LECTURE 10: CHANGES IN CHROMOSOME NUMBER

Reading: Ch 14, p 516-525

Problems: Ch 14 #29 – 31, 33, 38, 39

Announcements:

*I added office hours this week – Thursday, 9/25 from 12:30-2. Next week, office hours are Thursday 10/2 1:30-3:30 and Friday 10/3 from 10-noon.

*Beginning today (9/24), Section 114 (Wed 2-3 pm) moves from 2032 VLSB to 2038 VLSB.

*Those of you taking courses with midterms scheduled at the same time as our midterm(s) should see the website announcement page (on or after Monday, September 29th) for instructions.

*Our midterms will be given in multiple rooms. Please see the website (on or after Monday, September 29th) for your room assignment for the October 6th midterm.

*On Friday, we will review the major topics that we have covered so far in class. Beginning Monday, Professor Dernburg will begin lectures. I will be back at the end of October to give four lectures.

Last time, we talked about how aneuploids (**aneuploid** = organism in which a particular chromosome or chromosomal segment is over- or under-represented) can be generated by chromosomal rearrangements. Today, we'll talk about aneuploidy at the level of whole chromosomes and we'll talk about species whose genomes contain complete (but nondiploid) sets of chromosomes. However, first, we will review Robertsonian translocations by discussing a heritable form of Down's syndrome (see lecture notes from last time).

DEFINITIONS:

Ploidy: number of basic chromosome sets (a diploid has 2 sets; a hexaploid has 6 sets)
Euploid: organism containing multiples of the basic chromosome set
Monoploid: organisms with one chromosome set (in essentially diploid taxa)
Polyploid: organism containing more than two chromosome sets
Basic chromosome number, x (also called monoploid number): the number of different chromosomes that make up a single complete set. (In a diploid organism with 10 pairs of chromosomes, x = 10)
Haploid number, n: number of chromosomes in the gametes
(In diploid organisms n=x, but this is not true for polyploid species. Wheat is a hexaploid with 42 chromosomes; in this case x=7 and n=21.)
Allopolyploids: polyploids created by hybridization between different species (homeologous chromosomes)
Autopolyploids: polyploids created by chromosome duplication within a species

Most euploid species are diploids. There are a few examples of monoploidy. For example, in some species of bees, wasps, and ants, the females are diploid and males are monoploid. The males develop parthenogenetically from unfertilized eggs, thus receive a haploid set of chromosomes from their mothers. Males can produce gametes using a modified mitosis. Monoploids can also be made artificially in some species. For example, geneticists generate haploid embryos because genotype in haploids is revealed by phenotype. Haploid plants can also be used to select for recessive mutations that confer resistance to chemicals or pathogens. Haploid pollen grains are treated and plated onto agar plates. The haploid embryoids that result are treated with hormones to allow regeneration of a monoploid plant. Somatic cells from a monoploid plant are treated with mutagen and then plated onto agar containing the selective agent. The only cells that can generate embryoids and thus resistant monoploid plants are those resistant to the selective agent. The others die. Somatic cells from resistant monoploid plants are then treated with colchicine to block formation of the mitotic spindle and block cytokinesis, thereby allowing the cells to become diploid. Upon proper hormone treatment, the cells can develop into a diploid homozygous resistant and fertile plant.

There are a few examples of polyploidy in animals. It is rare probably because it interferes with sex-determination. Usually, polyploid animals have unusual reproductive cycles. Flatworms, leeches, brine shimps, some salamander and lizards, and goldfish are parthenogenetic. Triploid oysters have no sexual cycle (hence, no mating season when oysters are usually unpalatable!) Some polyploid amphibians (some frogs and toads) do use sexual reproduction.

Polyploidy is much more common in plants! Roughly one of three known species of flowering plants are polyploids. One-half of all known plant genera contain polyploid species and 2/3 of all grasses are polyploid. Examples are wheat (6x), alfalfa (4x), coffee (4x), peanuts (4x), strawberries (8x), and cotton (4x). Ornamentals such as roses, chrysanthemums, and tulips are also polyploid. Polyploidy is often associated with larger cells (because the nucleus is larger), and polyploids tend to be larger and more robust.

Some plant polyploids reproduce asexually because they are sterile. This is due to irregular segregation during meiosis, leading to aneuploid gametes. Triploidy is almost always sterile (bananas are propagated by cuttings, some apple species by grafts, and other species by bulbs). Triploids are often formed by fusion of a diploid (2x) gamete from a tetraploid (4x) parent with a normal gamete from a diploid parent. If x is large, you can imagine that meiosis would rarely result in balanced gametes; instead the gametes contain somewhere between x and 2x chromosomes. Because of the sterility, triploids rarely make seeds (recall a banana or seedless watermelon you ate recently). Remember the triploid oysters above.

Some tetraploids can provide viable offspring. Maintenance of a fertile tetraploid organism depends upon whether the plant can produce gametes with balanced sets of chromosomes. How are tetraploids usually generated?

Generation of Autopolyploids (polyploids created by chromosome duplication within a species): In diploid plants, sometimes sister chromatids fail to separate during mitosis, leading to tetraploid daughter cells; if these tetraploid cells give rise to reproductive tissues, then diploid (instead of haploid) gametes will be produced. A rare union of diploid gametes gives rise to a tetraploid individual; if this individual can self-pollinate, then a whole new species may arise. Autopolyploidy can also be induced artificially with colchicine.

Generation of Allopolyploids (polyploids created by hybridization between different species): Allopolyploids are generated by hybridization between two related species. The hybrid is sterile since the chromosomes (which can differ in shape, size, and number) cannot pair and segregate properly during meiosis. However, sometimes chromosomal doubling occurs, restoring fertility since doubling creates a proper pairing partner for each chromosome. If the two parental species are diploid, the resulting allopolyploid is called an **amphidiploid**. Meiosis is regular in amphidiploids because each chromosome now has a pairing partner, producing euploid gametes that can combine to allow normal sexual reproduction.

We will continue with a discussion of aneuploidy next lecture (see notes below):

MORE DEFINITIONS:

Aneuploid: Individuals have a numerical change in part of the genome. The chromosome number of aneuploids is not an exact multiple of the haploid number, n.
Hypoploid: an organism in which a chromosome (or part thereof) is underrepresented.
Hyperploid: an organism in which a chromosome (or part thereof) is overrepresented.

Aneuploidy is usually created by nondisjunction at meiosis I or II. If nondisjunction occurs at Meiosis I, none of the gametes produced have the haploid number of chromosomes. If non-disjunction occurs at Meiosis II, then 2 normal gametes and 2 aneuploid gametes are produced.

[Did not review in class, but one classic study was done by Blakeslee and Belling with Jimson weed. Jimson weed is diploid, having 12 pairs of chromosomes. A collection of 12 different mutants with peculiar inheritance patterns were isolated. The mutant phenotypes appeared to be caused by dominant factors transmitted mostly through the female. When the chromosomes were examined, it was shown that each of the twelve different mutant strains carried an extra copy of a particular chromosome (**trisomy, 2n+1**).]

Trisomies do occur in humans. The best known is Downs syndrome (trisomy of chromosome 21), which occurs in approximately 1/700 live births. Other much rarer trisomies are Patau Syndrome (trisomy 13) and Edwards Syndrome (trisomy 18). Individuals with trisomy 13 or 18 often die within the first few weeks of birth.

Trisomies of the sex chromosomes have less severe consequences. In humans, **Triplo-X** (XXX) females have normal genetilia, some fertility, and slight mental retardation, but otherwise are unaffected. **Klinefelter Syndrome**: XXY male (or XXXY, XXYY, XXXXY, XXXXXY) is subfertile with some feminization (those with >2 X's usu have mental impairment).

All other trisomies are embryonic lethals; in fact a large number of spontaneous abortions are due to abnormal chromosome abnormalites, especially trisomy, sex chromosome monosomy, and triploidy.

All human **monosomies (2n-1)**, except X, are embryonic lethals. A few 21 monosomics have survived beyond birth with severe multiple abnormalities. **Turner Syndrome**: (XO) female with retarded sexual development who is usually sterile. Somatic mosaics can also be the cause of Turner's syndrome and some cases of mild Down's syndrome (the severity of the syndrome will depend upon when during development the mitotic non-disjunction event occurred).

Aneuploidy for the autosomes seems generally to be more severe than aneuploidy for the X. In future lectures, we may discuss dosage compensation, which helps explain why this is true.