

# Genome evolution: extinction, continuation or explosion

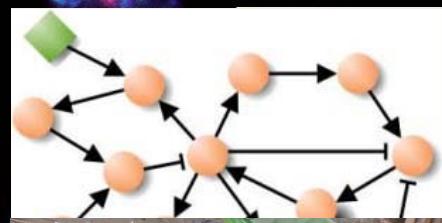
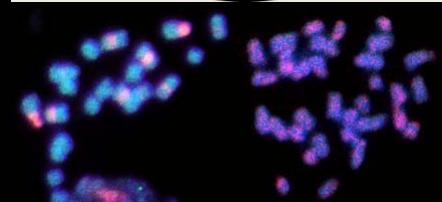
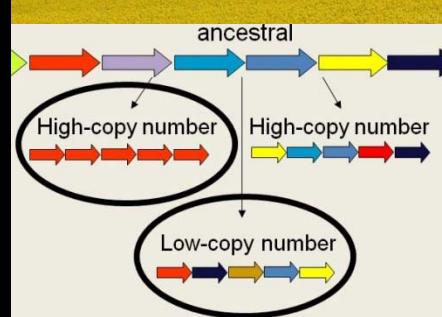
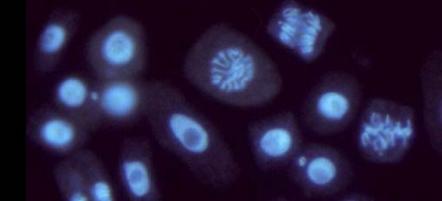
Pat Heslop-Harrison

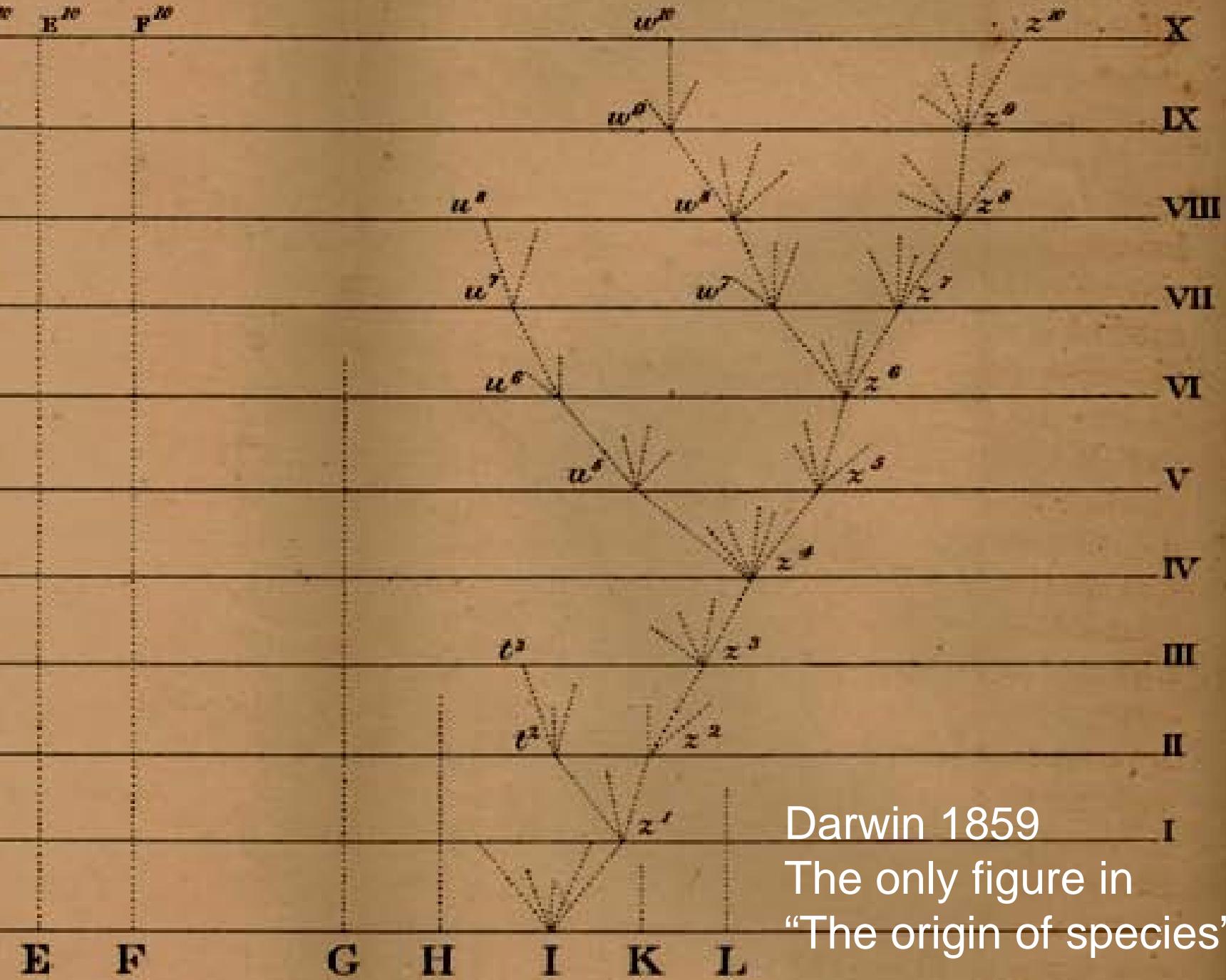
phh4@le.ac.uk

[www.molcyt.com](http://www.molcyt.com) pw/user: 'visitor'

Social media

Follow #PGE11 and Pathh1 on Twitter and  
[AoBBlog.com](http://AoBBlog.com)





# *Arabidopsis*

150 Mbp

# Wheat

17000 Mbp (x113 Arab.)

# Human

3300 Mbp (x22 *Arabidopsis*)

1 mid-sized human chromosome =

Arabidopsis genome

5 average wheat chromosomes =

Human genome



*Arabidopsis*

Wheat

Repetitive DNA is a major part of the genome

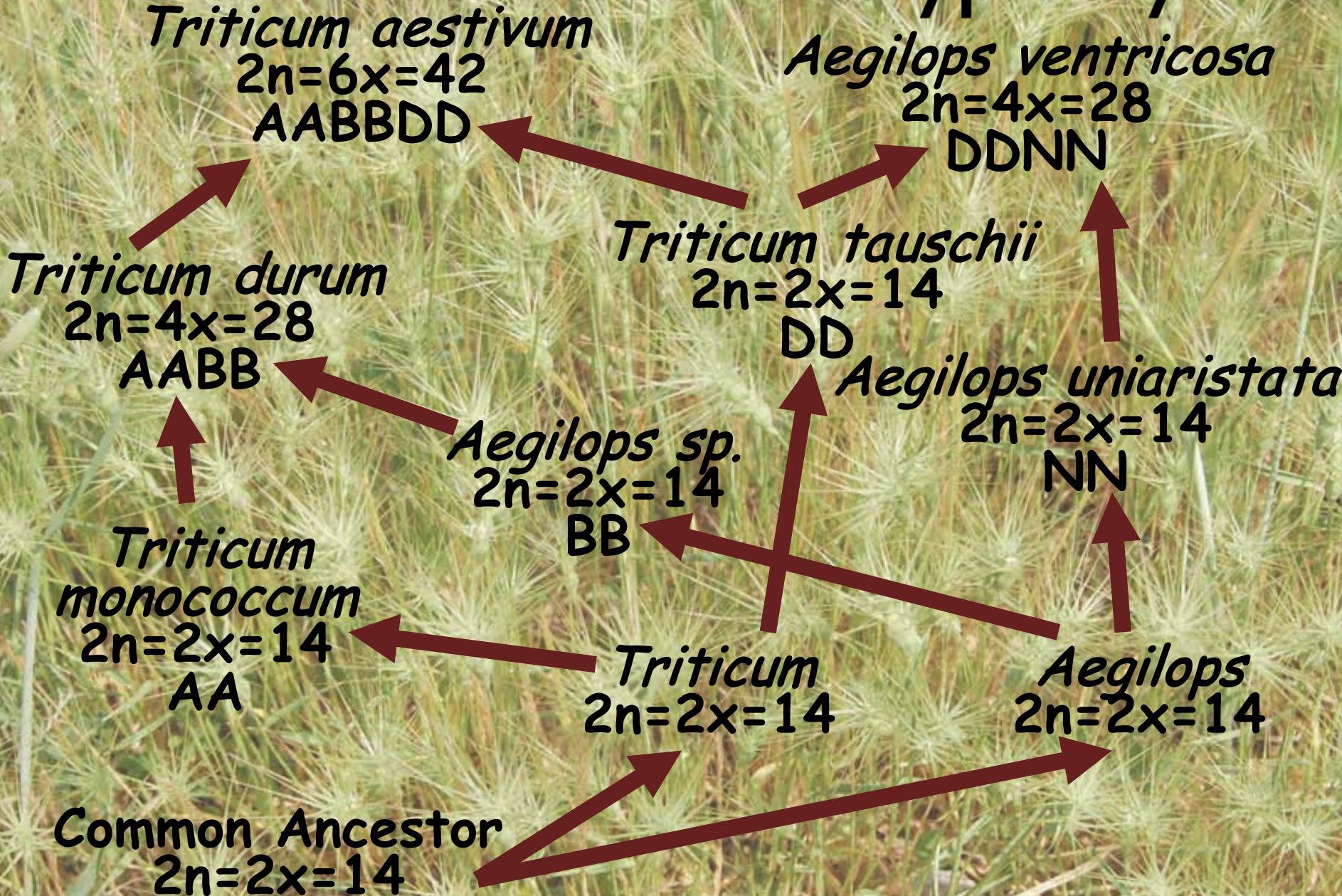
Localised sites – often tandem repeats

Dispersed widely – often transposable elements

Different families of these repeats have different evolutionary histories and consequences

Molecular cytogenetics, comparative genomics and sequence-based studies of species and hybrids

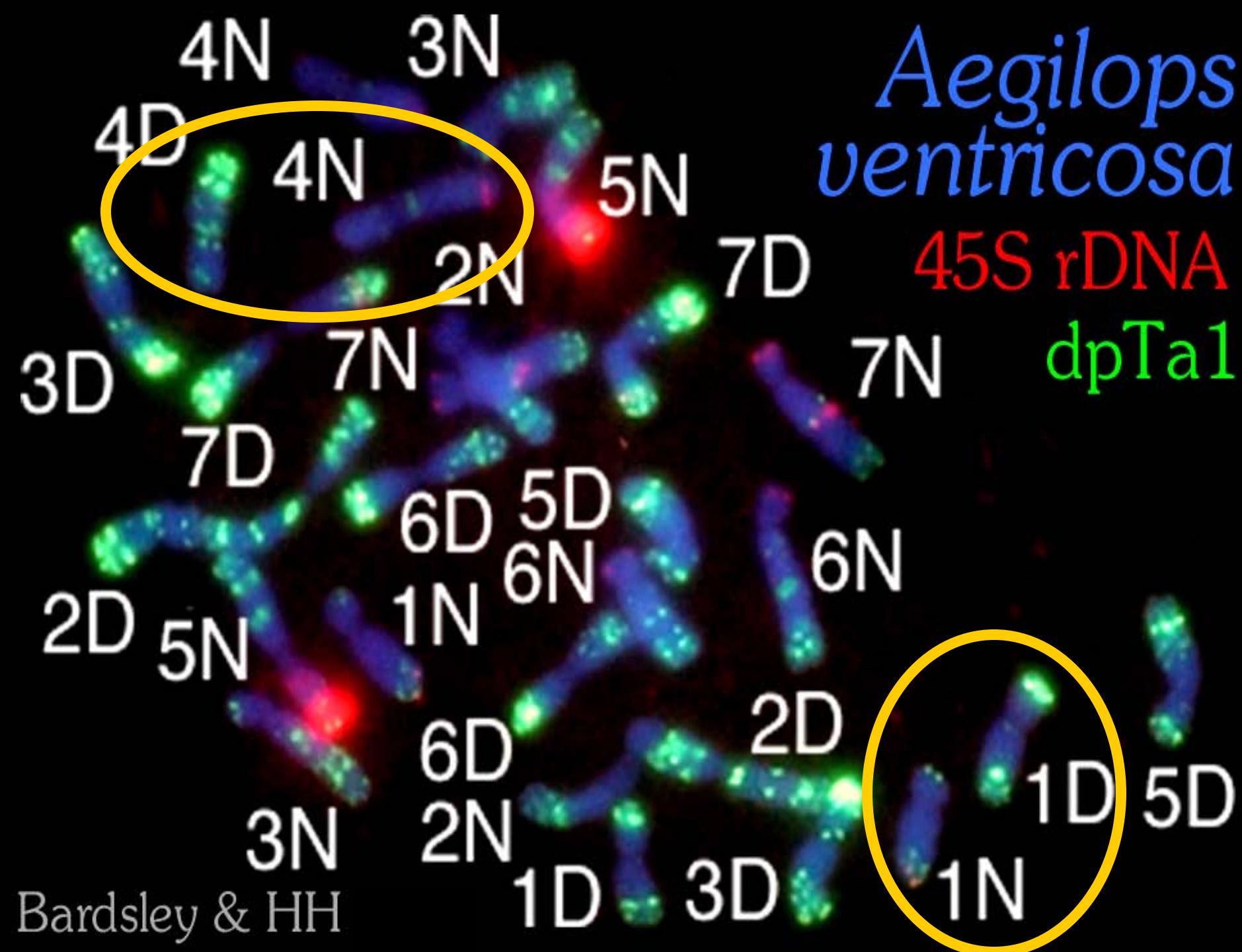
# Evolution of Wheats - Polyploidy



*Aegilops  
ventricosa*

45S rDNA

dpTa1





# Tandem Repeats

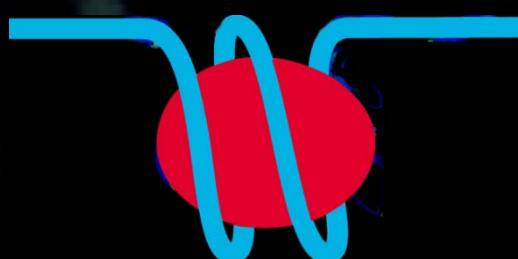


Where each arrow → is a single repeat unit

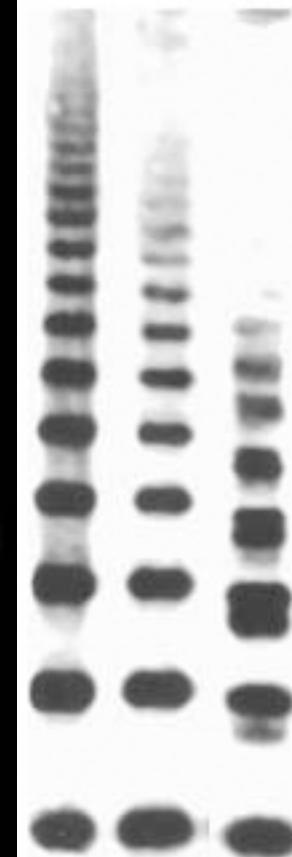
- often a multiple of 180 bp but up to 10kb long

## Head-to-tail organization

- CGACTAGT CGACTAGT  
CGACTAGT GGACTAGT  
CGACTAGT CGACTAGT



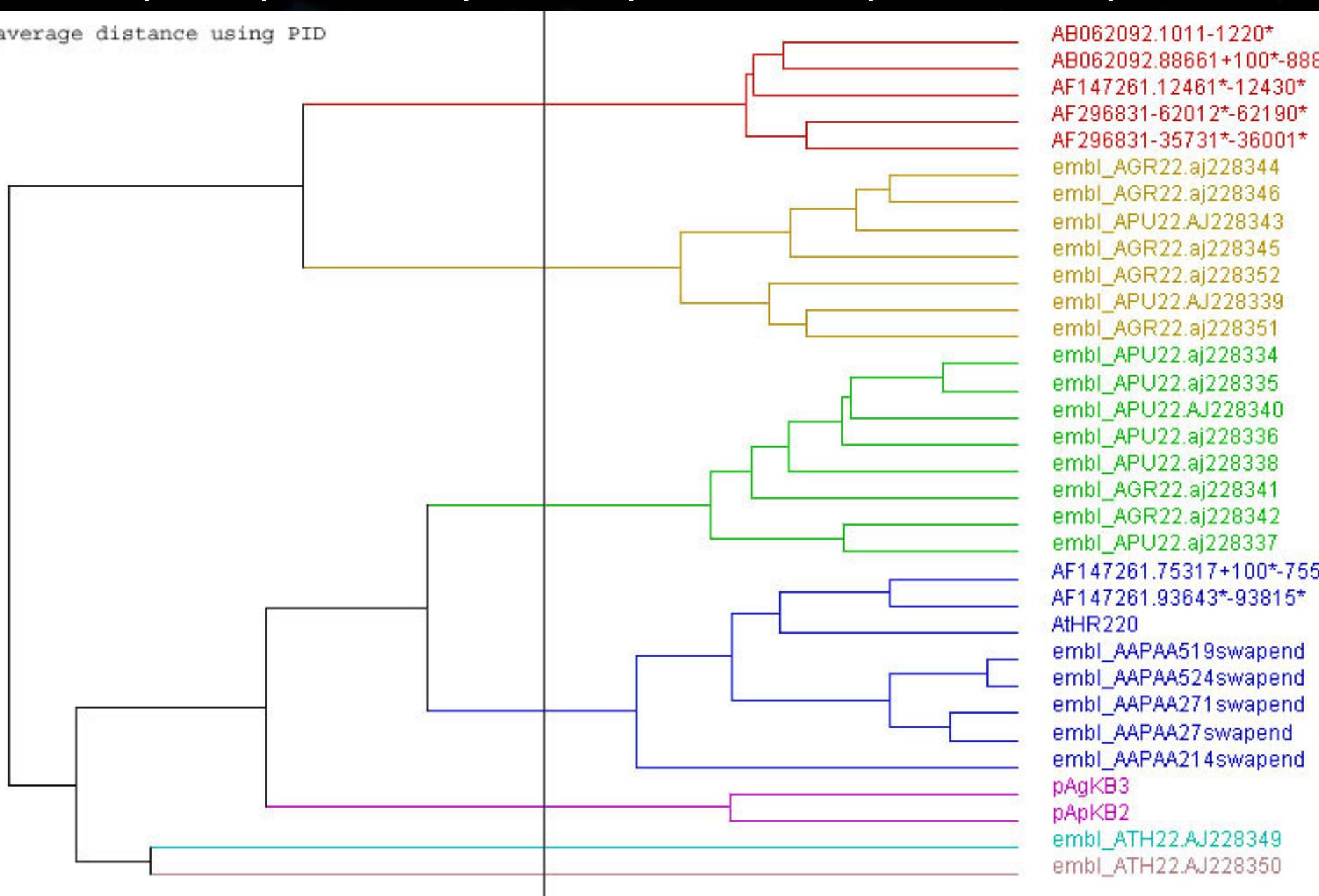
a b g



Crocus spp Probed pCvKB8 Frello, HH 2004

# *Arabidopsis* species – species specific 178bp tandem repeat motif

average distance using PID



# *Arabidopsis suecica*

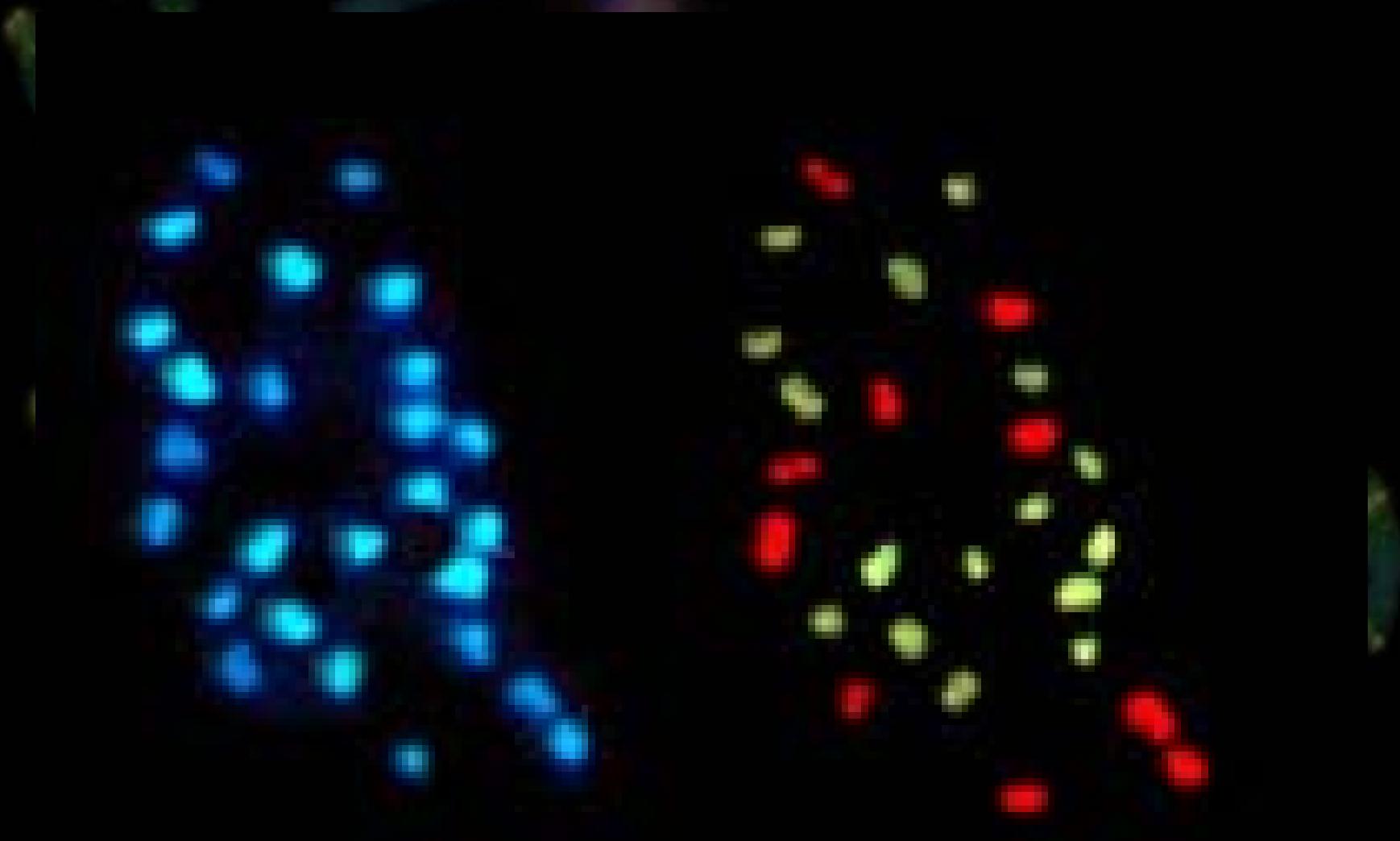
Hybrid of *A. arenosa* (pAa214 green)  
and *A. thaliana* (18obp red)

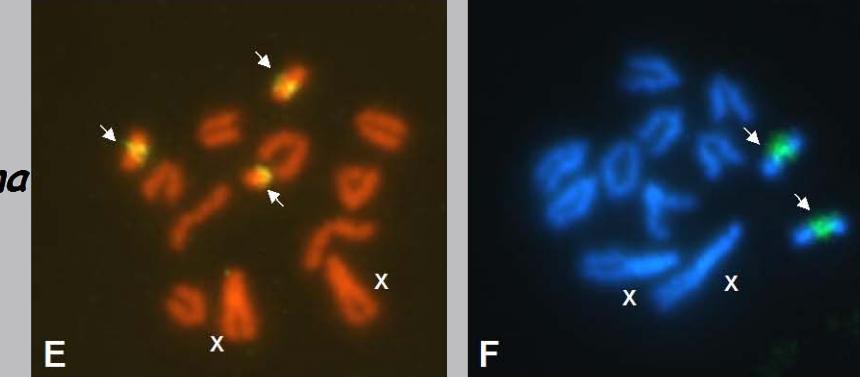
2n = 26

10 from *A. thaliana* 2n=2x=10

16 from *A. arenosa* 2n=2x=16

Kamm, HH et al.





*D. seriema*

High copy species

Low copy species

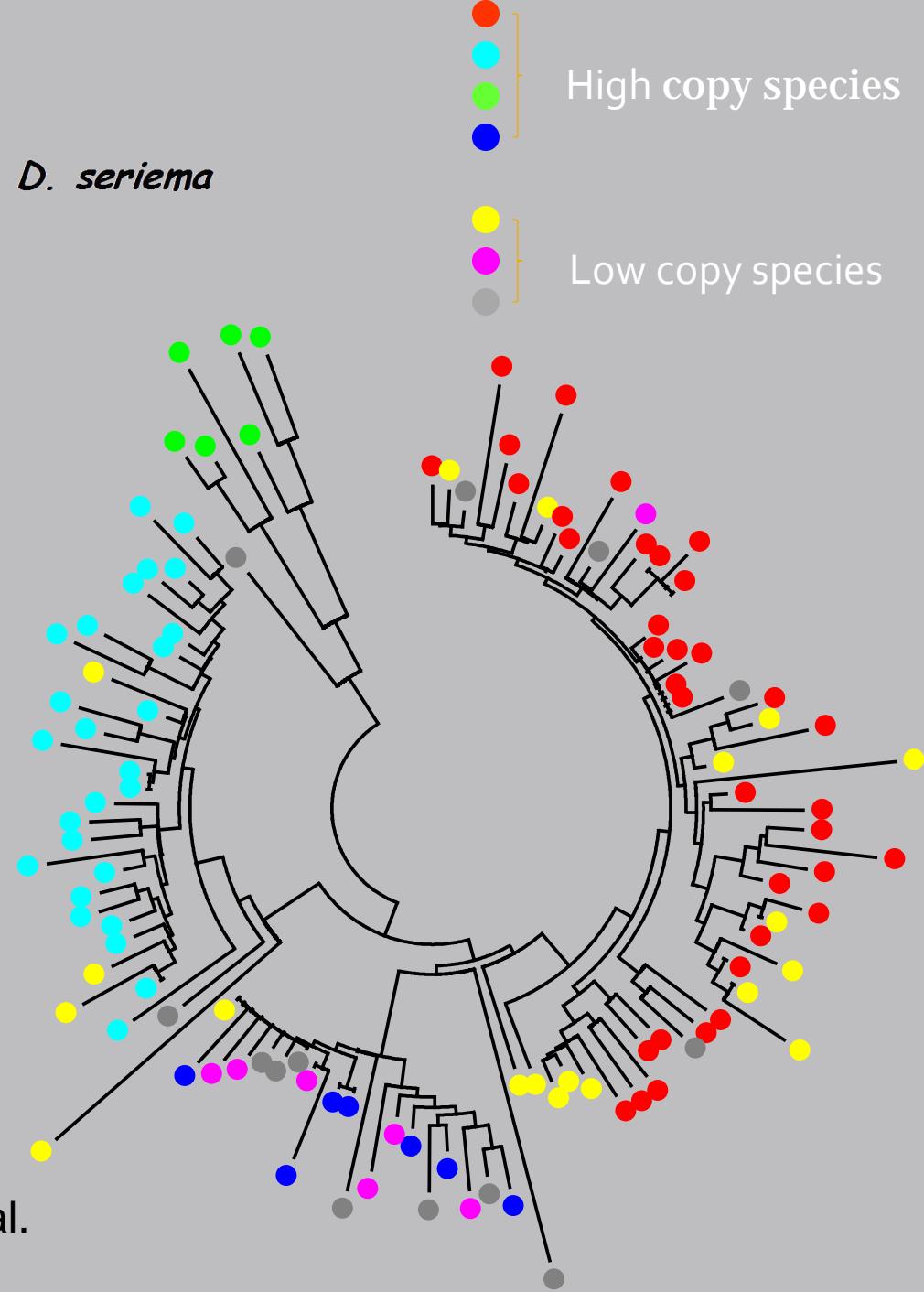
Dr119 repeats:  
showing  
homogenization and  
amplification of  
satellite tandem  
repeats

*Drosophila* spp: Kuhn, HH et al.

Mol Biol Evol 2011

Mol Gen Genet 2010

Chr Res 2008

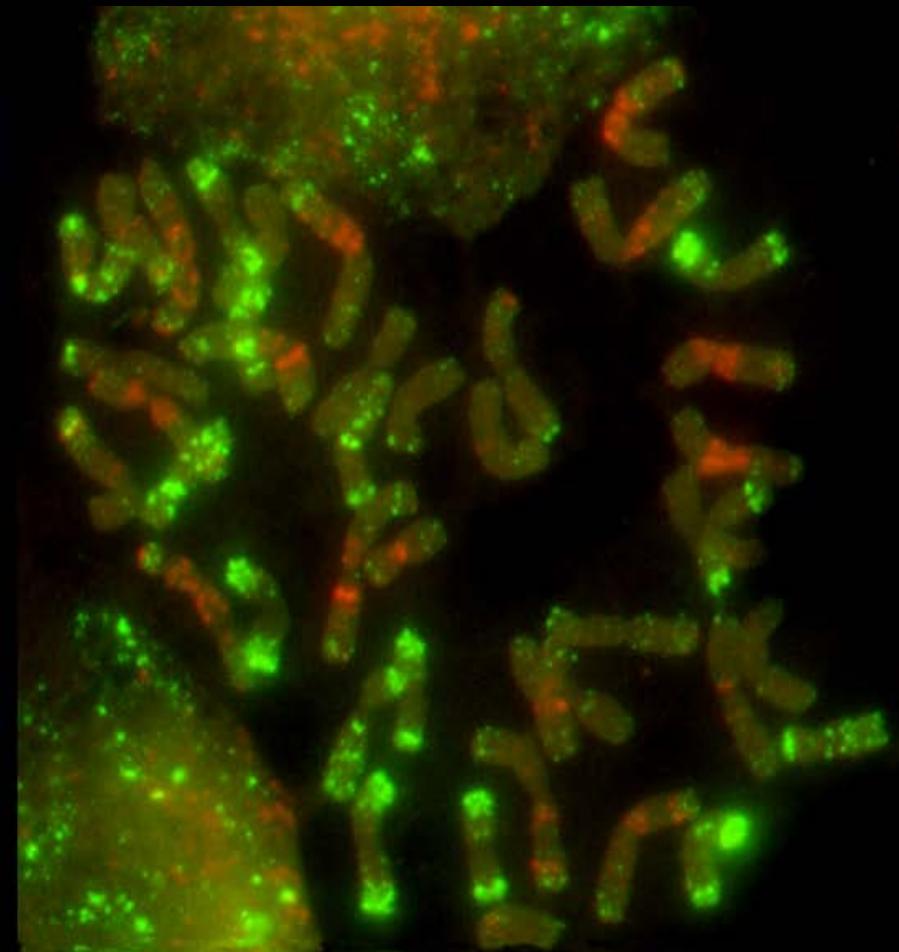


*Medicago*: watch this space: Mondin et al.

*Triticum aestivum*     $2n=6x=42$



DAPI



pSc119.2    dpTa1

High copy number – low diversity in each of 3 genomes

# Inheritance of Chromosome 5D

*Aegilops ventricosa*  
DDNN



$\times$  *Triticum persicum* Ac.1510  
AABB

ABDN

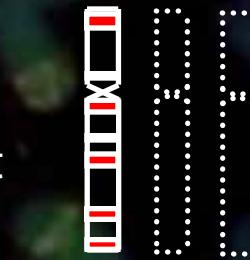
AABBDDNN



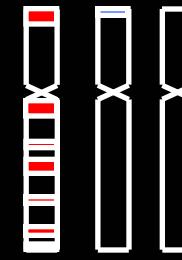
$\times$  Marne  
AABBDD



VPM1  $\times$  Hobbit

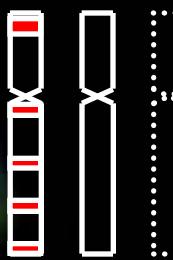


Dwarf A



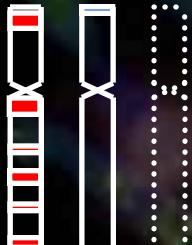
CWW1176-4

$\times$  Virtue



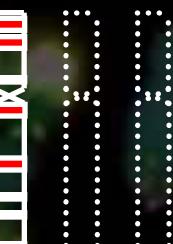
Rendezvous

$\times$  {Kraka  $\times$  (Huntsman  $\times$  Fruhgold)}



Piko

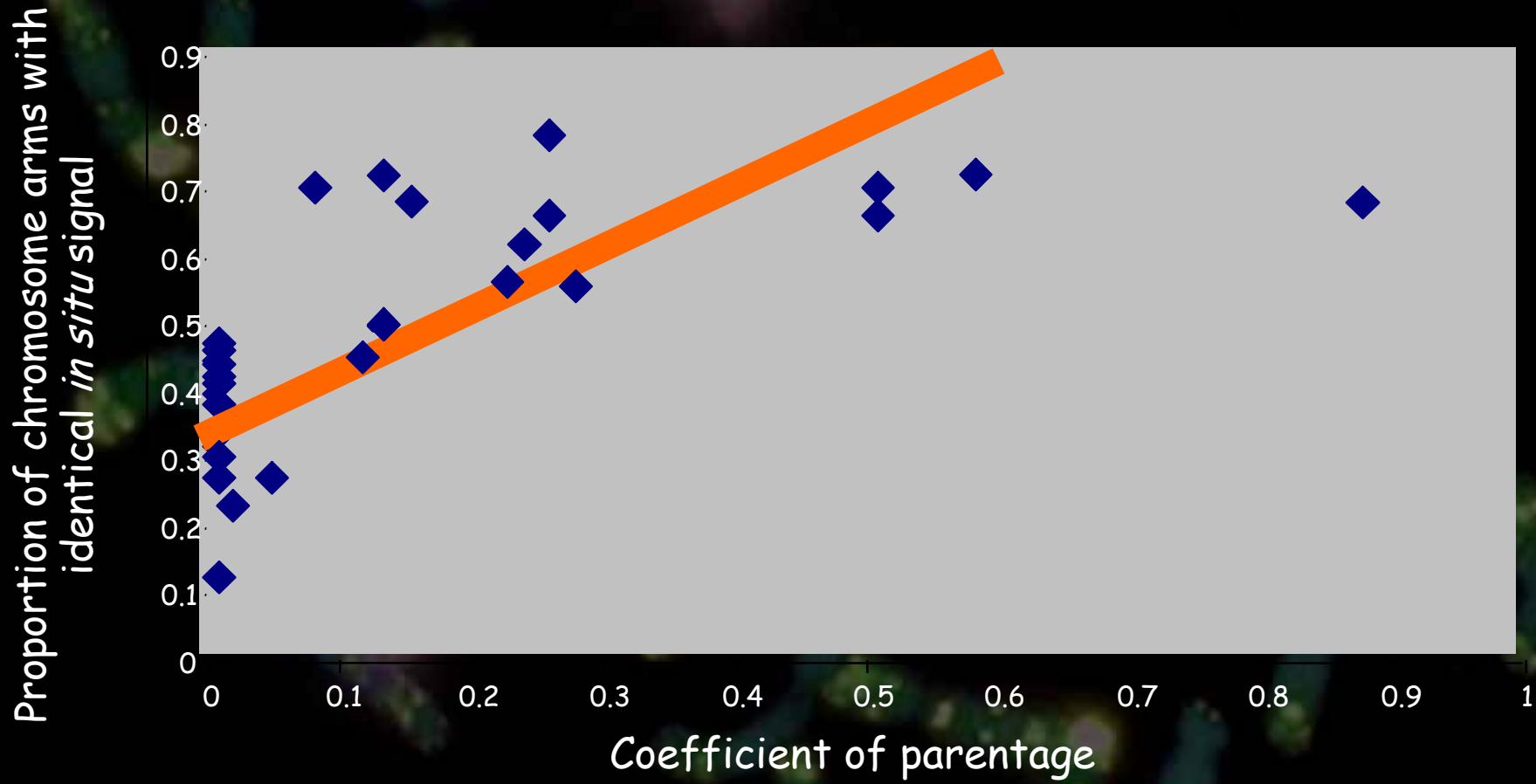
96ST61



dpTa1  
pSc119.2

Genomic Ae. ventricosa

# Correlation between genetic relationships and similarity of dpTa1 hybridization



- Eyespot (fungus *Pseudocercospora*) resistance from *Aegilops ventricosa* introduced to wheat by chromosome engineering
- Many diseases where *all* wheat varieties are highly susceptible



An aerial photograph of a agricultural field. The field is divided into three distinct sections: a dark green section on the left, a bright green section in the center, and a light brown section on the right. The boundary between the dark green and bright green sections is a sharp, straight line. The boundary between the bright green and light brown sections is a curved line. In the dark green section, there are several bright yellow spots. A red arrow points from the text "Crop standing" to the dark green section. A red line with an arrow points from the text "Lodging in cereals" to the curved boundary between the bright green and light brown sections.

Crop standing

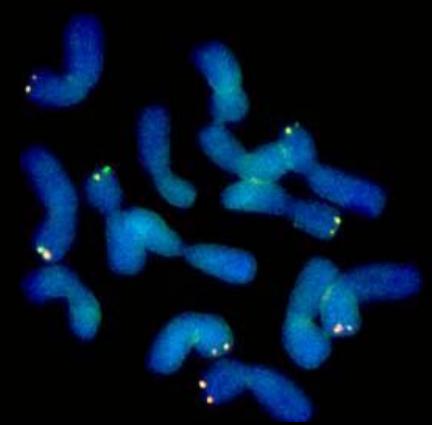
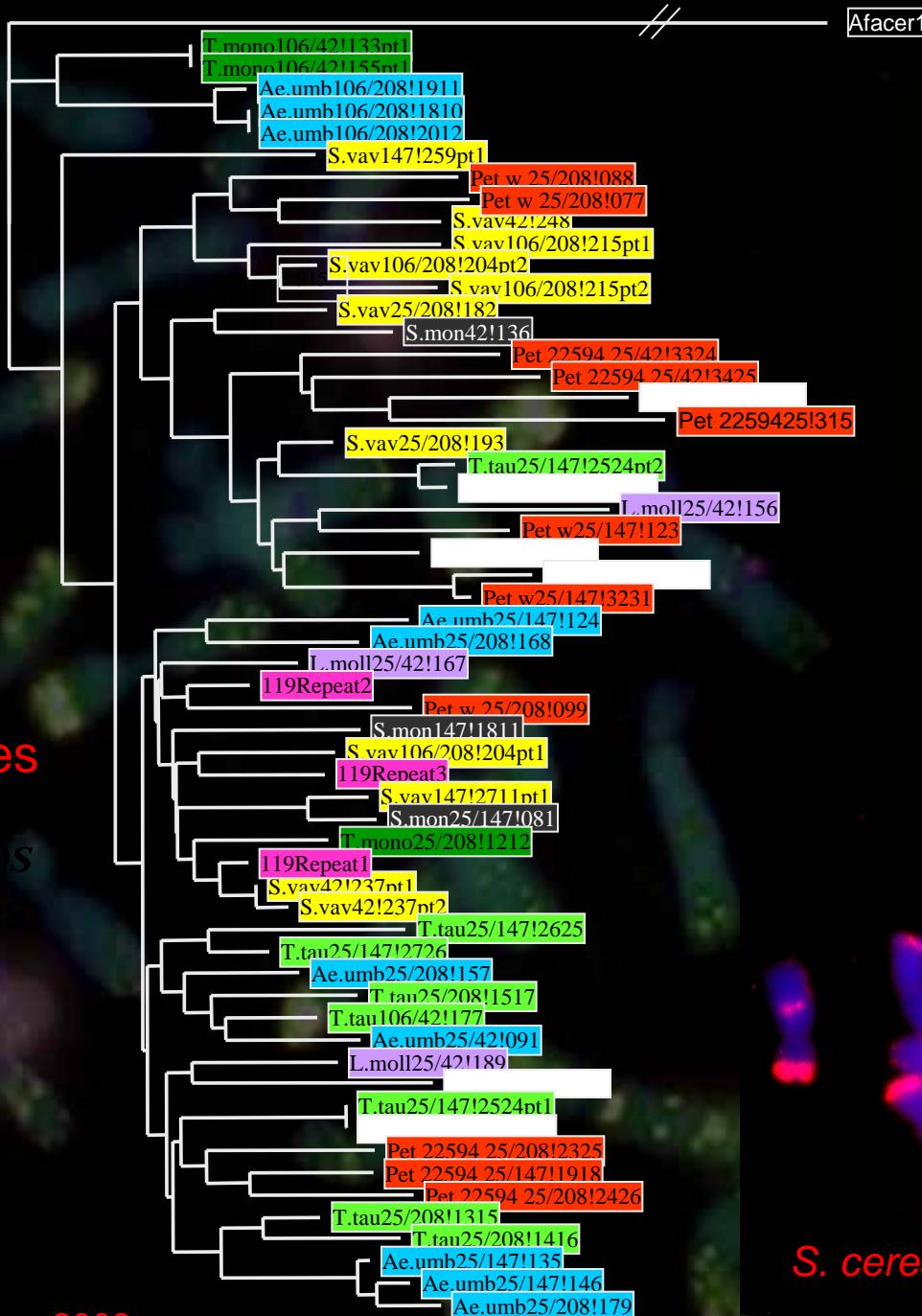
Lodging in cereals

Crop fallen

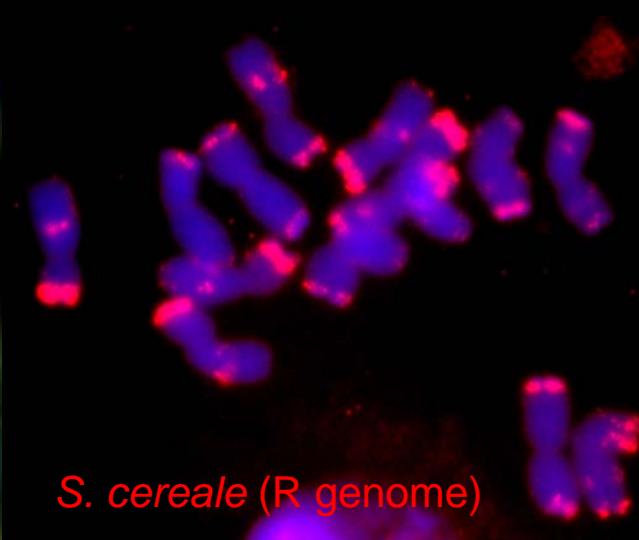
## 120bp repeat unit family

in *Triticum*,  
*Aegilops* and  
*Secale* species

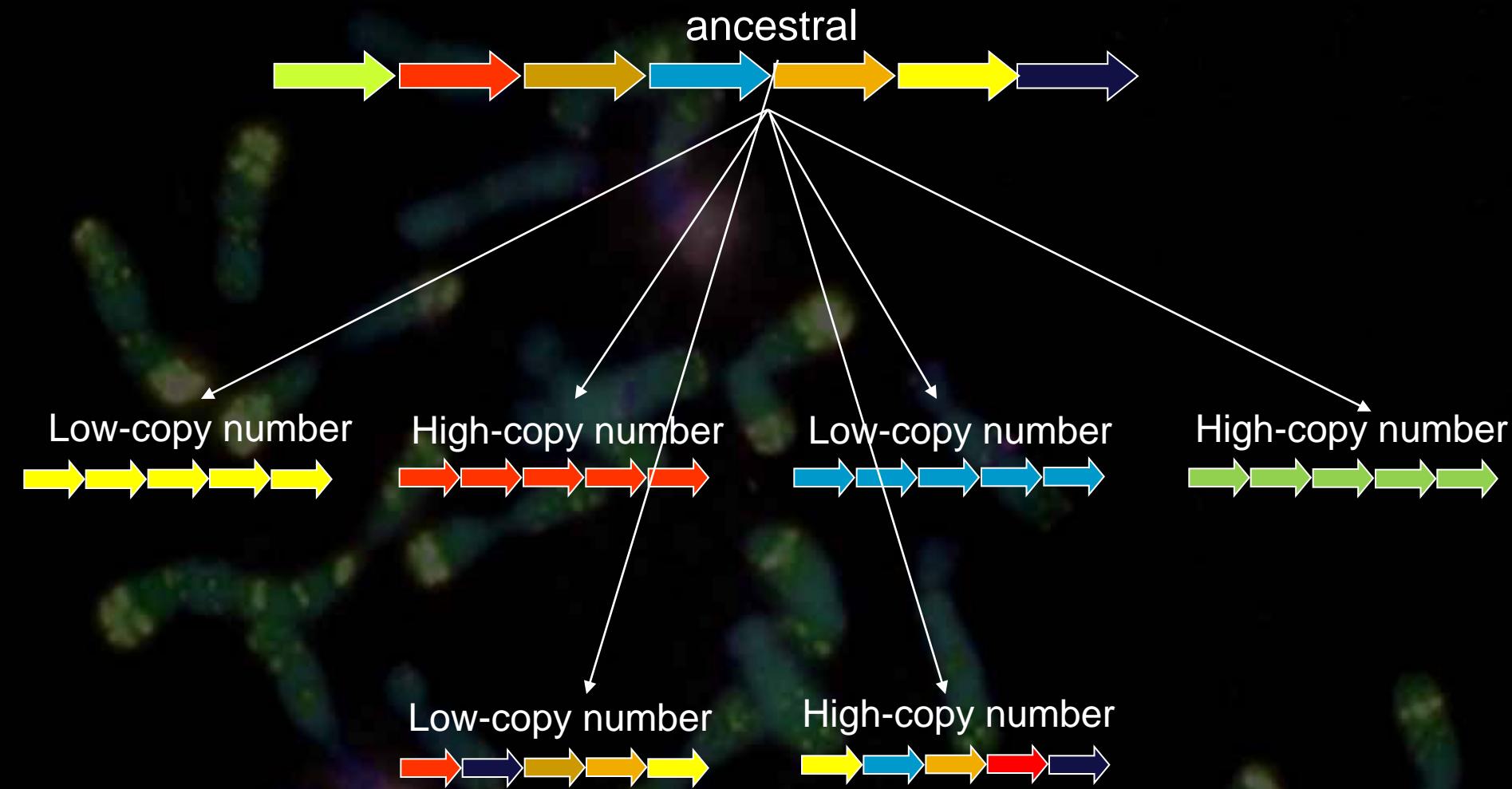
Colour blocks  
represent species  
70-100% in  
egress  
High copy,  
High diversity



*T.tauschii* (D genome)



*S. cereale* (R genome)

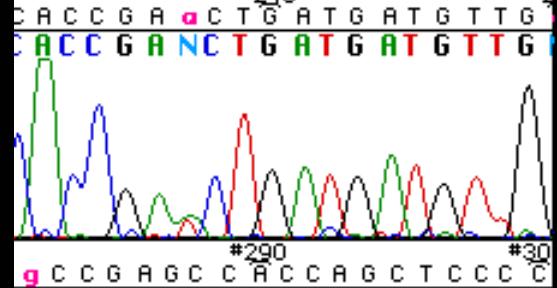


High copy spp: homogenized, amplification from a limited number of master copies

Low copy spp: much variation

# Levels

- Sequences
- Genes / motifs
- Repetitive DNA
- Chromosomes
- Chromosome sets  
(‘Genomes’)
- Genotypes/CVs
- Species
- Genera and above



... involving

- Mutation
- Rearrangement
- Duplication
- Deletion
- Homogenization

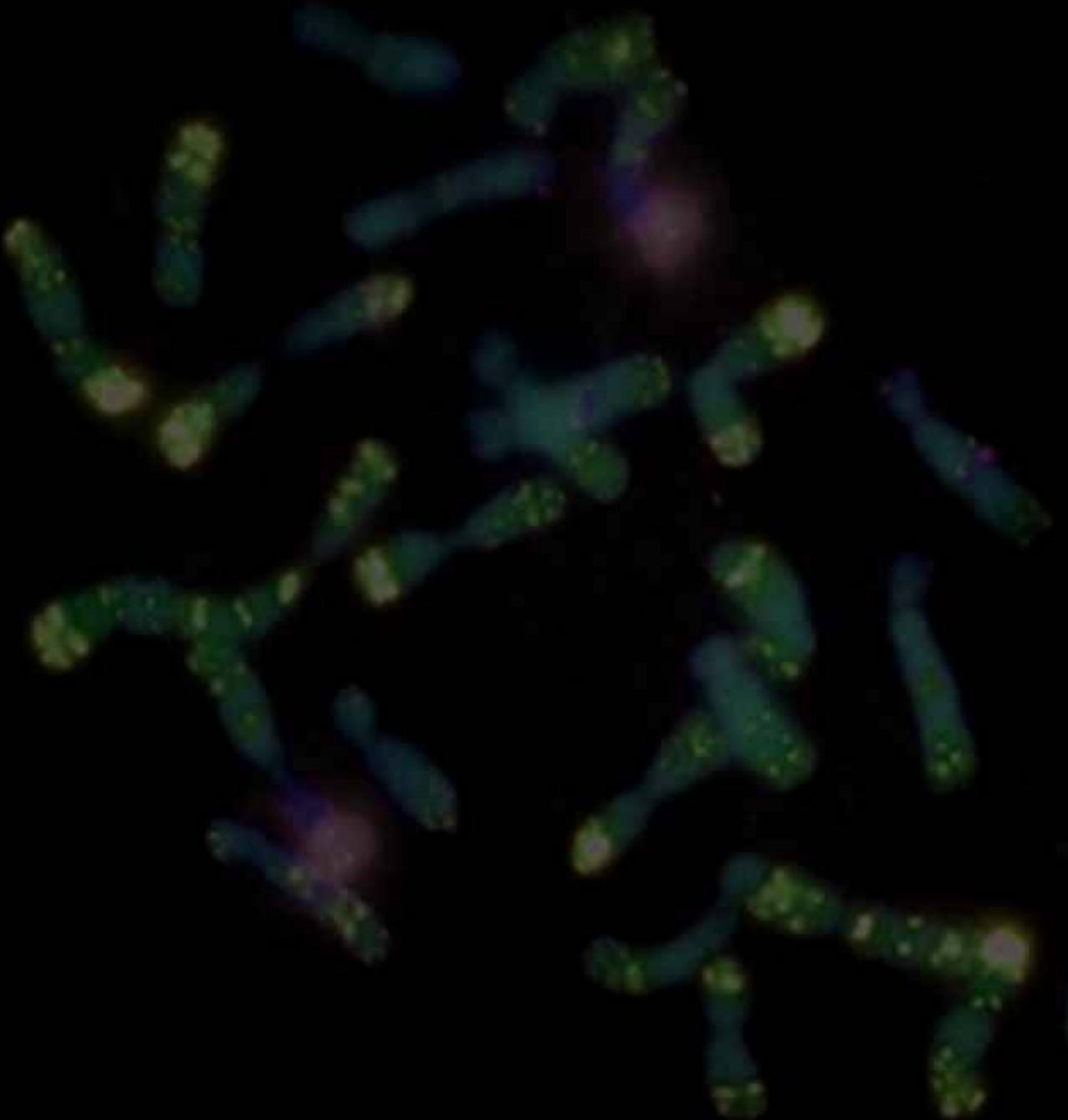
... consequences for

- Crops / wild species
- Selection
- Speciation



# Chromosome and genome engineering

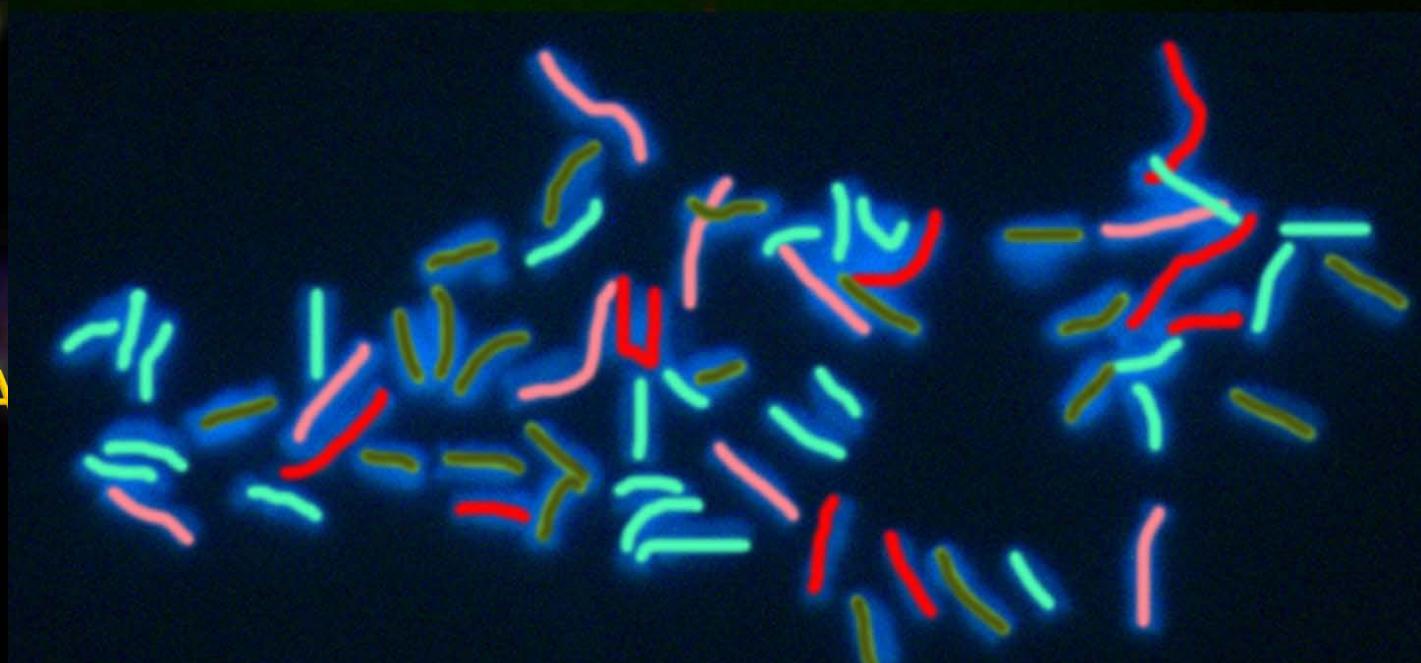
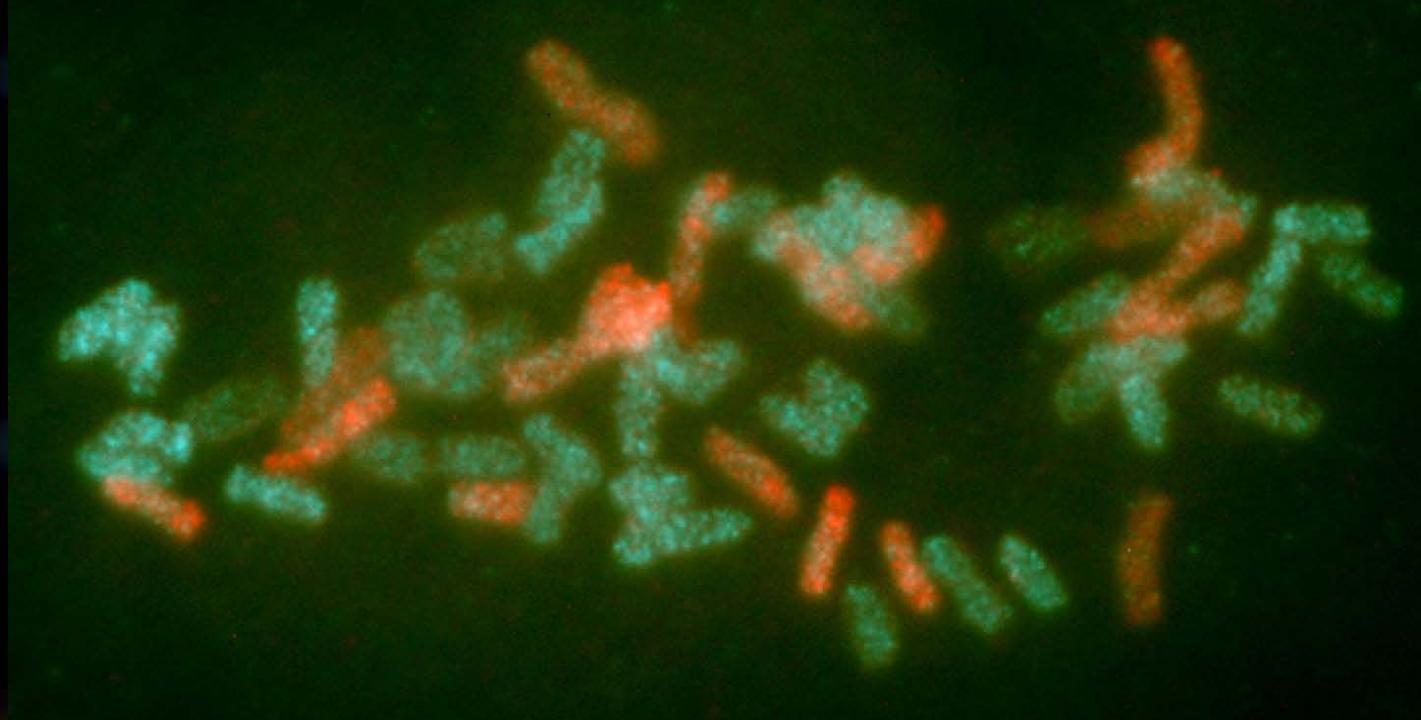
Cell fusion  
hybrid of two  
4x tetraploid  
tobacco  
species



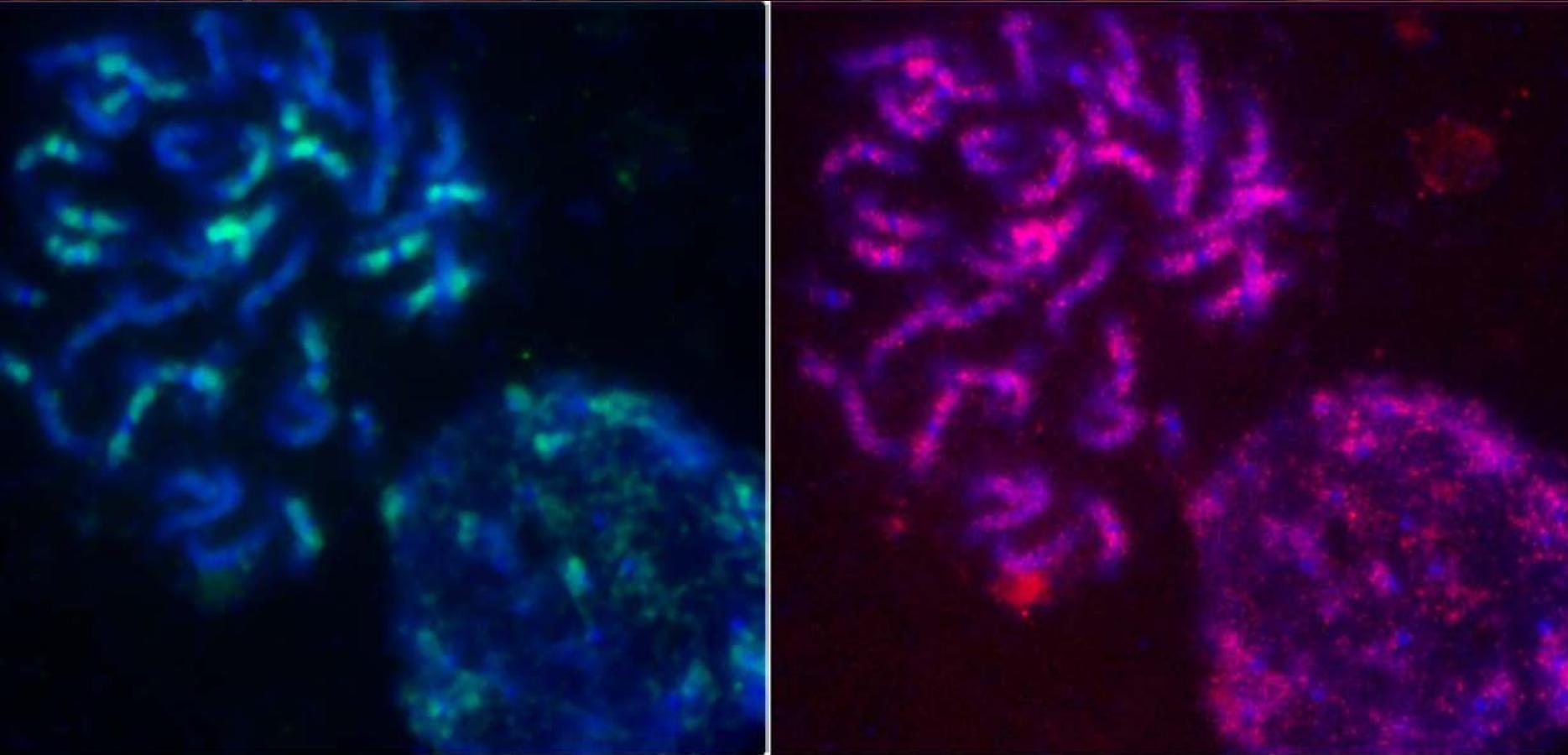
Patel, Badakshi, HH,  
Davey et al 2011  
Annals of Botany

*Nicotiana*  
hybrid  
 $4X + 4X$   
cell fusions

Each of 4  
chromosome  
sets has  
distinctive  
repetitive  
DNA when  
probed with  
genomic DNA



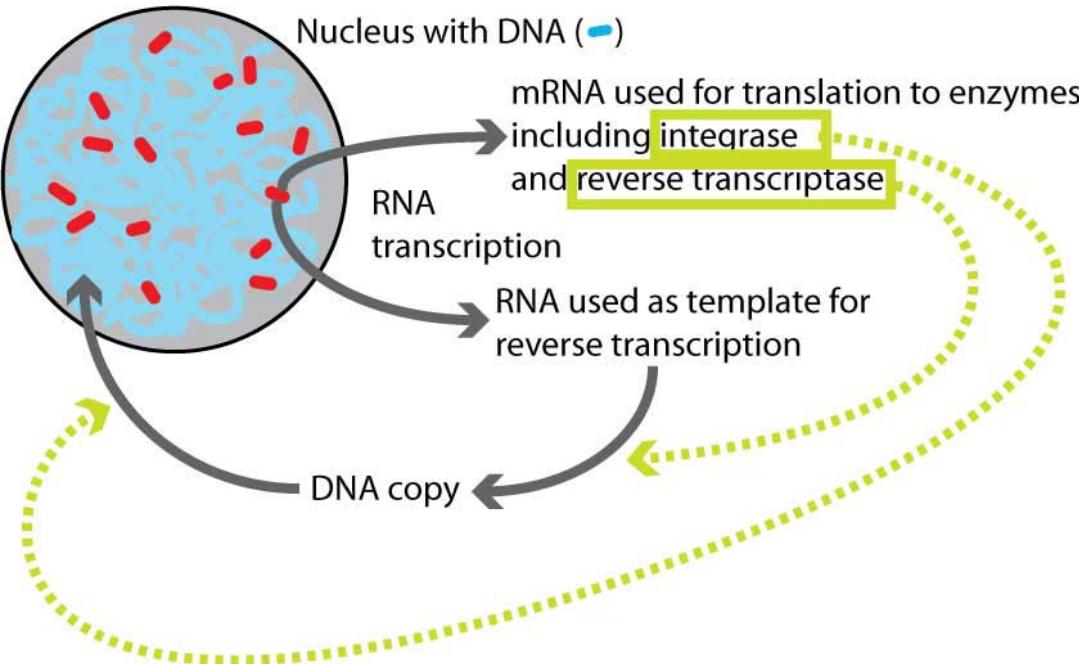
# Genome evolution



*Arachis hypogaea* –  $2n=4X=40$

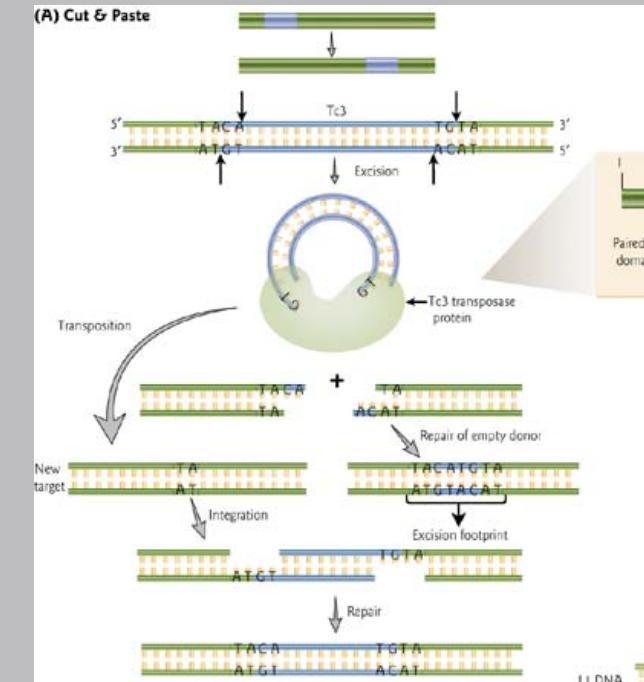
*In situ* hybridization of two BACs including repeats  
Contrasting distribution of their major repeat families

# Retrotransposons (●): The transposition cycle

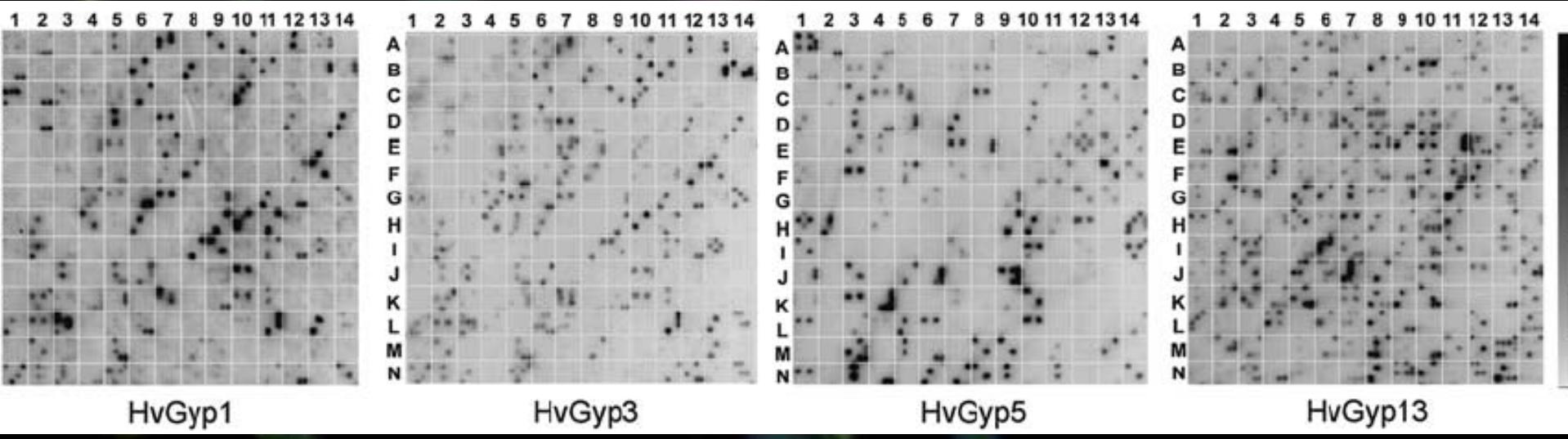


Retrotransposons  
Class I transposable elements  
RNA intermediate

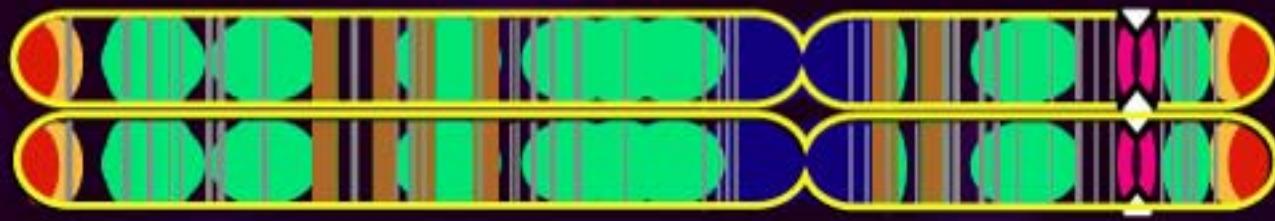
DNA transposons  
Class II transposable elements  
Cut-and-paste



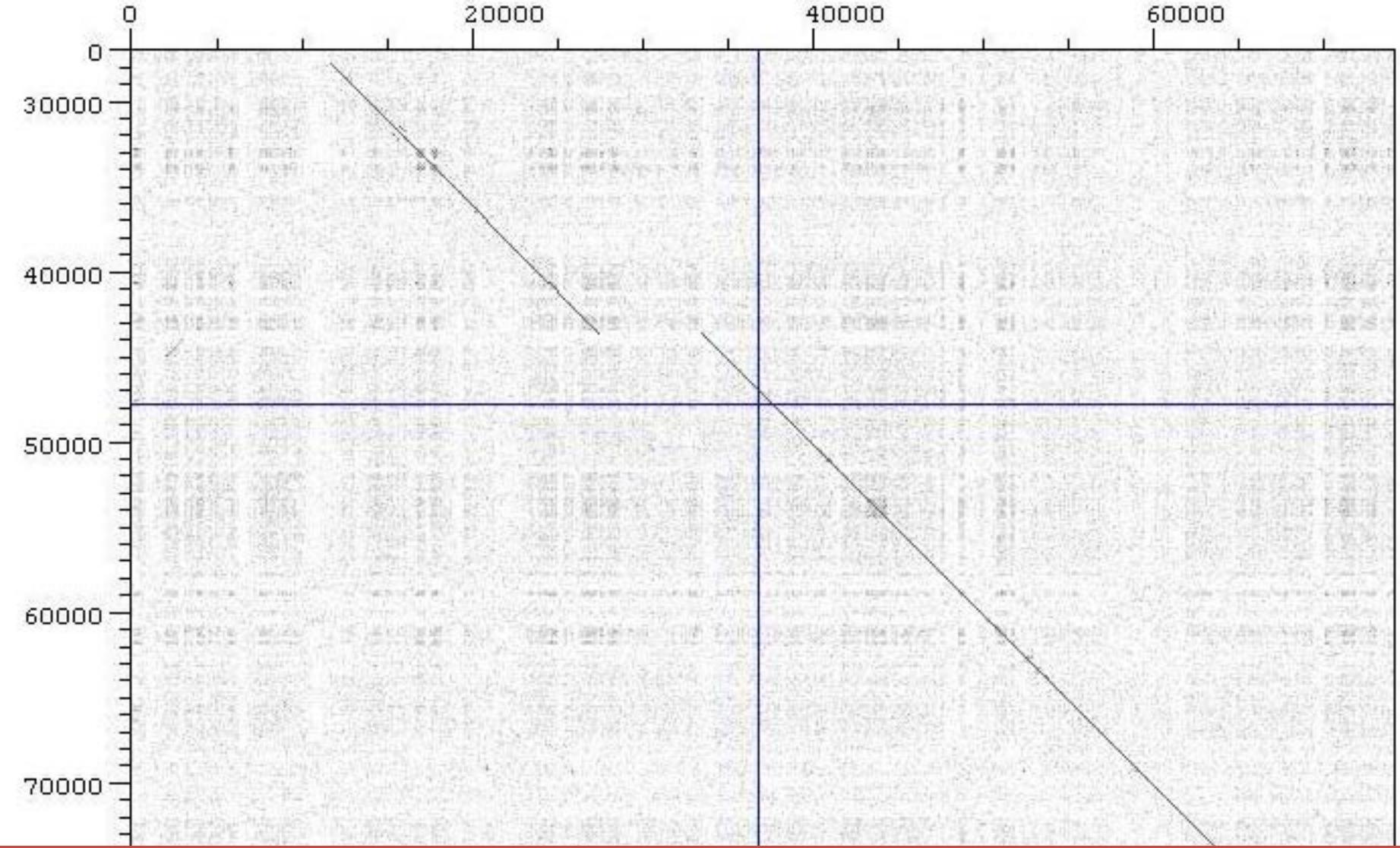
# Retroelement abundance and diversity in barley



Element abundance present > 5% of genome



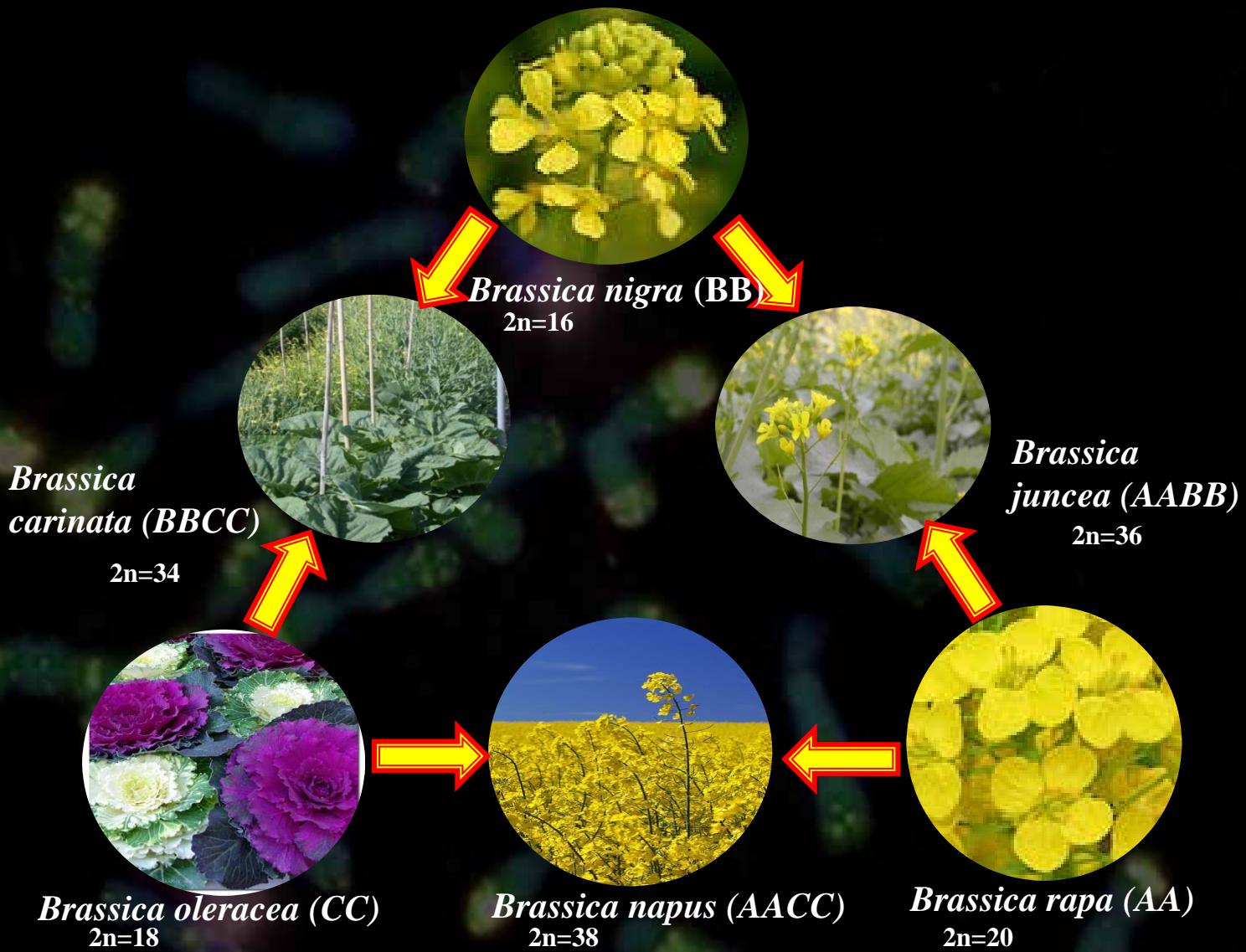
Barley gypsy: Vershinin, Druka, Kleinhofs, HH: Plant Mol Biol 2002;  
cf Brassica Alix & HH Plant Mol Biol 2005



Dotplot comparisons at scale of 10,000s bp

Two *Musa* chromosomes are >95% homologous with gaps

Faisal Nouroz 2011

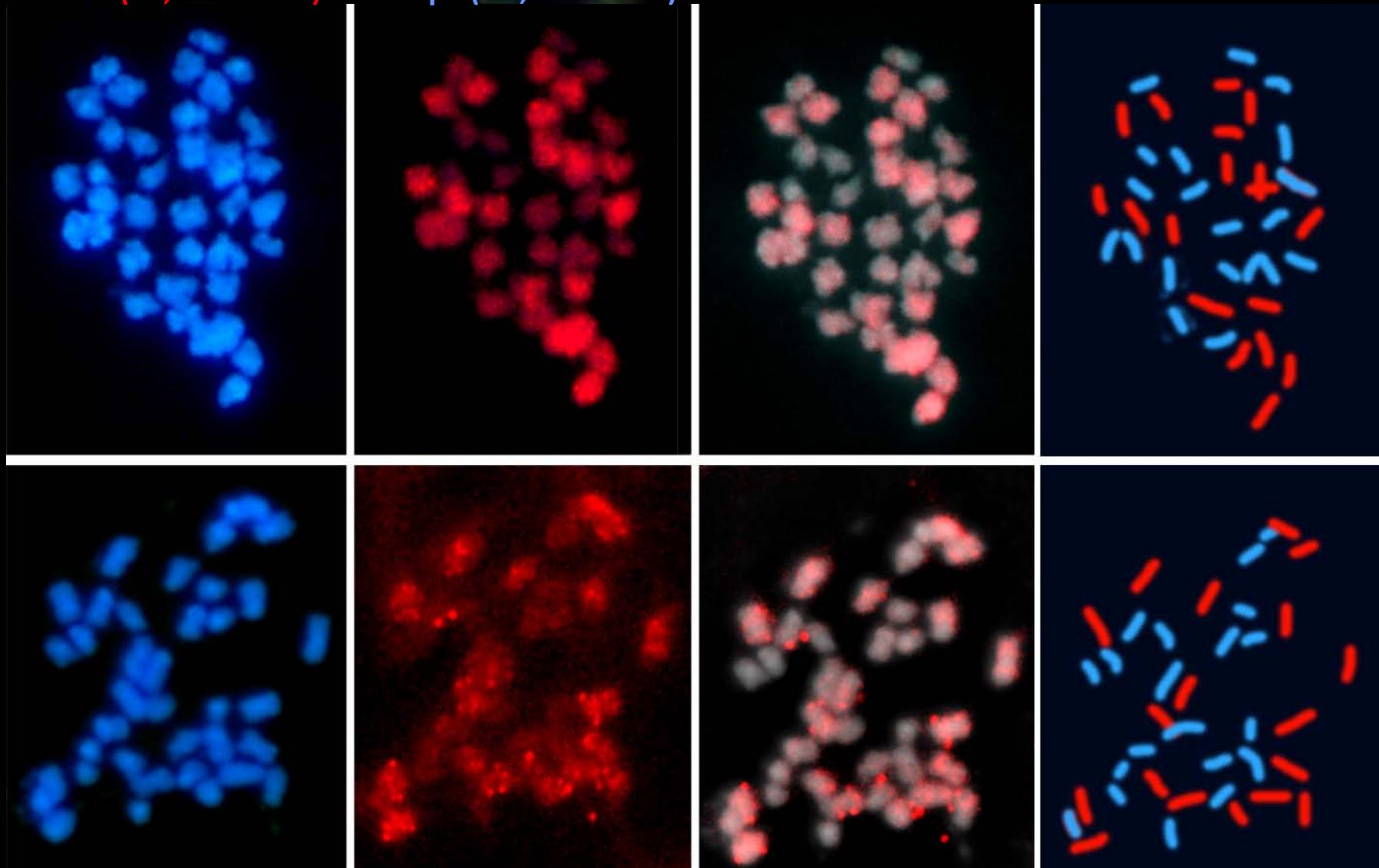


The *Brassica* genus is monophyletic, from single common ancestor.  
 What has changed in the DNA sequences?

# Genome Specificity of a CACTA Transposon

*B. napus* (AACC, 2n=4x=38) – hybridized with C-genome CACTA element red

*B. oleracea* (CC, 2n=2x=18)      *B. rapa* (AA, 2n=2x=20)

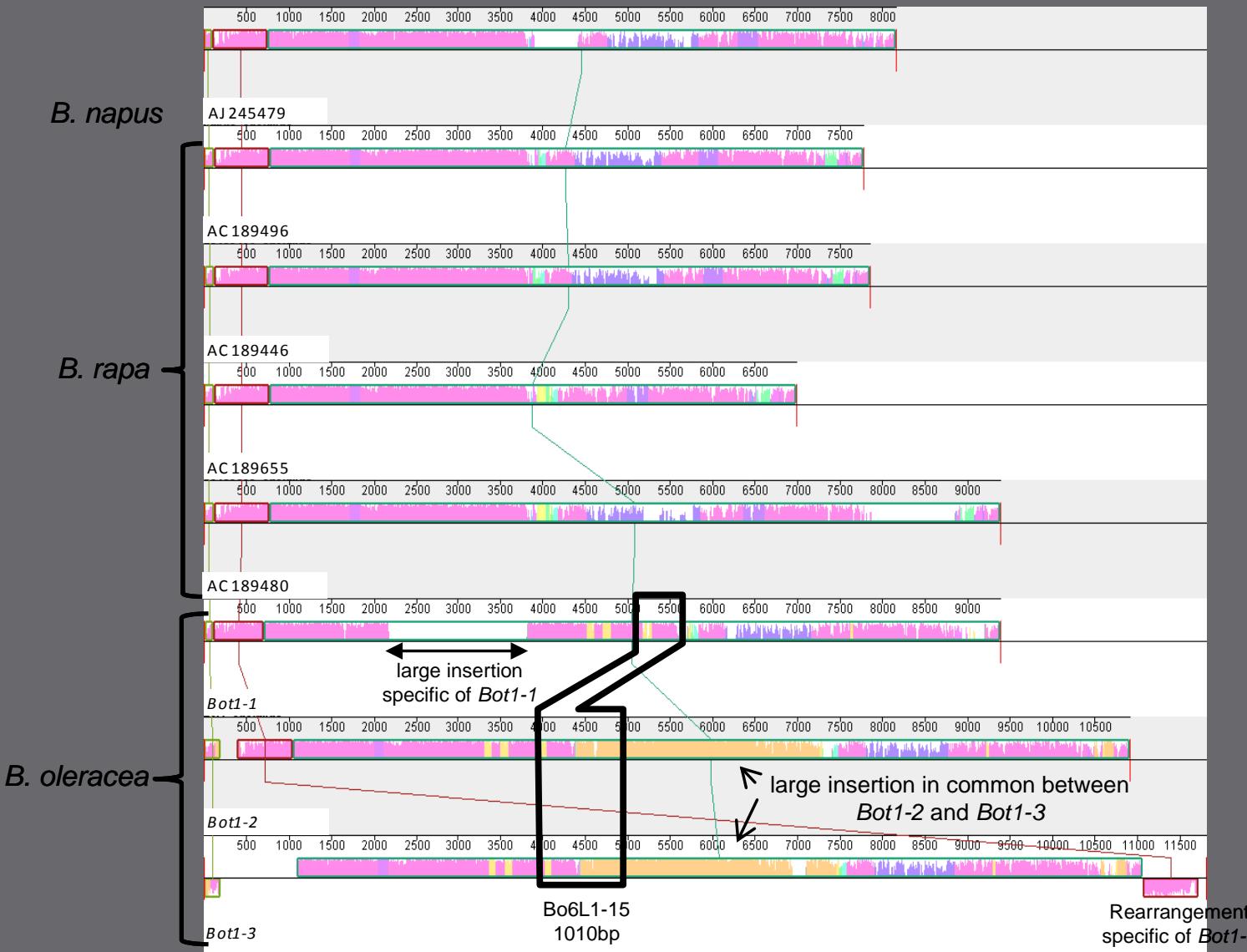


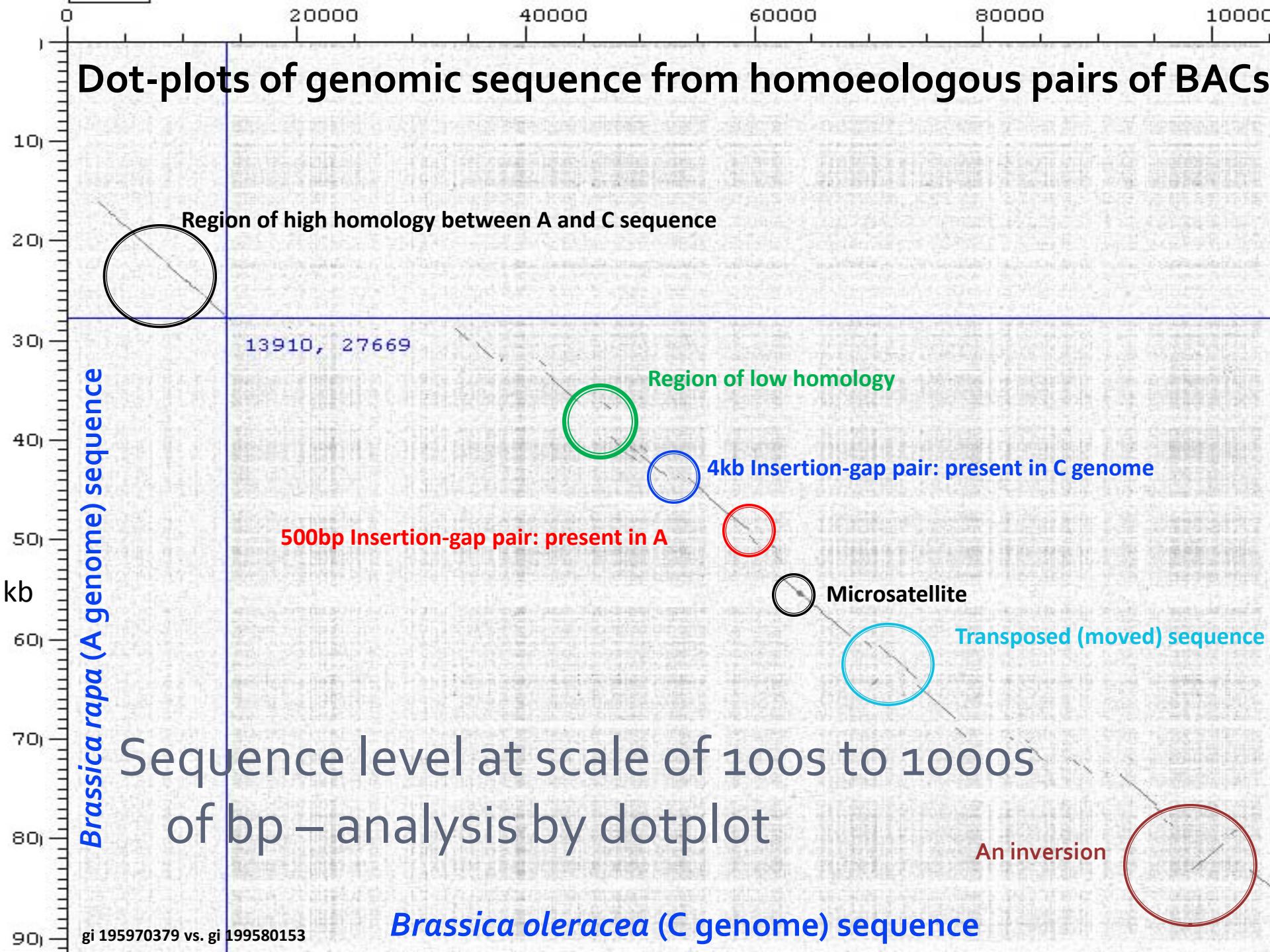
Alix et al. The CACTA transposon Bot1 played a major role in *Brassica* genome divergence and gene proliferation. Plant Journal

# Genome Specificity of a CACTA (En/Spm) Transposon

Carries a S locus-associated *SLL3* gene – involved in gene proliferation?

Transposons drive genome and gene evolution





4kb Insertion-gap pair: present in C genome

A

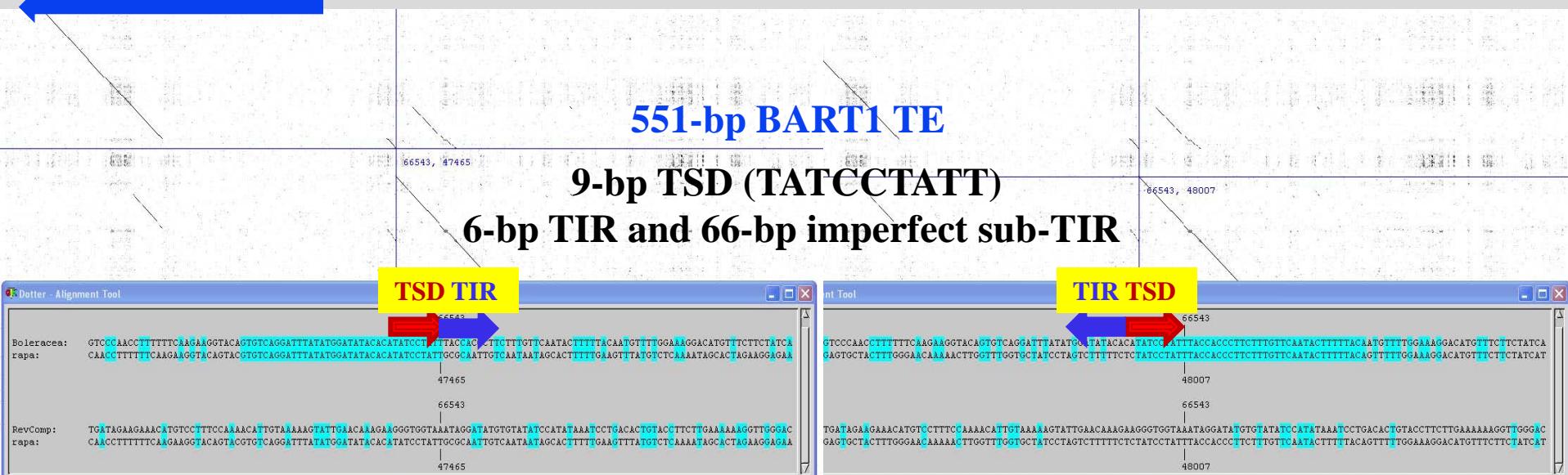
Microsatellite

Transposed (moved) sequence

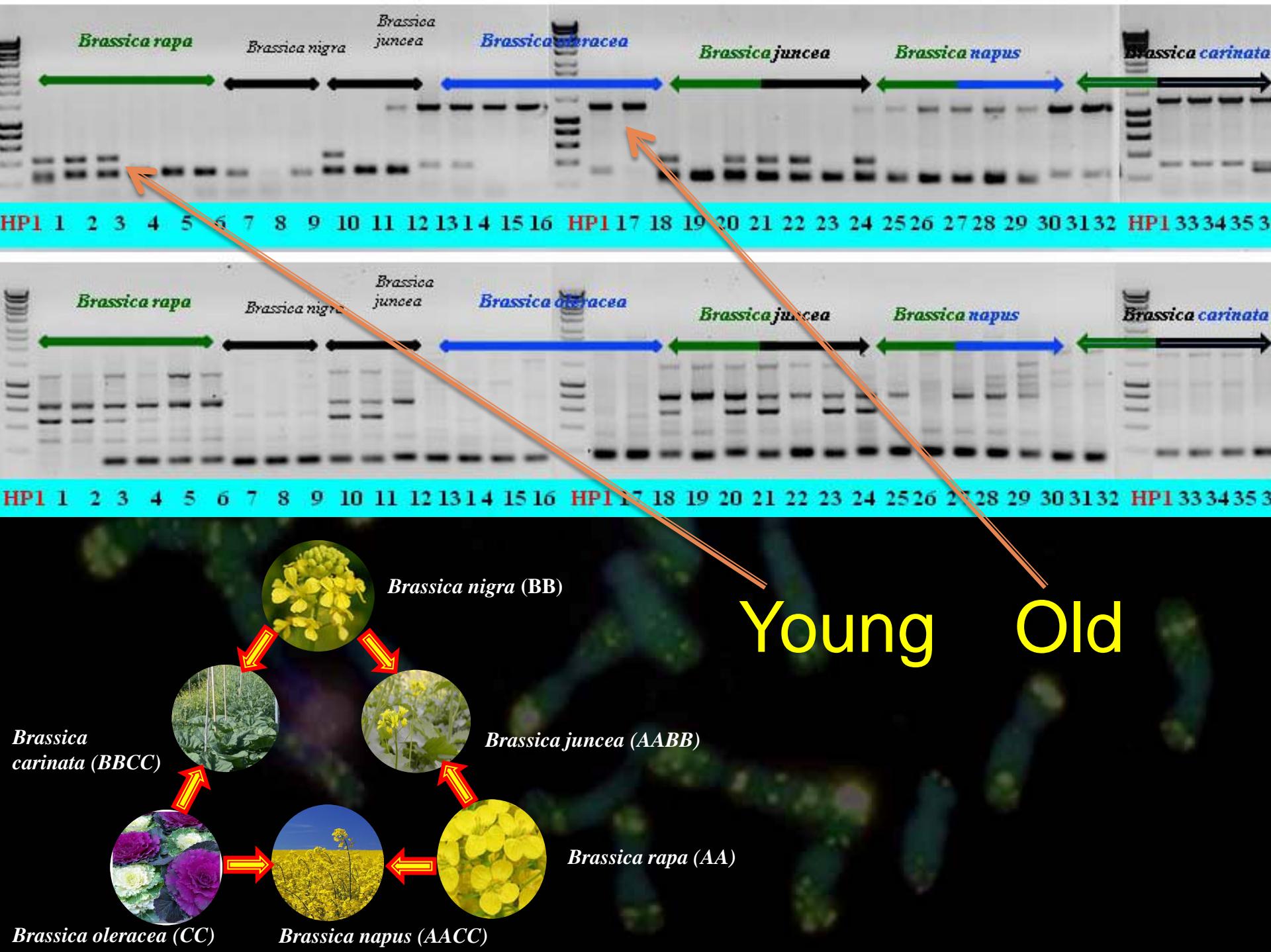
An inversion

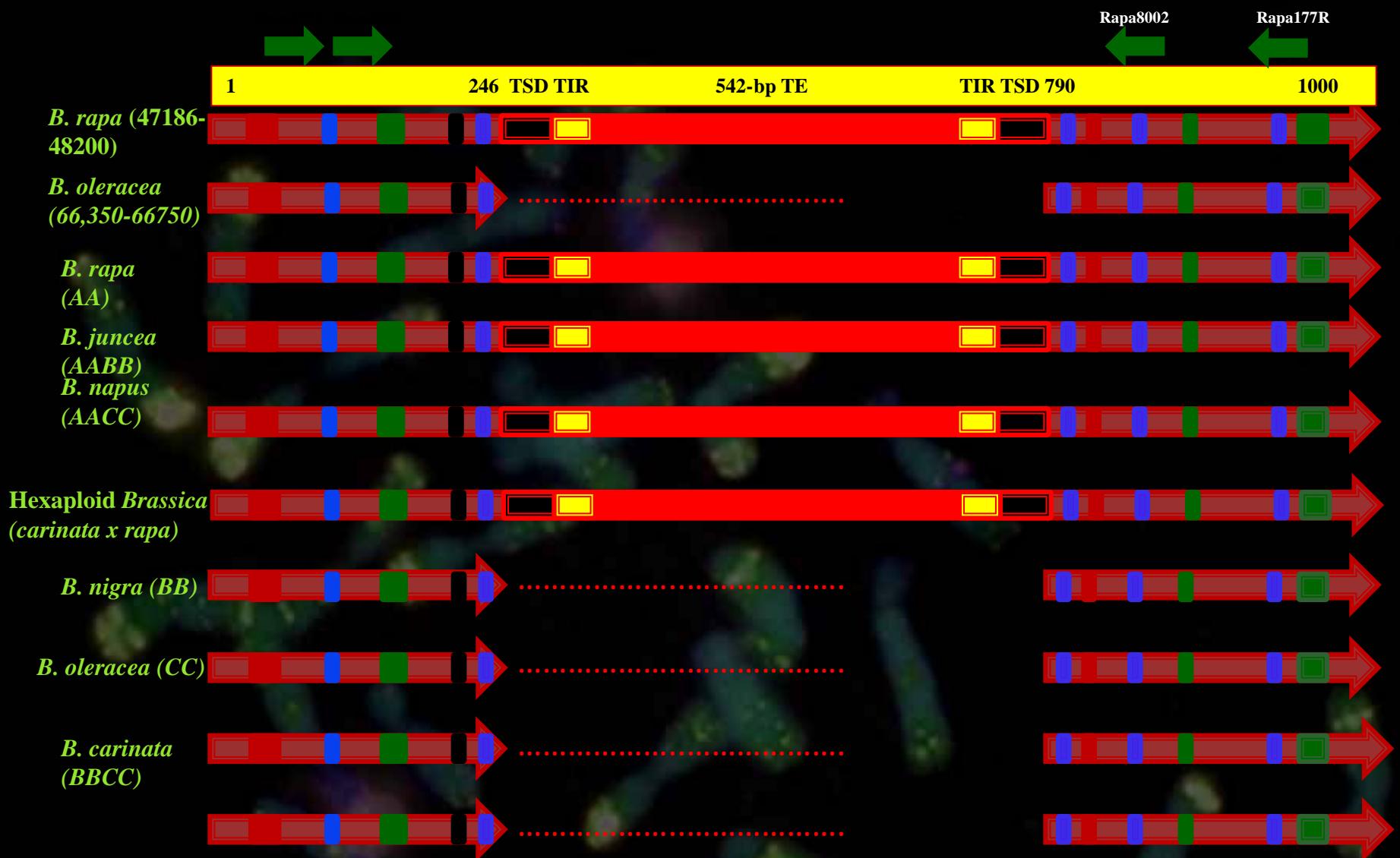
AAGTGAATGGATGCTCGCATTAGTTACTAT **GAGCCGATTCTCGCTTGC**AAAGCTAAAGAGGAAAAGGCCTTCG **CATTGCAGAAG**  
 AGCTGGCTGCCAGCGAGCAAGAGGTTTCATATT **GGCTTGTG**AAAATTGTTGCCACTTTGCTTACTAAGGAATGAAATAATAC  
 TTGTTTTTTTTCATGGTTAATATTAGAAGATATAATTCCCTTGAGTTAGATTACGTTCTTAT **GTCGACGAAGTGAAGAAATATT**  
**GTCTTGT**TTATGGTCCCTCTAGCCCCAACCTTTCAAGAAGGTACAGTACGTGTCAGGATTATGGATATACACA  
**TATCCTATTGCGCAATTGTCAATA** **ATAGCA**TTTGAG **TTTATGTCT**AAAATAGCACTAGAAGGAGAAAGTCACAAAAATGATATT  
**CATTAAAGGGTAAATAT**CTCTTATATCCTGGTT **AAAATTAA**AAAC **AAAAA**TAAT **AAAAA**TAAT **AAAAA**TAAT **AAAAA**ATGAAAAAA  
 AAGAAAT **TTTTTT**ATAGTTTCAGATTATGTTTCAGATTTCG **ATTTTTTT**ATTTTT **ATTTTTT**CGAA **ATTTTTTT**ATTTTTCA  
 AATTTCTTTTATAATT **AAAAA**ACTTTTGAAACTG **TTTTTTA**ATTTTATTTTATTAGTATT **ATTTTT**AT **AAAATTAAACCT**  
 AATTCCCAAACCCCCACCCCTTAACCTCAA **ACCCTAAGGTTGG**ATTAACCAATGGATATAA **GTGT**AT **ATTTACCTCTTAAATGA**  
**AACCT**ATTTTGACTTGAGTGC **ACTTGG**AAACAAAAA CTTGGTTGGTGC **AT** CCTAGTCTTTCTCTACCTATT

TACCACCCCTCTTGTCAATACTTTTACAGTTTGAAAGGACATGTTCTTCTATCATCACTTAATGGTTATATGTATGAGAAG  
 TTTGAAAGAGATTACACTGTTTGGATATTAAAAAAAGATATTACAAGATCTGATTGTTGTATTAAAATTCTACCAAATC  
 TCTCCTCAAAATCTG **GTCAAAGTCC**AAAATCCAAATATCTCAGTAAATTCCACCAATATGAAATCCTAAAACCTTCCAAAATA  
 GTTCAATAAGCCCT **TAGTG**TTGGTG



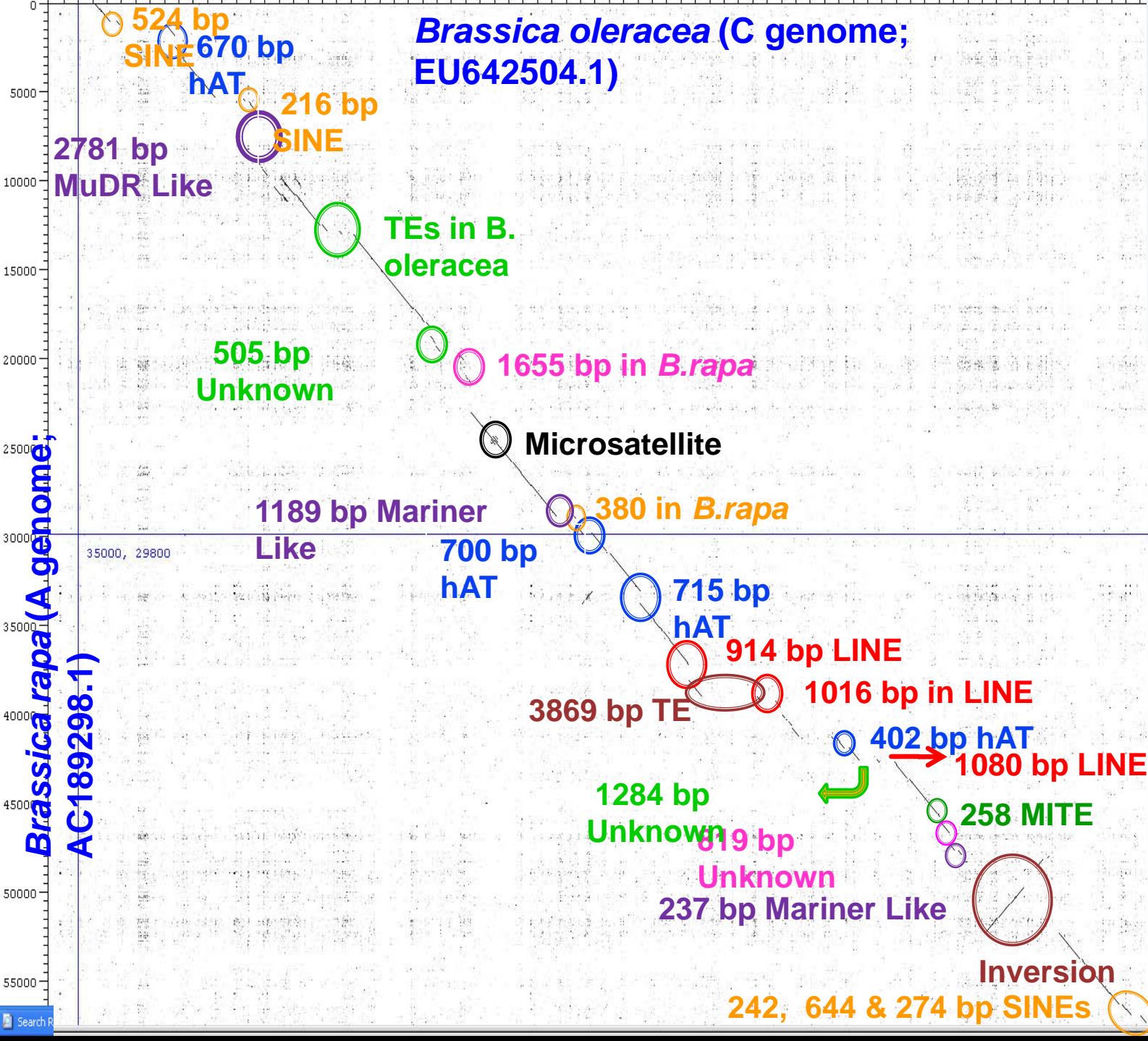
*Brassica rapa* with inserted 542bp sequence not present in *B. oleracea*. 9bp TSD (red letters and arrow) and TIR (blue). Flanking primers used in PCR (next slide) as blue arrows on sequence  
 Faisal Nouroz 2011





Mobile element insertion in *Brassica*

# *Brassica rapa* (A genome; AC189298.1)



# Repetitive DNA

rapid evolution in copy number, location and sequence, with diverse turnover mechanisms

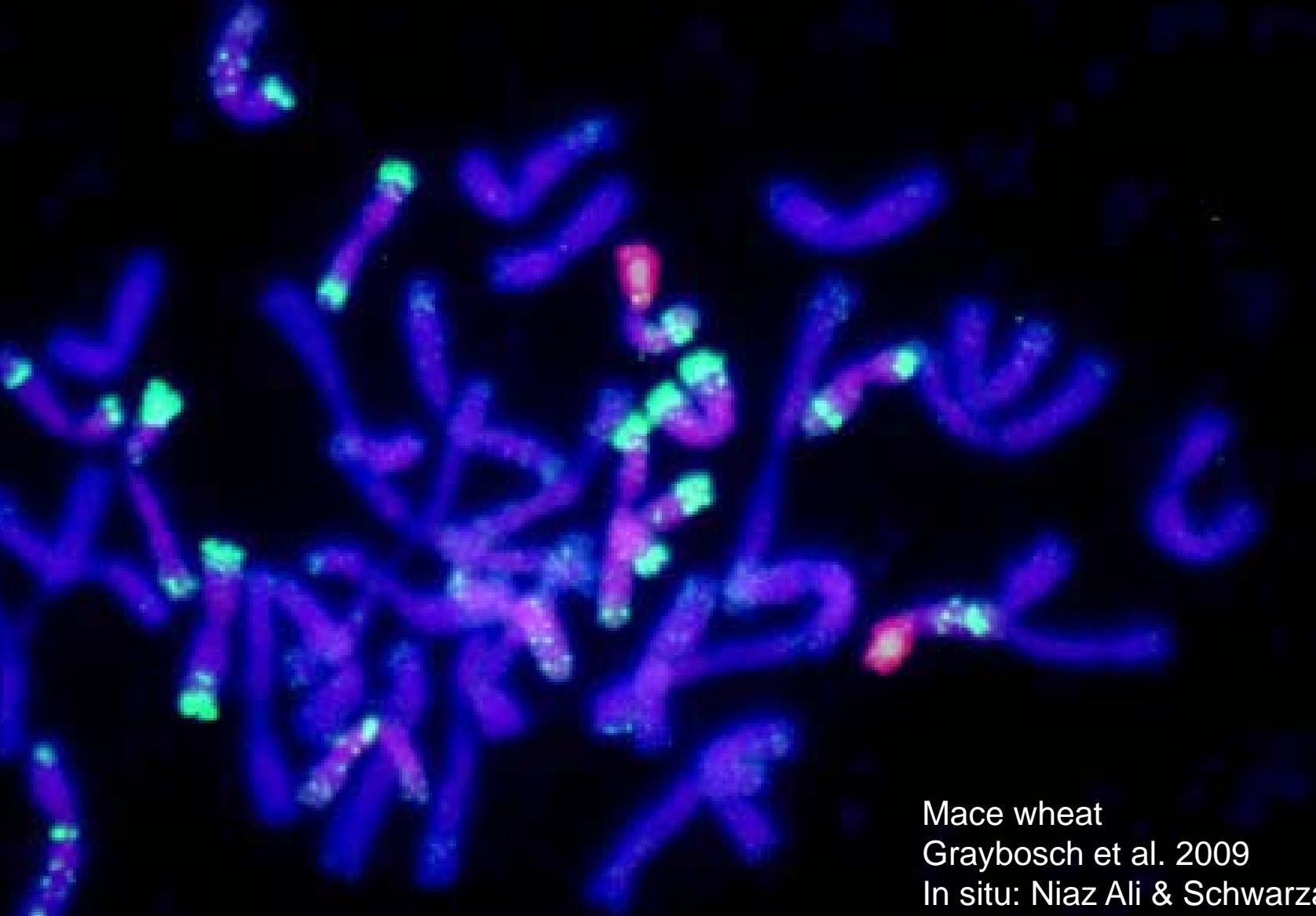
often marks the major differences between closely related species

it is hard to analyse by next generation or whole-genome sequencing methods

Many of the repetitive sequences are retrotransposons and DNA transposons

Some are microsatellite motifs

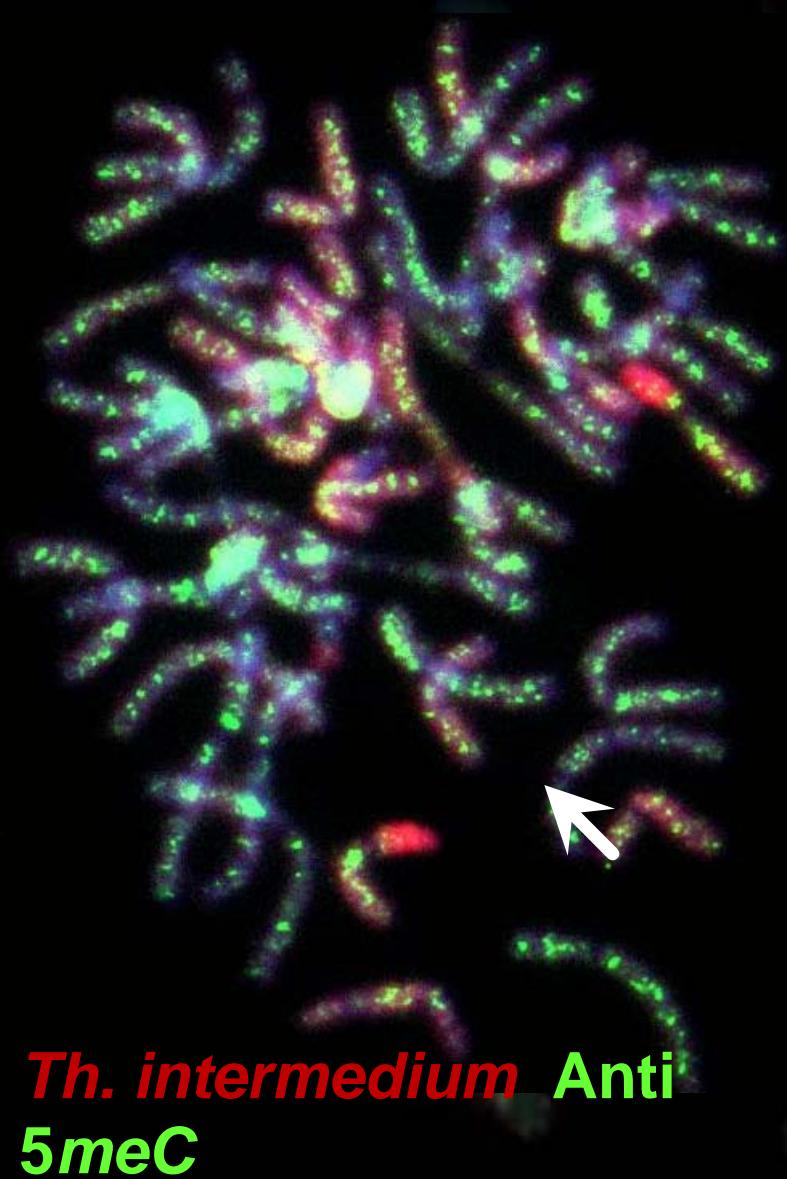
Some are satellites – including the most rapidly evolving sequences



Mace wheat  
Graybosch et al. 2009  
In situ: Niaz Ali & Schwarzacher

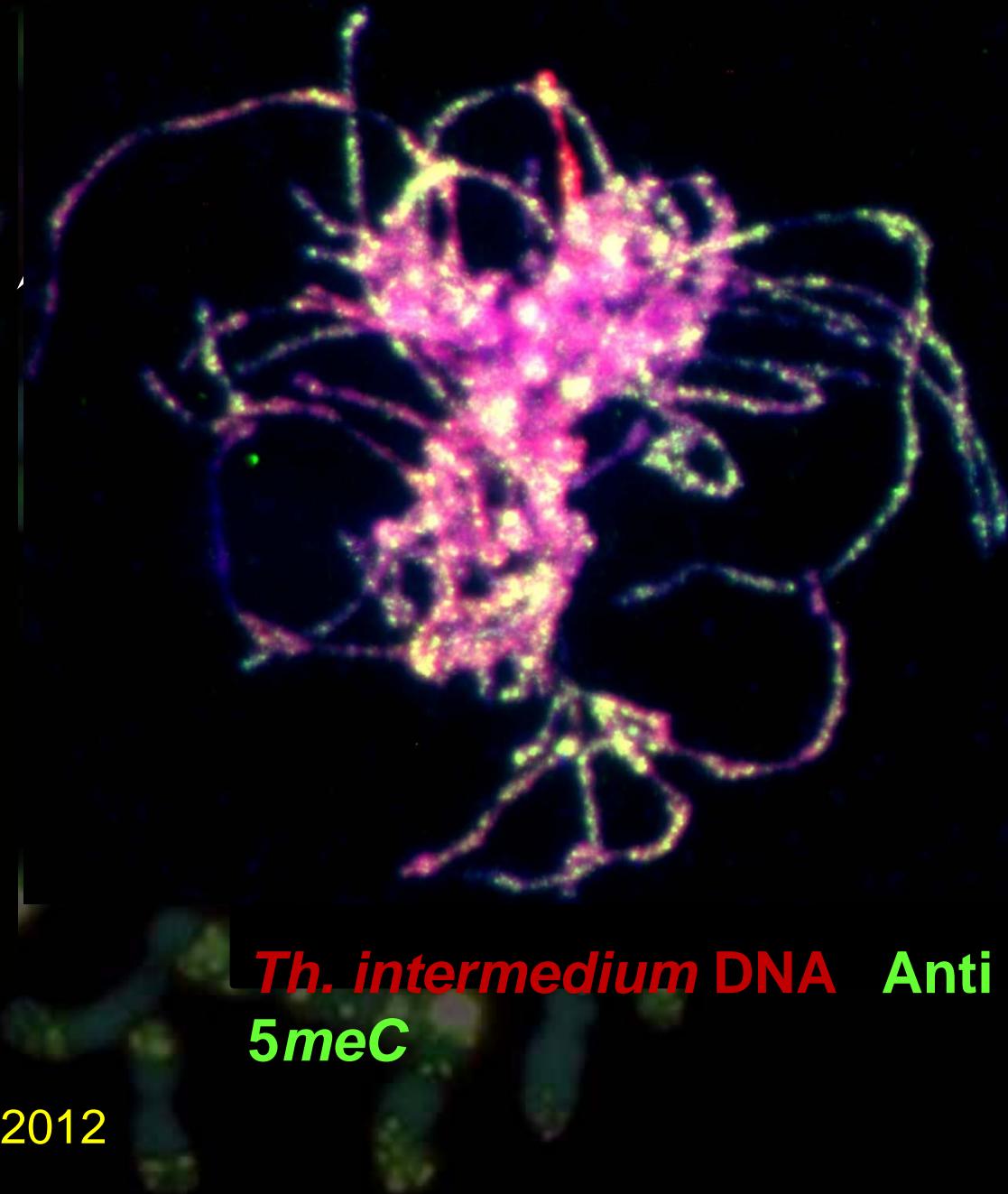


**Wsm-1: only highly effective source of resistance to WSMV**



*Th. intermedium* Anti  
5meC

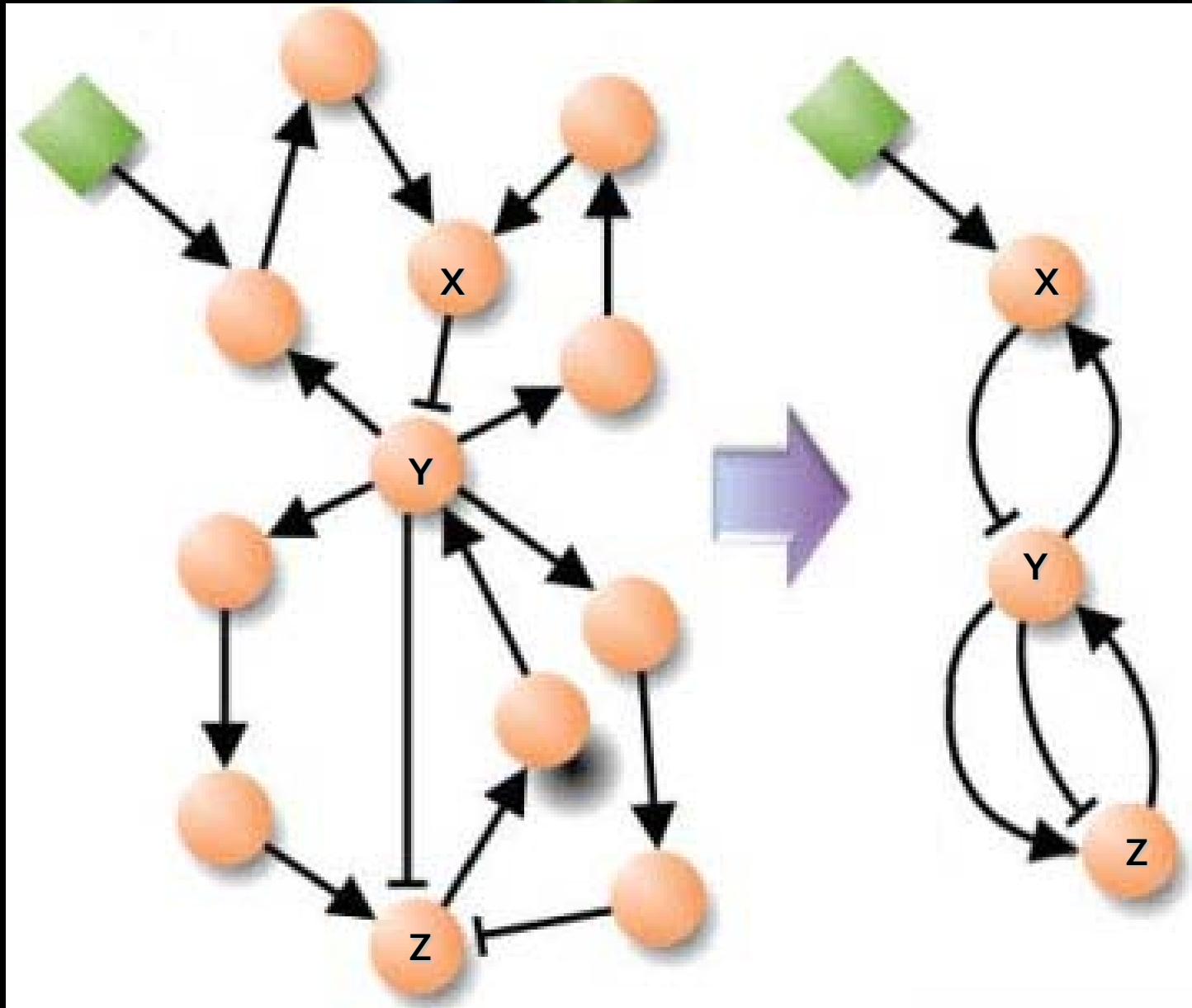
Niaz Ali, Trude Schwarzacher 2012



*Th. intermedium* DNA Anti  
5meC

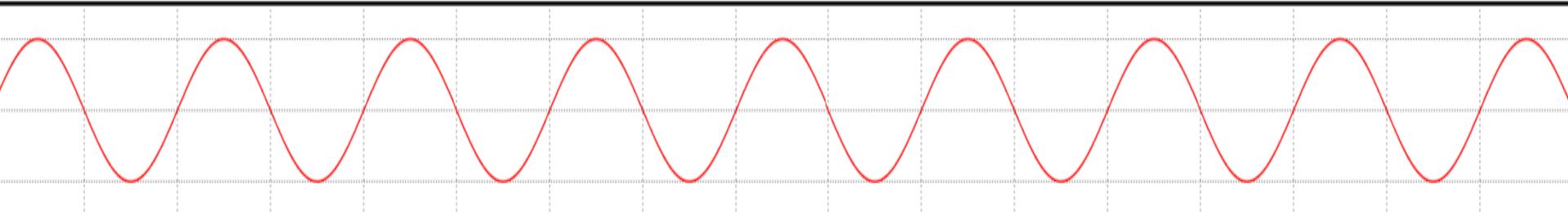
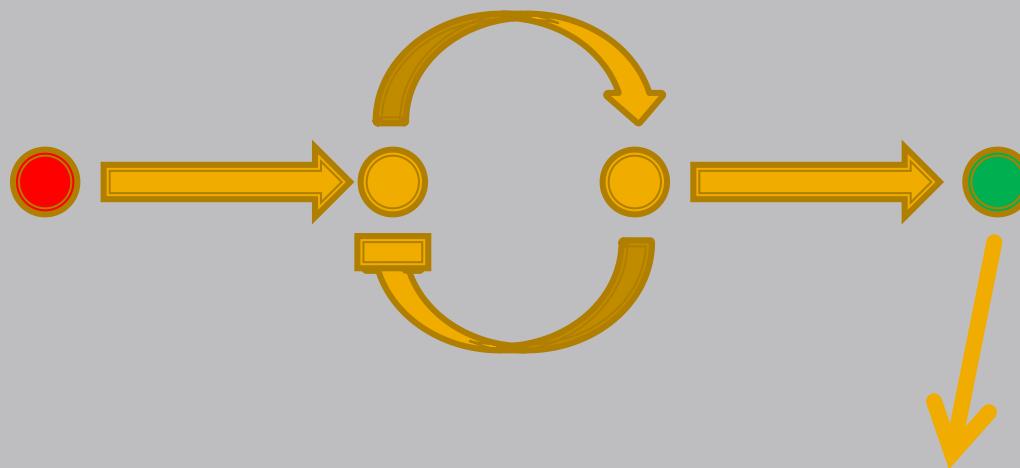
# Network reduction

Circadian Clock regulation  
after Leloup & Goldbeter  
cf Andrew Millar  
in Arabidopsis

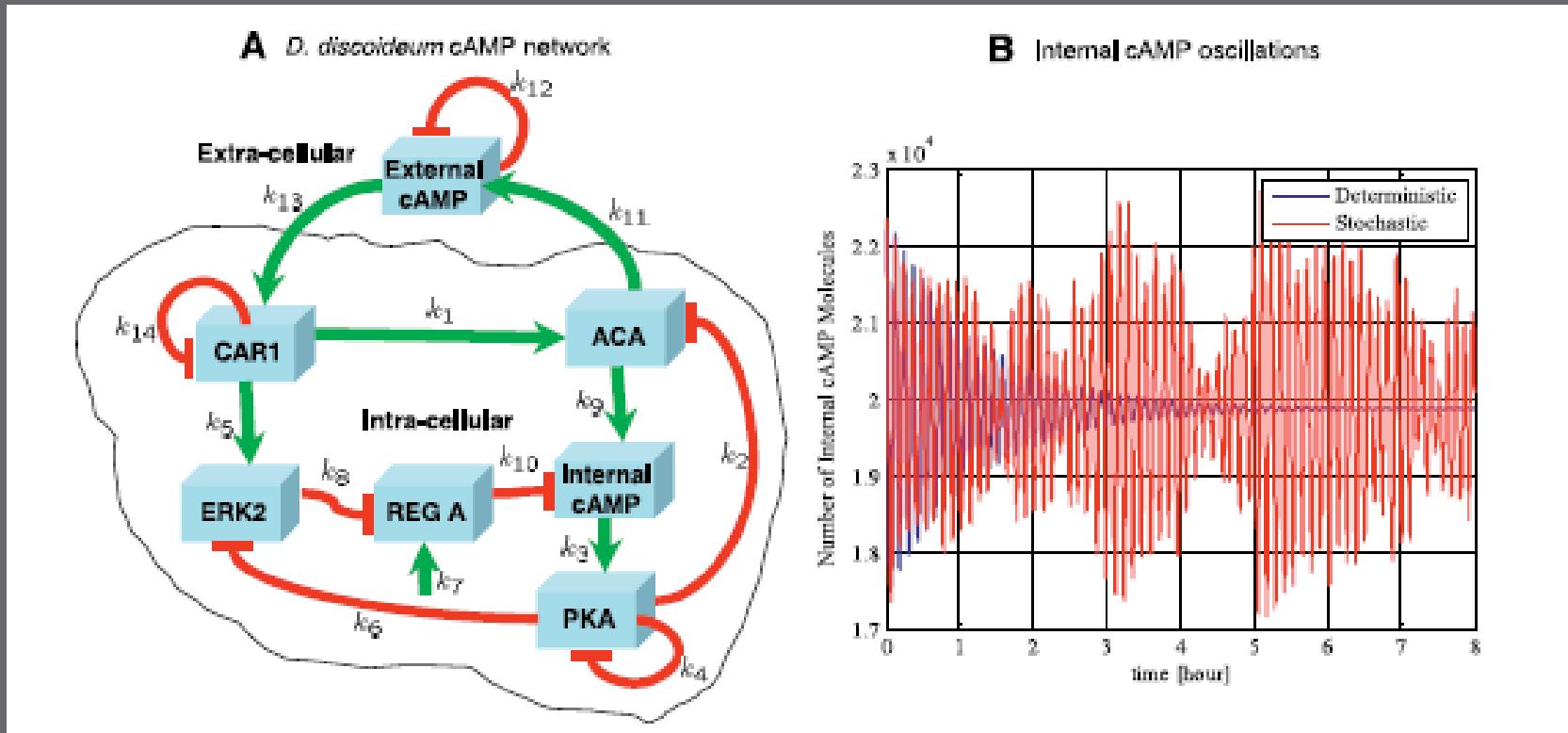


# Regulation of oscillations

- Robustness
- Synchronization without external regulators



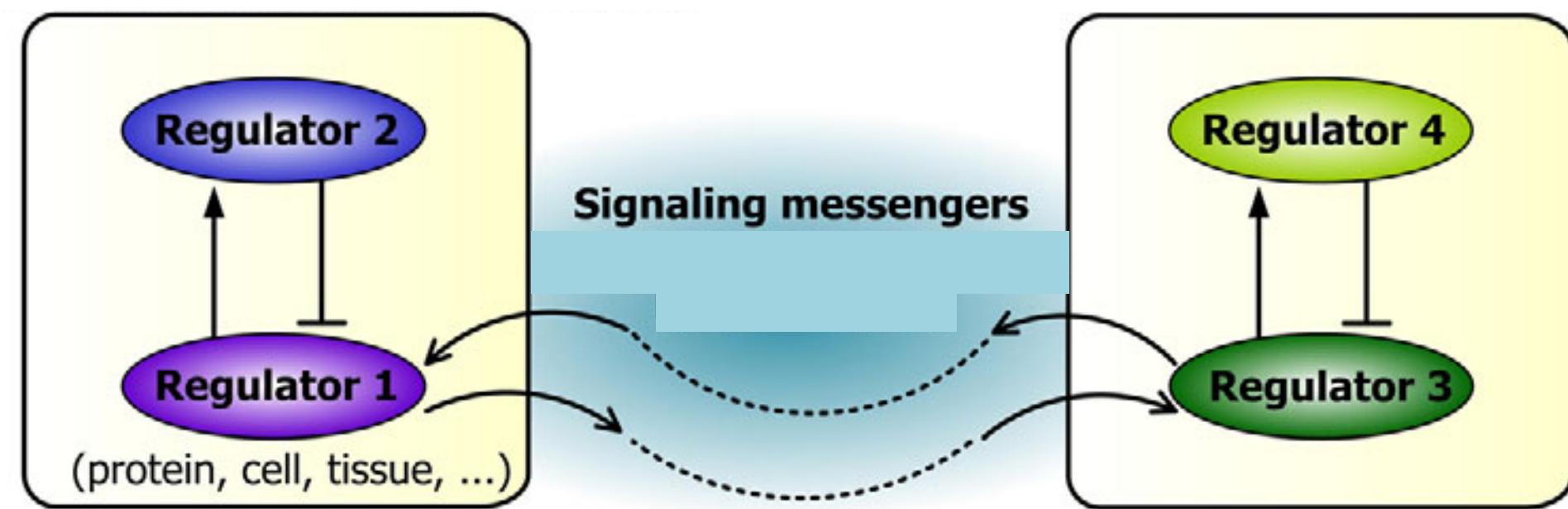
# Oscillations: noise and stability



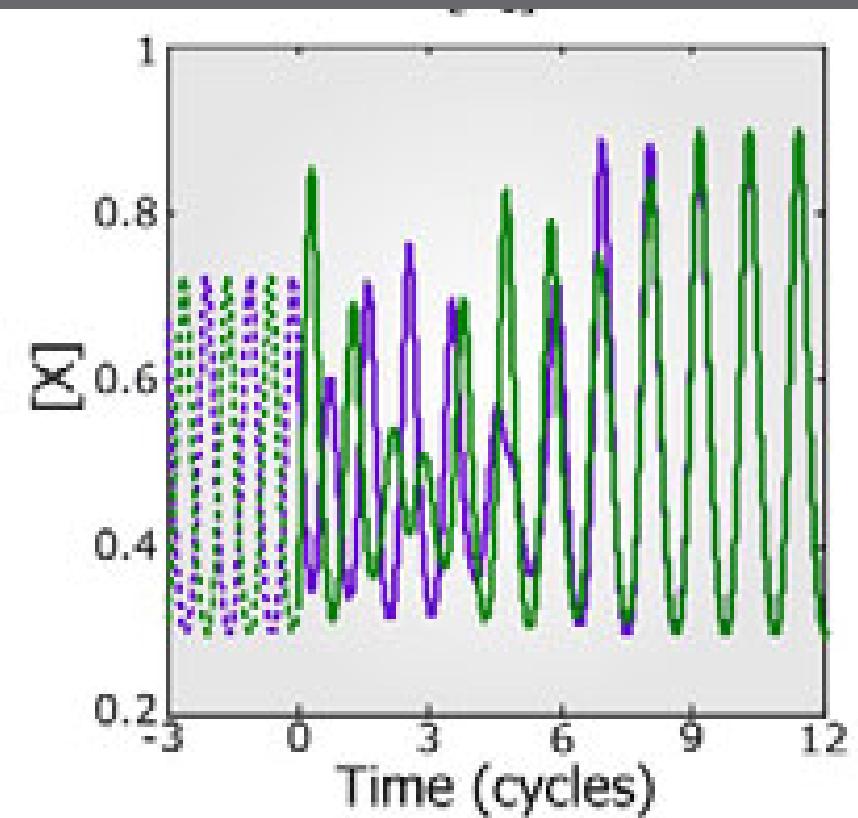
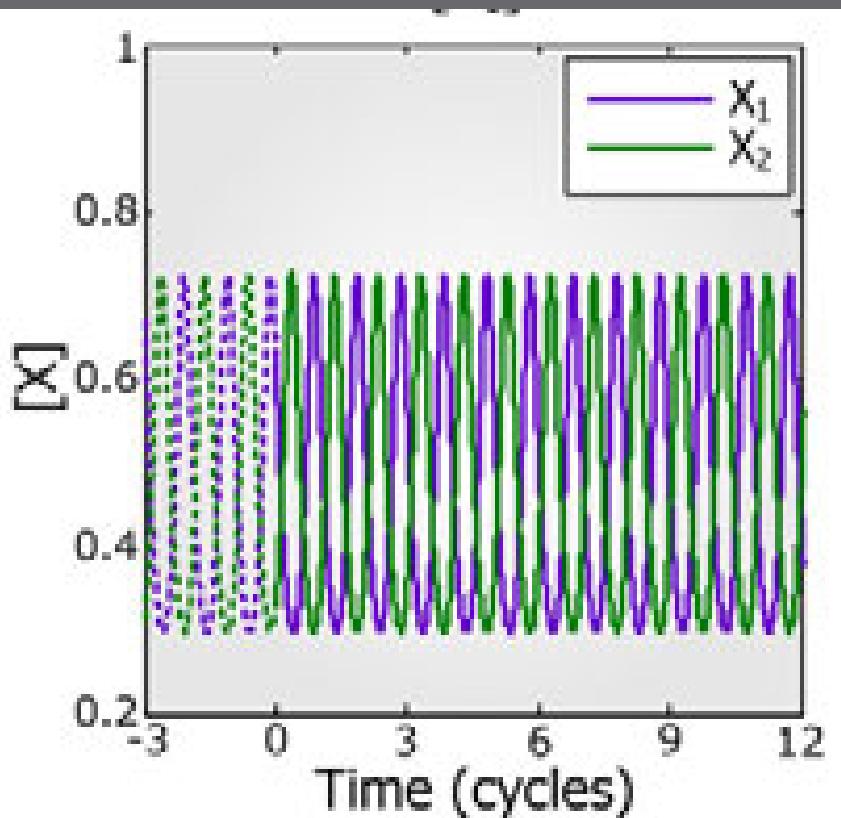
- Stochastic fluctuations
  - preserve stable oscillations
  - ensure robustness of the oscillations to cell-to-cell variations
- Robustness analysis requires stochastic simulation

## Oscillator 1

## Oscillator 2



# Weak Stronger Coupling



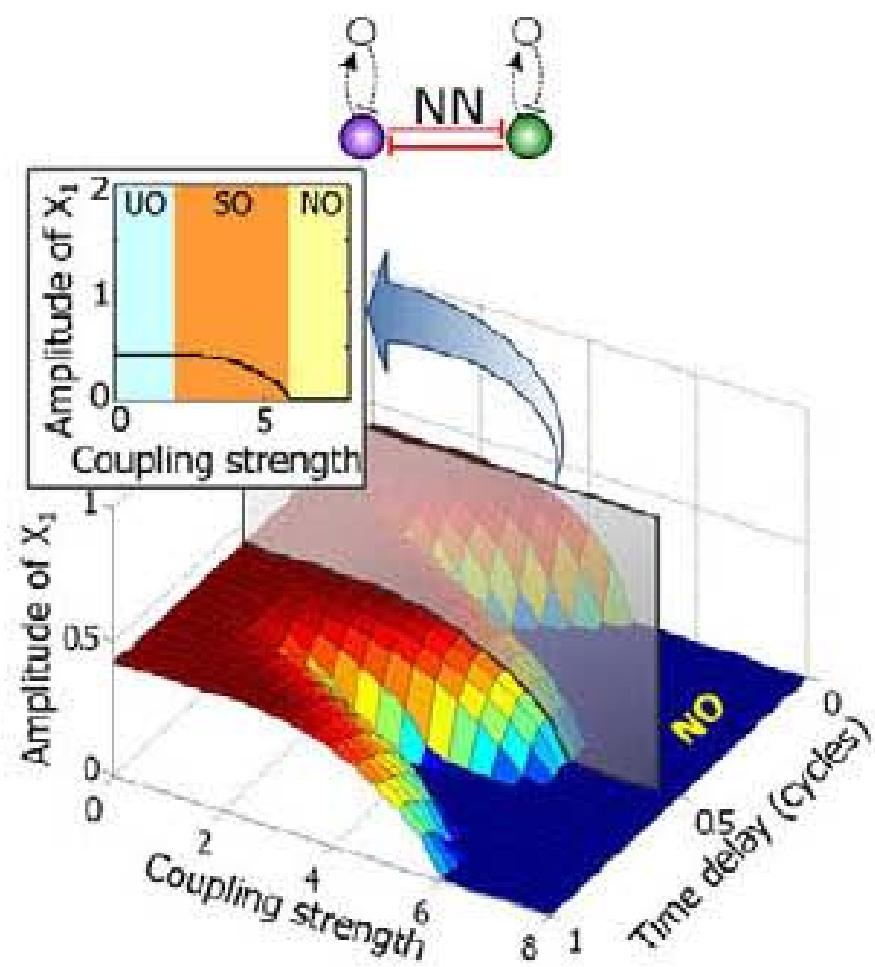
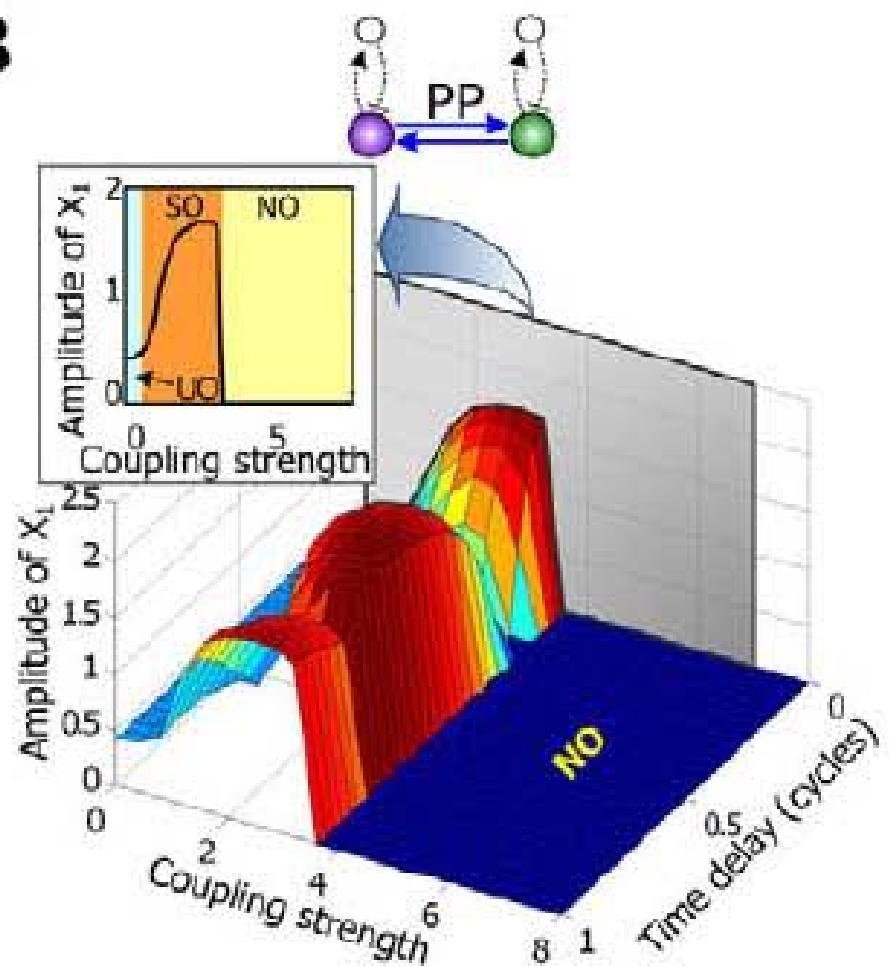
Live version: YouTube – pathh1 channel phaseportrait.avi

Jeong-Rae Kim, HH & Kwang-Hyun Cho. J Cell Sci 2010

# A design principle of synchronized oscillations

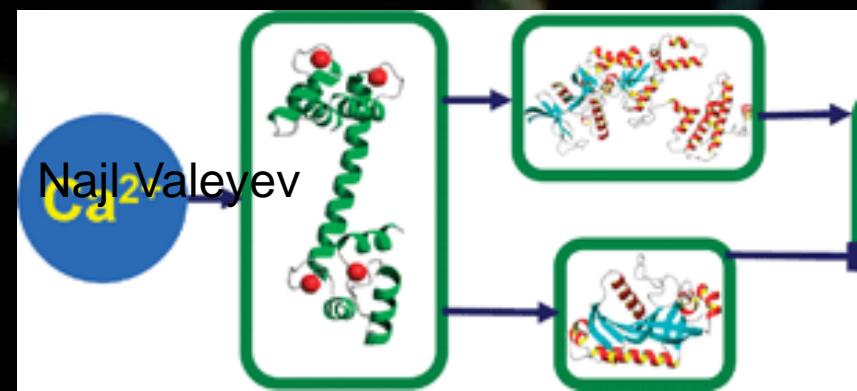
541

B

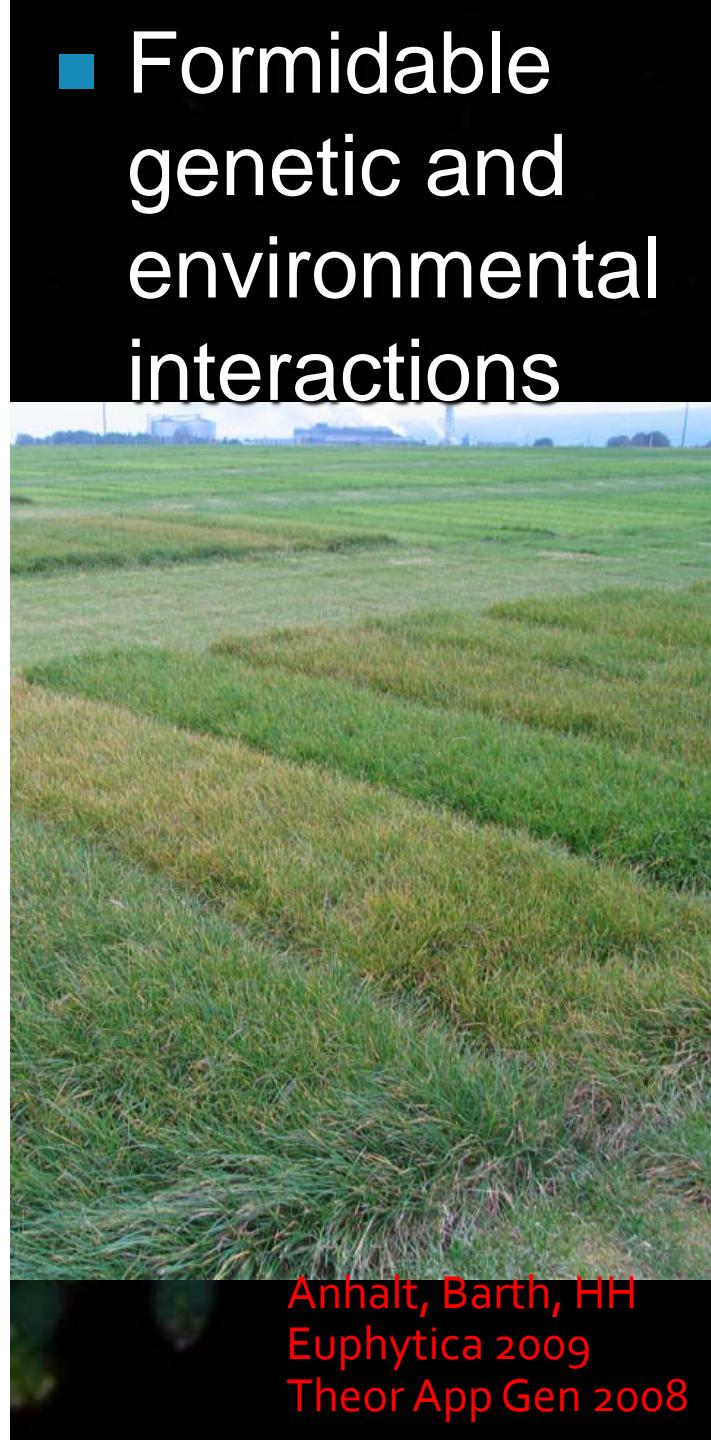
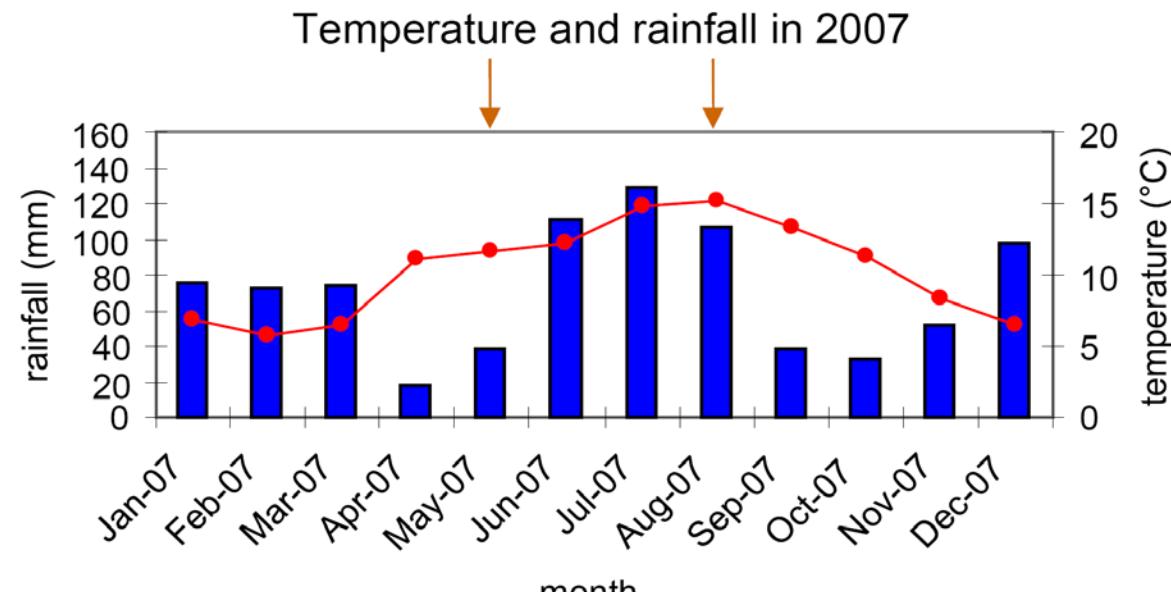
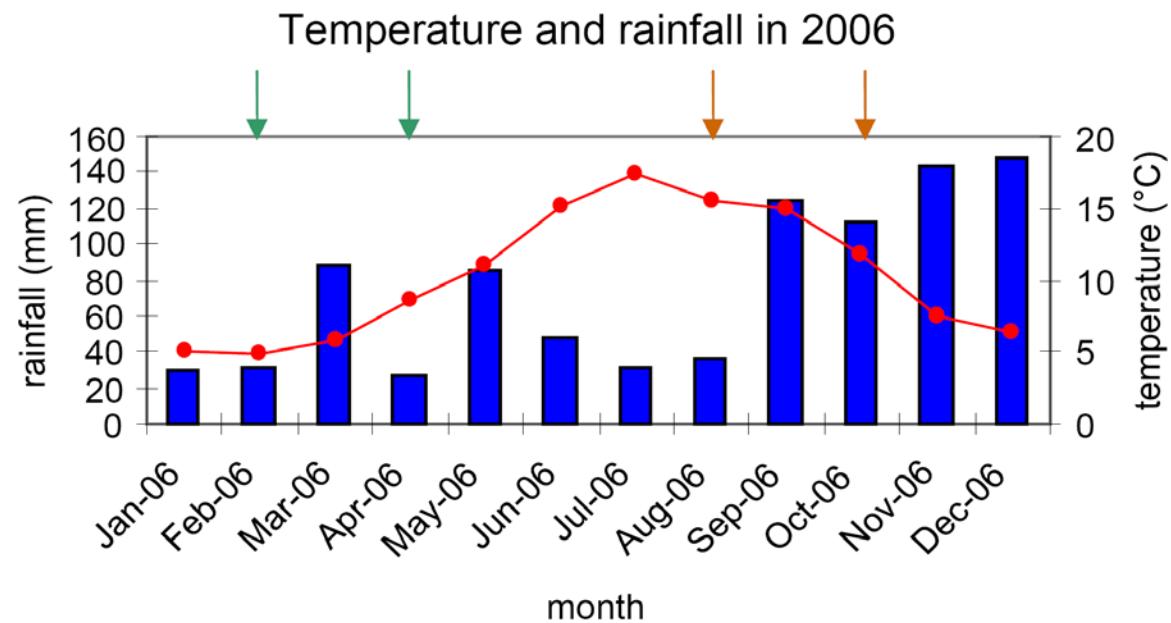


# Function and multifunction

- How many genes are there?
- 1990s: perhaps 100,000
- 2000: 25,000
- How does this give the range of functions and control?



# Formidable genetic and environmental interactions

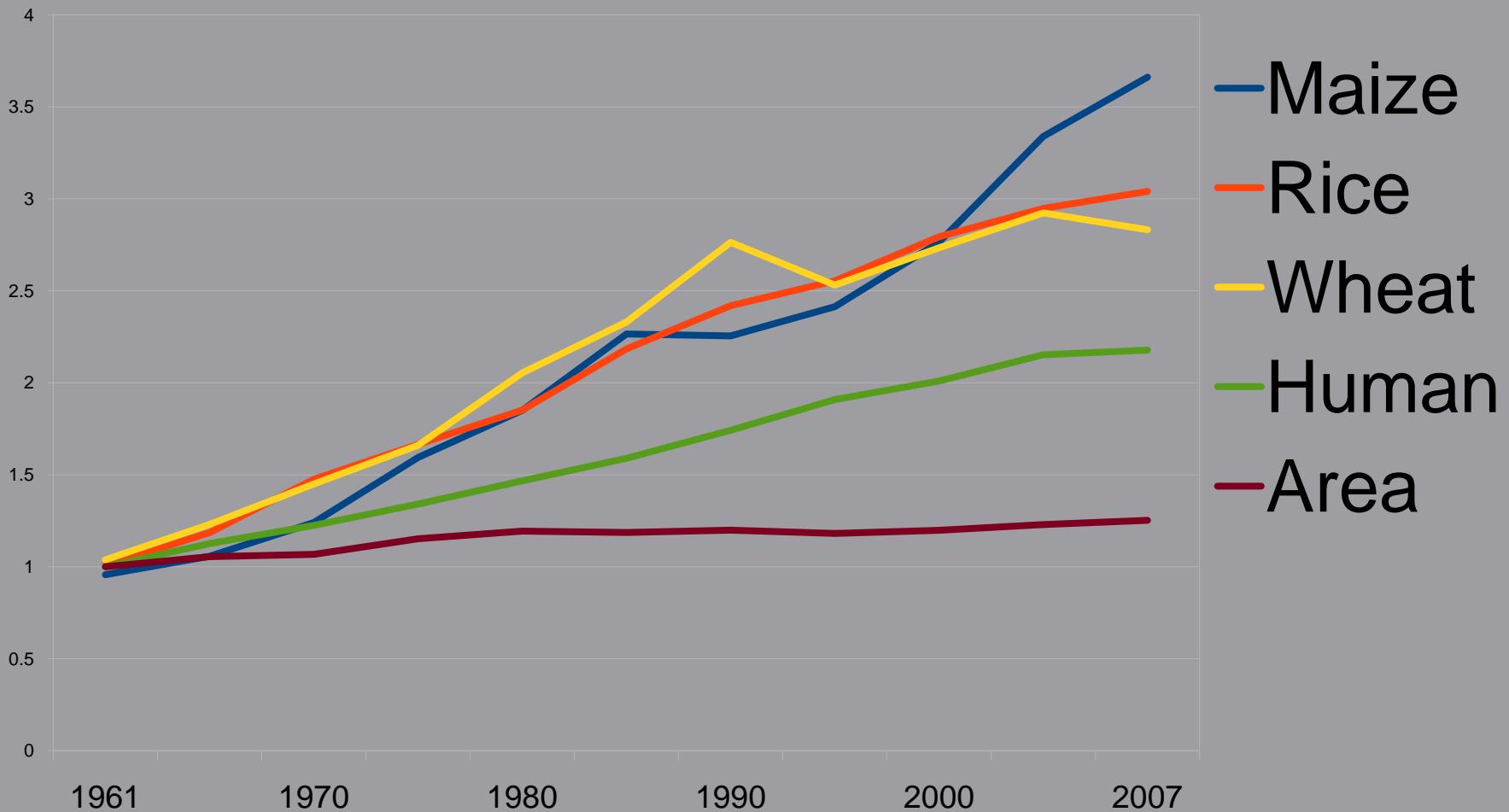


Anhalt, Barth, HH  
Euphytica 2009  
Theor App Gen 2008

# Nothing special about crop genomes?

Crop	Genome size	2n	Ploidy	Food
Rice	400 Mb	24	2	3x endosperm
Wheat	17,000 Mbp	42	6	3x endosperm
Maize	950 Mbp	10	4 (palaeo-tetraploid)	3x endosperm
Potato	900 Mbp	48	4	Modified leaf
Sugar beet	758 Mbp	18	2	Modified root
Cassava	770 Mbp	36	2	Tuber
Soybean	1,100 Mbp	40	4	Seed cotyledon
Oil palm	3,400 Mbp	32	2	Fruit mesocarp
Banana	500 Mbp	33	3	Fruit mesocarp

# 50 years of plant breeding progress



# United Nations

## Millennium Development Goals- MDGs

Goal 1 - Eradicate extreme poverty and hunger

Goal 2 - Achieve universal primary education

Goal 3 - Promote gender equity and empower women

Goal 4 - Reduce child mortality

Goal 5 - Improve maternal health

Goal 6- Combat HIV/AIDS, malaria and other diseases

Goal 7 - Ensure environmental sustainability

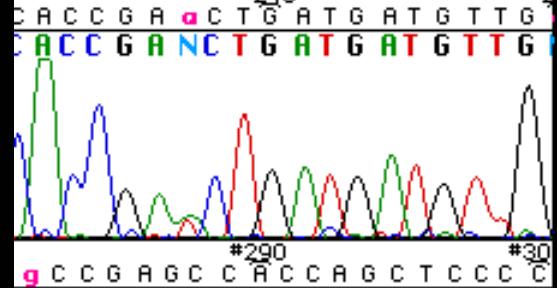
Goal 8 - Develop a global partnership for development





# Levels

- Sequences
- Genes / motifs
- Repetitive DNA
- Chromosomes
- Chromosome sets  
(‘Genomes’)
- Genotypes/CVs
- Species
- Genera and above



... involving

- Mutation
- Rearrangement
- Duplication
- Deletion
- Homogenization

... consequences for

- Crops / wild species
- Selection
- Speciation



# Genome evolution: extinction, continuation or explosion

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