Chapter 5

Hereditary Influences on Health Promotion of the Child and Family

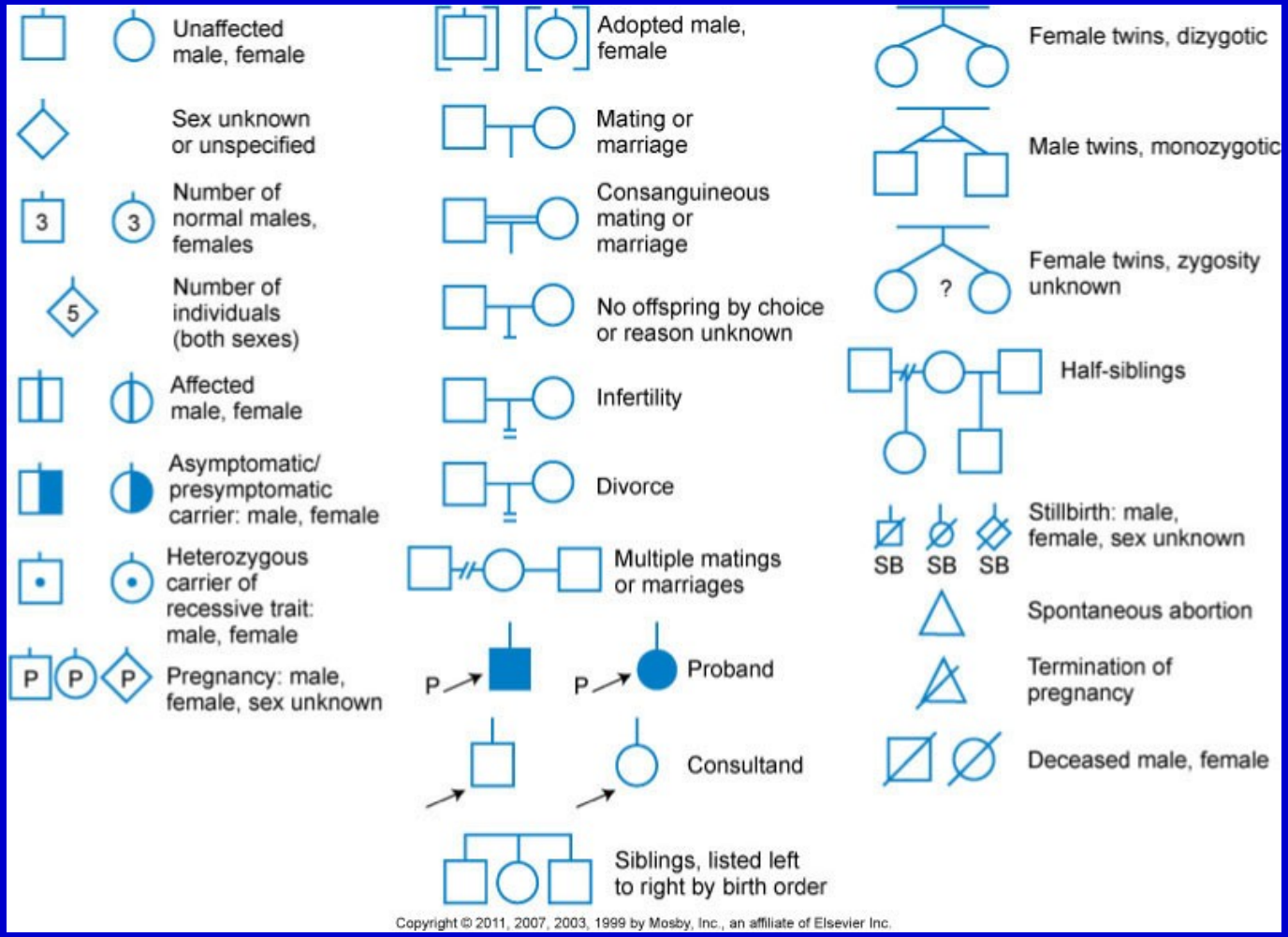
GENETIC INFLUENCES ON HEALTH

- Human Genome Project
- Pharmacogenetics/Pharmacogenomics
  - Allows individualized medication selection and dosing to improve efficacy and safety
- Heredity-environmental interplay in diseases
  - Genetic predisposition activated by an environmental trigger
    - Multifactorial diseases

Examples of Heredity and Environment Interplay in Human Diseases

- Single-gene disorders
  - PKU
- “Inherited” genetic mutations
- Anomalies from prenatal environmental causes
  - Congenital amputation caused by amniotic bands
  - Cleft lip and palate

Pedigree Symbols



Chromosomes

- Normal somatic cell has 23 pairs of chromosomes (46 total)

Chromosome Disorders

- Definition
  - Deviation in structure or number of a chromosome
- Result from
  - Duplication of genetic material or
  - Loss of genetic material
    - “Monogenetic” or “polygenetic” disorders

Chromosome Disorders—Types

- Structural abnormality
  - Loss, addition, rearrangement, or exchange of genes of a chromosome
  - Usually result from an error in cell division in sperm or ovum (parents are normal)

Numeric Chromosome Abnormalities

- Occur whenever entire chromosomes are added or deleted
- Euploidy: Uniform addition of chromosomes to all the original pairs
- Trisomy: Additional chromosome
- Monosomy: Deletion of one chromosome
  - Most types incompatible with life

Autosomal Inheritance Patterns

- Numeric alterations affecting the autosomes
- Examples include some of the most common trisomies:
  - Trisomy 21 (Down syndrome)
  - Trisomy 18 (Edwards syndrome)
  - Trisomy 13 (Patau syndrome)

Aneuploidy

- An abnormal chromosome pattern in which the total number of chromosomes is not a multiple of the haploid number (23)
- The most common aneuploidies in humans are *trisomies*

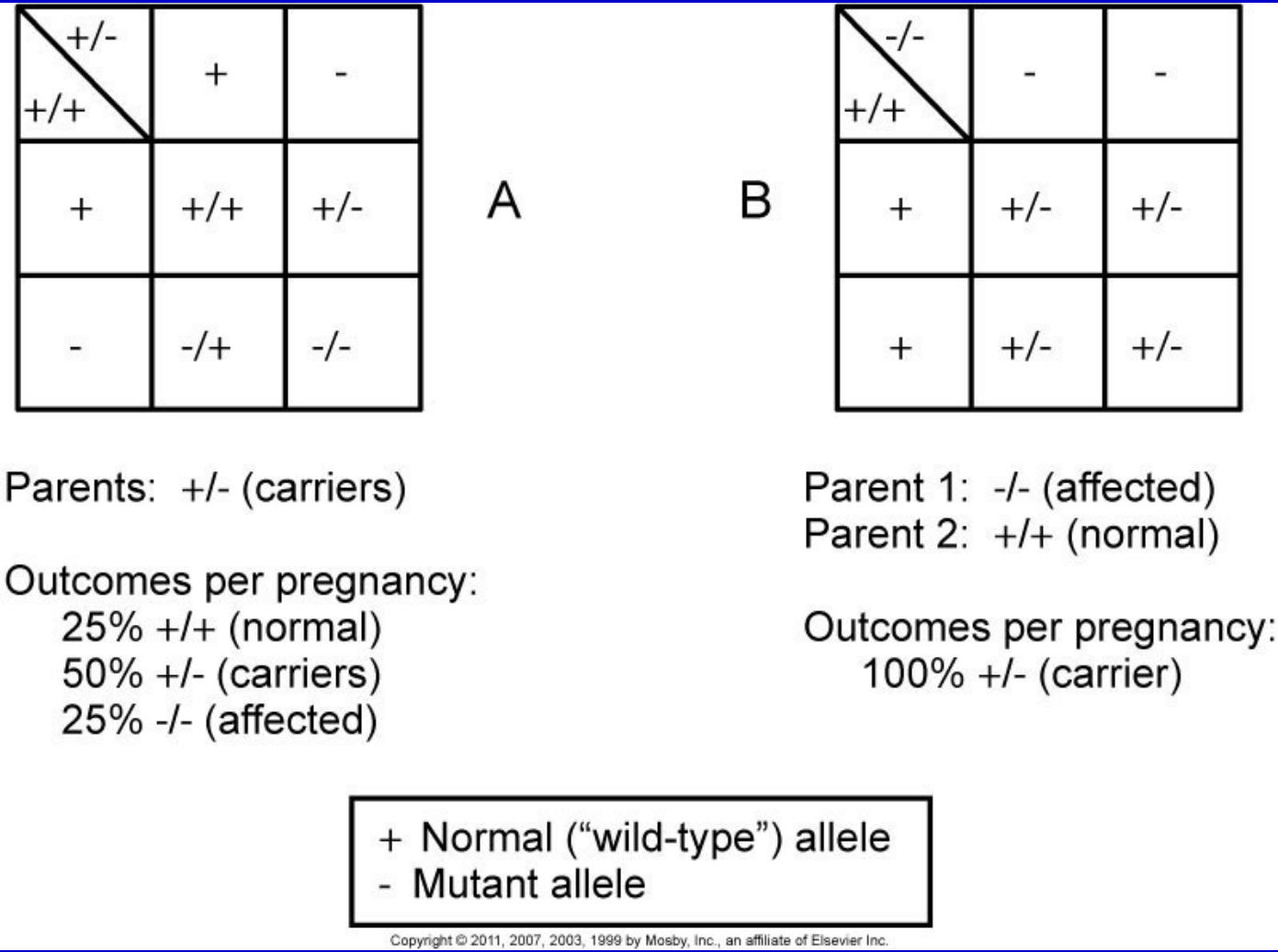
X-Linked Recessive Inheritance

- Affected individuals are principally males.
- All carrier females are normal.
- All affected males have symptoms of the disorder.
- Males are not carriers.

Sex Chromosome Aneuploidies

- Alteration in the number of sex chromosomes
- Profound effects are rare
- May have decreased intelligence or learning disabilities
- Examples
  - Klinefelter syndrome
  - Turner syndrome

Autosomal Recessive Inheritance



Chromosome Disorders

- Suffix “-somy”
  - Gain or loss of a chromosome
  - One missing = monosomy
  - Extra one = trisomy

Chromosome Disorders—cont'd

- Errors in cell division
  - Nondisjunction
  - Mosaicism
  - Translocation

Nondisjunction

- Failure of a pair of chromosomes to separate at meiosis
- Causes one daughter cell to have two chromosomes and the other to have none

Mosaicism

- Presence of two or more chromosomally distinct cell lines in the same individual
- Extent (expressed as percentage) depends on the stage of embryonic development during which the cell division error occurs

Translocation

- Alteration of a chromosome by transfer of a portion of it either to another chromosome or to another portion of the same chromosome

Translocation—cont'd

- No ultimate loss or gain of genetic material
  - "Carriers"
  - Phenotypically normal
  - May pass translocation to offspring

Unbalanced Translocation

- Typically lethal
- Spontaneous abortion occurs often

Congenital Anomalies

- Deformations
- Disruptions
- Dysplasias
- Malformations
- Syndrome
- Association
- Sequence

Syndrome

- A recognized pattern of malformations with a single, specific anatomic, physiologic, or biochemical cause

Trisomy 21 = Down Syndrome

- **Trisomy**—92% of all cases
  - Female with trisomy is 47,XX,+21
  - Male with trisomy is 47,XY,+21
  - The extra chromosome 21 is unattached and segregates freely during meiosis
  - The risk for this type increases with maternal age
- **Translocation Down syndrome**—4% of all cases
  - Male robertsonian translocation between acrocentric chromosomes 14 and 21 is 46,XY,t(14;21).
  - The majority of cases are without family history
  - Approximately 25% have one balanced translocation carrier parent

Trisomy 21 = Down Syndrome—cont'd

- **Mosaic Down syndrome**—Rare cases
  - Female with mosaic Down syndrome is 46,XX/47,XX,+21.
  - Results from mitotic nondisjunction during early embryonic development of a normal zygote. Affected children have mixed cell populations, some with the normal karyotype, others with the extra chromosome.
  - The proportion of trisomic cells affect the child's developmental potential and syndrome-associated potential health problems.

Trisomy 18

- Edwards syndrome
- Severe cognitive impairment and physical abnormalities
- Short life span

Trisomy 13

- Patau syndrome
- Severe malformations = greater gene imbalance
- Short life span

IMPACT OF HEREDITARY DISORDERS ON THE FAMILY

Prenatal Testing

- Prenatal screening tests
- Prenatal diagnostic procedures

Genetic Evaluation and Counseling

- Genetic services
- Estimation of risks
- Communicating risks

Role of Nurses in Genetics

- Nursing Assessment: Applying and Integrating Genetic and Genomic Knowledge
- Identification and Referral