Transmission Genetics

What is Transmission Genetics?

- Transmission Genetics is the study of traits as they are passed from generation to generation within families.
- Phenotype is the observation of a particular characteristic.
- In Transmission Genetics data collected is based on phenotype.

Human Pedigree Analysis

- Transmission Genetics can be visualized in a Human Pedigree Chart.
- The Human Pedigree Chart is an orderly presentation of a family’s information.
- The use of symbols and lines enable the passage of a trait within family members to be easily noticed and analyzed.

Human Pedigree Analysis

- Human Pedigree Charts may not be simple to compile. Data may be missing.
- Families may be reluctant to provide personal information.
- Information about some family members may not be possible to obtain due to death or separation or privacy issues.
Symbols are Used

The Symbols vital to constructing a Human Pedigree are found in your text on page 60.

What do the following symbols represent?

- [ ]
- [ ]
- [ ]
- [ ]

Traits and Genes

A Trait is a characteristic observed in an individual. It may be obvious, like your height or eye color. It may be deducible by medical testing or your state of being. Your are alive, therefore you must make Acid Maltase (an vital enzyme).

A Gene is the actual DNA sequence you possess, your Genotype. Order of ATGC s.

Chromatin Strands

- Sex Chromatin strands or Sex chromosomes are called X or Y.
- Human Female XX Human Male XY
- Autosomes are chromatin strands or chromosomes that are indicated by numbers. e.g. 1-22
- Genetic conditions can be caused by genes located on autosomes or sex chromatin strands

Gregor Mendel

- Czechoslovakian by birth
- Benedictine monk in Austria
- Studied inheritance in Pea Plants
- Analyzed the statistical results of thousands of plants.
- Established the Principles of Inheritance
- Known as the “Father of Modern Genetics”
Transmission Genetics

➢ Mendelian Inheritance was based on phenotype (observation).
➢ Mendel used letters, not geometric symbols with shadings and arrows.
➢ Mendel used the information collected to predict the possible traits in the next generation, not to explain the current family members.

Principles of Inheritance

I. Principle of Unit Factors
   In every organism there are single factors causing their features.

Current terminology

I. Genes = specific sequences of DNA nucleotides

Principles of Inheritance

II. Principle of Dominance
   In an organism some of the factors are seen, some are hidden. Dominant and Recessive

Current terminology

II. Dominant Genes - functional alleles
   recessive genes - non-functional alleles
   Alleles - different versions of the same gene.

Using Letters for Genes

➢ Dominant (Functional) Genes are written in uppercase letters.
   Melanin gene = M Pepsin gene = P
   Free ear lobe gene = F Polydactyly = P

➢ recessive (nonfunctional or dysfunctional) genes are written in lowercase letters.
   m = mutated melanin gene
   f = mutated free ear lobe gene
   p = nonfunctional polydactyly gene
Recessive

➢ Recessive conditions are due to the presence of nonfunctional or dysfunctional genes. \( mm = \) albinism
➢ A person needs two copies of the nonfunctional allele to have the condition.
➢ Examples: albinism, attached earlobes

Dominance

➢ Dominant conditions are caused by functional genes. \( Pp = \) polydactyly
➢ Only one copy of the gene is needed to cause the trait.
➢ Examples: Tongue rolling, hitchhiker thumb

Codominance - Multiple Alleles

➢ Genes may change or mutate. Different DNA sequences for a trait may exist.
➢ Co-Dominance means that all versions of the gene are functioning and contribute equally to the phenotype.
➢ This is also known as "Multiple Alleles".
➢ Allele means "the other", as in "other version".
➢ Examples: antigen type (AB), Sickle Cell Trait (HbB HbS)

Sex-linked

➢ Sex-linked traits are due to genes located on the X or Y chromatin.
➢ Genes on the X or Y chromatin strands may be recessive or dominant.
➢ The gender of the person affects the chance of experiencing a sex-linked trait.
➢ Examples: Red-Green Colorblindness, Hemophilia A
Pattern of Inheritance?

I
II
III

Recessive, Dominant, Sex-linked, Other?

Pattern of Inheritance?

I II III IV

Pattern of Inheritance?

I II III IV

Pattern of Inheritance?

I II III IV

Recessive, Dominant, Sex-linked, Other?
Case Study - Draw Pedigree Chart

1. Proband = Alexis, the only son of Czar Nicholas II & Czarina Alexandria of Russia has Hemophilia A.
2. Neither Nicholas nor Alexandria have Hemophilia A.
3. None of Alexis four older sisters have the condition.
4. His Grandparents Prince Albert & Queen Victoria of England do not have Hemophilia. They are 1st cousins
5. Albert & Victoria have 9 children in the following order: daughter, son, daughter, son, 2 daughters, 2 sons, one daughter.
6. Alexis’ Uncle Leopold, Duke of Albany, their last son has hemophilia.

Case Study - Draw Pedigree Chart

1. Parents seek genetic counseling when their third child, a boy (the proband), is born with a genetic condition.
2. Neither parents nor grandparents have the trait.
3. Father has a younger sister and brother without the condition.
4. Mother has one older brother without trait. His second daughter has the same genetic condition.

A Case Study will be on your Exam.
Value = 5%

Human Pedigree Chart Analysis

I will give you a Pedigree Chart. You will be asked to review the chart and answer questions based on what the Pedigree Chart shows you.