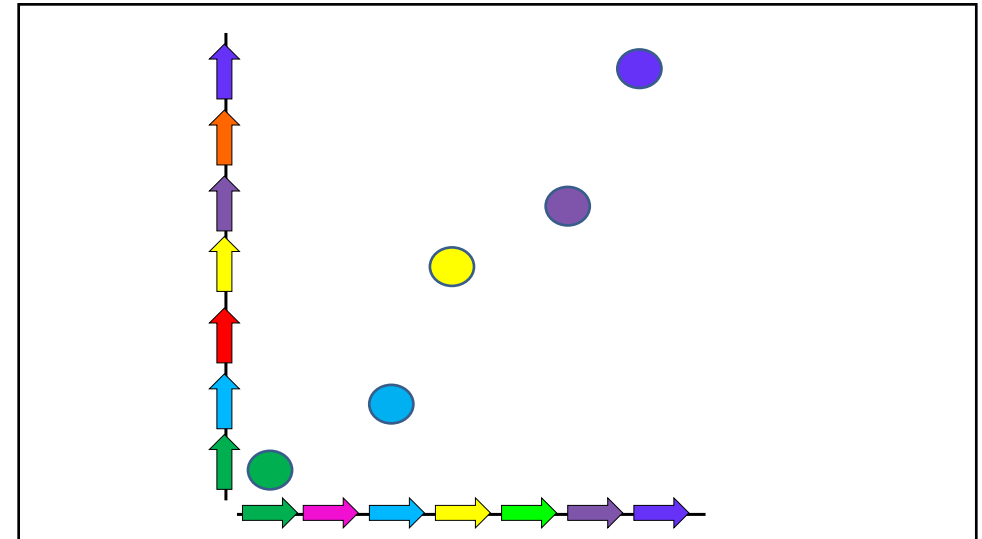
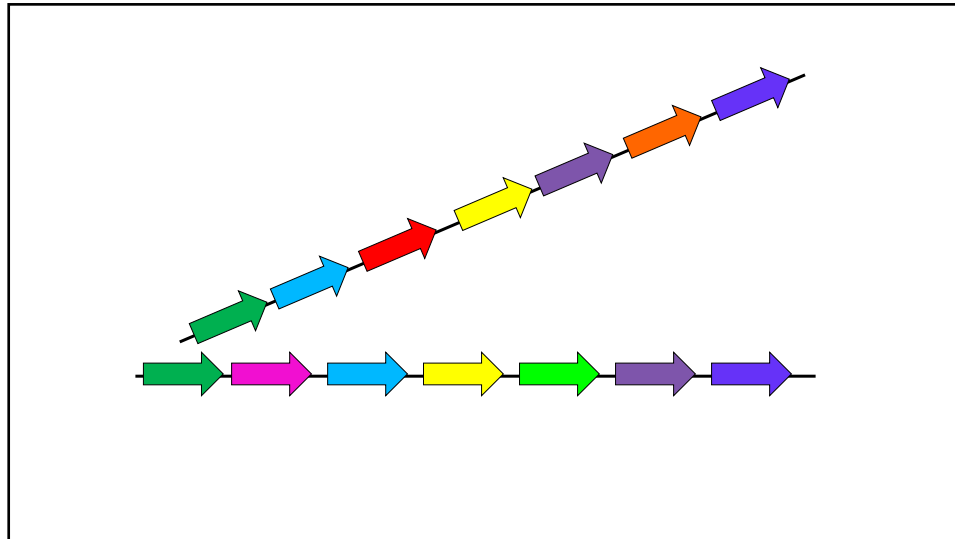


More time after genome duplication



Unaligned view (removing gaps just like in *cerevisiae* has occurred)





Saccharomyces cerevisiae

Molecular evidence for an ancient duplication of the entire yeast genome

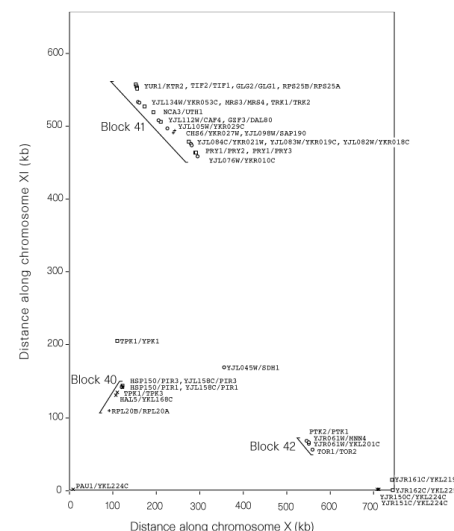
Kenneth H. Wolfe & Denis C. Shields
Departments of Genetics, University of Dublin, Trinity College, Dublin 2, Ireland

Gene duplication is an important source of evolutionary novelty^{1,2}. Most duplications are of just a single gene, but Ohno³ proposed that whole-genome duplication (polyploidy) is an important evolutionary mechanism. Many duplicate genes have been found in *Saccharomyces cerevisiae*, and these often seem to be phenotypically redundant⁴. Here we show that the arrangement of duplicated genes in the *S. cerevisiae* genome is consistent with Ohno's hypothesis. We propose a model in which this species is a degenerate tetraploid resulting from a whole-genome duplication that occurred after the divergence of *Saccharomyces* from *Kluyveromyces*. Only a small fraction of the genes were subsequently retained in duplicate (most were deleted), and gene order was rearranged by many reciprocal translocations between chromosomes. Protein pairs derived from this duplication event make up 1% of all yeast proteins, and include pairs of transcription factors, protein kinases, myosins, cyclins and phosphatases. Tetraploidy may have facilitated the evolution of anaerobic fermentation in *Saccharomyces*.

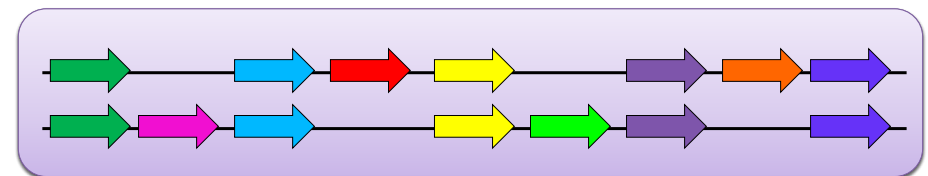
We searched systematically for duplicated regions^{5,6} in the complete yeast genome⁷ by using BLASTP⁸ amino-acid sequence similarity searches of all yeast proteins against one another, and plotted the results on dot matrices. Duplicate regions are visible as a diagonal series of 'hits' with conserved gene orientation. In the example shown in Fig. 1, three separate diagonals indicate three distinct regional duplications between chromosomes X and XI. Within each region the homologues are interspersed with genes that are not now duplicated. We propose that this is the result of random deletion of individual duplicated genes from one or other chromosome subsequent to the initial duplication of the whole region.

In the whole genome, 55 duplicate regions were identified

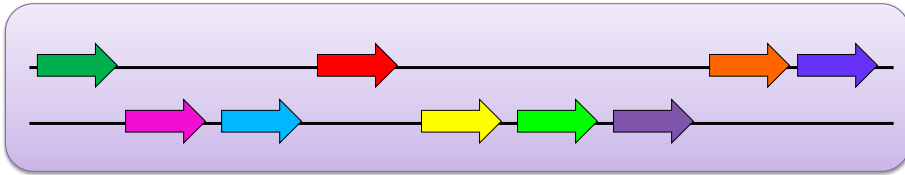
NATURE GENETICS 10, 107-112 (1997)



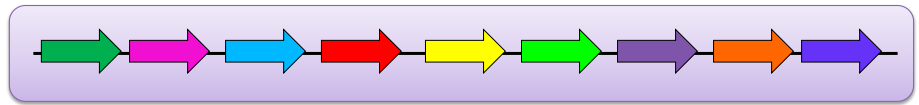
Problem reciprocal gene loss (illustrated with an extreme case); how to solve?



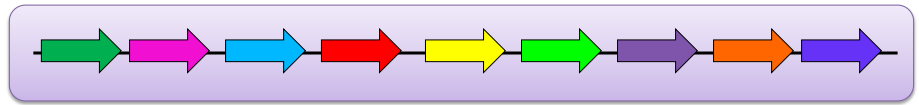
Problem reciprocal gene loss (illustrated with an extreme case); how to solve?



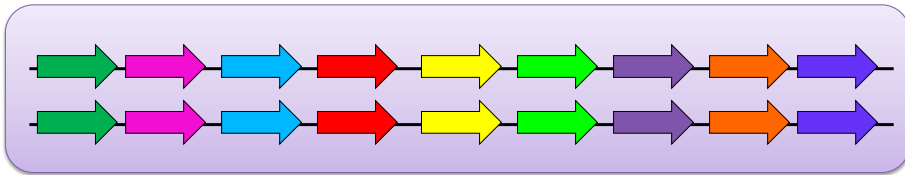
Just before genome duplication



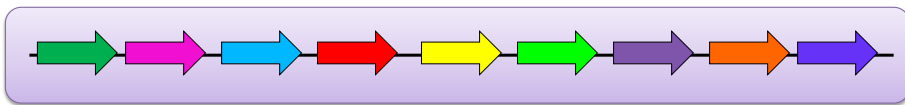
Outgroup!



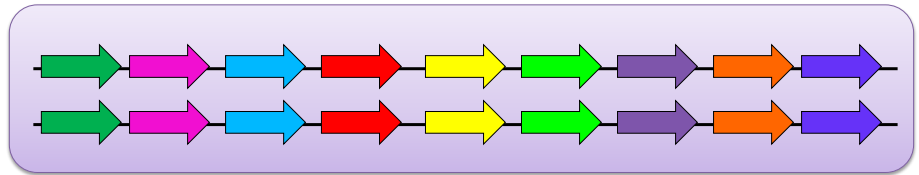
Just after genome duplication



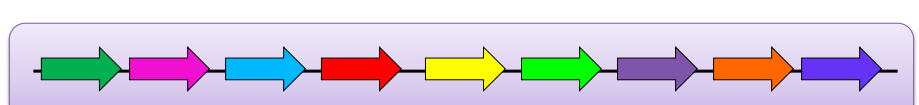
Outgroup



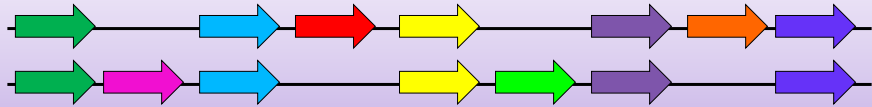
Just after genome duplication



Outgroup



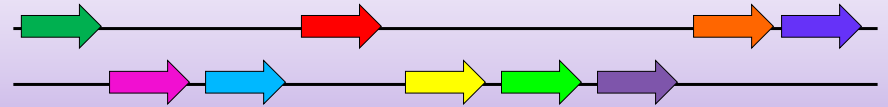
More time after genome duplication



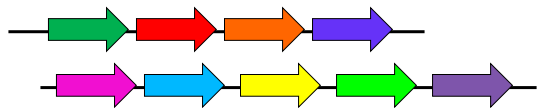
Outgroup



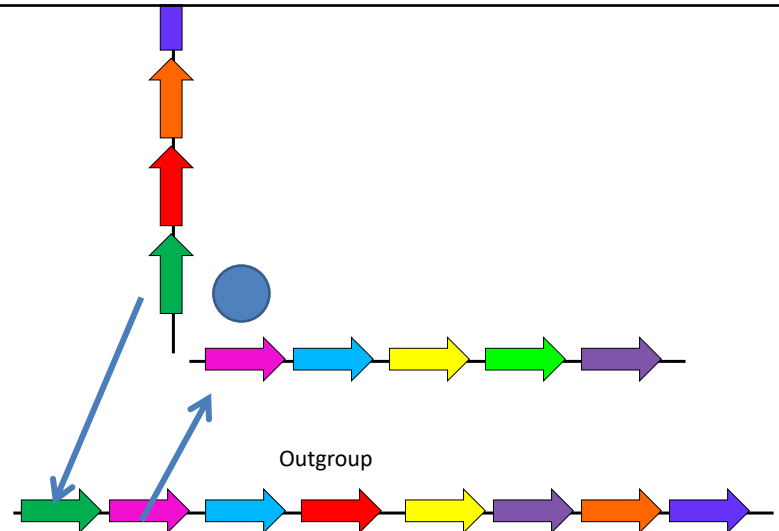
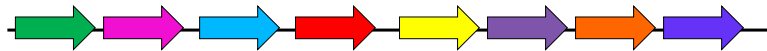
Problem reciprocal gene loss (illustrated with an extreme case); how to solve?



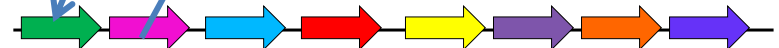
Outgroup

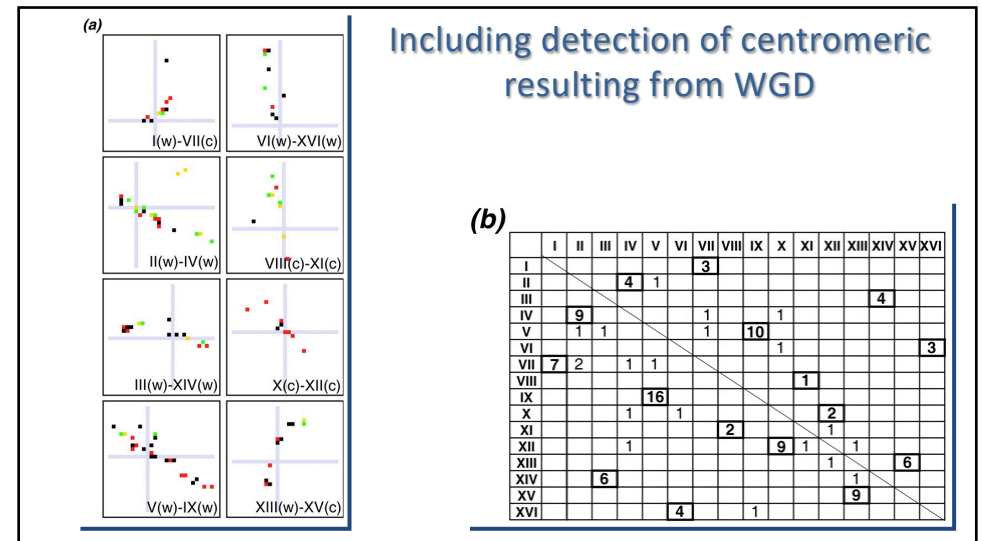
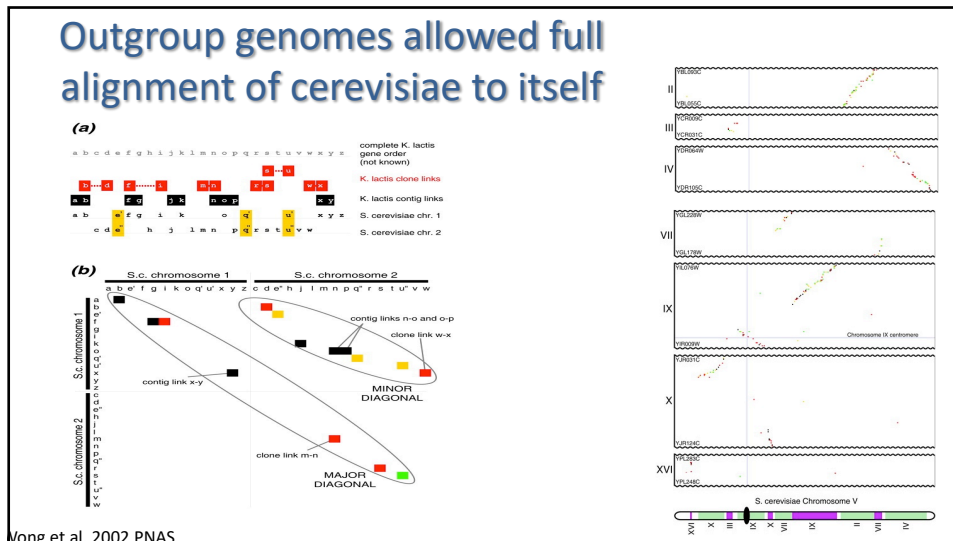
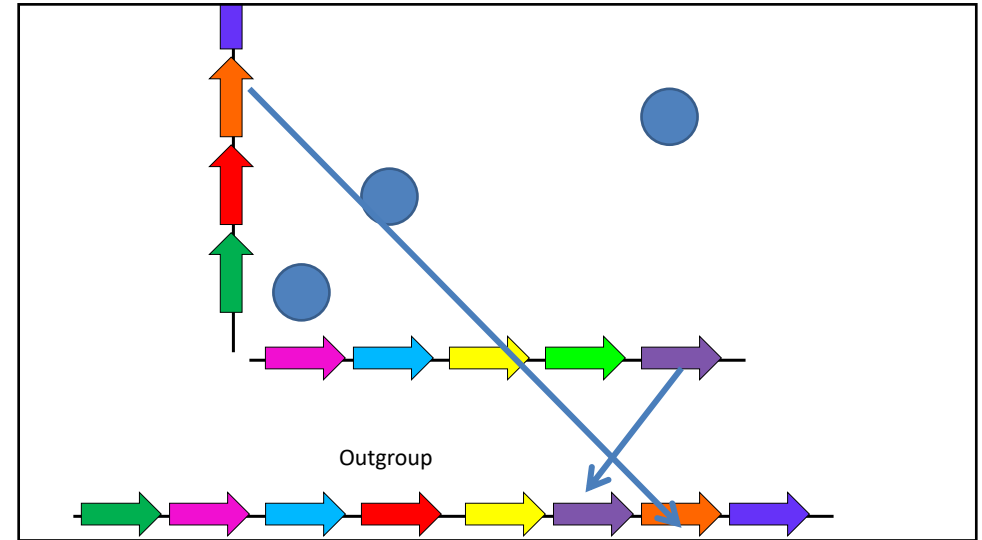
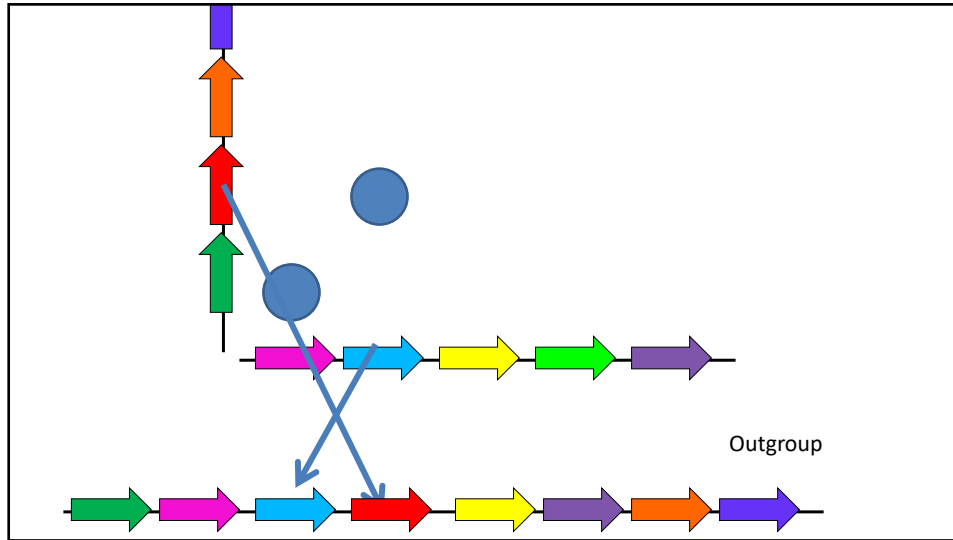


Outgroup



Outgroup





... maybe it was not a whole genome duplication but a hybridization?



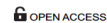
RESEARCH ARTICLE

Beyond the Whole-Genome Duplication: Phylogenetic Evidence for an Ancient Interspecies Hybridization in the Baker's Yeast Lineage

Marina Marcel-Houben^{1,2}, Toni Gabaldón^{1,2,3}*

¹ Centre for Genomic Regulation (CRG), Barcelona, Spain, ² Universitat Pompeu Fabra (UPF), Barcelona, Spain, ³ Institutó Catalana de Recerca i Estudis Avançats (ICREA), Barcelona, Spain

* tgabaldon@crg.es



Citation: Marcel-Houben M, Gabaldón T (2015)

Abstract

Whole-genome duplications have shaped the genomes of several vertebrate, plant, and fungal lineages. Earlier studies have focused on establishing when these events occurred

Clearer example of allopolyploidy (& genome dominance)

ARTICLE

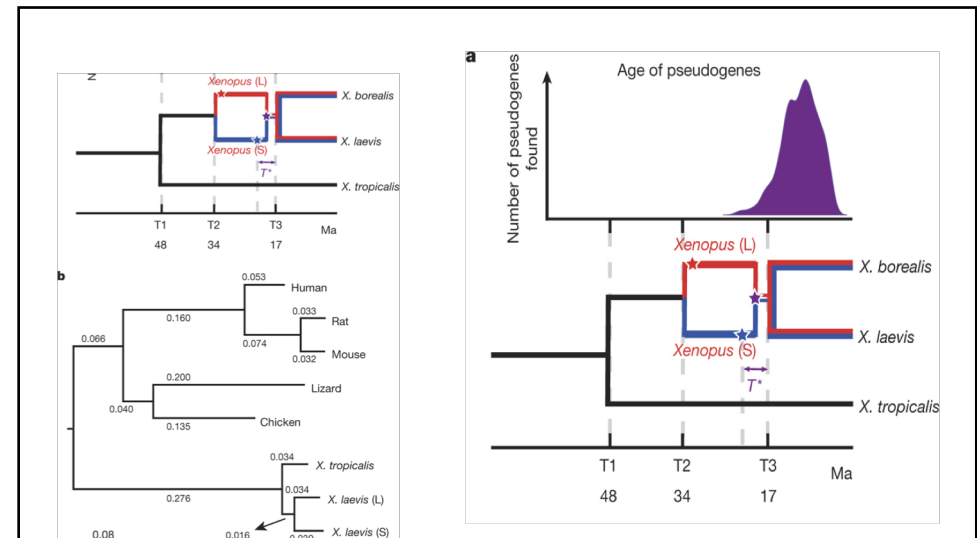
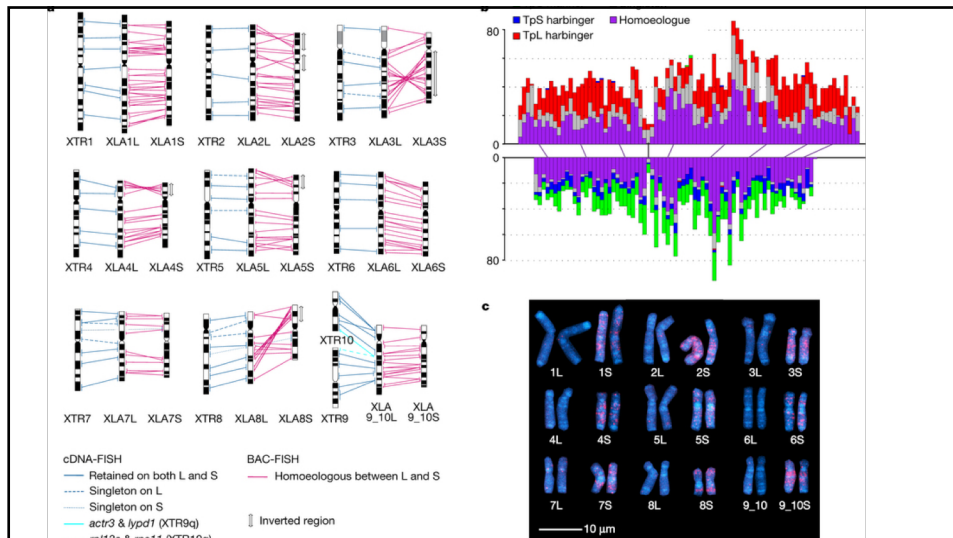
OPEN

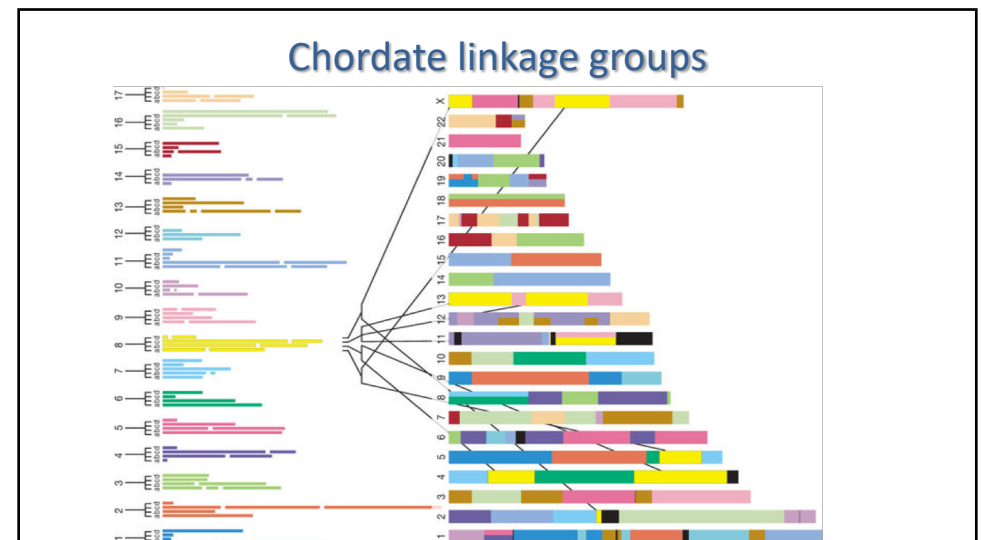
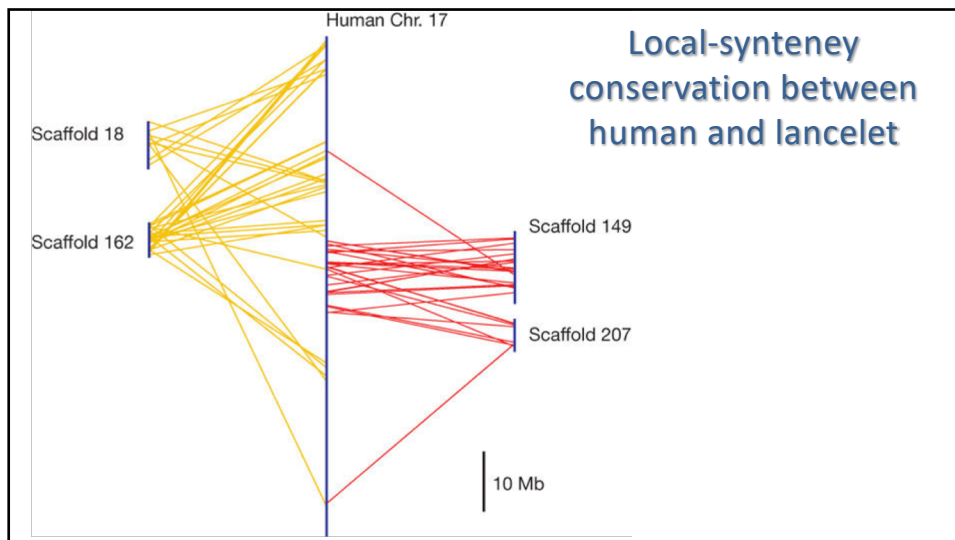
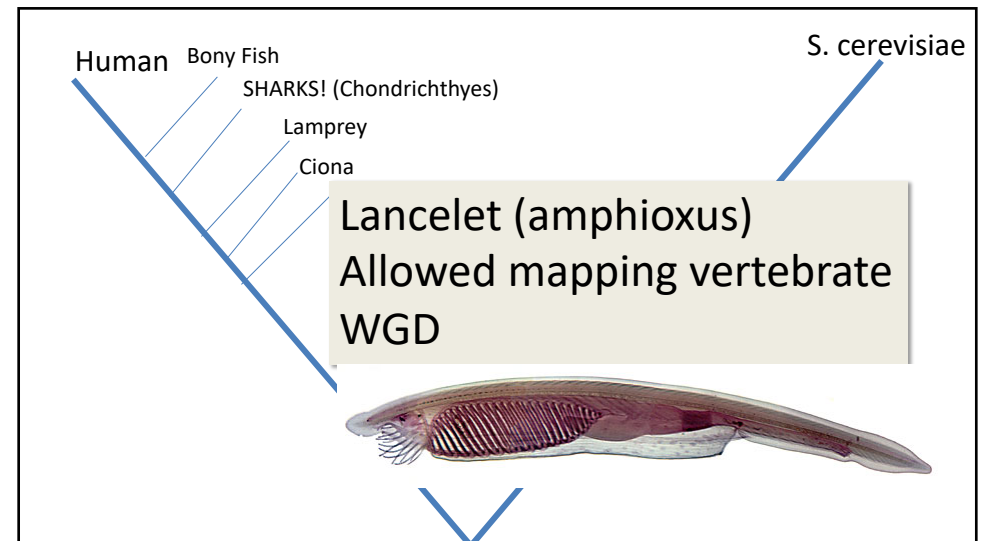
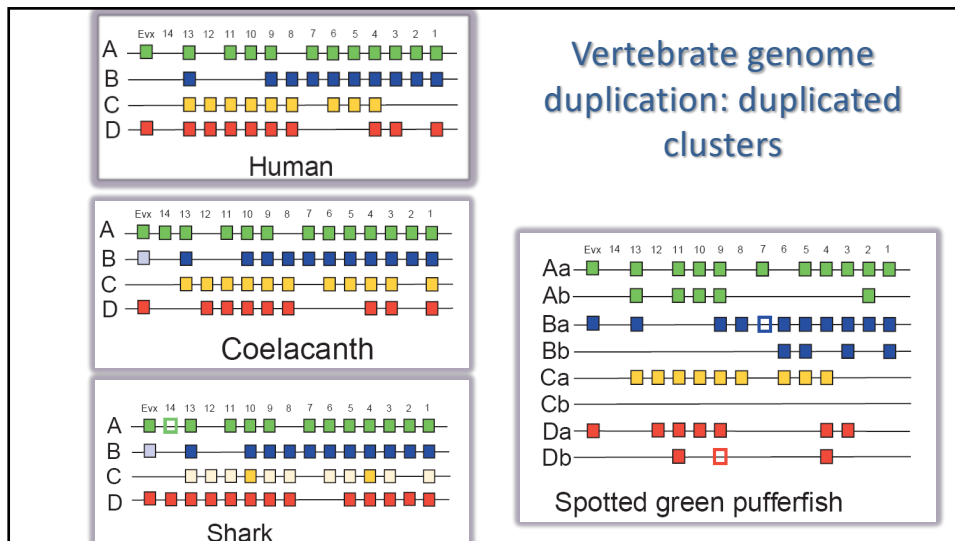
doi:10.1038/nature19640

Genome evolution in the allotetraploid frog *Xenopus laevis*

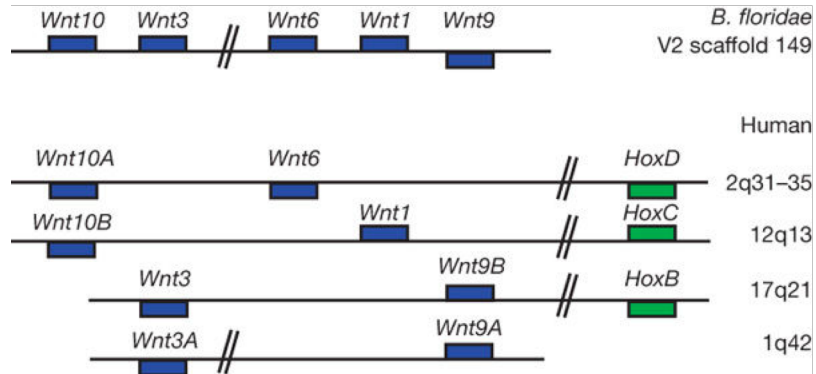
Adam M. Sessing^{1,2*}, Yoshinobu Uno^{3,4}, Taejeon Kwon^{1,5*}, Jarrod A. Chapman⁷, Atsushi Toyoda⁶, Shoji Takahashi⁷, Akimasa Fukui⁸, Akira Hikosaka⁹, Atsushi Suzuki⁷, Mariko Kondo¹⁰, Simon J. van Heeringen¹¹, Ian Quigley¹², Sven Heinz¹³, Hajime Ogino¹⁴, Haruki Ochi¹⁵, Uffe Hellsten⁷, Jessica B. Lyons⁷, Oleg Simakov¹⁶, Nicholas Putnam¹⁷, Jonathan Stites¹⁷, Yoko Kuroki¹⁸, Toshiaki Tanaka¹⁹, Tatsuo Michiue²⁰, Minoru Watanabe²¹, Ozren Bogdanovic²², Ryan Lister²³, Georgios Georgiou¹¹, Saita S. Paranjape¹¹, Ilja van Kruijsbergen¹¹, Shengqiang Shu⁷, Joseph Carlson⁷, Tsutomu Kinoshita²⁴, Yuko Ohta²⁵, Shunji Mawaribuchi²⁶, Jerry Jenkins^{2,26}, Jane Grimwood^{2,26}, Jeremy Schmutz^{2,26}, Therese Mitros¹, Sahar V. Mozaffari²⁷, Yutaka Suzuki²⁸, Yoshikazu Hara²⁹, Takamasa S. Yamamoto³⁰, Chiyo Takagi³⁰, Rebecca Heald³¹, Kelly Miller³¹, Christian Haudenschild³², Jacob Kitzman³³, Takuya Nakayama³⁴, Yumi Iizumi³⁵, Jacques Robert³⁶, Joshua Fortriede³⁷, Kevin Burns³⁸, Vaneet Lotay³⁹, Kanran Karim⁴⁰, Yumi Yasukawa⁴¹, Darwin S. Dichmann⁴², Martin F. Flajnik⁴³, Douglas W. Houston⁴⁴, Jay Shendure⁴⁵, Louis DuPasquier⁴⁶, Peter D. Vize⁴⁶, Aaron M. Zorn⁴⁷, Michihiko Ito⁴², Edward M. Marcotte⁴, John B. Wallingford⁴, Yuzuru Ito⁴⁸, Makoto Asashima⁴⁹, Naoto Ueno^{50,41}, Yoichi Matsuda⁴, Gert Jan C. Veenstra⁴¹, Asao Fujiyama^{4,44,45}, Richard M. Harland⁴, Masanori Taira⁴⁶ & Daniel S. Rokhsar^{1,2,16}

To explore the origins and consequences of tetraploidy in the African clawed frog, we sequenced the *Xenopus laevis* genome and compared it to the related diploid *X. tropicalis* genome. We characterize the allotetraploid origin of *X. laevis* by partitioning its genome into two homoeologous subgenomes, marked by distinct families of 'fossil' transposable elements. On the basis of the activity of these elements and the age of hundreds of unitary pseudogenes, we estimate that the two diploid progenitor species diverged around 34 million years ago (Ma) and combined to form an allotetraploid around 17–18 Ma. More than 56% of all genes were retained in two homoeologous copies. Protein function, gene expression, and the amount of conserved flanking sequence all correlate with retention rates. The subgenomes have evolved asymmetrically, with one chromosome set more often preserving the ancestral state and the other experiencing more gene loss, deletion, rearrangement, and reduced gene expression.

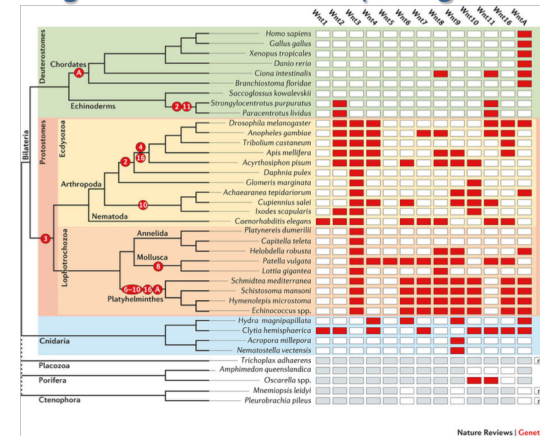




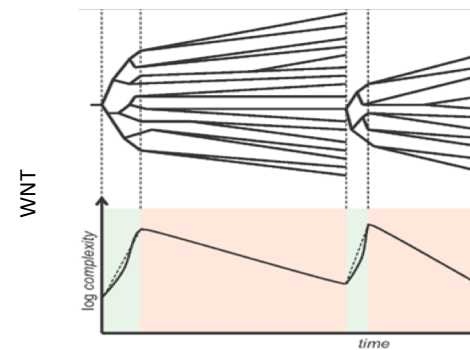
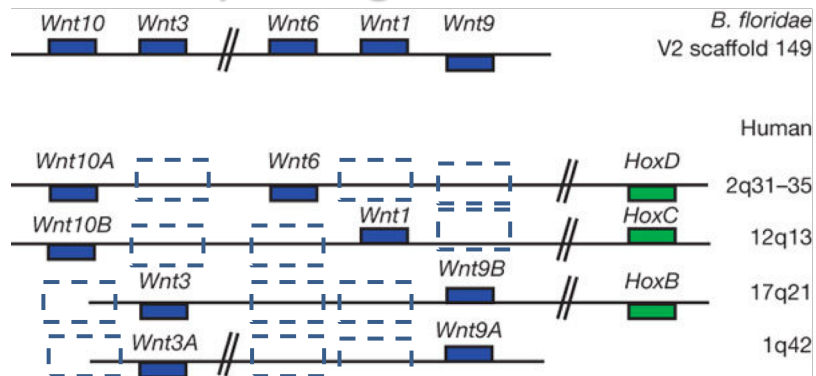
Duplicated wnt “clusters” detected via lancelet outgroup



wingless (Wnt) : a paradigmatic example of the pervasiveness of gene loss during metazoan evolution. (after gene duplication)

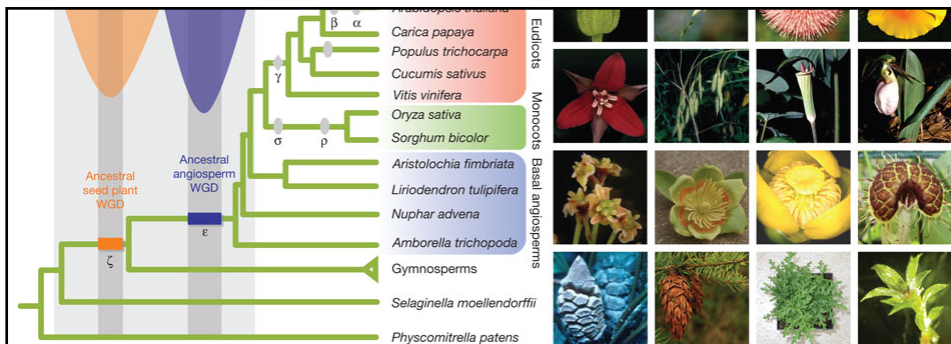


Wnt example of gene loss after WGD

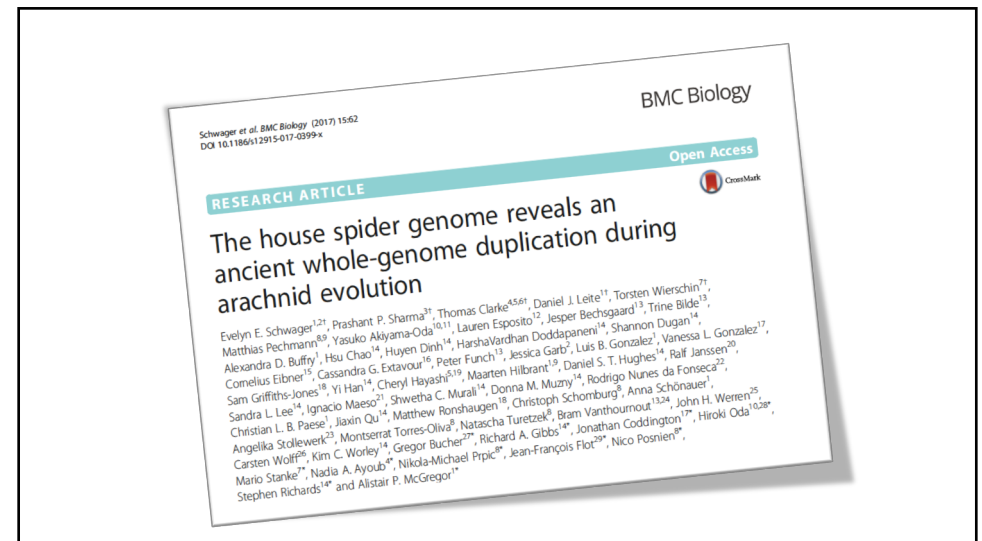


Tandem duplications at base of animals

WGD at base of vertebrates



Nature. 2011 Apr 10. [All from gene trees!!!] **Ancestral polyploidy in seed plants and angiosperms.** [Jiao Y](#), [Wickett NJ](#), [Ayyampalayam S](#), [Chanderbali AS](#), [Landherr L](#), [Ralph PE](#), [Tomsho LP](#), [Hu Y](#), [Liang H](#), [Soltis PS](#), [Soltis DE](#), [Clifton SW](#), [Schlarbaum SE](#), [Schuster SC](#), [Ma H](#), [Leebens-Mack J](#), [Depamphilis CW](#).



Is it always genome duplication?

Martens and Van de Peer *BMC Genomics* 2010, 11:353
http://www.biomedcentral.com/1471-2164/11/353

BMC Genomics
RESEARCH ARTICLE Open Access

The hidden duplication past of the plant pathogen *Phytophthora* and its consequences for infection

Cindy Martens^{1,2} and Yves Van de Peer^{1,2}

Abstract
Background: Oomycetes of the genus *Phytophthora* are pathogens that infect a wide range of plant species. For dicot hosts such as tomato, potato and soybean, *Phytophthora* is even the most important pathogen. Previous analyses of *Phytophthora* genomes uncovered many genes, large gene families and large genome sizes that can partially be explained by significant repeat expansion patterns.
Results: Analysis of the complete genomes of three different *Phytophthora* species, using a newly developed approach, unveiled a large number of small duplicated blocks, mainly consisting of two or three consecutive genes. Further analysis of these duplicated genes and comparison with the known gene and genome duplication history of ten other eukaryotes including parasites, algae, plants, fungi, vertebrates and invertebrates, suggests that the ancestor of *P. infestans*, *P. sojae* and *P. ramorum* most likely underwent a whole genome duplication (WGD). Genes that have survived in duplicate are mainly genes that are known to be preferentially retained following WGDs, but also genes important for pathogenicity and infection of the different hosts seem to have been retained in excess. As a result, the WGD might have contributed to the evolutionary and pathogenic success of *Phytophthora*.
Conclusions: The fact that we find many small blocks of duplicated genes indicates that the genomes of *Phytophthora* species have been heavily rearranged following the WGD. Most likely, the high repeat content in these genomes have played an important role in this rearrangement process. As a consequence, the paucity of retained larger duplicated blocks has greatly complicated previous attempts to detect remnants of a large-scale duplication event in *Phytophthora*. However, as we show here, our newly developed strategy to identify very small duplicated blocks might

GBE

Small Homologous Blocks in *Phytophthora* Genomes Do Not Point to an Ancient Whole-Genome Duplication

Jolien J.E. van Hooff^{1,4}, Berend Snel^{1,2}, and Michael F. Seidl^{1,2,3,*}

¹Theoretical Biology and Bioinformatics, Department of Biology, Utrecht University, The Netherlands
²Centre for BioSystems Genomics, Wageningen, The Netherlands
³Laboratory of Phytopathology, Wageningen University, Wageningen, The Netherlands
⁴Present address: Department of Molecular Cancer Research and Cancer Genomics Centre, University Medical Center Utrecht, Utrecht, The Netherlands

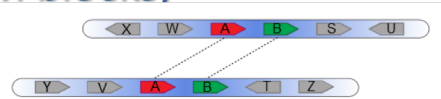
*Corresponding author: E-mail: michael.seidl@wur.nl

Accepted: April 15, 2014

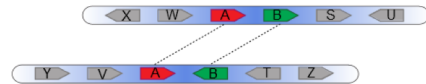
Abstract
Genomes of the plant-pathogenic genus *Phytophthora* are characterized by small duplicated blocks consisting of two consecutive genes (2HOM blocks) and by an elevated abundance of similarly aged gene duplicates. Both properties, in particular the presence of

Phytophthoras have many paralogous blocks (HOM blocks)

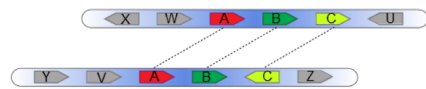
2HOM block with conserved orientation



2HOM block with unconserved orientation



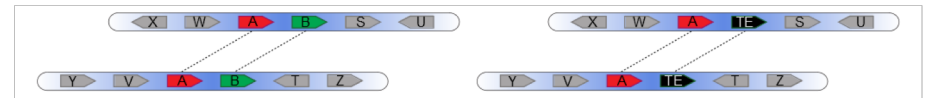
2HOM block being part of a 3HOM block



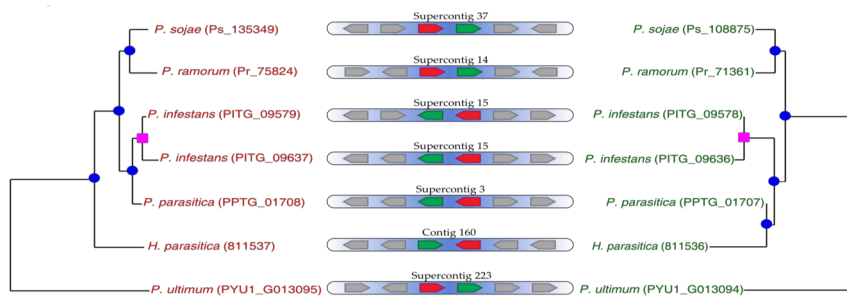
Adapted from: C Martens and Y van de Peer BMC Genomics (2010)

HOM block features do not point to a WGD

Species	2HOM blocks	TE-filtered			
		Full set	> 2 copies	Same scaffold	2 copies, different scaffolds
<i>P. infestans</i>	253	86	14	25	55
<i>P. ramorum</i>	128	31	1	16	15
<i>P. sojae</i>	207	35	4	15	18



majority originated from recent species-specific duplications

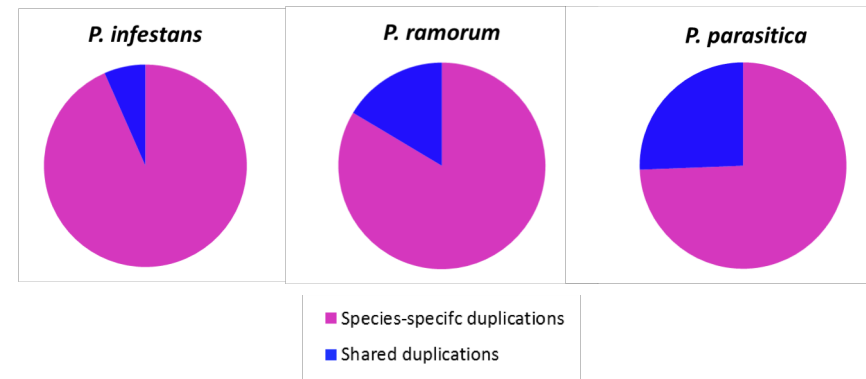


Family of CMGC/CDK protein kinases and riboflavin biosynthesis proteins

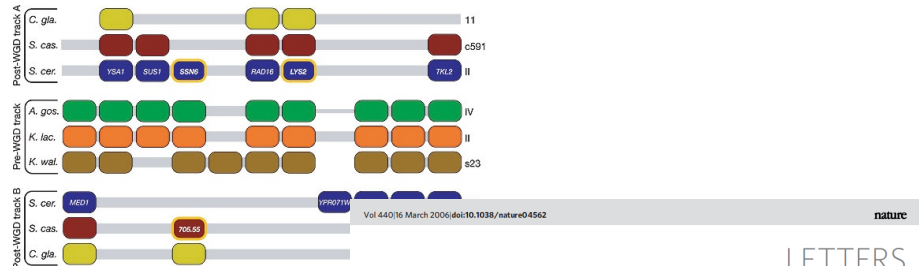
Family of vesicle transporters

E.g: in *P. infestans*, 132 out of 146 (90%) blocks analysed is species-specific, so cannot result from a WGD in the *Phytophthora* ancestor

Individual duplications are even more often species-specific



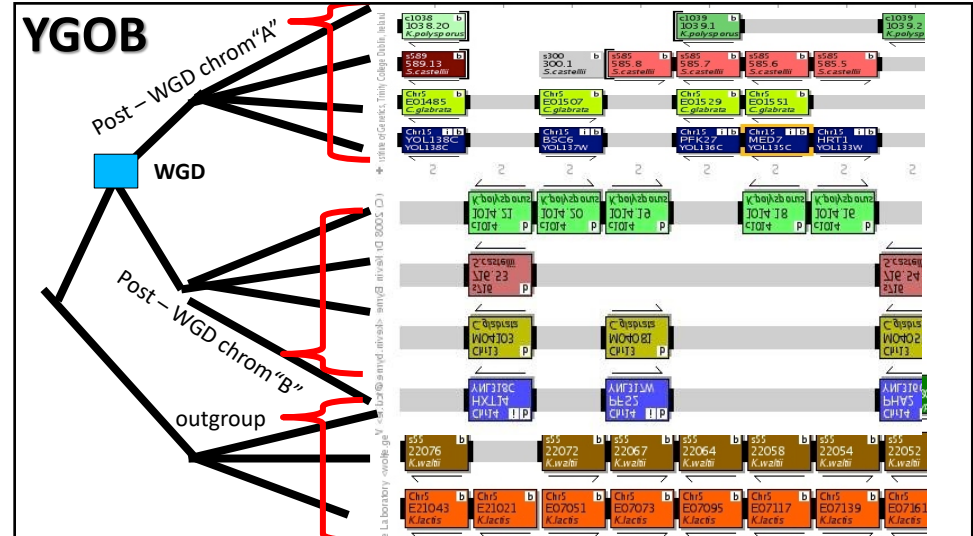
Reconstructed map of genome duplications allows unprecedented mapping of evolutionary history of genes in genomes



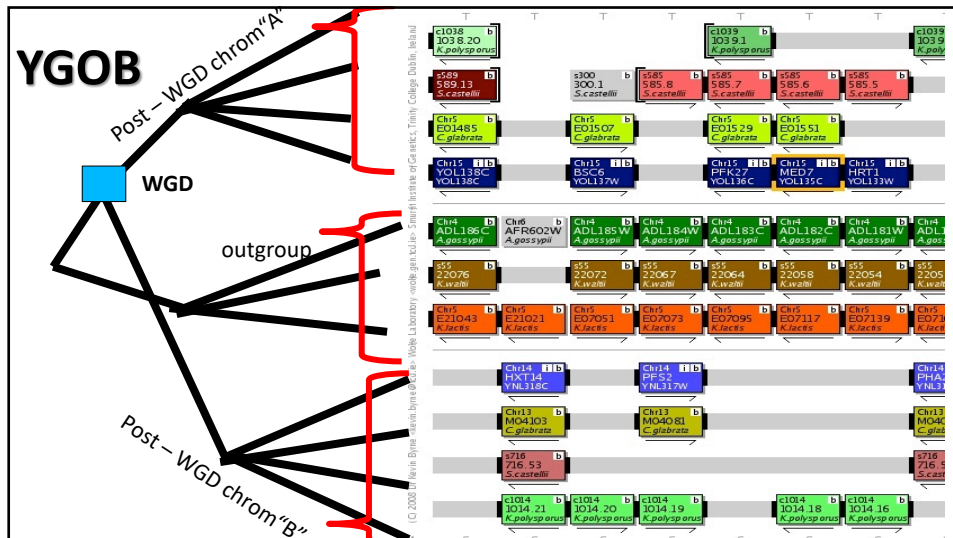
Multiple rounds of speciation associated with reciprocal gene loss in polyloid yeasts

Devin R. Scannell¹*, Kevin P. Byrne¹*, Jonathan L. Gordon¹, Simon Wong¹ & Kenneth H. Wolfe¹

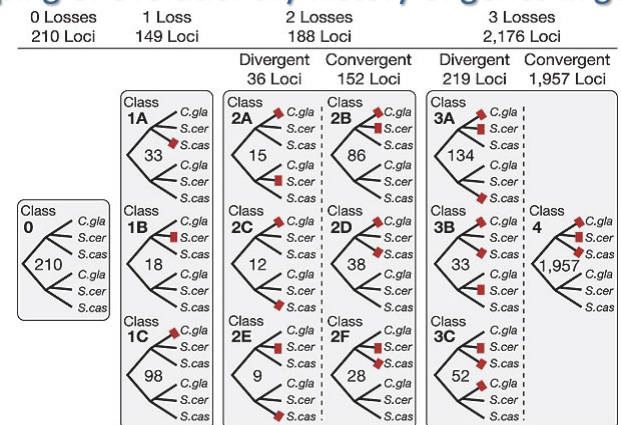
YGOB



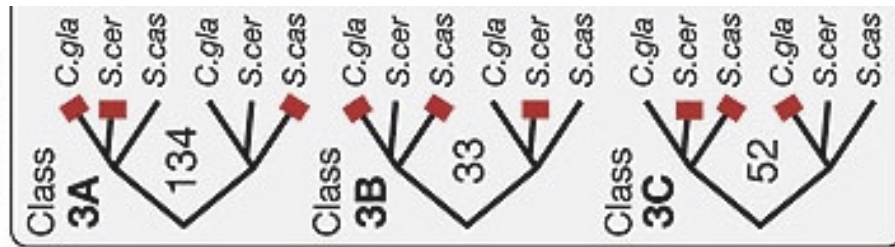
YGOB



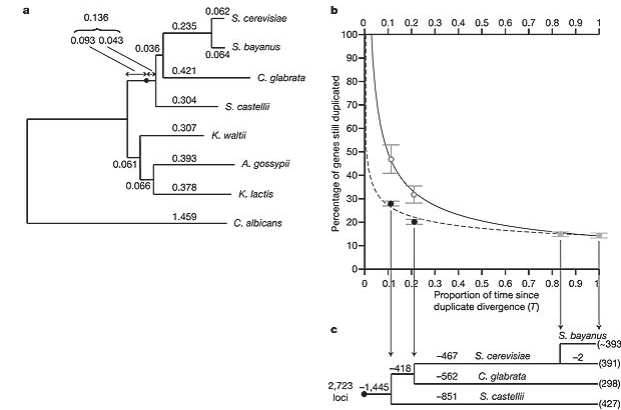
Reconstruction of genome duplications allows unprecedented mapping of evolutionary history of genes in genomes



Reconstructed map of genome duplications allows unprecedented mapping of evolutionary history of genes in genomes: “pseudo-orthology”



Major fate is gene loss



Paramecium genome duplications

Vol 444 | 9 November 2006 | doi:10.1038/nature05230

nature

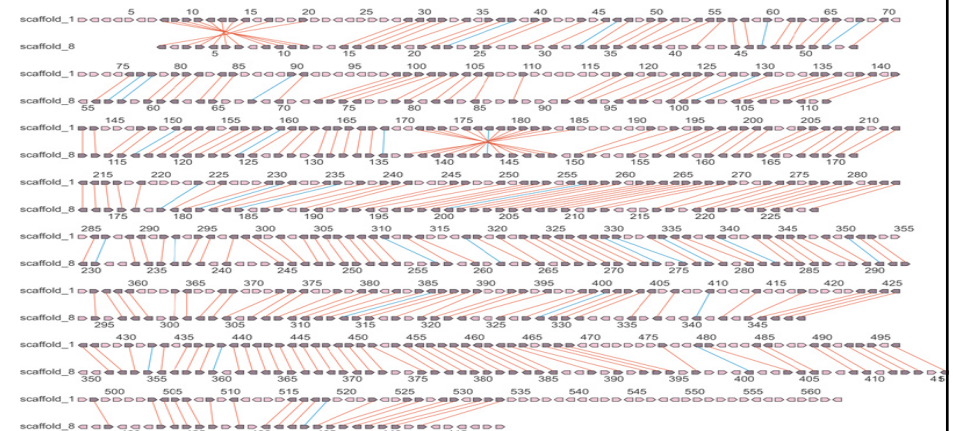
ARTICLES

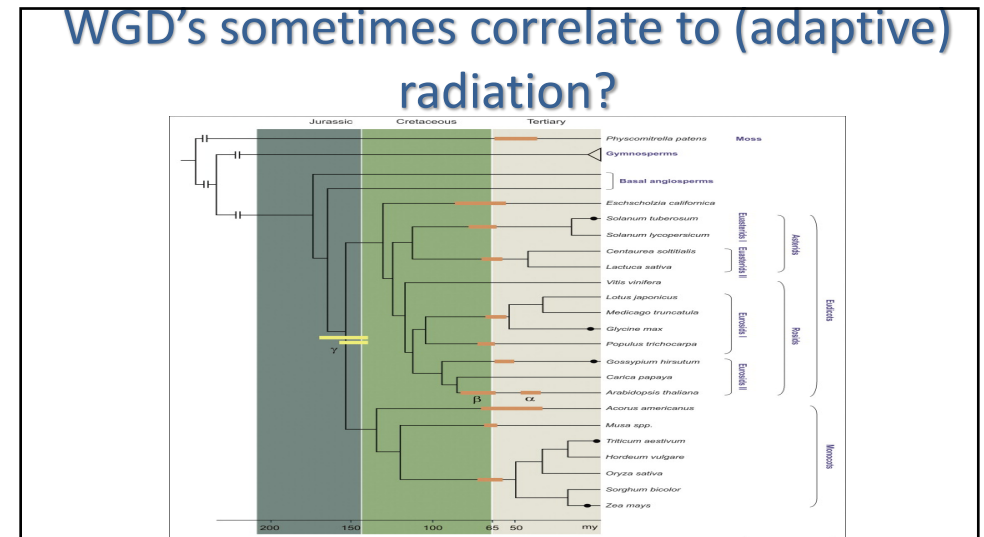
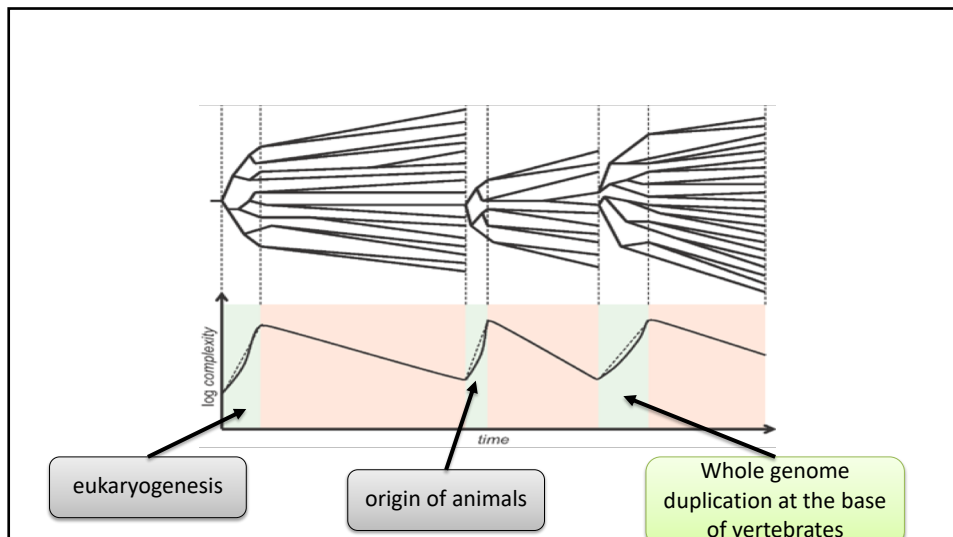
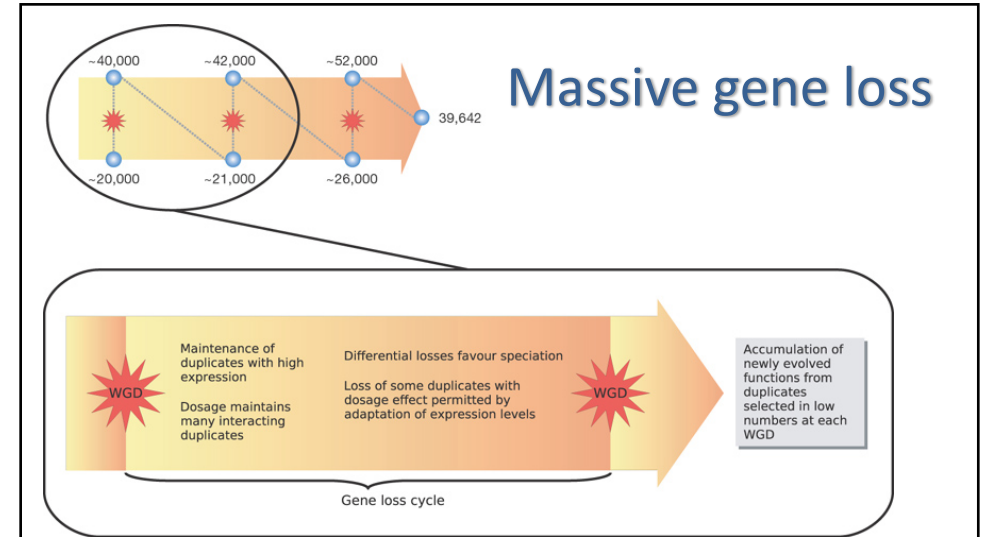
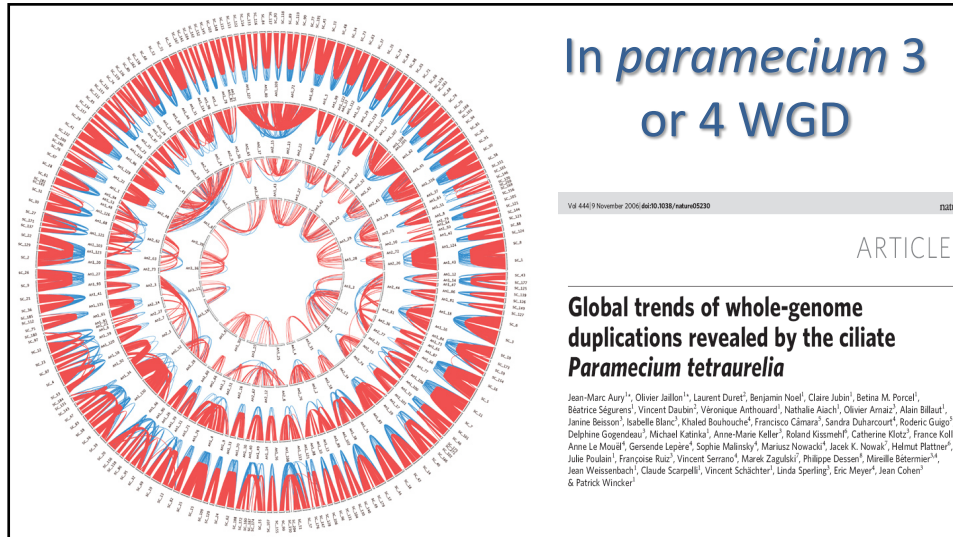
Global trends of whole-genome duplications revealed by the ciliate *Paramecium tetraurelia*

Jean-Marc Aury^{1,2}, Olivier Jaillon^{1,2}, Laurent Duret², Benjamin Noel¹, Claire Jubin¹, Betina M. Porcel¹, Béatrice Ségurens¹, Vincent Daubin², Véronique Anthouard¹, Nathalie Aiach¹, Olivier Arnaiz¹, Alain Billaut¹, Janine Beisson³, Isabelle Blanc³, Khaled Bouhouche³, Francisco Cámara³, Sandra Duhaucourt¹, Roderic Guigo³, Delphine Gogendeau³, Michael Katinka³, Anne-Marie Keller³, Roland Kissmehl³, Catherine Klotz³, France Koll¹, Anne Le Mouél³, Gersende Lepère³, Sophie Malinsky³, Mariusz Nowacki³, Jacek K. Nowak³, Helmut Plattner³, Julie Poulain³, Françoise Ruiz³, Vincent Serrano³, Marek Zagulski³, Philippe Dessen³, Mireille Bétermier^{3,4}, Jean Weissenbach¹, Claude Scarpelli¹, Vincent Schächter¹, Linda Sperling³, Eric Meyer³, Jean Cohen³ & Patrick Wincker¹

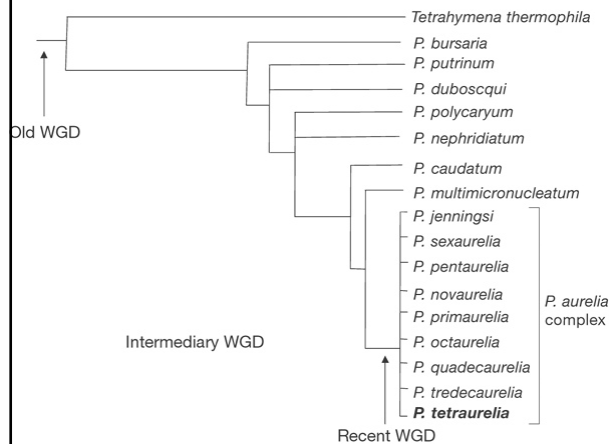


Comparison of two scaffolds originating from a common ancestor at the recent WGD



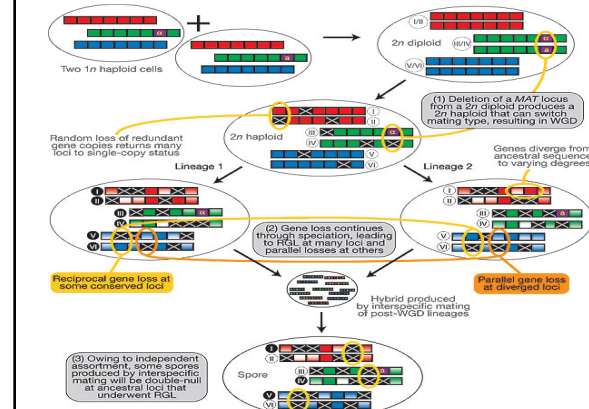


Correlation to (adaptive) radiation?

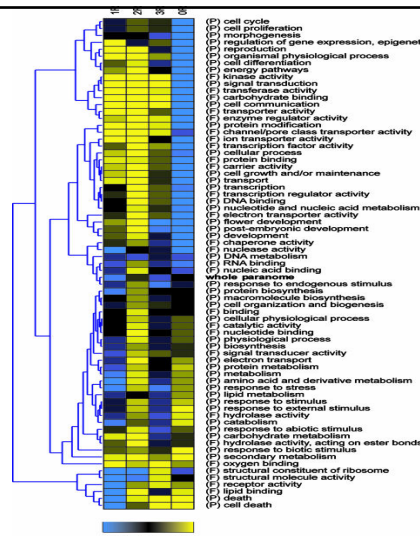
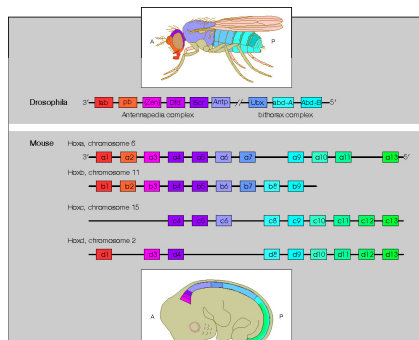


- Vertebrates
- Flowering plants
- Teleosts most species rich group of vertebrates
- Yeasts

Explanations for adaptive radiation? Incompatibility at essential loci



Explanations for adaptive radiation? Regulatory innovations



Non-sequitur? How this lecture fits in the course.

- Something to expect in gene trees.
 - If I see two paralogs in human my first guess is that they duplicated at base of vertebrates: provides scenario/search image
- *We want to detective What* has happened, this is generally difficult: here we are clear what happened because of synteny
- Some fringe / *in extremis* discussion of paralogy & orthology
- Much more well defined exposition of
 - Again importance of gene loss: provides nice example of expansion contraction / biphasic model of genome evolution
 - Changes in Speed of evolution