#### **Chromosomes and Karyotypes**







#### FIGURE 1-14

An electron micrograph of a human chromosome. Chromosome XII from a HeLa cell culture. (Courtesy of Dr. E. Du Praw.)

- 1838 Cell Theory, M.J. Schleiden and others (Schwann)
- 1842 chromosomes first seen by Nageli
- 1865 Charles Darwin, Pangenesis theory, blending inheritance
- **1865 Gregor Mendel** discovers, by crossbreeding peas, that specific laws govern hereditary traits. Each traits determined by pair of factors.
- **1869 -** Friedrich Miescher isolates DNA for the first time, names it nuclein.
- 1882 Walther Flemming describes threadlike 'chromatin' in the nucleus that turns red with staining, studied and named mitosis. The term 'chromosome' used by Heinrich Waldeyer in 1888.
- **1902** Mendel's work rediscovered and appreciated (DeVries, Corens, etc)
- 1903 Walter Sutton, the chromosomal theory of inheritance,

chromosomes are the carriers of genetic information

- 1944 Avery, MacLeod and McCarty show DNA was the genetic material
- 1953 James Watson and Francis Crick discover the molecular structure
  - of DNA: a double helix with base pairs of A + T and C + G.
- 1955 human chromosome number first established
- **1999 -** The first complete sequence of a human chromosome (22) was published.
- **2004 -** Complete sequencing of the human genome was finished by an international public consortium. **Craig Venter** etc.

#### Matthias Jakob Schleiden - 1838 proposes that cells are the basic structural elements of all plants.



#### **Cell Theory**

1. All living organisms are composed of one or more cells

 2. The cell is the basic unit of structure and organization of organisms
 3. All cells come from preexisting cells



Plate 1 from J. M. Schleiden, <u>Principles of</u> <u>Scientific Botany</u>, 1849, showing various

features of cell development







#### FIRST LAW:

- 1. Each trait due to a **pair of hereditary factors** which
- 2. segregate during gametogenesis SECOND LAW:
- 3. Multiple sets of hereditary factors assort independently

Mendel's work with peas showed that the "blending" explanation was wrong

# Walther Flemming - Mitosis





#### **Walter Sutton – Chromosome Theory of Inheritance**



-



| Chromo          | somes.          | Combinations in | Combinations in   |  |
|-----------------|-----------------|-----------------|-------------------|--|
| Somatic Series. | Reduced Series. | Gametes.        | Zygotes.          |  |
| 2               | I               | 2               | 4                 |  |
| 4               | 2               | 4               | 16                |  |
| 6               | 3               | 8               | 64                |  |
| 8               | 4               | 16              | 256               |  |
| 10              | 5               | 32              | 1,024             |  |
| 12              | 6               | 64              | 4,096             |  |
| 14              | 1 7             | 128             | 16,384            |  |
| 16              | 8               | 256             | 65,536            |  |
| 18              | 9               | 512             | 262,144           |  |
| 20              | 10              | 1,024           | 1,048,576         |  |
| 22              | II              | 2,048           | 4, 194, 304       |  |
| 24              | 12              | 4.096           | 16,777,216        |  |
| 26              | 13              | 8,192           | 07,108,864        |  |
| 28              | 14              | 16, 384         | 268,435,456       |  |
| 30              | 15              | 32,768          | 1,073,741,824     |  |
| 32              | 16 •            | 65.536          | 4,294,967,296     |  |
| 31              | 17              | 131,072         | 17, 179, 869, 184 |  |
| 36              | 18              | 202.144         | 68,719,476,736    |  |

Possible combinations of chromosome pairs at metaphase

#### Watson and Crick







Rosalind Franklin, X-ray diffraction of DNA

#### **Basic Definitions**

- gene basic unit of heredity; codes for a specific trait
- **locus** the specific location of a gene on a chromosome (locus plural loci)
- **chromosome** elongate cellular structure composed of DNA and protein they are the vehicles which carry DNA in cells
- **chromatid** one of two duplicated chromosomes connected at the centromere
- **centromere** region of chromosome where microtubules attach during mitosis and meiosis
- **diploid** (2n) cellular condition where each chromosome type is represented by two homologous chromosomes
- **haploid** (**n**) cellular condition where each chromosome type is represented by only one chromosome
- **homologous chromosome** chromosome of the same size and shape which carry the same type of genes

#### **Mitosis in Plants and Animals**









## **Cell Cycle**



# **Prophase**: the chromosomes begin to condense, while around the nucleus spindle fibres develop



**Metaphase**: the chromosomes line up along the equatorial plane of the cell



**Anaphase**: the chromosome pairs divide and the two groups migrate to opposite poles of the cell.



**Telophase** - a nuclear membrane forms, the chromosomes disperse and can no longer be distinguished. The spindle fibres dissolve. A new cell wall forms and the two cells separate.



#### Mitosis

#### Meiosis



#### 4 Haploid Gametes 1n







#### **Chromosome Number**

#### *Ophioglossum reticulatum* - 2N = 96X = 1440



Haplopappus (Machaeranthera) gracilis (Asteraceae)

2N = 2X = 4







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#### Index to Plant Chromosome Numbers (IPCN)

The *Index to Plant Chromosome Numbers* is an NSF funded project that aims to extract and index original plant chromosome numbers of naturally occurring and cultivated plants published throughout the world. A committee of voluntary contributing editors, located in various parts of the world, reviews sets of serial titles assigned to them and returns the information to the editors for collation in the *Index* and database. Chromosome indexes are published at two or three year intervals. The *Index to Plant Chromosome Numbers* project has been based at the Missouri Botanical Garden since 1978. Data from published indexes from 1979 onward are available for consultation through this facility.

For additional information, see the last supplement by Goldblatt & Johnson 2006. Index to Plant Chromosome Numbers 2001-2003. Monographs in Systematic Botany from the Missouri Botanical Garden 106.

An Index covering the years 2004-2006 is in preparation and will be published in the fall of 2008 as: Goldblatt & Johnson 2008. Index to Plant Chromosome Numbers 2004-2006. Monographs in Systematic Botany from the Missouri Botanical Garden.

Many but not all data in the printed version of the Index to Plant Chromosome Numbers (1979--) are available on the Web in the IPCN database. The printed indexes and the database provide references to chromosome counts reported in the original literature. We therefore request that the IPCN database itself not be cited as the source for chromosome counts. If there is a need to cite the IPCN database, we recommend the following:

Index to plant chromosome numbers. 1979-- . P. Goldblatt & D. E. Johnson, eds. Missouri Botanical Garden, St. Louis.





Peter Goldblatt MBG



Known chromosome numbers in Italian endemics range from 2n = 8 to 2n = 182. Mean chromosome number for Italian endemics is **2n = 30.68** 20.27 (median: 2n = 26, mode: 2*n* = 18).



#### **Chromosome Number**

**Variation exists within genera:** *Stylidium* (2N = 5-16, 26, 28, 30) *Cardamine* (2N = 16, 24, 28, 30, 32)

Variation exists within particular species: Rumex\_subgenus Rumex: 2N = 2X = 20, 2N = 4X = 40, 2N = 20X = 200. *Chaenactis douglasii*: 2N = 12-15, 18, 24-28, 36, 38.

0 6 6 6 7 7 XX 6 9 8

PROME 18.4 Drawings of metaphase chromosomes of Anemune (Rammulaceae). A. A. quinquirfuin, 2n=32; B. A. rivularis, 2n=16; C. A. richardsonii, 2n=14. x2500. (From Heimburger 1959:592)

Chamaecrista

Anemone

## Aneuploidy

a gain or loss of one or more chromosomes, e.g. 2N - 1, 2N + 1, 2N + 2, etc. The most common case are trisomies (sing. trisomy) where a single additional chromosome is present.



Fruits of *Datura*-Plants On top: Control plant (2n) Below: Mutants that are characterized by one additional chromosome each.

Loss of one or more chromosomes usually has more severe consequences



#### Aneuploidy

Claytonia virginica Walter Lewis (1970, 1971). Plants have different chromosome numbers in different parts of their ranges and even within same population. and within one individual from year to year.

"I would argue that if an organism does not take its chromosome number seriously, there is no reason why the systematist should" (Walter Lewis).



**FIGURE 19.2** Variation of diploid chromosome numbers in populations throughout the range (enclosed area) of *Claytonia virginica* (Portulacaceae). (Redrawn from Lewis, Oliver, and Suda 1967:154)

#### **Polyploidy – multiple sets of chromosomes**



Diploid – 2 sets Triploid – 3 sets (watermelon) Tetraploid – 4 sets (cotton) Hexaploids – 6 sets (wheat)

**Autopolyploids**: polyploids composed of multiple sets of chromosomes from the <u>same species</u>

**Allopolyploids**: polyploids that are a new species, composed of multiple sets of chromosomes from <u>closely related</u> species

#### Known Paleopolyploidy in Eukaryotes





#### **Hybrid Vigor**



An example of an allopolyploid that shows hybrid vigor over its diploid progenitors is resynthesized Brassica napus.

#### **Tripoids – 3 sets of chromosomes Triploid crops**: apple, banana, citrus, ginger, watermelon



| <i>Musa acuminata</i><br>(Asian Banana) | х | <i>Musa balbisiana</i><br>(Asian Banana) | = | <i>Musa X paradisiaca</i><br>(Hybrid Banana) |
|---|---|--|---|--|
| AA<br>(fertile)                         |   | BB<br>(fertile)                          |   | AAB or ABB (etc.)<br>(sterile)               |

Origin Of Triploid Banana From Asian Parents A = one haploid set of chromosomes from *M. acuminata* B = one haploid set of chromosomes from *M. balbisiana* 

#### Tetraploids – 4 sets of chromosomes





Failed meiosis, gametes 2N

**Tetraploid crops**: apple, durum or macaroni wheat, cotton, potato, cabbage, leek, tobacco, peanut, Pelargonium Hybridization followed chromosome doubling

#### **Examples of Polyploidy**



Current Biology

#### Hexaploid – 6 sets of chromosomes





**Autopolyploids**: polyploids composed of multiple sets of chromosomes from the <u>same species</u>

**Allopolyploids**: polyploids that are a new species, composed of multiple sets of chromosomes from <u>closely related</u> species









#### **Pre-treatment of Roots**

8-hydroxyquinoline -cause contraction and improve spreading of chromosomes

#### Fixation

Prevent autolysis of chromosome, denaturation of proteins.
Prevent bacterial decomposition of cells.
Prevents swelling and shrinking of cells and tissues.
Rapid penetration to reach the interior of the cell
Mixtures of chemicals are used, freshly prepared
Carnoy's: ethanol-acetic acid (3:1) or
Farmer's: ethanol-chloroform-acetic acid (6:3:1) (2:1:1)
Fixation time 30 minutes- 24 hours – 36 hours (maximum).
After fixation the material is preserved in 70% alcohol

#### **Aceto Carmine**

Carmine is a crimson dye, used in solution with 45% acetic acid known as **aceto-carmine**. Acetic acid is a good fixative for chromatin and is rapidly penetrating. The addition of a few drops of ferric chloride or ferric acetate intensifies the color..

Orcein

Fuelgen











#### Onion Root Tip Squash



#### Bean Root Tip Squash









# End