Chapter 11

Chromosomes and Human Inheritance
Human Chromosomes

- Human body cells have 23 pairs of homologous chromosomes
  - 22 pairs of autosomes
  - 1 pair of sex chromosomes
Autosomes and Sex Chromosomes

• Paired autosomes
  – Same in length and shape
  – Have the same centromere location
  – Carry the same genes along their length

• Sex chromosomes
  – Identical in human females (XX)
  – Nonidentical in human males (XY)
Autosomes and Sex Chromosomes

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Sex Determination

• SRY gene on the Y chromosome
  – Basis of male sex determination
  – Expression initiates synthesis of testosterone
  – Hormone causes embryo to develop into a male

• If an embryo has no Y chromosome (no SRY gene), it develops into a female
Sex Determination Pattern

meiosis, gamete formation in both female and male:

fertilization:

sex chromosome combinations possible in the new individual
Karyotyping

• Diagnostic tool that reveals missing or extra chromosomes and some structural changes in an individual’s chromosomes

• Metaphase chromosomes are prepared for microscopy, then imaged

• Chromosome images are arranged in sequence based on defining features
The image is reassembled: The chromosomes are paired by size, centromere position, and other characteristics.
Key Concepts:

AUTOSOMES AND SEX CHROMOSOMES

• All animals have pairs of autosomes—chromosomes identical in length, shape, and which genes they carry

• Sexually-reproducing species also have a pair of sex chromosomes (differ between females and males)
Key Concepts: AUTOSOMES AND SEX

CHROMOSOMES (cont.)

• A gene on one of the human sex chromosomes dictates the male sex

• Karyotyping, a diagnostic tool, reveals changes in the structure or number of an individual’s chromosomes
Autosomal Inheritance Patterns

• Some alleles on autosomes are inherited in simple Mendelian patterns associated with specific phenotypes.

• Certain mutated forms of alleles give rise to genetic abnormalities or genetic disorders.
Autosomal Dominant Inheritance

NORMAL MOTHER: aa
(meiosis, gamete formation)

AFFECTED FATHER: Aa
(meiosis, gamete formation)

A sperm, a egg

Aa affected child

aa normal child

Aa affected child

aa normal child

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Autosomal Dominant Inheritance

Examples

• Huntington’s Disease
• Achondroplasia
Autosomal Recessive Inheritance

CARRIER MOTHER

Aa (meiosis, gamete formation)

CARRIER FATHER

Aa (meiosis, gamete formation)

Sperm

A

eggs

A

Aa normal child

Aa carrier child

aa affected child
Autosomal Recessive Inheritance

- Example
- Galactosemia

It is a heritable metabolic disorder
Autosomal Recessive Disorders

- Galactosemia
Progeria

• Progeria
• Disorder arises by spontaneous mutation of a gene for lamin
• Distorts the nucleus
• Skeletal Muscles weakens
X-Linked Inheritance Patterns

• Certain dominant and recessive alleles on the X chromosome are inherited in Mendelian patterns

• Mutated alleles on the X chromosome contribute to more than 300 known genetic disorders

• Males can’t transmit recessive X-linked alleles to sons (son receives X chromosome from mother)
X-Linked Inheritance Patterns

CARRIER MOTHER

XX

(meiosis, gamete formation)

NORMAL FATHER

XY

(meiosis, gamete formation)

sperm

X

YY

normal daughter

normal son

carrier daughter

affected son

eggs
Hemophilia A

- X-linked recessive inheritance
- Hemophilia is an inherited disease that prevents the blood from clotting.
- People with hemophilia have a deficiency of a blood protein, also called a “clotting factor,” that is necessary to clot the blood and stop bleeding.
Color Blindness
Red – Green Color Blindness

• Condition in which an individual cannot distinguish some or all colors in the spectrum
• In red-green color blindness, some or all of the receptors that respond to red and green wavelength are weakened or absent
Key Concepts:

SEX-LINKED INHERITANCE

- Some traits are affected by genes on the X chromosome

- Inheritance patterns of such traits differ in males and females
Duchenne Muscular Dystrophy (DMD)

• DMD is one of a group of X-linked recessive disorders characterized by rapid degeneration of muscles.
• A recessive allele encodes dystrophin.
• When dystrophin is abnormal, cell cortex weakens and muscle cells die.
Heritable Changes in Chromosome Structure

- Duplication
  
  ![Diagram of Duplication]
  
  Normal chromosome

  ![Diagram showing one segment repeated]
  
  One segment repeated

- Deletion
  
  ![Diagram of Deletion]
  
  Segment C deleted

![Diagram showing segment C deleted]
Heritable Changes in Chromosome Structure

- **Inversion**

  ![Inversion Diagram]

  Segment G, H, I become inverted

- **Translocation**

  ![Translocation Diagram]

  reciprocal translocation
Evolution of Chromosome Structure

- Most alterations are harmful or lethal
- Over evolutionary time, many alterations with neutral or useful effects have accumulated in chromosomes of all species
Evolution of the Y Chromosome

- **a** Before 350 mya, sex was determined by temperature, not by chromosome differences.
- **b** SRY gene evolves 350 mya. Other mutations accumulate and the chromosomes of the pair diverge.
- **c** By 320–240 mya, the two chromosomes have diverged so much that they no longer can recombine in one region. The Y chromosome begins to degenerate.
- **d** Three more times, 170–130 mya, 130–80 mya, and 50–30 mya, the pair fails to recombine in another region. Each time, more changes accumulate, and the Y chromosome shortens. Today, the pair recombines only at a small region near the ends.
Key Concepts: **CHANGES IN CHROMOSOME STRUCTURE**

- On rare occasions, a chromosome may undergo permanent change in its structure, when a segment of it is deleted, duplicated, inverted, or translocated.
Heritable Changes in Chromosome Number

• Chromosome number of a parental cell can change permanently
• Often caused by **nondisjunction**
  – Failure of one or more pairs of duplicated chromosomes to separate during meiosis or mitosis
  – Nondisjunction affect the chromosome number at fertilization
Nondisjunction in Meiosis

1. Chromosome alignments at metaphase I
2. Nondisjunction at anaphase I
3. Alignments at metaphase II
4. Anaphase II
5. Chromosome number in gametes

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Aneuploidy

• Cells with too many or too few copies of a chromosome
  – In humans, the most common aneuploidy (trisomy 21) causes Down syndrome

• Most other human autosomal aneuploids die before birth
Aneuploidy:
Trisomy 21 (Down Syndrome)
Polyploidy

- **Polyploid** individuals inherit three or more of each type of chromosome from their parents.

- About 70% of all flowering plants, and some insects, fishes, and other animals are polyploid.
Sex Chromosome Abnormalities

• Change in number of sex chromosomes usually results in learning and motor skill impairment
  – Turner syndrome (XO)

• Problems can subtle and undiagnosed
  – XXX syndrome
  – Klinefelter syndrome (XXY)
  – XYY condition
Key Concepts: CHANGES IN CHROMOSOME NUMBER

• On rare occasions, the number of autosomes or sex chromosomes changes

• In humans, the change usually results in a genetic disorder
Human Genetic Analysis

• Geneticists construct pedigrees
  – Charts of genetic connections among individuals
  – Estimate chances that a couple’s offspring will inherit a certain trait
A Pedigree for Polydactyly
Prospects in Human Genetics

• Potential parents at risk of transmitting a harmful allele to their offspring have several options
  – Genetic counseling
  – Prenatal diagnosis
  – Preimplantation diagnosis (*in vitro*)
  – Phenotypic treatments
  – Abortions
  – Genetic screening
Prenatal Diagnosis: Fetoscopy

- a. Pulsed sound waves guide endoscope to umbilical cord
- b. Placement of needle with help of fiber optics
- c. Needle punctures tiny fetal vein in the cord
- d. Blood sample withdrawn by syringe