DEP 4053

Christine L. Ruva, Ph.D.

Heredity and Prenatal Development: Chapter 3

PRINCIPLES OF HEREDITARY TRANSMISSION

- Genotype
- Phenotype
- <u>Chromosomes</u>: in the nucleus of the cell store and transmit genetic information.
- **DNA** (deoxyribonucleic acid) molecules make up chromosomes.
- <u>Gene</u> is a segment of a DNA molecule which contains instructions for making proteins.
 Humans have approximately 30,000 genes (Human Genome Project, 2001).
- <u>Cell Division</u>
 - Mitosis:
 - somatic (body) cells
 - Meiosis:
 - <u>Steps of Meiosis</u>: It halves the number of chromosomes so each gamete contains 23.
 Why is this step necessary?
 - <u>Crossing over</u>: key part of first stage of meiosis
 - Why is this step necessary?

Boy or Girl?

- **<u>Fertilization</u>**: When sperm and ovum unite at conception, the resulting **zygote** has
- The 22 pairs of matching chromosomes within a human cell.
- The twenty-third pair consists of **sex chromosomes**.

PATTERNS OF GENETIC INHERITANCE

- Allele
- Homozygous
- Heterozygous

Possible Outcomes If Person Is Heterozygous For A Trait

- <u>dominant allele</u> & <u>recessive allele</u>.
 - What are the possible genotypes for the following phenotypes?
 - Blood type Phenotypes: A, B, O.
- <u>intermediate</u> (<u>blended</u>)
- <u>codominance</u>
- modifier genes

Possible Outcomes If Person Is Heterozygous For A Trait

• Complex traits follow **polygenic inheritance**

Gene Disorders Sickle Cell Anemia

- West Africa & Mediterranean descent
 - 1 in 600 African Am. are homozygous and 1 in 10 are heterozygous.
- Allele located on chromosome 11.
- Carriers of the trait are
 - Evolution
 - How environment & genes can interact

Other Gene Disorders

- <u>Phenylketonuria PKU</u>
 - environmental modifiable genetic disorder
 - mutation of gene on chromosome 12 (recessive)
 - Liver unable to produce an enzyme to break down phenylalanine.
 - Pregnant women with PKU should again reduce intake of phenylalanine ...
- Most sex or X-linked disorders occur when a recessive allele is carried on the X chromosome.
- Males are more likely to be affected Why?
- **Sperm** carry twice as many mutations as eggs, suggesting that:

Red-green color blindness

- for son to be color-blind, what must mother be?
- for daughter to be color blind what must parents be?

Hemophilia

Chromosomal Abnormalities

- Chromosomal defects occur during the process of **meiosis or mitosis** uneven segregation.
- Because these abnormalities involve more DNA than single-gene disorders, they often produce disorders with many mental and physical symptoms.

Sex Chromosome Abnormalities

- **Meiosis** of female cell should result in cells with single X chromosome, but could be XX or no X's.
- <u>Turner Syndrome</u>: If no X and fertilized by X sperm then XO.
- <u>Super female:</u> If XX and fertilized by X sperm then XXX.
- <u>Klinefelter Syndrome:</u> If XX and fertilized by Y sperm then XXY.

Chromosomal Abnormalities: Down Syndrome

• <u>Down syndrome or trisomy 21</u>

GENES AREN'T NECESSARILY = TO OUTCOMES (G vs. P) What reason can we give for are inability to predict completely from genotype phenotype?

- genotype is only a blueprint/recipe for development
- genotype only establishes a ______
- A ______ is a person's unique, genetically determined response to a range of environmental conditions. This accounts for how children respond in different ways to the same environment.
- <u>Canalization of trait</u>: is the tendency of heredity to restrict development to one or a few potential outcomes. Highly canalized traits require extreme environmental conditions to deter their genetically set outcomes.

RESEARCH ON GENE - ENVIRONMENT INTERACTION

Genetic-Environmental Correlation

- The concept of **genetic-environmental correlation** states that our genes influence the environments to which we are exposed.
- Passive and Evocative Correlation
 - In a **passive** correlation, a child has no control over the environment available to him or her. Parents create an environment compatible with their own heredity.
 - In an **evocative** correlation, a child behaves in ways consistent with his or her own heredity. The responses evoked from others will, in turn, strengthen the child's original response.
- Active Correlation
 - An **active** correlation is more common at older ages. As children extend their experiences beyond the immediate family, they choose environments that complement their genetic tendencies.
 - This tendency to actively choose environments that complement our heredity is called **niche-picking**.
 - With age, genetic factors may become more important in determining the environments we experience and choose for ourselves.

RESEARCH ON GENE - ENVIRONMENT INTERACTION

Environmental Influences on Gene Expression

- The relationship between heredity and environment is not a one-way street, from genes to environment to behavior.
- Rather, it is **bidirectional**; genes affect children's behavior and experiences, but their experiences and behavior also affect gene expression.
- Stimulation of both internal and external environments (to the child) triggers gene activity.