# Chapter 6 Cytogenetics: Karyotypes and Chromosomal Aberrations



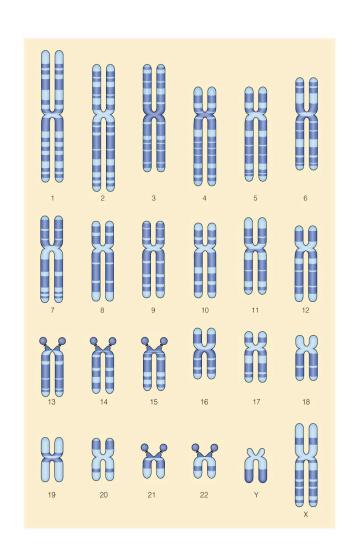
#### Chromosome Number Varies

Organism	Diploid Number (2 <i>n</i> )	Haploid Number ( <i>n</i> )
Human (Homo sapiens)	46	23
Chimpanzee (Pan troglodytes)	48	24
Gorilla (Gorilla gorilla)	48	24
Dog (Canis familiaris)	78	39
Chicken (Gallus domesticus)	78	39
Frog (Rana pipiens)	26	13
Housefly (Musca domestica)	12	6
Onion (Allium cepa)	16	8
Corn (Zea mays)	20	10
Tobacco (Nicotiana tabacum)	48	24
House mouse (Mus musculus)	40	20
Fruit fly (Drosophila melanogaster)	8	4
Nematode (Caenorhabditis elegans)	12	6



#### **Human Chromosomes**

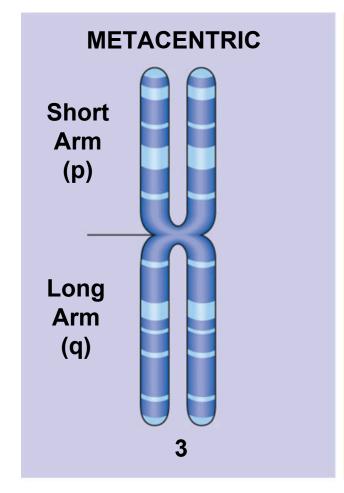
- Diploid number(2N) = 46
- 23 pairs
  - 22 pairs of autosomes
  - XX in females and XY in males
- Gametes (eggs and sperm) are haploid and have 23 chromosomes

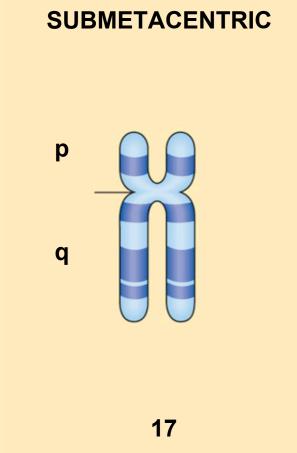


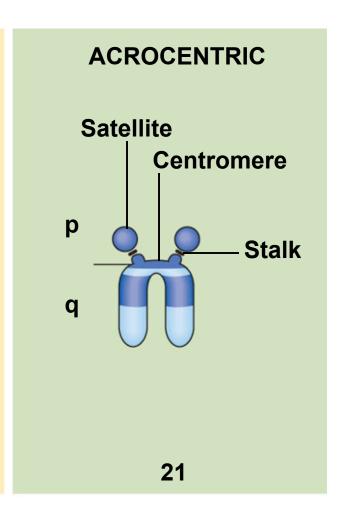




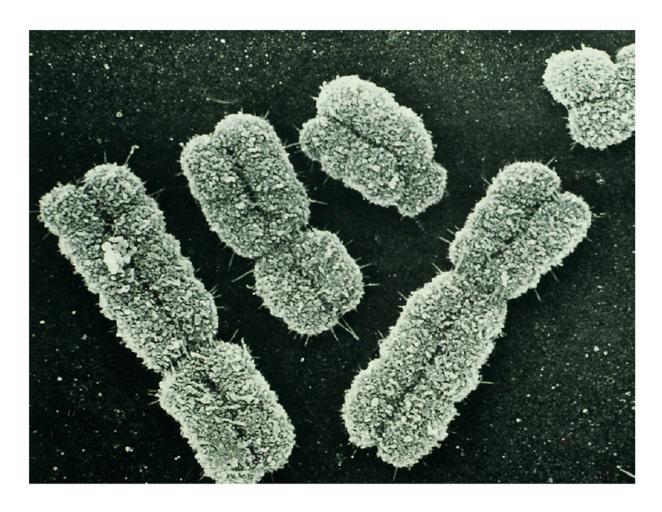
# The Centromere Divides the Chromosome into Two Arms





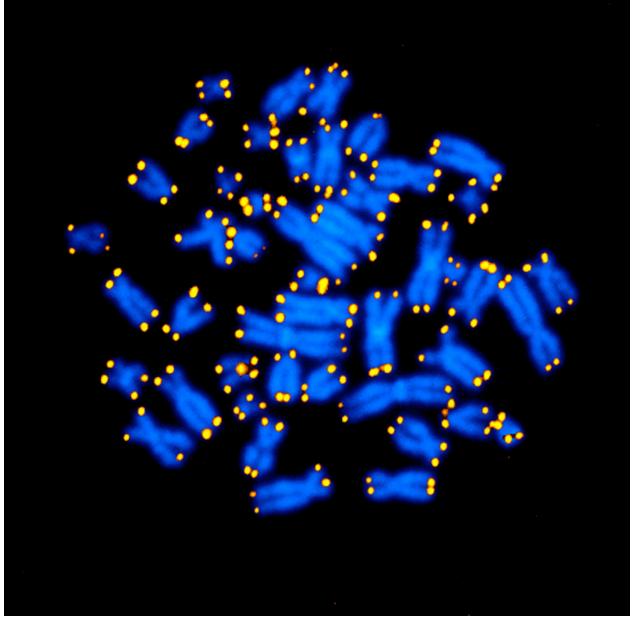


#### At metaphase of mitosis



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Telomeres (yellow)



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# Cells Used for Chromosomal Analysis

- Any cell with a nucleus
- Lymphocytes
- Skin cells
- Tumor cells
- Amniotic cells
- Chorionic villi
- Rare fetal cells from maternal blood



#### Karyotype

 Chromosomes photographed during metaphase and arranged in a standard sequence

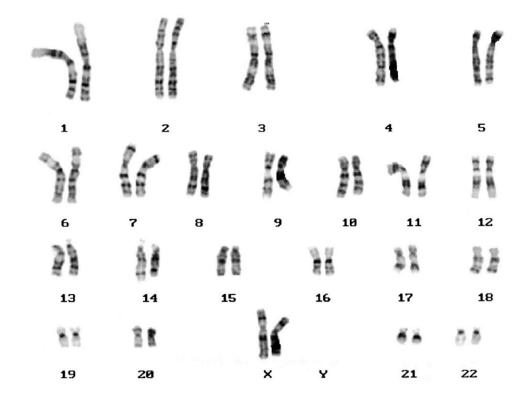


Fig. 6.3



### Creating a Karyotype

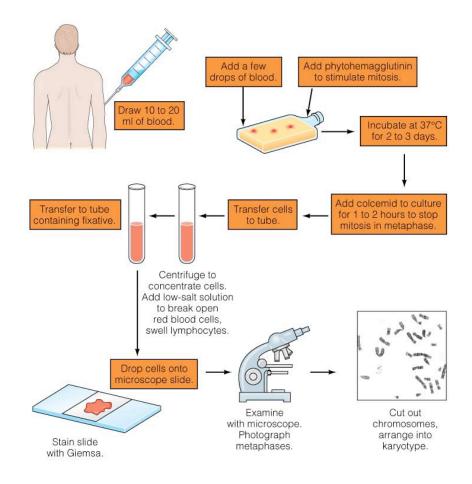


Fig. 6.6



#### Stains and Dyes

- Used to produce a pattern of bands specific to each type of chromosome
- One common method is G-banding
  - Treated with trypsin
  - Stained with Giemsa stain
  - Metaphase chromosomes approximately 550 bands
  - More bands can be produced in early metaphase and late prophase chromosomes



# Banding Techniques

Stains and dyes are used to identify the chromosomes

Banding technique

**G-banding** — Treat metaphase spreads with enzyme that digests part of chromosomal protein. Stain with Giemsa stain. Observe banding pattern with light microscope.

Q-banding — Treat metaphase spreads with the chemical quinacrine mustard. Observe fluorescent banding pattern with a special ultraviolet light microscope.

R-banding — Heat metaphase spreads at high temperatures to achieve partial denaturation of DNA. Stain with Giemsa stain. Observe with light microscope.

C-banding — Chemically treat metaphase spreads to extract DNA from the arms but not the centromeric regions of chromosomes. Stain with Giemsa stain and observe with light microscope.

Appearance of chromosomes



Darkly stained G bands.



➤ Bright fluorescent bands upon exposure to ultraviolet light; same as darkly stained G bands.

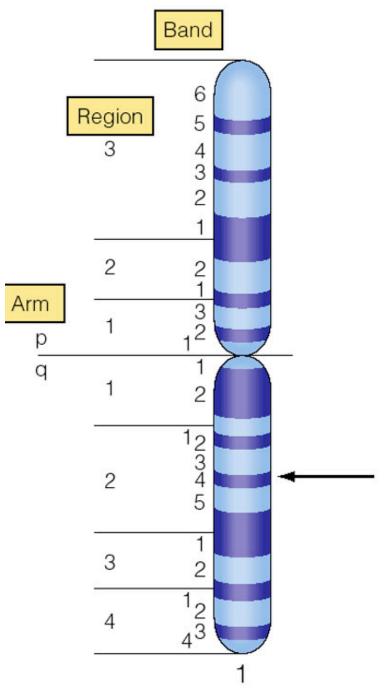


Darkly stained R bands correspond to light bands in G-banded chromosomes. Pattern is the reverse of G-banding.



Darkly stained C band centromeric region of the chromosome corresponds to region of constitutive heterochromatin.





Banding patterns allow individual chromosomes to be identified

Provide location of genes

Information about structural aberrations

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Fig. 6-5, p.124