BIS101 F2014: Chromosomal Evolution

Go back over P-elements in Drosophila and C-Ds in maize.

Changes in chromosome number:

Euploid -- multiples of a full set of chromosomes

- haploid -- number of chromosomes in a gamete ?
- diploid ?
- monoploid (males in bees, wasps, ants.) why might be selected against? ? deleterious recessives
- tetraploid ?

Polyploidy

autopolyploid - from single parent, doubles. can be made with colchicine injection which arrests mitosis. how do chromosomes pair? sometimes as sets of **bivalents** or often as **quadrivalents**

allopolyploid - hybrid of two different parents then doubles. **homeologous** chromosomes

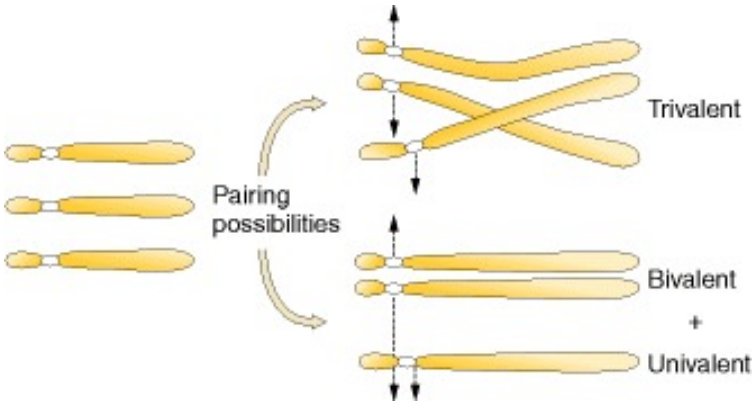
DRAW with color auto vs. allo

end result of most polyploids is fractionation -- reduction down to previous numbers. corn is an ancient tetraploid 10 -> 20 -> 10 but still has 2 copies of copies of many genes in its haploid genome.

What about odd multiples of ploidy? triploidy? pentaploidy?

with odd multiples problem is ?

draw triploid. explain **bivalents** and segregation of **univalents** in meiosis resulting in **aneuploid** gametes (can be one tri (bi + uni) or normal bi + uni



Aneuploidy

Incomplete sets, too many or few of one or more chromosomes

- monosomic, disomic, trisomic etc.
- e.g. ? Down's syndrome or trisomy 21.
- the process that creates this is **nondisjunction** usually at meiosis
- turns out an important function of crossing over is to hold chromosomes together.
- mutants that are achiasmatic or have other strange biases in meiosis sometimes also have increase rates of nondisjunction
- aneuploidy causes

In human females ovules are arrested in prophase of meiosis at birth, and only finish meiosis during ovulation

as women age, their meiotic products have increasing risk of nondisjunction, thought to be because of failing or loss of cohesion of chiasmata

increase risk of chromosomal aberrations and aneuploidy like downs syndrome of older women.

X Females -- Turner syndrome nondisjunction in father

Kleinfelter's syndrome: XXY (low testosterone, sterile)

Gene balance

relative expression levels of genes matters.

think of the whole elaborate systems of expression control we've described.
now throw the whole thing out of whack doubling some genes and not others!

- imagine gene that controls edge of leaf and one that controls center of leaf. could end up with some strange looking leaves if genes are out of balance
- double every gene in the genome, what's the effect on the relative ratio? (none)
- double copies of a gene, what effect on transcription? (double)

- examples of Datura in book -- extra chromosome copy disrupts gene balance alters morphology of fruitshape
- but most organisms not so tolerant

however, can you think of a "normal" case of aneuploidy ❓

sex chromosomes! In drosophila fixed by 2x expressing everything on X in males because of TE!.

In humans one X is deactivated in each cell randomly so expresses same amount. this is called **dosage compensation**

calico cats

O orange o black, in heterozygous Oo cat, which X is silenced leads to coloration differences. additional genes determine white spots.

if i clone a calico will it have same coat color❓ No.

Structural rearrangements

Figure 17-19 in book does a great job showing these.

indels

deletion and insertion we've discussed to some degree — but can happen to large regions

- why would a very large (100's of kb) deletion very likely be deleterious? not having lots of genes is really bad in homozygote,
- why bad in het ❓ recessive deleterious expressed. (**hemizygous**)

rearrangements

DSBs common. if multiple breaks and wrong ends get stuck together -> rearrangement

Also faulty recombination. Redraw genes and TEs on 1 chromosomes, but could happen even between chromosomes in rare occasions.

inversions

17-26, 17-28, 17-29 great figures showing effects of inversions and how meiosis works in inversion heterozygote.

simply a piece of DNA that flips. has any effect on gene number ❓ NO

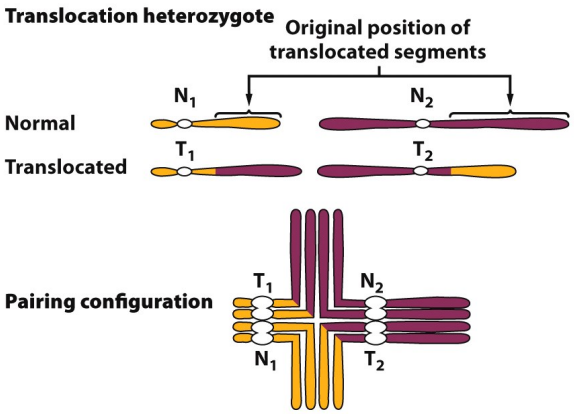
may have phenotypic effects because it rearranges or breaks genes (could move a gene into new chromatin, near enhancer, etc.)

prevents recombination because of loop structures that form in meiosis for pairing to occur. recombination results end with deletions (see above figures)

are being shown to be important in adaptation! (mimulus example)

translocation

two chromosomes switch arms (**reciprocal**). leads to some very strange pairing in meiosis



Two types of segregations:

Adjacent-1		Final meiotic products	
Up	$T_1 + N_2$	Duplication of purple, deletion of orange translocated segment	Often inviable
Down	$N_1 + T_2$	Duplication of orange, deletion of purple translocated segment	
Alternate			
Up	$T_1 + T_2$	Translocation genotype	Both complete and viable
Down	$N_1 + N_2$	Normal	

Figure 17-30
Introduction to Genetic Analysis, Tenth Edition

depending on how meiosis finishes, can produce inviable or viable gametes. diagonal good, off-diag bad!

nonreciprocal translocation (one of the translocation copies or one arm piece lost). leads to gene imbalance. once version of this is Robertsonian translocation. involves **acrocentric** chromosomes (compare to **metacentric**) moving long arms together, losing small arms often not necessary common and thought to be important for speciation in e.g. mice