A genotype refers to person’s genetic heritage. For example:

- Gene for brown eyes (B)
- Gene for blue eyes (b)

Diagram showing genetic inheritance:
- Parent: B b
- Offspring: B B, B b, b b

B = Gene for brown eyes
b = Gene for blue eyes
Phenotype

• The phenotype is one’s genotype expressed in characteristics that can be observed and measured (e.g., blue eyes).

• It includes physical traits (e.g., height, weight) as well as psychological characteristics (intelligence, personality).
Genetic Principles

- **Genotype**

- **Phenotype**

**Single pair inheritance:**

- Dominant-recessive genes principle involves the ability of dominant genes to override potential influence of recessive genes.

- Recessive genes exert influence only if two genes of a pair are recessive.
Polygenic Principle of Inheritance

- Many genes interact to produce a particular characteristic.
- Most complex human behaviors and traits are influenced by many pairs of genes in interaction with the environment.
Selected Genetic Disorders

- Phenylketonuria
- Down syndrome
- Sickle-cell anemia
- Hemophilia
- Fragile X
- Klinefelter
- Turner syndrome
- Anencephaly
- Cystic fibrosis
- Spina bifida
- Tay-Sachs
Genetic Disorders.

- Some disorders are entirely hereditary and are passed on according to principles of inheritance - e.g. hemophilia, Huntington’s disease.
- Other disorders are not inherited but may result from errors during cell division, due to radiation, drugs or aging, e.g. Down syndrome.
Both parents, usually unaffected, carry a normal gene (D), which dominates its abnormal recessive counterpart (r).

Rearranged Inheritance of a Birth Defect

The odds for each child are:
1. 25% risk of inheriting a "double dose" of r genes which may cause a serious birth defect
2. 25% chance of inheriting two D's, thus being unaffected
3. 50% chance of being a carrier as both parents are

Possible hereditary results

Normal
Carrier
Carrier
Affected
Cystic fibrosis

- Cystic fibrosis is a glandular dysfunction that interferes with mucus production, breathing, and digestion and generally shortens the life span.
- It can be treated by physical therapy, oxygen therapy, synthetic enzymes, and antibiotics.
- Amniocentesis can detect CF prenatally.
- Carrier detection is by family history and DNA analysis.
Sickle-cell Anemia

- Sickle-cell anemia is a type of disorder in which sickle-shaped red blood cell limits oxygen supply to joints and organs.
- It can lead to joint swelling and heart or kidney failure.
- Groups usually affected include African and Latino.
- Sickle-cell anemia is treatable by penicillin, antibiotics, and blood transfusions.
- Prenatal diagnostic methods include CVS, amniocentesis. Carrier detection is by a blood test.
Chromosome abnormalities

Down Syndrome
Turner Syndrome
Kleinfelter Syndrome
Down Syndrome

- Extra 21st chromosome.
- Mental retardation and motor impairment.
- Physical abnormalities
- Highest risk- first birth after age 35.
- Diagnosis is by AFP, CVS, or amniocentesis, carrier detection: family history, and chromosomal analysis.
Klinefelter

• Klinefelter syndrome is a chromosomal disorder in which males have an extra “X” in the sex chromosome, resulting in the “XXY” pattern.
• Characteristics include undeveloped testes, enlarged breasts, and tall stature.
• Typically only males are affected.
• Treatment includes hormone therapy.
• Prenatal diagnosis is by CVS or amniocentesis.
Turner syndrome

• Turner syndrome is a chromosomal disorder in which a female is missing an “X” chromosome (“XO”).
• Such females are short, have webbed necks, may be mentally retarded, and sexually underdeveloped.
• Only females are affected. Hormone therapy is used to treat this syndrome.
• Although there is no prenatal diagnosis, there is a blood test for carrier detection.
Sex-Linked Disorders

- Sex-linked inheritance patterns
- Mother carries gene on X chromosome:
  - Daughters 50% chance of becoming carriers - but will not have disease – normal X chromosome protects
- Males - 50% chance of inheriting the disease (No X chromosome to mask)
Behavior Genetics

- The degree and nature of behavior’s hereditary basis.
- Twin studies - identical (monozygotic) vs. fraternal twins (dizygotic)
- Identical Twins raised apart
- Adoption studies
Genotype-Environment Interactions

- Passive interaction between heredity and the environment - genetic parents also provide the rearing environment.
- Evocative interactions: the child’s genotype elicits certain types of physical and social environments.
- Active (niche-picking) refers to the tendency of children to seek out environments that they find compatible and stimulating.
Between-group differences (cause: the soils in which the plants were grown)

Within-group differences (cause: genetic variation in the seeds)
Shared and Nonshared Environmental Influences

• Shared environmental influences include siblings common experiences such as parents’ personalities, family’s environment, SES, and the neighborhood.

• Nonshared environmental influences refer to each child’s own unique experiences (in and outside of family) that are not shared with a sibling.
A reaction range is the range of possible phenotypes for each genotype and is determined by the range within which the environment can modify the trait.
Models of Gene/Environment Interaction.

- **Gottesman’s ‘Limit-Setting Model’ (Reaction Range) (1974):** Genes establish a reaction range within which development will occur. E.g., genes have set limits on potential basketball skills in that very short individuals will not become proficient regardless of their environment. Very tall people have the greatest opportunity to benefit from a particular environment.
Conclusions About Heredity-Environment Interaction

• The relative contributions of heredity and environment operate together; they are not additive so it is not possible to say what percentage of anything is due to either heredity or environment.

• Complex behaviors may have a genetic loading that predisposes one to a particular development. But the actual development of the characteristic depends on complex interaction with the environment.
Prenatal Testing for Abnormalities

- Amniocentesis.
- Ultrasound- new more powerful methods
- Chorionic villus biopsy
- Alpha-fetoprotein (AFP)
- New types of blood work/screening