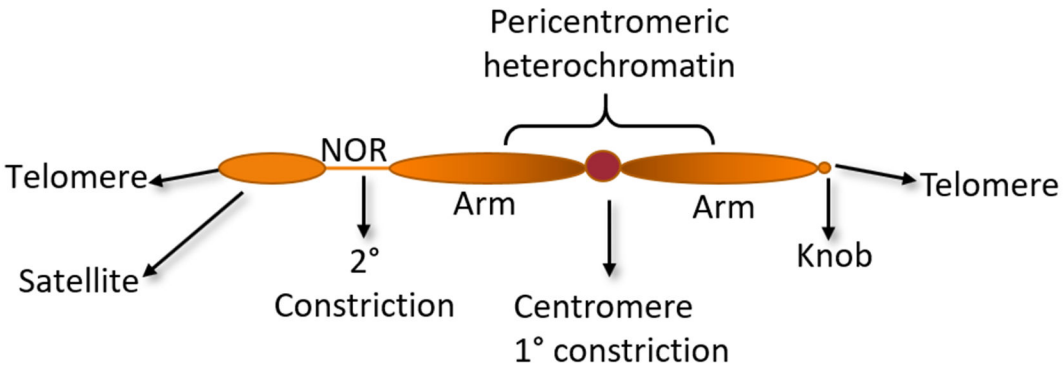
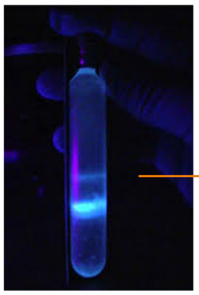
Landmarks and terminology -See review by **Heslop-Harrison and Schwarzacher, 2011**

"Chromo" = colored

"some" = body

-only visible during cell division when DNA condenses & nuclear membrane disappears





- Do not confuse the chromosome satellite with satellite DNA

- The centromere is also known as the 1º constriction.
- Site of the kinetochore, the protein complex which attaches the spindle fibers to the centromere

- After DNA replication, the chromosome is made up of 2 chromatids
  - Sister chromatids
  - After centromere division, each chromatid becomes a chromosome

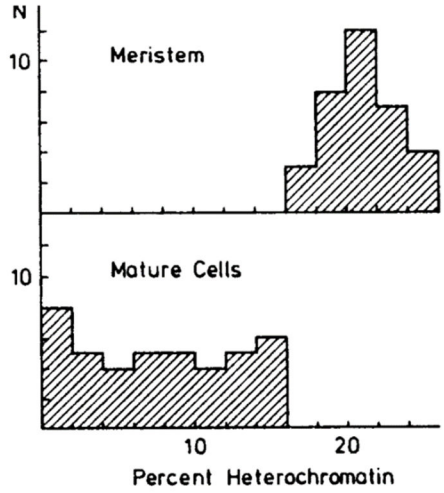
Chromosomes can be categorized according to

		
Metacentric Isobrachial	Submetacentric Acrocentric Heterobrachial	Telocentric

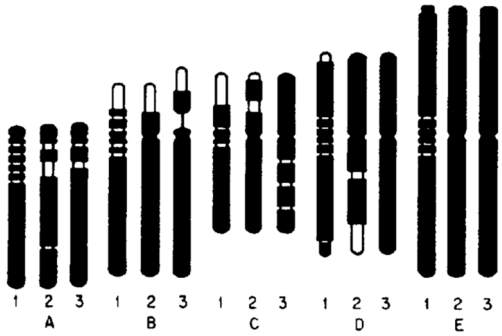
- The material chromosomes are made of is called chromatin, so named by Flemming (on a temporary basis, but it stuck) due to its affinity for dyes.

- **Heitz** (1928, 1933, 1934) discovered euchromatin and heterochromatin

- Euchromatin: Stains lightly; represents areas of the chromosome with genes that are usually transcribed
    - Condensed for mitosis, decondensed during interphase
- Heterochromatin: Stains darkly; always condensed, consists of tightly coiled repetitive DNA
    - Brown & Nelson-Rees (1961, Genetics 46:983-1007)
    - Constitutive: heterochromatin from ♀ and ♂ - derived chromosomes behaves the same during development
    - Facultative: heterochromatin from ♀ and ♂ - derived chromosomes does not behave the same during development. Can also be stage-specific



Changes in heterochromatization of DNA of Dendrobium. Nagl, 1983



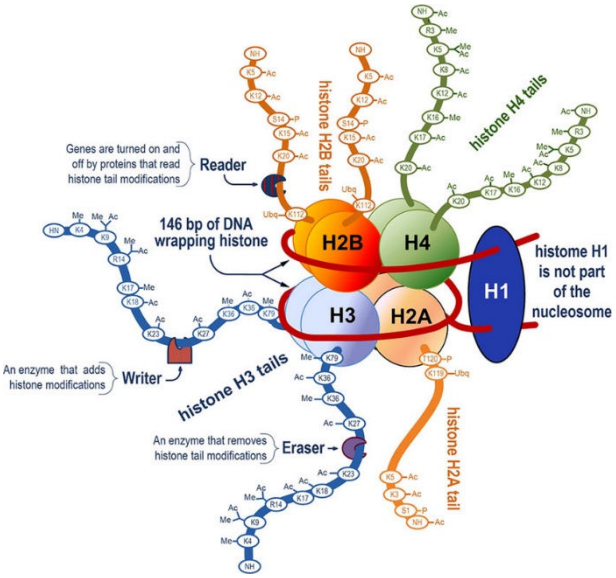
Location of heterochromatin in 1) Trillium erectum, 2) T. grandiflorum, and 3) T. undalatum

Chromatin is about 50% DNA and 50% protein

- 5% = **nonhistone** proteins = acidic proteins
    - DNA binding proteins (Transcription factors)
    - topoisomerases: relax and coil DNA
        - Type I: break single DNA strands
        - Type II: break double DNA strands = gyrases

- 45% of a chromosome is made of **histone** (basic) proteins:
    - H4, H3, H2A, H2B, and H1types
    - H3 and H4 core are highly conserved in eukaryotes

- have a role in chromosome structure and regulation of function
- acetylation, phosphorylation, ubiquination, and methylation of the tails

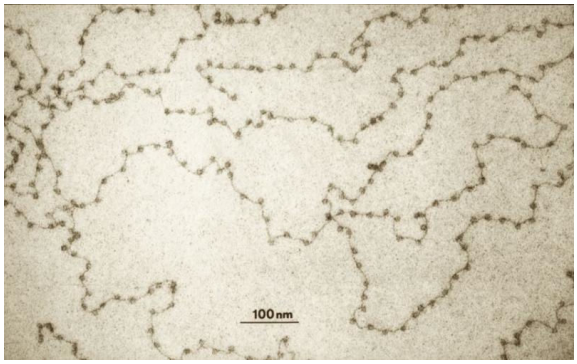


https://doi.org/10.1021/acs.analchem.7b05007

Chromatin organization

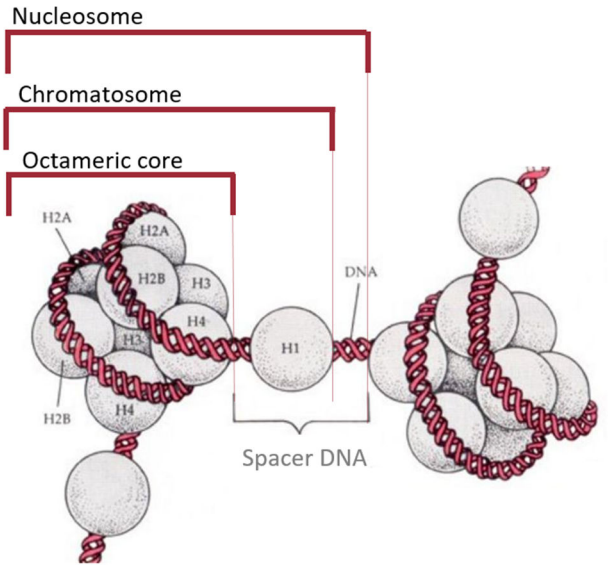
- The nucleosome
  - discovered by Hewish and Burgoyne in 1973
  - basic structural unit of chromatin

A string of nucleosomes forms the **10-nm fiber**



https://www.ornl.gov/blog/ornl-review/beads-string-discovering-nucleosome

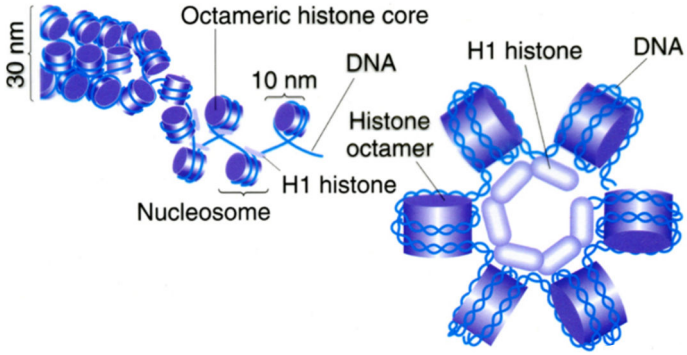
- **Core particle:** 146 bp DNA + histone octamer
- **Chromatosome:** Core particle + H1 + 22 bp DNA tail
- **Nucleosome:** about 200 bp DNA + chromatosome



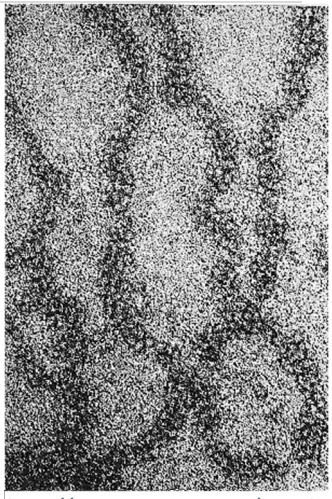
1Oak.cats.ohiou.edu/~ballardh/pbio475/Heredity/Hierarchical-arrangement.JPG

Interphase configuration

The 10-nm fiber is in turn coiled into the **30-nm fiber** or **solenoid**-- helical configuration-- normal arrangement in the chromosome



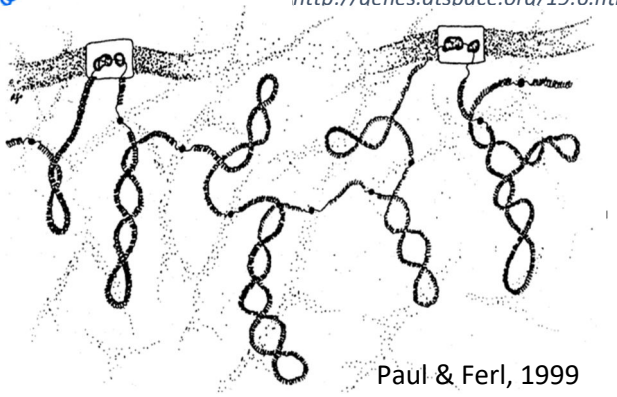
http://www.discoveryandinnovation.com/BIOL202/notes/lectu



http://genes.atspace.org/19.6.html

The 30-nm fiber in turn forms loops attached to the nuclear protein core.

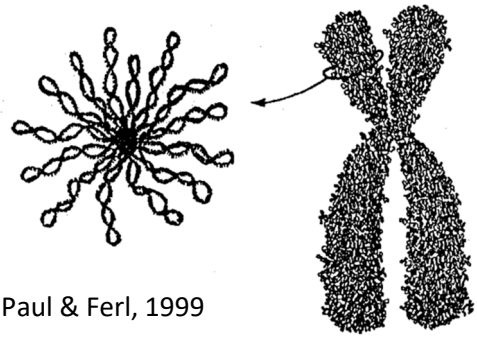
SARs = Scaffold attaching regions delineate the loops. Also called **MARs = matrix attaching regions**. - Loops are 4-13 kb long, and are thought to delineate transcriptional domains.



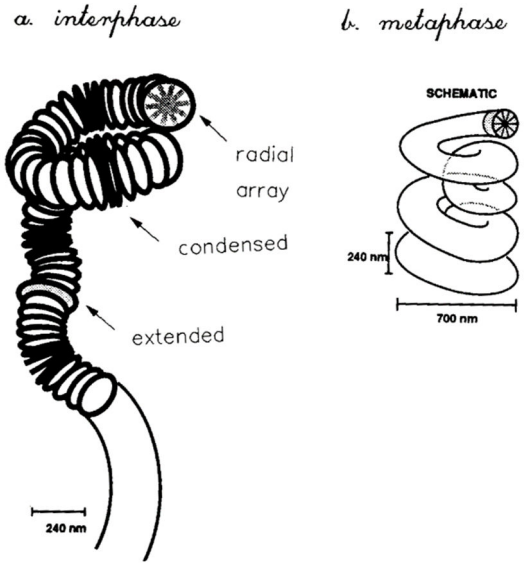
Paul & Ferl, 1999

Mitotic configuration

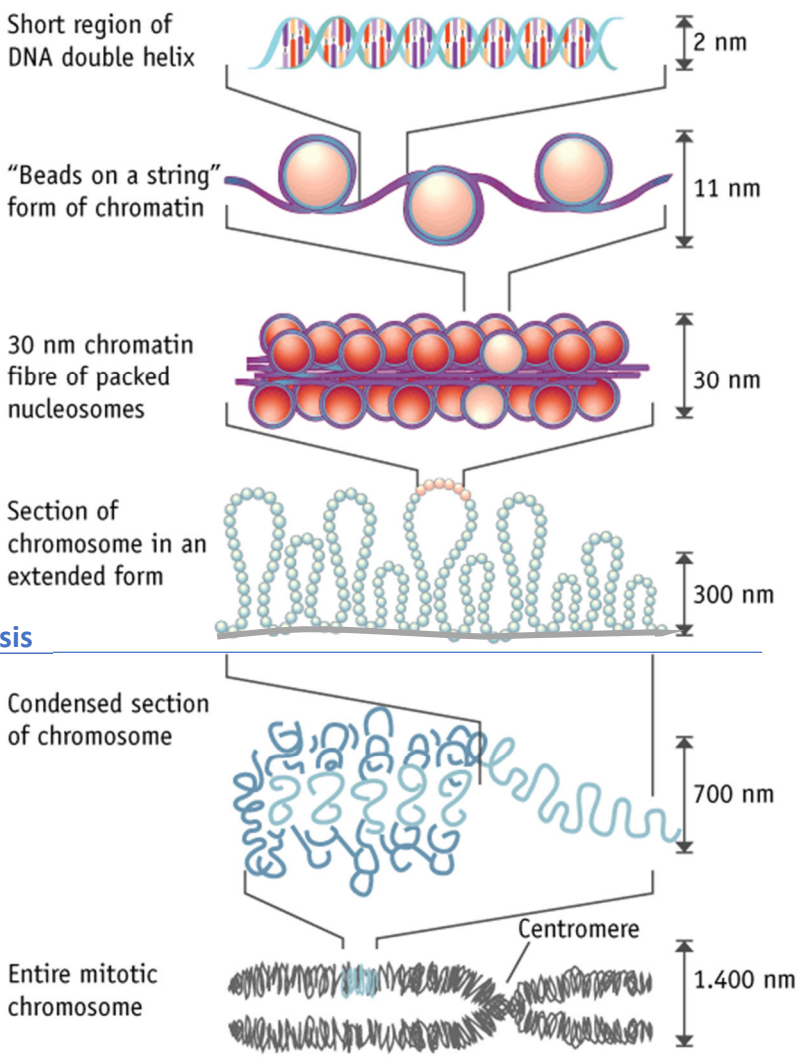
Finally, the solenoid is itself coiled into 240-nm fibers (the normal interphase chromosome) and a 700 nm fiber at metaphase.



Paul & Ferl, 1999

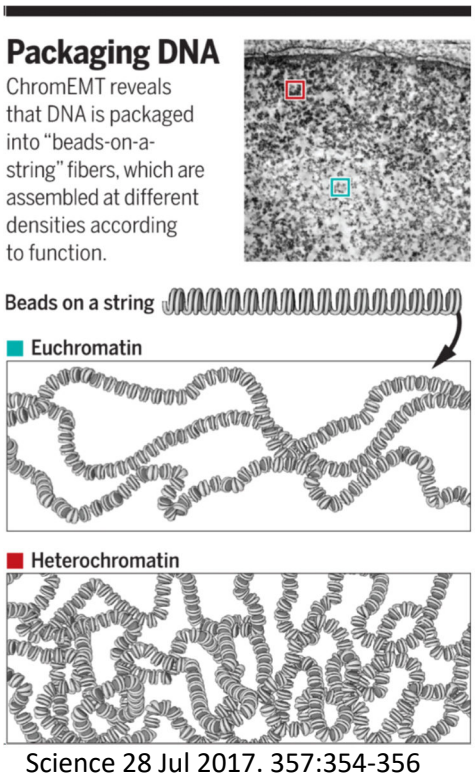


- Interphase configuration appears to be an artifact of isolated DNA



May only exist in isolated DNA

Chromatin organization

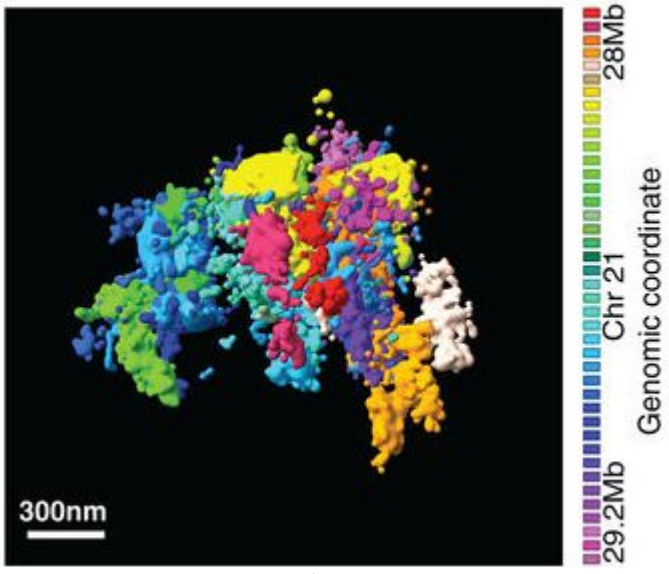


Science 28 Jul 2017. 357:354-356

- The emerging view for interphase is that Eu- and Hetero chromatin differ by the density of the 10-nm fibers

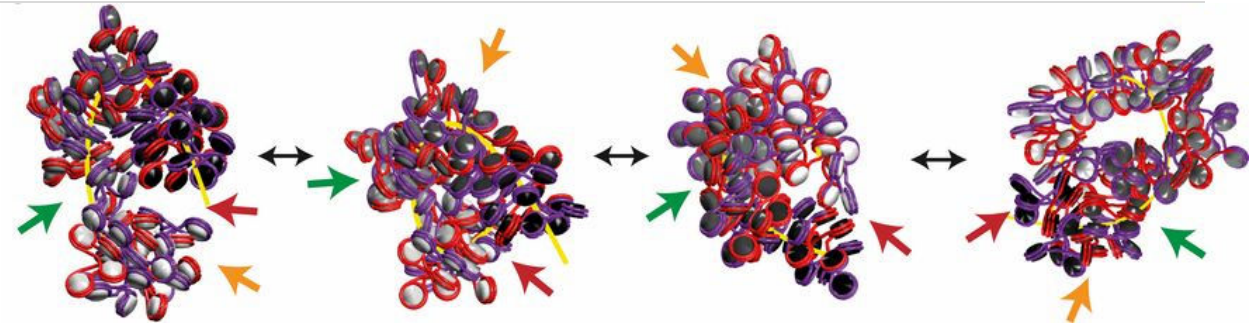
Chromatin organization

- Chromatin forms clusters



Bintu et al., 2018; DOI: 10.1126/science.aau1783

- These clusters are dynamic & mediated by H1



Sridhar et al, 2020. https://doi.org/10.1073/pnas.1910044117

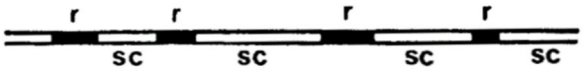
Landmarks & DNA arrangements

CHROMOSOME LANDMARKS

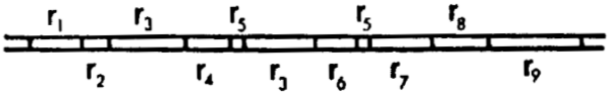
- The major chromosome landmarks are associated with repetitive DNA (Flavell 1980):

a. **Interspersed**:

- short pieces (50-2000 bp) interspersed with single copy DNA



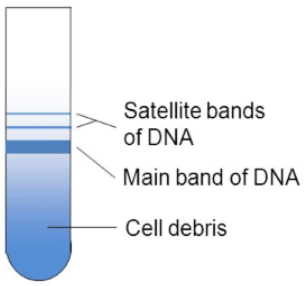
-unrelated short repeats interspersed with each other in different permutations



b. **Tandem**

- identical repeating units

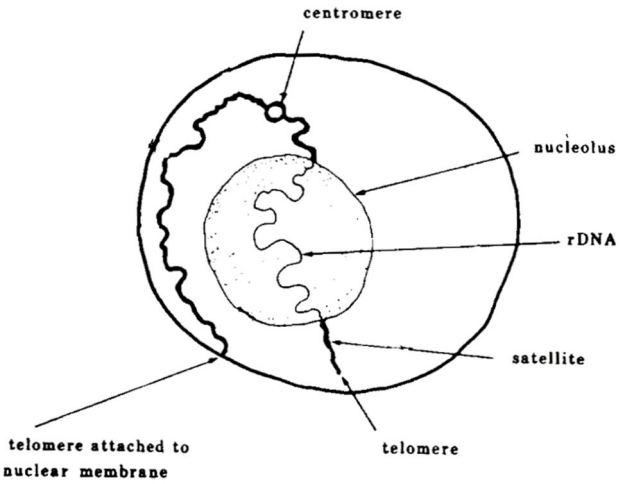
- Tandem repeats are frequently associated with satellite DNA, hence the original name of microsatellites for these markers



http://duwyjaza17.dva4.ru.net/go/ebevaruwu-satellite-dna-ppt.html.

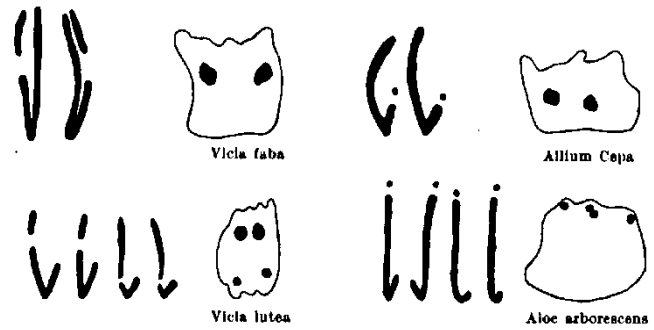
THE NUCLEAR ORGANIZING REGION

- The nucleolus is a spherical organelle found inside the nucleus
- It forms in late telophase, and disappears in late prophase—

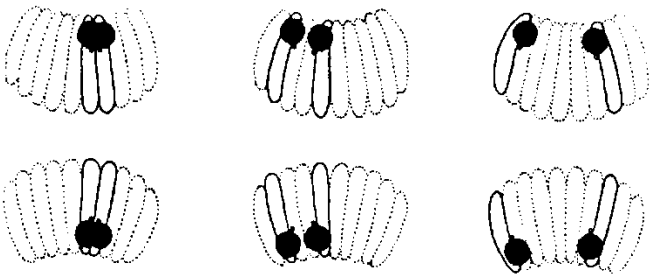


1931: Heitz

- Looked at 12 different plant species
- Concluded that individuals had a constant maximum number of nucleoli, and wondered why.



- Maximum number of nucleoli = the maximum number of 2̂ constrictions



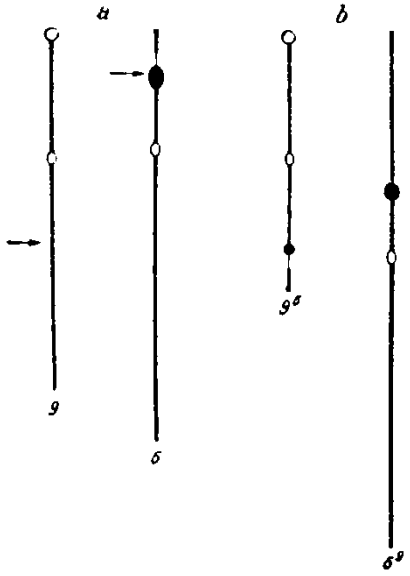
- Nucleoli form in specific regions during late telophase

From *Passarge 1979*

1934: McClintock

Question - what does the 2° constriction do?

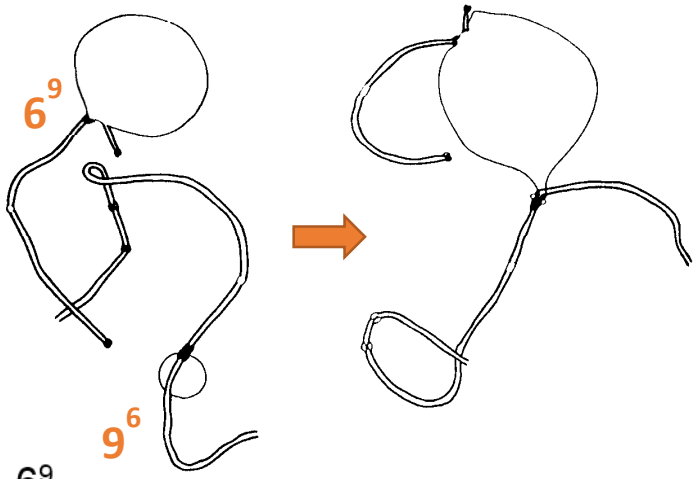
- Induced translocations between chromosomes 6 and 9 of maize:



- Chromosome 6 normally contains the 2°

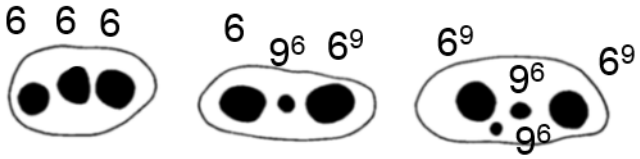
- Break points were as indicated:
- Both 6^9 and 9^6 now had a portion of the 2°

- Result was that both 6^9 and 9^6 would form nucleoli
- The two nucleoli would later coalesce into one



- Chromosome 6^9 has the larger 2°, and consequently, the larger nucleolus.

Root-tip cells of trisomics:



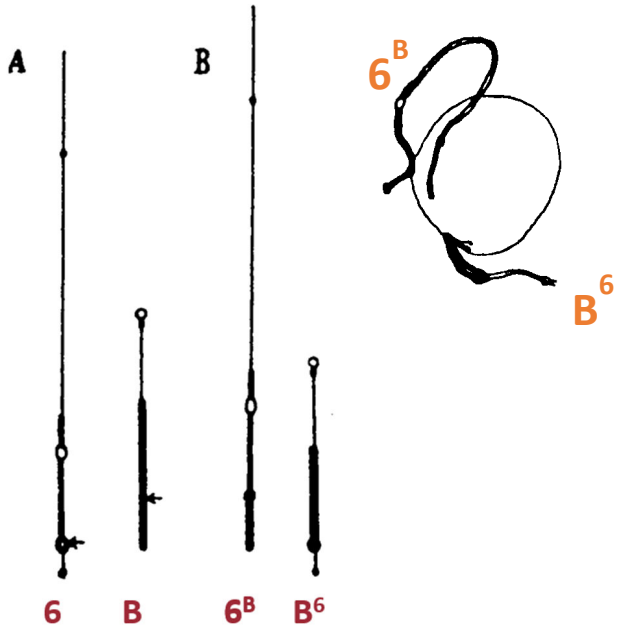
- After meiosis, breaks in the chromosomes with translocations resulted in microscopes without any 2°.
- These had no nucleolus, but had droplets of nucleolar material throughout the cell

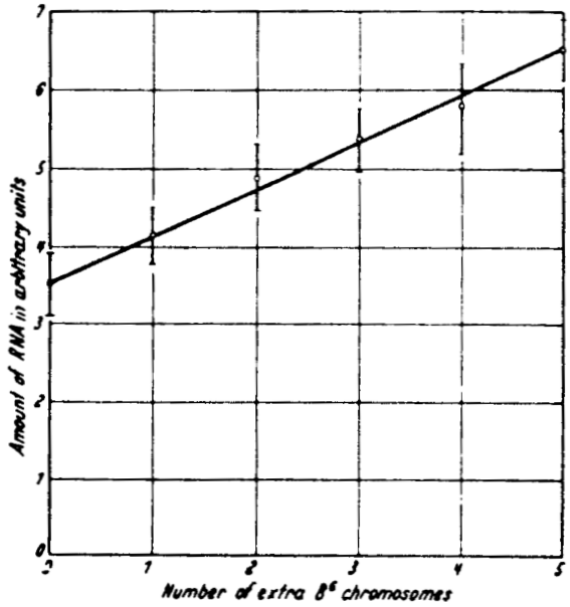
- Take home message– the size and number of nucleoli corresponded to the size and number of 2° constrictions.
- The inference is that the 2° constriction is where the nucleolus forms.
- Therefore the 2° constriction = the nucleolar organizing region (NOR)

1955: Lin

Question - what does the NOR do?

- Used chromosome 6/B chromosome translocations in maize:

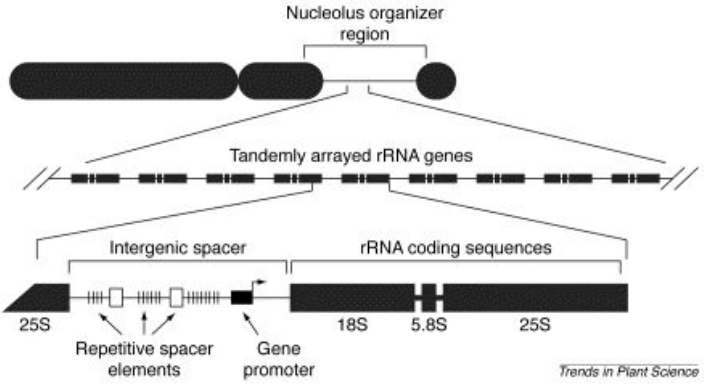




- Obtained cells with 1-5 extra NORs, using B^6 chromosomes
- Microsporocytes have only one nucleolus at prophase I, regardless of number of NORs
- Measured the amount of RNA by absorption of UV light at 2600 nm:
    - Got ~14% increase in RNA concentration per B^6 chromosome.
    - Adding another B chromosome (not a B^6) resulted in only a 2% increase in [RNA]
    - ∴ **conclude** that the NOR controls the amount of RNA in the nucleus.

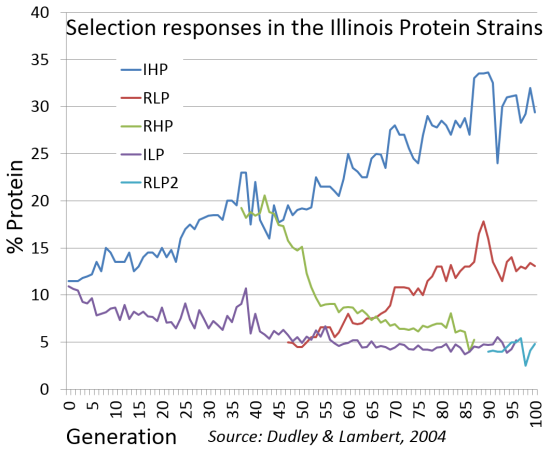
- From other work, know that the nucleolus is the site of rRNA synthesis.
- NOR is the site of the rRNA genes = rDNA
    - It forms on the 2° constriction of chromosomes

- The 2° consists of the rRNA genes
- Is site of ribosome synthesis



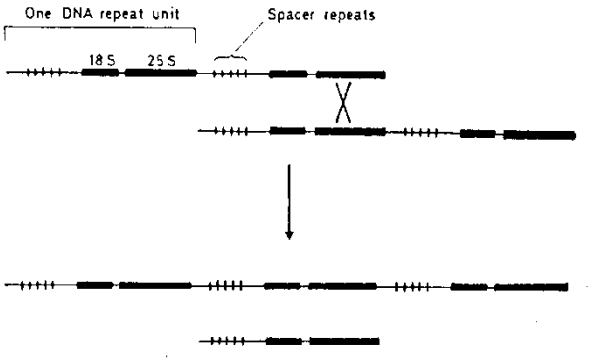
1971, 1978: Phillips

- Curcurbits have 27,000 rRNA genes
- Jerusalem artichoke has 1400 rRNA genes
- Corn:
  - "Sticky mutant" 3,300 copies
  - W23 5,000 copies
  - W127 17,000 copies
  - "Reverse high protein" 23,100 copies



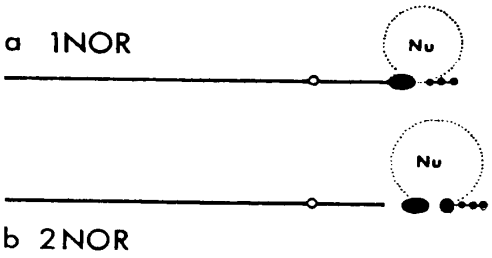
The Illinois long term selection experiment started in 1896 and continues to the present.

- Such variability is a hallmark of repetitive DNA, which varies considerably in copy number due to displaced pairing prior to crossing over - Flavell, 1985:



- Found a stock with a duplicated 2° constriction region:

  (The 2° constriction is immediately to the right of a heterochromatic region, here depicted as a large knob. The duplication includes part of the heterochromatic region in addition to the 2° constriction).

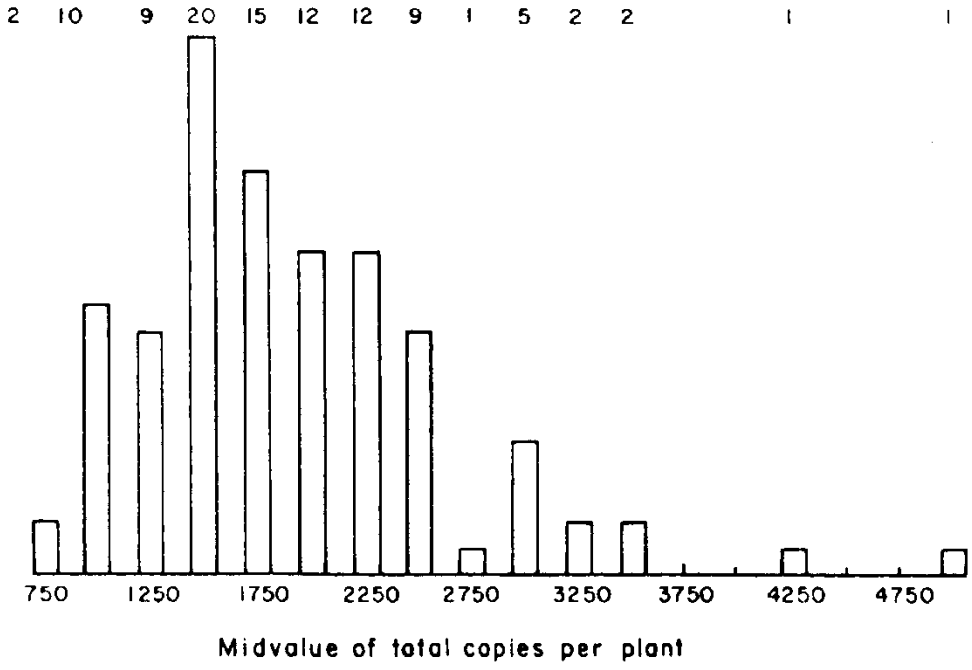


  - This line had twice the amount of rRNA genes.

- Also looked at:
  - Trisomic for chromosome 6: had 50% more rDNA copies
  - Monosomic for chromosome 6: had a 50% reduction in rDNA copy number
  - B^{6NOR}: Changed number of rRNA genes with the addition of NORs

1990: Zhang et al (Allard):

- Surveyed a natural population of *Hordeum spontaneum* for rDNA copy number:



- Excess number of rDNA meets need for peak demands
- Organisms with a low number of rRNA genes resort to some sort of amplification of this region

- Above a minimum number of copies, copy number does not affect fitness of the individual. Particular alleles may affect fitness, but not their number, i.e., quality is more important than quantity.

Fultz et al, 2023

Arabidopsis NORs 1st to be sequenced

- Arabidopsis has 2 NORs = ~6.5% of genome
    - Each has unique sequence variations
    - Expression correlated with sequence homogeneity
        - Does concerted evolution depend on gene transcription?

- NOR2 -~5.5 Mbp
    - Most genes epigenetically silenced
    - In Columbia, but not other genotypes
- NOR4 - ~3.9 Mbp
    - rRNA gene activity in central region

TELOMERES

Reviews by **Peska & Garcia, 2020** & **Shakirov et al., 2022**

Herman Muller, 1938, coined term from (*telos* = end & meros = *part*) [Collect. Net 13:181-198]

- The ends of the chromosomes consist of tandem repeats which have been highly conserved throughout evolution

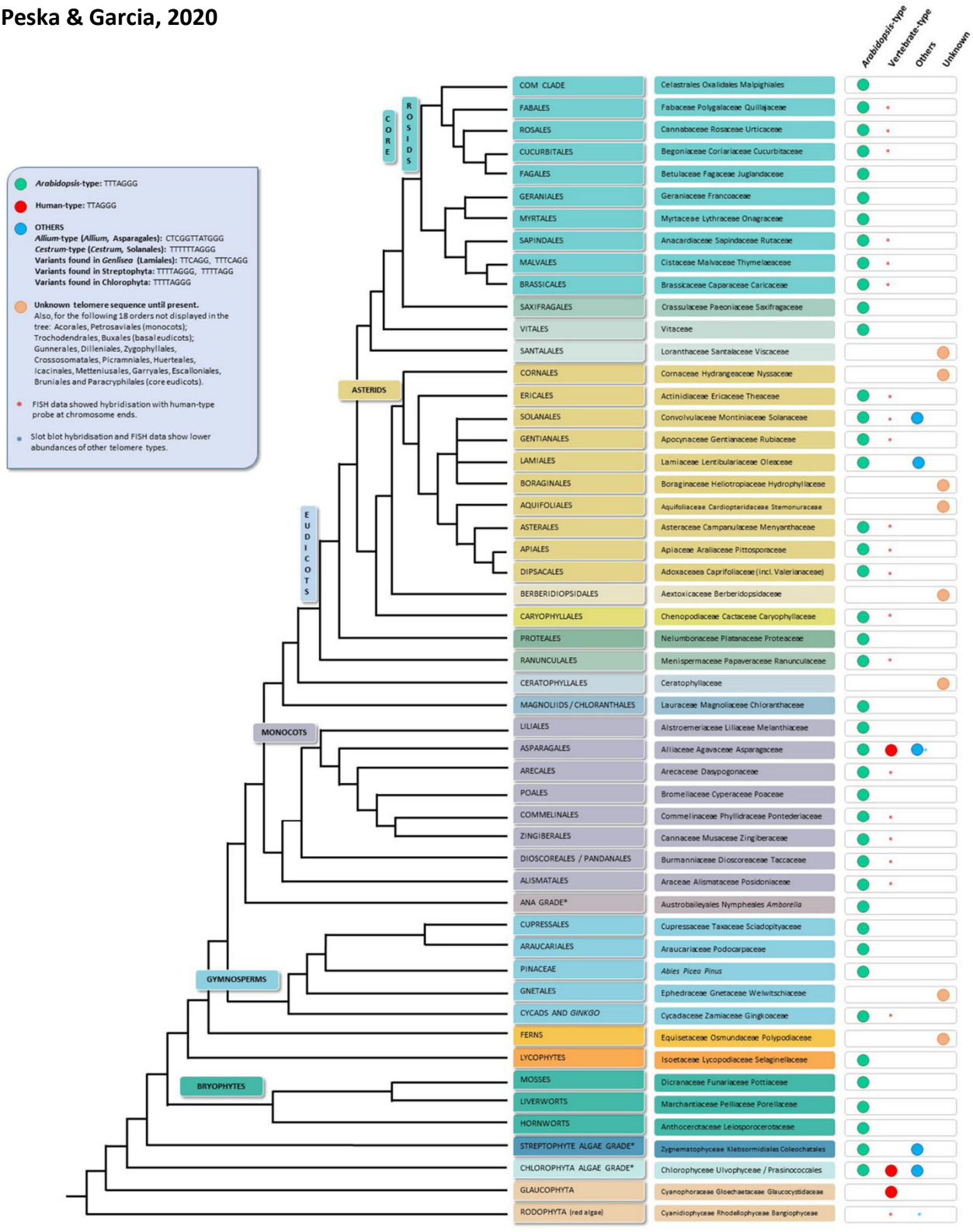
Organism	Type	Telomere sequence
Euplotes	protozoan	TTTTGGG
Tetrahymena thermophila	protozoan	TTGGGG
Vertebrates, slime molds, and trypanosomes		TTGGGA
Arabidopsis thaliana	angiosperm	TTTAGGG
Dictyostelium		AG_{1-8}
Plasmodium		(C/T)TTGGGA
Baker's yeast		TG_{1-3}

- The number of copies differs for between species and between chromosomes:

Maize		= 150-380 copies/chromosome
Rye & barley	1-6 kb/chromosome	= 140 - 850 copies/chromosome
Humans	3-20 kb/chromosome	= 500 - 3000+ copies/chromosome

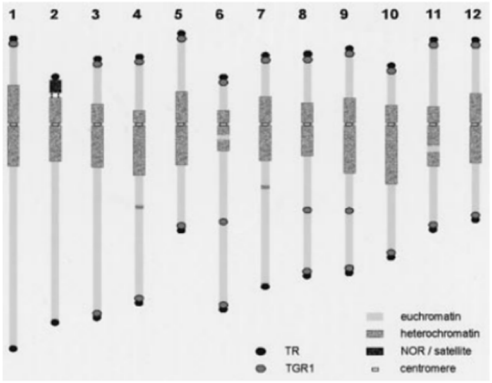
- Copy number is stable within a genotype, but length appears to be controlled by 3 genes in maize (Burr et al., 1992, Plant Cell 4:953-960)
- Amaryllidaceae (onions et al), Solanaceae, & Lentibulariaceae lack arabidopsis-type telomeres, these having been replaced by either transposon DNA or rDNA (Peska & Garcia, 2020)

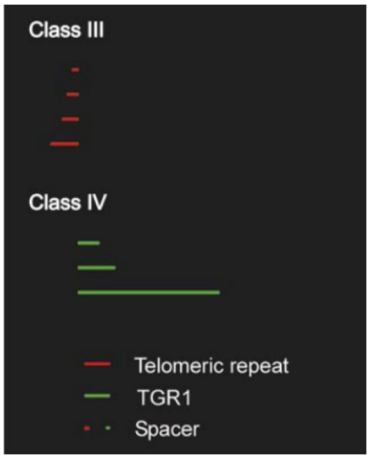
Peska & Garcia, 2020



Subtelomere repeats – Zhong et al, 1998

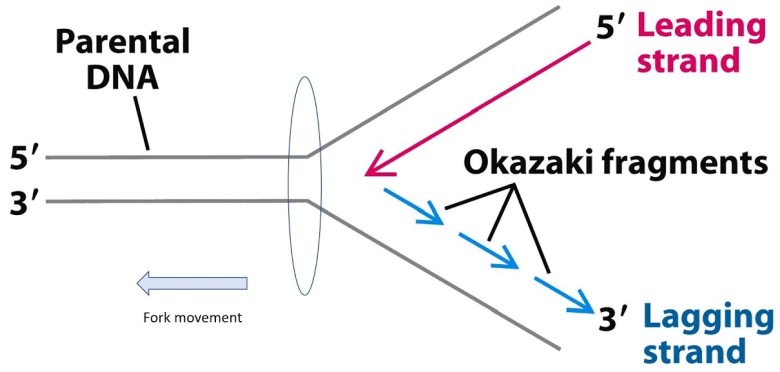
- Often associated with more complex repeats, called subtelomeric repeats
  - are species-specific
  - E.g., tomato, Zhong et al, 1998
    - 162-bp subtelomeric repeat present in 20 of 24 ends
    - 27 combinations of telomere/subtelomere arrangements
    - each chromosome has an individual arrangement
    - separated from telomere by at least 13 kb
    - telomere, spacer, & subtelomeric repeat
    - 2% of genome





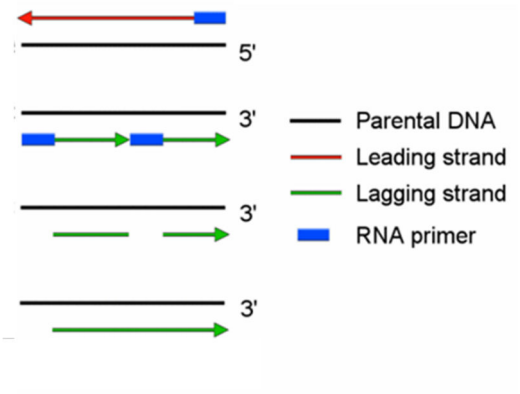
Function:

1. Protect chromosome ends from degradation, fusion, and recombination.
2. Attach to nuclear membrane
3. De novo synthesis by telomerase to replace base pairs lost during replication:
    - DNA synthesis is always 5' to 3'. This will copy bottom strand from one end to the other



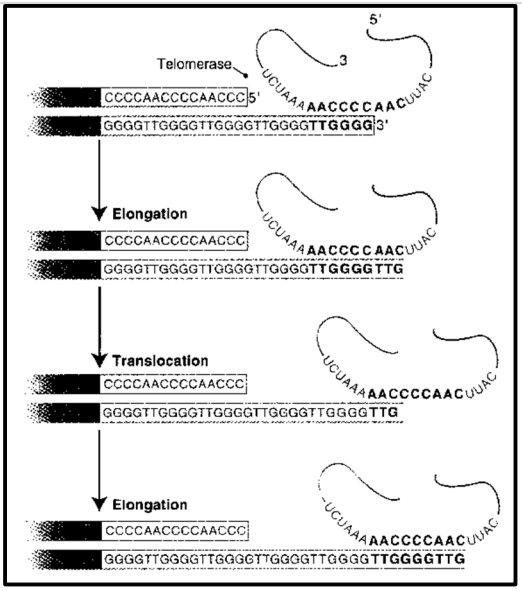
Youtube.com/watch?v=pz3vZ7HDnvQ

- Top strand synthesis requires RNA primers. These are removed, and the gaps, filled, but there is no gap to fill at the end, resulting in the loss of terminal DNA.



- Once the telomere is gone, start losing genes from end of chromosome

- A cell line without telomerase cannot be immortal
  - Lack of telomerase is probably the main reason animal cells are difficult to grow in culture
  - Cancer cells reactivate telomerase, and thus become immortal



Function:

4. Meiosis pairing

Pairing combination	% chiasmate association in the deleted arm
Control	
t1BL / 1B	97
Long arm deletion homozygous	
del1BL-4 (0.97)	83
del1BL-5 (0.84)	86
Long arm deletion hetrozygous	
t1BL / del1BL-4 (0.97)	11
t1BL / del1BL-5 (0.84)	6

Gill & Friebe

Lukaszewski, 1997

- Used small terminal deletions, when heterozygous in isochromosomes of wheat, prevent pairing.

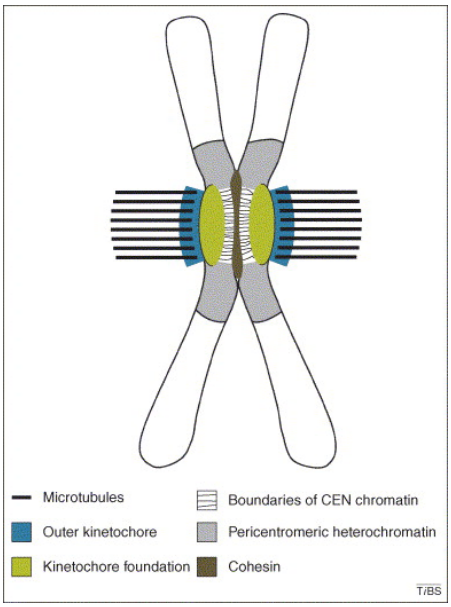
- No interference with pairing if deletions are homozygous

CENTROMERES

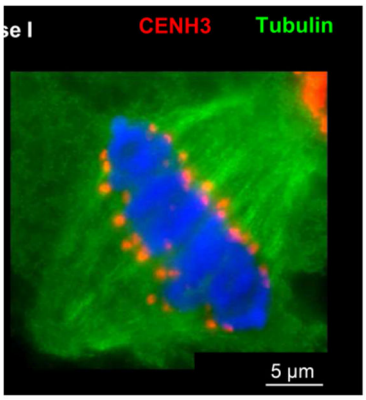
Based on **Zhang et al., 2005; Topp et al., 2004** (Dawe Lab), and **Birchler et al., 2011; Presting, 2018; Naish, 2024**

1º constriction in a chromosome, which is also defined:

- As an area with no recombination
- As having the ability to recruit **kinetochore** proteins to which spindle fibers attach
  - spindle can then separate chromosomes or chromatids during mitosis and meiosis
  - kinetochore proteins are highly conserved
- By its unique H3 centromeric histone, called CenH3
  - Means centromeres are epigenetically defined
  - In maize, serine50 gets phosphorylated in active centromeres



- Once thought of as heterochromatin
  - Now known to be euchromatic, and to have expressed genes in it
  - Non-genic portions consist of a mixture of centromeric retrotransposons (**CRX**s) and satellite repeats (**CentX**), where X= name of species
- In maize, some CRM subfamilies have the most CenH3
- Non-genic centromeric sequences are actively transcribed, and RNA accumulates at the kinetochore, being bound to the histone proteins



CenH3 localization in mitosis. V Schubert, A Ruban & A Houben. 2016. Chromatin ring formation at plant centromeres. Frontiers in Plant Science. 7: 28

Based on ***Richards & Dawe, 1998; Jin et al., 2004***

3 main types of centromeres

1) Yeast centromeres – 125 bp unique sequence
   - Centromere forms anywhere sequence is present
2) Animal centromeres – simple class of repetitive DNA – eg, human α satellite DNA
   - Satellite DNA not to be confused with a chromosome satellite
   - Term comes from a satellite band of DNA formed upon centrifugation in CsCl
3) Other eukaryotes- complex patterns

- Plants - have tandem repeat families
- Also contain low-copy sequences, transposable elements, and telomere repeats
  - devote several 100 kb to Mb of DNA
  - eg, arabidopsis centromeres are 3.55 to 4.4 Mbp/chromosome
  - no conservation between major lineages of plants

- In general, reference genomes of crop plants do not include the centromere. But, there are a few high-quality plant centromeres sequenced

***Nagaki et al.*, 2004; *Wu et al.*, 2004**

Rice centromere 8 has been cloned and sequenced.

Wu et al:
- 1.97 Mb long
- defined by pericentromeric flanking sequences
- mostly retrotransposons
- 201 ORFs

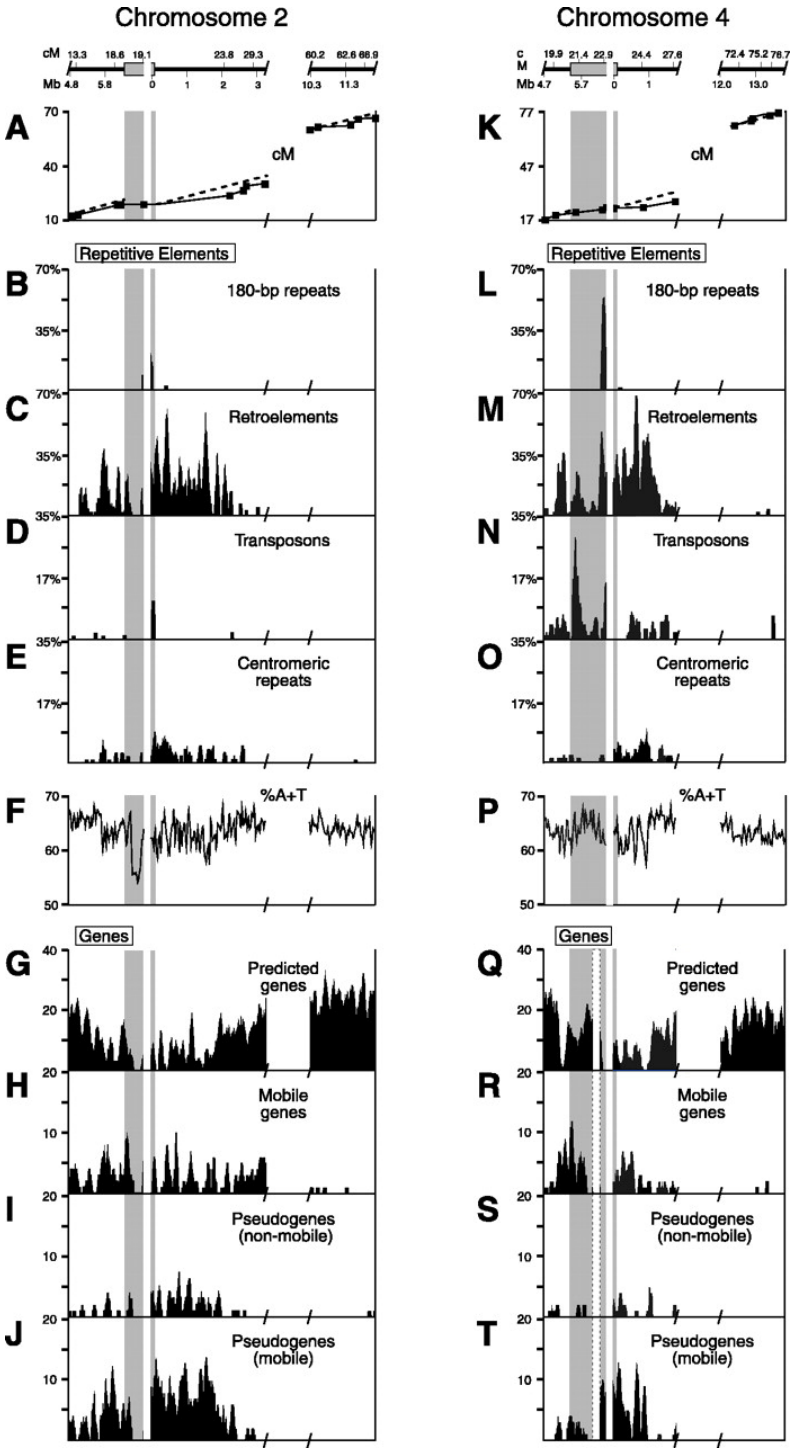
Nagaki et al:
- 1.67 Mb long
- defined by pericentromeric flanking sequences + CenH3
  - CenH3 = ~750 kb, including CentO regions
- mostly retrotransposons
- 14% of MITES, unknown repeats, truncated transposons
- 72% repetitive CentO satellite sequences in three 40 to 80-kb clusters
- 47 putative genes, 14 of which are in the kinetochore binding region

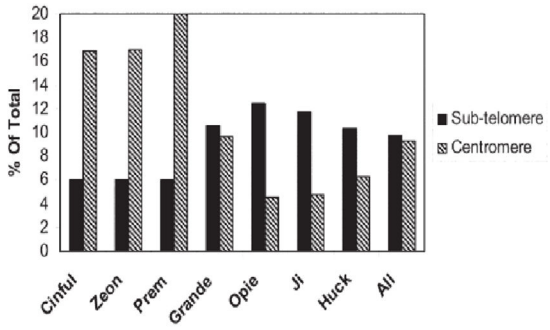
Furthermore, segmental duplications and numerous centromere-proximal inversions distinguish CEN8 of different rice species or subspecies (Bennetzen lab, as cited in Wolfgruber 2016)

***Copenhaver et al.*, 1999**

The arabidopsis centromeres

- Limited homology between centromeres of different chromosomes
- Major component = 180 bp tandem repeat
- CEN sequences- homologous to retrotransposons
- Contain expressed genes





Fengler et al., 2007

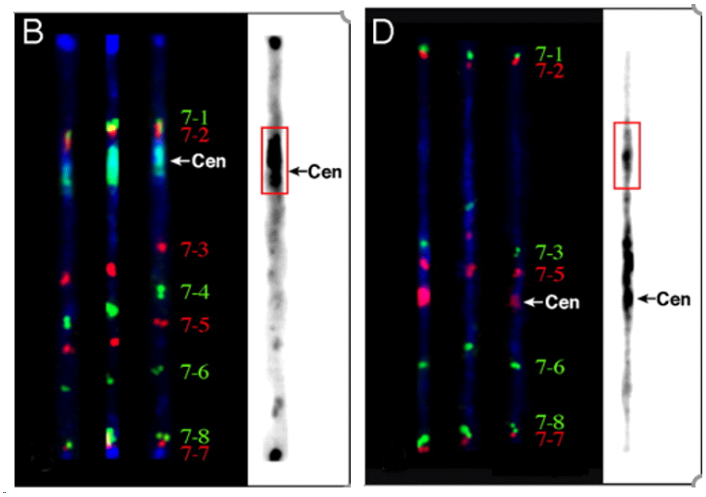
- Note that some retrotransposons have a preference for centromeres; others avoid centromeres.

***Nakano et al., 2003*, *Black et al.*, 2004; *Birchler* et al., 2011**

- Phosphorylation of the centromere-specific CenH3 appears to be a prerequisite for centromeric sequences to gain centromeric function
- Active gene expression appears to be a prerequisite for CenH3 activation
- Centromeres form wherever on the DNA CenH3 gets placed during cell division

Han et al., 2009

- Cucumber and melon: a case where a centromere became inactivated in its original spot, and a new spot became activated
- Pericentric heterochromatin is not being maintained where the centromere lost function



Han et al., 2009. B= cucumber chromosome 7. C, its melon corresponding chromosome, II. The red box in D is where the melon centromere used to be. Note that the flanking markers have not moved.

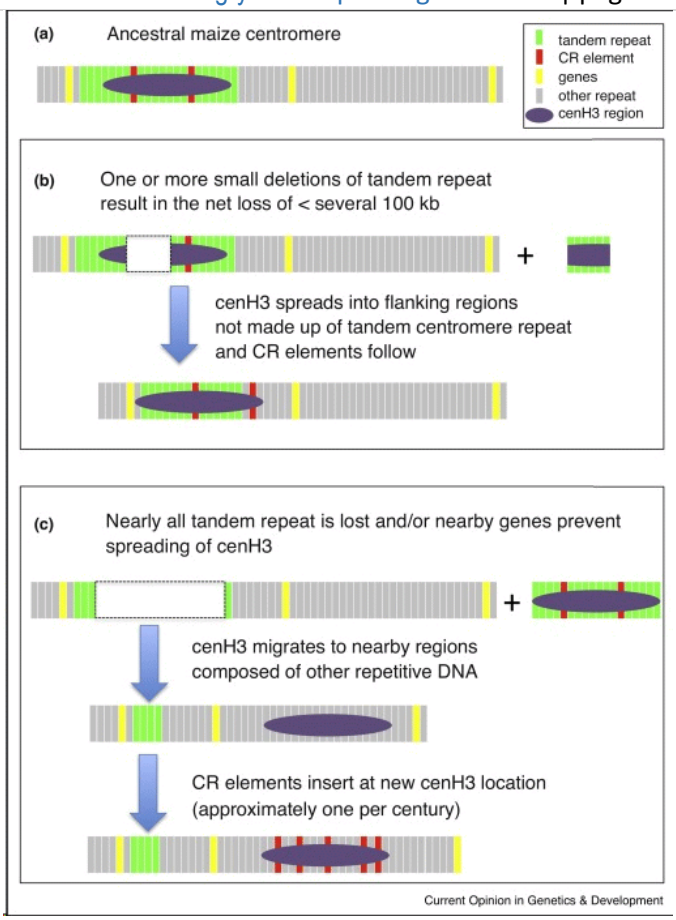
Additional evidence for epigenetic control

- Gao et al., 2011. In corn, a dicentric chromosome created from a translocation inactivates a centromere.

- Nasuda et al., 2005; Zhang et al, 2013. In barley and corn, when centromere is lost, new sequences start acting like a centromere.

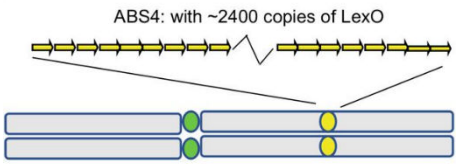
Centromere dynamics

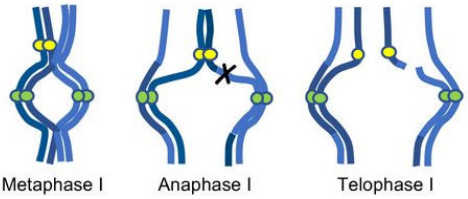
Presting, 2018

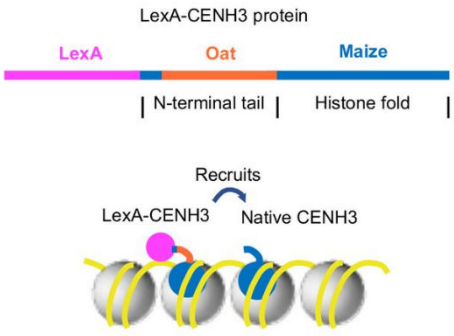


Synthetic centromeres

Dawe et al., 2022

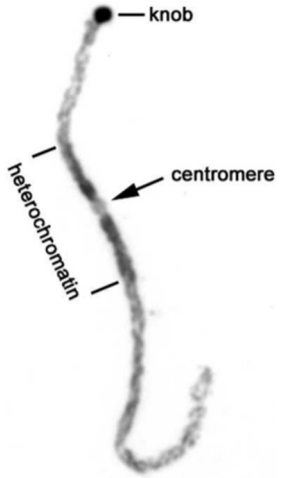






Depends on LexO/LexA binding. Engineer with LexO- makes large array. Then add LexA fused to CenH3. These in turn recruit more CenH3, leading to centromeric function. Dicentric chromosomes break during division.

Pericentromeric heterochromatin



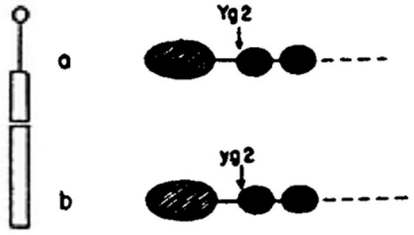
Anderson et al, 2007. Gen Res 16:115-122.

- Centromeres are flanked by large regions of heterochromatin
  - Called <u>pericentromeric heterochromatin</u>
- In plants, makes up 20-50% of chromosome
- Is not recombinogenic
- Contains ~30% of genes
- Breeding implications?

KNOBS

After **Ananieval et al, 1998**

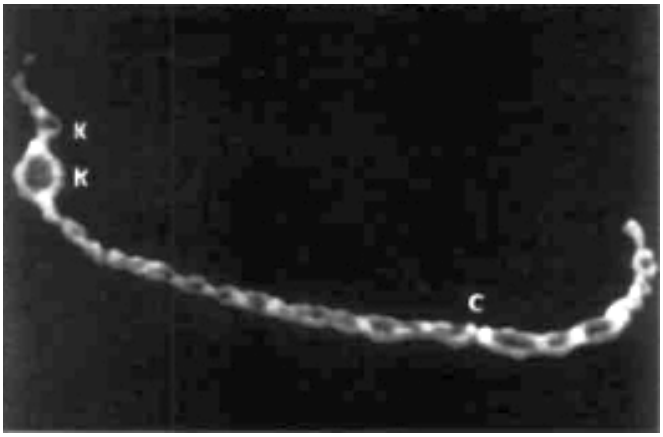
- Appear as short, thickened areas on a chromosome
- Discovered by McClintock, 1929
- In maize, made up of 180-bp tandem repeat
  - Interspersed with transposable elements– up to 1/3 of length
  - 358-bp tandem array (=TR-1 element)
- Different knobs differ in the relative amounts of both repeats



McClintock, 1929- 1st drawing of a knob. 1944- A terminal knob and two chromomeres.

Chromomeres vs Knobs

The term applied by EB Wilson (1896) to each of the serially aligned granules of a chromosome, best seen when the thread is relatively elongated as in early prophase 1 of meiosis (Gk. chroma, colour; meros, part).



Caixeta and de Carvalho. 2000. Digested pachytene chromosomes with trypsin. Chromomeres (bright spots) and knobs (K) on chromosome 8 of maize

Looking at knobs & chromomeres at the molecular level
Ananiev, EV, RL Phillips, and HW Rines, 1998

- Both appear to consist of an 180-bp repeat and the TR-1 element
- *Thus the difference is one of scale*

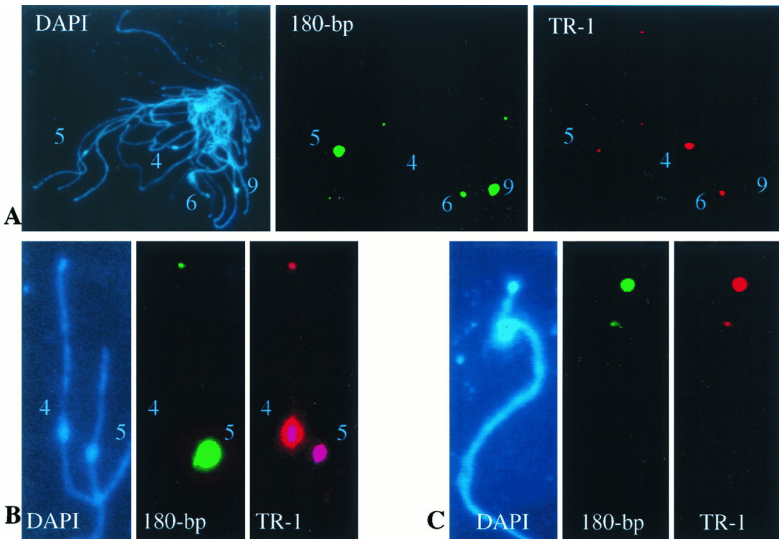


Image: The 180-bp repeat and TR-1 element form two clusters: a big one in the knob at the terminus of the short arm and a small one probably in the first chromomere of the satellite of chromosome 6.

Knobs can be slow-replicating during mitosis and can get left behind during division.

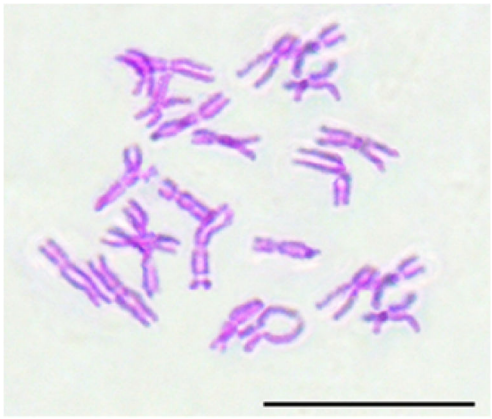
The identification of individual chromosomes

Landmarks are often not enough to distinguish chromosomes from each other

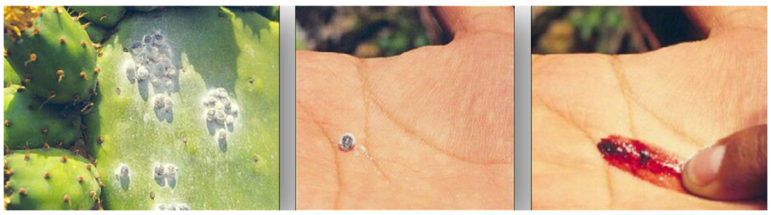
Yang F, L Lingjiao, E Rong, Y-b He, X Zhao & Y Wu. 2015. Karyotype analysis of obtained tetraploid in medicinal plant (Platycodon grandiflorus). Journal of Medicinal Plants Research. 9: 294-300

The acetocarmine stain

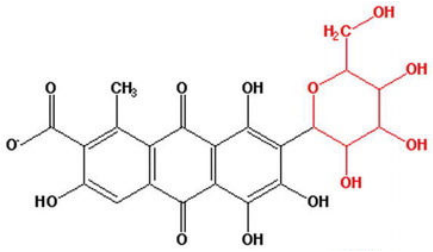
Dapson, 2007



Mirzaghaderi G. 2010. A simple metaphase chromosome preparation from meristematic root tip cells of wheat for karyotyping or in situ hybridization. African J Biotechnol 93: 314-318



Carmine is made by cochineal insects feeding on Opuntia cactus. Originally used to dye clothes. Still used as a natural food coloring.



- Still not certain on chemical structure. It binds with aluminum, and in the presence of iron ions forms coordinate covalent bond with DNA

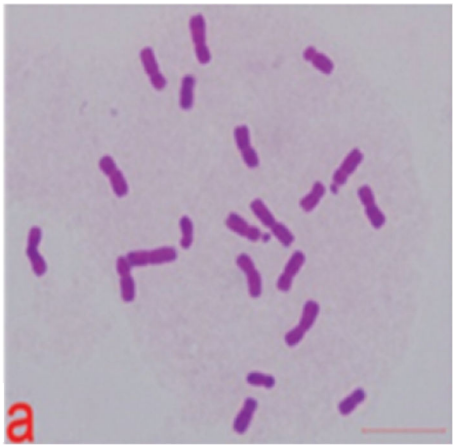
The carbol fuchsin stain

- Phenol + basic fuchsin
- Aniline (coal tar dye)
- Discovered by von Hoffman

August Wilhelm von Hofmann

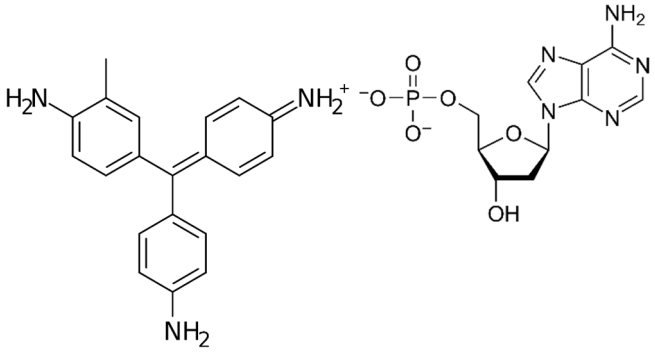
1818 - 1892

Wikipedia commons



Iqbal MZ, Cheng M, Zhao Y, Wen X, Zhang P, Zhang L, Ali A, Rong T, Tang QL (2018) Mysterious meiotic behavior of autopolyploid and allopolyploid maize. Comparative Cytogenetics 12(2): 247-265.

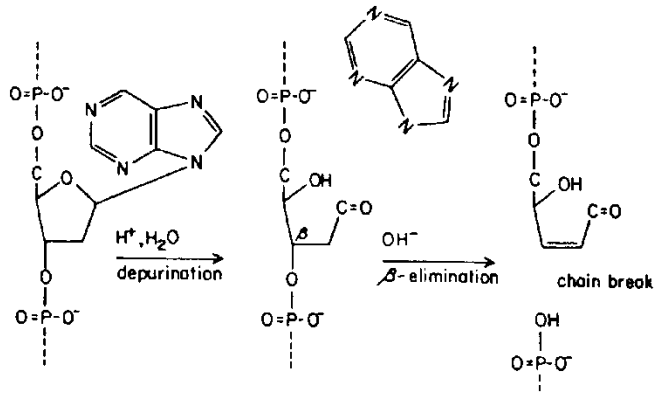
- Has a secondary amine group with a positive charge that binds to the negatively charged phosphate groups on DNA
- In general, DNA has a high affinity for cations - due to PO_4 groups on DNA



DNA pretreatments

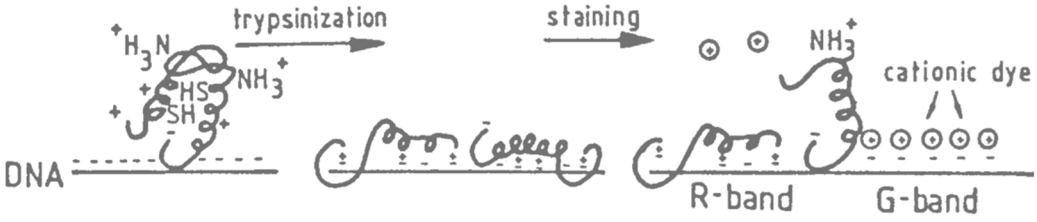
Acid hydrolysis

- Depurinates DNA
- Opens the pentose in the DNA up, leaving an aldehyde that can react with stains



But, these dyes stain the whole chromosome, does not help with the identification of individual chromosomes.

Trypsinization



- Based on the fact that heterochromatin is not as accessible to trypsinization as euchromatin
- Mode of action is not understood well, and above is only one model
- However, some of the phosphates are not accessible to cations because of proteins bound to the DNA

- During the staining process, the protein on the DNA is rearranged, such that additional phosphate groups are no longer available to bind with the stain

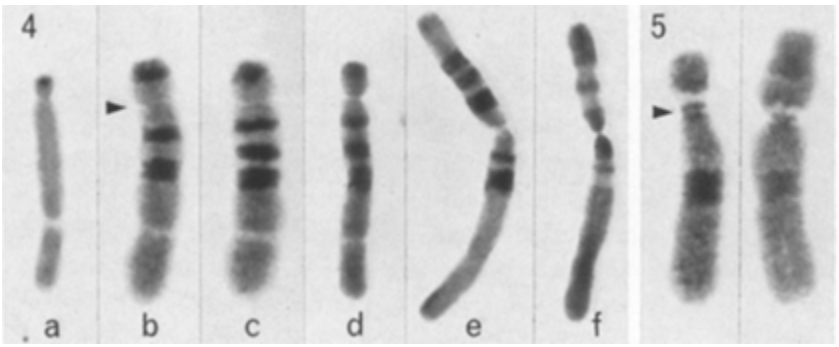
1) Adding trypsin nicks and denatures the protein bound to the DNA
2) The positive charges of the denatured protein are now free to bind with the phosphate groups on the DNA
3) Correspond well to AT rich regions
4) When the stain is added, it can compete with the protein to create G-bands, but R-band proteins are bound too tightly to be displaced by the stain.

- Different stains can target proteins and/or DNA to give different banding patterns, and thus identify individual chromosomes based on their banding pattern
- DNA is broken up into pieces small enough to diffuse out of their proteinaceous entrapments

G bands

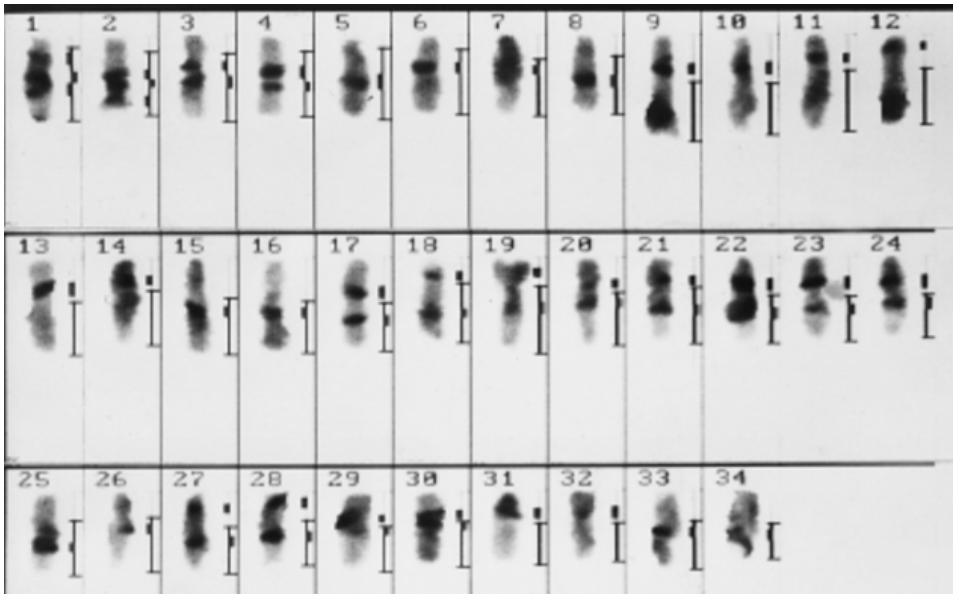
Schweizer, 1973

- Giemsa + alkali: = G bands
- Giemsa at high temperature: DNA rich in CG pairs = R bands (for reverse Giemsa)



G-banded chromosomes of Fritillaria recurva

- Resolution in plants was never good, as compared to animals. EG, G-banding in sunflower

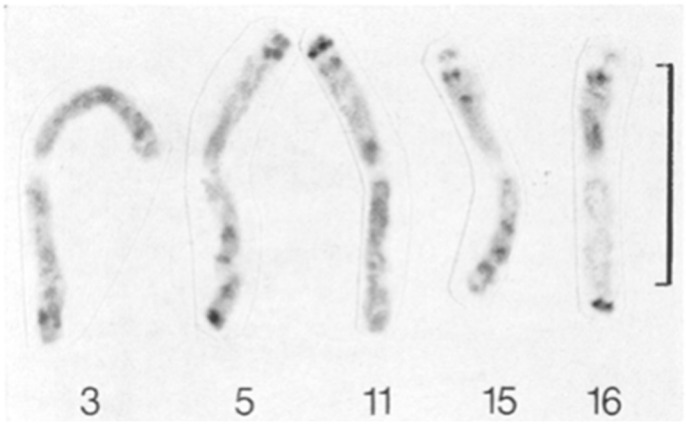


Schrader O, R Ahne, J Fuchs & Schubert. 1997. Karyotype analysis of Helianthus annuus using Giemsa banding and fluorescence in situ hybridization. Chromosome Research 5:451-456

C bands (Hy bands/Feulgen bands)

Developed by Greilhuber, 1973

- Acid hydrolysis for depurination
- ß-elimination of depurinated DNA by hot salt
- Renaturation in SSC or $Ba(OH)_2$ buffer
- Stain with Acetocarmine or Feulgen



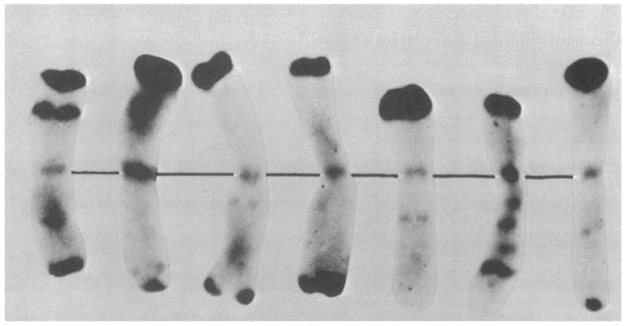
C-banded chromosomes of Allium carinata

C+G bands

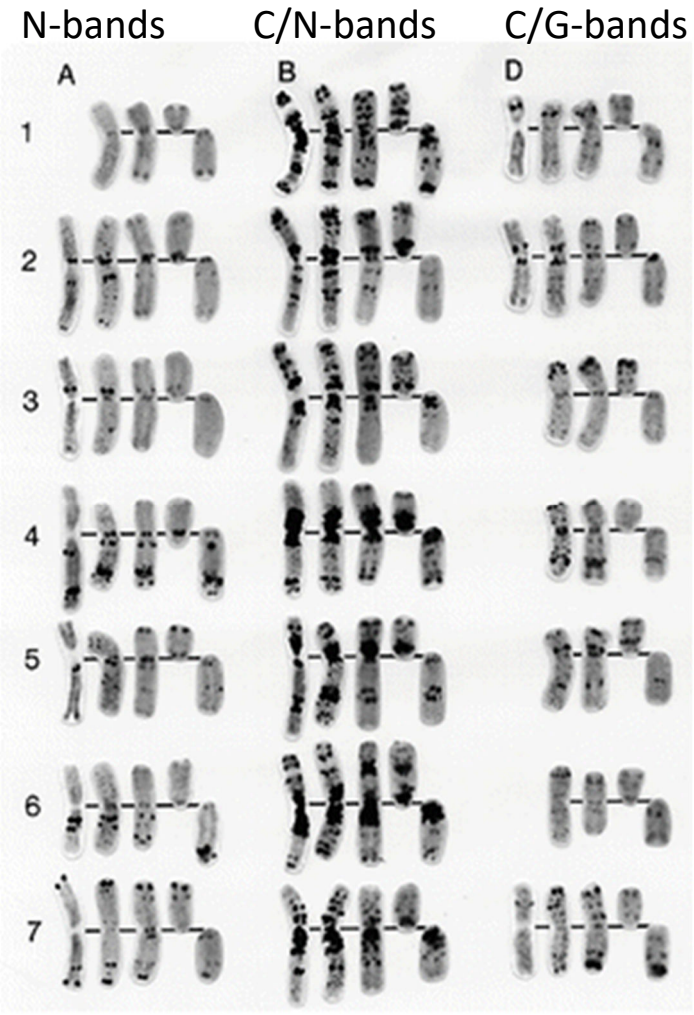
Gill + Kimber, 1973

- Gives improved resolution

C+G banded chromosomes of rye



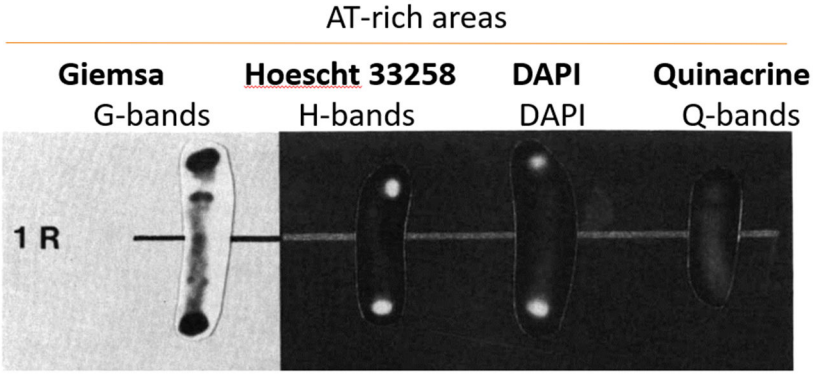
Other stains



Gill BS, Friebe B, Endo TR (1991) Standard karyotype and nomenclature system for description of chromosome bands and structural aberrations in wheat (Triticum aestivum). Genome 34:830–839

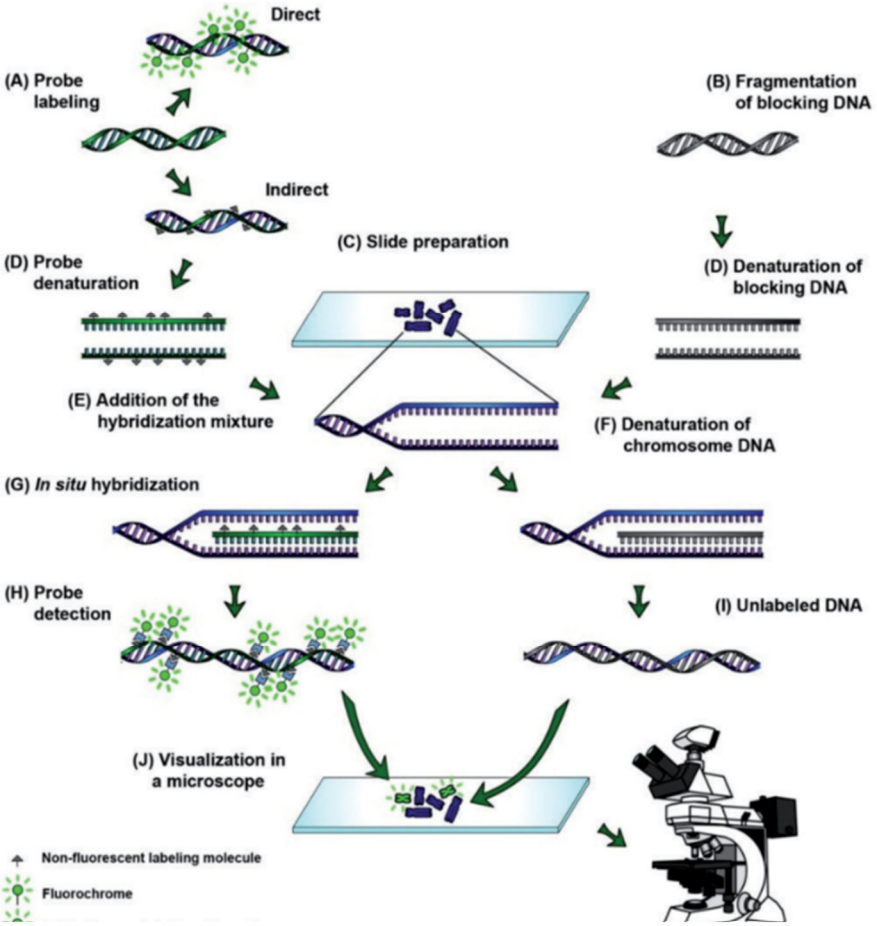
- N = acetocarmine → hot NaH2PO4 → Giemsa stain
- C/N = acetocarmine → hot NaH2PO4 → Barium hydroxide → Giemsa stain
- C/G = acetic acid → dehydration → acid hydrolysis → Giemsa stain

- Giemsa and other stains in general target AT-rich areas



Martin J & CU Heseman. 1988. Cytogenetic investigations in wheat, rye and triticale. I. Evaluation of improved Giemsa C- and fluorochrome banding techniques in rye chromosomes. Heredity. 61: 459-467.

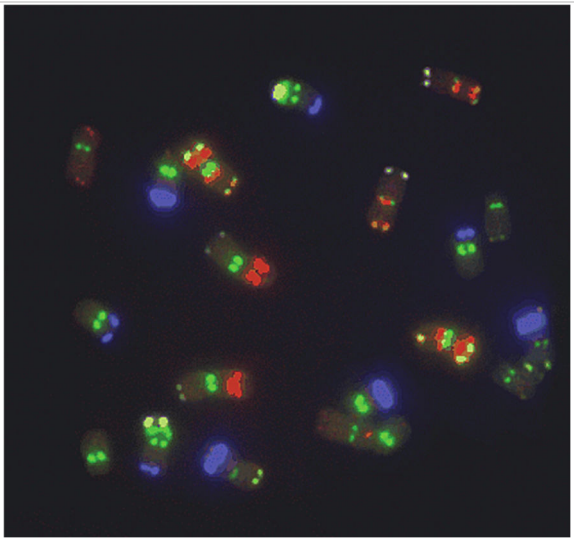
GISH



2Brammer et al., 2013

FISH

None of the banding techniques based on stains give outstanding results, particularly with small chromosomes. Hence the advent of FISH-fluorescent in-situ hybridization. Centromeric, telomeric, satellite, specific repeat sequences, NOR, etc can be linked to fluorescent dyes and hybridized to the chromosome.



Kato et al, 2004

Individual chromosomes in a genome can now be identified by:

1) Relative length of chromosomes
2) Landmarks, e.g., knobs, satellites, heterochromatin and constrictions
3) Position of centromere
4) Bands (G, R, Q, C, N)
5) Chromosome painting
   - FISH & GISH

- Note: some people use arm-length ratios, but this is not universally accepted because this is too susceptible to changes in arm length depending on pretreatments, fixatives, etc.

KARYOTYPES (whole genomes)

- Genome (**Winkler**, 1920): From **Gen**e + Chromos**ome**. Also from Greek 'genomai' for "I become, I am born, to come into being" –> A set of 1 of every type of chromosome

"I propose the expression Genom for the haploid chromosome set, which, together with the pertinent protoplasm, specifies the material foundations of the species ..."

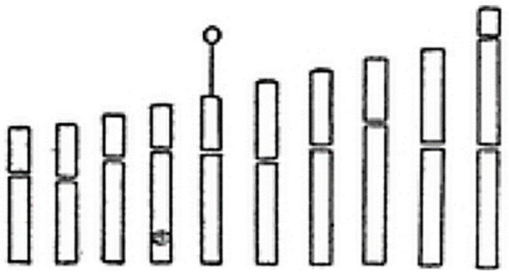
Hans Karl Albert Winkler

1877 - 1945

www.ranker.com/list/famous-botanists-from-germany/reference

McClintock, 1929 - Drew the first karyotype, using maize, to demonstrate the premise that each chromosome was identifiable based on its morphology

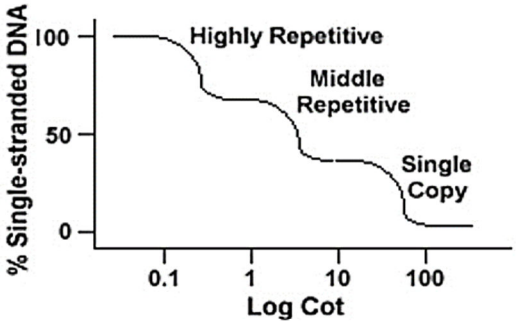
- "the author is convinced that every chromosome of the set is morphologically identifiable"



McClintock, B. 1929. Chromosome morphology in Zea mays. Science, 69, 629

Whole genomes can be identified by:

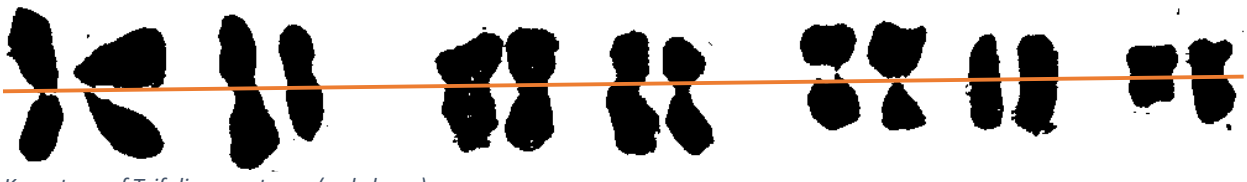
1) Chromosome number
2) Kind of centric activity (diffuse, localized, or polycentric)
3) Size of localized centromeres & number of microtubule attachment sites
4) Arm ratios (caution!)
5) Number, size, & position of 2º constrictions and satellites
6) Absolute size of chromosomes = DNA content
7) Size difference within a complement
8) Position, number, size, and distribution of heterochromatin
9) Banding patterns
10) Annealing rates
11) Total amount of DNA
12) Molecular markers
13) Number of sub genomes
14) Sex chromosomes



www.slideshare.net/StevensonThabah/repetitive-sequences-in-the-eukaryotic-genome

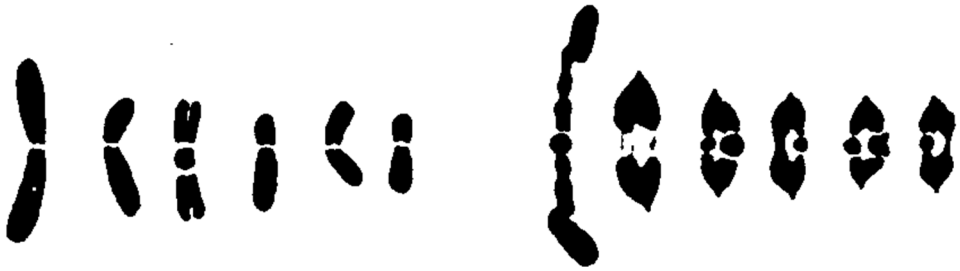
Karyotypes have been used for taxonomic purposes. Caution is warranted as "Changes in chromosome morphology may lag behind or precede changes in external phenotype." (Jackson)

Karyotype: An actual photograph of the chromosomes, with the homologues paired, and arranged by size with the centromeres lined up. Homologues are chromosomes with the same sequence of genes on them.



Karyotype of Trifolium pratense (red clover)

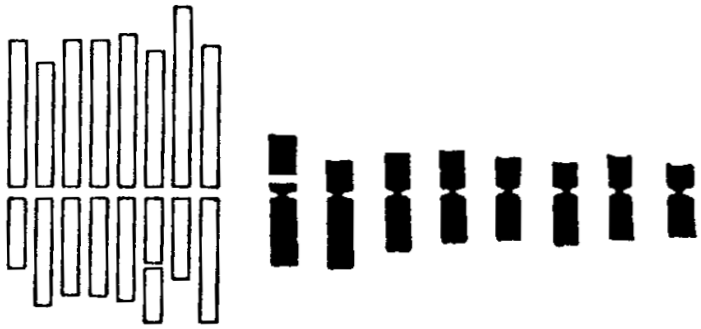
Idiogram: Drawings of chromosomes as they are seen through a microscope. They are rare today, having been largely supplanted by karyotypes and, especially, idiotypes.



Idiograms of root tip (left) and meiotic (right) chromosomes of Trifolium subeterraneum (sub clover).

- A diagram can be more informative than a photo or portrait:

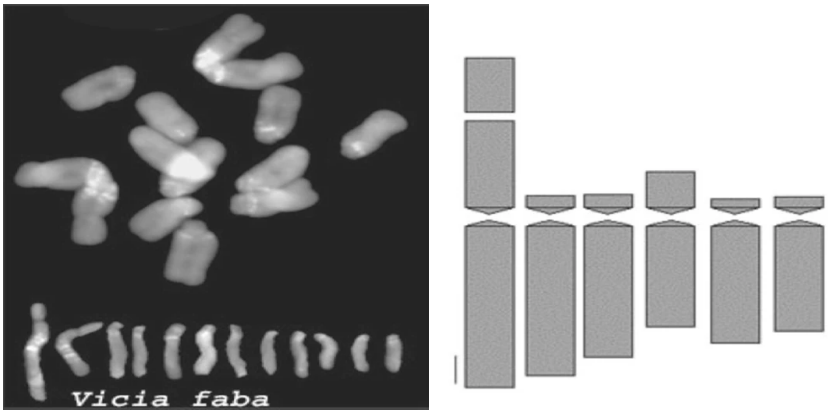
Idiotypes: Schematic representations of chromosome sets. NOR depicted as a gap.



Idiotypes of Trifolium subterraneum (left) and T. masaiense (right)

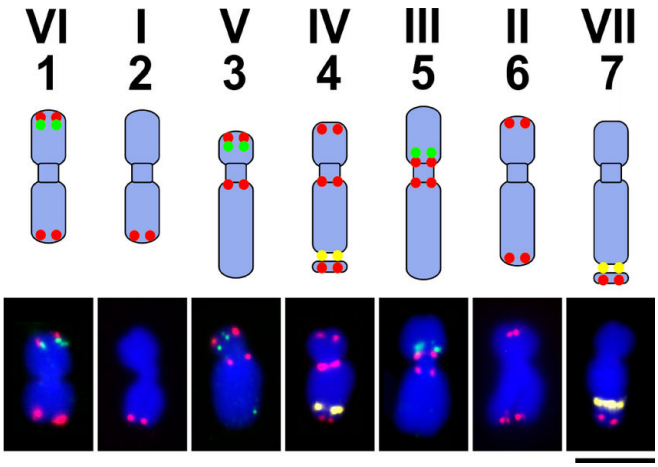
- A diagram can be more informative than a photo or portrait, particularly if landmarks are added

Hybrid presentations



Faba bean karyotype & idiotype. Osman, S.A., Ali, H.B., El-Ashry, Z.M. et al. 2020. Karyotype variation and biochemical analysis of five Vicia species. Bull Natl Res Cent 44, 91

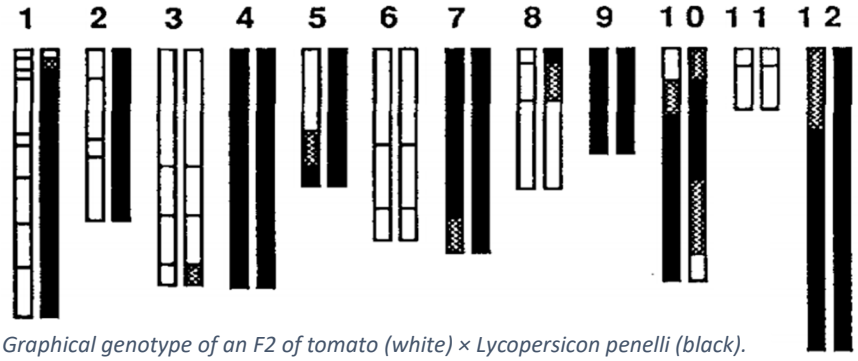
Pea idiotype and karytoype.
PisTR-B (red), 5S rDNA (green), and 45S rDNA (yellow)



Smýkal P; Aubert G; Burstin J; Coyne CJ; Ellis NTH; Flavell AJ; Ford R; Hýbl M; Macas J; Neumann P; McPhee KE; Redden RJ; Rubiales D; Weller JL; Warkentin TD. 2012. Pea (Pisum sativum L.) in the Genomic Era. Agronomy 2: 74-115.

Graphical genotypes

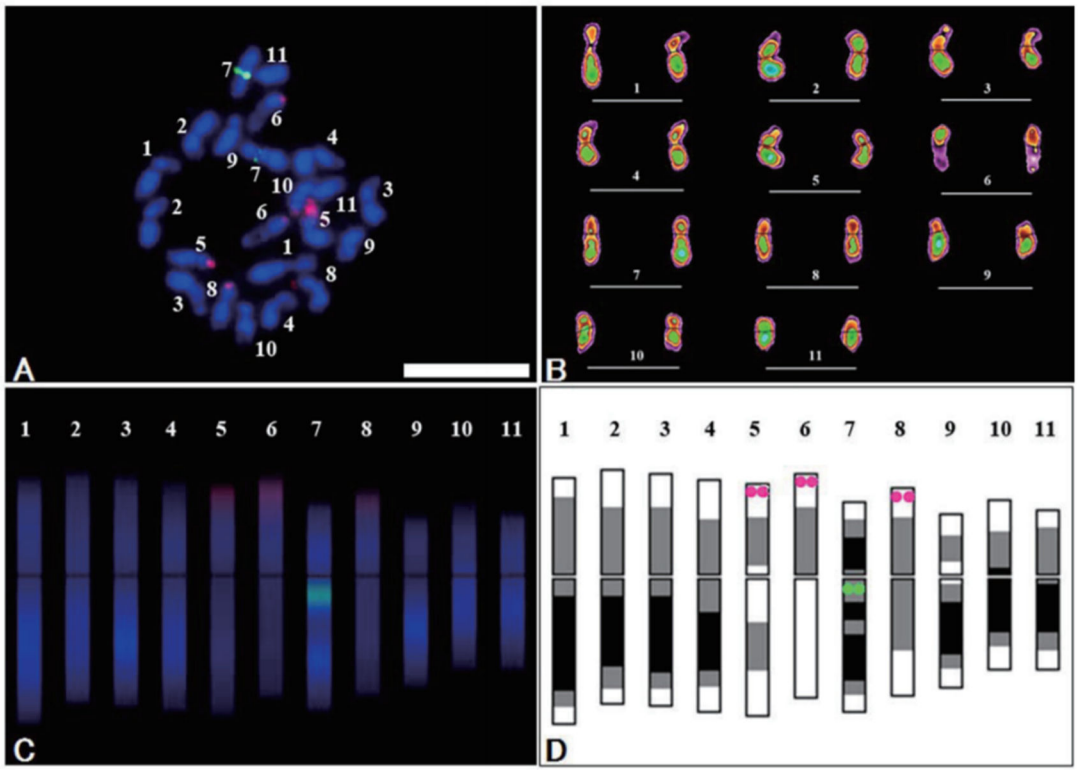
Young and Tanksley, 1989



Graphical genotype of an F2 of tomato (white) × Lycopersicon penelli (black).

CHIAS – Chromosomal Image Analysis System

Based on Fukui & Iijima, 1991

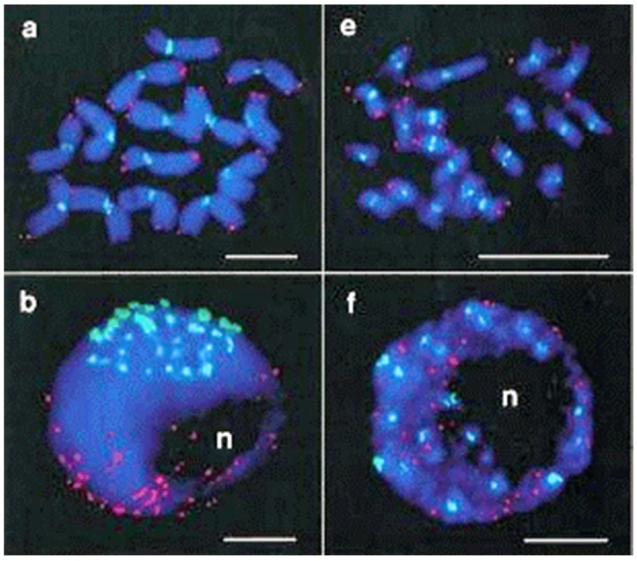


Ohimodo et al., 2013. Chromosome Science 16: 17-21, 2013

Nuclear architecture

Rabl 1885

- Noticed that during zygotene, the centromeres all cluster on one end of the nucleus, and the telomeres cluster on the opposite side = "**Bouquet stage**"
    - This arrangement apparently maintained through interphase
- the area with the centromeres is known as the **polfeld**
- leads to unequal DNA distribution within the nucleus
- In mitosis, telomeres remain clustered after cell division = **Rabl configuration**



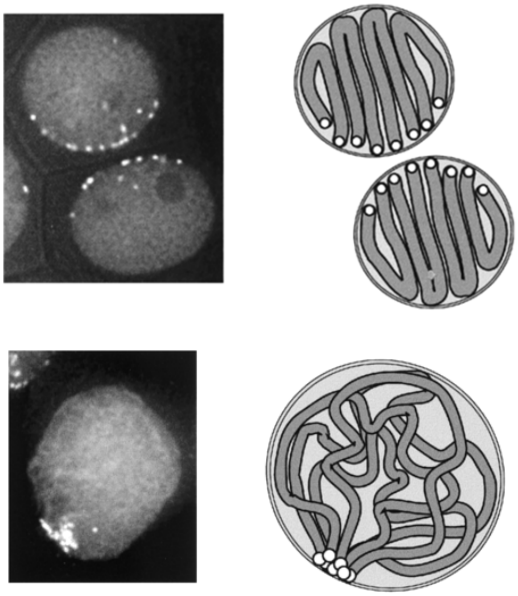
Dong & Jiang, 1998. A&b = rye; e & f = sorghum. A& e = metaphase; b& f = interphase

Dong & Jiang, 1998

- Found in rye, wheat, barley, oat
- Not found in maize, rice or sorghum

Cowan et al., 2000

- The Rabl configuration (right) represents the telomere and centromere positions from anaphase
- The bouquet is the clustering of telomeres during meiotic prophase– related to pairing?



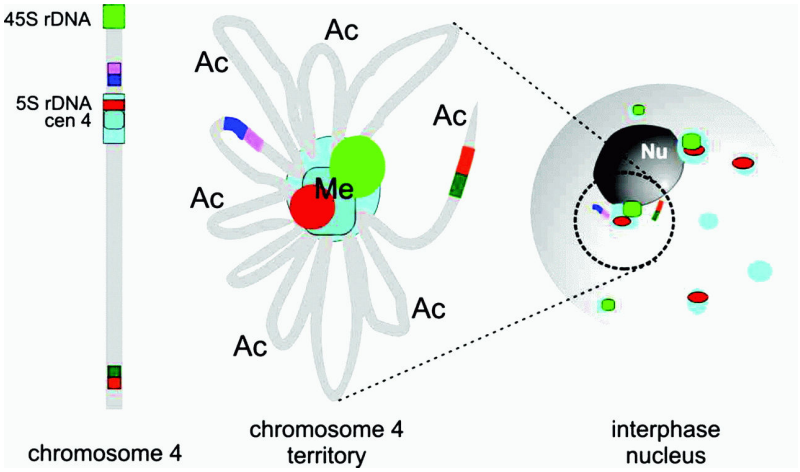
Chromosomal arrangements

Long-time question: - Are chromosomes arranged in any particular order relative to each other, ie, does each chromosome occupy a specified "chromosome territory"?

Fransz et al., 2002

Arrangement of arabidopsis chromosome 4 in the nucleus.

- Histone 4 is acetylated in euchromatin
- Not localized in the nucleus → random location in nucleus



Berr & Schubert, 2007

Used FISH and 3-D imaging to study chromosome arrangement in arabidopsis

- Association of non-NOR chromosomes is random
- I.e., homologs occupy different domains in the nucleus
- After mitosis, sister chromatids in daughter cells are symmetrically arranged for a brief period