Genetic Influences: Terms and Patterns of Transmission

Genetics
- The field of science that studies heredity.
- The basis of initial human development depends on genetics and the genetic codes produced by the mating of two people.

Heredity
- The biological transmission of traits and characteristics from one generation to another. Inborn factors inherited.

Genetic Influences: Terms and Patterns of Transmission

Environment
- External influences, i.e., family, community, personal experiences affecting development.

Genes
- Biochemical material regulating the development of traits.
- Determines inherited characteristics.

Genetic Influences: Terms and Patterns of Transmission

Genes
- Short segments of DNA
- Packets of instructions that tell cells how to behave.
- Part of the chromosome. Contains a segment of the DNA.
- Every cell contains a segment of DNA.
• Genetic Influences: Terms and Patterns of Transmission

• Genes
  - Every cell contains an identical set of 50,000 to 100,000 genes.
  - Some are active in each cell – others are not.

• Chromosomes
  - Genetic structures found in the cell’s nucleus.

• Genetic Influences: Terms and Patterns of Transmission

• Chromosomes
  - More than 100,000 genes are distributed among the 46 chromosomes.
  - Chromosomes are threadlike structures that carry the body’s DNA.
  - Our genetic blueprint
  - Determines individual characteristics

• The Process of Fertilization

• Genetic Influences: Terms and Patterns of Transmission

• DNA
  - Chemical components that carry the genetic code.

• Zygote
  - The fertilized ovum (egg) that carries genetic messages from both parents.

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• Monozygote
  - A cell that divides into two cells. Identical twins. Incomplete division can lead to conjoined twins.

• Dizygote
  - When two ova are released and fertilized
  - in the same month. Fraternal twins.
• Genetic Influences: Terms and Patterns of Transmission

• **Recessive Traits**
  - Usually not expressed.

• **Phenotypes**
  - A set of observable characteristics of an individual or group, e.g., color.

• **Genotypes**
  - The genetic pattern carried internally.

• **PATTERNS OF GENETIC TRANSMISSION** - Mendel’s Law

• **The Law of Dominant Inheritance**
  - Expressed trait = dominant
  - Trait not revealed = recessive

• **Alleles**
  - A gene that governs alternative expressions of a characteristic. Offspring receives one from each parent.

• Genetic Transmission

• **Homozygous**
  - when both alleles are the same.

• **Heterozygous**
  - when both alleles are different.
  - in a heterozygous situation, the dominant allele is expressed.

• Genetic Transmission

• **Recessive Inheritance**
  - occurs when a homozygous organism receives the same recessive allele from each parent. The recessive trait appears.
• **Incomplete Dominance**
  - occurs when two colors are crossbred and a muted color results. Example: red and white snapdragons=pink; black and white=brown or caramel.

• **Genetic Transmission**

• **Incomplete Dominance**
  - Persons with blood type AB
  - Polygenic inheritance or the interaction of several genes.

• **Sex-Linked Inheritance**

• **Recessive genes** carried on an X chromosome.

• **Red-green color blindness** is a recessive trait that usually occurs in males.

• **Hemophilia** – when the female has one abnormal gene (x) and one normal gene (y). Impacts males most severely. Affected females=second carrier. Fathers can pass on to daughters.

• **Huntington's Disease** -> transmission from father severe.

• **What Determines Sex?**

• **Father**
  - 120 to 170 males are conceived for every 100 females.
  - **Males** more readily abort spontaneously or are stillborn.
  - Only 106 males are born for every 100 females.
  - Males are more susceptible to disorders and die earlier in life.

• **Genetic Abnormalities**
• Birth defects can be caused by sex-linked abnormalities in the chromosomes.

• Can also be caused by hormonal imbalances and other chemical imbalances. Also environmental.

• More than 4,200 diseases caused by defects in genes have been identified.

• Genetic Abnormalities

• Not all are seen at birth.

• Tay-Sachs, Sickle–Cell Anemia may not appear until 6 months or later.

• Cystic Fibrosis may not appear until age 4.

• Glaucoma and Huntington’s not until late 30s, 40s, or 50s.

• Dominant Gene Transmission

• Abnormal traits can be carried by dominant genes.

• When a mother has one normal recessive gene and one abnormal dominant gene and the father has 2 normal genes, each offspring has a 50/50 chance of inheriting the abnormal gene from the mother and having a defect.

• Dominant Gene Transmission

• Two dominant gene disorders are: achondroplasia (a type of dwarfism) and Huntington’s disease.

• Recessive Gene Transmission

• Recessive gene transmission often fatal in infancy.

• Examples: Tay-Sachs – a degenerative disease of the central nervous system.
- **PKU or (Phenylketonuria)** – an enzyme disorder transmitted by a recessive gene. Babies can not metabolize the protein phenylalanine. Damages CNS.

- Recessive Gene Transmission
  - Hemophilia
  - Sickle-Cell
  - Anemia
  - Color –blindness
  - Cystic fibrosis
  - Recessive traits appear when an offspring receives the same recessive gene from each parent.

**Recessive Gene Transmission**

- When only one parent has the faulty recessive gene, no offspring will be defective. They can be carriers of the recessive gene.

- Each child has a 50/50 chance of being a carrier like the parent and of passing the recessive gene on to their offspring.

**Recessive Gene Transmission**

- When both parents carry a recessive gene they are capable of passing it on to their offspring. The offspring has a 25 percent chance of a birth defect and a 50 percent chance of being a carrier.

- Sex-Linked Characteristics
  - Hemophilia
• Carried on one of the X chromosomes of an unaffected mother or father.

• Sex-linked recessive traits almost always appear in male children. Males have a 50 percent chance of inheriting the abnormal X chromosome and a 50 percent chance of not.

• **Sex-Linked Characteristics**

• An affected father cannot pass the gene on to his sons since he contributes the Y chromosome, but can pass it on to their daughters who become carriers and pass it on to their male offspring.

• **Chromosomal Abnormalities**

• Many are inherited.

• **Klinefelter’s Syndrome**

• **Turner’s Syndrome**

• **XYY and XXX Syndrome**

• **Down Syndrome**

• **Fragile X Syndrome**

• **Alternative Methods of Conception**

• **Artificial Insemination**
  – Female receives injections of husband’s (or donor’s) sperm into cervix.

• **In-Vitro Fertilization**
  – Egg and sperm (fresh or frozen) is fertilized in a petri dish/incubator and is then implanted as an embryo into the mother’s uterus. The first baby conceived through IVF was Louise Brown in 1978.

• **GIFT (Gamete Intra-Fallopian Transfer)**
  – Sperm and unfertilized eggs are inserted together in the fallopian tube via a laparoscopy to become fertilized.

• **ZIFT (Zygote Intra Fallopian Transfer)**
Sperm and unfertilized eggs are fertilized in a petri dish/incubator and then the zygote is injected into the fallopian tubes.

- **Frozen Embryo Transfer**
  - Egg and sperm is fertilized and matures in a petri dish/incubator and develops into an embryo. The embryo is frozen until such time the couple wishes to conceive. The embryo is then transferred into the uterus.

- **Donated Eggs**
  - An ovum is taken from a fertile woman and donated to another woman to enable conception.

- **Surrogate Motherhood**
  - Another woman or female relative is inseminated and carried the fetus to term until birth, usually under a contractual agreement.

- **Preimplantation Genetic Diagnosis**
  - A new prenatal technique. Can identify genetic defects in embryos of from 4 to 8 cells, which were conceived by in vitro fertilization. Can detect such diseases as cystic fibrosis. Defective embryos are not implanted.

- **Intracytoplasmic Sperm Injection (ICSI)**
  - A form of artificial insemination that involves injecting an individual sperm directly into an egg in a laboratory dish.
  - This procedure has raised concern that boys born to fathers who use the technique may inherit their father’s infertility.

- **Genetic Engineering**
  - The passive or active attempt to systematically control the regeneration of organisms.

**Examples of Techniques:**

- **Eugenics**
  Selective breeding. Phenotypic characteristics are changed by selectively mating parent to obtain desired genes.

- **Parthenogenesis**
  Process where a single cell is produced without fertilization (and produces a new organism).

- **Cloning**
  Process where an ordinary somatic nucleus is stimulated to produce a new individual.

- **Euphenics**
  The changing of the phenotype by altering the early developmental process.
• Genetic Engineering
  • Gene Therapy
    – The capability to eradicate hereditary and incurable diseases, to improve physical characteristics, to make those improvements permanent in future generations and to mold the overall characteristics of the human species. (Currently being performed at NIH.)
    
    – Somatic Cell Gene Therapy
      (non reproductive cells). Insertion of a gene to correct a genetic defect. Example: Sickle-cell anemia, hemophilia.

    – Germ-line Therapy
      Besides genetic defects, could treat other serious diseases including cancer and heart disease.

  • Enhancement Engineering
    – The insertion of the gene into the reproductive tissue of the patient in such a way that the disorder in his or her offspring would also be corrected.
    – Performed in animal experiments. Also on clinical trials on humans.

  • Eugenic Genetic Engineering
    – The attempt to alter or “improve” complex human traits, each of which is coded by a large number of genes.
    – Examples: personality, intelligence, character, formation of body organs, et.
    – Currently not feasible.

• Questions About Genetic Engineering
  • Is genetic engineering morally proper?
  • Who would decide which genes to alter?
  • How would one distinguish between a serious disease and a minor disease?
  • Is it normal to be shorter than 5 feet? Should a person be given a gene to make him taller?
  • What if a gene could be found that affects memory? Should the gene be given to people who are already intelligent to make them more intelligent? Or, to people with low intelligence? Normal intelligence?
• Prenatal Assessment and Influences

• Assessment Techniques

• Prenatal Diagnosis of Birth Defects: Techniques/Procedures
  • **Amniocentesis**
    — Drawing of sample of amniotic fluid (16th to 18th week).
  • **Chorionic Villus Sampling**
    — A culture of tissue from around the membrane of the embryo (9th to 12th week).
  • **Alpha Fetoprotein Test (AFP)**
    — Blood test—analyzed for AFP levels (16th to 18th week).
  • **Ultrasound**
    — High frequency sound waves directed into abdomen of pregnant woman to yield sonogram—a picture of uterus, fetus and placenta.
  • **Umbilical Cord Assessment**
    — Insertion of needle in umbilical cord with use of ultrasound to draw fetal blood.
  • **Fetoscopy or Embryoscopy**
    — Insertion of lighted lens into uterus to look at fetus parts and determine abnormalities.

• Prenatal Development

• Stages of Development

• *Environmental Influences*

• *Maternal and Paternal Factors*

• *Environmental Influences*

• *Infertility*
• *Sperm Count & Quality*
• *Chemicals*
• **Foods**
• **Alcohol**
• **Drugs**
• **Pesticides**
• **Illness and Disease**
• **Radiation**

• NO DRUGS!
• Many harmful drugs to fetus:
  • Antibiotics (Streptomycin & tetracycline)
  • Sulfonamides
  • Vitamins A, B, C.D. & K, in excess.
  • Barbiturates, opiates, certain hormones and Accutane.
  • Aspirin & Birth Control Pills
  • DES, caffeine, nicotine, alcohol, marijuana, cocaine
• **Harmful Illnesses**
  • Rubella, infections, diabetes, TB, Syphilis, AIDS, Blood type-RH positive vs. RH negative

• **Environmental Hazards**
• **Industrial chemicals**
• **Lead contamination**
• **Radiation**
• Video Display terminals
• Polluted Air/Water
• Paternal Transmissions
• Exposure to lead, drugs, nicotine, alcohol, radiation, DES, pesticides, and more.
• Exposure to electromagnetic fields
• Fathers who smoke
• Father’s age
• Critical Factors
• For Ideal Prenatal Development
• Good Nutrition is Important!
• Breakfast important and essential.
• Need an extra 300 to 500 calories daily and 30 more grams of protein.
• Malnutrition impacts fetus.

• Prenatal Abilities
• Hearing and Learning

• Prenatal Abilities
• Fetal Hearing
  — Responds to bells and vibrations
  — Can discriminate between different strokes.
  — Fetuses can blink at sounds.
• Fetal Learning
— Can remember and discriminate what they have heard while in the womb.
— Can develop preferences for the kinds of sounds they hear before birth—particularly for mother’s voice.
— Can remember stories, lessons recited to them while in the womb.

• **Development of Embryo and Fetus**

• **Month to Month**

• **Critical Questions**
  • Do you think you would be a better parent if you could choose whether your child would be a girl or boy?
  • In what ways is sex selection by adoption different from sex selection by sperm manipulation?
  • If you were expecting a baby how would you decide which kinds of interventions might make your delivery as satisfactory as possible?
  • What information would you need to make choices?