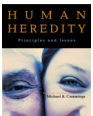


Chapter 6

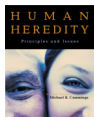
Cytogenetics: Karyotypes and Chromosomal Aberrations



Chromosome Number Varies

Table 6.1 Chromosome Number in Selected Organisms

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

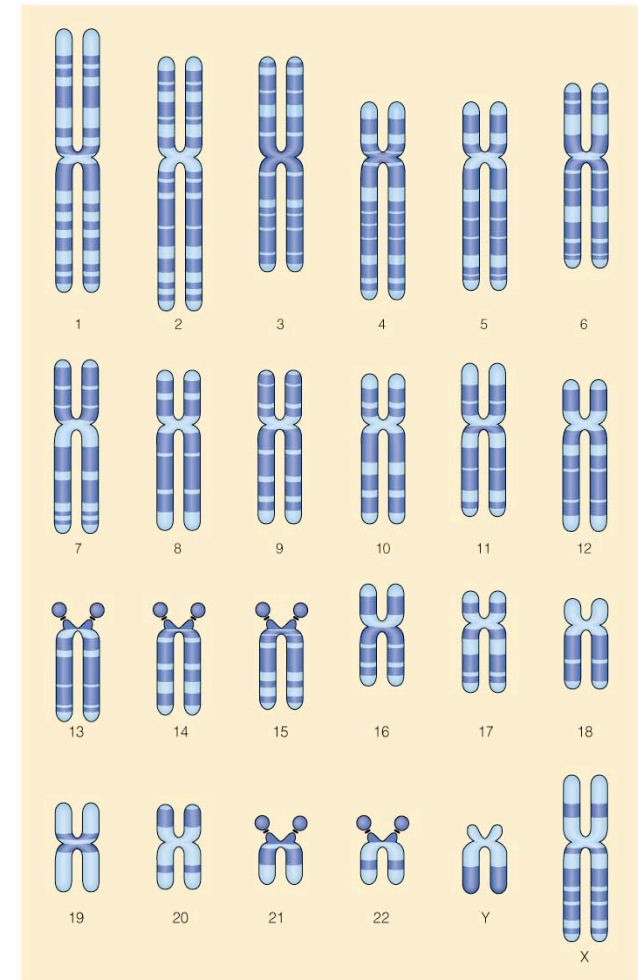
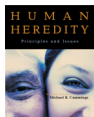
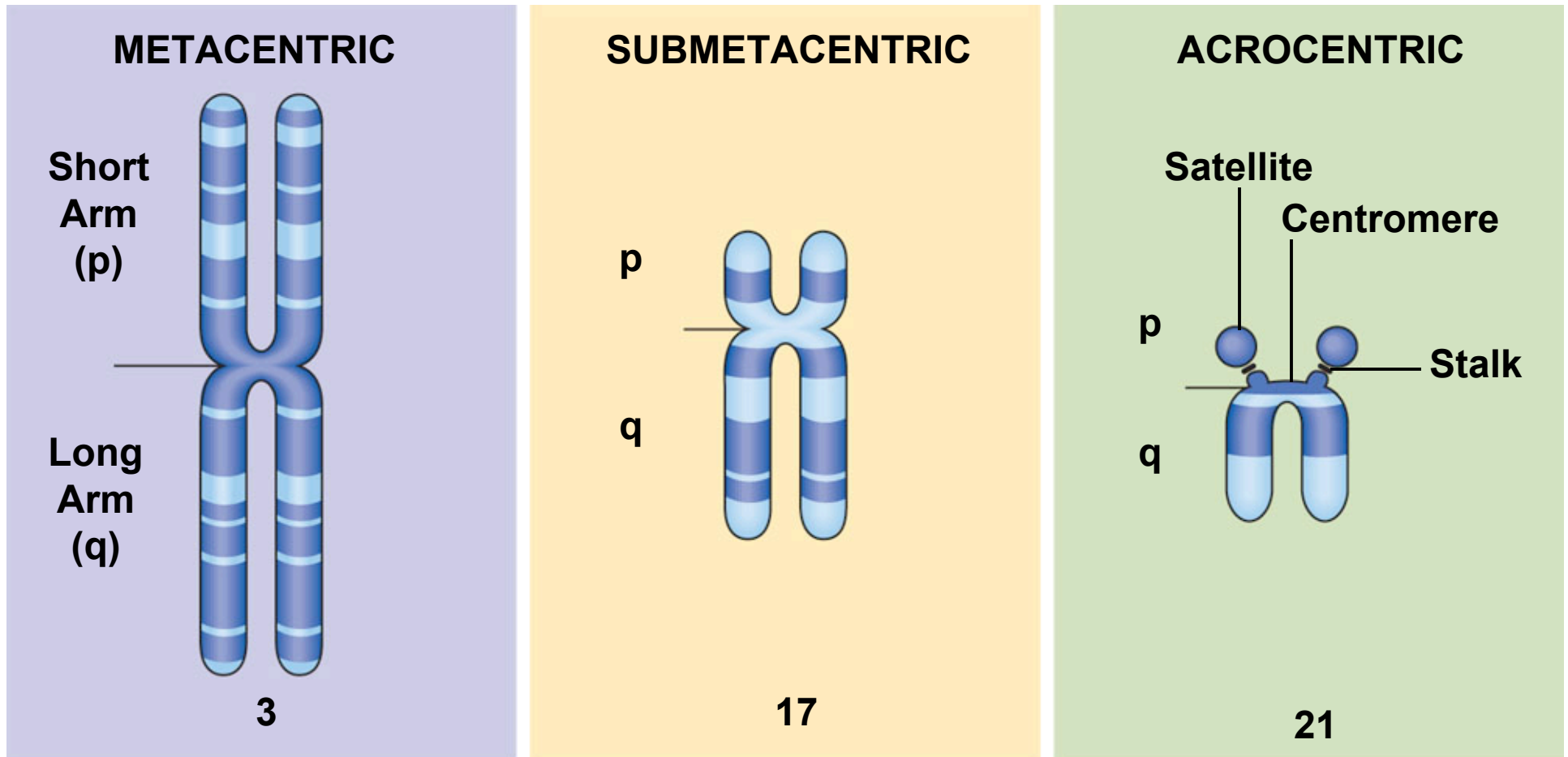


Fig. 6.4



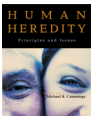
The Centromere Divides the Chromosome into Two Arms



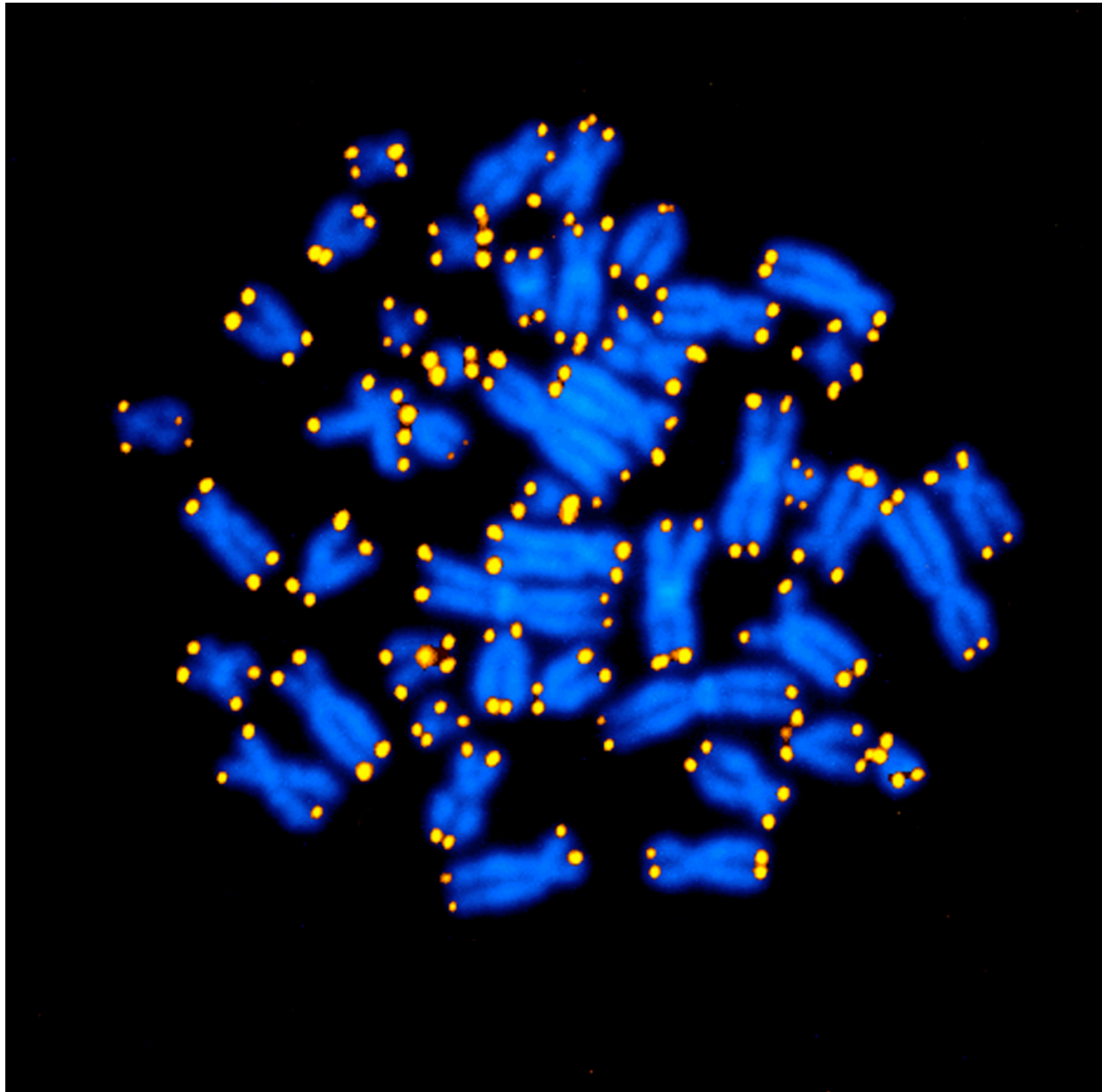
At metaphase of mitosis



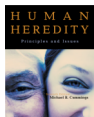
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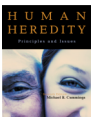


Telomeres
(yellow)



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

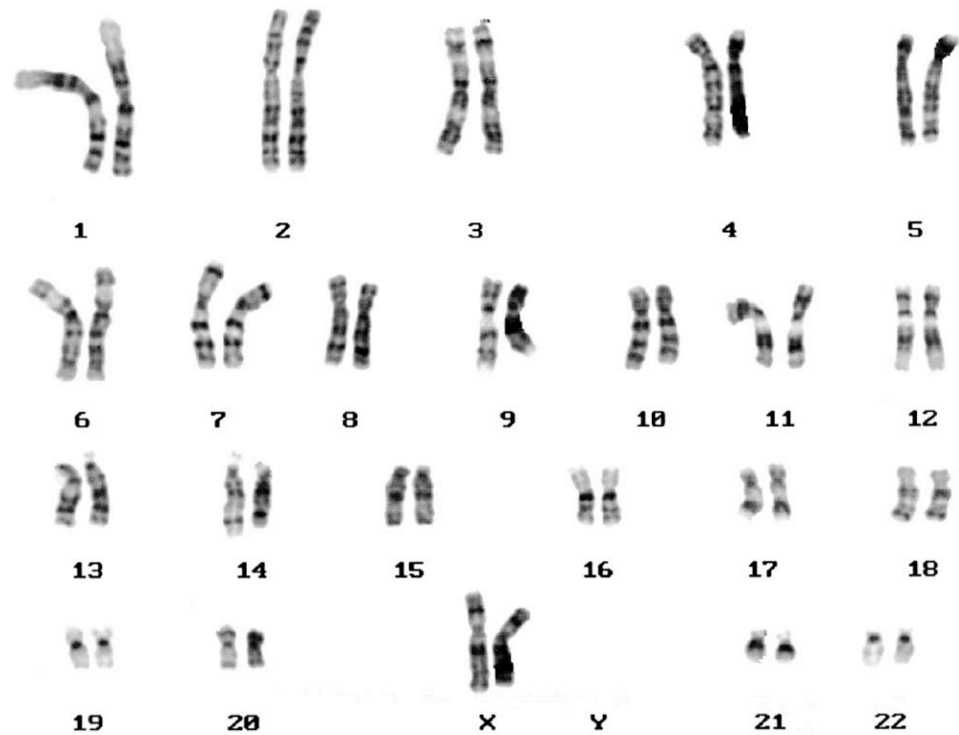
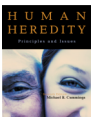


Fig. 6.3



Creating a Karyotype

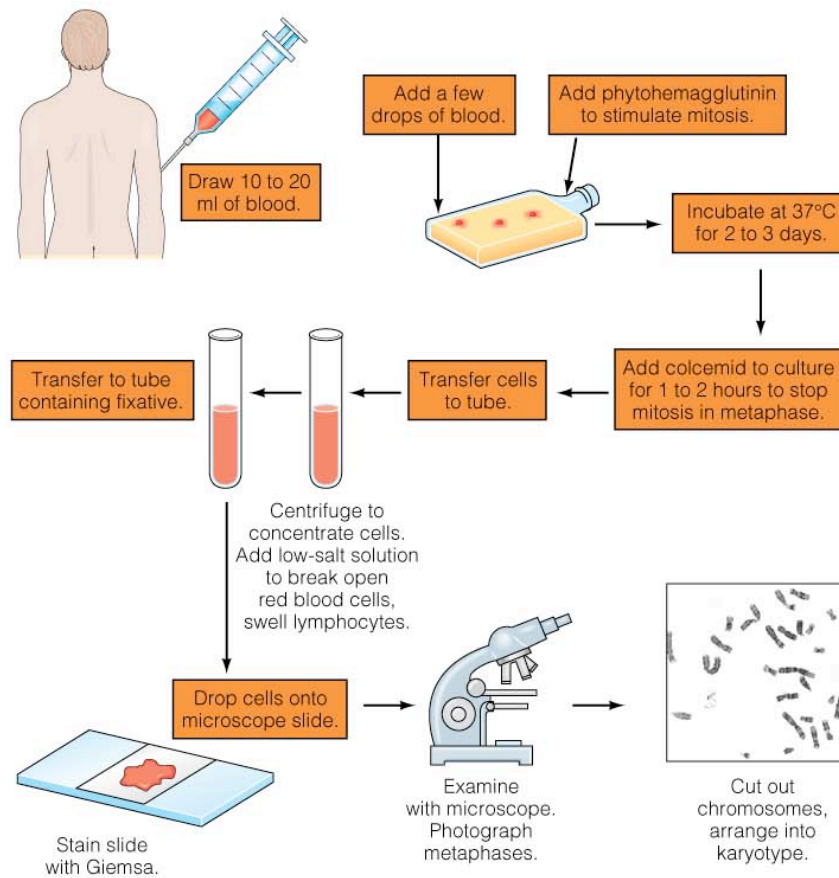
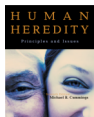
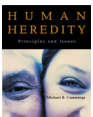


Fig. 6.6



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

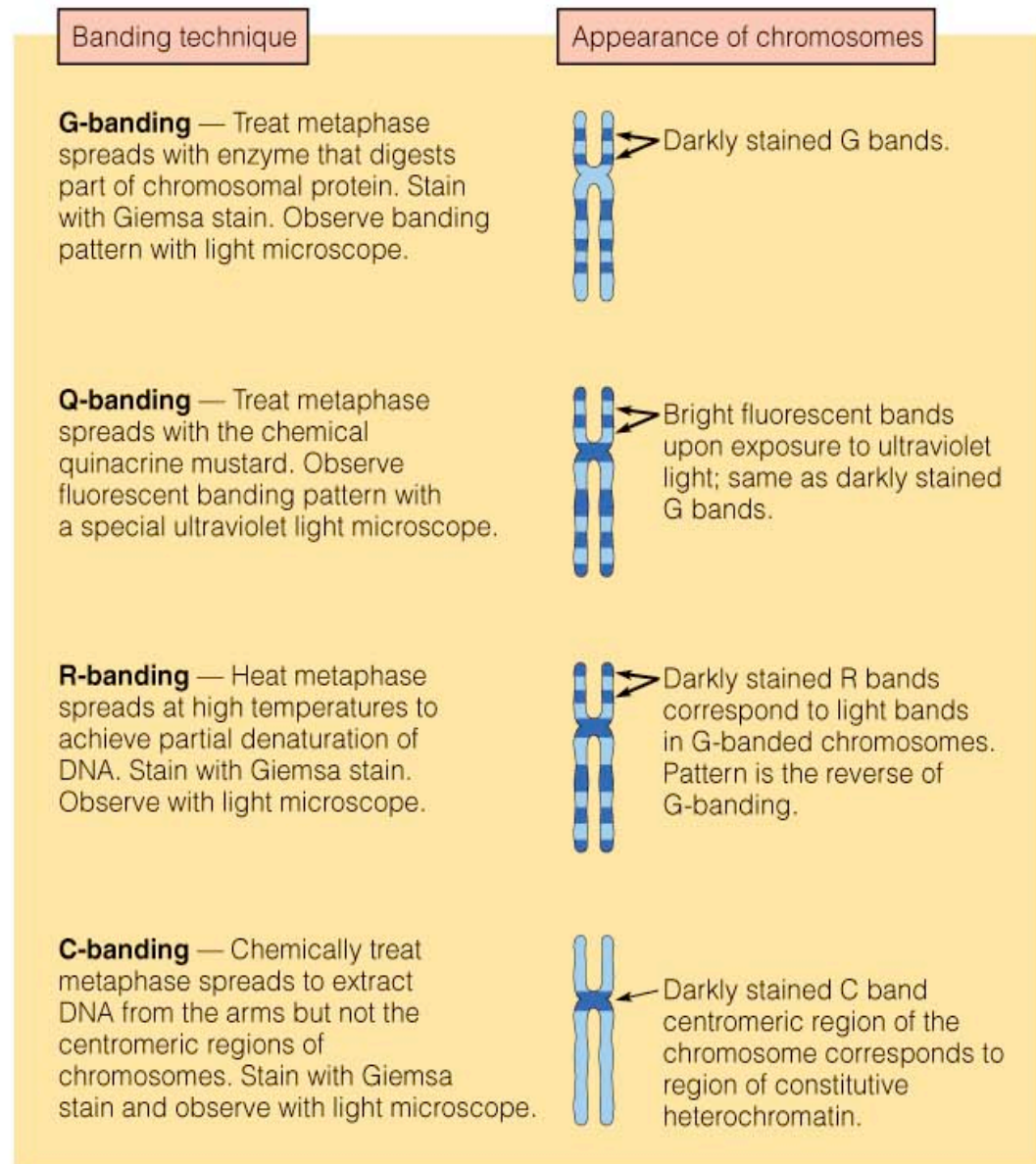
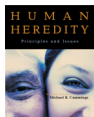
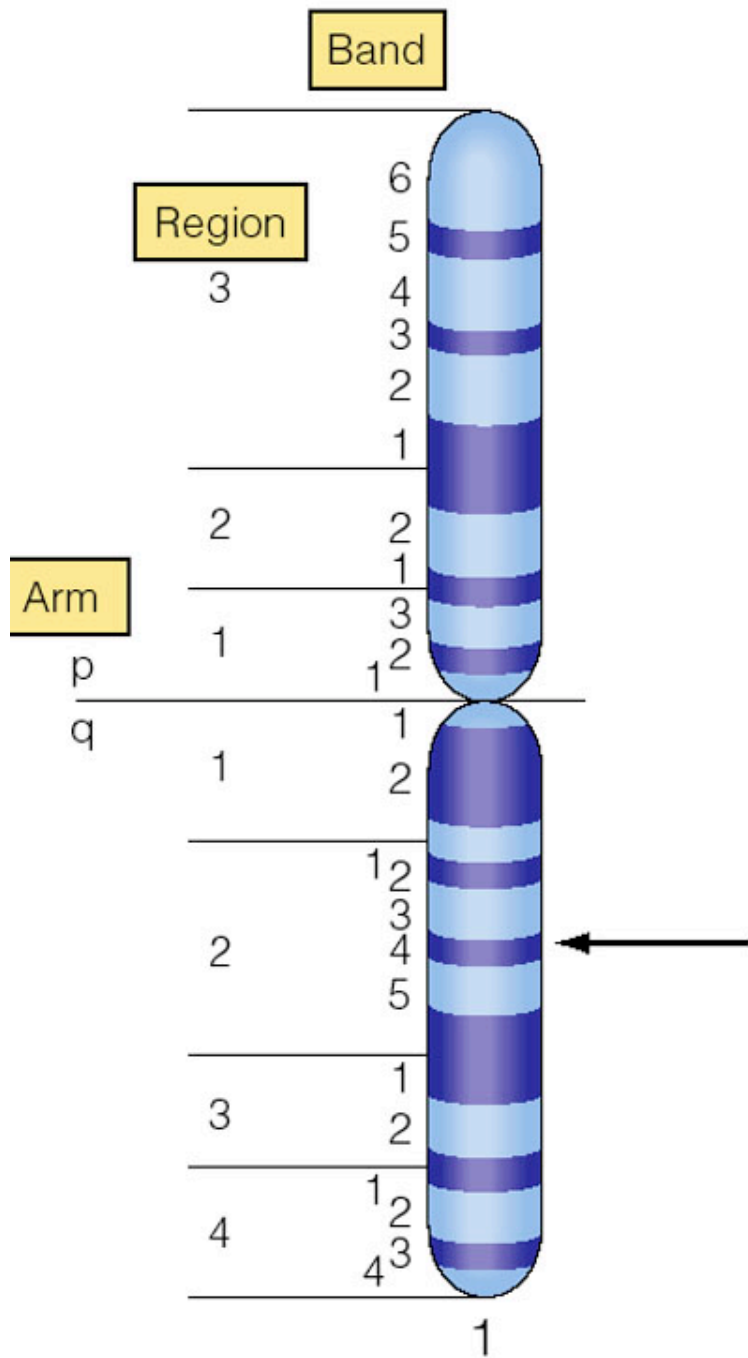


Fig. 6.8





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