



Lecture 8

Inheritance patterns

Patterns of inheritance Definition:

Its the **manner** in which a particular **genetic trait** or **disorder** is passed from one generation to the next.

Modes of Inheritance:

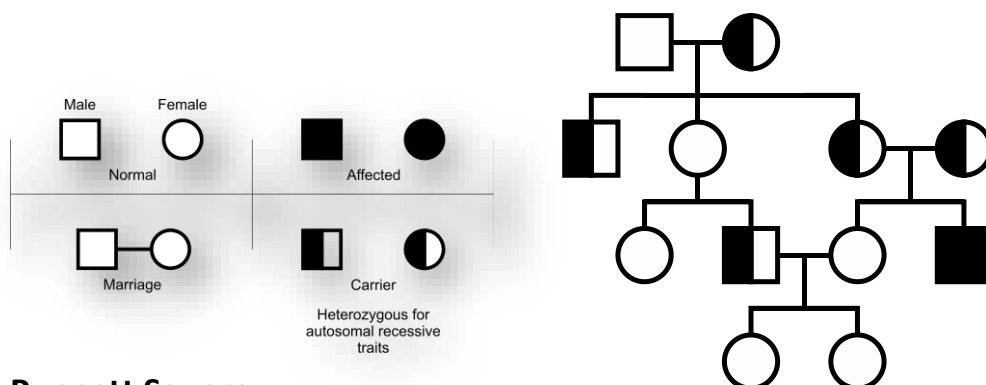
1. **Single Gene or Mendelian** ✓
2. Multifactorial
3. Mitochondrial

Important definitions:

Gene	A section of DNA that occupies a specific location on a chromosome and codes for a protein product(trait)
Locus	The position of a gene on a chromosome.
Allele	Different versions of the same gene are called alleles(one from each parent).
Genotypes	Genotype is the genetic make-up (alleles) of an individual organism
Phenotype	Phenotype is the <u>observable</u> physical or biochemical characteristics of an individual organism.
Heterozygous	having two different alleles at a given gene locus=When an individual's two alleles for a gene are different, he or she is said to be heterozygous for that gene.
Homozygous	having identical alleles at a given gene locus.= When an individual's two alleles for a gene are the same, he or she is said to be homozygous for that gene.
Dominant	Traits that are manifest in individuals with just one copy of the allele (heterozygotes)
Recessive	Traits that are only manifest in individuals who have two copies of the allele (homozygous)
Autosomal inheritance	A trait or disorder that is determined by a gene on an autosome (non-sex chromosomes)
sex- linked inheritance	A trait or disorder determined by a gene on one of the sex chromosomes
Trait	In genetics, a feature of a living thing is called a " <u>trait</u> ".

Important definition: Pedigree:

A pedigree is a chart of the **genetic history** of family over several generations.



Punnett Square:

- **Punnett invented a technique for the prediction of genotypes.**
- The Punnett square is a way of representing the genotypes of the parental gametes and all the possible offspring they produce.
- Making a Punnett Square Is One Way to Determine Likely Outcome of Genetic Cross

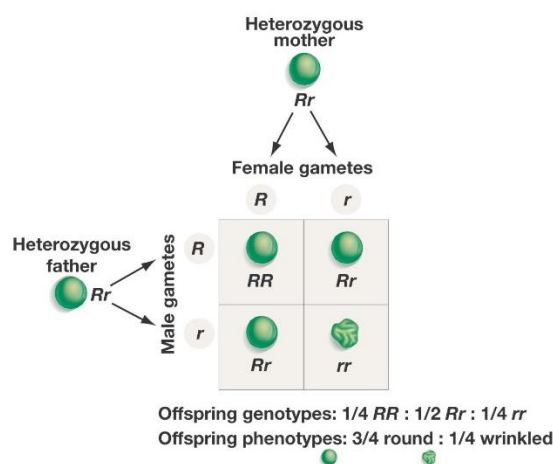
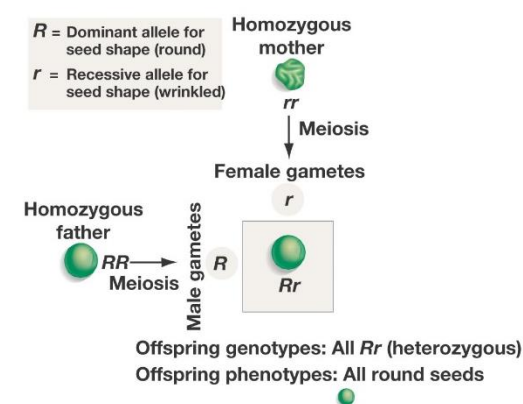
Mendelian disorders:

- **Single gene** inheritance is also referred to as Mendelian inheritance.
- More than 16,000 traits or disorders in humans exhibit single gene or mendelian inheritance.
- Gregor Mendel: Austrian botanist (1822-84)
- Mendel studied hereditary traits in pea plants.

Mendelian inheritance

There are four types of Mendelian inheritance patterns

1. Autosomal inheritance (Dominant)
2. Autosomal inheritance (Recessive)
 1. Sex-linked inheritance(Dominant)
3. Sex-linked inheritance(Recessive)





Autosomal Dominant:

Autosomal Dominant Inheritance:

- Dominant conditions are **expressed** in individuals who have just **one copy** of the allele (i.e. expressed in heterozygous state).
- When one parent is affected (het.) and the other parent is unaffected, approx. 1/2 of the offspring will be affected.

Punnett's square showing possible gamete combinations for an autosomal dominant allele 'A'

The offspring have 50% chance to have the disease (Aa).

50% of offspring will be normal (aa)

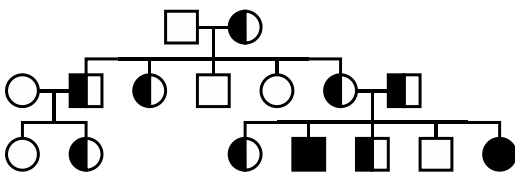
		Normal Parent	
		a	a
Affected Parent	A	Aa	Aa
	a	aa	aa

2. Autosomal Recessive:

Autosomal Recessive Inheritance:

Recessive conditions are clinically manifest only when an individual has **two copies** of the mutant allele (homozygous).

- When just one copy of the mutant allele is present, an individual is a **carrier** of the mutation, but does not develop the condition.
- **Both** male and female are **affected**.
- **Both** male and female can **transmit** the disease
-



Punnett's square showing possible gamete combinations for an autosomal recessive allele 'a'

When both parents are heterozygous, approx. 1/4 of the progeny will be affected.



		Carrier Parent	
		A	a
Carrier	A	AA	Aa
	a	Aa	aa

Example → Sickle cell anemia:

- This is an example of sickle cell for a woman who is *heterozygous* for the disease and a man who is also *heterozygous*.
- Sickle cell anemia is recessive (s)
- Normal hemoglobin is dominant (S)

Sex-Linked inheritance:

X-linked recessive inheritance:

- Mutant genes are on the X chromosome
- All men who inherit the mutation are affected (only one X chromosome)
- Women must inherit 2 mutated copies to be affected.
- More males than females are affected.
- It is never passed from father to son.
- Affected sons are usually born to unaffected mothers, thus the trait skips generations.
- Approximately 1/2 of carrier mothers' sons are affected.
- All daughters of affected fathers are carriers.
- Only males are affected
- Transmission through unaffected females to male
- No male to male transmission

X-linked dominant inheritance:

- Mutations only in X chromosome.
- Both males and females are affected.
- Affected males transmit the disease to their daughters (male to female only).
- Affected females transmit the disease to daughters and sons
- More females than males
- All daughters of affected males are affected, but no sons.
- A child of an affected female has 50% risk of disease.



Rules of Inheritance X-Linked Dominant:

- Both males and females are affected; often more females than males are affected.
- Does not skip generations.
 - Affected sons must have an affected mother;
 - Affected daughters must have either an affected mother or an affected father.
- Affected fathers will pass the trait on to all their daughters
- Affected mothers if heterozygous will pass the trait on to 1/2 of their sons and 1/2 of their daughters.

Y-linked inheritance:

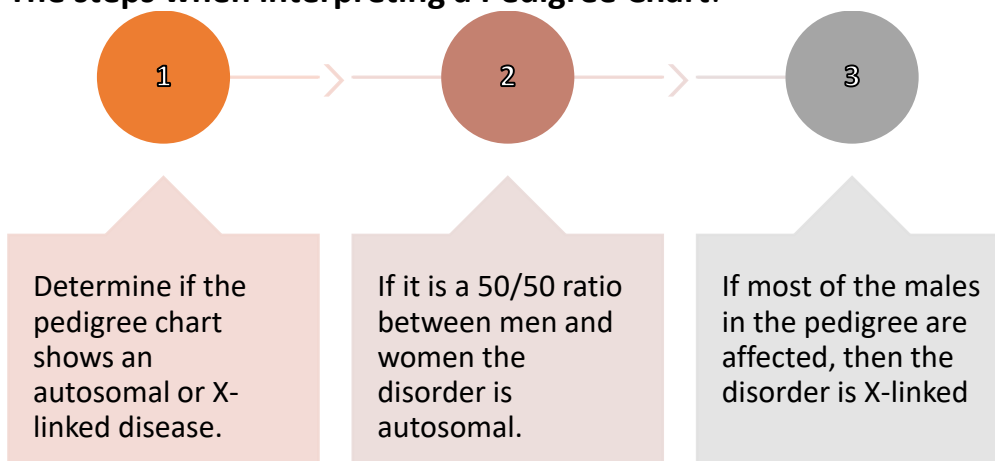
- Affects only males
- Affected males always have affected fathers
- All sons of an affected male are affected
 - Only males are affected
 - It is passed from father to all sons
 - It does not skip generations

Mitochondrial inheritance:

Rules of Inheritance Mitochondrial:

- Trait is inherited **from mother only**.
- All **children** of a mother are affected.

The steps when interpreting a Pedigree Chart:



Interpreting a Pedigree Chart:

Determine whether the disorder is **dominant or recessive**.

- If the disorder is dominant, one of the parents must have the disorder.
- If the disorder is recessive, neither parent has to have the disorder because they can be heterozygous.