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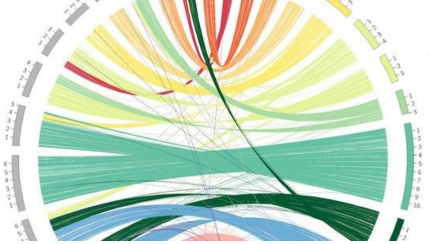
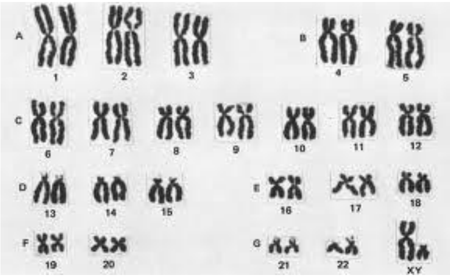


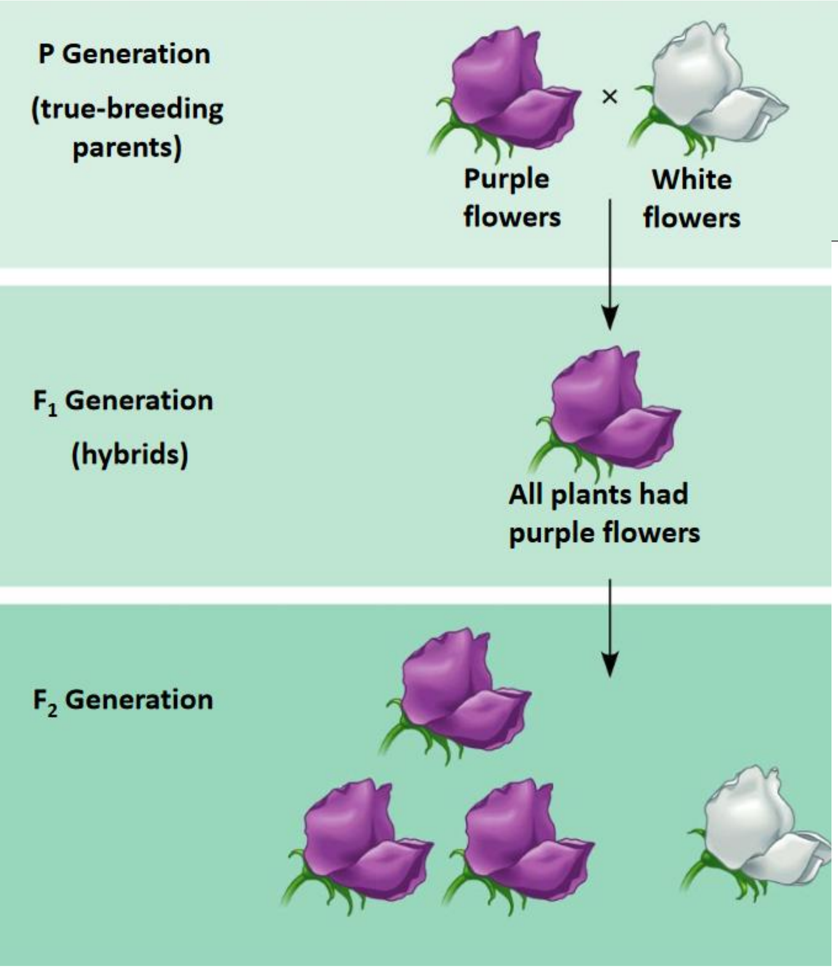
Curso Genomica

La transmisión, segregación y recombinación del material hereditario

9 setiembre 2024
Fernando Alvarez

La herencia en los organismos diploides: la leyes de Mendel, los cromosomas y la meiosis





Cuadrado de Punnet para predecir el resultado de un cruzamiento dihíbrido

Por la primera Ley de Mendel:

gametas Y = gametas y = 1/2

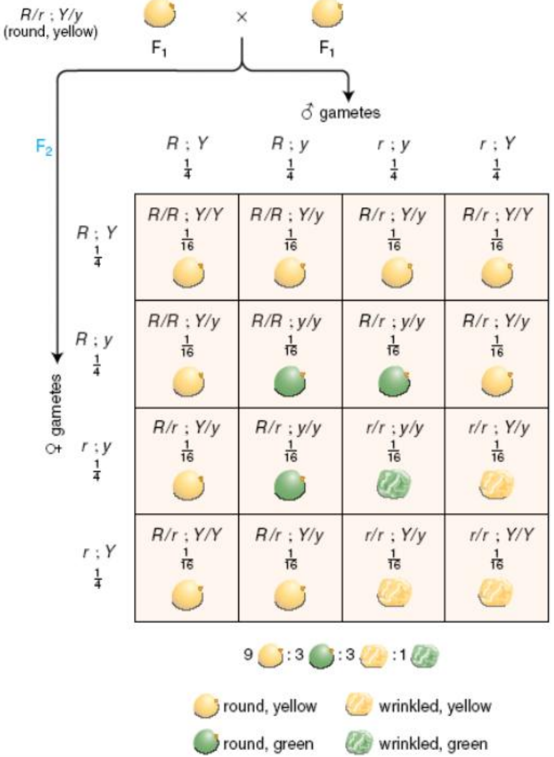
gametas R = gametas r = 1/2

p (RY) = 1/2 x 1/2 =1/4

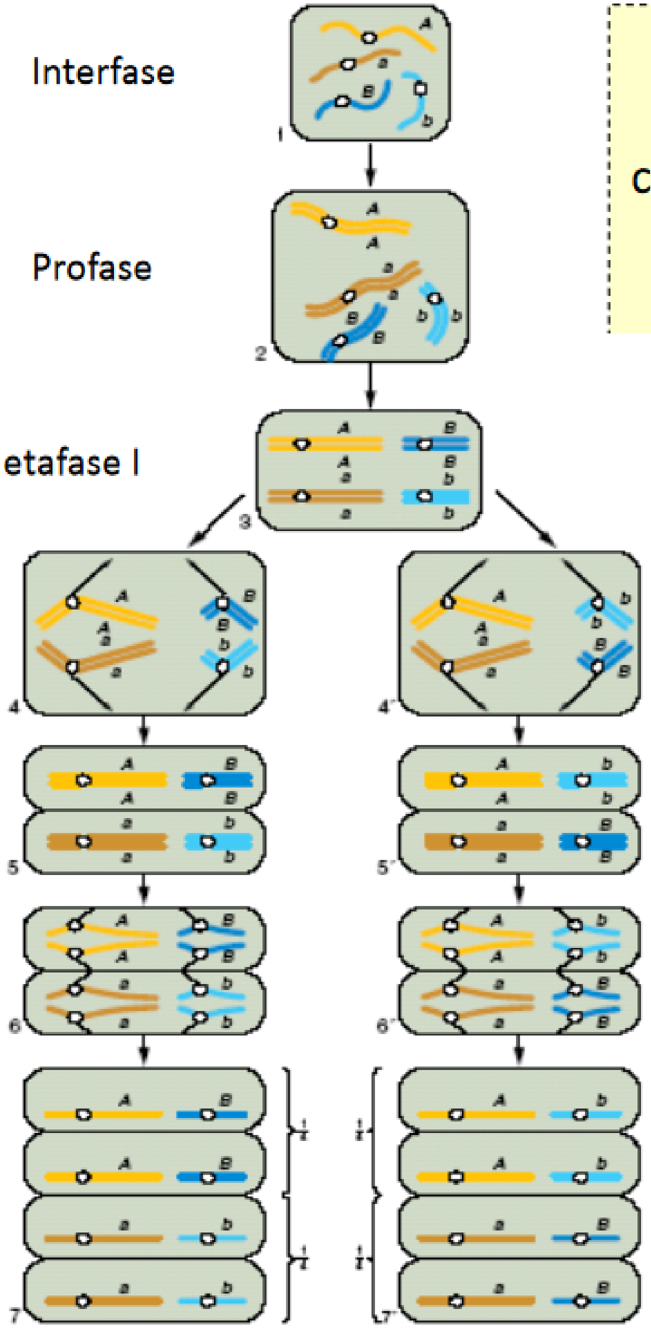
p (Ry) = 1/2 x 1/2 =1/4

p (rY) = 1/2 x 1/2 =1/4

p (ry) = 1/2 x 1/2 =1/4



Teoría cromosómica de la herencia (Sutton-Boveri): el paralelismo entre el comportamiento de los genes (Mendel) y los cromosomas llevó a pensar que los genes están situados en cromosomas.



Interfase

Profase

Metafase I

Anafase I

Telofase I

Anafase II

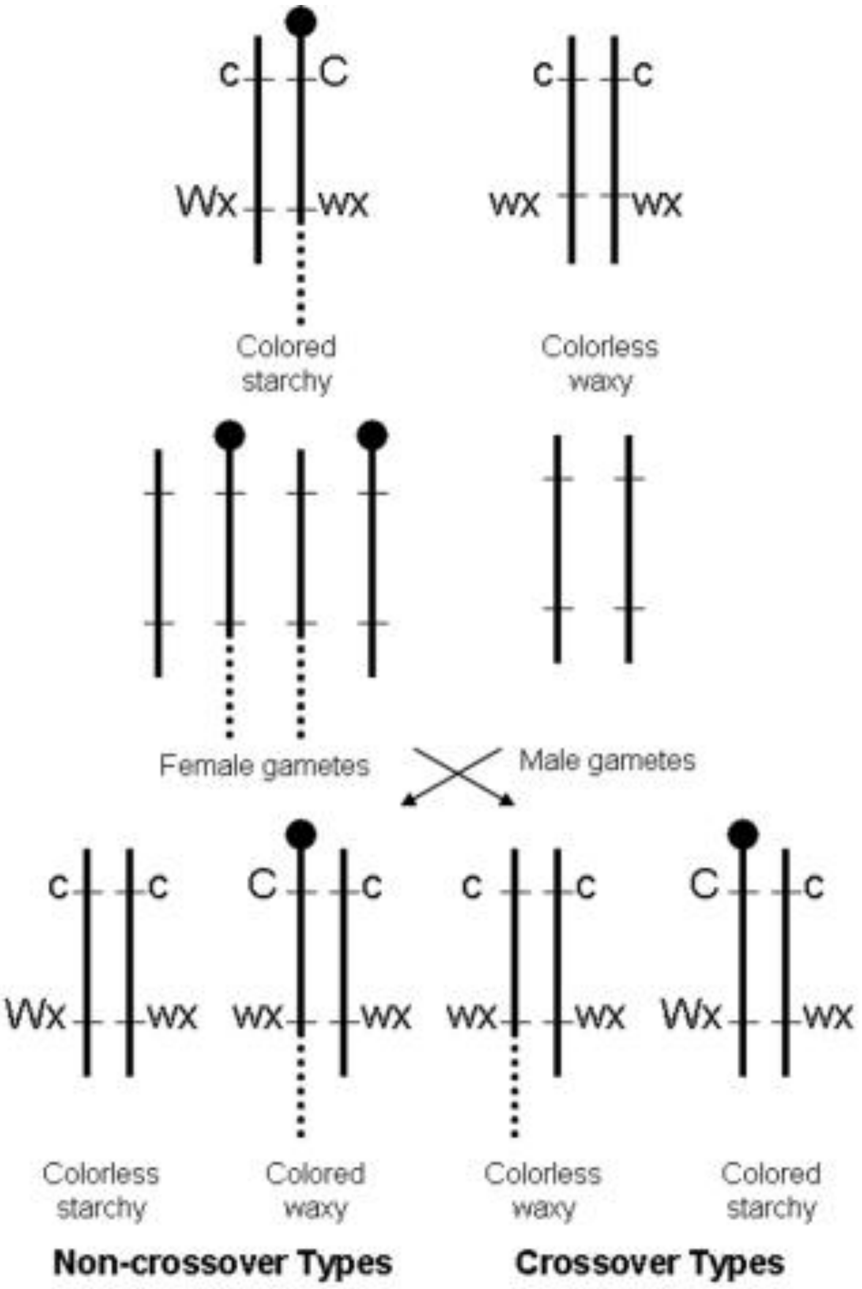
Telofase II

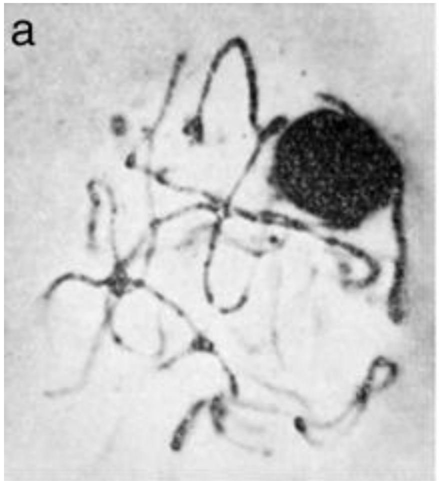
Explica la distribución igualitaria y la segregación independiente

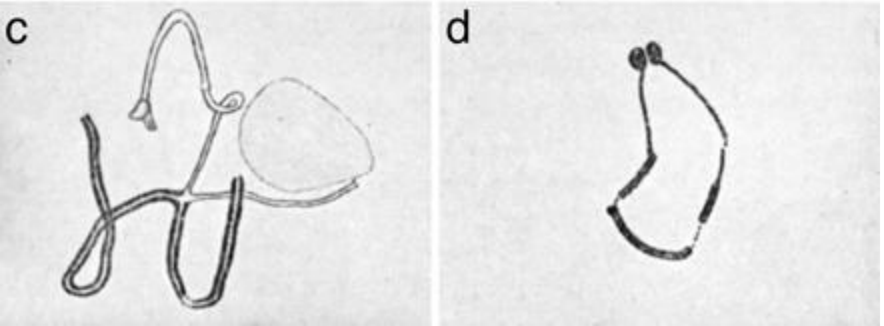
Meiosis de una célula diploide con genotipo A/a:B/b

Walter Sutton (left) and Theodor Boveri (right) independently developed the chromosome theory of inheritance in 1902.

Creighton, H.B., & McClintock, B. (1931)







Creighton, H.B., & McClintock, B. (1931). "A correlation of cytological and genetic crossing over in *Zea Mays*." *Proceedings of the National Academy of Sciences, 17*(8), 492-497.

Teoría cromosómica de la herencia

Origen de la Variación Genética que introduce la Meiosis

<u>- A nivel de la organización intracromosómica:</u> se forman nuevas variantes cromosómicas mediante **entrecruzamiento**.

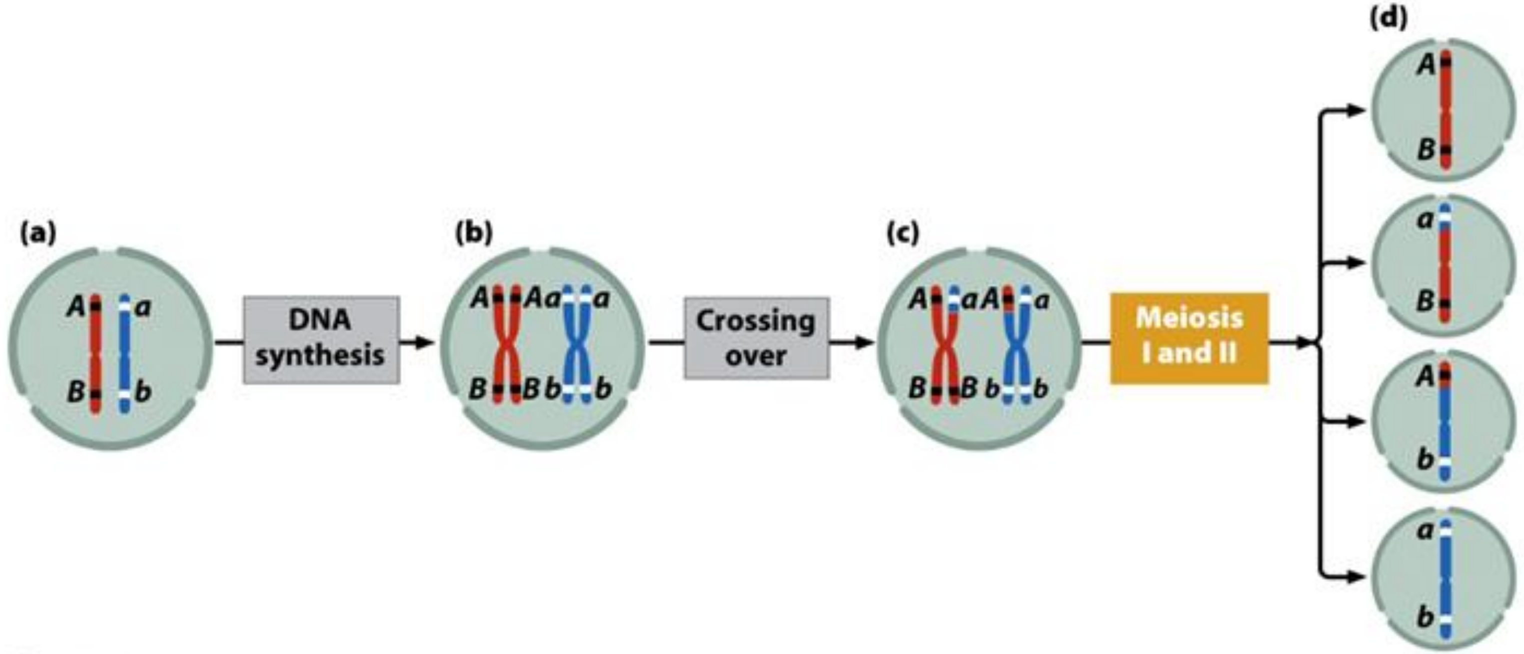
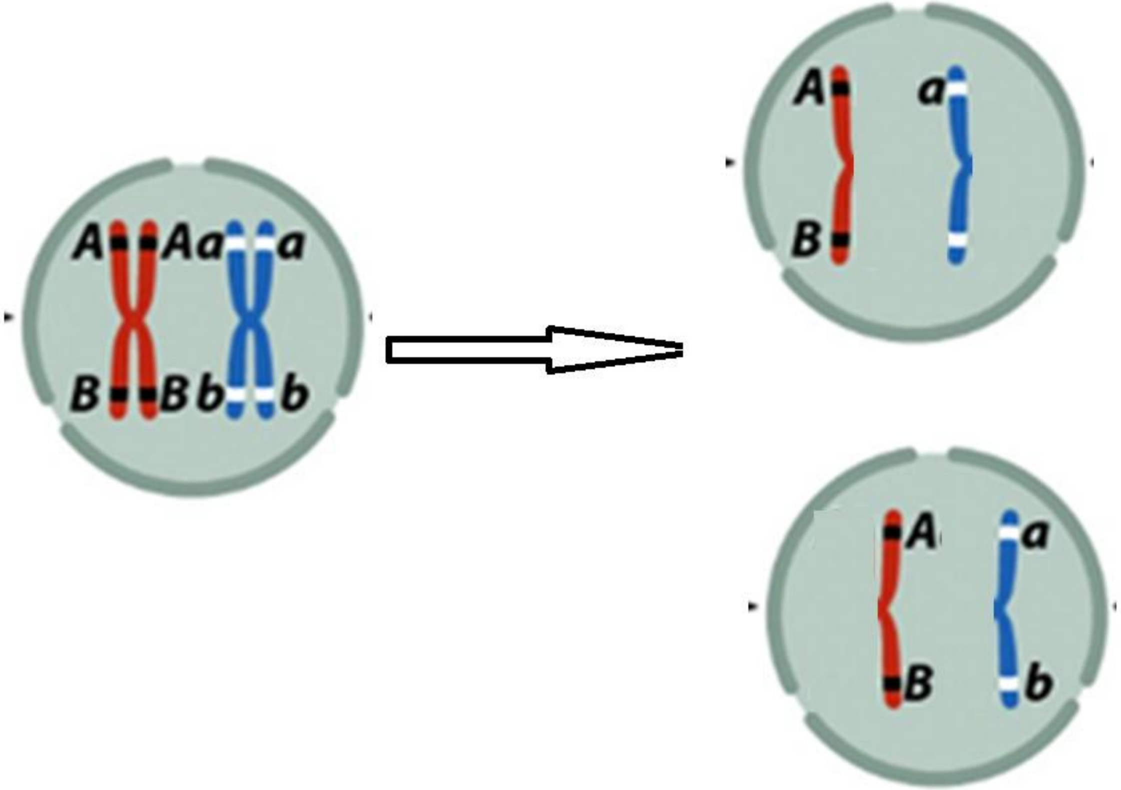


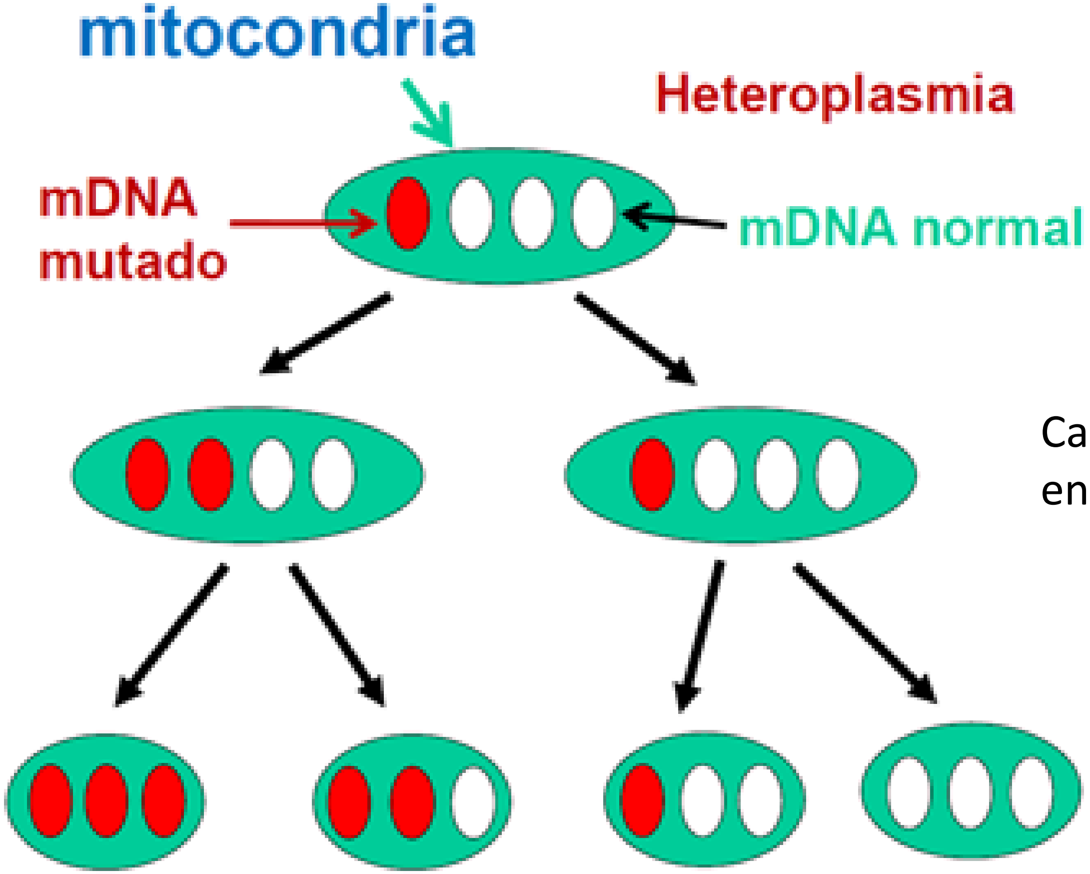
Figure 2-16
Genetics: A Conceptual Approach, Third Edition

Mitosis



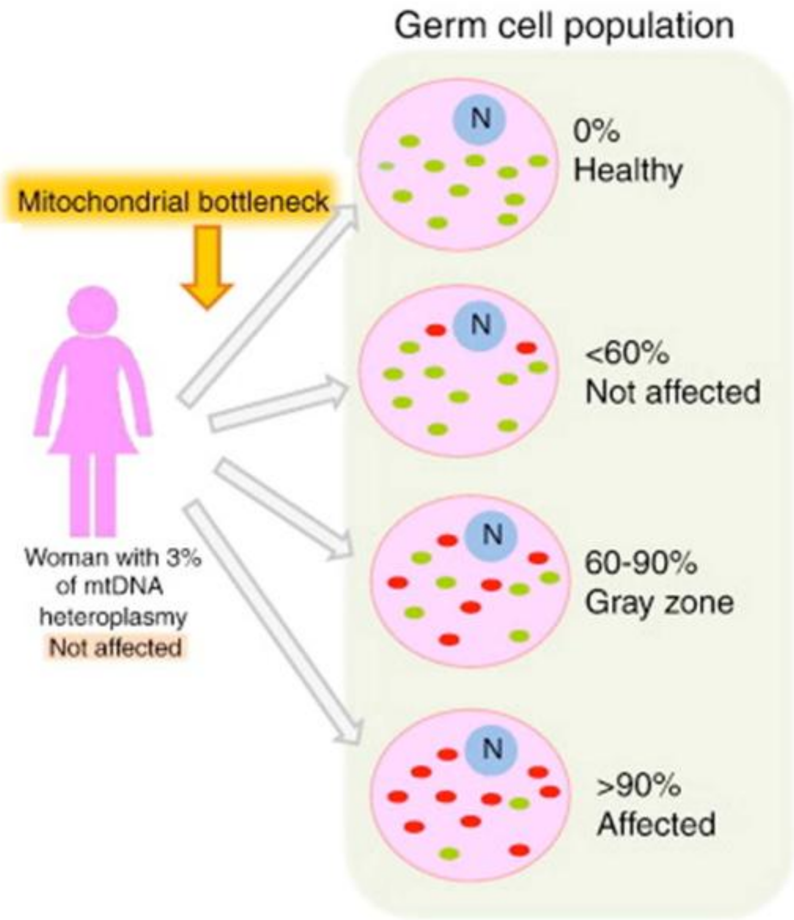
Ausencia de sistema de segregación preciso

Heteroplasmia y la segregación deriva mitocondrial



Cada mitocondria tiene entre 2 y 10 mtDNA s

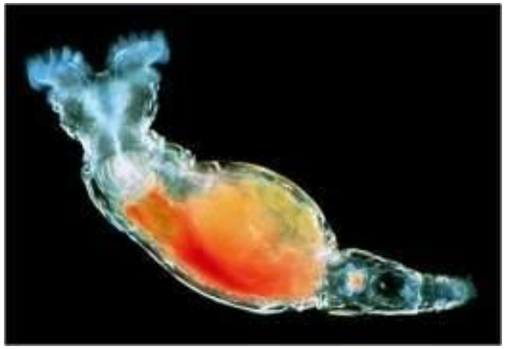
Cuello de botella materno

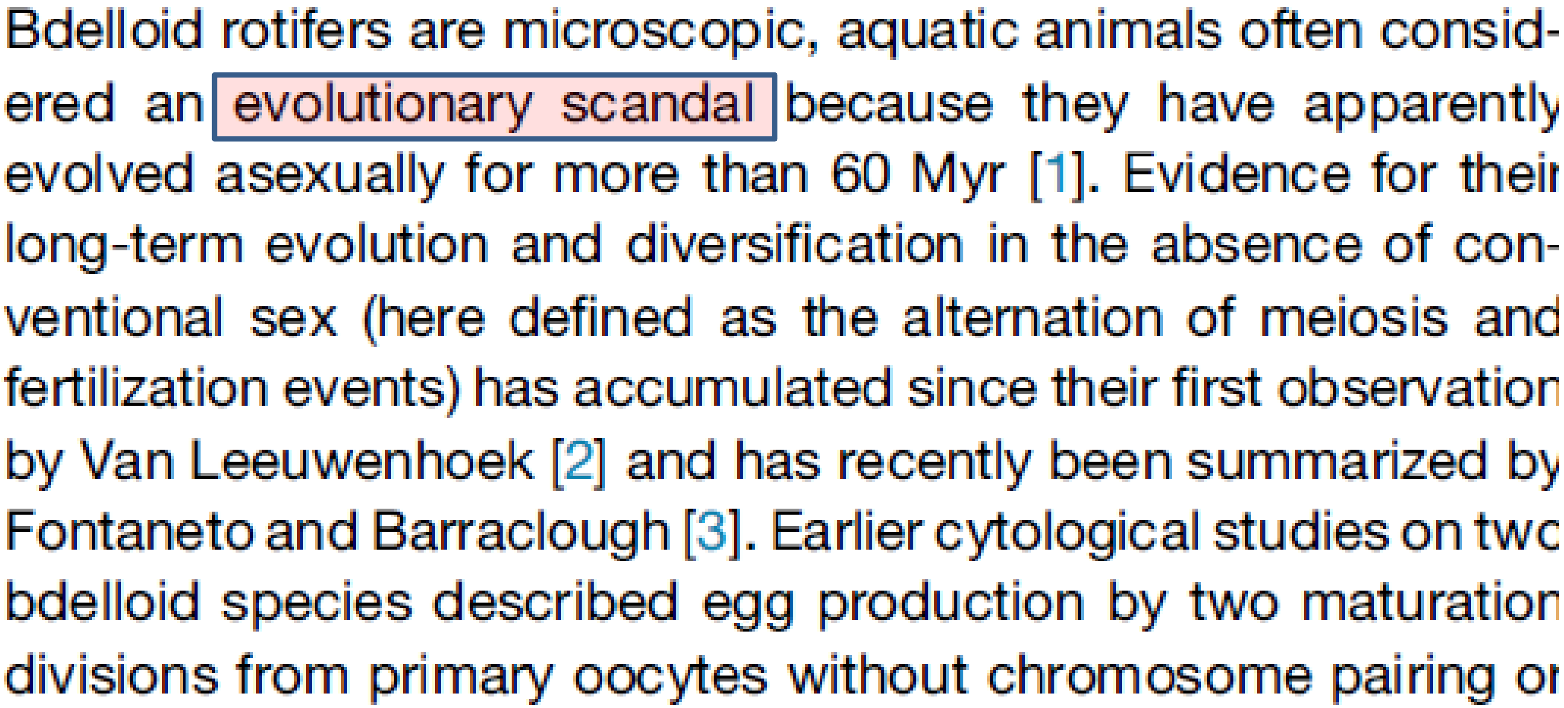


Yabuuchi et al. 2011 BBA 1820: 637

Ausencia de meiosis (recombinación)

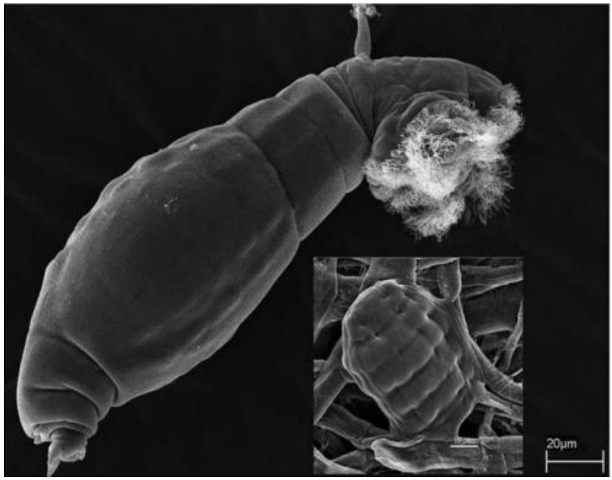
Los rotíferos delloideos:
un escándalo asexual





Bdelloid rotifers are microscopic, aquatic animals often considered an evolutionary scandal because they have apparently evolved asexually for more than 60 Myr [1]. Evidence for their long-term evolution and diversification in the absence of conventional sex (here defined as the alternation of meiosis and fertilization events) has accumulated since their first observation by Van Leeuwenhoek [2] and has recently been summarized by Fontaneto and Barraclough [3]. Earlier cytological studies on two bdelloid species described egg production by two maturation divisions from primary oocytes without chromosome pairing or

Los rotíferos delloideos: un escándalo asexual



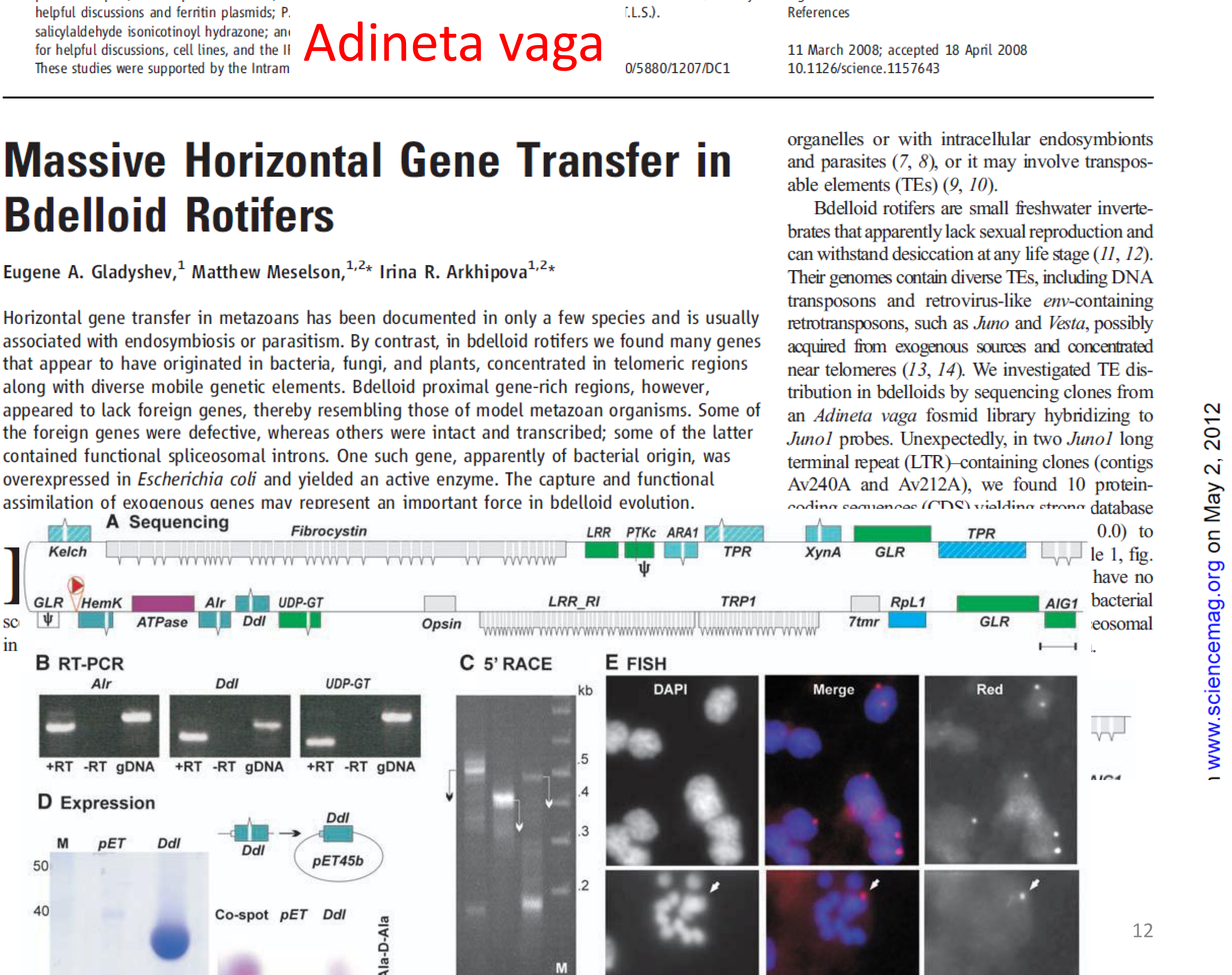
Bdelloid rotifers reproduce exclusively by parthenogenesis, whereby eggs are formed by 2 successive mitotic divisions of resting oocytes, in the absence of chromosome pairing or reduction in chromosome numbers

Deshidratacion y HGT

A second key feature is that most bdelloid species are remarkably resilient to desiccation, and can survive the loss of almost all cellular water at any stage in their life cycle, including as adults. On desiccation, animals contract their body into a flat, ellipsoid “tun” shape and enter a dormant state called anhydrobiosis, during which all metabolic activities associated with life are suspended . Individuals can remain in this condition for long periods, usually days or weeks but occasionally several years.

It may be that HGT is facilitated by membrane disruption and DNA fragmentation and repair associated with the repeated desiccation and recovery experienced in typical bdelloid habitats, allowing DNA in ingested or other environmental material to enter bdelloid genome

Adineta vaga



helpful discussions and ferritin plasmids; P.
salicylaldehyde isonicotinoyl hydrazone; an
for helpful discussions, cell lines, and the I
These studies were supported by the Intram

.L.S.).

0/5880/1207/DC1

References

11 March 2008; accepted 18 April 2008
10.1126/science.1157643

Massive Horizontal Gene Transfer in Bdelloid Rotifers

Eugene A. Gladyshev,[1] **Matthew Meselson,**[1,2]* **Irina R. Arkhipova**[1,2]*

Horizontal gene transfer in metazoans has been documented in only a few species and is usually associated with endosymbiosis or parasitism. By contrast, in bdelloid rotifers we found many genes that appear to have originated in bacteria, fungi, and plants, concentrated in telomeric regions along with diverse mobile genetic elements. Bdelloid proximal gene-rich regions, however, appeared to lack foreign genes, thereby resembling those of model metazoan organisms. Some of the foreign genes were defective, whereas others were intact and transcribed; some of the latter contained functional spliceosomal introns. One such gene, apparently of bacterial origin, was overexpressed in *Escherichia coli* and yielded an active enzyme. The capture and functional assimilation of exogenous genes may represent an important force in bdelloid evolution.

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organelles or with intracellular endosymbionts and parasites (*7*, *8*), or it may involve transposable elements (TEs) (*9*, *10*).

Bdelloid rotifers are small freshwater invertebrates that apparently lack sexual reproduction and can withstand desiccation at any life stage (*11*, *12*). Their genomes contain diverse TEs, including DNA transposons and retrovirus-like *env*-containing retrotransposons, such as *Juno* and *Vesta*, possibly acquired from exogenous sources and concentrated near telomeres (*13*, *14*). We investigated TE distribution in bdelloids by sequencing clones from an *Adineta vaga* fosmid library hybridizing to *Juno1* probes. Unexpectedly, in two *Juno1* long terminal repeat (LTR)–containing clones (contigs Av240A and Av212A), we found 10 protein-coding sequences (CDS) yielding strong database

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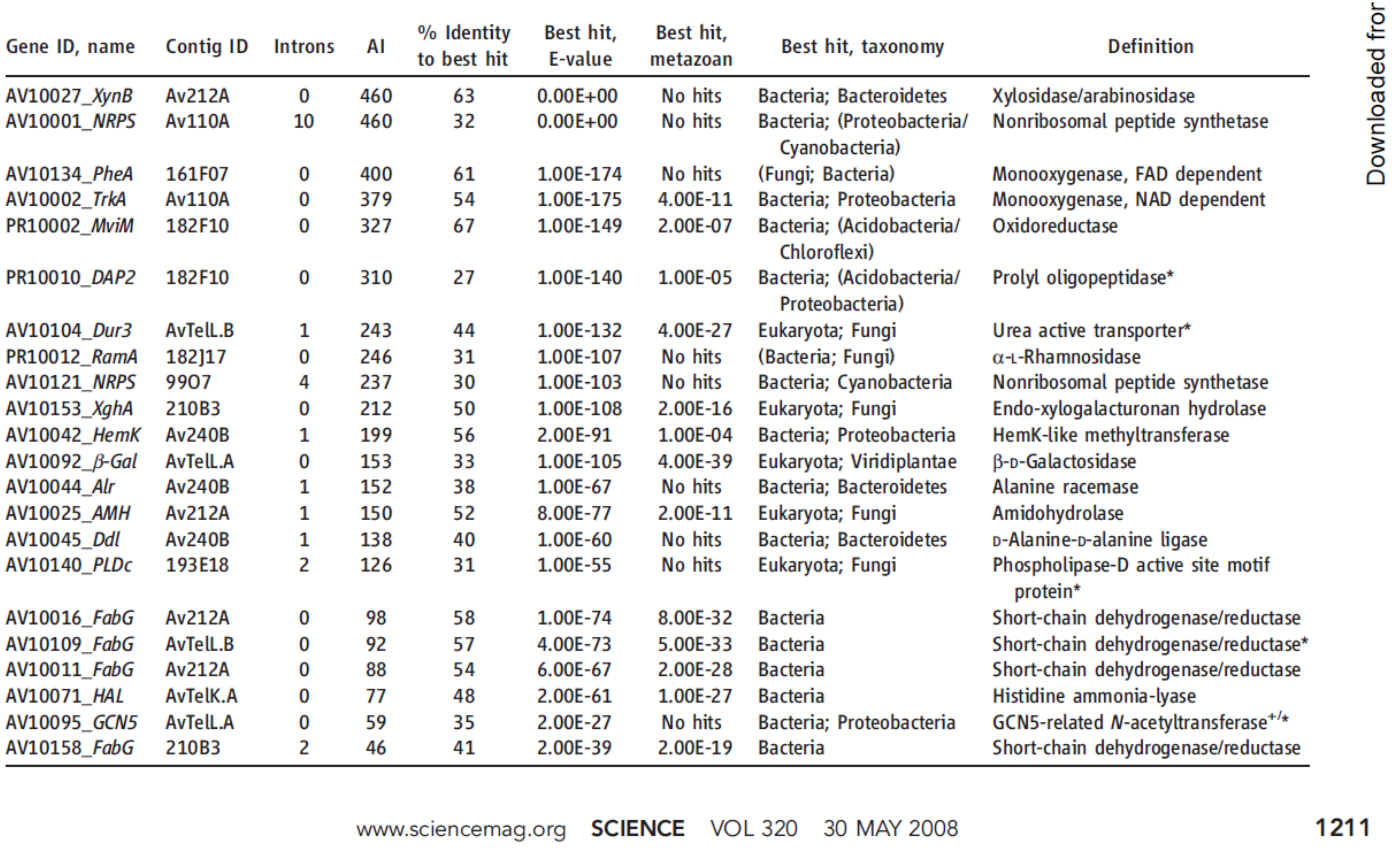
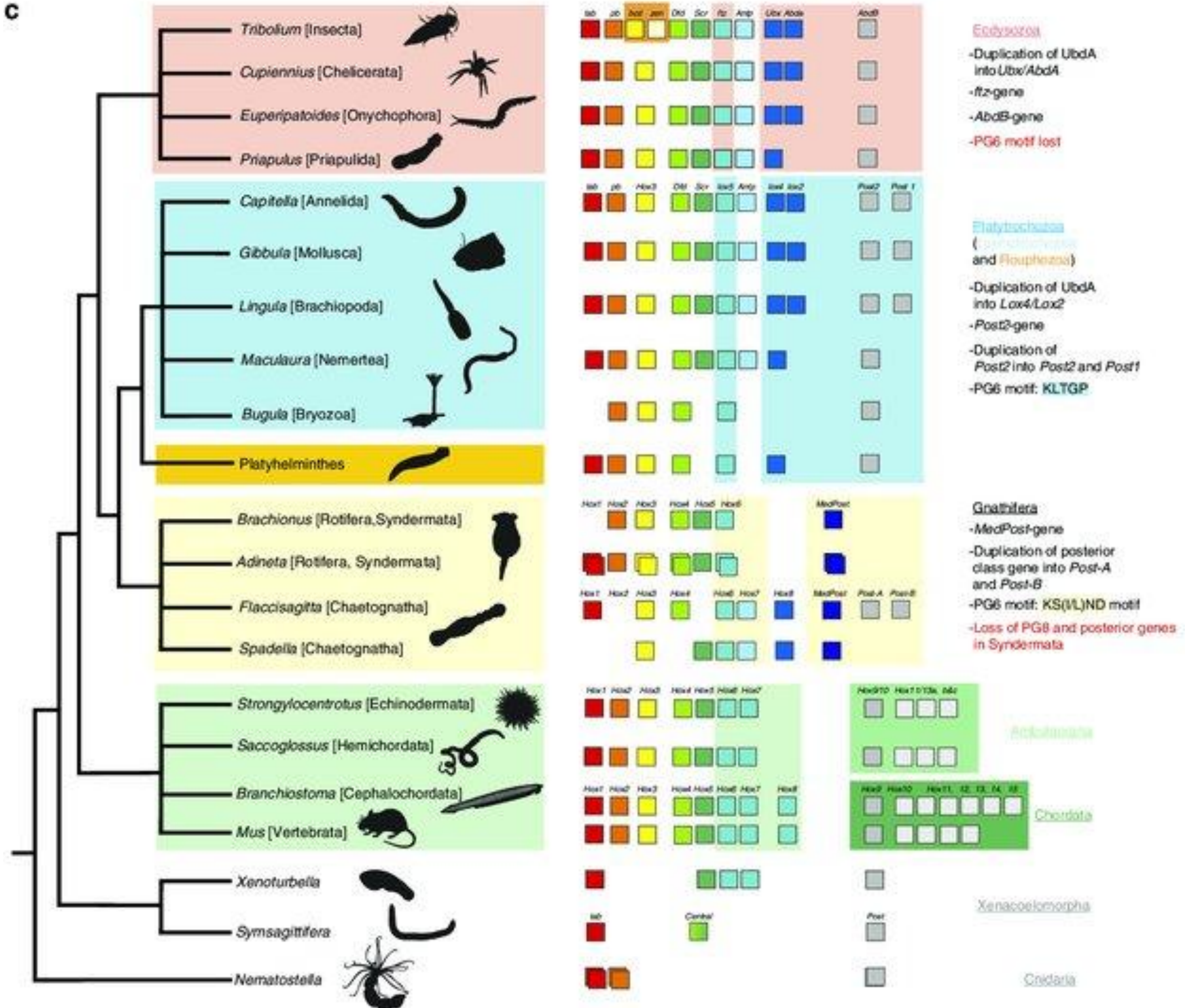
Tabla resultado de BLASTP



Gene ID, name	Contig ID	Introns	AI	% Identity to best hit	Best hit, E-value	Best hit, metazoan	Best hit, taxonomy	Definition
AV10027_*XynB*	Av212A	0	460	63	0.00E+00	No hits	Bacteria; Bacteroidetes	Xylosidase/arabinosidase
AV10001_*NRPS*	Av110A	10	460	32	0.00E+00	No hits	Bacteria; (Proteobacteria/ Cyanobacteria)	Nonribosomal peptide synthetase
AV10134_*PheA*	161F07	0	400	61	1.00E-174	No hits	(Fungi; Bacteria)	Monooxygenase, FAD dependent
AV10002_*TrkA*	Av110A	0	379	54	1.00E-175	4.00E-11	Bacteria; Proteobacteria	Monooxygenase, NAD dependent
PR10002_*MviM*	182F10	0	327	67	1.00E-149	2.00E-07	Bacteria; (Acidobacteria/ Chloroflexi)	Oxidoreductase
PR10010_*DAP2*	182F10	0	310	27	1.00E-140	1.00E-05	Bacteria; (Acidobacteria/ Proteobacteria)	Prolyl oligopeptidase*
AV10104_*Dur3*	AvTelL.B	1	243	44	1.00E-132	4.00E-27	Eukaryota; Fungi	Urea active transporter*
PR10012_*RamA*	182J17	0	246	31	1.00E-107	No hits	(Bacteria; Fungi)	α-L-Rhamnosidase
AV10121_*NRPS*	99O7	4	237	30	1.00E-103	No hits	Bacteria; Cyanobacteria	Nonribosomal peptide synthetase
AV10153_*XghA*	210B3	0	212	50	1.00E-108	2.00E-16	Eukaryota; Fungi	Endo-xylogalacturonan hydrolase
AV10042_*HemK*	Av240B	1	199	56	2.00E-91	1.00E-04	Bacteria; Proteobacteria	HemK-like methyltransferase
AV10092_*β-Gal*	AvTelL.A	0	153	33	1.00E-105	4.00E-39	Eukaryota; Viridiplantae	β-D-Galactosidase
AV10044_*Alr*	Av240B	1	152	38	1.00E-67	No hits	Bacteria; Bacteroidetes	Alanine racemase
AV10025_*AMH*	Av212A	1	150	52	8.00E-77	2.00E-11	Eukaryota; Fungi	Amidohydrolase
AV10045_*Ddl*	Av240B	1	138	40	1.00E-60	No hits	Bacteria; Bacteroidetes	D-Alanine-D-alanine ligase
AV10140_*PLDc*	193E18	2	126	31	1.00E-55	No hits	Eukaryota; Fungi	Phospholipase-D active site motif protein*
AV10016_*FabG*	Av212A	0	98	58	1.00E-74	8.00E-32	Bacteria	Short-chain dehydrogenase/reductase
AV10109_*FabG*	AvTelL.B	0	92	57	4.00E-73	5.00E-33	Bacteria	Short-chain dehydrogenase/reductase*
AV10011_*FabG*	Av212A	0	88	54	6.00E-67	2.00E-28	Bacteria	Short-chain dehydrogenase/reductase
AV10071_*HAL*	AvTelK.A	0	77	48	2.00E-61	1.00E-27	Bacteria	Histidine ammonia-lyase
AV10095_*GCN5*	AvTelL.A	0	59	35	2.00E-27	No hits	Bacteria; Proteobacteria	GCN5-related *N*-acetyltransferase$^{+/*}$
AV10158_*FabG*	210B3	2	46	41	2.00E-39	2.00E-19	Bacteria	Short-chain dehydrogenase/reductase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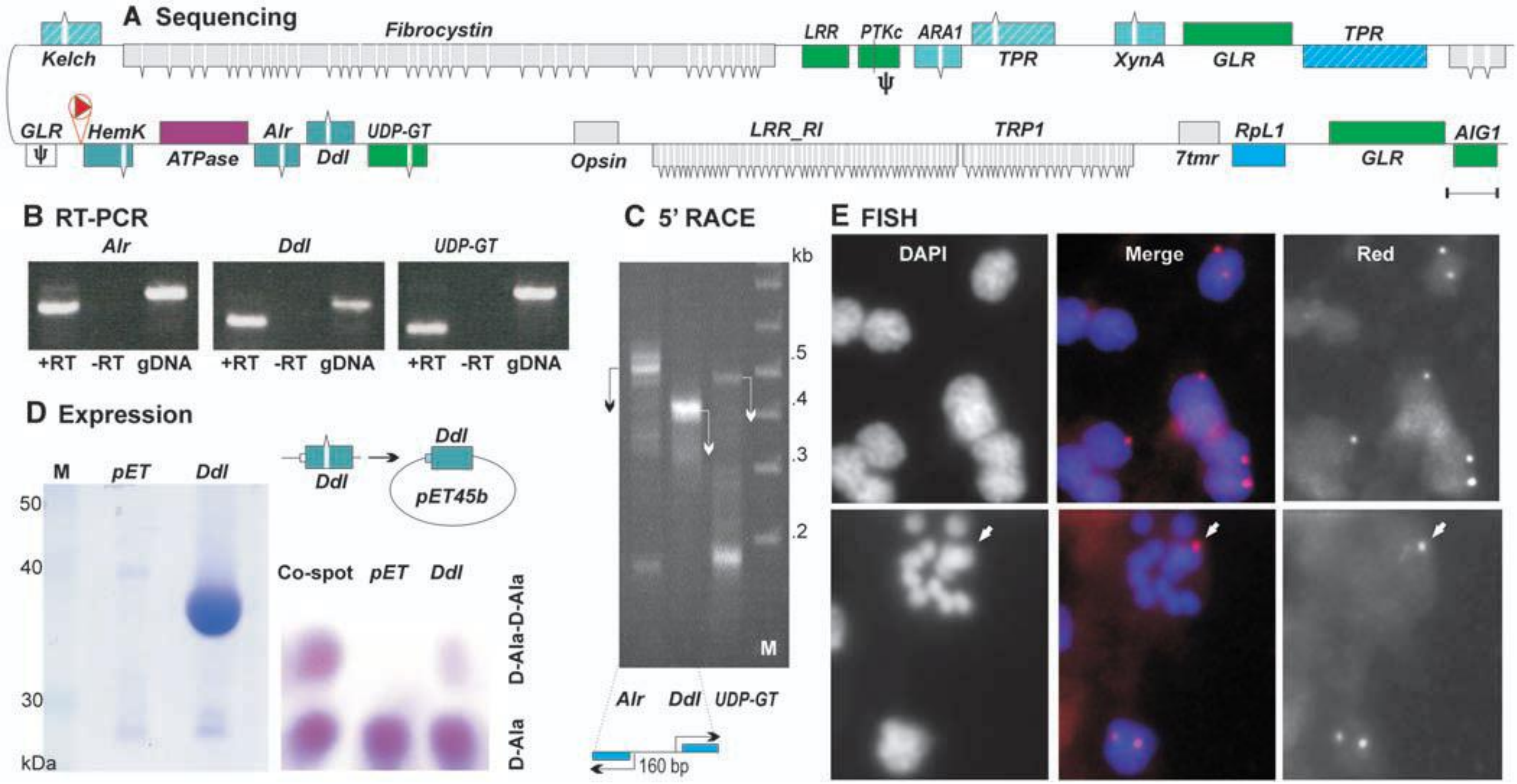
Tribolium [Insecta]
Cupiennius [Chelicerata]
Euperipatoides [Onychophora]
Priapulus [Priapulida]
Capitella [Annelida]
Gibbula [Mollusca]
Lingula [Brachiopoda]
Maculaura [Nemertea]
Bugula [Bryozoa]
Platyhelminthes
Brachionus [Rotifera,Syndermata]
Adineta [Rotifera, Syndermata]
Flaccisagitta [Chaetognatha]
Spadella [Chaetognatha]
Strongylocentrotus [Echinodermata]
Saccoglossus [Hemichordata]
Branchiostoma [Cephalochordata]
Mus [Vertebrata]
Xenoturbella
Symsagittifera
Nematostella
lab
pb
bcd
zen
Dfd
Scr
ftz
Antp
Ubx
Abda
AbdB
lab
pb
Hox3
Dfd
Scr
lox5
Antp
lox4
lox2
Post2
Post 1
Hox1
Hox2
Hox3
Hox4
Hox5
Hox6
MedPost
Hox1
Hox2
Hox3
Hox4
Hox6
Hox7
Hox8
MedPost
Post-A
Post-B
Hox1
Hox2
Hox3
Hox4
Hox5
Hox6
Hox7
Hox9/10
Hox11/13a
b&c
Hox1
Hox2
Hox3
Hox4
Hox5
Hox6
Hox7
Hox8
Hox9
Hox10
Hox11
12
13
14
15
lab
Central
Post
Ecdysozoa
-Duplication of UbdA into Ubx/AbdA
-ftz-gene
-AbdB-gene
-PG6 motif lost
Platytrochozoa
(
and Rouphozoa)
-Duplication of UbdA into Lox4/Lox2
-Post2-gene
-Duplication of Post2 into Post2 and Post1
-PG6 motif: KLTGP
Gnathifera
-MedPost-gene
-Duplication of posterior class gene into Post-A and Post-B
-PG6 motif: KS(I/L)ND motif
-Loss of PG8 and posterior genes in Syndermata
Ambulacraria
Chordata
Xenacoelomorpha
Cnidaria

Contigs subtelomericos contiene genes adquiridos por HTG en ***Adineta vaga***



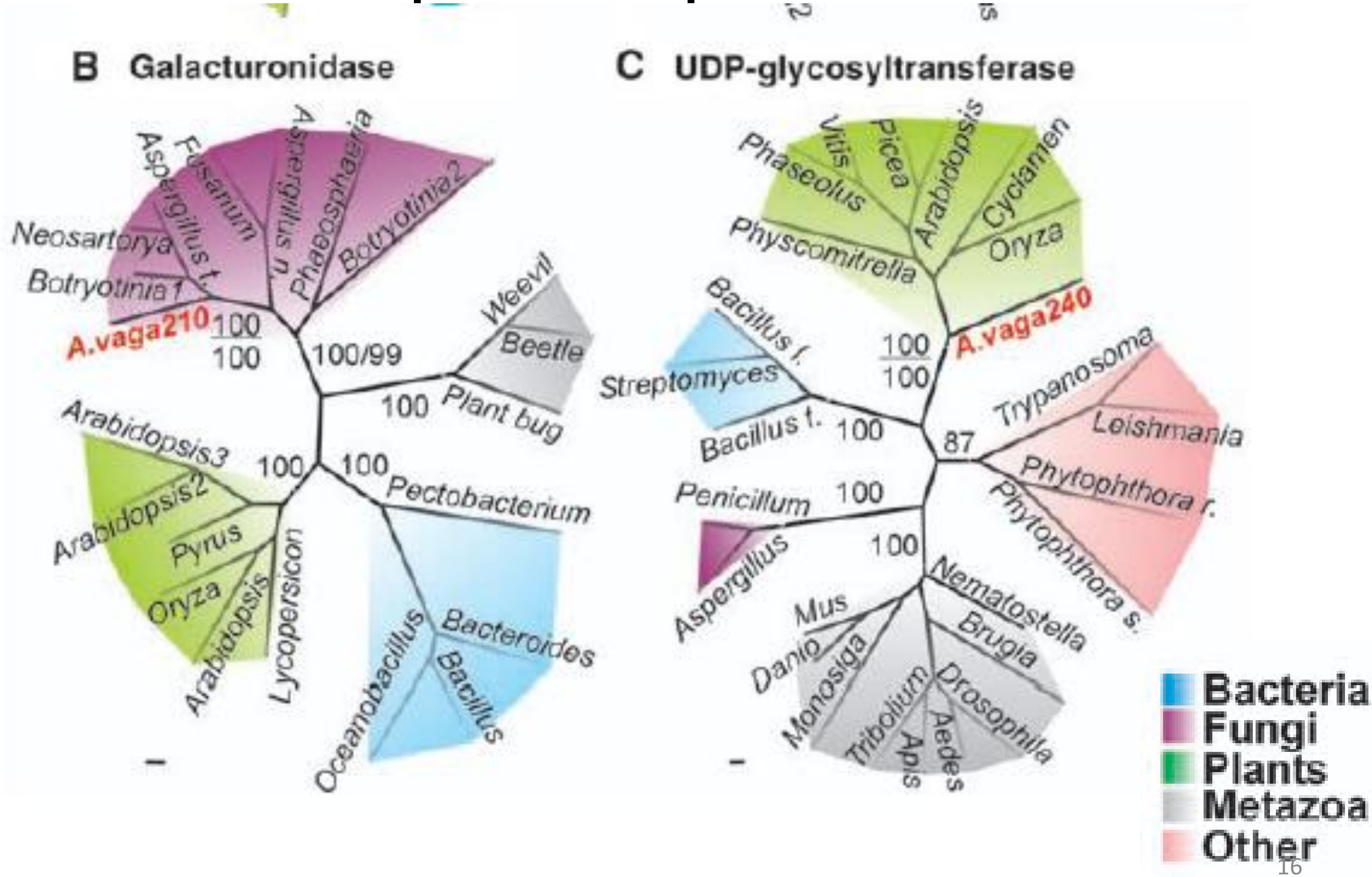
⬜ Origen de metazoario

🟦 Origen bacteriano

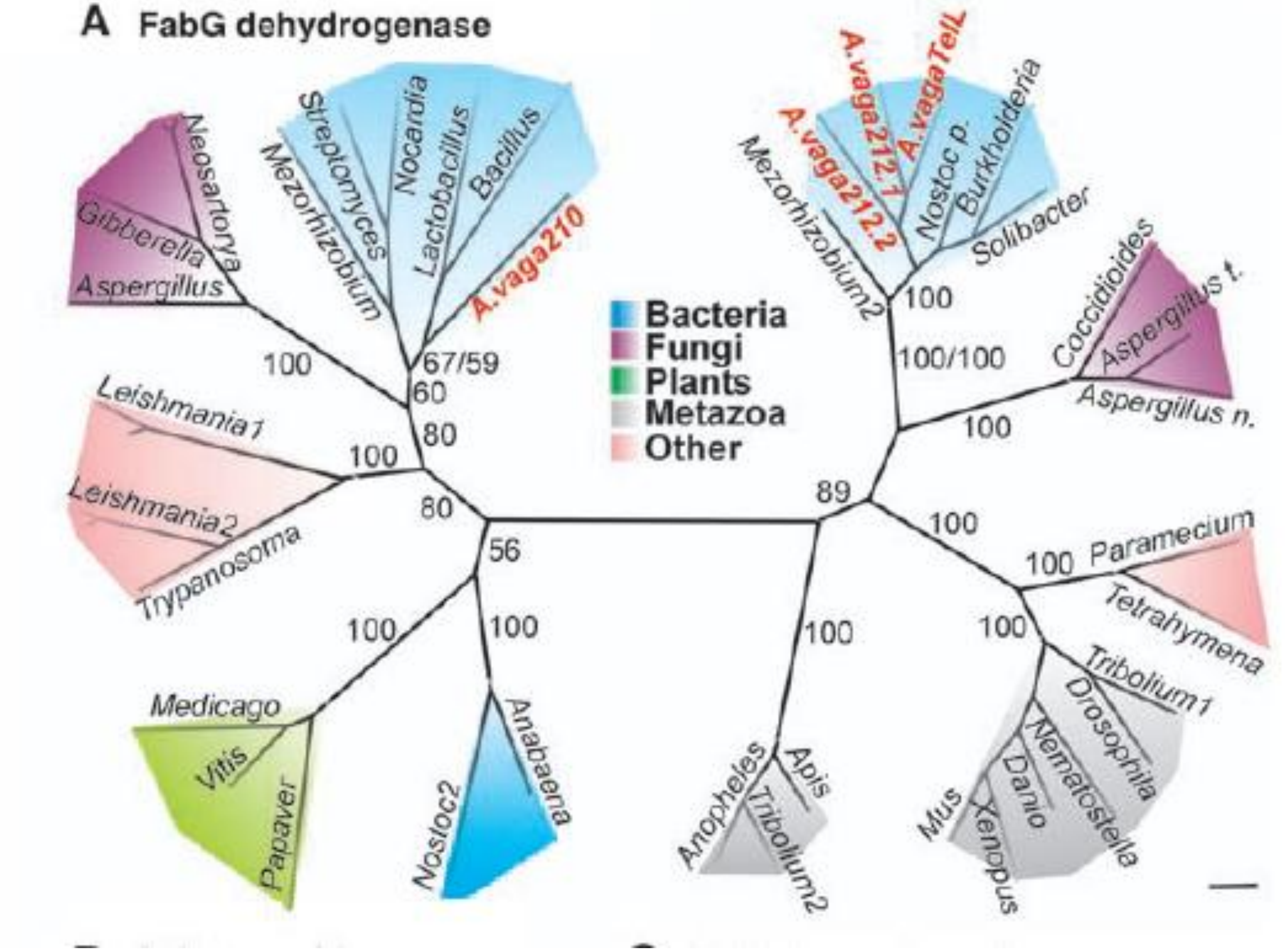
🟪 Origen de hongos

🟩 Origen de plantas

Filogenias de genes presumiblemente adquiridos por HGT

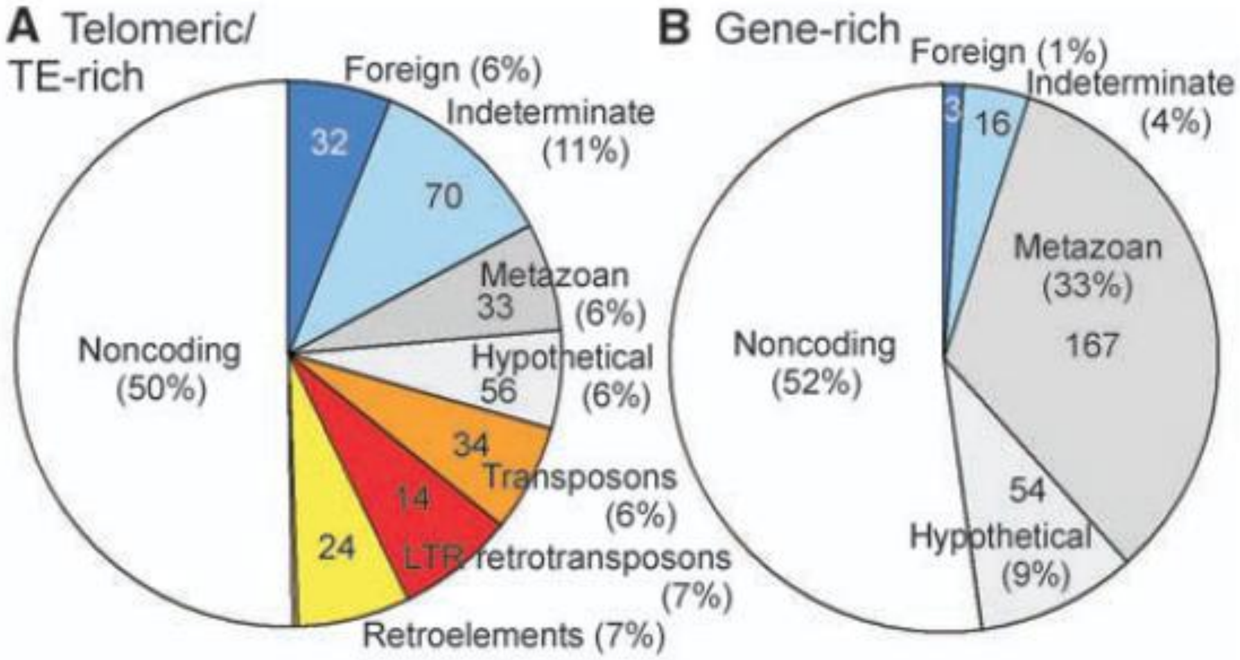


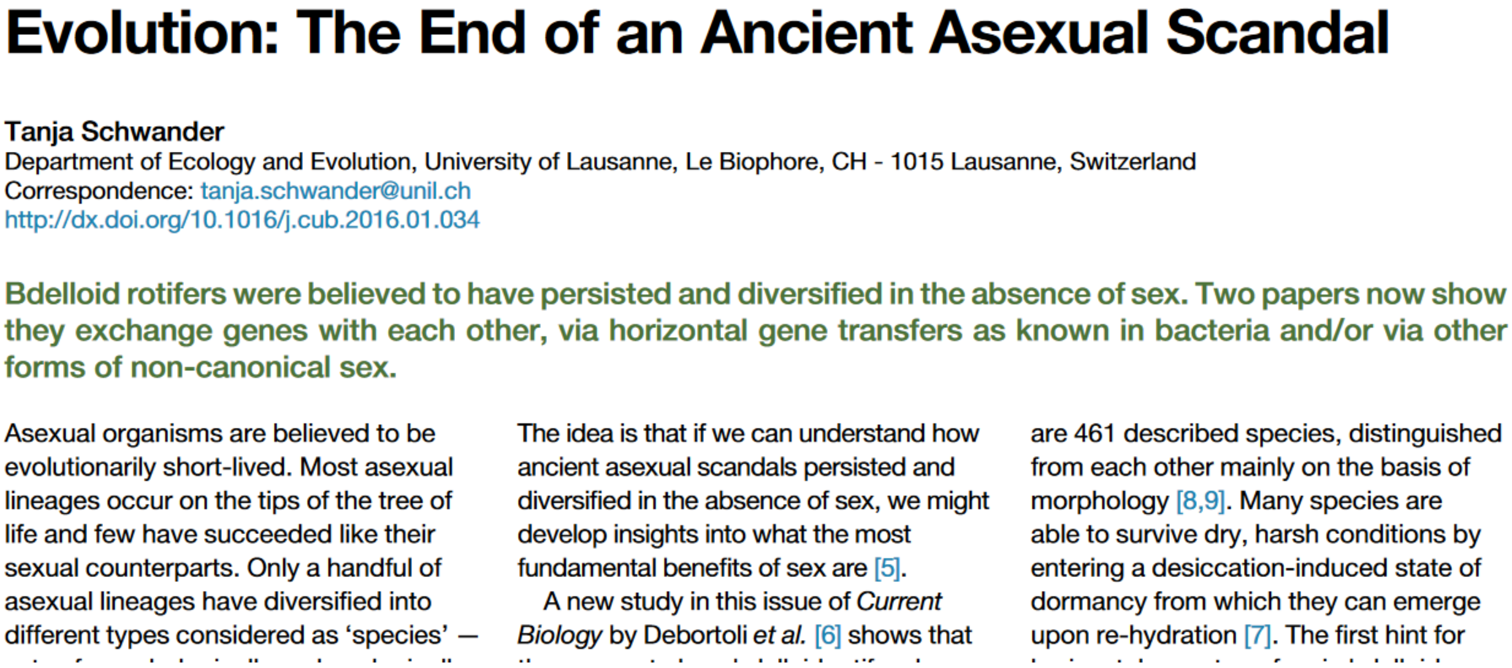
Filogenia de FabG presumiblemente adquirida por HGT



Los genes adquiridos mediante HGT presentan ubicación preferentemente telomérica

Fig. 3. Comparison of bdelloid telomeric/TE-rich regions (**A**) with bdelloid gene-rich regions (**B**) and with other model invertebrates (**C**). Pie charts were built with 921,903 base pairs (bp) (A) and 661,316 bp (B) of genomic DNA, respectively (excluding one of the two colinear partners). For TE count, ~1.3 Mb of genomic DNA in each data set was analyzed (including both colinear partners). The size of each sector corresponds to the percentage of the total length





Evolution: The End of an Ancient Asexual Scandal

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http://dx.doi.org/10.1016/j.cub.2016.01.034

Bdelloid rotifers were believed to have persisted and diversified in the absence of sex. Two papers now show they exchange genes with each other, via horizontal gene transfers as known in bacteria and/or via other forms of non-canonical sex.

Asexual organisms are believed to be evolutionarily short-lived. Most asexual lineages occur on the tips of the tree of life and few have succeeded like their sexual counterparts. Only a handful of asexual lineages have diversified into different types considered as 'species' —

The idea is that if we can understand how ancient asexual scandals persisted and diversified in the absence of sex, we might develop insights into what the most fundamental benefits of sex are [5].

A new study in this issue of *Current Biology* by Debortoli *et al.* [6] shows that

are 461 described species, distinguished from each other mainly on the basis of morphology [8,9]. Many species are able to survive dry, harsh conditions by entering a desiccation-induced state of dormancy from which they can emerge upon re-hydration [7]. The first hint for

Article

CellPress

Genetic Exchange among Bdelloid Rotifers Is More Likely Due to Horizontal Gene Transfer Than to Meiotic Sex

Nicolas Debortoli,[1] Xiang Li,[1] Isobel Eyres,[2] Diego Fontaneto,[3] Boris Hespeels,[1] Cuong Q. Tang,[2,4] Jean-François Flot,[5,*] and Karine Van Doninck[1,*]
[1]Laboratory of Evolutionary Genetics and Ecology, URBE, NAXYS, University of Namur, Rue de Bruxelles 61, 5000 Namur, Belgium
[2]Department of Life Sciences, Imperial College London, Silwood Park Campus, Ascot, Berkshire SL5 7PY, UK
[3]Institute of Ecosystem Study, National Research Council, Largo Tonolli 50, 28922 Verbania Pallanza, Italy
[4]Natural History Museum, Cromwell Road, London SW7 5BD, UK
[5]Evolutionary Biology & Ecology, Université Libre de Bruxelles, C.P. 160/12, Avenue F.D. Roosevelt 50, 1050 Bruxelles, Belgium
*Correspondence: jflot@ulb.ac.be (J.-F.F.), karine.vandoninck@unamur.be (K.V.D.)
http://dx.doi.org/10.1016/j.cub.2016.01.031

SUMMARY

Although strict asexuality is supposed to be an evolutionary dead end, morphological, cytogenetic, and genomic data suggest that bdelloid rotifers, a clade of microscopic animals, have persisted and diversified for more than 60 Myr in an ameiotic fashion. Moreover, the genome of bdelloids of the genus *Adineta* comprises 8%–10% of genes of putative non-metazoan origin, indicating that horizontal

reduction in chromosome number [4, 5]. This absence of meiosis was corroborated by the recent publication of the draft genome of *Adineta vaga*, which appears devoid of homologous chromosomes, hence ruling out the possibility of conventional meiosis [6]. However, these results could not dismiss the presence of alternative mechanisms of genetic exchange among bdelloid rotifers.

The idea that bdelloids acquire genes horizontally was first suggested by the observation that 8%–10% of the genes found within *Adineta*'s genome [6, 7] and transcriptome [8] are of putative non-metazoan origin, indicating that bdelloids are receptive

Bebortoli et al 2016

Haplogrupos de COI (576 individuos, 40 haplogrupos 6 especies cripticas A-F)

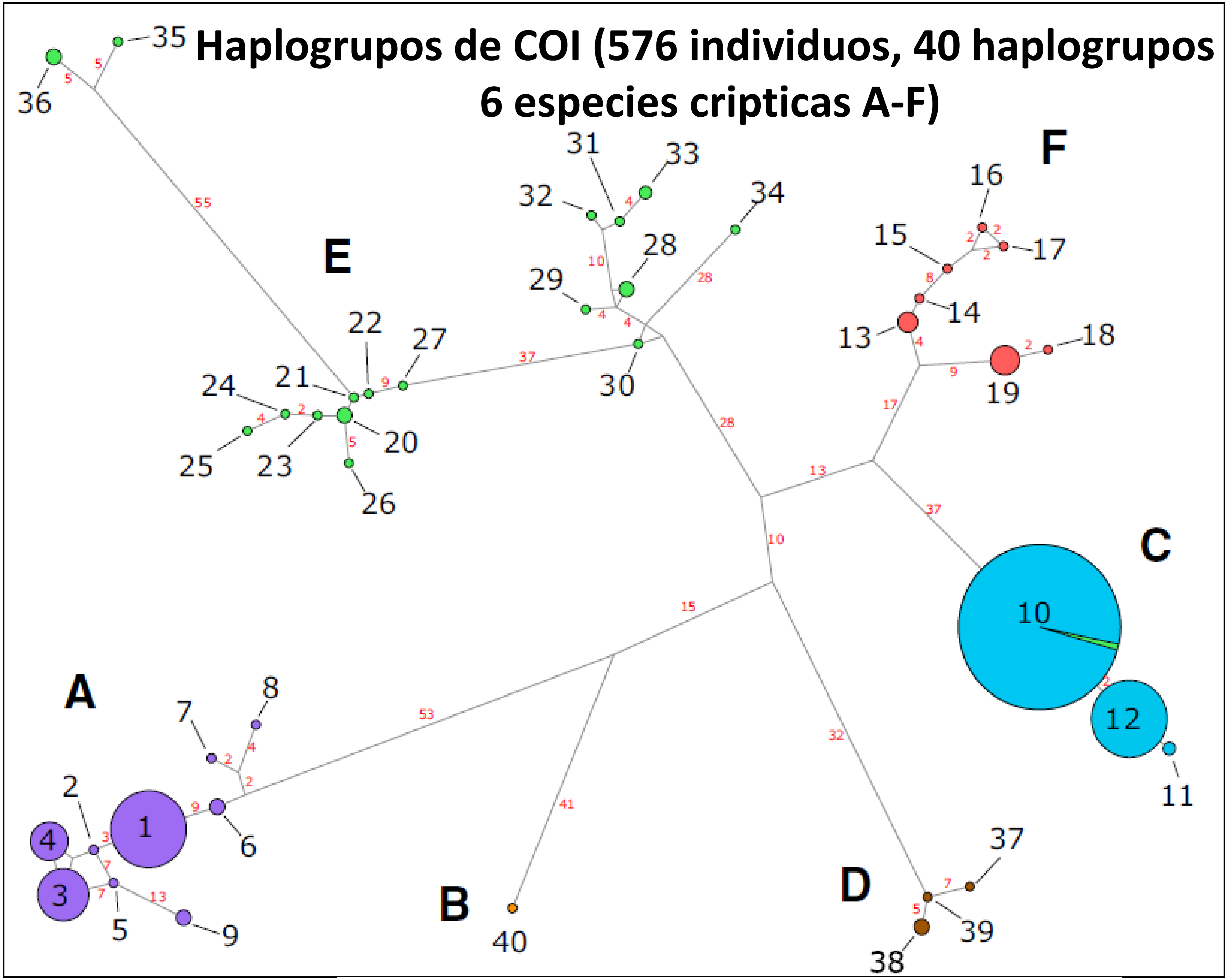
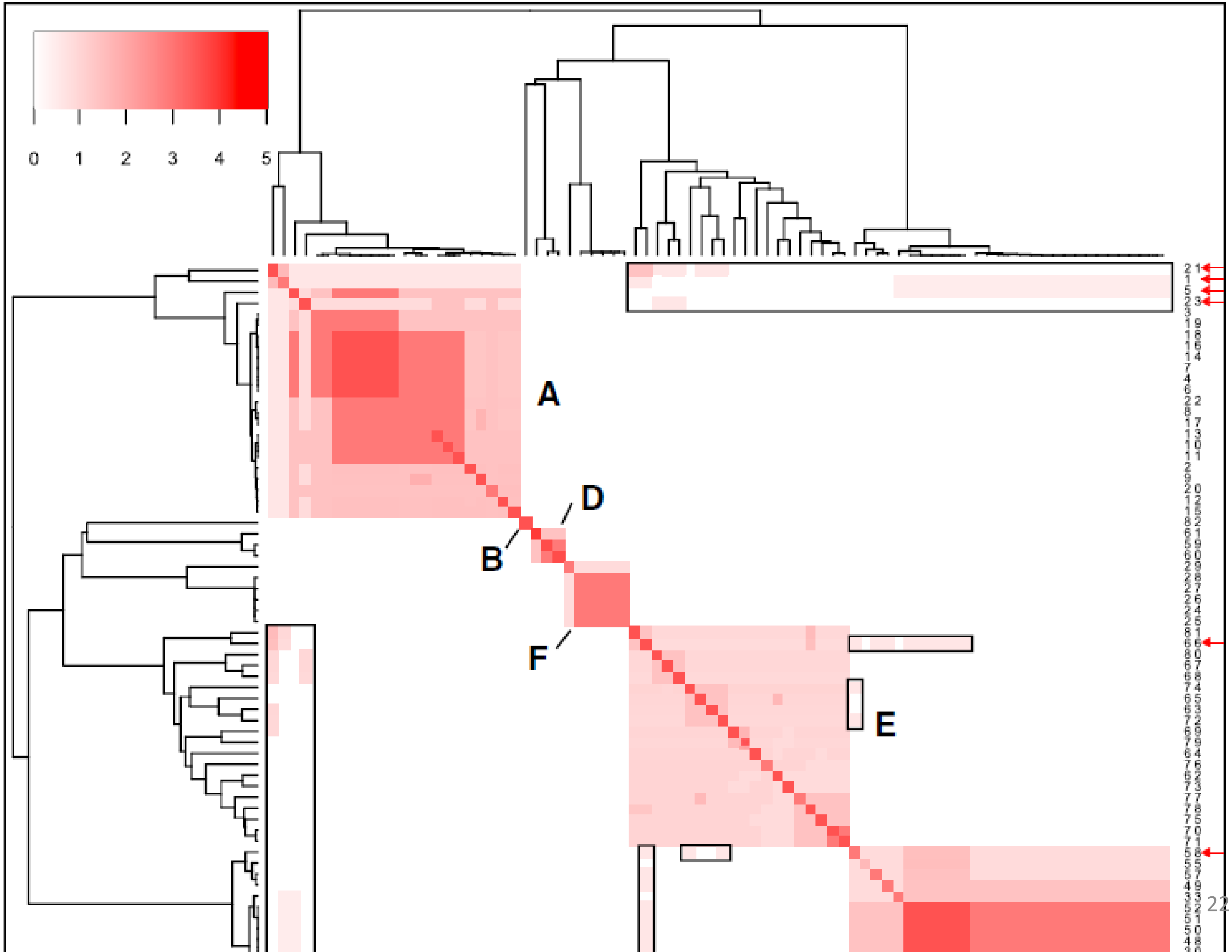


Figure 1. Median-Joining Haplotype Network of the COI Sequences Obtained from the 576 *A. vaga* Individuals Collected

Bebortoli et al 2016



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	sample ID	species	COI (species)	28S (species)	EPIC25 (species)	EPIC63 (species)	Nu1054 (species)
Ind1	B14	A	Hap6 (A)	Hap1 (A)	Hap9-Hap10 (C)	Hap16-Hap20 (E)	
Ind2	B15	A	Hap6 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap2 (A)
Ind3	C1T1	A	Hap3 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	
Ind4	C3T3	A	Hap3 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap1-Hap5 (A)
Ind5	D14	A	Hap3 (A)	Hap1 (A)	Hap10 (C)	Hap1 (A)	Hap1 (A)
Ind6	D12	A	Hap3 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1 (A)	Hap1-Hap6 (A)
Ind7	D13	A	Hap3 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1 (A)	Hap1-Hap7 (A)
Ind8	E1B1	A	Hap4 (A)	Hap1 (A)	Hap4 (A)	Hap1 (A)	
Ind9	E1B3	A	Hap4 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1 (A)	Hap1-Hap4 (A)
Ind10	H001	A	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap1-Hap8 (A)
Ind11	H3-03	A	Hap1 (A)	Hap1 (A)	Hap1-Hap2 (A)	Hap1 (A)	Hap1-Hap2 (A)
Ind12	H3-04	A	Hap2 (A)	Hap1 (A)	Hap2-Hap4 (A)	Hap1-Hap3 (A)	Hap2-Hap9 (A)
Ind13	HB01	A	Hap1 (A)	Hap1 (A)	Hap1-Hap2 (A)	Hap1 (A)	Hap1-Hap2 (A)
Ind14	C28	A	Hap3 (A)	Hap1 (A)	Hap4 (A)	Hap1 (A)	Hap2 (A)
Ind15	C33	A	Hap9 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1-Hap3 (A)	Hap2-Hap3 (A)
Ind16	C31	A	Hap3 (A)	Hap1 (A)	Hap4 (A)	Hap3 (A)	Hap2 (A)
Ind17	C1T2	A	Hap4 (A)	Hap1 (A)	Hap2-Hap4 (A)	Hap2-Hap3 (A)	
Ind18	D11	A	Hap3 (A)	Hap1 (A)	Hap2-Hap4 (A)	Hap1 (A)	Hap2 (A)
Ind19	E11	A	Hap3 (A)	Hap1 (A)	Hap4 (A)	Hap3 (A)	
Ind20	E31	A	Hap5 (A)	Hap1 (A)	Hap3-Hap5 (A)	Hap1 (A)	Hap1-Hap3 (A)
Ind21	B11	A	Hap6 (A)	Hap1 (A)	Hap35 (E)	Hap16 (E)	Hap22 (E)
Ind22	B33	A	Hap7 (A)	Hap1 (A)	Hap1-Hap5 (A)	Hap1 (A)	
Ind23	B3B1	A	Hap1 (A)	Hap1-Hap2 (A)	Hap35 (E)	Hap1 (A)	
Ind24	C210	F	Hap15 (F)	Hap4 (F)	Hap16-Hap19 (F)	Hap11-Hap12 (F)	
Ind25	C211	F	Hap14 (F)	Hap4 (F)	Hap16-Hap22 (F)	Hap11-Hap12 (F)	
Ind26	C21	F	Hap13 (F)	Hap4 (F)	Hap17-Hap20 (F)	Hap11-Hap13 (F)	
Ind27	C24	F	Hap16 (F)	Hap4 (F)	Hap17-Hap22 (F)	Hap11-Hap13 (F)	
Ind28	C27	F	Hap17 (F)	Hap4 (F)	Hap16-Hap19 (F)	Hap11-Hap12 (F)	

	sample ID	species	COI (species)	28S (species)	EPIC25 (species)	EPIC63 (species)	Nu1054 (species)
Ind1	B14	A	Hap6 (A)	Hap1 (A)	Hap9-Hap10 (C)	Hap16-Hap20 (E)	
Ind2	B15	A	Hap6 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap2 (A)
Ind3	C1T1	A	Hap3 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	
Ind4	C3T3	A	Hap3 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap1-Hap5 (A)
Ind5	D14	A	Hap3 (A)	Hap1 (A)	Hap10 (C)	Hap1 (A)	Hap1 (A)
Ind6	D12	A	Hap3 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1 (A)	Hap1-Hap6 (A)
Ind7	D13	A	Hap3 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1 (A)	Hap1-Hap7 (A)
Ind8	E1B1	A	Hap4 (A)	Hap1 (A)	Hap4 (A)	Hap1 (A)	
Ind9	E1B3	A	Hap4 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1 (A)	Hap1-Hap4 (A)
Ind10	H001	A	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap1 (A)	Hap1-Hap8 (A)
Ind11	H3-03	A	Hap1 (A)	Hap1 (A)	Hap1-Hap2 (A)	Hap1 (A)	Hap1-Hap2 (A)
Ind12	H3-04	A	Hap2 (A)	Hap1 (A)	Hap2-Hap4 (A)	Hap1-Hap3 (A)	Hap2-Hap9 (A)
Ind13	HB01	A	Hap1 (A)	Hap1 (A)	Hap1-Hap2 (A)	Hap1 (A)	Hap1-Hap2 (A)
Ind14	C28	A	Hap3 (A)	Hap1 (A)	Hap4 (A)	Hap1 (A)	Hap2 (A)
Ind15	C33	A	Hap9 (A)	Hap1 (A)	Hap1-Hap4 (A)	Hap1-Hap3 (A)	Hap2-Hap3 (A)
Ind16	C31	A	Hap3 (A)	Hap1 (A)	Hap4 (A)	Hap3 (A)	Hap2 (A)
Ind17	C1T2	A	Hap4 (A)	Hap1 (A)	Hap2-Hap4 (A)	Hap2-Hap3 (A)	
Ind18	D11	A	Hap3 (A)	Hap1 (A)	Hap2-Hap4 (A)	Hap1 (A)	Hap2 (A)
Ind19	E11	A	Hap3 (A)	Hap1 (A)	Hap4 (A)	Hap3 (A)	
Ind20	E31	A	Hap5 (A)	Hap1 (A)	Hap3-Hap5 (A)	Hap1 (A)	Hap1-Hap3 (A)
Ind21	B11	A	Hap6 (A)	Hap1 (A)	Hap35 (E)	Hap16 (E)	Hap22 (E)
Ind22	B33	A	Hap7 (A)	Hap1 (A)	Hap1-Hap5 (A)	Hap1 (A)	
Ind23	B3B1	A	Hap1 (A)	Hap1-Hap2 (A)	Hap35 (E)	Hap1 (A)	
Ind24	C210	F	Hap15 (F)	Hap4 (F)	Hap16-Hap19 (F)	Hap11-Hap12 (F)	
Ind25	C211	F	Hap14 (F)	Hap4 (F)	Hap16-Hap22 (F)	Hap11-Hap12 (F)	
Ind26	C21	F	Hap13 (F)	Hap4 (F)	Hap17-Hap20 (F)	Hap11-Hap13 (F)	
Ind27	C24	F	Hap16 (F)	Hap4 (F)	Hap17-Hap22 (F)	Hap11-Hap13 (F)	
Ind28	C27	F	Hap17 (F)	Hap4 (F)	Hap16-Hap19 (F)	Hap11-Hap12 (F)	

In Brief

Debortoli et al. analyze patterns of allele sharing to delineate cryptic species in the bdelloid rotifer ***Adineta vaga***. They find evidence of inter- and intraspecific genetic exchanges interspersed with chromosome regions bearing signatures of asexual evolution, suggesting that bdelloids exchange DNA horizontally rather than via meiotic sex.

Current Biology
Report

Cross-Contamination Explains “Inter and Intraspecific Horizontal Genetic Transfers” between Asexual Bdelloid Rotifers

Christopher G. Wilson,[1,2,*] Reuben W. Nowell,[1] and Timothy G. Barraclough[1]
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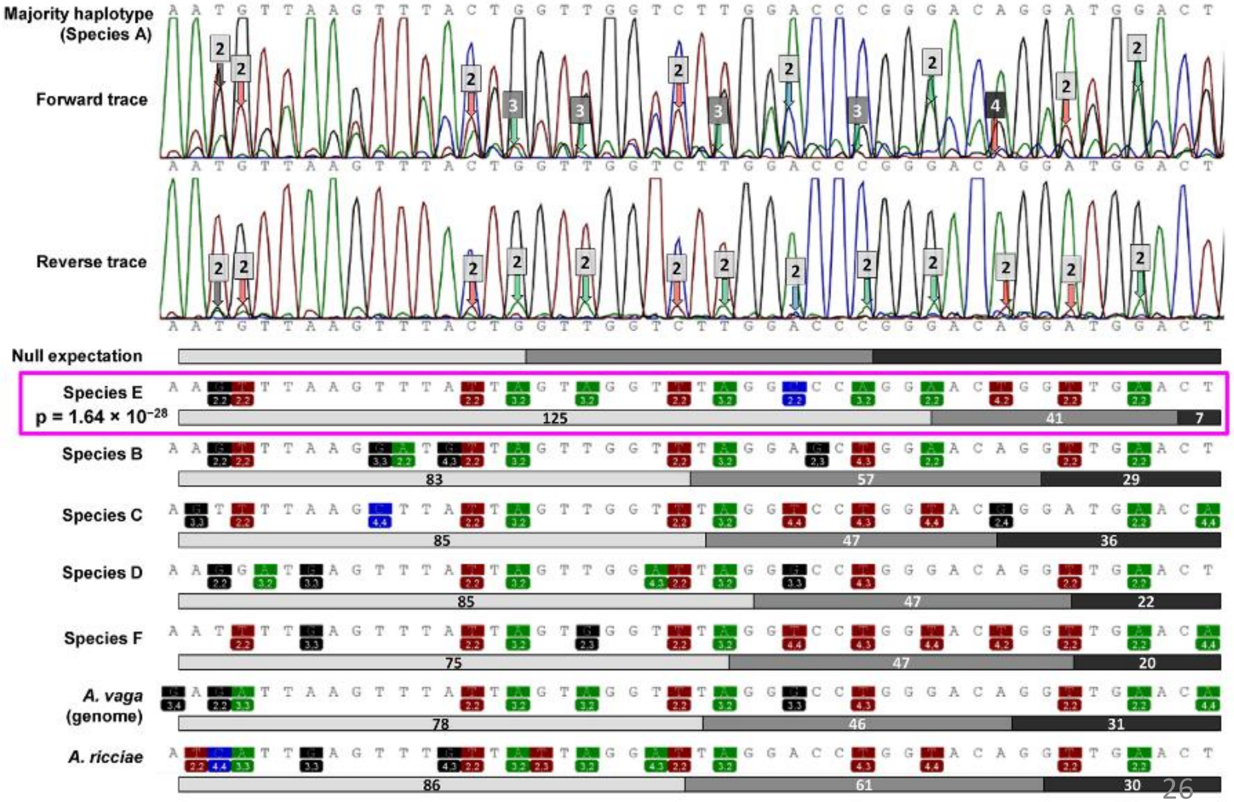
*Correspondence: chris.wilson@imperial.ac.uk
https://doi.org/10.1016/j.cub.2018.05.070

SUMMARY

A few metazoan lineages are thought to have
for millions of years without sexual reproduct
they would offer important clues to the ev
paradox of sex itself [1, 2]. Most “ancient



CellPress
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Report

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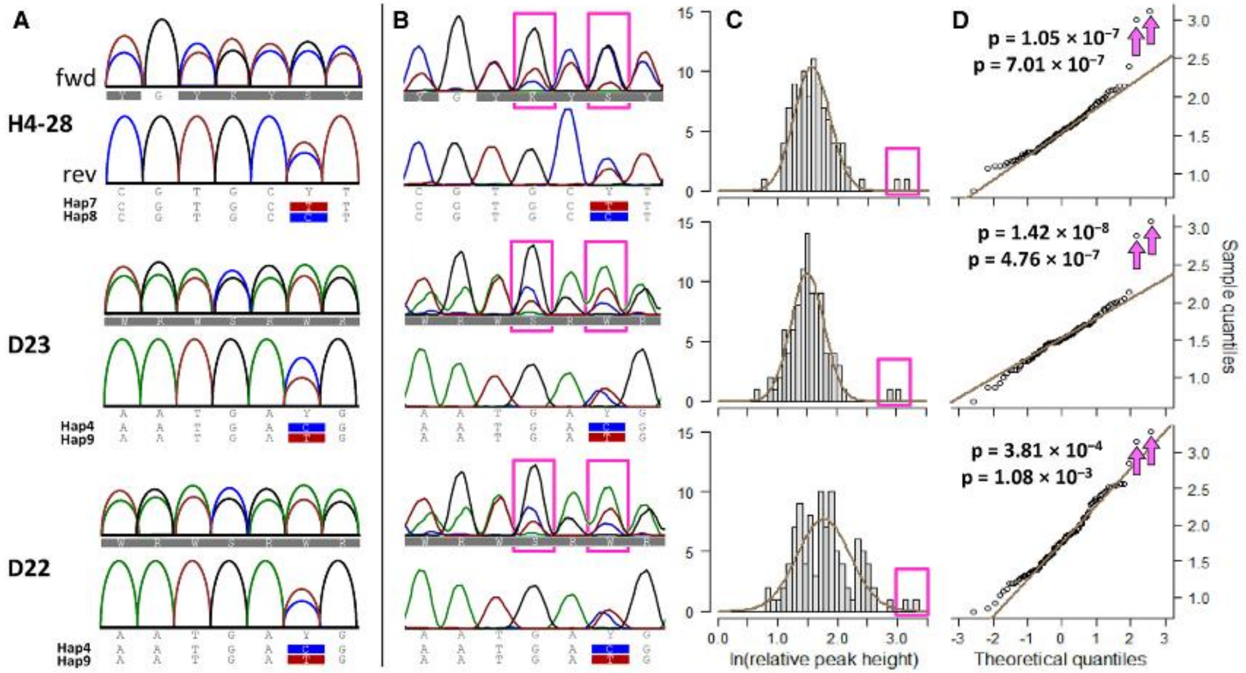
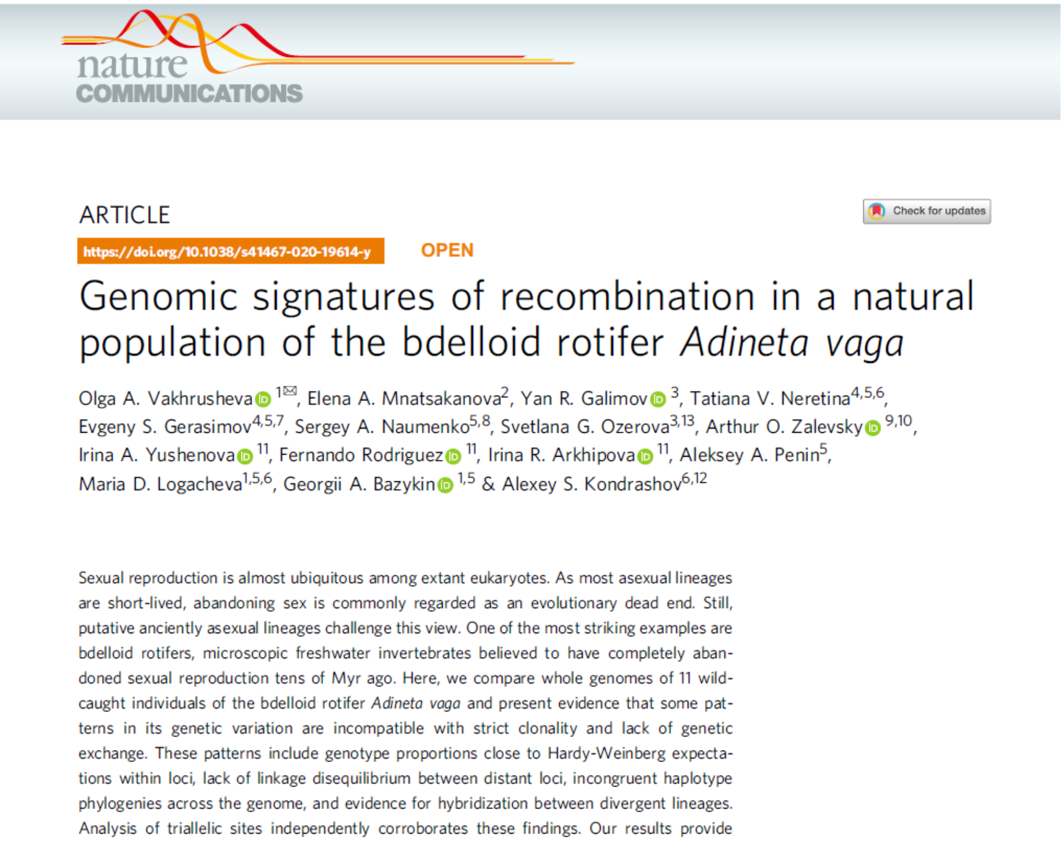


Figure 2. Conspecific Contamination Is Indicated by Triple Peaks in EPIC63 Chromatograms for Samples H4-28, D23, and D22, Revealing Four Alleles where Only Two Are Expected for a Single Animal

(A) Predicted appearance of overlapping chromatogram peaks for the two alleles reported by Debortoli et al. [18], which differ by a C/T SNP and a 2 bp indel earlier in the sequence that has shifted their forward reads 2 bp out of phase.

(B) In each case, the data show two unexpected triple peaks (pink boxes), indicating that C and T variants each occur as both long and short alleles. Four different copies therefore are present (long/C, long/T, short/C, and short/T), whereas a single animal would only have two. This is hidden in the reverse reads (and at other loci) because all copies are in phase at the SNP site.



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ARTICLE

https://doi.org/10.1038/s41467-020-19614-y OPEN

Check for updates

Genomic signatures of recombination in a natural population of the bdelloid rotifer *Adineta vaga*

Olga A. Vakhrusheva 1✉, Elena A. Mnatsakanova[2], Yan R. Galimov 3, Tatiana V. Neretina[4,5,6], Evgeny S. Gerasimov[4,5,7], Sergey A. Naumenko[5,8], Svetlana G. Ozerova[3,13], Arthur O. Zalevsky 9,10, Irina A. Yushenova 11, Fernando Rodriguez 11, Irina R. Arkhipova 11, Aleksey A. Penin[5], Maria D. Logacheva[1,5,6], Georgii A. Bazykin 1,5 & Alexey S. Kondrashov[6,12]

Sexual reproduction is almost ubiquitous among extant eukaryotes. As most asexual lineages are short-lived, abandoning sex is commonly regarded as an evolutionary dead end. Still, putative anciently asexual lineages challenge this view. One of the most striking examples are bdelloid rotifers, microscopic freshwater invertebrates believed to have completely abandoned sexual reproduction tens of Myr ago. Here, we compare whole genomes of 11 wild-caught individuals of the bdelloid rotifer *Adineta vaga* and present evidence that some patterns in its genetic variation are incompatible with strict clonality and lack of genetic exchange. These patterns include genotype proportions close to Hardy-Weinberg expectations within loci, lack of linkage disequilibrium between distant loci, incongruent haplotype phylogenies across the genome, and evidence for hybridization between divergent lineages. Analysis of triallelic sites independently corroborates these findings. Our results provide

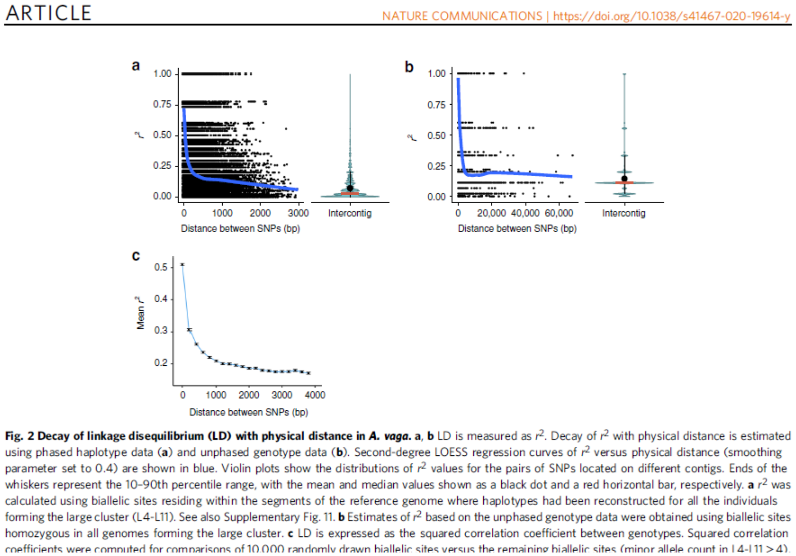


Fig. 2 Decay of linkage disequilibrium (LD) with physical distance in *A. vaga*. **a**, **b** LD is measured as r^2. Decay of r^2 with physical distance is estimated using phased haplotype data (**a**) and unphased genotype data (**b**). Second-degree LOESS regression curves of r^2 versus physical distance (smoothing parameter set to 0.4) are shown in blue. Violin plots show the distributions of r^2 values for the pairs of SNPs located on different contigs. Ends of the whiskers represent the 10–90th percentile range, with the mean and median values shown as a black dot and a red horizontal bar, respectively. **a** r^2 was calculated using biallelic sites residing within the segments of the reference genome where haplotypes had been reconstructed for all the individuals forming the large cluster (L4-L11). See also Supplementary Fig. 11. **b** Estimates of r^2 based on the unphased genotype data were obtained using biallelic sites homozygous in all genomes forming the large cluster. **c** LD is expressed as the squared correlation coefficient between genotypes. Squared correlation coefficients were computed for comparisons of 10,000 randomly drawn biallelic sites versus the remaining biallelic sites (minor allele count in L4-L11 > 4).

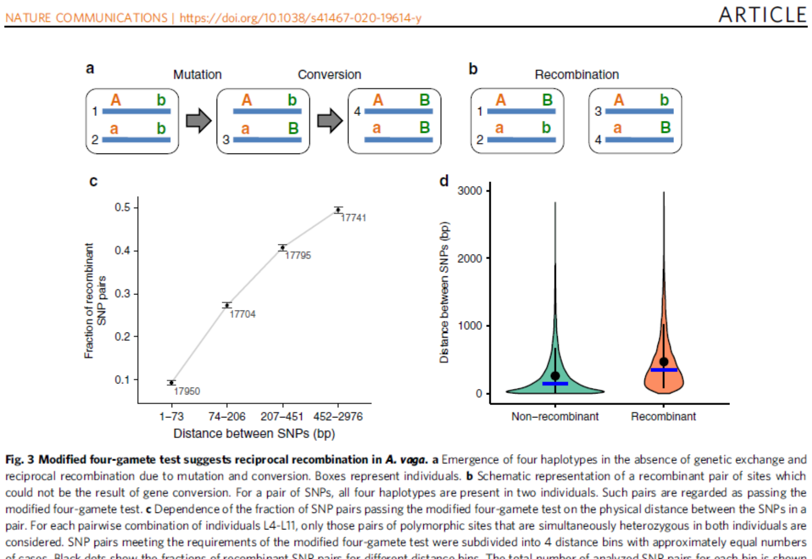
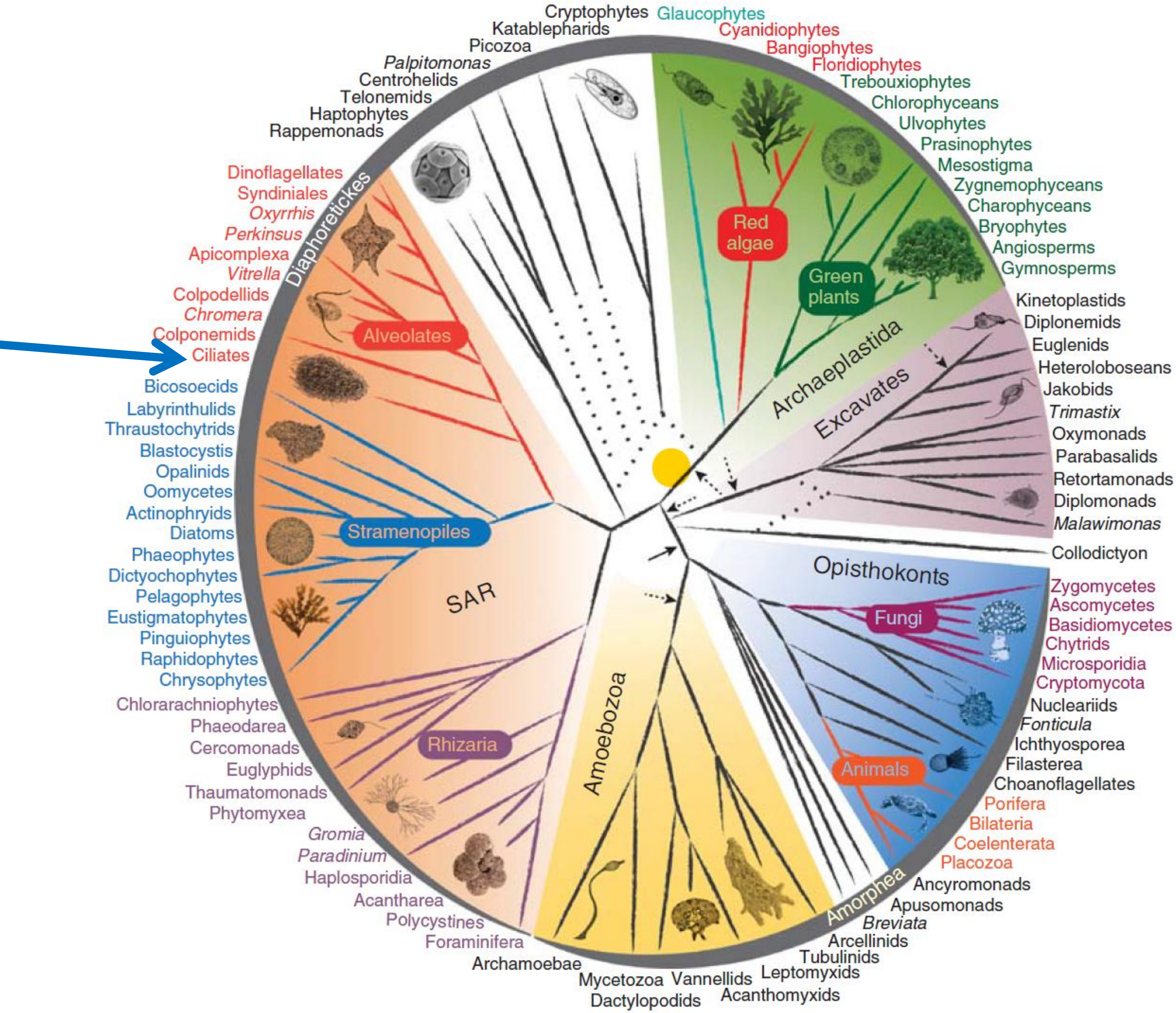


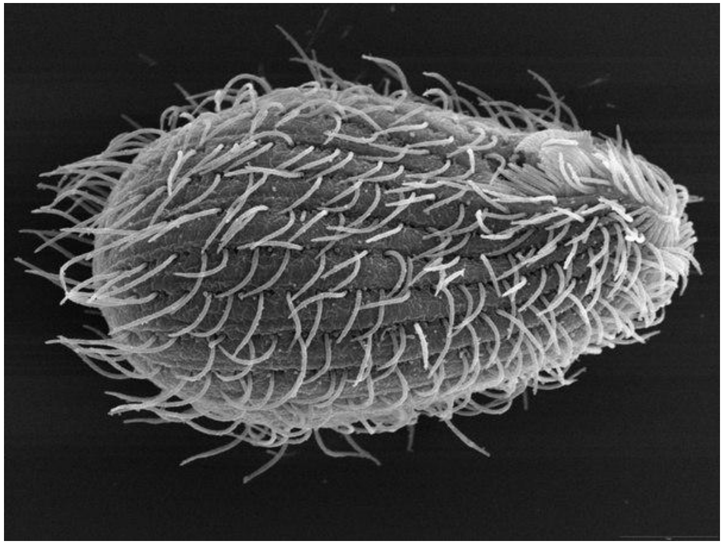
Fig. 3 Modified four-gamete test suggests reciprocal recombination in *A. vaga*. **a** Emergence of four haplotypes in the absence of genetic exchange and reciprocal recombination due to mutation and conversion. Boxes represent individuals. **b** Schematic representation of a recombinant pair of sites which could not be the result of gene conversion. For a pair of SNPs, all four haplotypes are present in two individuals. Such pairs are regarded as passing the modified four-gamete test. **c** Dependence of the fraction of SNP pairs passing the modified four-gamete test on the physical distance between the SNPs in a pair. For each pairwise combination of individuals L4-L11, only those pairs of polymorphic sites that are simultaneously heterozygous in both individuals are considered. SNP pairs meeting the requirements of the modified four-gamete test were subdivided into 4 distance bins with approximately equal numbers of cases. Black dots show the fractions of recombinant SNP pairs for different distance bins. The total number of analyzed SNP pairs for each bin is shown



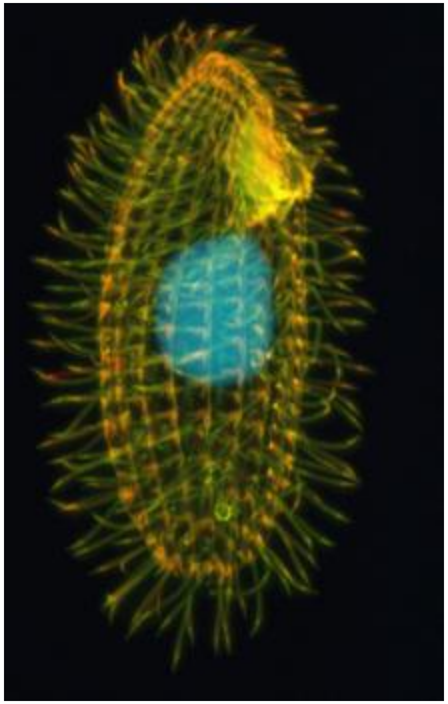
Cryptophytes
Glaucophytes
Cyanidiophytes
Bangiophytes
Floridiophytes
Trebouxiophytes
Chlorophyceans
Ulvophytes
Prasinophytes
Mesostigma
Zygnemophyceans
Charophyceans
Bryophytes
Angiosperms
Gymnosperms
Katablepharids
Picozoa
Palpitomonas
Centrohelids
Telonemids
Haptophytes
Rappemonads
Red algae
Green plants
Archaeplastida
Diaphoretickes
Dinoflagellates
Syndiniales
Oxyrrhis
Perkinsus
Apicomplexa
Vitrella
Colpodellids
Chromera
Colponemids
Ciliates
Alveolates
Bicosoecids
Labyrinthulids
Thraustochytrids
Blastocystis
Opalinids
Oomycetes
Actinophryids
Diatoms
Phaeophytes
Dictyochophytes
Pelagophytes
Eustigmatophytes
Pinguiophytes
Raphidophytes
Chrysophytes
Stramenopiles
SAR
Chlorarachniophytes
Phaeodarea
Cercomonads
Euglyphids
Thaumatomonads
Phytomyxea
Gromia
Paradinium
Haplosporidia
Acantharea
Polycystines
Foraminifera
Rhizaria
Kinetoplastids
Diplonemids
Euglenids
Heteroloboseans
Jakobids
Trimastix
Oxymonads
Parabasalids
Retortamonads
Diplomonads
Malawimonas
Excavates
Collodictyon
Opisthokonts
Fungi
Zygomycetes
Ascomycetes
Basidiomycetes
Chytrids
Microsporidia
Cryptomycota
Nucleariids
Fonticula
Ichthyosporea
Filasterea
Choanoflagellates
Animals
Porifera
Bilateria
Coelenterata
Placozoa
Ancyromonads
Apusomonads
Breviata
Amorphea
Amoebozoa
Arcellinids
Tubulinids
Leptomyxids
Acanthomyxids
Vannellids
Dactylopodids
Mycetozoa
Archamoebae

Ciliados

Tetrahymena



Paramecium



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Los ciliados tienen un micronúcleo (línea germinal) y un macronúcleo (vegetativo).
El ciclo de vida alterna la reproducción sexual y la vegetativa.

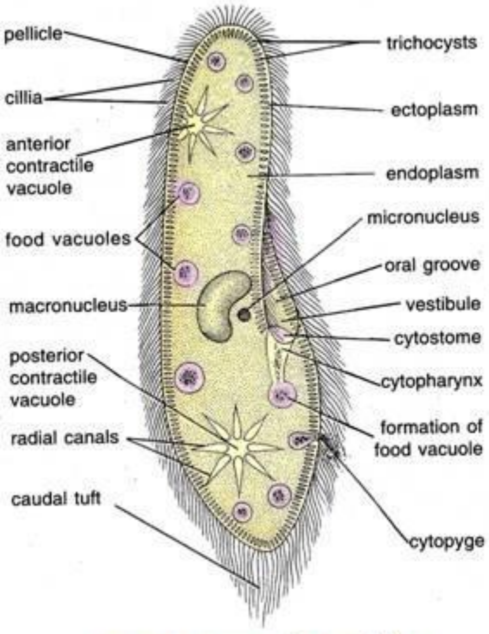
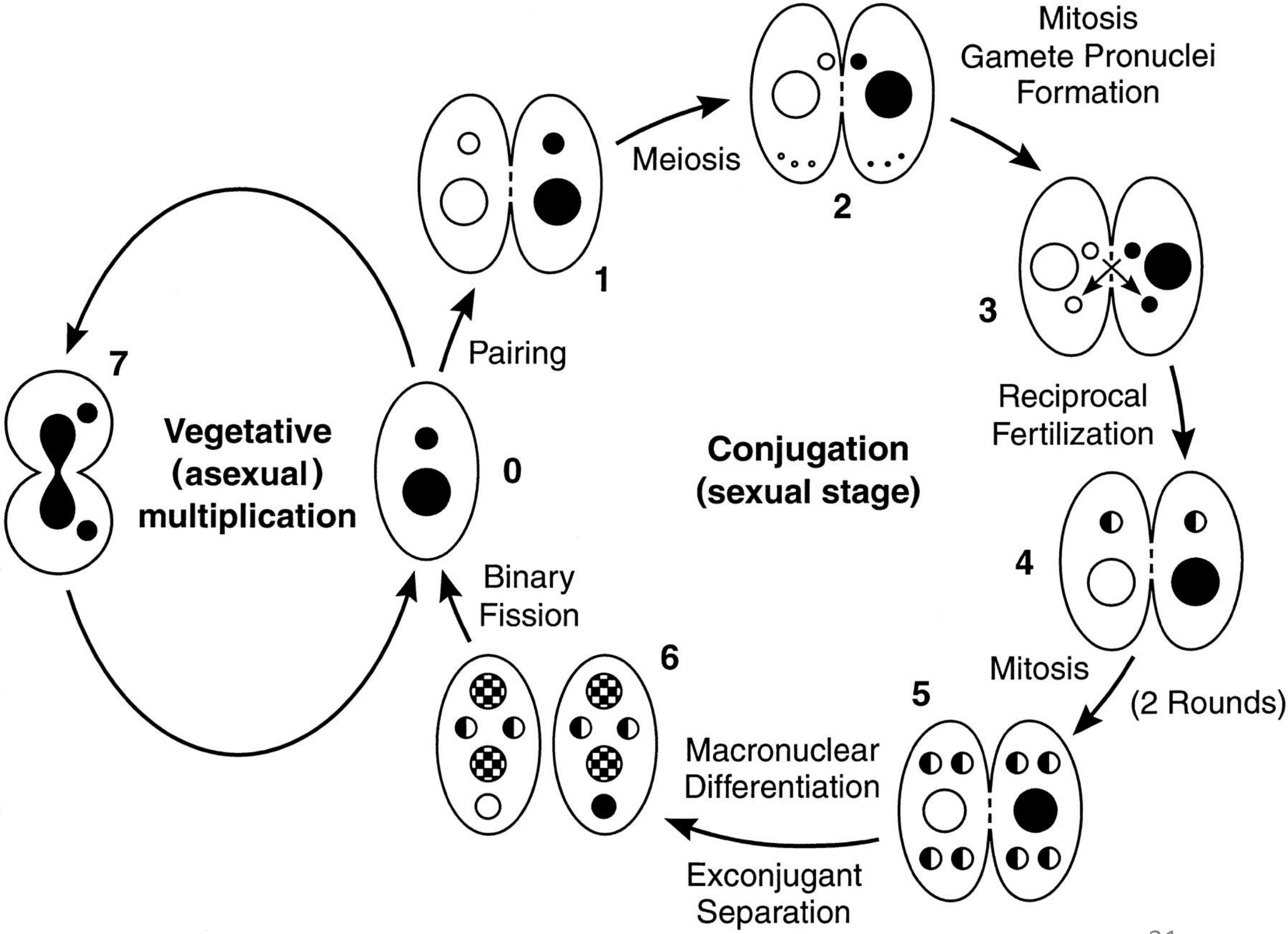
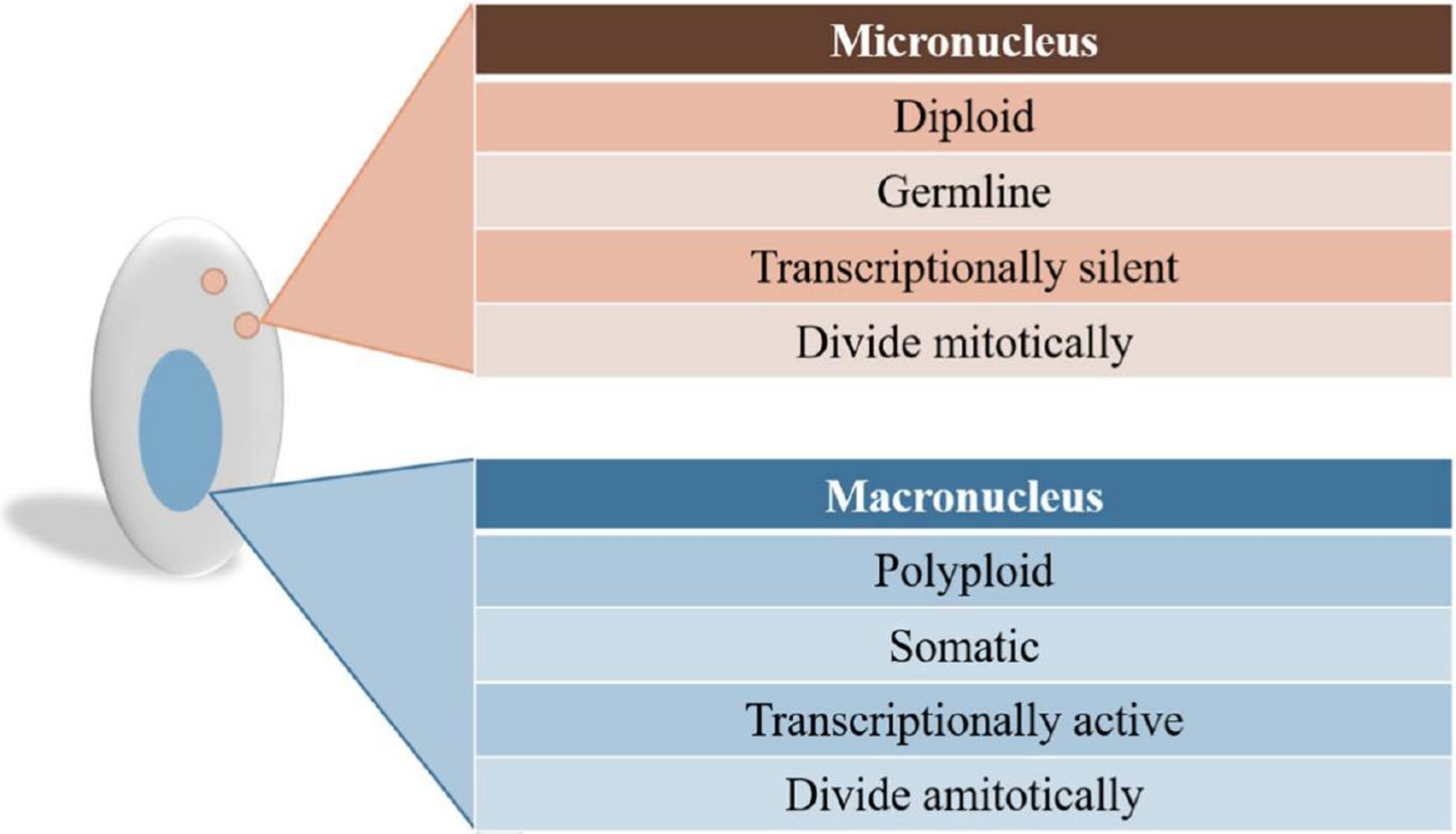


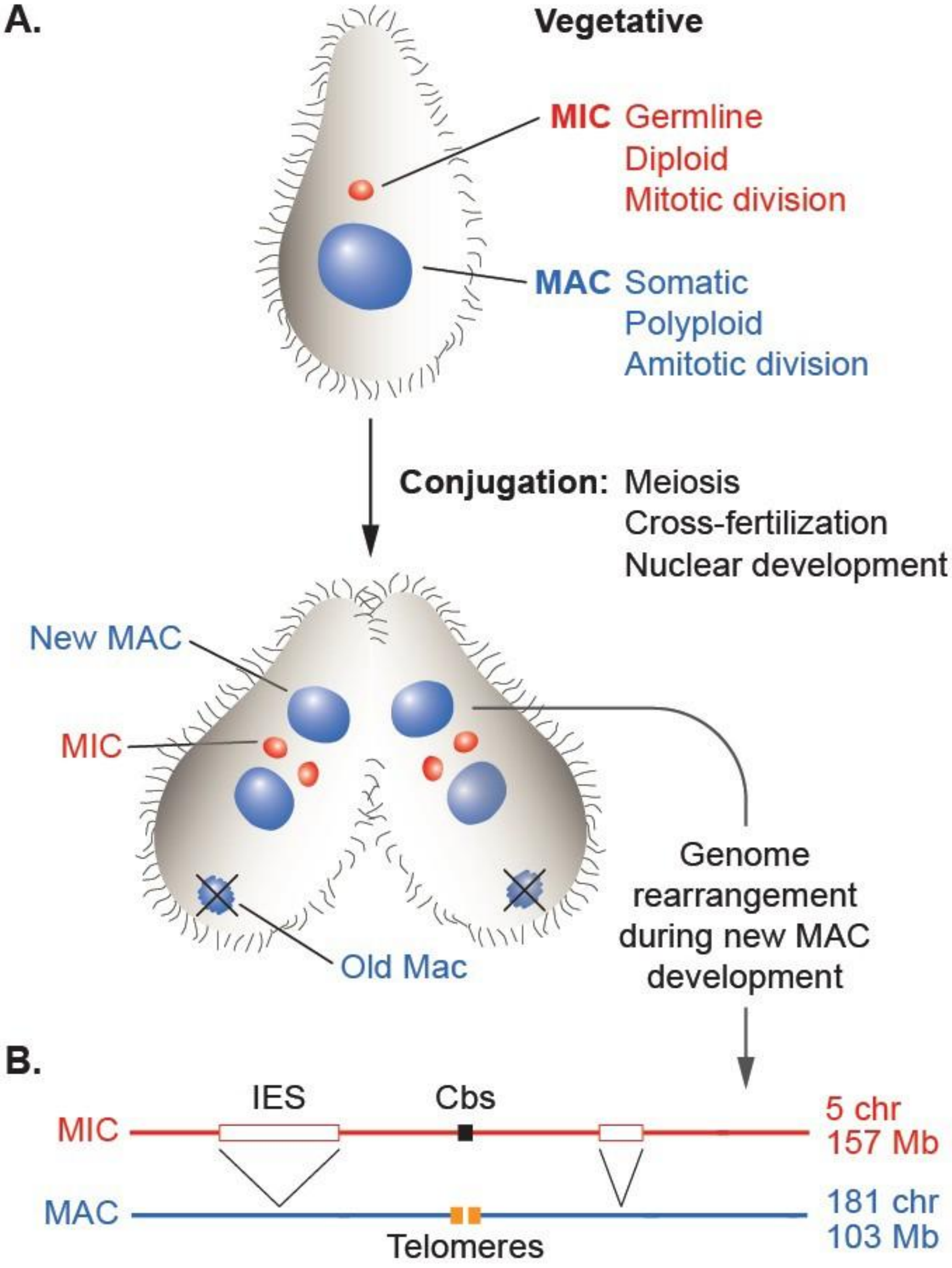
Fig. 20.1. *Paramecium caudatum.*





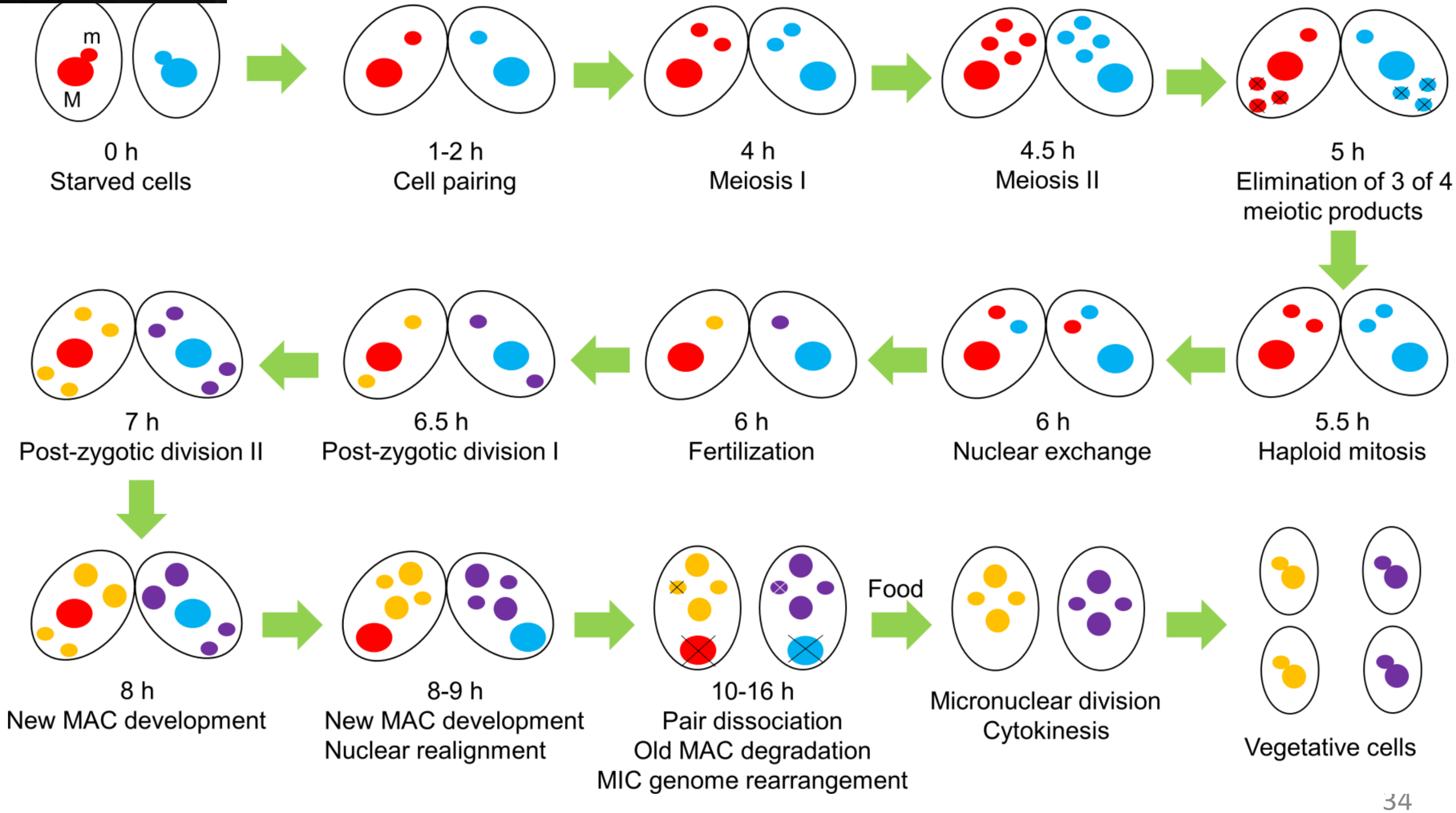
Micronucleus
Diploid
Germline
Transcriptionally silent
Divide mitotically

Macronucleus
Polyploid
Somatic
Transcriptionally active
Divide amitotically


A.
Vegetative
MIC Germline
Diploid
Mitotic division
MAC Somatic
Polyploid
Amitotic division
Conjugation: Meiosis
Cross-fertilization
Nuclear development
New MAC
MIC
Old Mac
Genome
rearrangement
during new MAC
development
B.
IES
Cbs
MIC
5 chr
157 Mb
MAC
181 chr
103 Mb
Telomeres

Apareamiento, meiosis y conjugación

Nucleos (n)



0 h
Starved cells

1-2 h
Cell pairing

4 h
Meiosis I

4.5 h
Meiosis II

5 h
Elimination of 3 of 4 meiotic products

5.5 h
Haploid mitosis

6 h
Nuclear exchange

6 h
Fertilization

6.5 h
Post-zygotic division I

7 h
Post-zygotic division II

8 h
New MAC development

8-9 h
New MAC development
Nuclear realignment

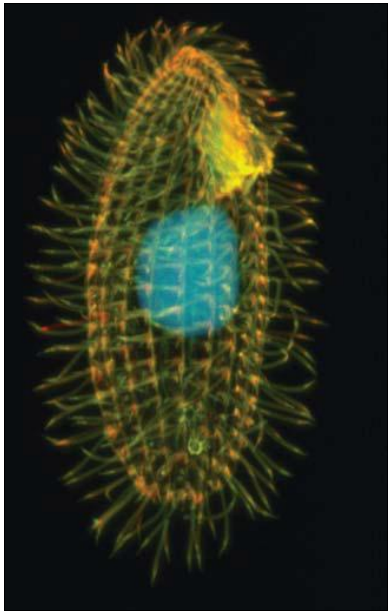
10-16 h
Pair dissociation
Old MAC degradation
MIC genome rearrangement

Micronuclear division
Cytokinesis

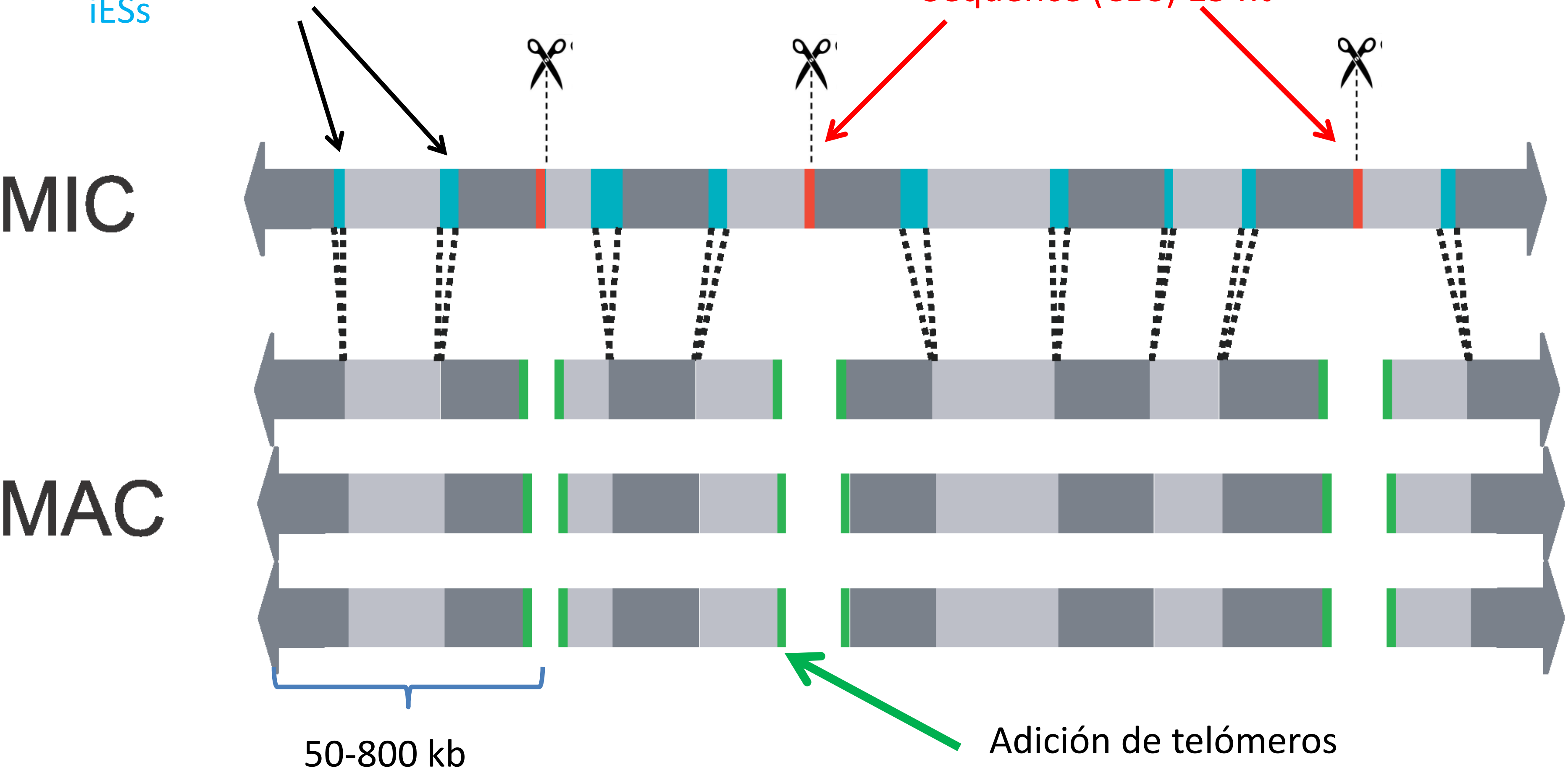
Vegetative cells

Genoma *Tetrahymena*

MIC chromosome	MIC chromosome length (Mb)	Cen midpoint (Mb)
1	36.32	18.61
2	25.51	12.33
3	31.52	15.15
4	31.72	15.29
5	27.47	13.88
Total	**152.54**	



Formación del macronúcleo: proceso de diferenciación del macronucleo a partir del micronucleo



MIC-specific **IESs** (**I**nternal **E**liminated **S**equences, 28-800 nt) Se eliminan unas 6 mil iESs

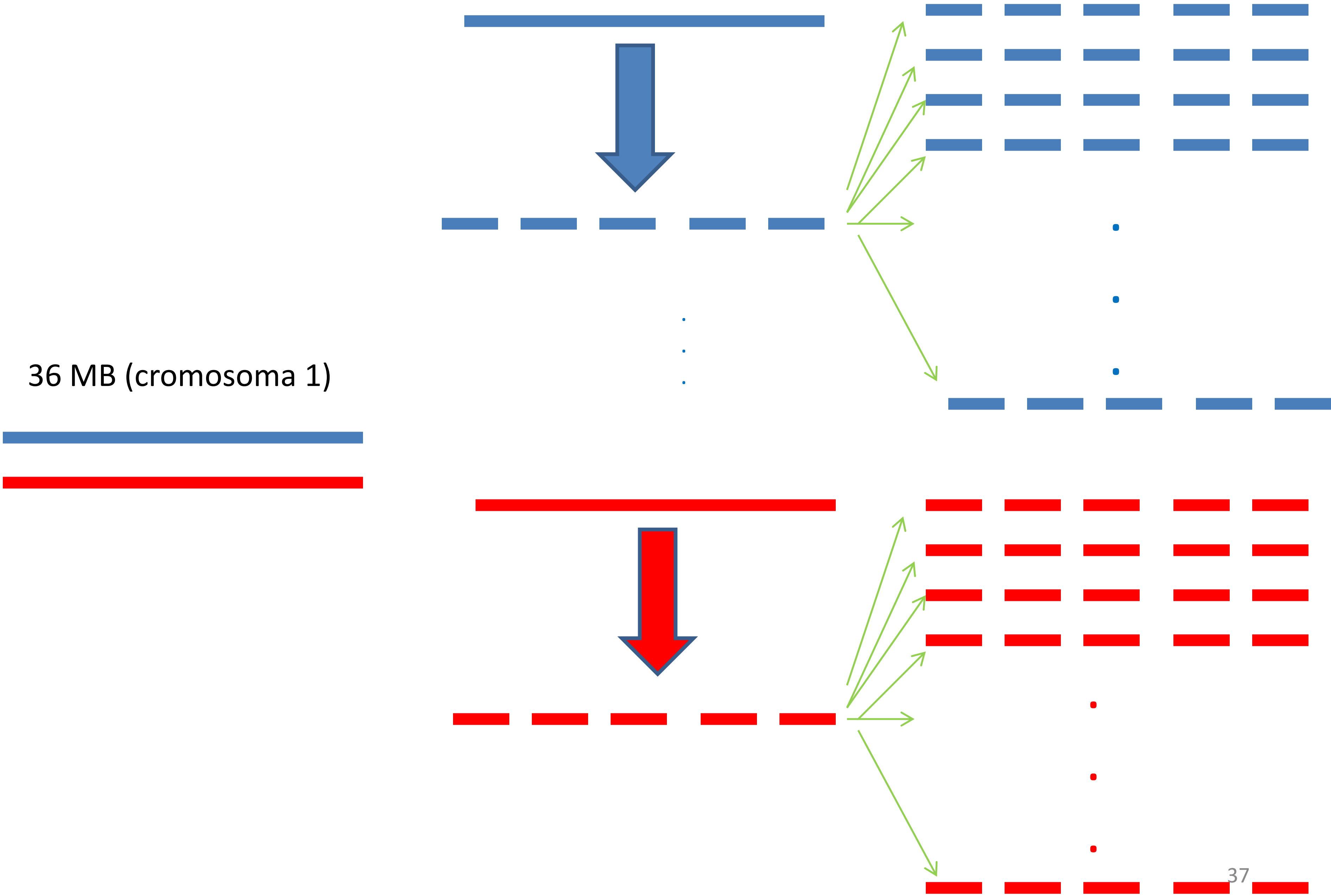
Chromosome **B**reakage **S**equence (CBS) 15 nt

50-800 kb

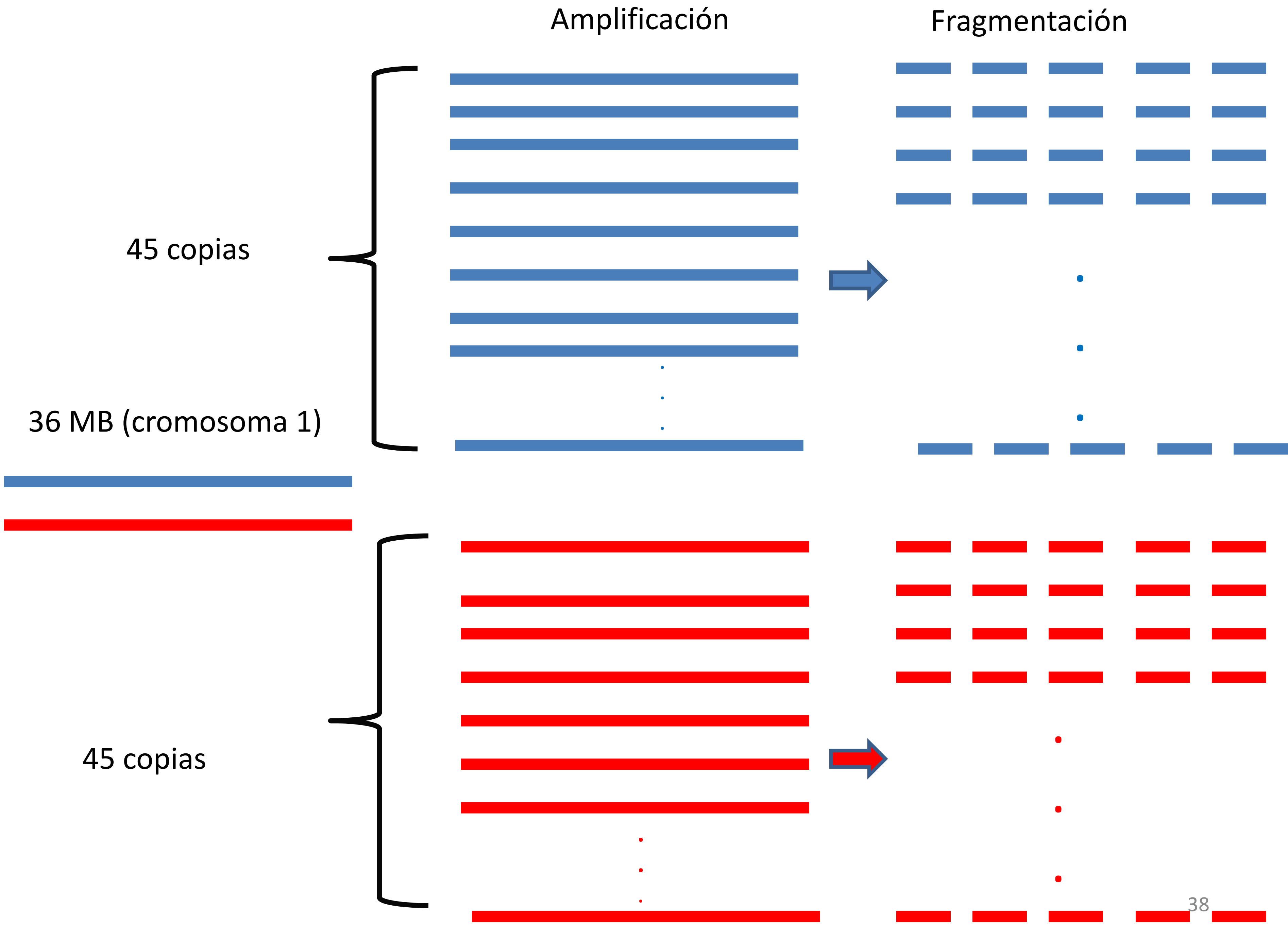
Adición de telómeros

Fragmentacion

Amplificacion



36 MB (cromosoma 1)



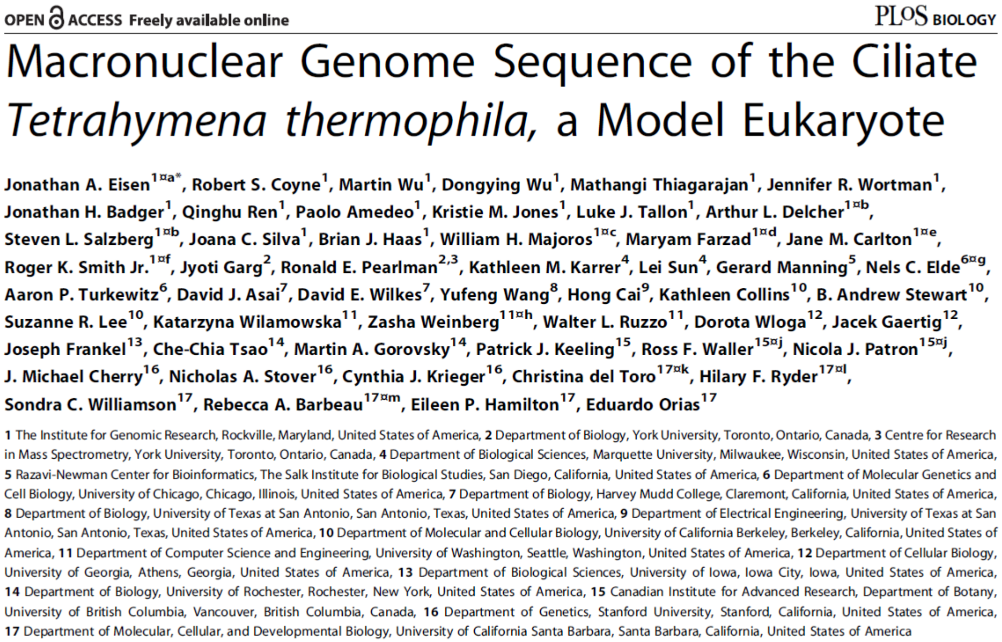
Amplificación

Fragmentación

45 copias

36 MB (cromosoma 1)

45 copias



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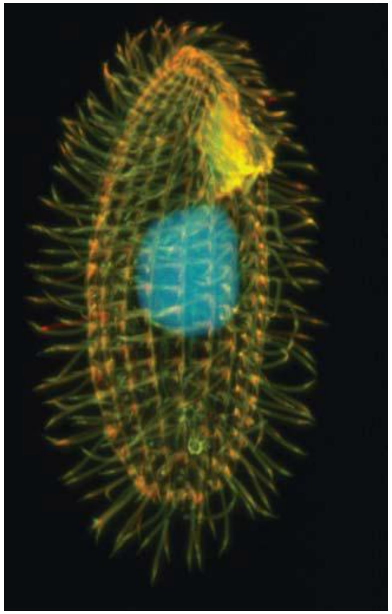
Macronuclear Genome Sequence of the Ciliate *Tetrahymena thermophila*, a Model Eukaryote

Jonathan A. Eisen[1¤a*], Robert S. Coyne[1], Martin Wu[1], Dongying Wu[1], Mathangi Thiagarajan[1], Jennifer R. Wortman[1], Jonathan H. Badger[1], Qinghu Ren[1], Paolo Amedeo[1], Kristie M. Jones[1], Luke J. Tallon[1], Arthur L. Delcher[1¤b], Steven L. Salzberg[1¤b], Joana C. Silva[1], Brian J. Haas[1], William H. Majoros[1¤c], Maryam Farzad[1¤d], Jane M. Carlton[1¤e], Roger K. Smith Jr.[1¤f], Jyoti Garg[2], Ronald E. Pearlman[2,3], Kathleen M. Karrer[4], Lei Sun[4], Gerard Manning[5], Nels C. Elde[6¤g], Aaron P. Turkewitz[6], David J. Asai[7], David E. Wilkes[7], Yufeng Wang[8], Hong Cai[9], Kathleen Collins[10], B. Andrew Stewart[10], Suzanne R. Lee[10], Katarzyna Wilamowska[11], Zasha Weinberg[11¤h], Walter L. Ruzzo[11], Dorota Wloga[12], Jacek Gaertig[12], Joseph Frankel[13], Che-Chia Tsao[14], Martin A. Gorovsky[14], Patrick J. Keeling[15], Ross F. Waller[15¤j], Nicola J. Patron[15¤j], J. Michael Cherry[16], Nicholas A. Stover[16], Cynthia J. Krieger[16], Christina del Toro[17¤k], Hilary F. Ryder[17¤l], Sondra C. Williamson[17], Rebecca A. Barbeau[17¤m], Eileen P. Hamilton[17], Eduardo Orias[17]

1 The Institute for Genomic Research, Rockville, Maryland, United States of America, **2** Department of Biology, York University, Toronto, Ontario, Canada, **3** Centre for Research in Mass Spectrometry, York University, Toronto, Ontario, Canada, **4** Department of Biological Sciences, Marquette University, Milwaukee, Wisconsin, United States of America, **5** Razavi-Newman Center for Bioinformatics, The Salk Institute for Biological Studies, San Diego, California, United States of America, **6** Department of Molecular Genetics and Cell Biology, University of Chicago, Chicago, Illinois, United States of America, **7** Department of Biology, Harvey Mudd College, Claremont, California, United States of America, **8** Department of Biology, University of Texas at San Antonio, San Antonio, Texas, United States of America, **9** Department of Electrical Engineering, University of Texas at San Antonio, San Antonio, Texas, United States of America, **10** Department of Molecular and Cellular Biology, University of California Berkeley, Berkeley, California, United States of America, **11** Department of Computer Science and Engineering, University of Washington, Seattle, Washington, United States of America, **12** Department of Cellular Biology, University of Georgia, Athens, Georgia, United States of America, **13** Department of Biological Sciences, University of Iowa, Iowa City, Iowa, United States of America, **14** Department of Biology, University of Rochester, Rochester, New York, United States of America, **15** Canadian Institute for Advanced Research, Department of Botany, University of British Columbia, Vancouver, British Columbia, Canada, **16** Department of Genetics, Stanford University, Stanford, California, United States of America, **17** Department of Molecular, Cellular, and Developmental Biology, University of California Santa Barbara, Santa Barbara, California, United States of America



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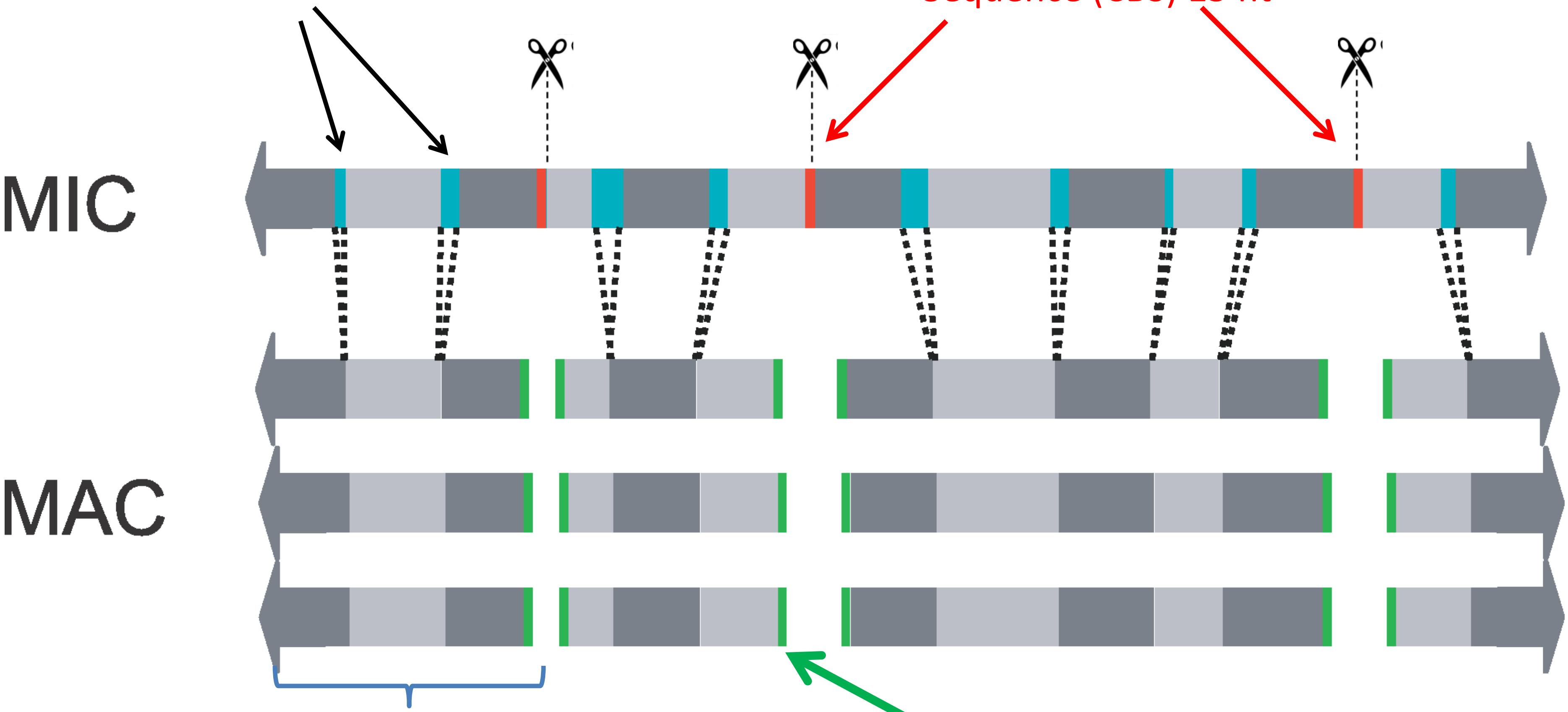


MIC-specific **IESs** (**I**nternal **E**liminated **S**equences, 28-800 nt)

Chromosome **B**reakage **S**equence (CBS) 15 nt

50-800 kb

Adición de telómeros

Conservación de los CBSs

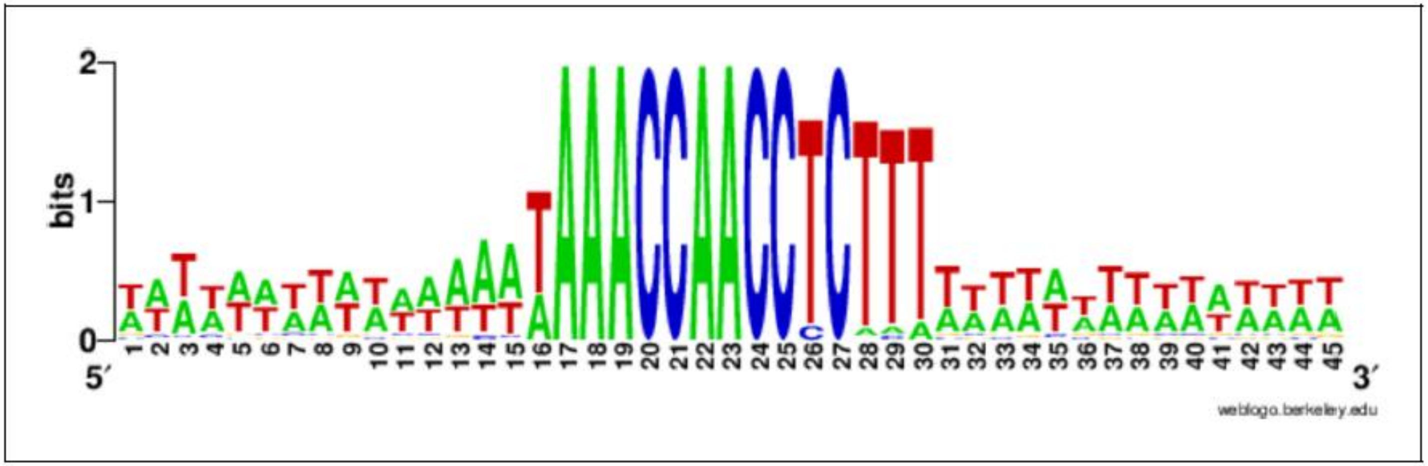
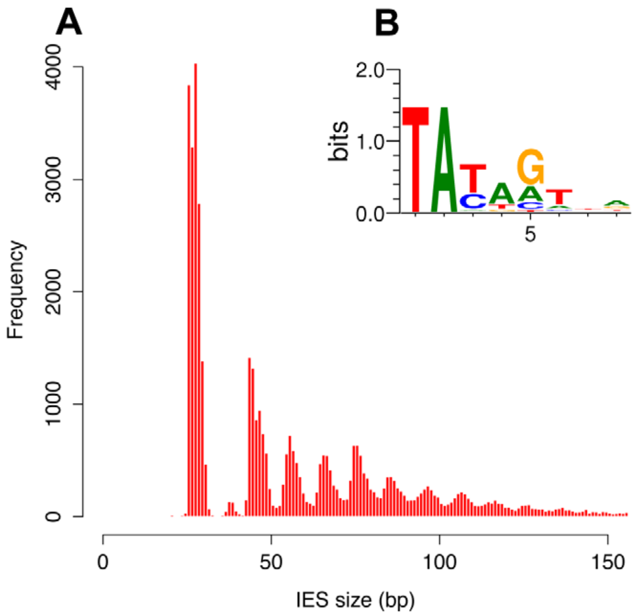


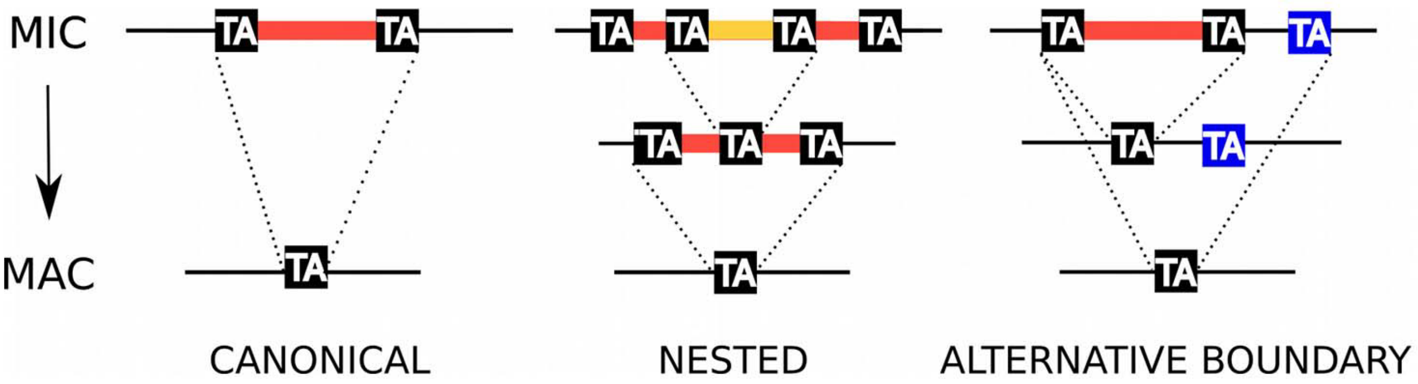
Figure 3. Conservation of the 15 bp chromosome breakage sequence. Nucleotide conservation was calculated at every position, as described in (***Hamilton et al., 2006a***), for the 225 Cbs's and their 15 bp flanking sequences, aligned on the C-rich Cbs strand. The Cbs element occupies positions 16 to 30. At any given position in the logo plot, two bits represent maximum conservation (only one nucleotide occupies that position), and 0 bits corresponds to no conservation (all four nucleotides are equally frequent).

MIC-specific IESs (internal eliminated sequences, 28-800 nt)

Domesticated piggyBac transposase (Tpb2p.)

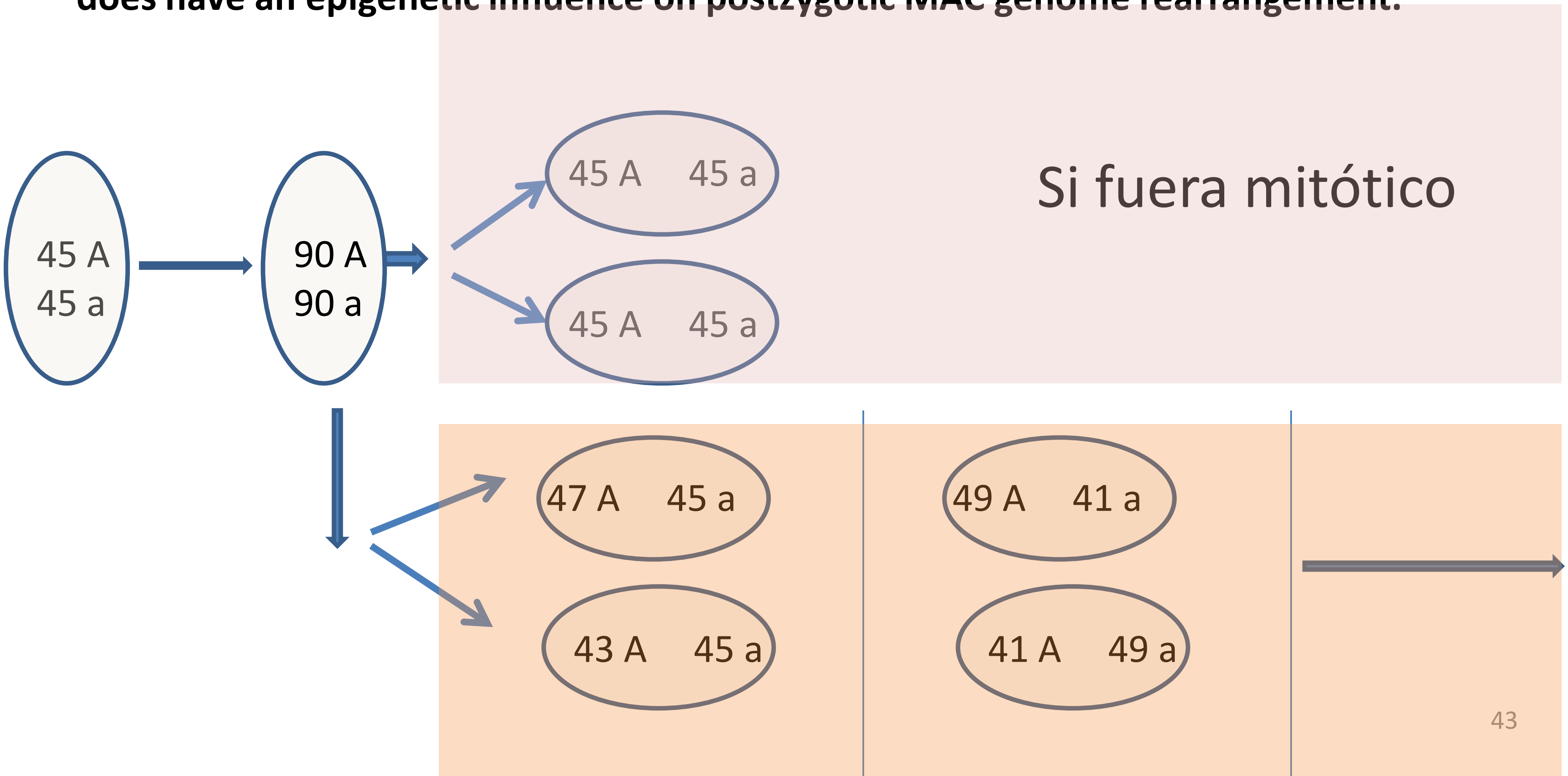
TATA-specific, short repeat





Segregación de material genético

During asexual reproduction, the MAC divides amitotically, with apparently random distribution of chromosome copies that behave as if acentromeric. In contrast, MIC chromosomes are metacentric and are distributed mitotically. Parental MAC DNA is not transmitted to sexual progeny, although it does have an epigenetic influence on postzygotic MAC genome rearrangement.



45 A
45 a

90 A
90 a

45 A 45 a

45 A 45 a

Si fuera mitótico

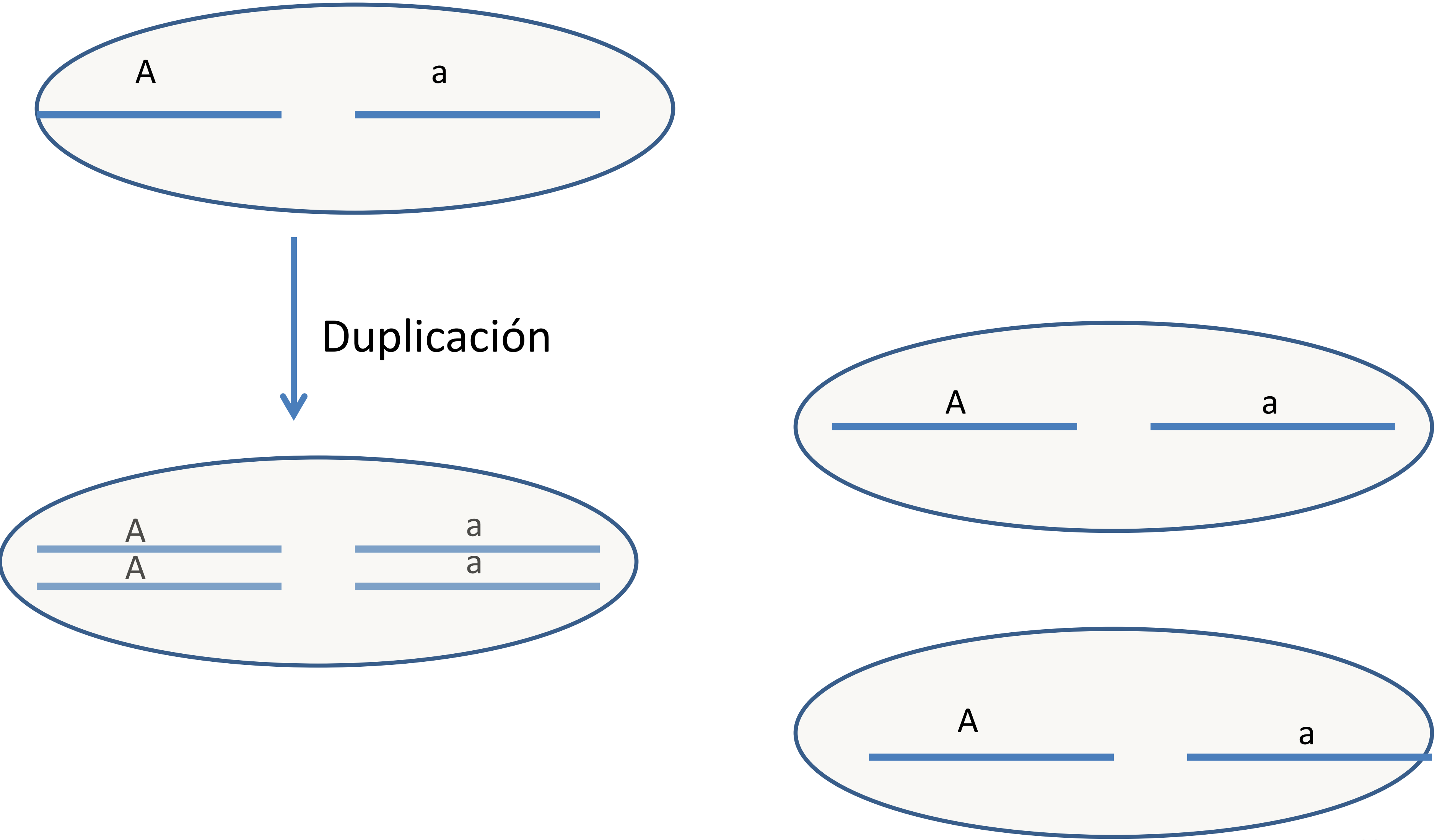
47 A 45 a

43 A 45 a

49 A 41 a

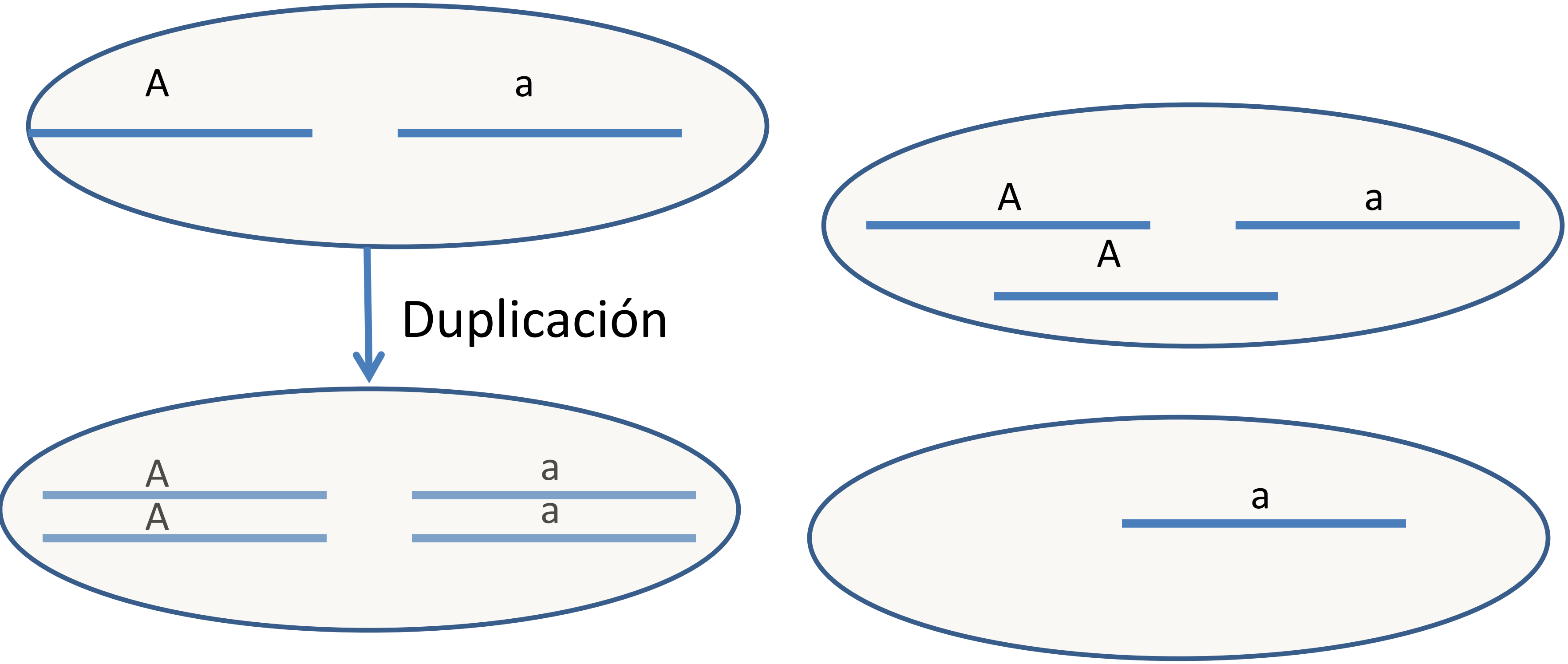
41 A 49 a

Si fuera mitotico



Duplicación

Si fuera no mitotico



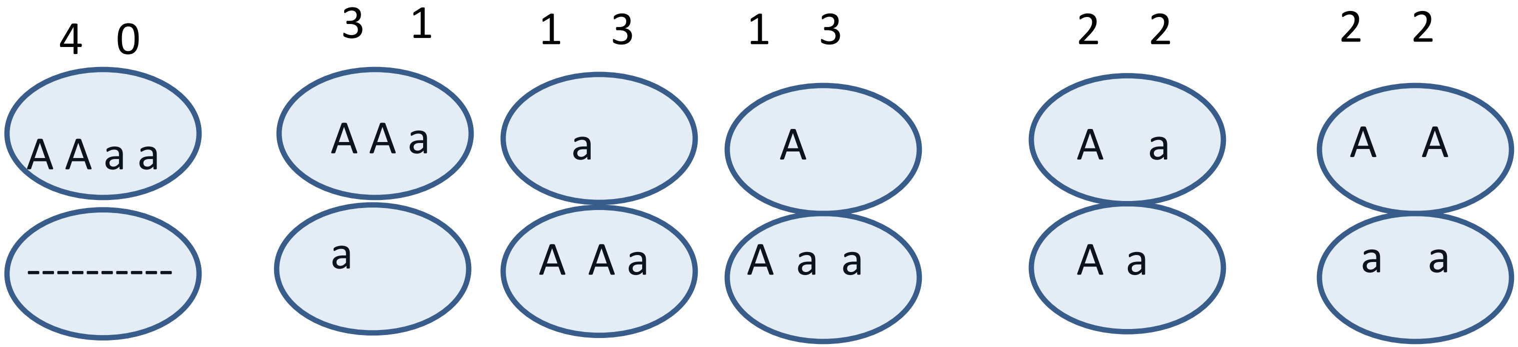
A a

Duplicación

A A a a

A A a

a



4 0
A A a a
----

3 1
A A a
a

1 3
a
A A a

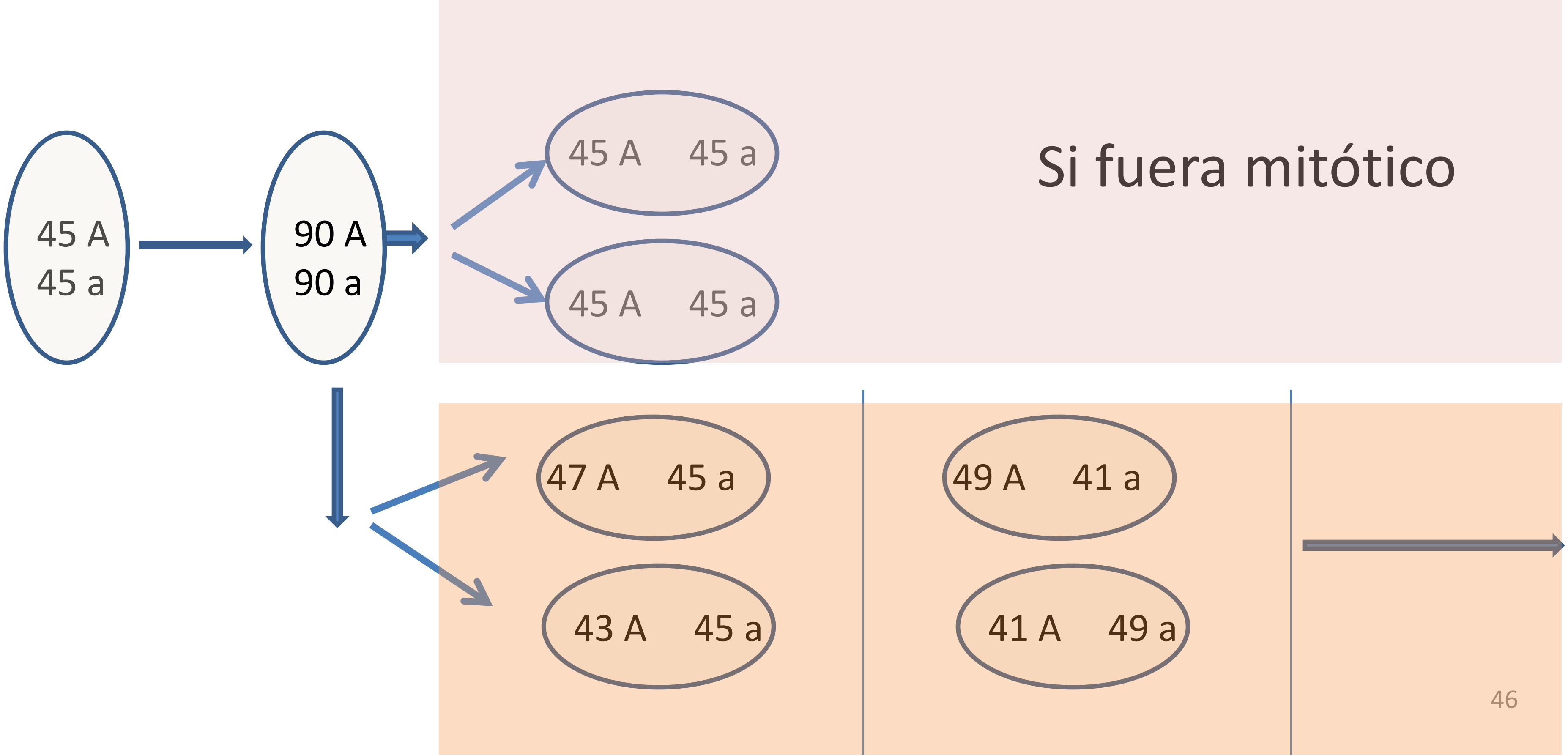
1 3
A
A a a

2 2
A a
A a

2 2
A A
a a

Segregación de material genético

During asexual reproduction, the MAC divides amitotically, with apparently random distribution of chromosome copies that behave as if acentromeric. In contrast, MIC chromosomes are metacentric and are distributed mitotically. Parental MAC DNA is not transmitted to sexual progeny, although it does have an epigenetic influence on postzygotic MAC genome rearrangement.



45 A
45 a

90 A
90 a

45 A 45 a

45 A 45 a

Si fuera mitótico

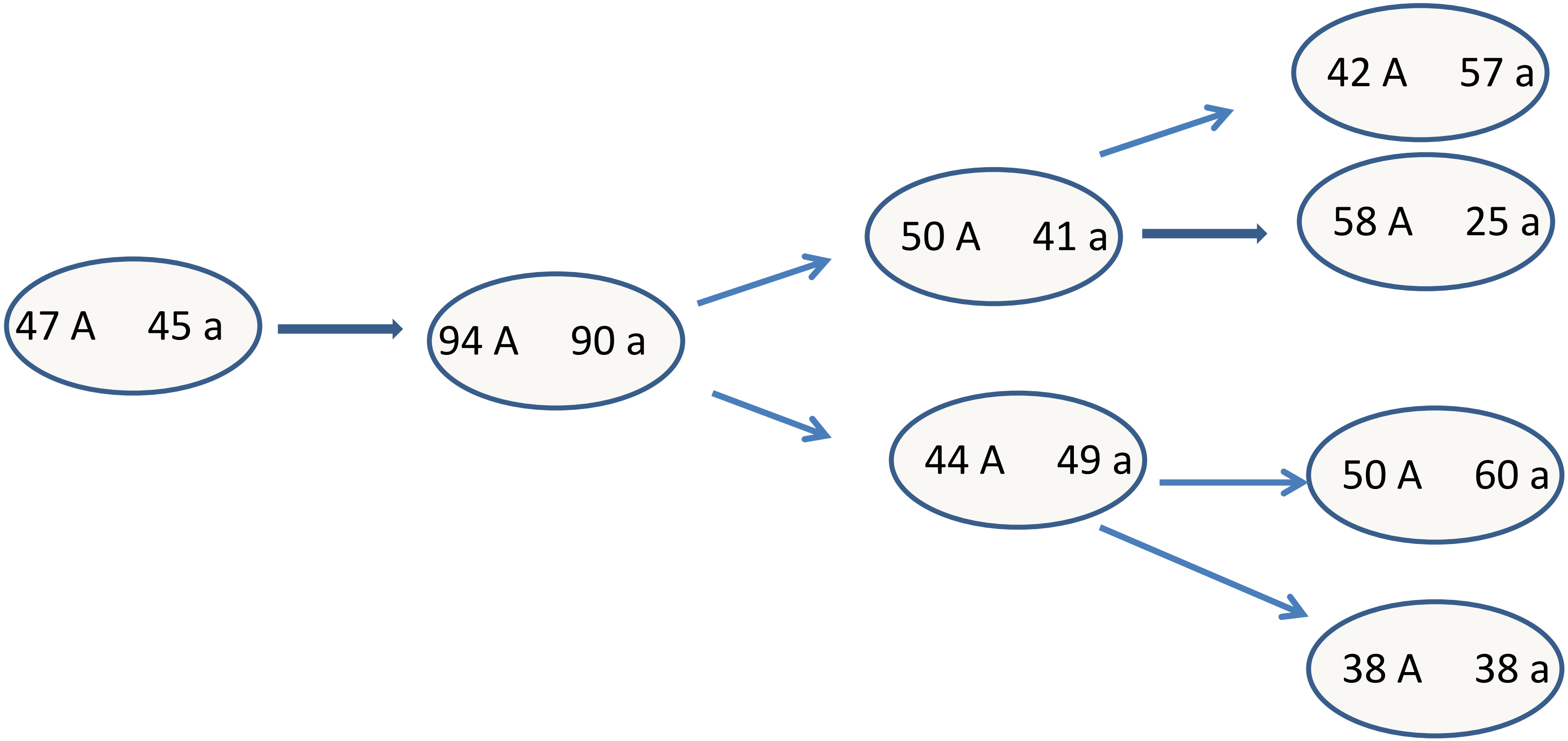
47 A 45 a

43 A 45 a

49 A 41 a

41 A 49 a

Segregación no-mitótica



47 A 45 a

94 A 90 a

50 A 41 a

44 A 49 a

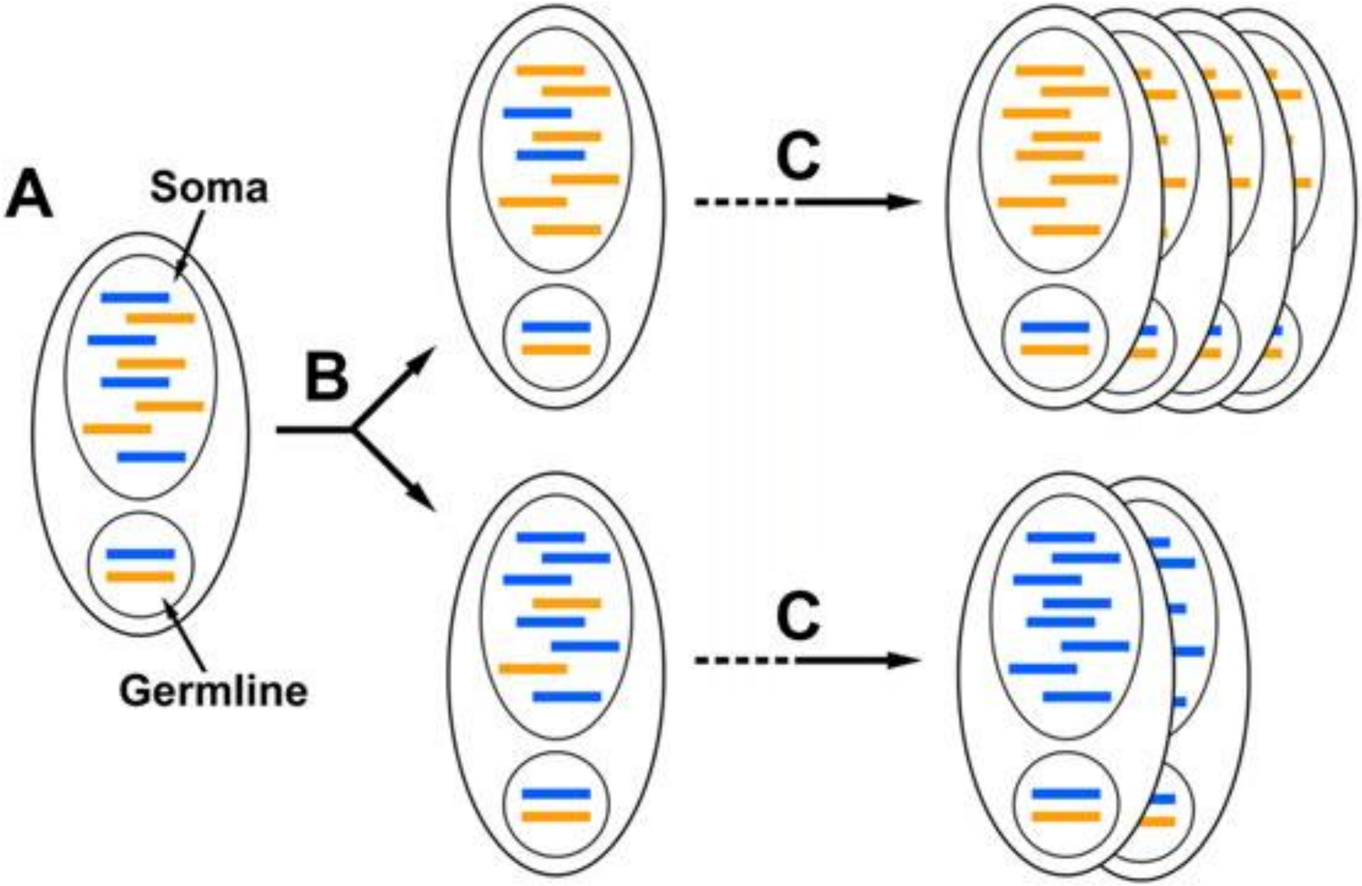
42 A 57 a

58 A 25 a

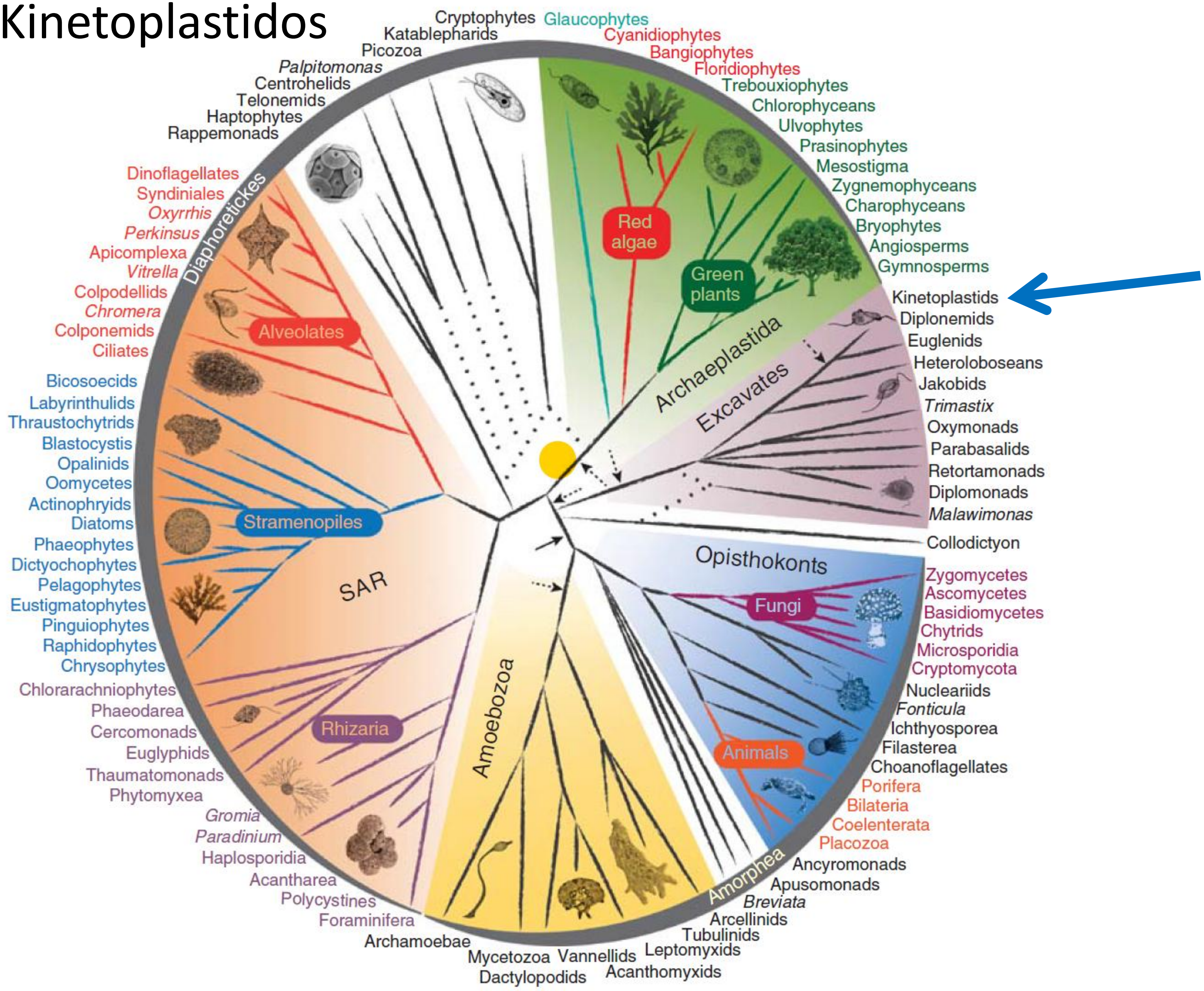
50 A 60 a

38 A 38 a

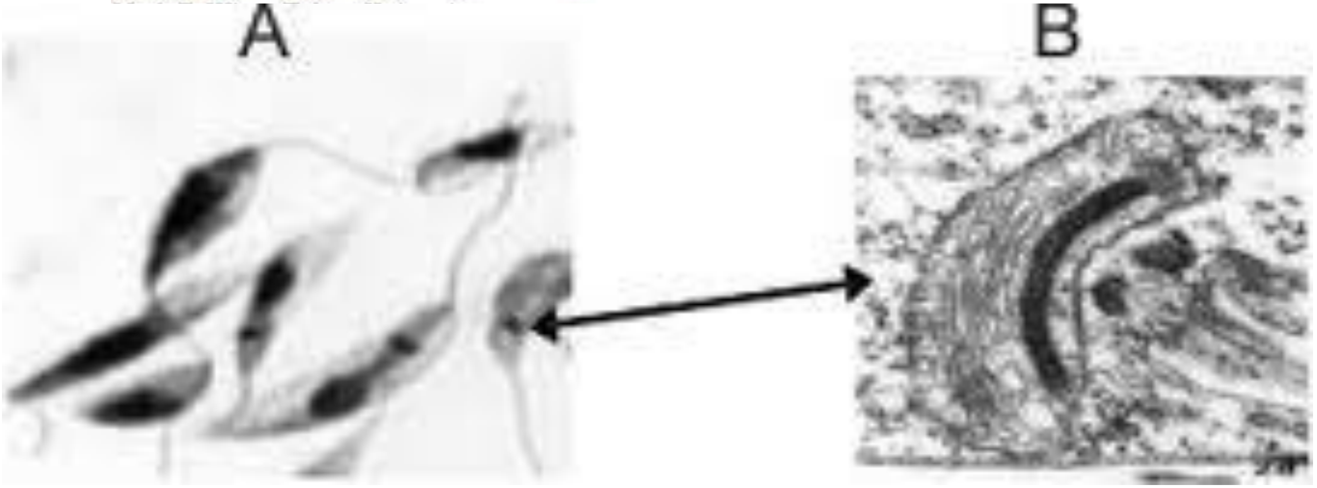
Cuando consideramos muchos cromosomas simultaneamente



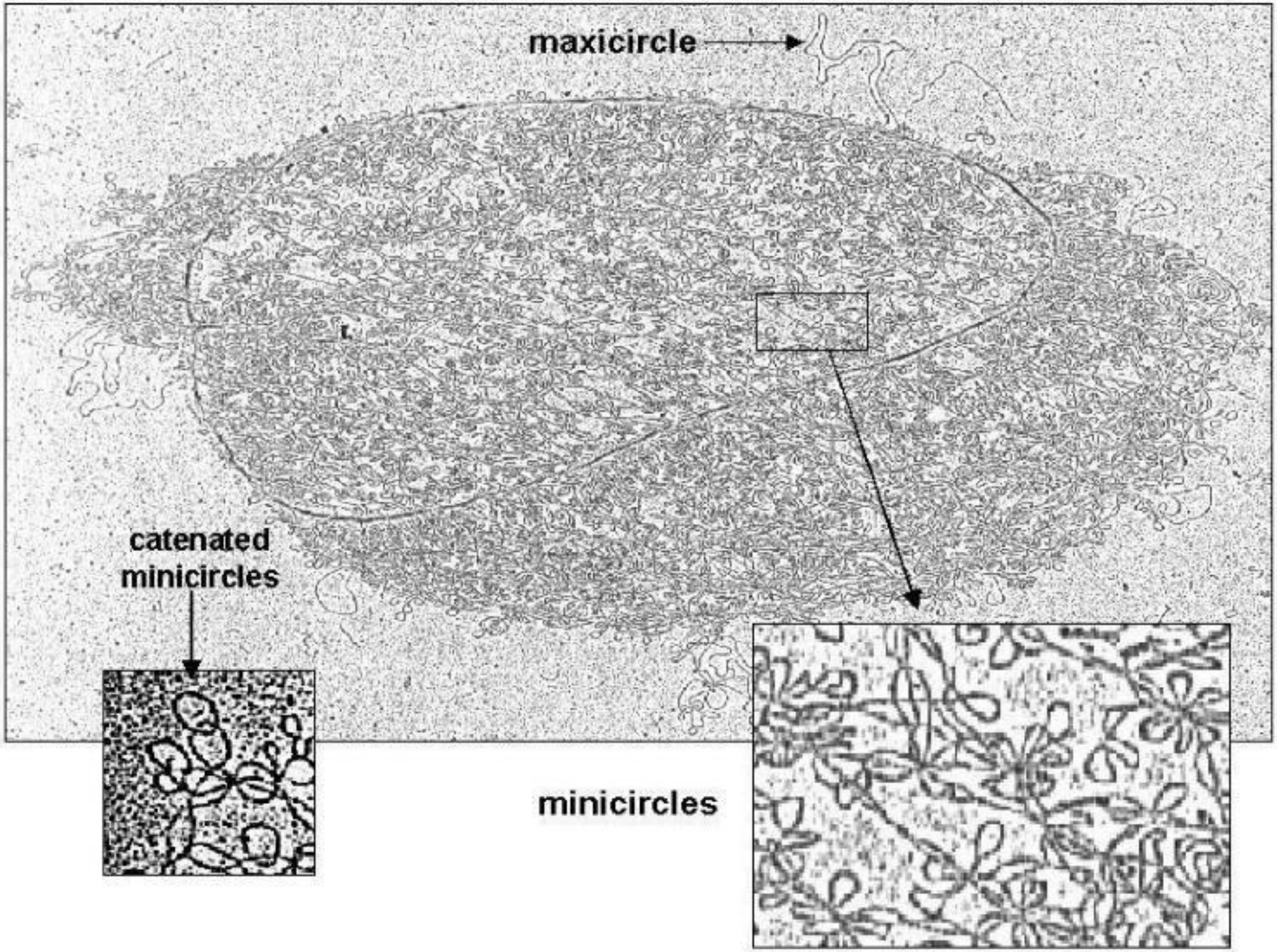
Kinetoplastidos



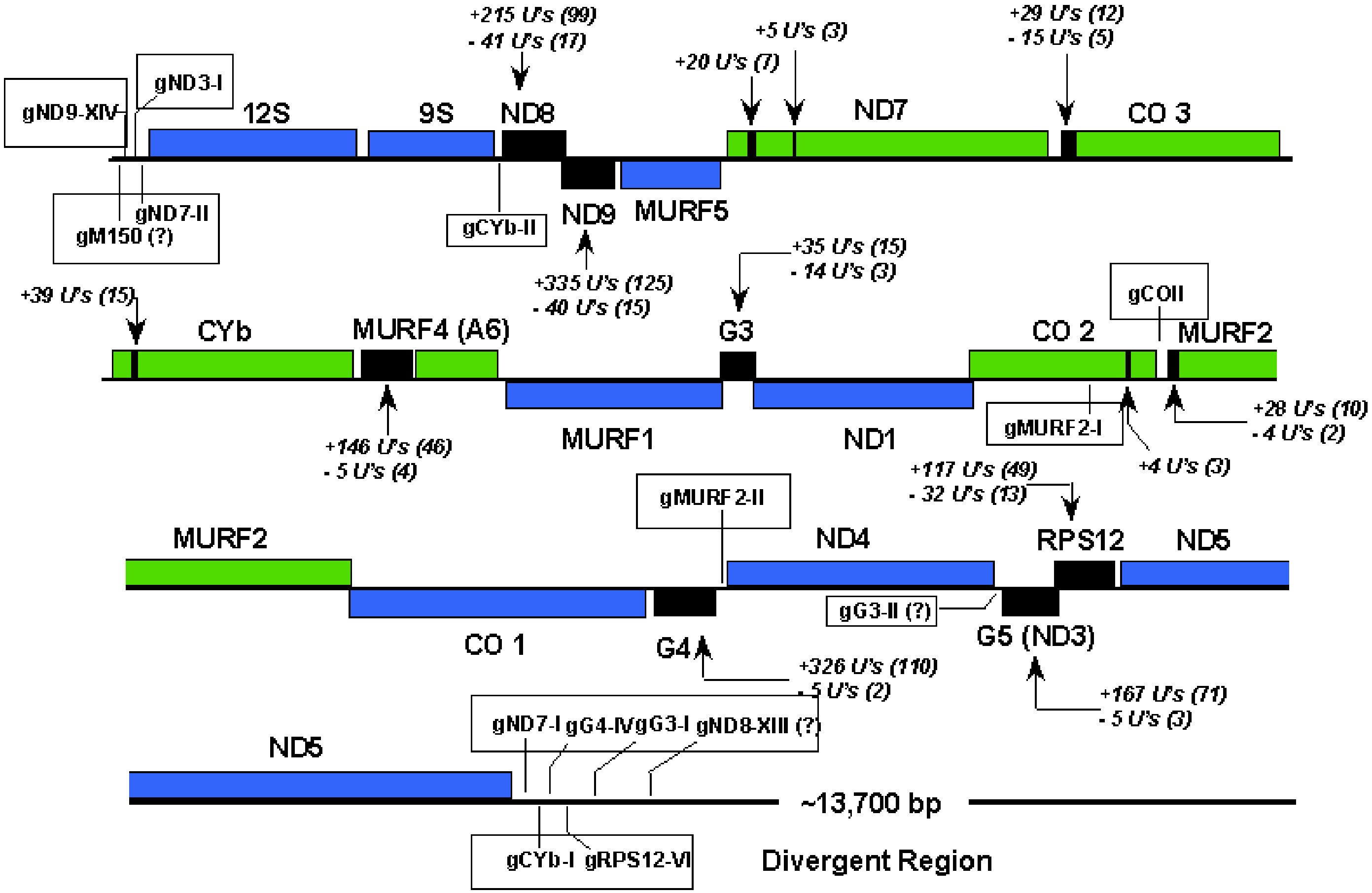
Kinetoplastidos



kDNA network from *L. tarentolae*



Maxicircle genome of Leishmania tarentolae



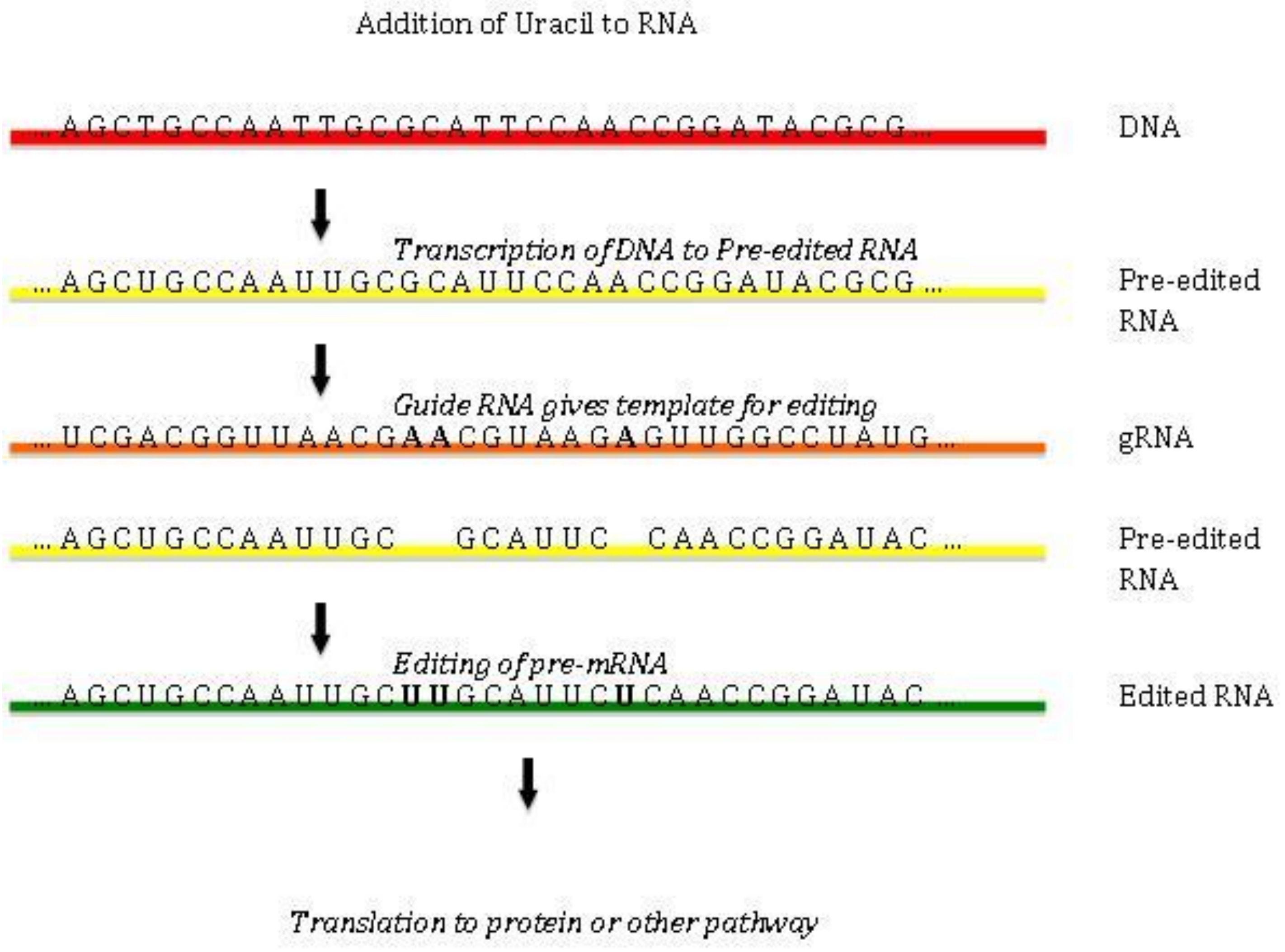
🟩 Edited gene (unedited region)

⬛ Edited gene (pre-edited region)

🟦 Unedited genes

gRNA genes are boxed

RNA editing


Addition of Uracil to RNA
...AGCTGCCAATTGCGCATTCCAACCGGATACGCG...
DNA
Transcription of DNA to Pre-edited RNA
...AGCUGCCAAUUGCGCAUUCCAACCGGAUACGCG...
Pre-edited RNA
Guide RNA gives template for editing
...UCGACGGUUAACG**AA**CGUAAG**A**GUUGGCCUAUG...
gRNA
...AGCUGCCAAUUGC GCAUUC CAACCGGAUAC ...
Pre-edited RNA
Editing of pre-mRNA
...AGCUGCCAAUUGC**UU**GCAUUC**U**CAACCGGAUAC ...
Edited RNA
Translation to protein or other pathway

Editing in trypanosomatids

Alignment: Genomic vs Edited (mRNA) sequences

```
COII_genomic      ATGAGTTTTATATTATCCTTTTGATTATTATTTTTAATAGATTCTGTAATAGTTTTATTA
COII_edited       ATGAGTTTTATATTATCCTTTTGATTATTATTTTTAATAGATTCTGTAATAGTTTTATTA

COII_genomic      TCTTTATCTTTTTTTCTCGTTTTATGAATATGTATTTTATTAATTTCTTCTATATTATGT
COII_edited       TCTTTATCTTTTTTTCTCGTTTTATGAATATGTATTTTATTAATTTCTTCTATATTATGT

COII_genomic      CAATGGTATTGAGTTTATTTTTTATTTGGTGACACAACTATTTTTAGTAATTTAATATTA
COII_edited       CAATGGTATTGAGTTTATTTTTTATTTGGTGACACAACTATTTTTAGTAATTTAATATTA

COII_genomic      GAAAGTGATTACATTTTAGGAGATTTAAGAATATTGCAATGTAATCATGTGCTAACTTTA
COII_edited       GAAAGTGATTACATTTTAGGAGATTTAAGAATATTGCAATGTAATCATGTGCTAACTTTA


COII_genomic      TTAAGTTTAGTAATTTATAAATTATGATTATCTGCAGTAGATGTAATACATTCATTCTCA
COII_edited       TTAAGTTTAGTAATTTATAAATTATGATTATCTGCAGTAGATGTAATACATTCATTCTCA

COII_genomic      TTATCAAGTTTAGGTATAAAAGTAGA--G-A-ACCTGGTAGATGTAATGAAATAATTTTA
COII_edited       TTATCAAGTTTAGGTATAAAAGTAGAuuGuAuACCTGGTAGATGTAATGAAATAATTTTA

COII_genomic      TATGCATCAAATGCAGCAACTATTTATGGTCAGTGTAGTGAATTATGTGGTGTATTACAT
COII_edited       TATGCATCAAATGCAGCAACTATTTATGGTCAGTGTAGTGAATTATGTGGTGTATTACAT

COII_genomic      GGATTTATGCCTATTGTAATAAATTTTATATAA
COII_edited       GGATTTATGCCTATTGTAATAAATTTTATATAA
```

Alignment: Genomic vs Edited (mRNA) sequences

```
MURF2_genomic     A-G---GG--G----ATTTTA---AG----A-----G-GCTTTGATTG-AGCCGTATTTT
MURF2_edited      AuGuuuGGuuGuuuuA----AuuuAGuuuuAuuuuuGuGCTTTGATTGuAGCCGTATTTT

MURF2_genomic     TGATTTATTATGTATTAGAACATATGATTTTATATTATGATGATTTGATTTAGATTTCAT
MURF2_edited      TGATTTATTATGTATTAGAACATATGATTTTATATTATGATGATTTGATTTAGATTTCAT

MURF2_genomic     TTTATACGATTTCGTTTTTGATTTTGTTGTTTGCATAACTTTTATCTTTATTTTTGTGTT
MURF2_edited      TTTATACGATTTCGTTTTTGATTTTGTTGTTTGCATAACTTTTATCTTTATTTTTGTGTT

MURF2_genomic     GGGTTTTTTTTTGAGGATTTTTTTCAGTTTTGTATTTGTCTTATTATTTATAACATTTTT
MURF2_edited      GGGTTTTTTTTTGAGGATTTTTTTCAGTTTTGTATTTGTCTTATTATTTATAACATTTTT

MURF2_genomic     TGGTTTTTTTGCAACAGCTTTATTATATTCTGGATATTATTTATTTTATATATATATATT
MURF2_edited      TGGTTTTTTTGCAACAGCTTTATTATATTCTGGATATTATTTATTTTATATATATATATT
```

Pan editing (criptogenes)

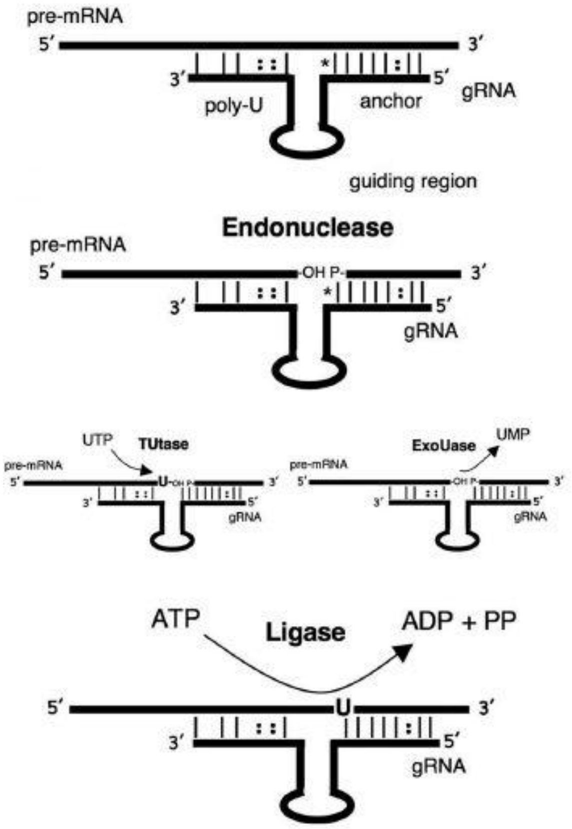
A6 Genomic, Edited (mRNA) sequences

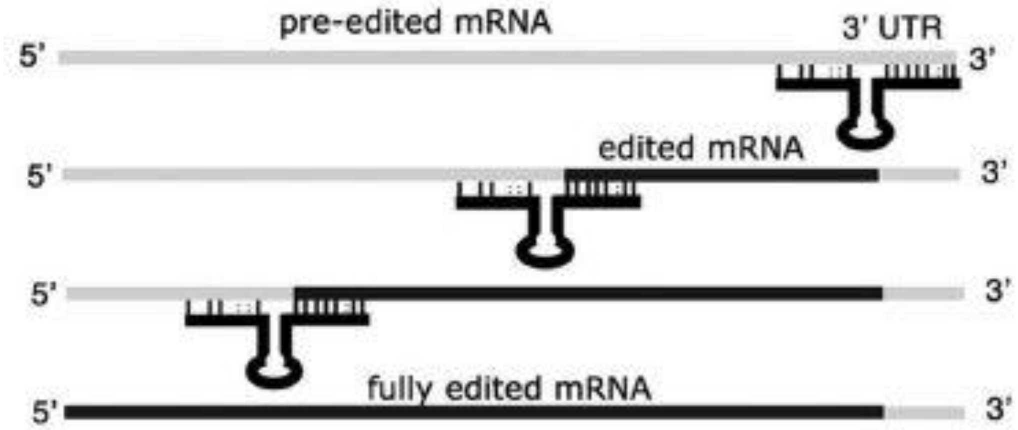
```
Un A-G-----G-----------G-GA---GTTTTGTG--GCG---G--ATTTTTTA-GTG-A-A--A--G-G-G-GA-C-AGG--A-G
Ed AuGuuuuuGuuuuuuuuuuuGuGAuuuGuuuuG-GuuGCGuuuGuuATT----AuGTGuGuAuuAuuGuGuGuGAuCuAGGuuAuG

Un ----G--G-G-A----AATTTTTG---GA-G--AA-----GTA------G--G----G---G---GA---G-A---G---A--GG
Ed uuuuGuuGuGuAuuuuAATT---GuuuGAuGuuAAuuuuuG-AuuuuuuGuuGuuuuGuuuGuuuGAuuuGuAuuuGuuuAuuGG

Un ---A-G---A-----G--A--G-GG---A-G--G---ATTTTA---G-A-AG---GATTT-G-A--ATTTG-A—-ACCTTTTTA-
Ed uuuAuGuuuAuuuuuGuuAuuGuGGuuuAuGuuGuuuA----AuuuGuAuAGuuuGATTTuGuAuuATT-GuAuuACCT----Au
```

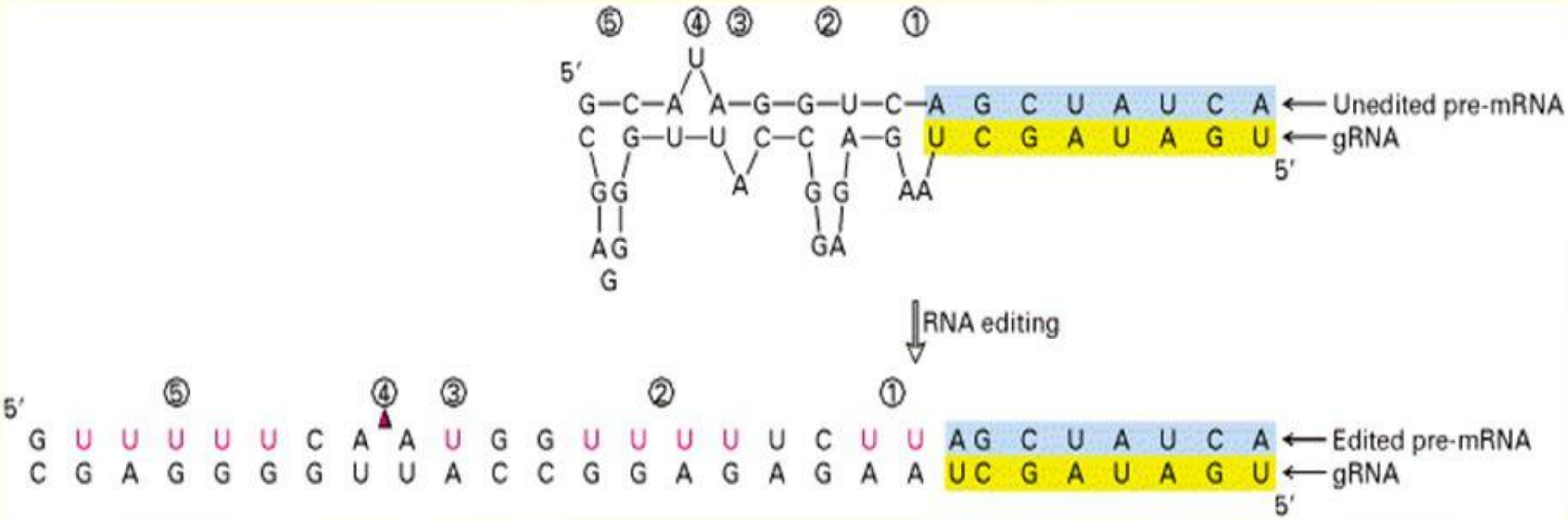
Editing mechanism



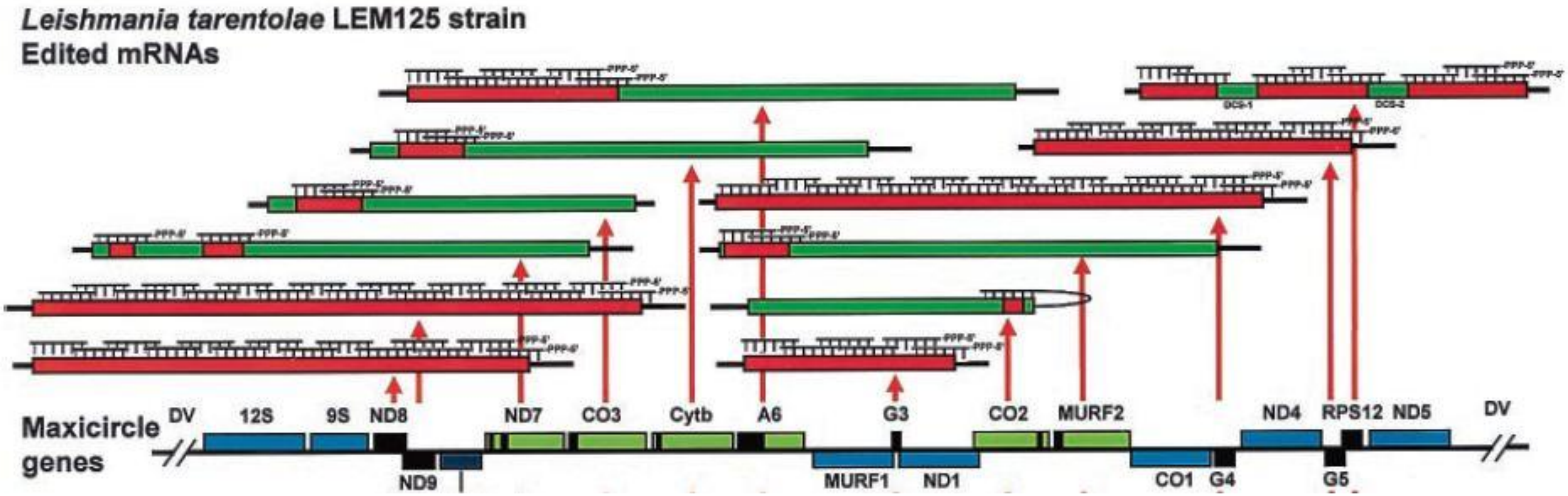


Guide-RNAs (encoded in minicircles)

Mechanism of RNA editing in kinetoplast pre-mRNAs of trypanosomes



En base al número de eventos de editing que se requieren para madurar todos los ARNms, se estima que se requieren entre 200 and 600 tipos de gRNAS



Colloquium

Evolution of RNA editing in trypanosome mitochondria

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