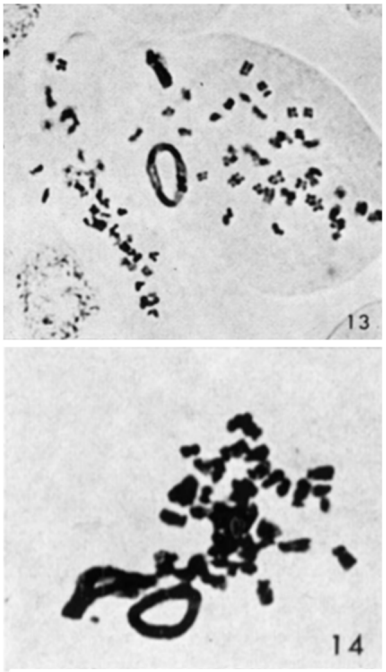
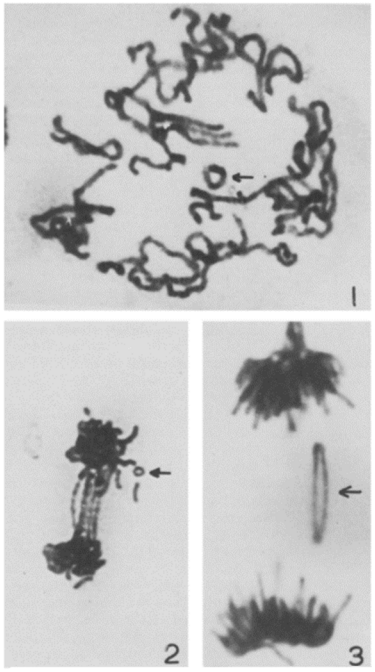
Ring chromosomes

Review by **Yu, 2018**

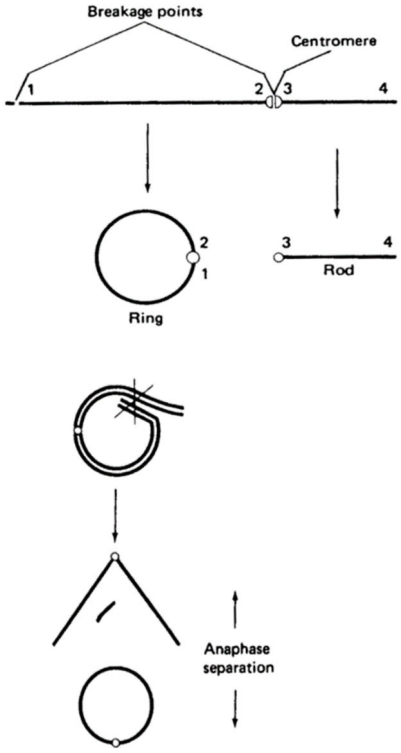
- Arise through double breakage, one near the end, the other at the centromere
- Can also be formed by a crossover within a duplication

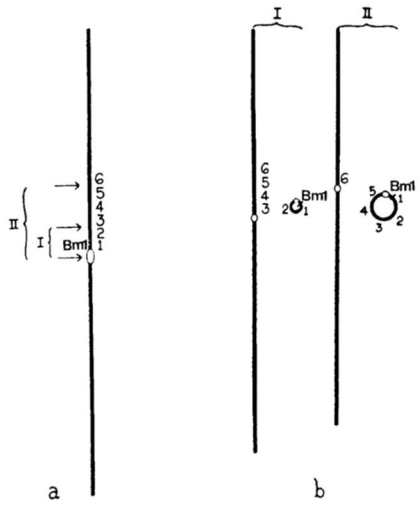


Ring chromosomes in tobacco Gerstel & Burns, 1967



Ring chromosomes in onion, Gohil & Kaul, 1983. Experentia 39: 1152-3.





McClintock, 1938

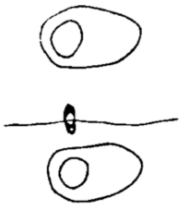
Devised a system of deficient rods with complementary rings on chromosome 5 of maize

- Loss of a ring would give plants variegated for normal and brown midrib (whenever rings get lost)
- In a plant of genotype *bm1 bm1*, loss of a ring with *Bm1* results in brown tissue
- In a plant hemizygous for *bm1* (i.e., one homologue is deficient for the *bm1* allele), loss of a ring with *Bm1* results in dead tissue
- In a homozygous state, the deficiency is lethal.
- The rings were not transmitted through the gametophyte

Small rings behave differently than large rings:

- Lost more frequently (right: lag behind, and don't get included in a nucleus)
- Change in size less frequently
- Reduced frequency of double-sized rings
- Increase in number - 2 rings go to the same nucleus

Ring size	Freq. double-sized rings
Same as rod	15-20%
$^1/_{10}$ as big	1%
$^1/_{25}$ as big	0.2%

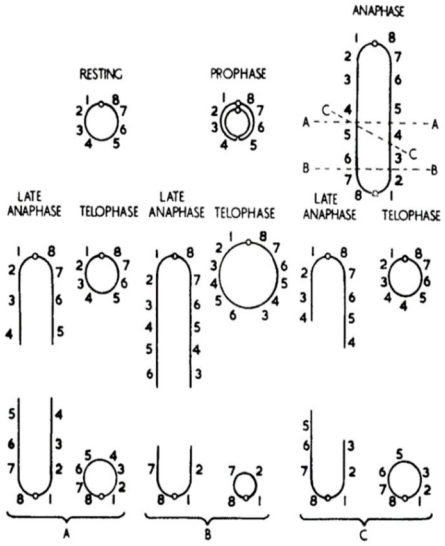


McClintock, 1941

Ring chromosomes become unstable if sister-strand crossing over occurs

- increase in size through duplications
- decrease in size through deletions
- can be lost
- can increase in number

- A = breakage into equal halves followed by fusion
- B = breakage into unequal parts followed by fusion (altered size)
- C = breakage into unequal halves followed by fusion (similar size, but altered genetic content)



Breakage-Fusion-Bridge cycles

McClintock, 1941; review by Jones, 2005

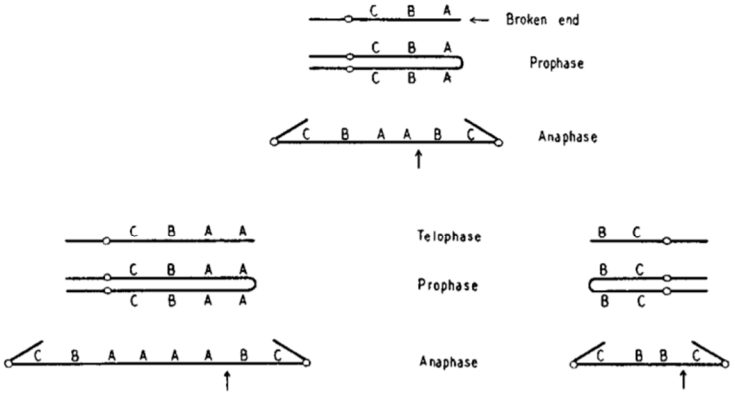
2 main types:

- chromosome (breakage of ring chromosomes in somatic tissues)
- chromatid

Chromosome breakage fusion bridge cycles:

When a chromosome with its end broken off replicates, its ends may fuse.

- This leads to the formation of a bridge during anaphase.
- The bridge breaks, and the cycle gets repeated
- When the break takes place, the resulting chromosomes will either have duplicate or deficient material.



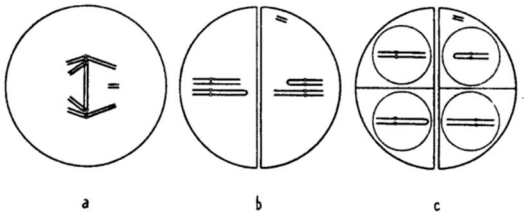
from McClintock 1941

BFB in meiosis

McClintock, 1938

To study BFB cycles in meiosis, need 2 broken ends in a cell

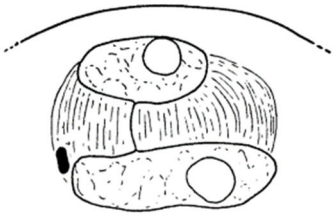
- Created these by a 4-strand double crossover within a large paracentric inversion in chromosome 4 of maize
- Gives a bridge and fragment after the first division, and 2 bridges during the second division



Breakage of the bridge takes place during anaphase or telophase II.

- About 25% of the microspores receive the broken chromosome.
- About 7.5% receive both the broken chromosome and the resulting fragment, thus resulting in a full genomic complement

- Bridges form during the mitotic division of microgametogenesis
  - This is chromatid breakage fusion bridge cycle



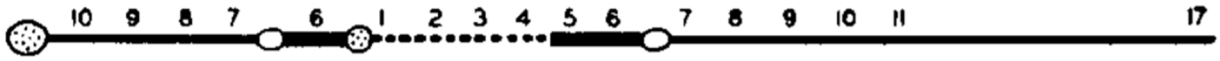
- At right is the first mitotic division of the microspore.
  - Note the bridge remaining between the daughter nuclei.
  - One of the fragment halves remains in the spindle, the other was included in the larger nucleus.

BFB in the endosperm

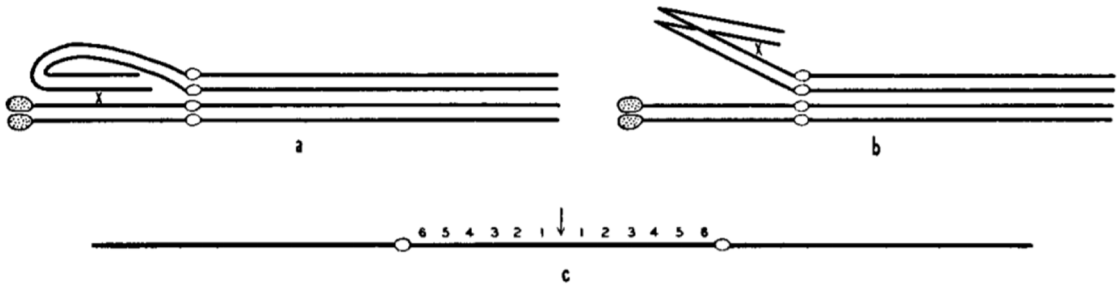
McClintock, 1941

The difficulty in transmitting this to the next generation is that the inversion causes duplicate-deficient gametes.

- Consequently, McClintock devised a system that, even if a break occurred, the chromosome would not be deficient
- Used a chromosome 9 (with dominant genetic markers) that had an inverse duplication of in the short arm:



The duplicated fragment can either cross over with its homologue (a) or with itself (b), producing a dicentric chromosome with an inverted duplication in the middle (c):



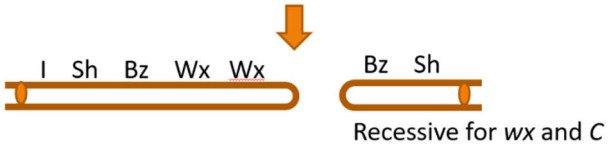
(a) leads to the formation of a bridge and fragment at anaphase I
(b) leads to a bridge at anaphase II

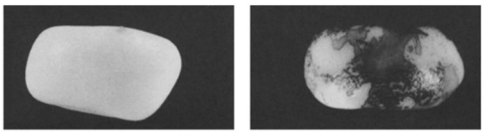
- A break can still occur as indicated by the arrow, and still have intact chromosomes

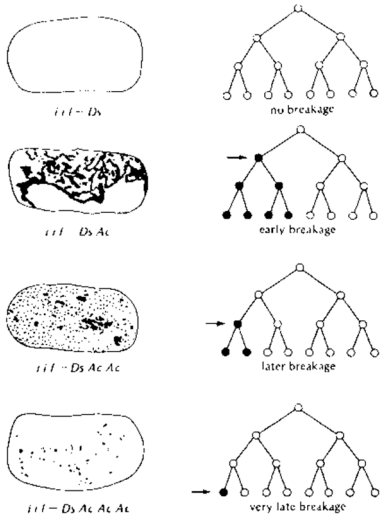
McClintock, 1951

The markers make this easy to follow:

- The BFB cycle continued in the endosperm, but not in the embryo
- Leads to formation of variegated endosperm
    - Get all types of variegation, depending on when and where breaks occur:





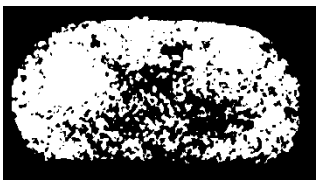


from Strickberger, 1972, after McClintock 1951

If the cell is heterozygous for appropriate markers, the loss of genetic material results in a readily identifiable cell lineage

- This phenomenon does not occur in the embryos, as the broken ends heal rather than fuse.
- The original breakage is due to excision of the *Ds* transposable element. Increasing copy number of *Ac* decreases excision and breakage, leading to smaller spots (right) [*did not know @ time*].

- The maize kernel below was heterozygous for the *I* gene, the dominant allele of which inhibits color formation. Loss of the dominant allele leads to colored sectors (from McClintock 1951)



Rings and herbicide resistance

Palmer amaranth, the king of weeds, cripples new herbicides

Scientists in the US sound the alarm about a crop-smothering weed that is growing resistant to multiple herbicides

by Melody M. Bomgardner

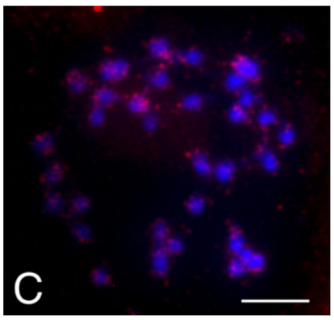
August 3, 2019 | A version of this story appeared in Volume 97, Issue 31

https://cen.acs.org/business/specialty-chemicals/Palmer-amaranth-king-weeds-cripples/97/i31

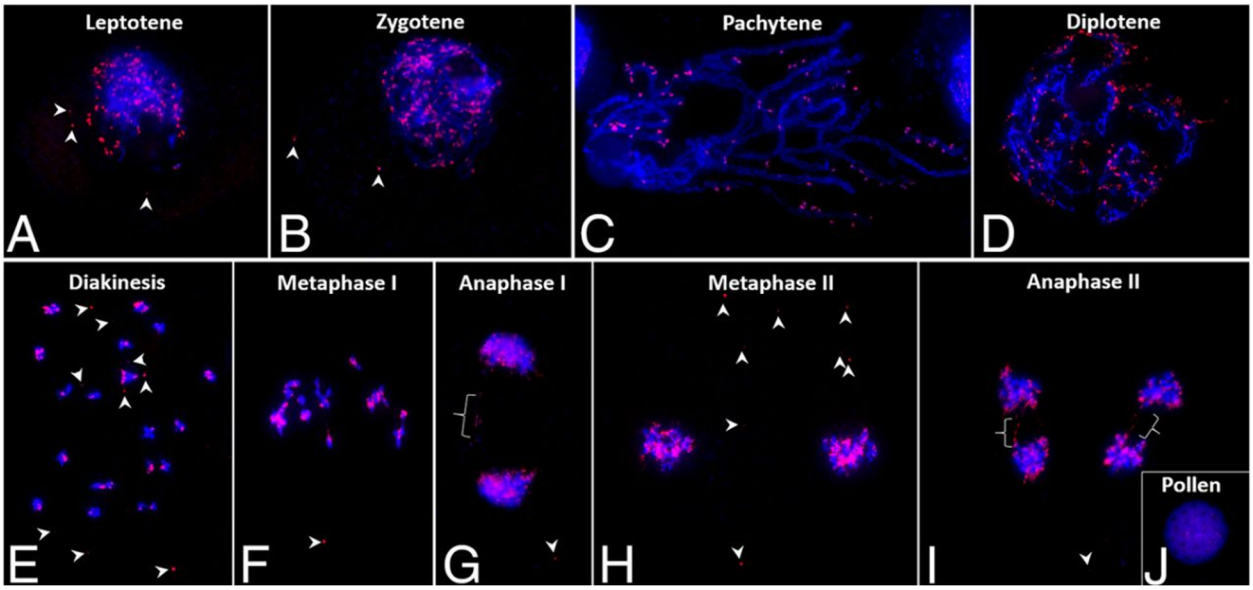
https://cropwatch.unl.edu/2017/status-herbicide-resistant-weeds-nebraska/

Koo et al, 2018a; Molin et al, 2020

- Resistant plants have 40-100 copies of EPSPS gene dispersed on all the chromosomes
- Increase genome size by 11%
- ~400-kb extrachromosomal circular DNA (eccDNA) that harbors the EPSPS gene and 58 other genes
- Transmitted by chromosome tethering during mitosis and meiosis



Gaines et al, 2009



Tethered and unthethered eccDNAs during meiosis. From Koo et al, 2018

Rings & BFB in herbicide tolerance

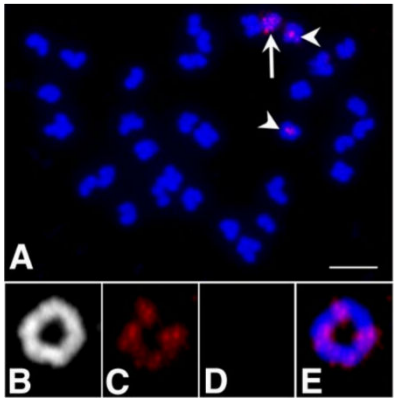
Koo et al., 2018b

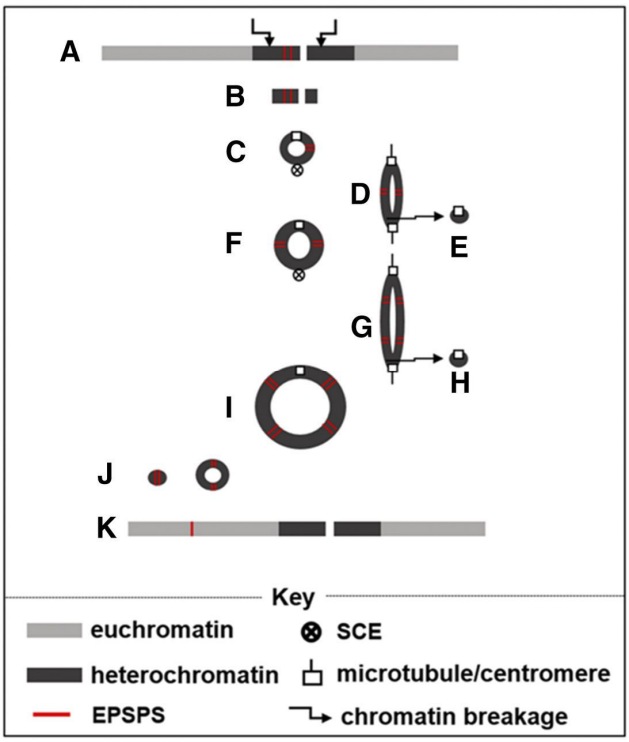
Water hemp, *Amaranthus tuberculatus*, is one of the weeds that has become resistant to glyphosate

Photo shows a glyphosate resistant *A. tuberculatus* plant showed chromosome constitution of 2n = 32+ 1 circle (arrow)

FISH for EPSPS shows presence on two somatic chromosomes (arrow heads) and a cluster on the ring chromosome.

https://www.youtube.com/watch?v=DgpaxUkBeZA





Proposed model:

- Amplification of EPSPS-containing pericentromeric heterochromatin onto another chromosome
- Breakage leading to ring chromosome
- SSCOs lead to dicentric rings
- These break into different sized rings in continuing rounds of BFB cycles
- Some rings can reinsert into genome, forming new EPSPS loci