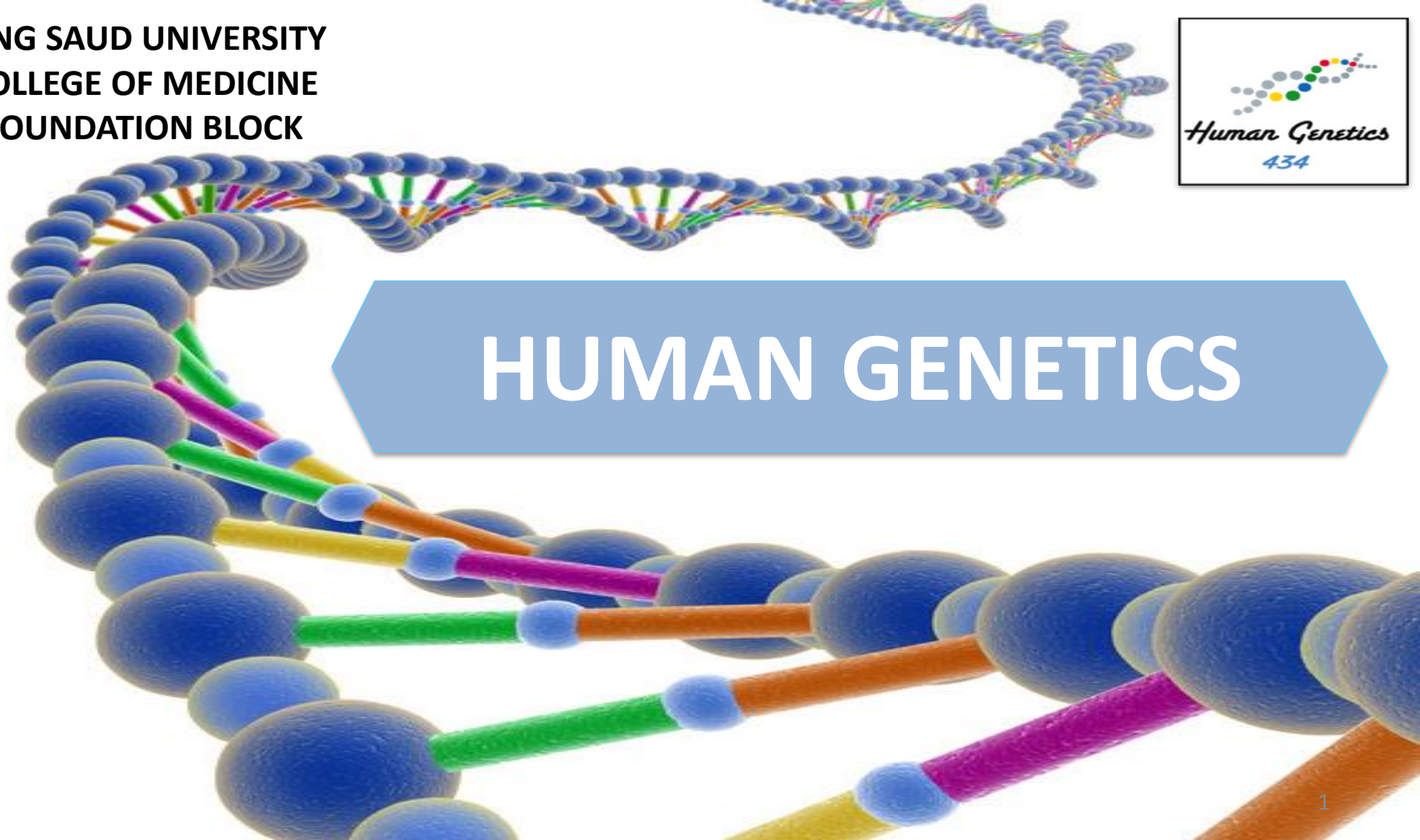
KING SAUD UNIVERSITY
COLLEGE OF MEDICINE
FOUNDATION BLOCK

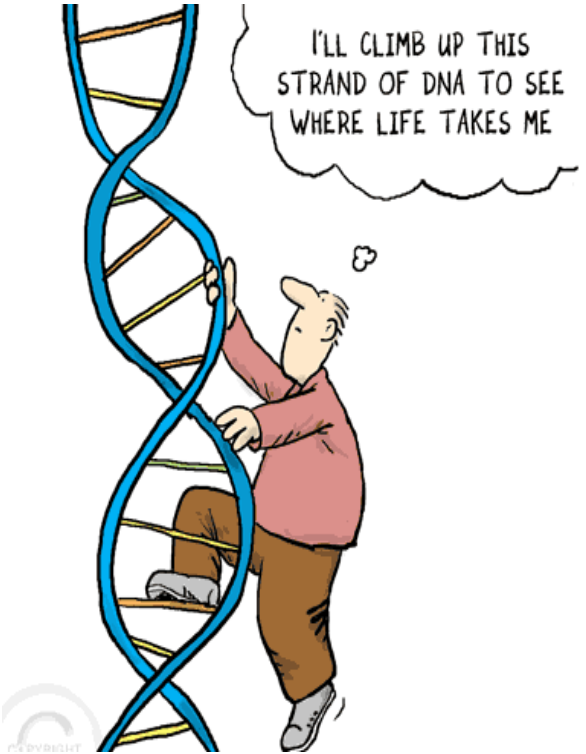
Human Genetics
434



HUMAN GENETICS

OBJECTIVES :

- Asses Mendel's laws of inheritance
- Understand the bases of Mendelian inheritance
- Define various patterns of single gene inheritance using family pedigree and Punnett's squares



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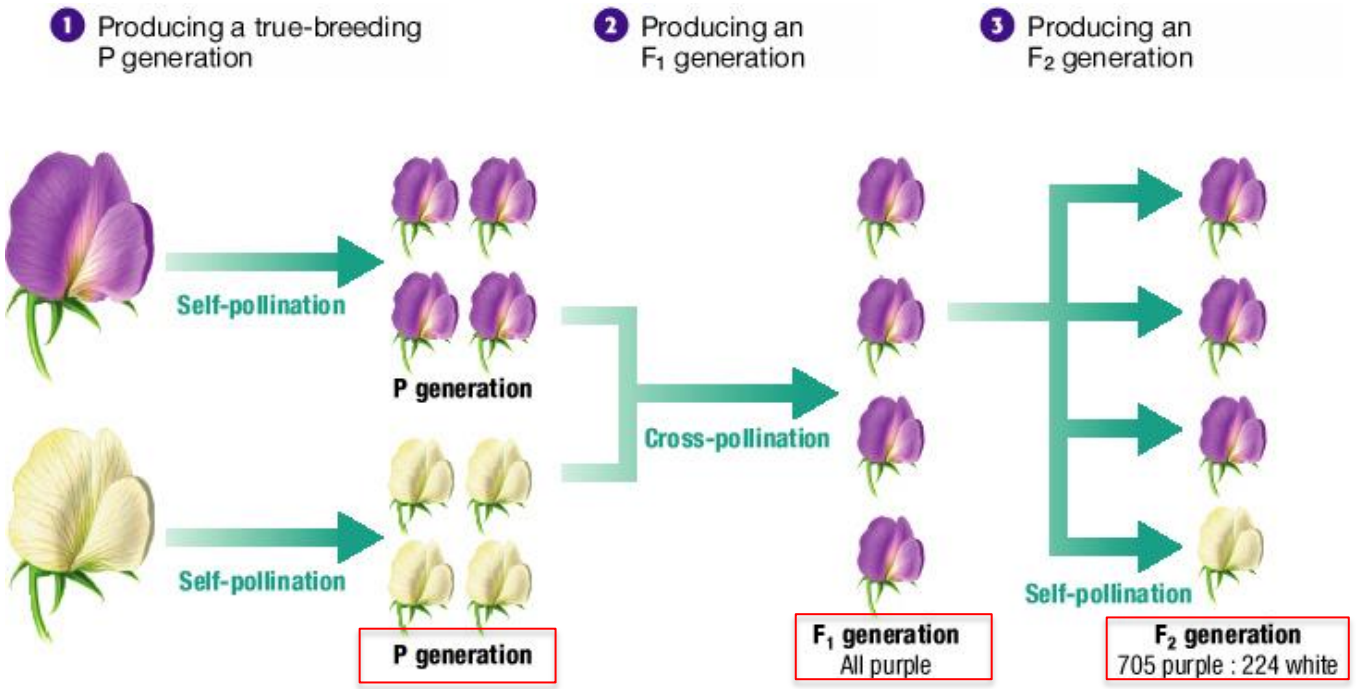
MENDELIAN INHERITANCE



Three Steps of Mendel's Experiments

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Mendel's breeding experiments results:

- The plant characteristics being studied were each controlled by factors (traits(1)) that occur in pairs. one of which was inherited from each parent.
- The pure-bred plants, with two identical genes, used in the initial cross would now be referred to as homozygous.
- The hybrid F1 plants, each of which has one gene for tallness and one for shortness, would be referred to as heterozygous (different genes).
- The genes responsible for these contrasting characteristics are referred to as *allelomorphs*, or alleles for short.

Further Explanation:

An individual inherits two alleles for each gene(**character)**, one from each parent. If the two alleles are the same, the individual is homozygous for that gene. If the alleles are different, the individual is heterozygous.

(1) a genetically determined characteristic.

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GENOTYPES AND PHENOTYPES

- If the two alleles differ, one of them will be Dominant, and the other is Recessive.

Homozygous(2) dominant **(TT)** — (2) Homo=the same

Homozygous(3) recessive (**tt**) — (3) Hetero=different

Heterozygous(4) (**Tt**) — (4) **COMPLETE DOMINANCE** - one allele is dominant to another allele

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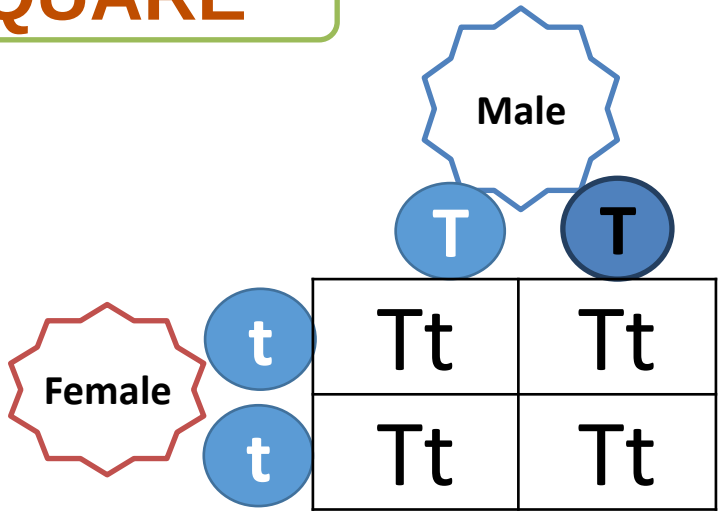
PUNNETT SQUARE



Each parent can only contribute one allele per gene. ✧

✧ These genes are found on the chromosomes carried in **the sex cells**.

✧ Offspring will inherit two alleles to express that gene.

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RECALL MENDEL'S 1st EXPERIMENTS:

CROSS: Purebred purple female x White male (**P1 generation** = **PP** x **pp**)

Result: F1 generation

- **Genotypic ratio**: 1 **Pp** (heterozygous dominant)
- **Phenotypic ratio**: 1 **Purple**

IMPORTANT

Mendel's **Law of Dominance** states that recessive alleles will always be masked by **dominant** alleles. Therefore, a cross between a homozygous **dominant** and a homozygous recessive will always express the **dominant** phenotype, while still having a heterozygous genotype.

F2 generation are:
25% homozygous dominant
50% heterozygous
25% homozygous recessive.

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MENDEL'S FIRST LAW : law of segregation

CROSS: Two F1 generation offspring with each other. (**P1 generation = Pp x Pp)**

Result: F2 generation

- **Genotypic ratio: 1PP:2Pp:1pp**
- **Phenotypic ratio 3Purple:1white**

In the **monohybrid** cross (mating of two organisms that differ in only one character), one version disappeared and will reappear in later generation..

IMPORTANT

- Mendel's experiments that followed the inheritance of flower color or other characters focused on only a single character via monohybrid crosses (law of segregation)
- He conducted other experiments in which he followed the inheritance of two different characters (a dihybrid cross) (law of independent assortment)

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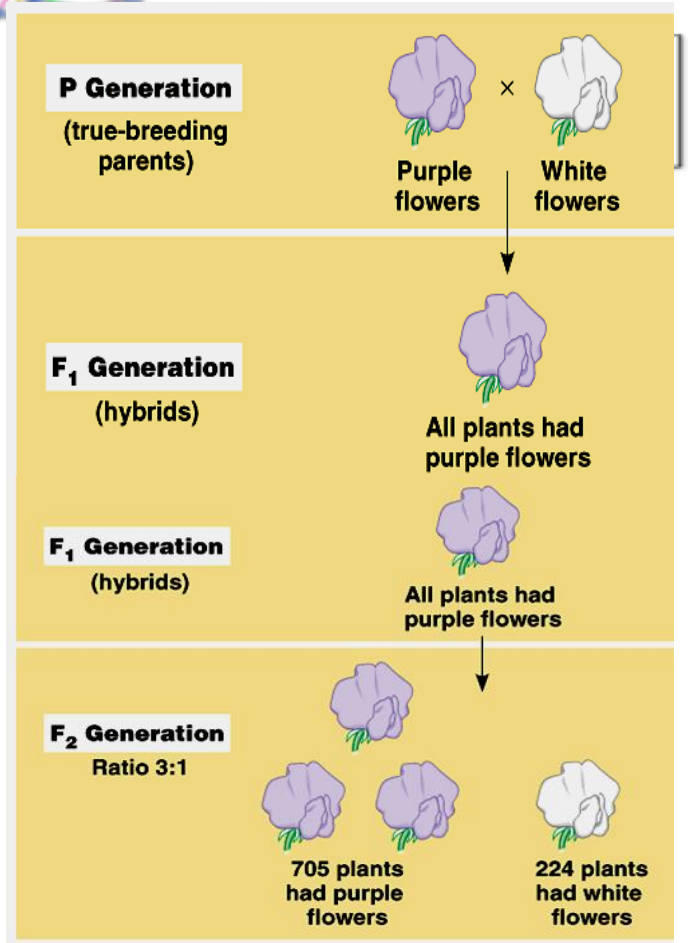
RECALL MENDEL'S 2nd EXPERIMENTS:

- The two alleles for a gene (character) are isolated into separate gametes (The genes determine the organism's traits, and are inherited from its parents. As the pair of chromosomes separate, each gamete only receives one of each allele)

- Mendel also noted that alleles of a gene could be either **dominant** or **recessive**.

Rare Exception to this rule: Occur when two allelic genes fail to separate because of **chromosome non-disjunction** at the first meiotic division.



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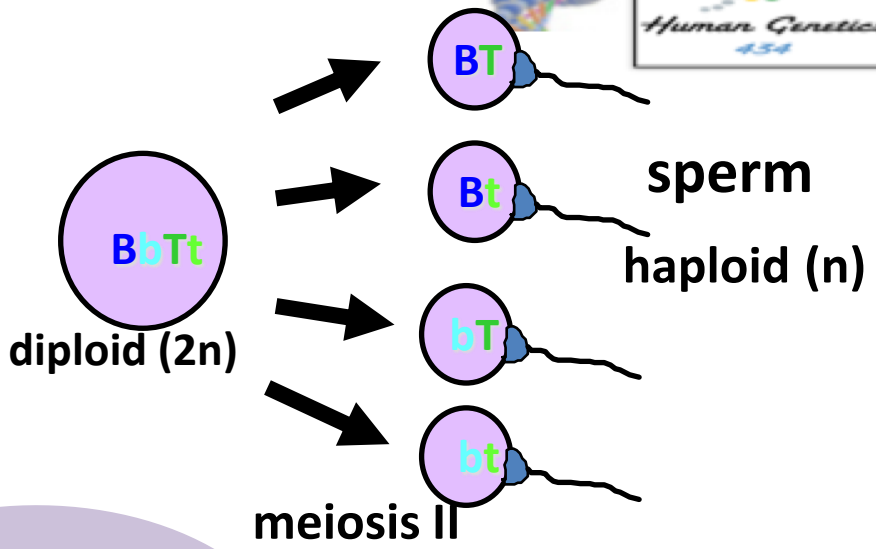
MENDEL'S SECOND LAW : law of independent assortment

- Alleles of different genes separate independently of one another when gametes are formed. (the principle state that when two or more characteristics are inherited, individual hereditary factors assort independently during gamete production, giving different traits an equal opportunity of occurring together.)

- In dihybird crosses you will see more combinations of the two genes



This is not always true as genes that are close together on the same chromosome tend to be inherited together "they **are linked**".

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MENDEL'S SECOND LAW :
law of independent assortment Continue..

Phenotypic ratio:

9 round, green: 3 round, yellow: 3 wrinkled, green: 1 wrinkled, yellow

→ **(9:3:3:1)**

Genotypic ratio:

1 RRGG: 2 RRGg: 2 RrGG: 4 RrGg: 1 RRgg: 2 Rrgg: 2 rrGg: 1 rrGG: 1 rrgg

Male Gametes

Female Gametes

	RG	Rg	rG	rg
RG	RRGG	RRGg	RrGG	RrGg
Rg	RRGg	RRgg	RrGg	Rrgg
rG	RrGG	RrGg	rrGG	rrGg
rg	RrGg	Rrgg	rrGg	rrgg

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MENDELIAN INHERITANCE (single gene inheritance)

- Over 11,000 traits/disorders in humans exhibit single gene unifactorial or Mendelian inheritance.
- A trait or disorder that is determined by a gene on an autosome is said to show autosomal inheritance
- A trait or disorder determined by a gene on one of the sex chromosomes is said to show sex-linked inheritance.

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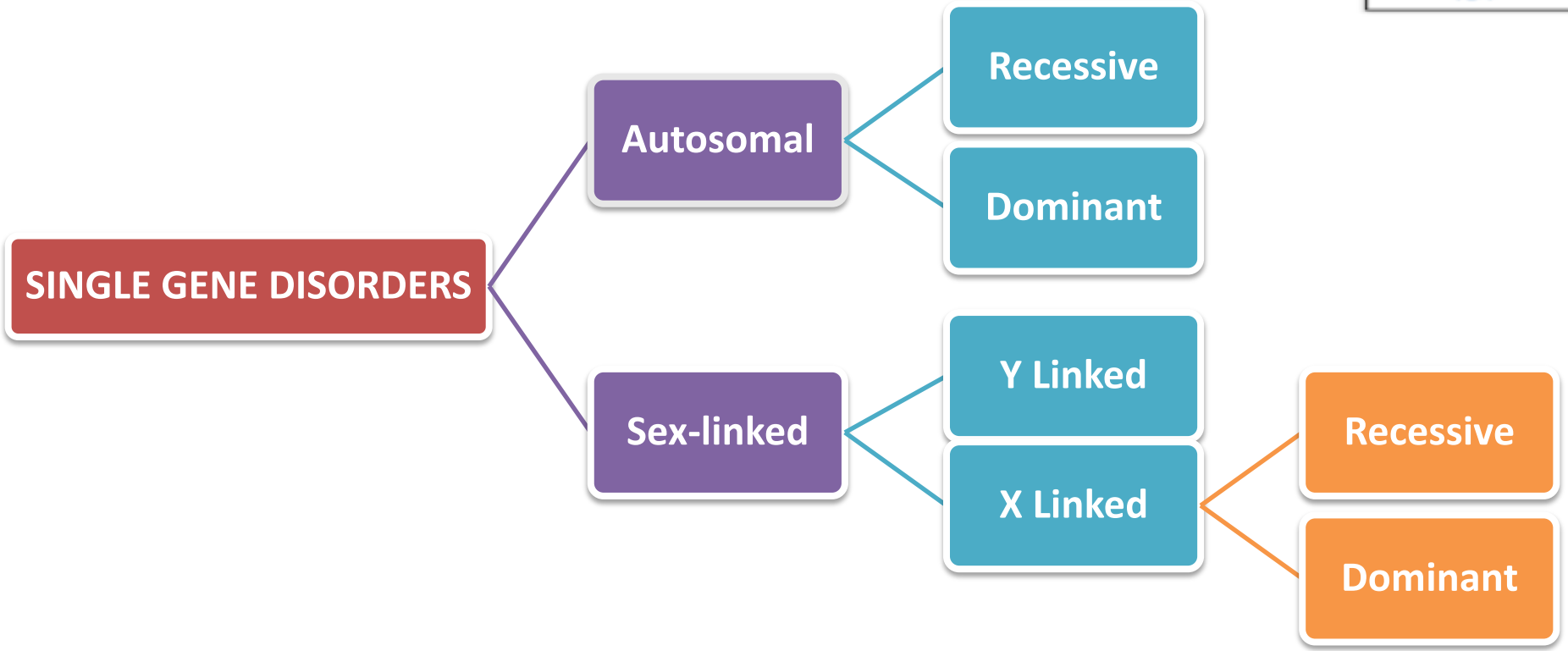
SIMPLE PATTERN OF INHERITANCE (Mode of Inheritance)



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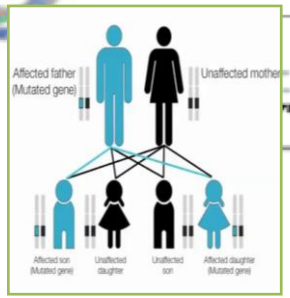
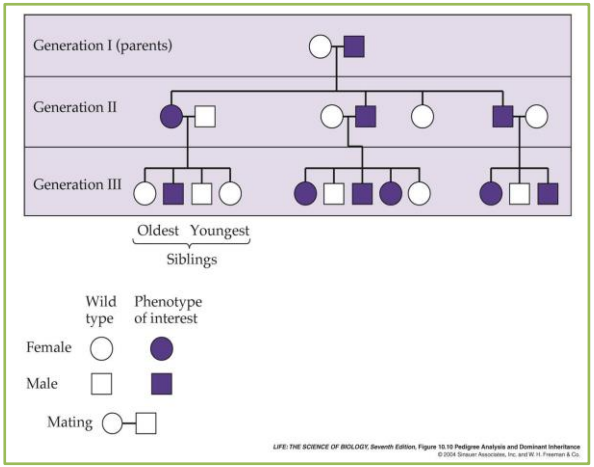
AUTOSOMAL DOMINANT MODE OF INHERITANCE:

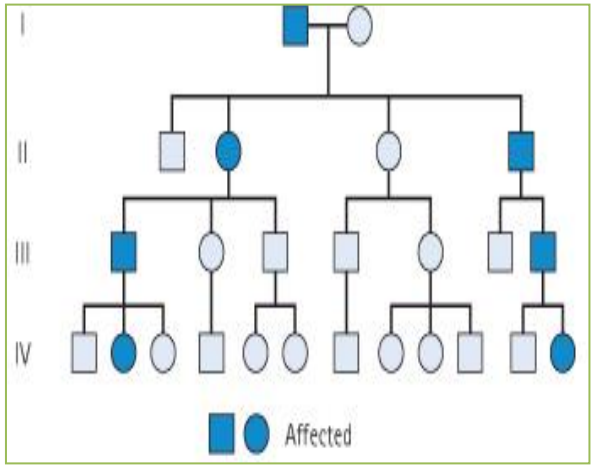


- The trait (character, disease) appears in **every generation**.
- Unaffected persons do not transmit the trait to their children.
- Examples: Huntington disease, Myotonic dystrophy, Marfan syndrome etc.

A Pedigree Analysis for Huntington's disease



Family Tree of an Autosomal Dominant Mode of Inheritance



Note:

The presence of male-to-male (i.e. father to son) transmission

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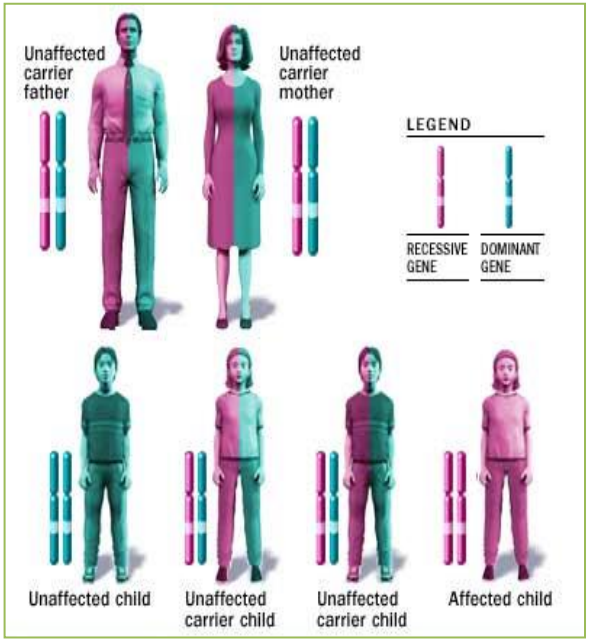
AUTOSOMAL RECESSIVE MODE OF INHERITANCE:

- The trait (character, disease) is recessive.
- The trait expresses itself only in homozygous state.
- Unaffected persons (heterozygotes) may have affected children (if the other parent is heterozygote).
- The parents of the affected child maybe related (consanguineous).
- Males and female are equally affected.

- Examples:

Cystic fibrosis
Phenyketonuria
Sickle cell anaemia
Thalassaemia.. etc



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PUNNETT SQUARE
showing Autosomal Recessive inheritance:

(1) Both Parents Heterozygous:

Father \ Mother	**A**	**a**
A	AA	Aa
a	Aa	aa

- **25% offspring affected “Homozygous”**
- **50% Trait “Heterozygous normal but carrier”**
- **25% Normal**

(2) One Parent Heterozygous:

Father \ Mother	**A**	**a**
A	AA	Aa
A	AA	Aa

- **50% normal but carrier “Heterozygous”**
- **50% Normal**

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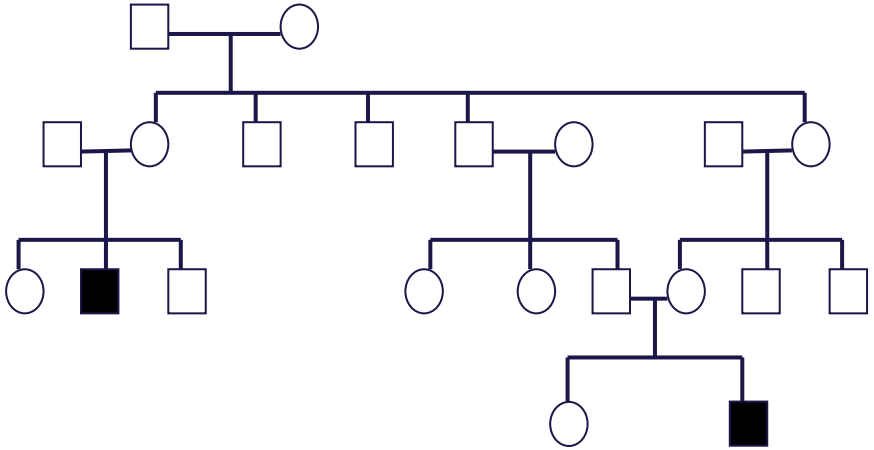
PUNNETT SQUARE
showing Autosomal Recessive inheritance:

(1) Both Parents Homozygous:

Mother / Father	**A**	**A**
a	Aa	Aa
a	Aa	Aa

✧ **100% offsprings carriers.**

*** Family tree of an Autosomal Recessive Disorder:**



✧ **Sickle cell disease (SS)**

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Sex – Linked inheritance

- This is the inheritance of a gene present on the sex chromosomes.
- The Inheritance Pattern is **different** from the autosomal inheritance.
- Inheritance is different in the males and females.

Y – Linked **Inheritance**

- The gene is on the **Y** chromosomes.
- The gene is passed from **fathers to sons** only
- Daughters are **not affected**
- Hairy ears in India
- Male are Hemizygous, the condition exhibits itself
- whether dominant or recessive.

Father / Mother	**X**	**Y***
X	XX	XY*
X	XX	XY*

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X – Linked inheritance

- The gene is present on the **X** chromosome
- The inheritance follows **specific** pattern
- **Males** have one X chromosome, and are **hemizygous**
- **Females** have **2** X chromosomes, they may be **homozygous or heterozygous**
- These disorders may be **: recessive or dominant**

X – Linked Recessive Inheritance

- The incidence of the **X-linked** disease is higher in male than in female
- The trait is passed from an **affected man** through all his daughters to half their sons
- The trait is **never** transmitted directly from father to sons
- An **affected** women has **affected sons** and **carrier daughters**
- X - Linked Recessive Disorders: Albinism, Fragile X syndrome, Hemophilia, Muscular dystrophy, Retinitis pigmentosa

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PUNNETT SQUARE
showing X-Linked Recessive inheritance:

(1) Normal female, affected male

Mother / Father	X	X
X*	XX*	XX*
Y	XY	XY

- All sons are normal
- All daughters carriers (not affected)

(2) Carrier female, normal male:

Mother / Father	X*	X
X	XX*	XX
Y	X*Y	XY

- 50% sons affected
- 50% daughters carriers

(3) Homozygous female, normal male

Mother / Father	X*	X*
X	XX*	XX*
Y	X*Y	X*Y

- All sons affected
- All daughters carriers

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X – Linked Dominant Inheritance

- The gene is on **X** Chromosome and is **dominant**
- The trait occurs at the same frequency in both males and females
- **Hemizygous** male and **heterozygous** females express the disease.

PUNNETT SQUARE showing X-Linked Dominant inheritance:

(1) Affected male and normal female

Father \ Mother	X	X
X*	XX*	XX*
Y	XY	XY

All daughters affected
All sons normal

(2) Affected female (heterozygous) and normal male

Father \ Mother	X*	X
X	XX*	XX
Y	X*Y	XY

50% sons affected
50% daughters affected

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YouTube HELPFUL VIDEOS

- How Mendel's Pea Plants Helped Us Understand Genetics
http://www.youtube.com/watch?v=Mehz7tCxjSE

- Heredity
http://www.youtube.com/watch?v=CBezq1fFUEA

- pedigrees, Patterns of Genetic Inheritance, Autosomal Dominant Recessive X-Linked Mitocondrial
http://www.youtube.com/watch?v=BG6atAkSejc

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Take Home Message

- An accurate determination of the family pedigree is an important part of the workup of every patient
- Pedigrees for single-gene disorders may demonstrate a straightforward, typical mendelian inheritance pattern
- These patterns depend on the chromosomal location of the gene locus, which may be autosomal or sex chromosome-linked, and whether the phenotype is dominant or recessive

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