

Genetics Lecture 7

1. Human Genetics and Mendelian Inheritance

Although humans are difficult subjects for genetic studies because:

- Long generation time
- Few offspring per family
- Controlled breeding experiments are impossible/unethical

Mendelian laws still form the basis of human genetics.

2. Pedigree Analysis (VERY IMPORTANT)

A pedigree is a family tree used to trace inheritance of traits through generations.

-Used for:

- Identifying inheritance patterns
- Predicting risk in future children
- Diagnosing genetic diseases
- Family counseling

-Standard Pedigree Rules:

- Each horizontal row = one generation
- Older generations on top
- Generations labeled with Roman numerals
- Individuals labeled with Arabic numbers
- Siblings arranged oldest to youngest
- Male usually left, female right

3. Important Pedigree

Symbols

Square = Male

Circle = Female

Filled = Affected

Empty = Unaffected

Half-filled = Carrier

Slash = Dead

Arrow = Proband

4. Important Genetic Terms

Gene = Basic unit of inheritance

Locus = Location of gene

Allele = Different form of gene

Genotype = AA, Aa, aa

Phenotype = Observable trait

Homozygous = AA or aa

Heterozygous = Aa

Compound heterozygote = two different mutant alleles

5. Chromosomal Location of

Traits

Autosomal = chromosomes 1-22

X-linked = X chromosome

Y-linked = Y chromosome

6. Mendel's Laws

Law of Segregation:

Two alleles separate during meiosis.

Law of Independent Assortment:

Genes on different chromosomes assort independently.

Exception: linked genes.

7. Dominant vs

Recessive

Dominant:
Aa or AA affected
Recessive:
aa affected
Aa carrier

8.Codominance

Both alleles expressed equally.
Example: AB blood group.

9.X-linked Inheritance

X-linked dominant:
One mutant allele enough.

X-linked recessive:
Male XaY affected.
Female XaXa usually affected.
Female XAXa carrier.

Hemizygous:
Male has one X-linked allele.

10.Autosomal Dominant Features

- Vertical transmission
- Both sexes equally affected
- Both sexes transmit
- Heterozygotes common
- Variable expressivity
- Variable age of onset
- New mutations possible
- Structural protein defect common

11.Autosomal Dominant Probability

Aa x aa = 50% affected
Aa x Aa = 75% affected

12.Examples of Autosomal Dominant Diseases

Familial Hypercholesterolemia
Huntington disease
NF1
Marfan syndrome
Achondroplasia
Myotonic dystrophy

13.Autosomal Recessive Features

- Horizontal transmission
- May skip generations
- Both sexes equally affected
- Both sexes transmit
- Consanguinity common
- Enzyme deficiency common

14.Autosomal Recessive Probability

Aa x Aa:
25% aa affected
50% Aa carrier
25% AA normal

Unaffected child chance to be carrier = 2/3

15.Examples of Autosomal Recessive Diseases

Cystic fibrosis
Sickle cell disease
Tay-Sachs disease
Phenylketonuria (PKU)

16.Consanguinity

Marriage between relatives increases recessive disease risk.

17.Variable Expressivity vs Penetrance

-Variable expressivity:
Severity differs

-Penetrance:
Mutation present but no symptoms

18.Pleiotropy

One gene affects multiple systems.
Example: Marfan syndrome

19.High Yield Points

Vertical transmission = AD
Horizontal transmission = AR
2/3 rule important
Male with X-linked recessive = hemizygous
Huntington = late onset AD
Marfan = pleiotropy
NF1 = variable expressivity
Achondroplasia = de novo mutation
CF = chloride channel defect
Sickle trait protects from malaria

20.

Autosomal Dominant:

- Every generation
- Vertical
- Structural proteins
- Rare consanguinity

Autosomal Recessive:

- May skip generations
- Horizontal
- Enzyme defects
- Common consanguinity