Variations in Chromosome Structure & Function

Ch. 8
Variation in Chromosome Number

- Chromosome numbers can vary in two main ways
  - Euploidy
    - Variation in the number of complete sets of chromosome
    - Variations occur occasionally in animals and frequently in plants
  - Aneuploidy
    - Variation in the number of particular chromosomes within a set
    - Variations are always regarded as abnormal conditions
Polyploid organisms have three or more sets of chromosomes.

Individual is said to be trisomic.

(b) Variations in euploidy

(c) Variations in aneuploidy

Individual is said to be monosomic.
The phenotype of every eukaryotic species is influenced by thousands of different genes
   – The expression of these genes has to be intricately coordinated to produce a phenotypically normal individual

Aneuploidy commonly causes an abnormal phenotype
   – It leads to an imbalance in the amount of gene products
In most cases, these effects are detrimental. They produce individuals that are less likely to survive than a euploid individual.
Aneuploidy

- Alterations in chromosome number occur frequently during gamete formation
  - ~5-10% of embryos have an abnormal chromosome number
  - ~50% of spontaneous abortions are due to such abnormalities

- In some cases, an abnormality in chromosome number produces an offspring that can survive
<table>
<thead>
<tr>
<th>Condition</th>
<th>Frequency</th>
<th>Syndrome</th>
<th>Characteristics</th>
</tr>
</thead>
<tbody>
<tr>
<td><em>Autosomal</em></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Trisomy 21</td>
<td>1/800</td>
<td>Down</td>
<td>Mental retardation, abnormal pattern of palm creases, slanted eyes, flattened face, short stature</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>1/6,000</td>
<td>Edward</td>
<td>Mental and physical retardation, facial abnormalities, extreme muscle tone, early death</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>1/15,000</td>
<td>Patau</td>
<td>Mental and physical retardation, wide variety of defects in organs, large triangular nose, early death</td>
</tr>
<tr>
<td><em>Sex Chromosomal</em></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>XXY</td>
<td>1/1,000 (males)</td>
<td>Klinefelter</td>
<td>Sexual immaturity (no sperm), breast swelling</td>
</tr>
<tr>
<td>XYY</td>
<td>1/1,000 (males)</td>
<td>Jacobs</td>
<td>Tall</td>
</tr>
<tr>
<td>XXX</td>
<td>1/1,500 (females)</td>
<td>Triple X</td>
<td>Tall and thin, menstrual irregularity</td>
</tr>
<tr>
<td>XO</td>
<td>1/5,000 (females)</td>
<td>Turner</td>
<td>Short stature, webbed neck, sexually undeveloped</td>
</tr>
</tbody>
</table>
Aneuploidy

- The autosomal aneuploidies compatible with survival are trisomies 13, 18 and 21
  - These involve chromosomes that are relatively small

- Aneuploidies involving sex chromosomes generally have less severe effects than those of autosomes
  - This is explained by X inactivation
    - All additional X chromosomes are converted into Barr bodies

- The phenotypic effects listed in Table 8.1 may be due to
  - 1. The expression of X-linked genes prior to embryonic X-inactivation
  - 2. An imbalance in the expression of pseudoautosomal genes
Aneuploidy

- Some human aneuploidies are influenced by the age of the parents
  - Older parents more likely to produce abnormal offspring
  - Example: Down syndrome (Trisomy 21)
    - Incidence rises with the age of either parent, especially mothers
Down syndrome is caused by the failure of chromosome 21 to segregate properly
- This **nondisjunction** most commonly occurs during meiosis I in the oocyte

The correlation between maternal age and Down syndrome could be due to the age of oocytes
- Human primary oocytes are produced in the ovary of the female fetus prior to birth
  - They are however arrested in prophase I until the time of ovulation
- As a woman ages, her primary oocytes have been arrested in prophase I for a progressively longer period of time
  - This added length of time may contribute to an increased frequency of nondisjunction
Euploidy

- Most species of animals are diploid (2n)

- In many cases, changes in euploidy are not tolerated
  - Polyploidy in animals is generally a lethal condition

- Some euploidy variations are naturally occurring
  - Female bees are diploid
  - Male bees (drones) are monoploid
    - Contain a single set of chromosomes
In contrast to animals, plants commonly exhibit polyploidy
  – 30-35% of ferns and flowering plants are polyploid
  – Many of the fruits and grain we eat come from polyploid plants

In many instances, polyploid strains of plants display outstanding agricultural characteristics
  – They are often larger in size and more robust
Euploidy

- Polyploids having an odd number of chromosome sets are usually sterile
  - These plants produce highly aneuploid gametes
    - Example: In a triploid organism there is an unequal separation of homologous chromosomes (three each) during anaphase I

Each cell receives one copy of some chromosomes and two copies of other chromosomes
Euploidy

- Sterility is generally a detrimental trait
- It is agriculturally desirable because it may result in
  - 1. Seedless fruit
    - Seedless watermelons and bananas
      - Triploid varieties
    - Asexually propagated by human via cuttings
  - 2. Seedless flowers
    - Marigold flowering plants
      - Triploid varieties
      - Developed by Burpee (Seed producers)
There are three natural mechanisms by which the chromosome number of a species can vary:

1. Meiotic nondisjunction
2. Mitotic abnormalities
3. Interspecies crosses
**Meiotic Nondisjunction**

- **Nondisjunction** refers to the failure of chromosomes to segregate properly during anaphase.

- Meiotic nondisjunction can produce haploid cells that have too many or too few chromosomes.
  - If such a gamete participates in fertilization:
    - The resulting individual will have an abnormal chromosomal composition in all of its cells.
During fertilization, these gametes produce an individual that is **trisomic** for the missing chromosome.

During fertilization, these gametes produce an individual that is **monosomic** for the missing chromosome.

(a) Nondisjunction in meiosis I

**All four gametes are abnormal**
Meiotic

Nondisjunction

Nondisjunction in meiosis II

50% Abnormal gametes

50% Normal gametes

(n + 1) (n - 1) n n

(b) Nondisjunction in meiosis II
Meiotic Nondisjunction

- In rare cases, all the chromosomes can undergo nondisjunction and migrate to one daughter cell.

- This is termed **complete nondisjunction**
  - It results in a diploid cell and one without chromosomes.
  - The chromosome-less cell is nonviable.
  - The diploid cell can participate in fertilization with a normal gamete.
    - This yields a triploid individual.
Aneuploidy

First meiotic division
Nondisjunction during first meiotic division

Second meiotic division
Gametes
Fertilization
Zygote

Trisomic (2n + 1)
Monosomic (2n - 1)
Monosomic (2n - 1)
Trisomic (2n + 1)
Diploid 2n
Mitotic Abnormalities

- Abnormalities in chromosome number often occur after fertilization
  - In this case, the abnormality occurs in mitosis not meiosis

  1. **Mitotic disjunction**
     - Sister chromatids separate improperly
       - This leads to trisomic and monosomic daughter cells

  2. **Chromosome loss**
     - One of the sister chromatids does not migrate to a pole
       - This leads to normal and monosomic daughter cells
Changes in Chromosome Number

Euploidy: Cells that contain only complete sets of chromosomes
- Diploidy ($2x$): Two copies of each homolog
- Monoploidy ($x$): One copy of each homolog

Polyploidy: More than the normal diploid number of chromosome sets
- Triploidy ($3x$): Three copies of each homolog
- Tetraploidy ($4x$): Four copies of each homolog

Aneuploidy: Loss or gain of one or more chromosomes producing a chromosome number that is not an exact multiple of the haploid number
- Monosomy ($2n - 1$)
- Trisomy ($2n + 1$)
- Tetrasomy ($2n + 2$)
Homework Problems

- Chapter 8

- # 1, 20, 21, 22, 23

- DON’T forget to take the online QUIZ!!
- DON’T forget to submit the online iActivity
  – “Karyotypes”