BIO 201

GENETICS I

LINKAGE AND RECOMBINATION

Linkage is the coexistence of two or more genes in the same chromosome and their inheritance in group.

Linked genes do not assort independently but tend to stay together in the same combination (form a linkage group) as they were in the parents.

Because, all the genes of a chromosome have their identical genes on the homologous chromosome, therefore linkage groups of a homologous pair of chromosome is considered as one.

Linkage reduces the possibility of variability in gametes unless crossing over occurs.

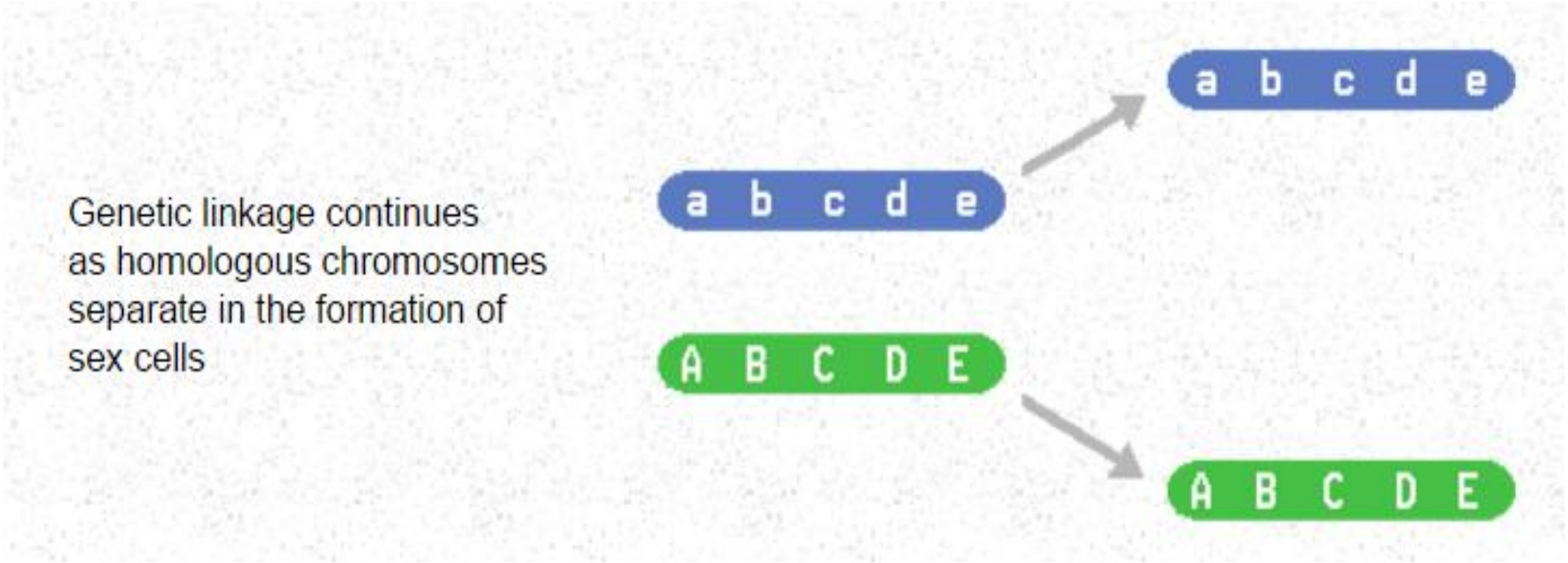
LINKAGE AND RECOMBINATION

Linked genes in a dihybrid may be present in one of two basic conformations;

Cis conformation - The two dominant or wild-type alleles are present on the same homolog.

Trans conformation - The two dominant or wild-type alleles are on different homologs

- Cis *A B/a b*
- Trans *A b/a B*



Source: http://anthro.palomar.edu/biobasis/bio_3.htm

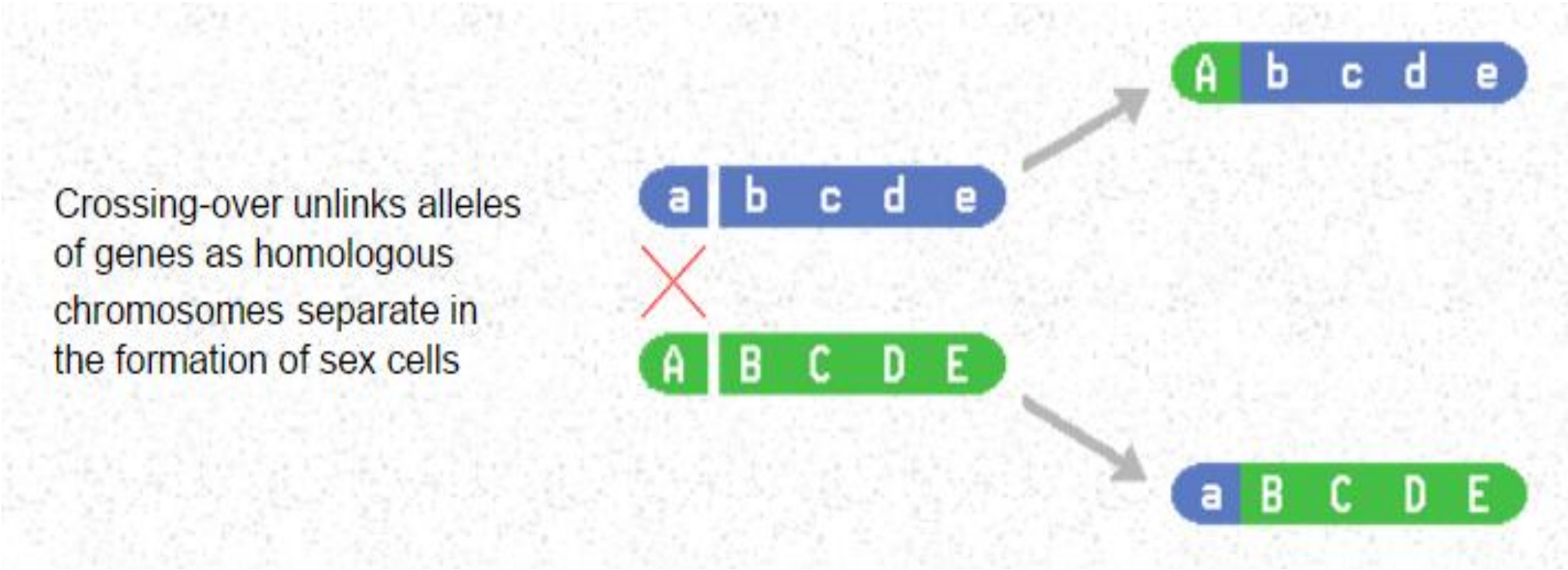
LINKAGE AND RECOMBINATION

Recombination is accomplished through a process known as crossing over.

The process produces new combinations (recombinations) of genes by interchanging of corresponding segments between non-sister chromatids of homologous chromosomes.

Crossing-over or recombination occurs at two levels; chromosomal level and at DNA level (genetic recombination)

The frequency of crossing over appears to be closely related to physical distance between genes on chromosome and serves as a tool in constructing genetic maps of chromosomes.



Source: http://anthro.palomar.edu/biobasis/bio_3.htm

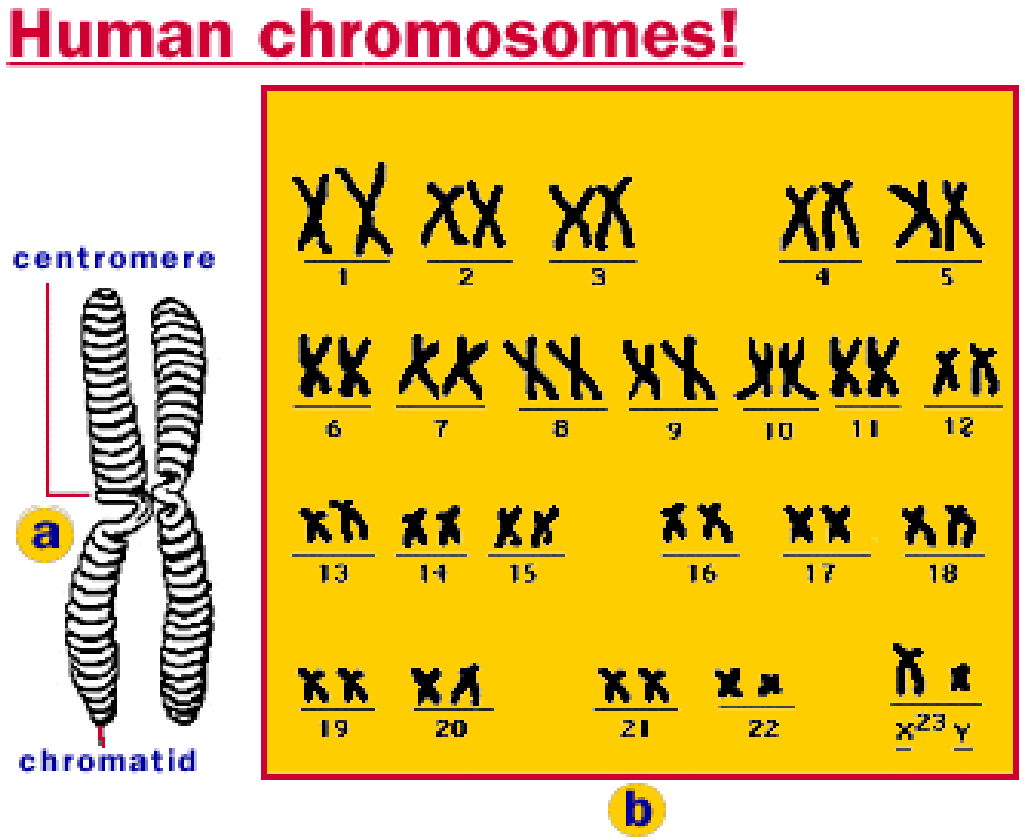
SEX CHROMOSOMES AND SEX-LINKED INHERITANCE

Most animals and many plants show sexual dimorphism

Presence of special pair of **sex chromosomes.**

For example, human body cells have 46 chromosomes: 22 homologous pairs of autosomes plus 2 sex chromosomes.

- Females – XX chromosomes showing homogametic sex.
- Males – XY chromosomes showing heterogametic sex.



Source: http://learningon.theloop.school.nz/moodle/mod/book/view.php?id=14895

SEX CHROMOSOMES AND SEX-LINKED INHERITANCE

.Fruit flies have three pairs of autosomes plus a pair of sex chromosomes.

- Females – XX chromosomes
- Males – XY chromosomes

*In *Drosophila,* **the *number of X chromosomes* determines sex**: two X's result in a female and one X results in a male. In mammals, **the *presence of the Y* determines maleness** and the absence of a Y determines femaleness.

The dioecious plant *Melandrium album* has 22 chromosomes per cell: 20 autosomes plus 2 sex chromosomes, with XX females and XY males.

Sex-linked patterns of inheritance

X and Y chromosomes are divided into homologous and differential regions.

Homologous regions contain DNA sequences that are substantially similar on both sex chromosomes. The *differential* regions contain genes that have no counterparts on the other sex chromosome.

The **X chromosome contains many hundreds of genes with no counterparts on the Y.**

The Y chromosome contains only a few dozen genes which are more involved in male sexual function. Some of these genes have counterparts on the X, but some do not.

Sex-linked patterns of inheritance

Genes on the differential regions are said to show **sex linkage**

- Genes in the differential region of the X show an inheritance pattern called **X linkage;** those in the differential region of the Y show **Y linkage**

This pattern contrasts with the inheritance patterns of genes on the autosomes

X-linked Inheritance

A good example of X linkage is the eye color in *Drosophila controlled* by two alleles of a gene located on the differential region of the X chromosome.

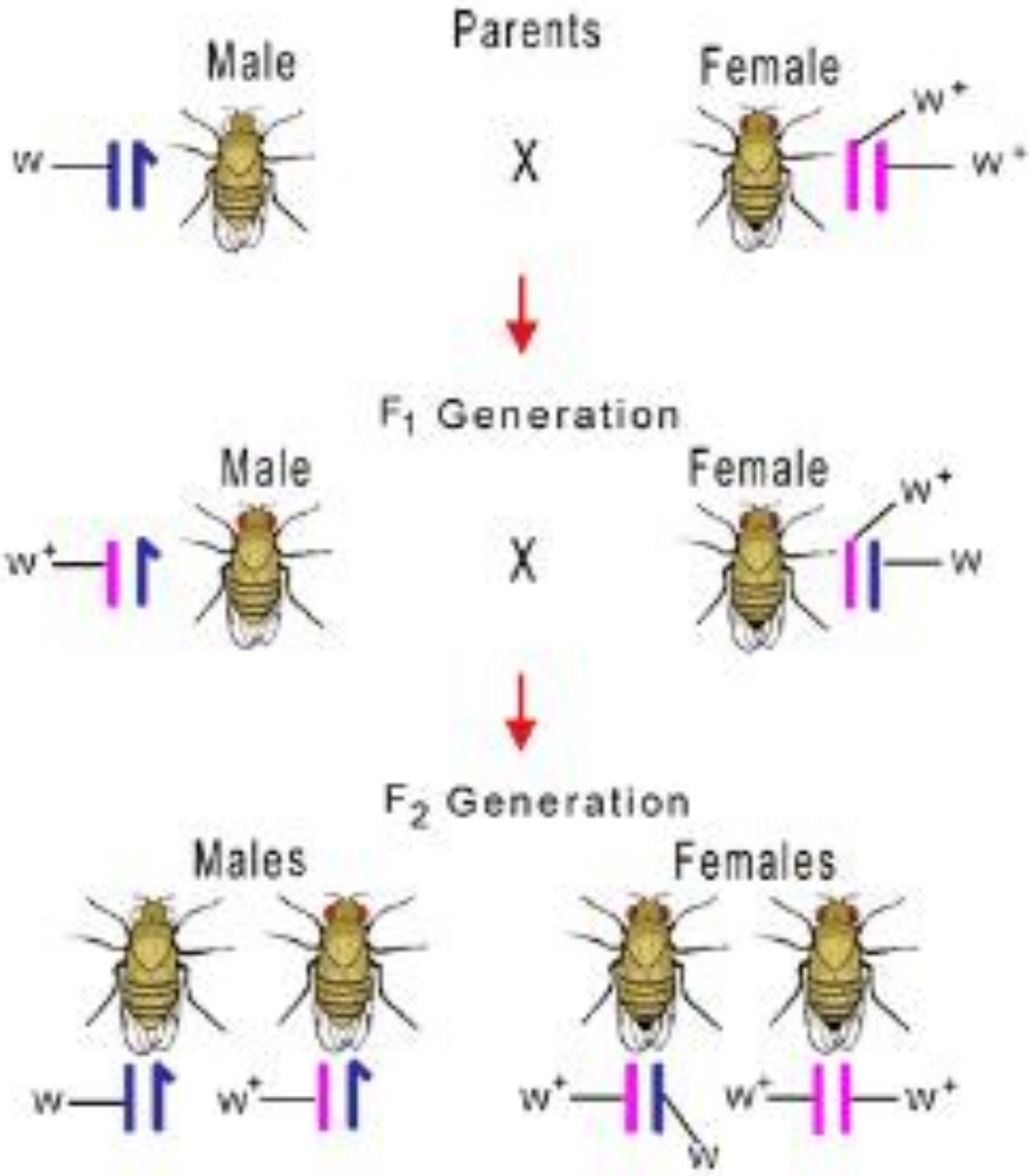
The mutant allele is *w* for white eyes (the lowercase shows it is recessive), and the corresponding wild-type allele is w^+.

In sex linkage, different ratios exist in different sexes and also there are differences between reciprocal crosses.

X-linked Inheritance

For example, when white-eyed males (X^wY) are crossed with red-eyed females ($X^{w+}X^{w+}$), all the F_1 progeny have red eyes, showing that the allele for white is recessive. Crossing these red-eyed F_1 males and females produces a 3:1 F_2 ratio of red-eyed to white-eyed flies, but all the white-eyed flies are males.

A reciprocal cross between white-eyed females (X^wX^w) and red-eyed males ($X^{w+}Y$) gives an F_1 in which all the females are red-eyed, but all the males are white-eyed. The F_2 consists of half red-eyed and half white-eyed flies of both sexes.



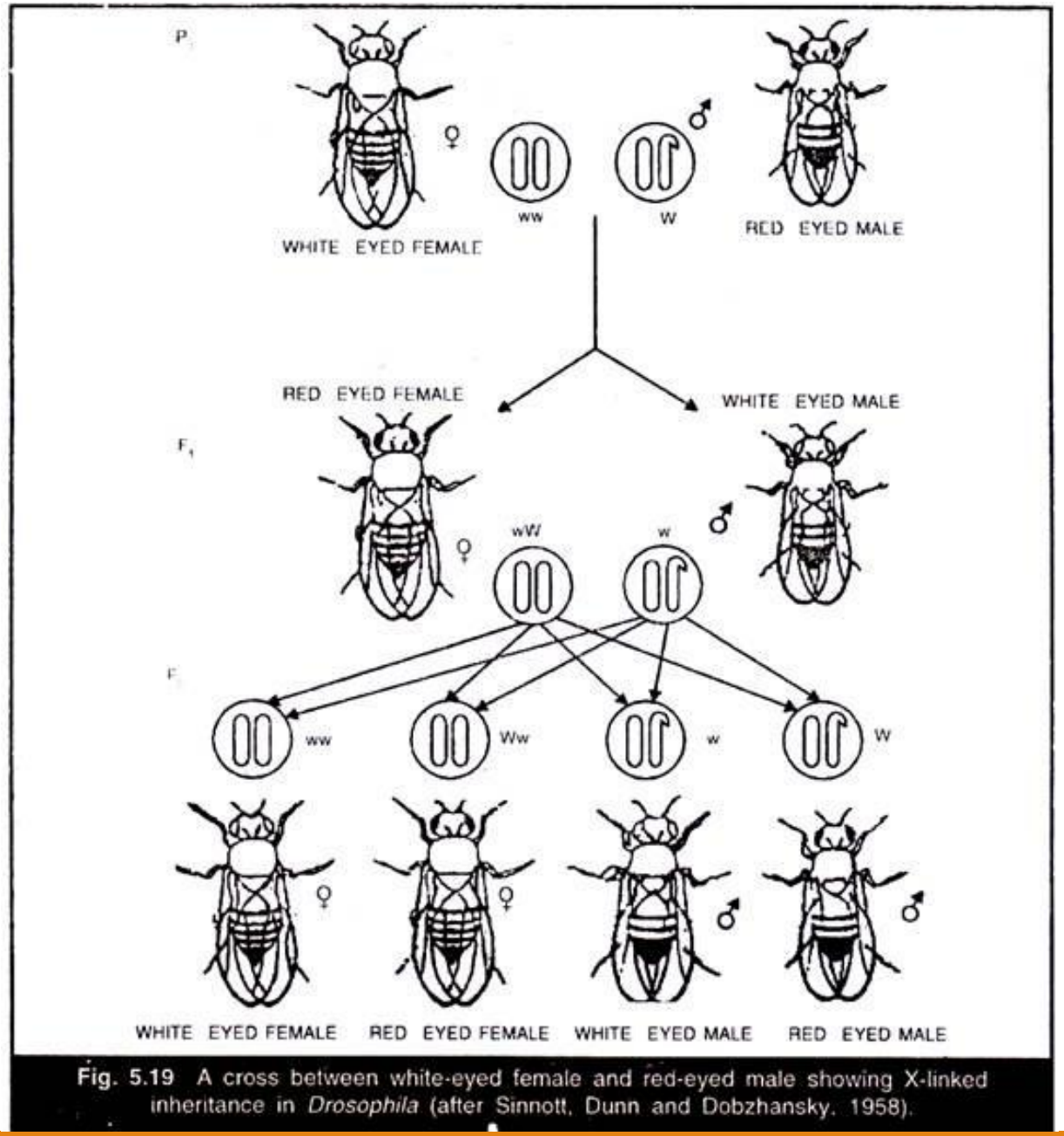


Fig. 5.19 A cross between white-eyed female and red-eyed male showing X-linked inheritance in *Drosophila* (after Sinnott, Dunn and Dobzhansky, 1958).

Source: http://www.biologydiscussion.com/genetics/sex-linked-inheritance-sex-linkage-in-drosophila-and-man-with-diagram/5241

Source: https://wikispaces.psu.edu/display/bio110/Chromosome+Behavior+and+Sex+Chromosomes

Pedigree Analysis of X-linked Recessive Disorders

1. Many more males than females show the rare phenotype under study. A female will show the phenotype only if both her mother *and* her father bear the allele (for example, $X^A X^a$ x X^a Y).

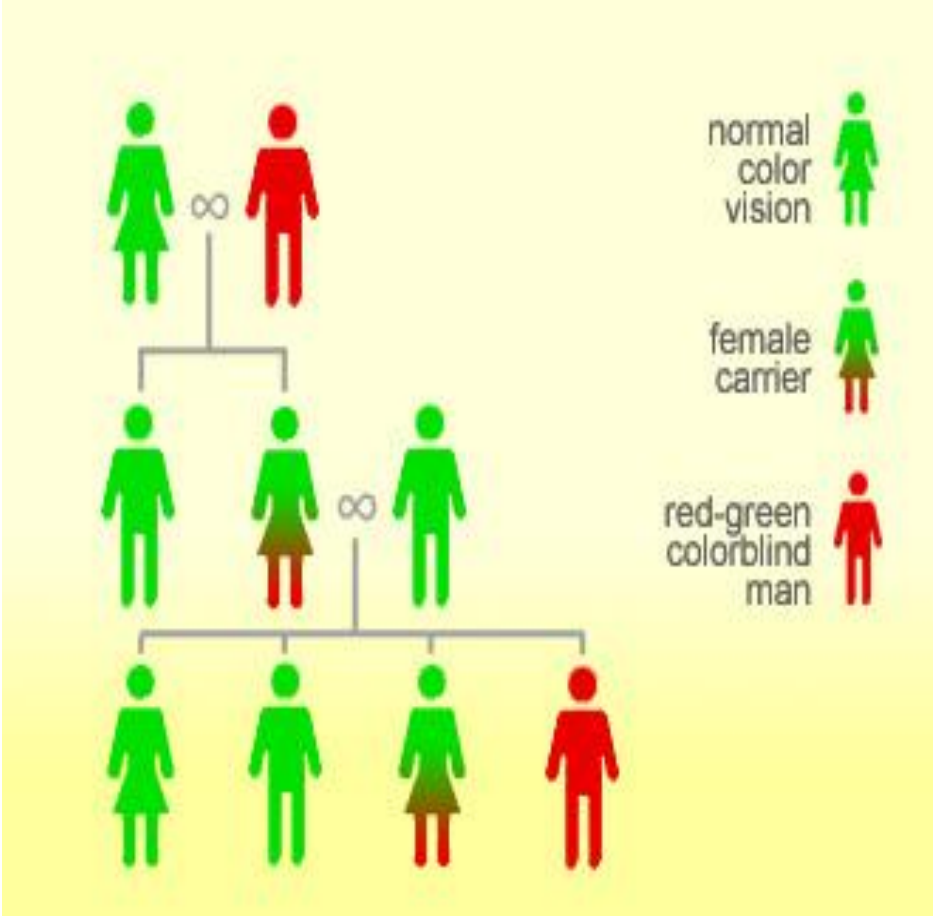
2. None of the offspring of an affected male show the phenotype, but all his daughters are "carriers," who bear the recessive allele masked in the heterozygous condition. Half the sons of these carrier daughters show the phenotype. This pattern might be obscured by inheritance of the recessive allele from a heterozygous mother as well as the affected father.

3. None of the sons of an affected male show the phenotype under study, nor will they pass the condition to their offspring.

***In the pedigree analysis of rare X-linked recessives, a normal female of unknown genotype is assumed to be homozygous unless there is evidence to the contrary.**

Examples of X-linked Recessive Disorders

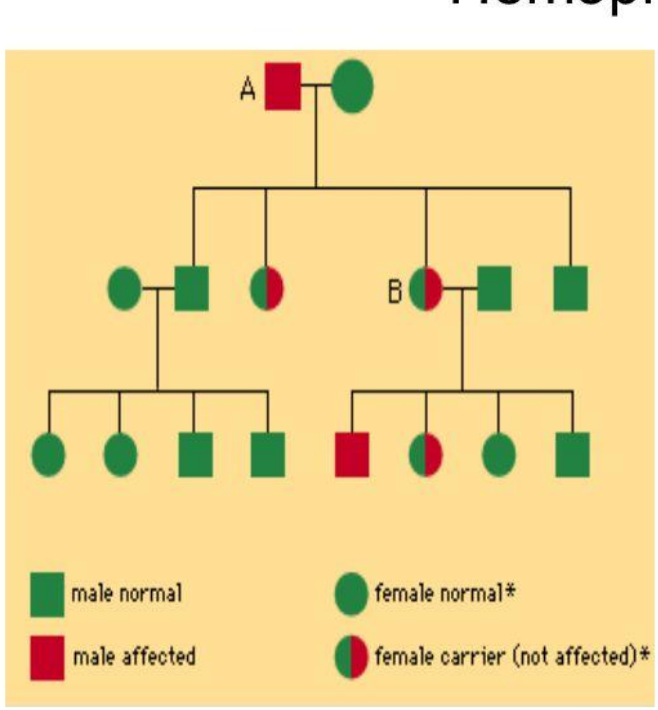
Red-green colour blindness - People with this condition are unable to distinguish red from green. Colour vision is based on three different kinds of cone cells in the retina, each sensitive to red, green, or blue wavelengths. The genetic determinants for the red and green cone cells are on the X chromosome. As with any X-linked recessive, there are many more males with the phenotype than females.



Source: http://www.color-blindness.com/what-is-color-blindness/

Examples of X-linked Recessive Disorders

Haemophilia - This is the failure of blood to clot. The most common type of haemophilia is caused by the absence or malfunction of one of the proteins called *factor VIII* which act in sequence to make blood clot. The most well known cases of haemophilia are found in the pedigree of interrelated royal families in Europe.



Transmission of hemophilia

(A) Mating of affected hemophilic man and normal woman—all sons normal, all daughters carriers.
(B) (B) Mating of carrier woman and normal man—half of sons normal and half affected;
(C) half of daughters carriers, half normal.

Examples of X-linked Recessive Disorders

Testicular feminization syndrome - People afflicted with this syndrome are chromosomally males, having 44 autosomes plus an X and a Y, but they develop as females. They have female external genitalia, a blind vagina, and no uterus. Testes may be present either in the labia or in the abdomen. Although many such persons marry, they are sterile.

Pedigree Analysis of X-linked Dominant Disorders

1. Affected males pass the condition to all their daughters but to none of their sons

2. Affected heterozygous females married to unaffected males pass the condition to half their sons and daughters.

There are few examples of X-linked dominant phenotypes in humans. One example is *hypophosphatemia,* a type of vitamin D–resistant rickets.

Y-linked Inheritance

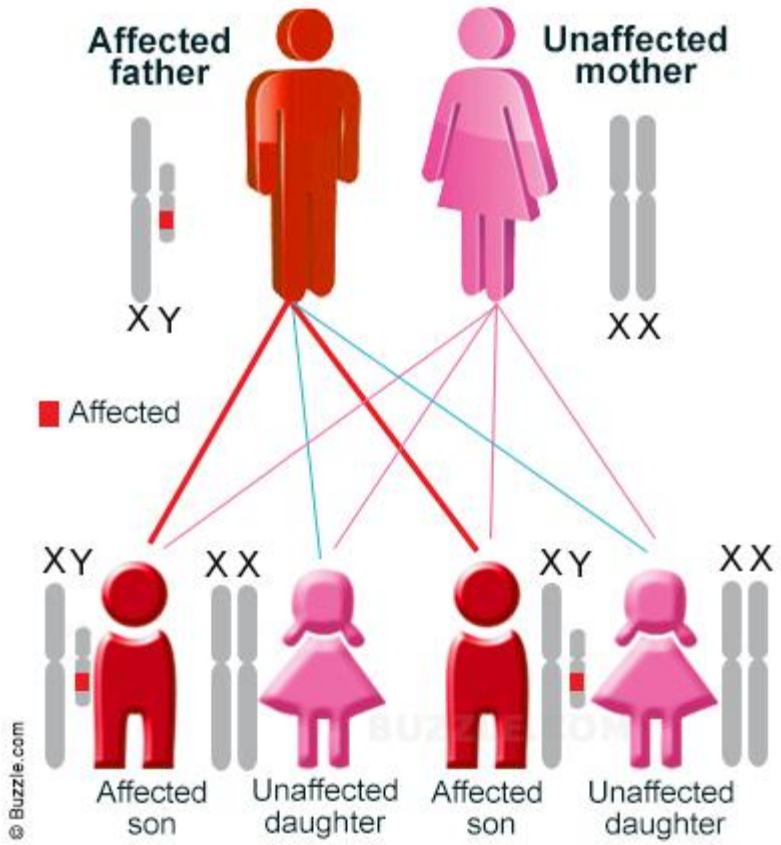
Only males inherit genes on the differential region of the human Y chromosome, with fathers transmitting the genes to their sons.

Example is the *SRY* gene located on the differential region of the Y chromosome. It is sometimes called the *testis-determining factor*.

Some cases of male sterility have been shown to be caused by deletions of Y chromosome regions containing sperm-promoting genes.

There have been no convincing cases of non-sexual phenotypic variants associated with the Y.

Hairy ear rims has been proposed as a possibility. In some (but not all) families hairy ear rims are transmitted exclusively from father to son.



CYTOPLASMIC INHERITANCE OR MATERNAL INHERITANCE

Occurrence of DNA molecules in the cytoplasm of many eukaryotic and prokaryotic cells.

For example, bacterial cells such as *E. coli* possess extra-DNA elements called plasmids in the cytoplasm. Likewise, the eukaryotic cells have extra-DNA molecules or small circular chromosomes in their mitochondria and chloroplasts.

Mitochondrial genes are concerned with the mitochondrion's task of energy production, whereas chloroplast genes are needed for the chloroplast to carry out its function of photosynthesis.

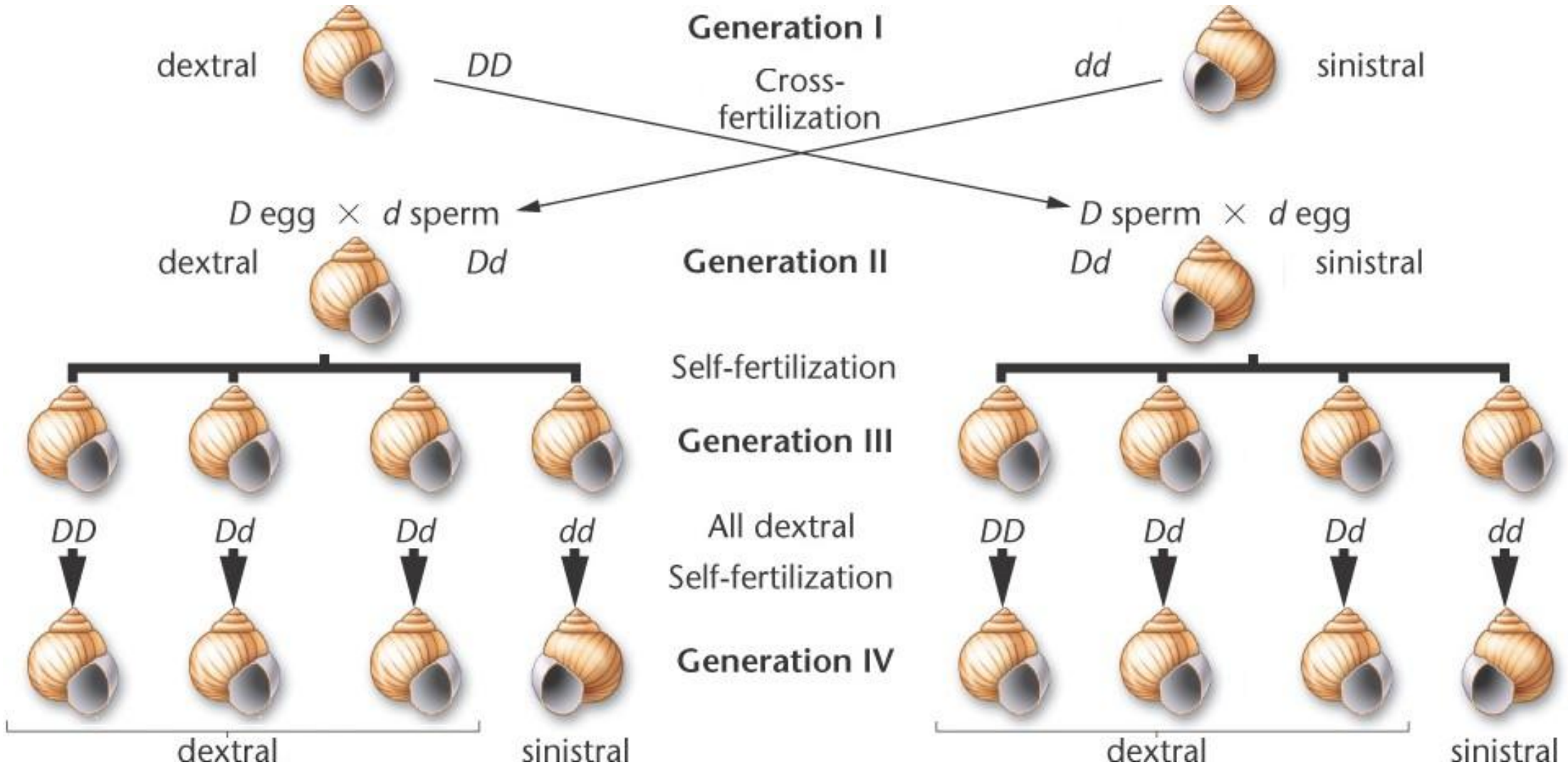
CYTOPLASMIC INHERITANCE OR MATERNAL INHERITANCE

These cytoplasmic genes have a characteristic pattern of inheritance referred to as maternal, uniparental or cytoplasmic inheritance.

This is because the male and female gametes do not contribute cytoplasm equally to the zygote. The egg contributes the bulk of the cytoplasm and the sperm essentially none. Hence, the female parent contributes the organelles along with the cytoplasm.

This is the basis for maternal inheritance where the progeny always resemble one parent, the female.

Examples are shell coiling in *Limnaea peregra,* chloroplast inheritance in *Mirabilis jalapa,* streptomycin resistance in *Chlamydomonas,* mitochondrial inheritance in yeast etc.


Generation I
dextral
DD
Cross-fertilization
dd
sinistral
D egg × d sperm
D sperm × d egg
dextral
Dd
Generation II
Dd
sinistral
Self-fertilization
Generation III
DD
Dd
Dd
dd
All dextral
Self-fertilization
DD
Dd
Dd
dd
Generation IV
dextral
sinistral
dextral
sinistral

Get a sheet of paper!

The gene for yellow body colour y in Drosophila is recessive and sex-linked. Its dominant allele y^+ produces wild type body colour. What phenotypic ratios are expected from the following crosses?

A. yellow male X yellow female

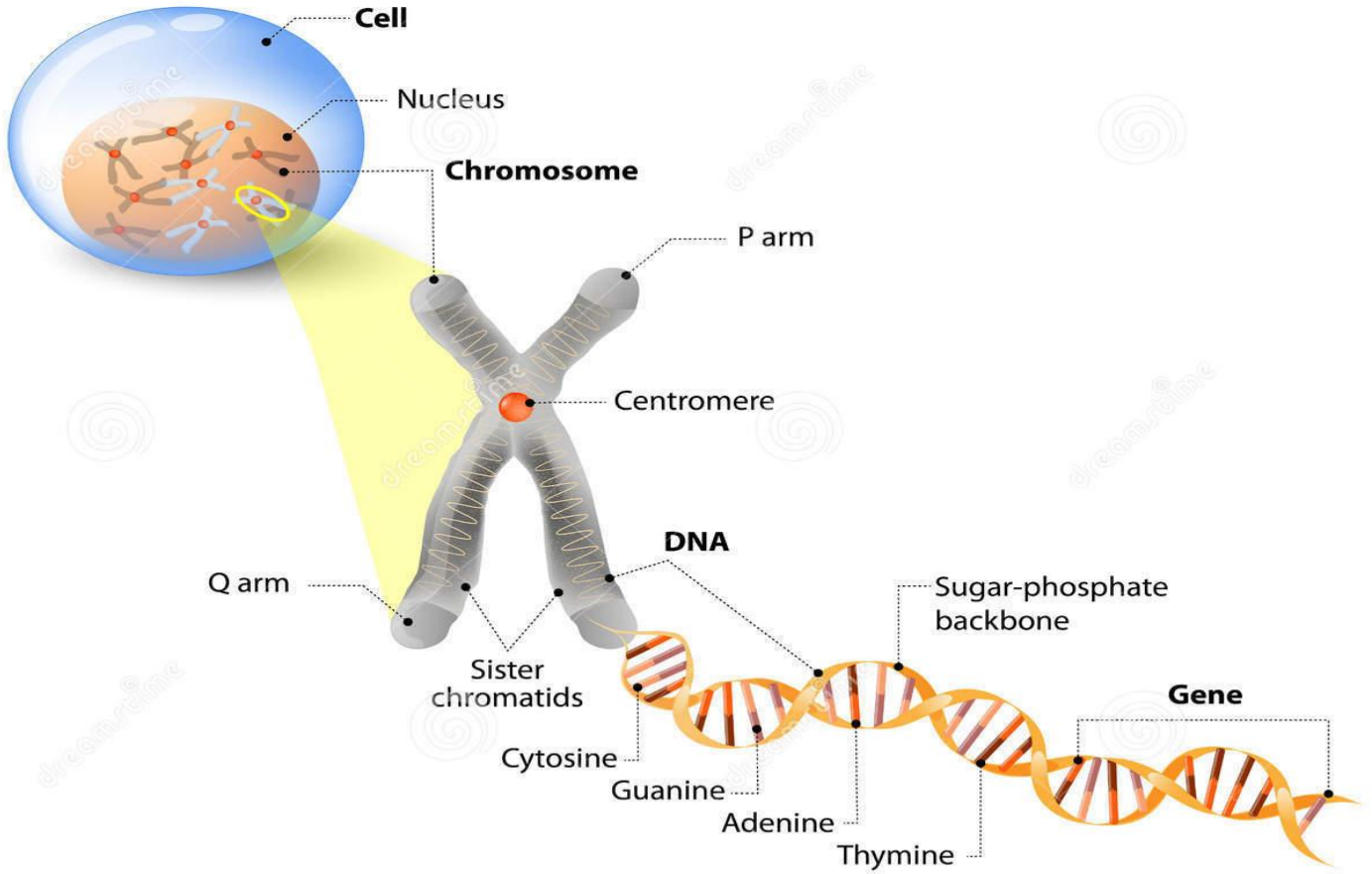
B. yellow female X wild type male

C. wild female (homozygous) X yellow male

D. wild type (carrier) female X yellow male

E. wild type (carrier) female X wild type male

VARIATION IN GENOME STRUCTURE



VARIATION IN GENOME STRUCTURE

Genetic variation is a term used to describe the variation in the DNA sequence in each of our genomes.

It results in different forms, or alleles, of genes which makes us all unique, whether in terms of hair colour, skin colour or even the shape of our faces.

Genetic variation among individuals provides the raw material for evolution.

The ultimate source of all variation is mutation.

Mutation is a change in the DNA sequence of a gene that affects the phenotype.

VARIATION IN GENOME STRUCTURE

New alleles arise in all organisms, some spontaneously, others as a result of exposure to radiation and chemicals in the environment.

Genetic variations can range in size from macroscopic to sub-microscopic.

They can take the form of **gross structural changes in chromosomes**, missing or extra genetic information, or **minute errors in the spelling of the genetic code.**

In the cellular environment, DNA molecules are not absolutely stable; each base pair in a DNA double helix has a certain probability of mutating known as gene mutations.

Chromosome Mutations

The changes in the genome involving chromosome parts, whole chromosomes, or whole chromosome sets are called chromosome aberrations or chromosome mutations.

Oftentimes, they cause abnormalities in cell and organism function.

Most of these abnormalities stem from changes in gene number or gene position.

In some cases, a chromosome mutation results from chromosome breakage. If the break occurs within a gene, the result is functional disruption of that gene.

Chromosome mutations can basically be divided into two groups:

- **Changes in chromosome number-** These are not associated with structural alterations of any of the DNA molecules of the cell. Rather, it is the number of these DNA molecules that is changed, and this change in number is the basis of their genetic effects. This includes
- Loss or gain of whole chromosome set (euploidy)

i. Loss of an entire set of chromosome (monoploidy)

ii. Addition of one or more sets of chromosomes (polyploidy)

iii. Loss or gain of a part of the chromosome set (aneuploidy)

Monoploids

*They have a single basic set of chromosomes.

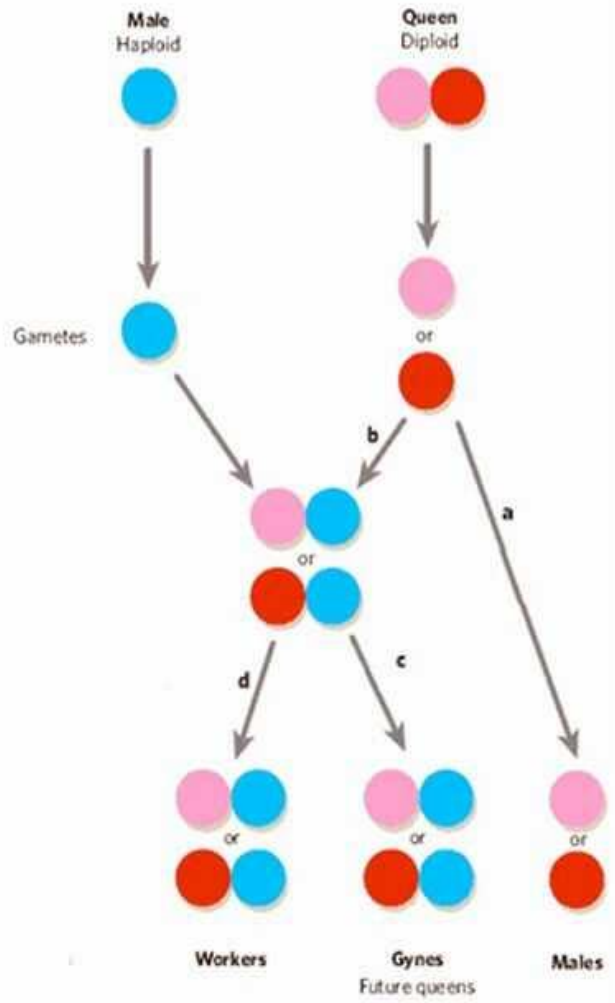
*During anaphase, each chromosome move independently of the other and goes to either of the two poles.

*Gametes containing less than the haploid number of chromosomes are normally not viable. Therefore monoploid organisms are highly sterile. bees, wasps, and ants.

*Monoploid plants have reduced size of all vegetative and floral parts.

Monoploid

- male wasps, bees and ants have only 1 haploid genome,
  - males develop from unfertilized eggs,
    - gametes are formed by mitosis.



Bees (example)

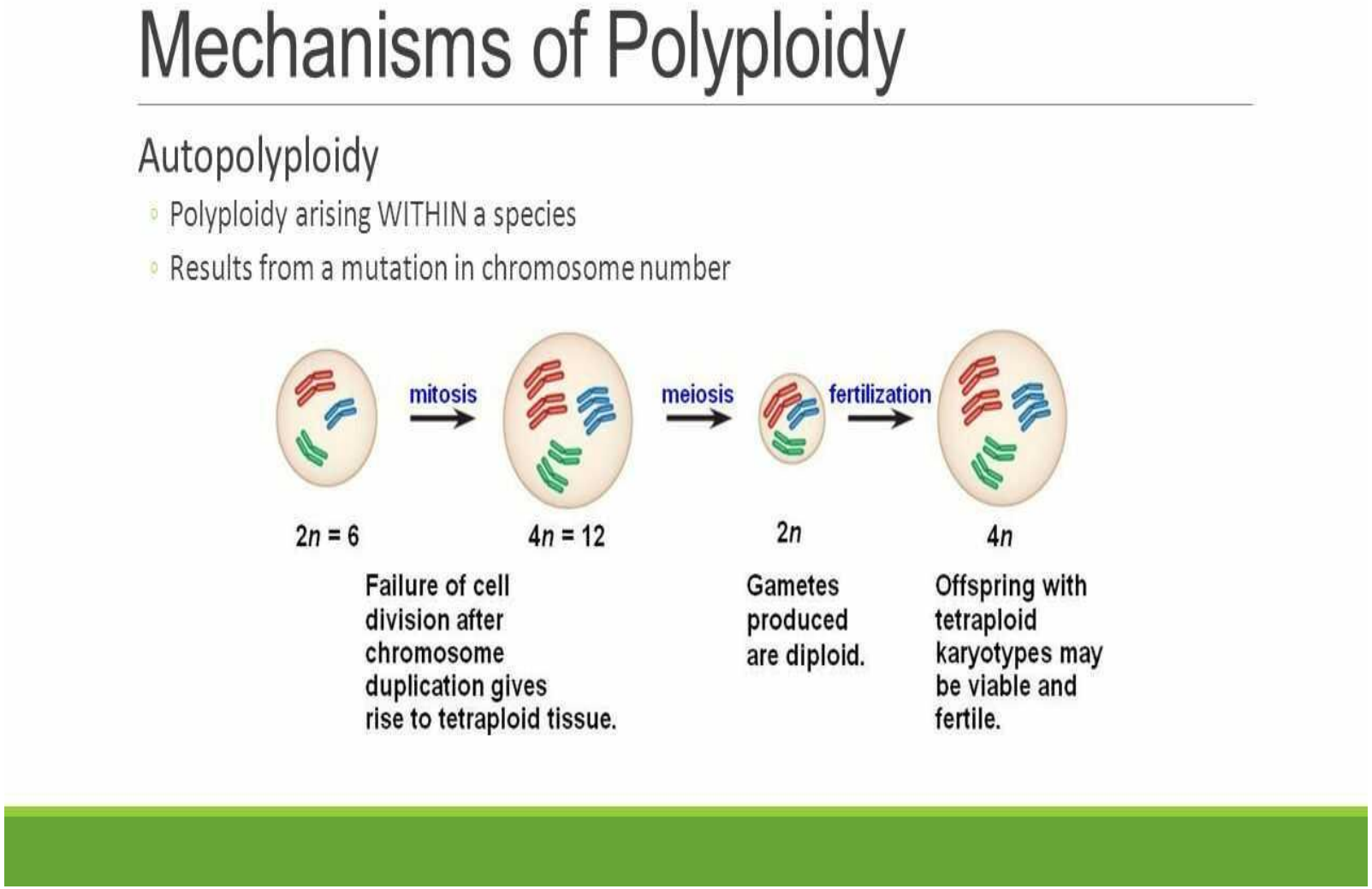
Autopolyploids

*They have multiple chromosome sets originating from within one species.

*They arise spontaneously in nature but may be produced artificially by means of chemicals, radioactive substances and temperature shocks.

*Triploids are usually autopolyploids and are characteristically sterile.

*Leaves, flowers and fruits of an autopolyploid are larger in size than diploid plant.

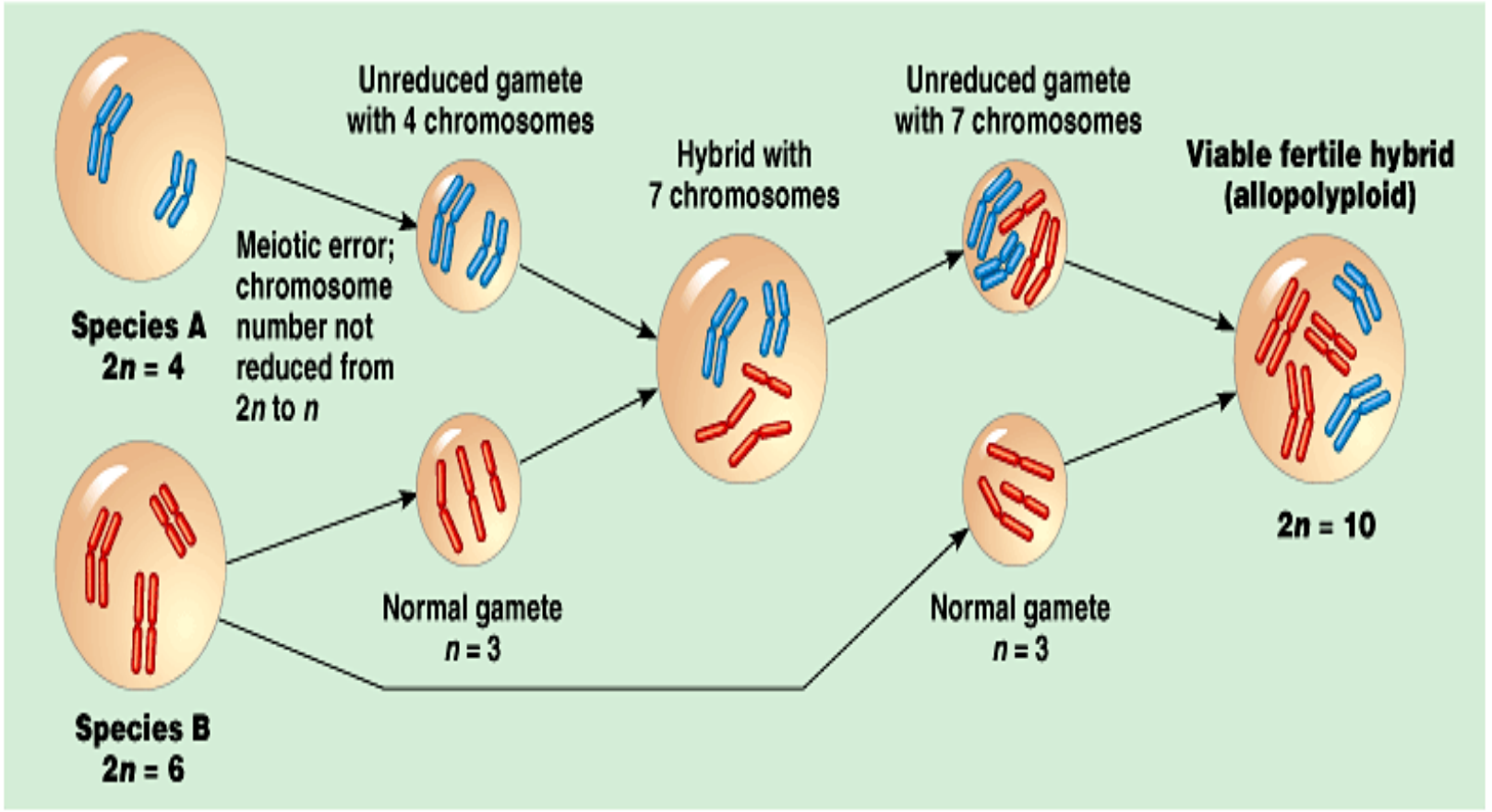


Allopolyploids

*occur due to the doubling of chromosome number in a F_1 hybrid which is derived from two distinctly different species.

*A cross between radish (*Raphanus sativum,* 2n=18) and cabbage (*Brassica oleracea,* 2n=18) produced F_1 got sterile (diploid) hybrids. Eventually, one part of the hybrid plant produced some seeds.

*On planting, these seeds produced fertile individuals with 36 chromosomes. These fertile tetraploids were called *Raphanobrassica.*



Polyploidy

Segmental allopolyploids – different genomes of some allopolyploids are not quite different from each other indicating that segments of chromosomes and not the whole chromosomes are homologous. Segmental allopolyploids are intermediate between autopolyploids and allopolyploids.

Aneuploidy

Aneuploidy is the second major category of chromosomal aberrations in which the chromosome number is abnormal.

Generally, the aneuploidy chromosome set differs from the wild type by only one chromosome or by a small number of chromosomes.

An aneuploid can have a chromosome number either greater or smaller than that of the wild type.

For autosomes in diploid organisms, the aneuploidy 2n+1 is trisomic, 2n---1 is monosomic, and 2n-2 (the -2 represents the loss of both homologs of a chromosome) is nullisomic. In haploids, n+1 is **disomic.**

Special notation is used to describe sex chromosome aneuploids because it must deal with the two different chromosomes. The notation merely lists the copies of each sex chromosome, such as XXY, XYY, XXX, or XO.

Aneuploid organisms result mainly from nondisjunction during a parental meiosis.

Monosomy

*Monosomics are missing one copy of a chromosome.

In humans, monosomics for any of the autosomes die in utero.

*Many X chromosome monosomics also die in utero, but some are viable.

- A human chromosome complement of 44 autosomes plus a single X produces a condition known as **Turner syndrome,** represented as XO.
- *Affected persons have a characteristic phenotype.
- *Although their intelligence is near normal, some of their specific cognitive functions are defective.

Turner syndrome

- **Monosomy X or X0**
  - 1 in every 5000 births
  - varied degree of effects
  - webbed neck
  - short stature
  - immature sterile females

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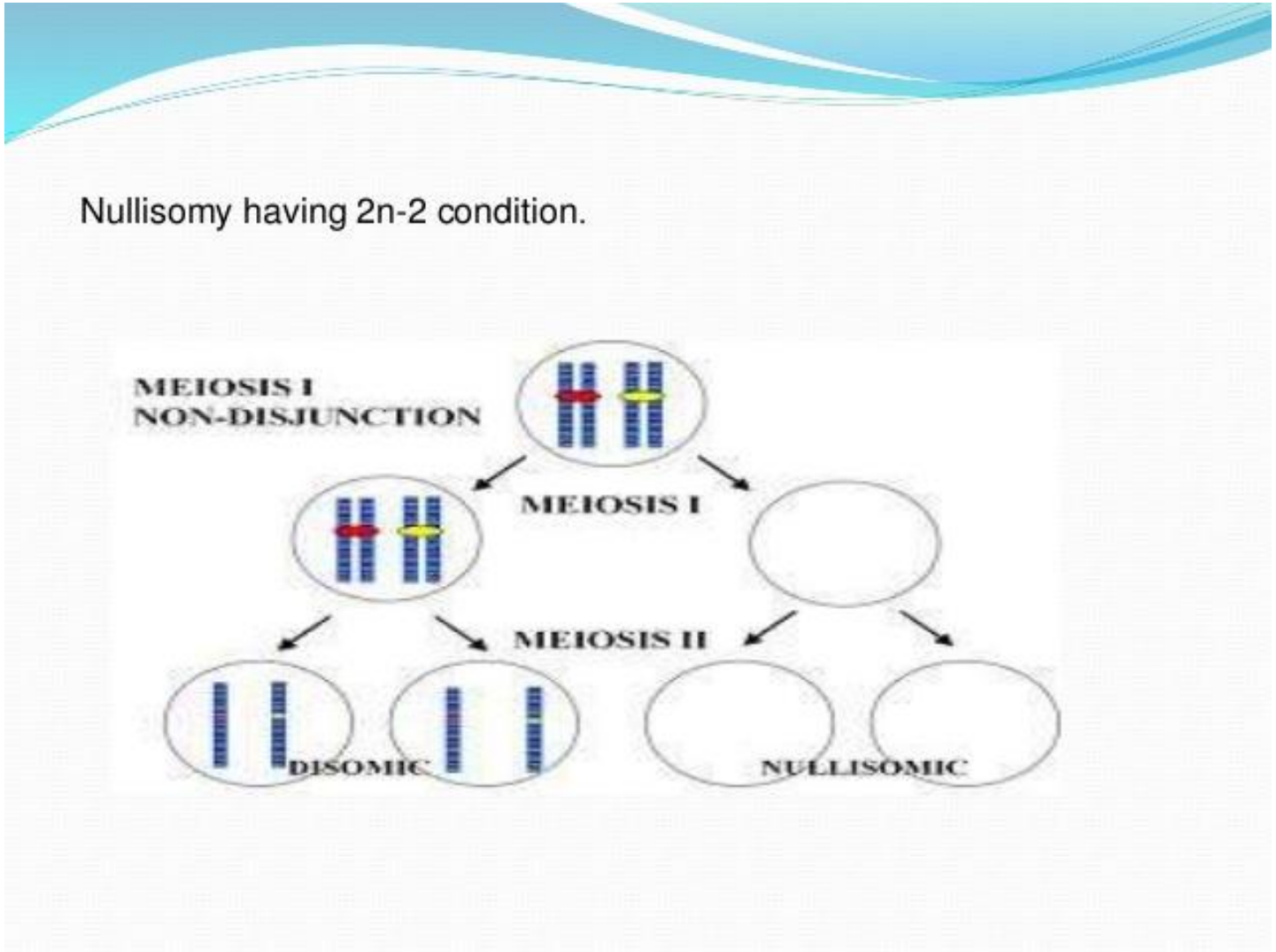
2005-2006

Nullisomy

*An organism which has lost a chromosome pair is a nullisomic with genomic formula 2n-2.

*A nullisomic diploid often does not survive, however, a nullisomic polyploid may survive but exhibit reduced vigour and fertility.

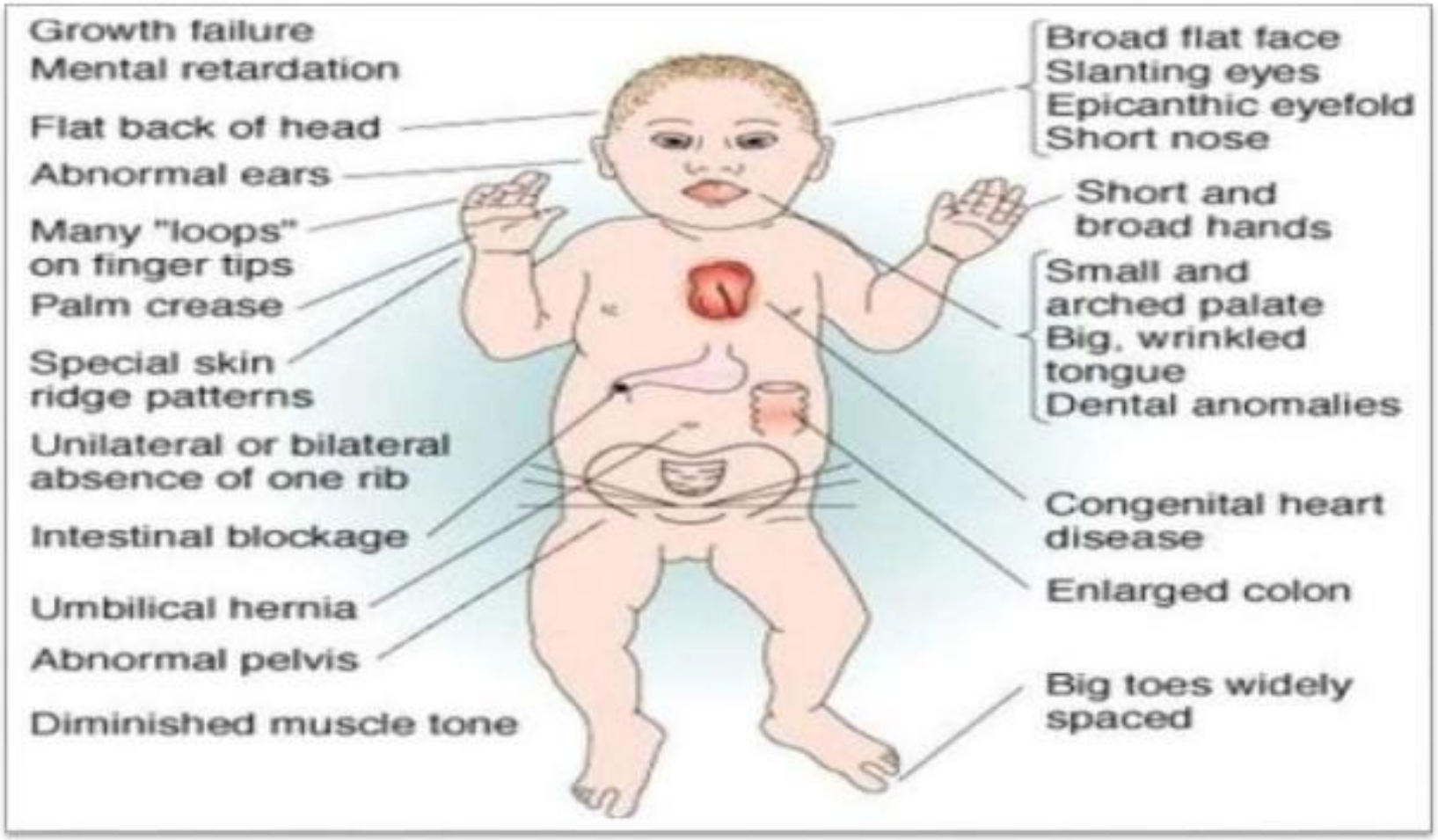


Trisomy

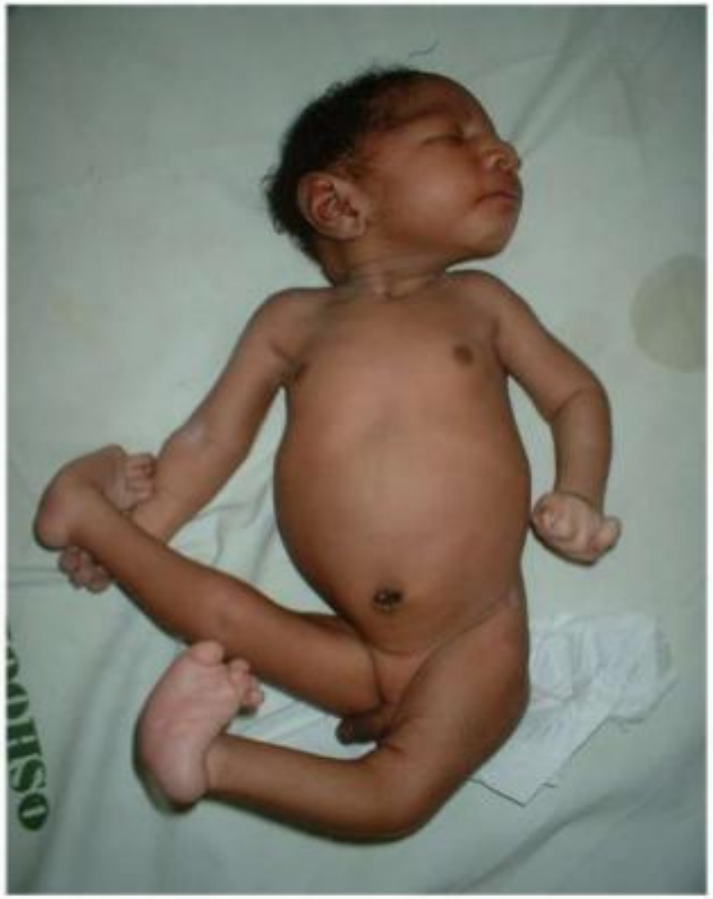
Trisomics contain an extra copy of one chromosome (2n + 1).

- In diploid organisms generally, the chromosomal imbalance from the trisomic condition can result in abnormality or death.
- There are many examples of viable trisomics.
- Furthermore, trisomics can be fertile. There are several examples of viable human trisomies.

Down's syndrome – Trisomy 21.

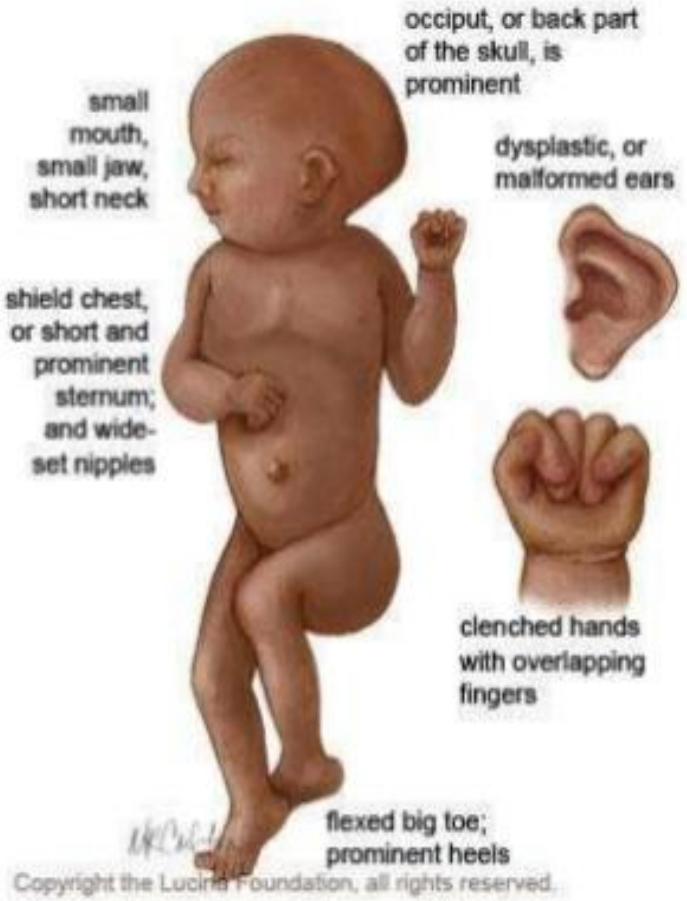


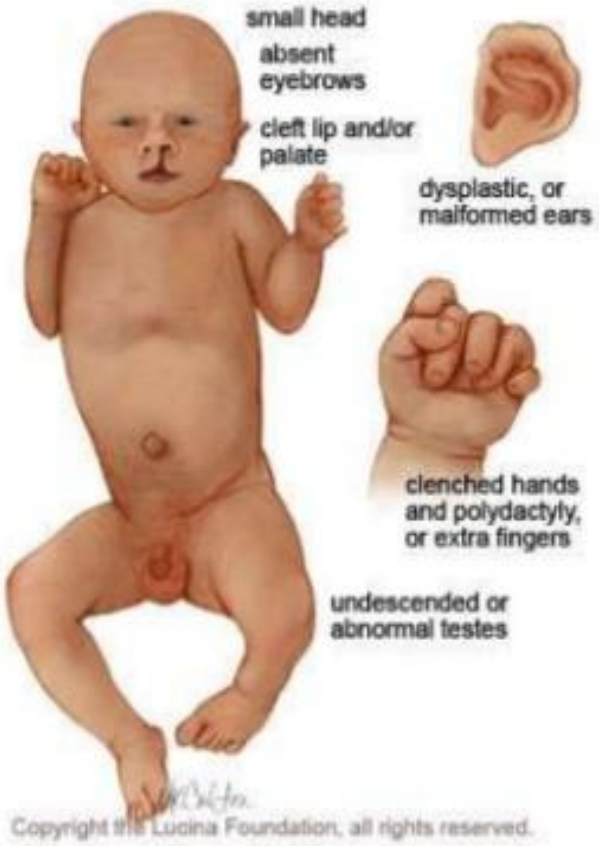
Trisomy 18 (Edward's Syndrome)



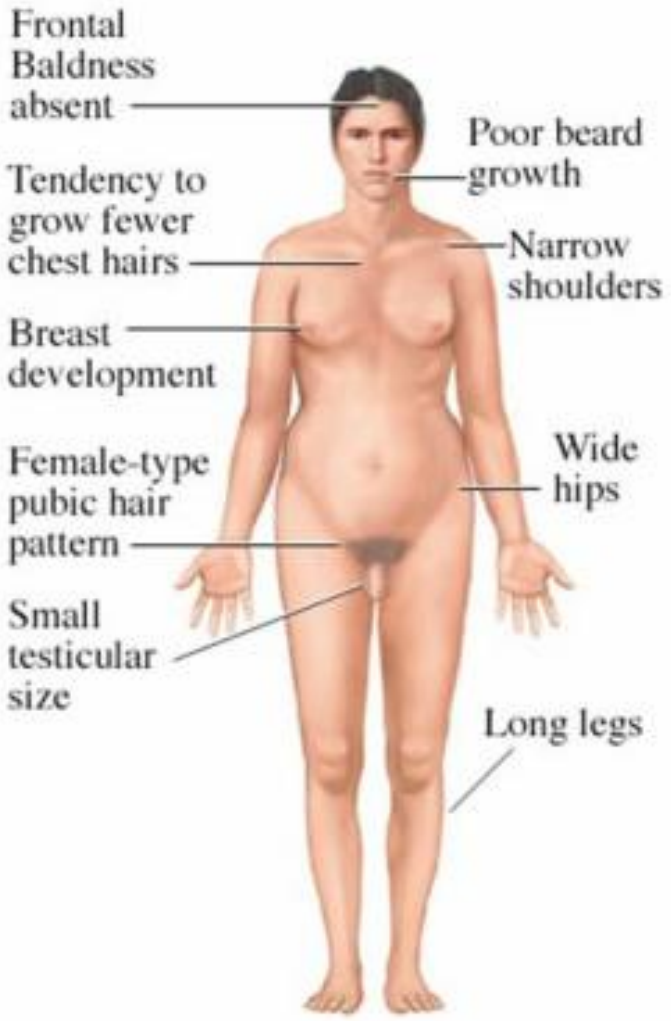
- unusually small head
- back of the head is prominent
- ears are malformed and low-set
- mouth and jaw are small (may also have a cleft lip or cleft palate
- hands are clenched into fists, and the index finger overlaps the other fingers
- Clubfeet (or rocker bottom feet) and toes may be webbed or fused

Patau's syndrome. (Trisomy 13)



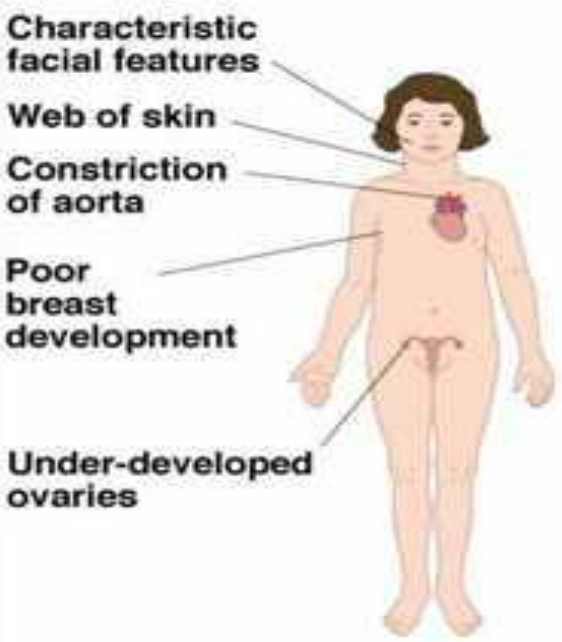


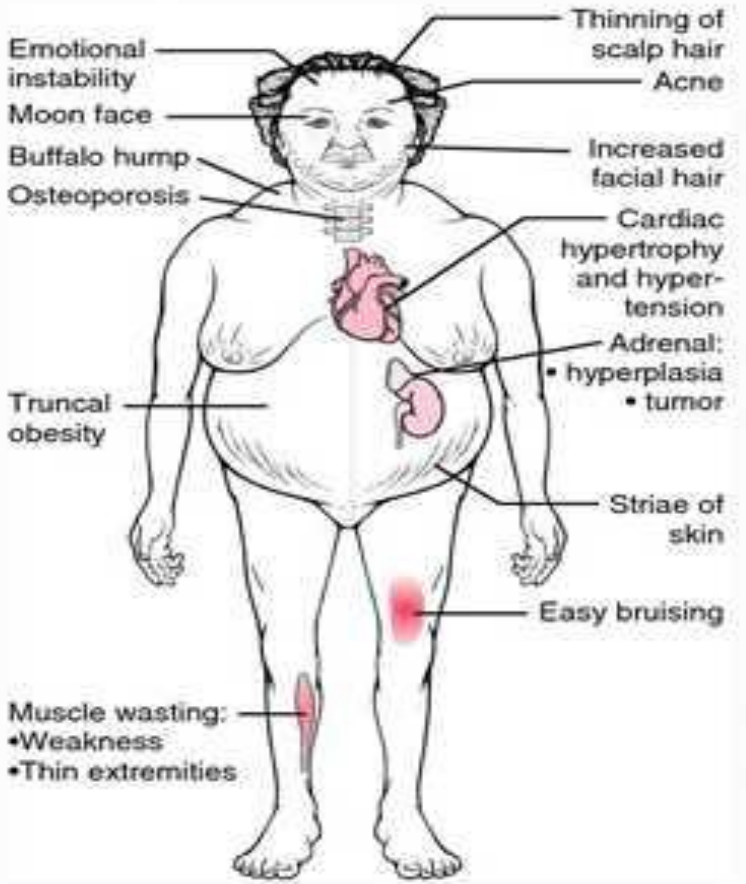
Klinefelter syndrome

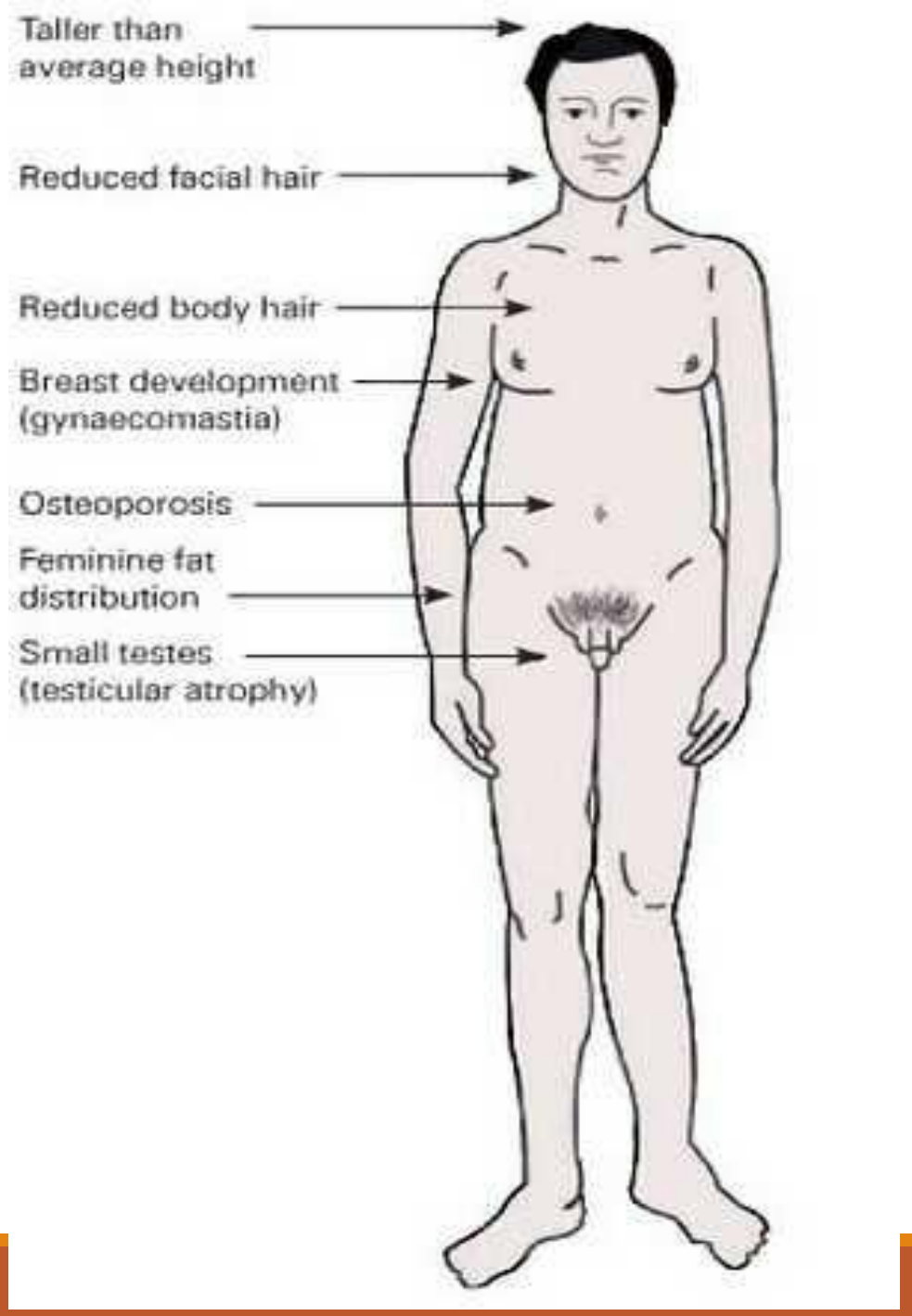


- Lower IQ than sibs
- Tall stature
- Poor muscle tone
- Reduced secondary sexual characteristics
- Gynaecomastia (male breasts)
- Small testes/infertility

Triple X syndrome
(in child and adult life)






Taller than average height
Reduced facial hair
Reduced body hair
Breast development (gynaecomastia)
Osteoporosis
Feminine fat distribution
Small testes (testicular atrophy)

Chromosome mutations

Changes in chromosome structure or structural aberrations - Changes in chromosome structure, on the other hand, result in novel sequence arrangements within one or more DNA double helices.

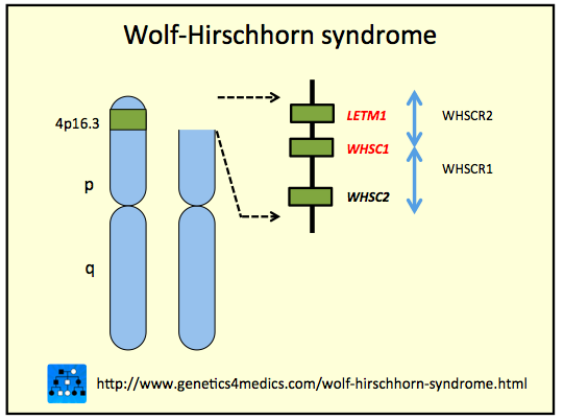
i. Deletion – terminal or intercalary.

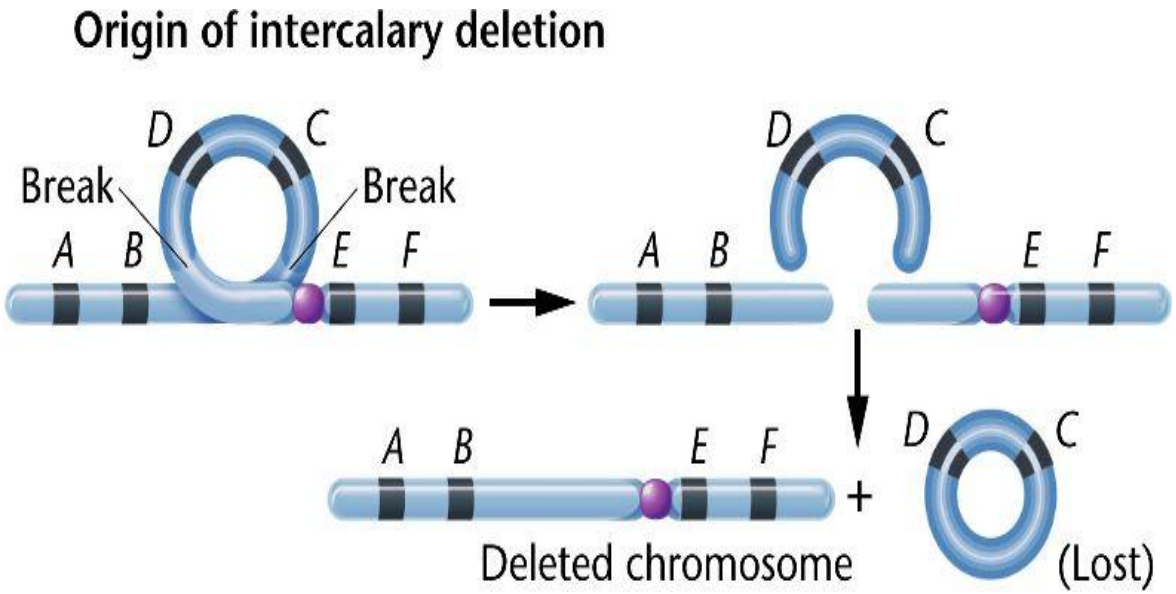
ii. Duplication

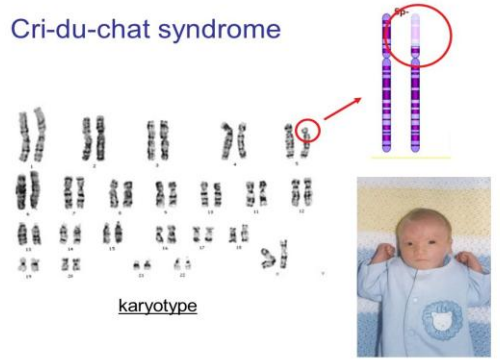
iii. Inversion – pericentric or paracentric

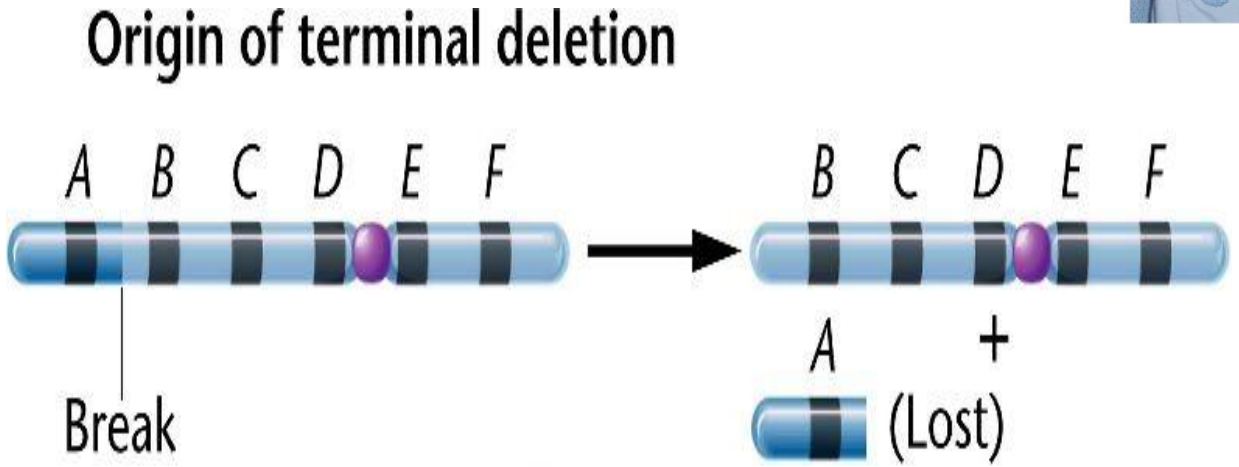
ii. Translocation – reciprocal or Robertsonian

Structural aberrations - Deletion



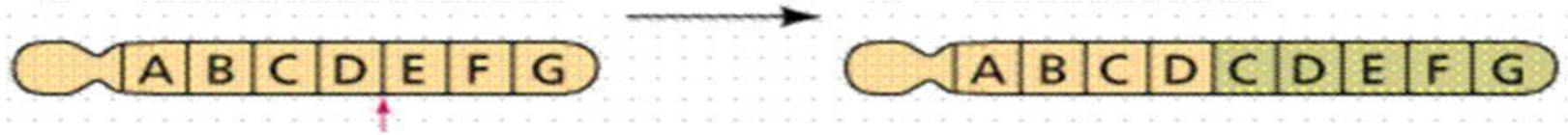






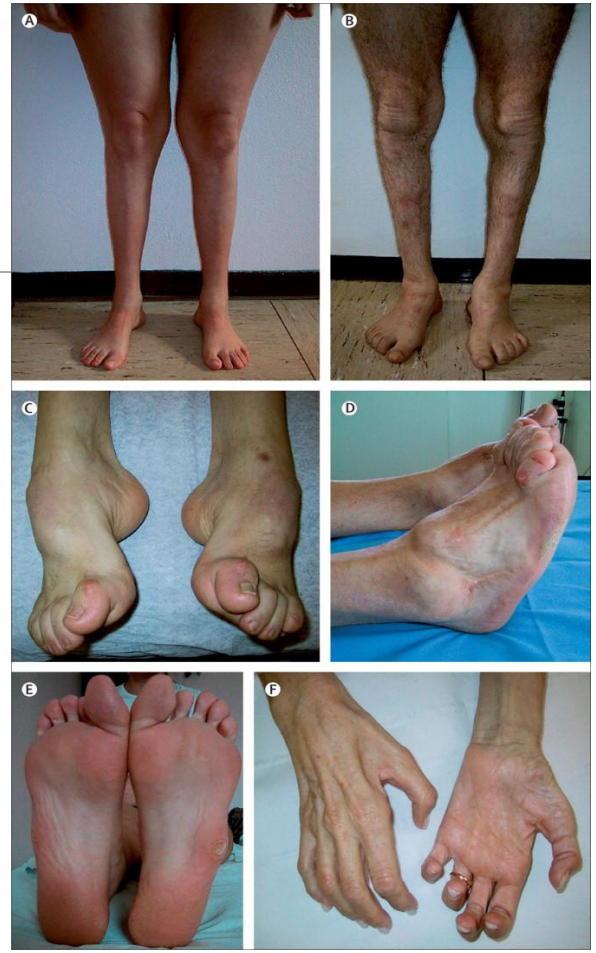
Structural aberrations - Duplication

Chromosomal Duplication

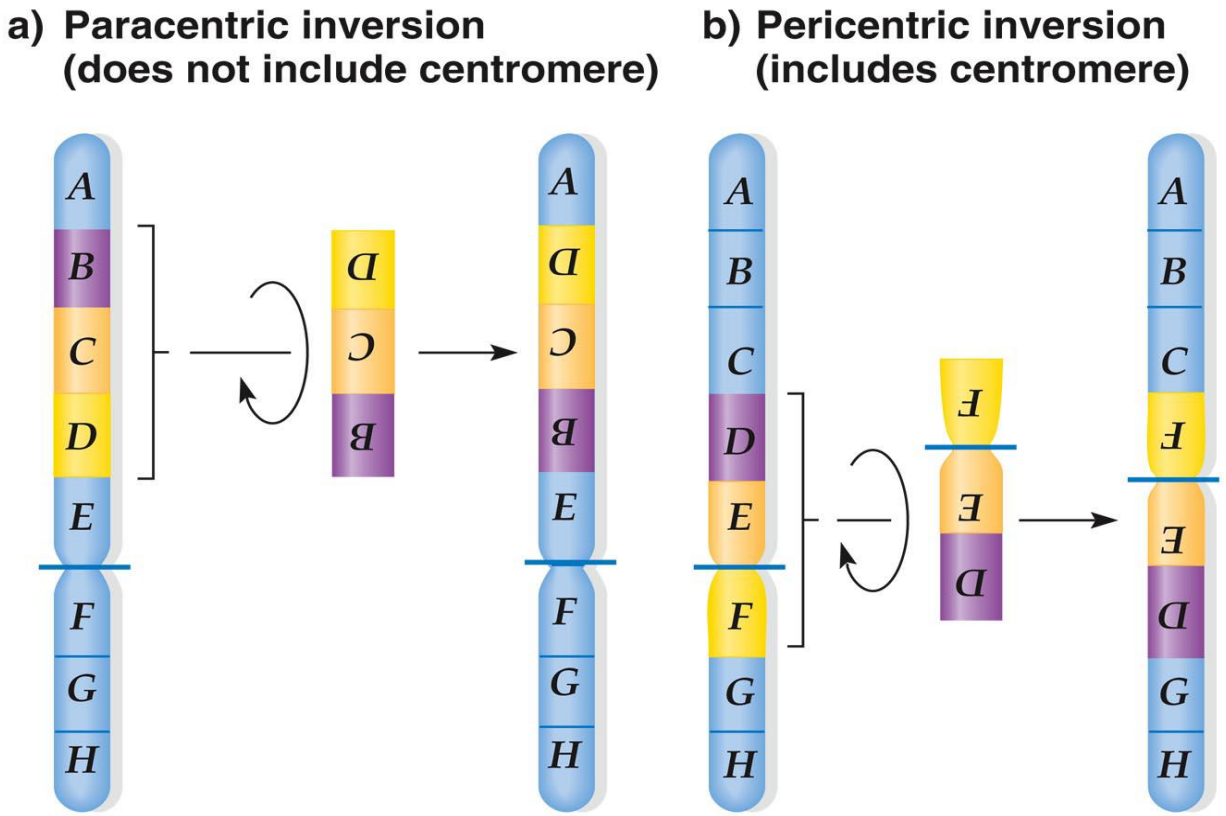


A segment of genes is copied twice and added to the chromosome

Causes:
Charcot–Marie–Tooth disease
(high arched foot, claw feet, confined to a wheelchair)

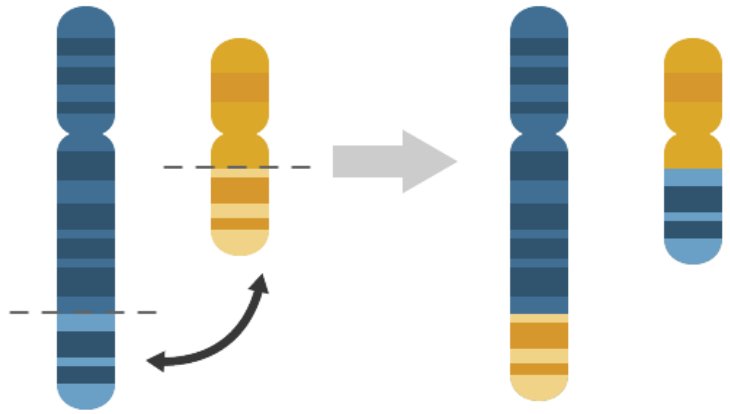


Structural aberrations - Inversion

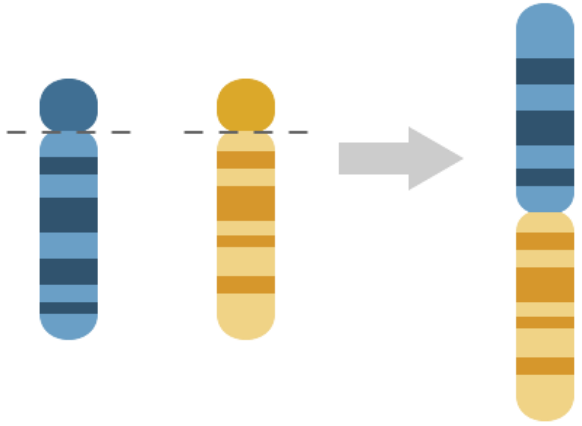


Structural aberrations - Translocation

Reciprocal translocation



Robertsonian translocation



Gene Mutations

A gene mutation is defined as an alteration in the sequence of nucleotides in DNA. This change can affect a single nucleotide pair or larger gene segments of a chromosome.

Types of mutations

Spontaneous mutations – these are mutations that occur under natural conditions without the action or effect of an external agent. They are presumably the ultimate source of natural genetic variation that is seen in populations.

Induced mutations – these involve the action of certain environmental agents, called mutagens that increase the rate at which mutations occur.

Gene Mutations - Classifications

- Point mutations
- Base-pair insertions or deletions.

Point mutations (base-pair substitution)

This type of mutation changes a single nucleotide base pair. There are two subtypes:

Transition - is the replacement of a base by the other base of the same chemical category (purine replaced by purine; pyrimidine replaced by pyrimidine).

For example, G--C →A--T A--T→G--C C--G→ T--A T--A → C--G

Transversion - is the replacement of a base of one chemical category by a base of the other (pyrimidine replaced by purine: T to G; purine replaced by pyrimidine).

For example, A--T→ C--G or T--A T--A → G--C or A--T C--G → G--C or A--T

G--C→ C--G or T—A

Categories of Point Mutations

Silent Mutation: The mutation changes one codon for an amino acid into another codon for that same amino acid. For example, if the DNA sequence **CGC** is changed to **CGA**, the amino acid arginine will still be produced.

Missense Mutation: This type of mutation alters the nucleotide sequence so that a different amino acid is produced altering the resulting protein. The change may not have much effect on the protein, may be beneficial to protein function, or may be dangerous. For example, if the codon for arginine **CGC** is changed to **GGC**, the amino acid glycine will be produced instead of arginine.

Nonsense Mutation: This type of mutation alters the nucleotide sequence so that a stop codon is coded for in place of an amino acid. A stop codon signals the end of the translation process and stops protein production. If this process is ended too soon, the amino acid sequence is cut short and the resulting protein is most always non-functional.

Base-pair insertion or deletion

Base insertion or deletion – The simplest of these mutations are single base-pair additions or single-base-pair deletions.

For example, AAGACTCCT→AAGAGCTCCT (addition)

AAGACTCCT→AAACTCCT (deletion)

Insertions and deletions can cause frame shift mutations when base pairs that are not a multiple of three are added to or deleted from the sequence.

Frameshift Mutation

Frameshift Mutation – This type of gene mutation alters the template from which amino acids are read.

Original Sequence: CGA-CCA-ACG-GCG...

Amino Acids Produced: Arginine - Proline - Threonine - Alanine
Inserted Base Pairs (GA): CGA-CCA-**GA**A-CGG-CG...

Amino Acids Produced: Arginine - Proline - Glutamic Acid - Arginine

Frameshift mutations typically result in complete loss of normal protein structure and function.

Assignment – Group work

With the aid of a chart, show a structured arrangement of the types of mutation.

QUANTITATIVE INHERITANCE

The phenotypic traits of different organisms may be of two kinds viz., qualitative and quantitative.

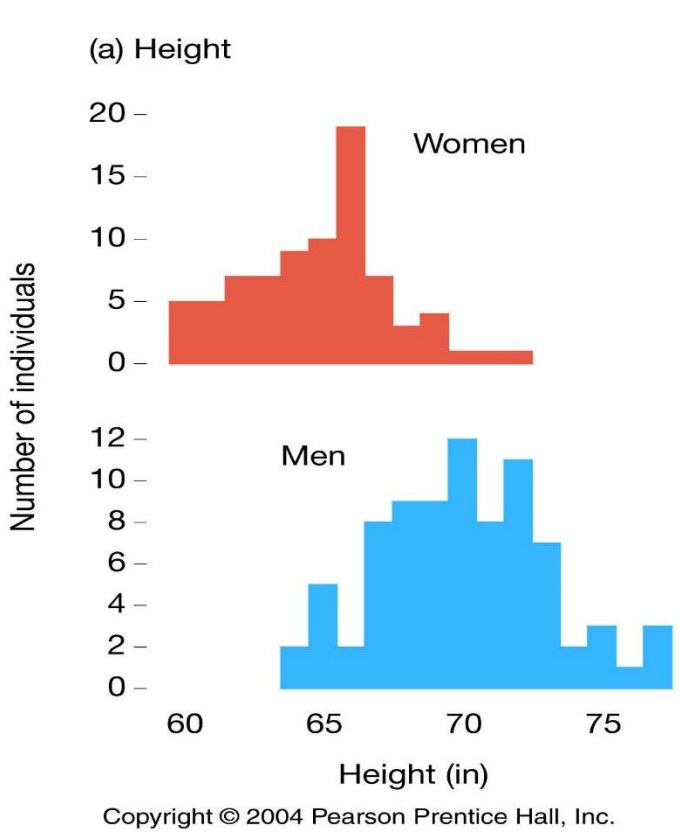
Qualitative traits bear distinctive classes of phenotypes and are controlled by two or many alleles of a single gene with little or no environmental modifications.

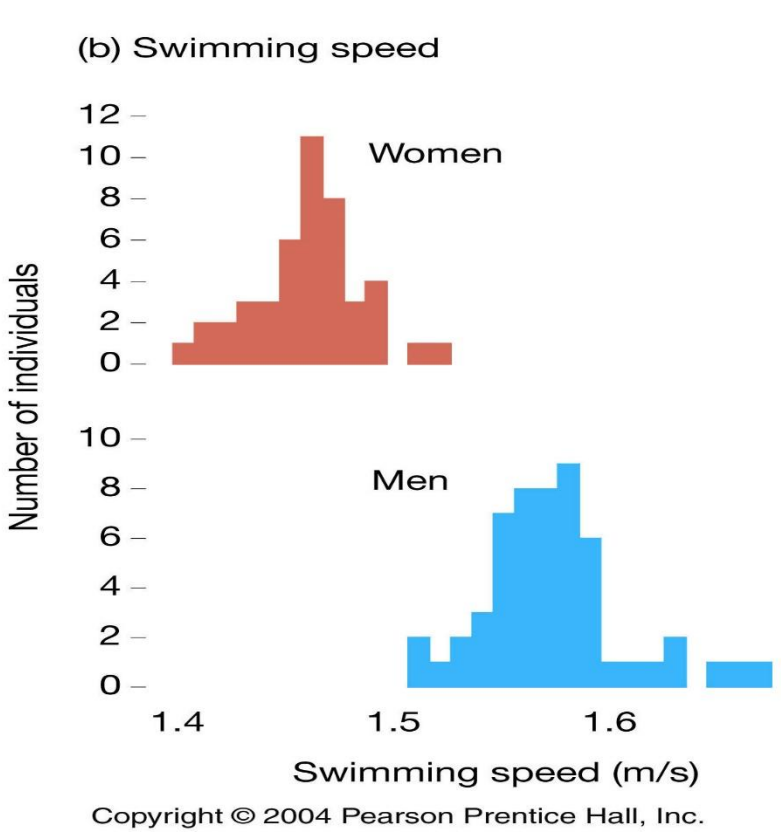
Quantitative inheritance involve the contributions of many different genes and are often influenced by environmental factors.

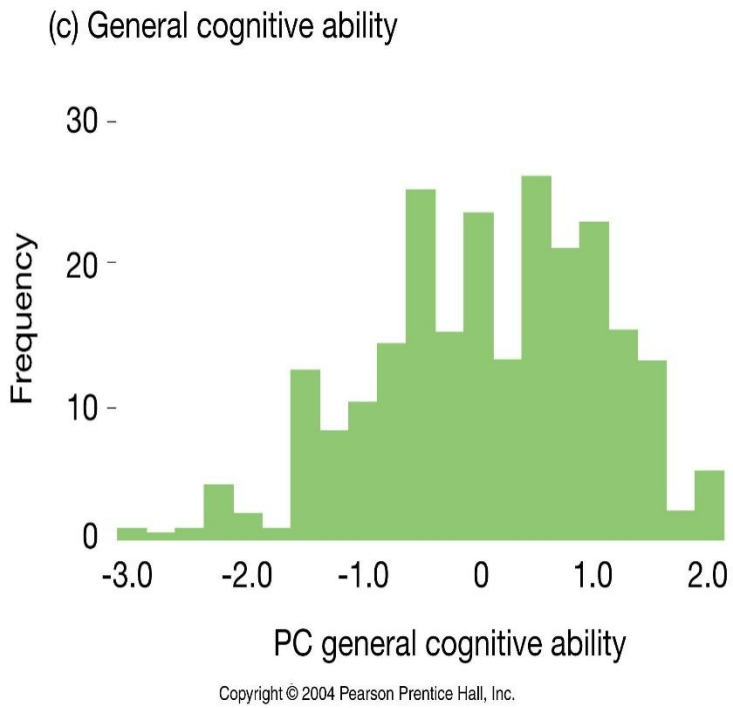
In other words, the phenotype of an individual depended on its genotype at all the relevant loci with each allele adding (or subtracting) a small amount.

Quantitative Inheritance is also referred to as **cumulative gene action** or **polygenic inheritance**.

Some quantitative traits in humans







Assumptions

1. The effect of each contributing alleles on the phenotype is equal .
2. The effects of the contributing alleles are additive.
3. There is no dominance at each locus; one allele does not obscure the effect of the other alleles and the effect of homozygosity of alleles is greater than that of heterozygosity of allele.

   *Different genotypes will have the same phenotype if the total number of contributing alleles is the same in all the genotypes.
4. There is no epistasis (masking effect of one gene locus upon the expression of another). It is assumed that the genotype at one locus does not mask the effect of the genotype at another locus. In the absence of epistasis there is a direct correspondence between the genotype and the phenotype.

Assumptions

5. There is no linkage among the loci controlling the trait.

6. The environment has no effect on the genotype. In other words the phenotype is entirely attributable to the genotype.

*Certainly this assumption is a great simplification of the true situation. For example, malnourishment is known to affect height and weight.

*However, the environmental effect is not assigned any value because its true or even approximate value is variable and difficult to quantify consistently, ignoring it therefore, greatly facilitates quantification of the effect of the alleles involved.

Examples of Quantitative Inheritance

1. Kernel Colour in Wheat
   a) One – locus control
   b) Two – locus control
   c) Three – locus control
   d) Multilocus control
2. Skin Colour in Human Beings

One – locus control

P_1	:	Red	X	White
		AA	↓	aa
F_1	:		Intermediate colour	
			Aa	
F_2	:		1 Red : 2 Intermediate : 1 White	
			1AA : 2 Aa : 1aa	

Two – locus control

P_1	:	dark red	X	white
		AABB	↓	aabb
F_1	:		AaBb	
			medium red	

F_2 :	**AB**	**Ab**	**aB**	**ab**
AB	AABB dark red	AABb medium dark red	AaBB medium dark red	AaBb medium red
Ab	AABb medium dark red	AAbb medium red	AaBb medium red	Aabb light red
aB	AaBB medium dark red	AaBb medium red	aaBB medium red	aaBb light red
ab	AaBb medium red	Aabb light red	aaBb light red	aabb white

Three – locus control

P_1 : Red X White

AABBCC aabbcc

F_1 : Intermediate colour

AaBbCc

F_2 :

	ABC	**ABc**	**AbC**	**aBC**	**Abc**	**aBc**	**abC**	**abc**
ABC	6	5	5	5	4	4	4	3
ABc	5	4	4	4	3	3	3	2
AbC	5	4	4	4	3	3	3	2
aBC	5	4	4	4	3	3	3	2
Abc	4	3	3	3	2	2	2	1
aBc	4	3	3	3	2	2	2	1
abC	4	3	3	3	2	2	2	1
abc	3	2	2	2	1	1	1	0

Skin Colour in Human Beings

P_1 : AABB (Negro) X aabb (White)

P_1 gametes: AB, ab

F_1 : AaBb

mulattoes, intermediate skin colour

Intercross: AaBb (mulattoes) X AaBb (mulattoes)

F_2 :	**AB**	**Ab**	**aB**	**ab**
AB	AABB	AABb	AaBB	AaBb
	like negro	darker than mulattoes	darker than mulattoes	like mulattoes
Ab	AABb	AAbb	AaBb	Aabb
	darker than mulattoes	like mulattoes	like mulattoes	lighter than mulattoes
aB	AaBB	AaBb	aaBB	aaBb
	darker than mulattoes	like mulattoes	like mulattoes	lighter than mulattoes
ab	AaBb	Aabb	aaBb	aabb
	darker than mulattoes	lighter than mulattoes	lighter than mulattoes	like white

Multilocus control

Number of pairs of genes	Fraction of F_2 like either parent	Number of genotypic class in F_2
1	1/4	3
2	1/16	9
3	1/64	27
n	$(1/4)^n$	3^n

Realised Heritability

This implies the degree to which phenotypic variation among individuals is due to genetic differences.

It also means the degree to which offspring resemble their parents.

Heritability is defined in the following equation:

$$h^2 = \frac{Y_0 - \bar{Y}}{Y_p - \bar{Y}} = \frac{\text{gain}}{\text{selection differentiated}}$$

where h^2 = heritability, Y_0 = offspring yield, $\bar{Y}$ = mean yield of the population, Y_p = parental yield.

* A low h^2 (< 0.01) occurs when the offspring of the selected parents differ little from the original population while a high h^2 (> 0.6) occurs when the offspring of the selected parents differ from the original population.

Example

A farmer wants to increase the average body weight in a herd of cattle. She begins with a herd having a mean weight of 595 kg and chooses individuals to breed that have a mean weight of 625 kg. Twenty offspring were obtained, having the following weights in kilograms: 612, 587, 604, 589, 615, 641, 575, 611, 610, 598, 589, 620, 617, 577, 609, 633, 588, 599, 601, and 611. Calculate the realized heritability in this herd with regard to body weight.

Ans. 0.3

Introduction to Population Genetics

All the individuals of a species constitute a **population.** The genetic of the inheritance of phenotypic traits in a given population is called **population genetics.** Certain fundamental aspects of population genetics are:

- Mendelian population
- Gene pool and gene frequency
- Chance mating or Panmixis
- Hardy-Weinberg Law

Mendelian Population

This is a group of sexually reproducing organisms with a relatively close degree of genetic relationship (such as species, subspecies, breed, variety, strain, etc.) residing within defined geographical boundaries where interbreeding occurs.

Gene pool and gene frequency

The percentages of gametes in the gene pool for a pair of alleles (A and a) depend upon the genotypic frequencies of the parental generation whose gametes form the pool. Proposed hypoyheses are:

Classical hypothesis – this proposes that the gene pool of a population consists of a wild-type allele at each gene locus with a frequency approaching one. Mutant alleles in very low frequencies may also exist at each locus

Balance hypothesis – there is generally no single wild-type or ‘normal’ allele. The gene pool of a population is envisioned as consisting at most loci an array of alleles in moderate frequencies. A typical individual is heterozygous at a large proportion of its gene loci.

Chance Mating or Panmixis

Every male gamete in the gene pool has an equal opportunity of uniting with every female gamete. Zygotic frequencies expected in the next generation from such random gametic unions may be predicted from a knowledge of the allelic frequencies in the gene pool of the parental population.

For example, if p stands for percentage of A alleles in the gene pool and q stands for the percentage of a alleles, the checkerboard of both alleles may predict possible chance combinations of A and a gametes as follows:

$(p+q)^2 = p^2 + 2pq + q^2$

	p(A)	q(a)
p(A)	(AA) p^2	(Aa) pq
q(a)	(Aa) pq	(aa) q^2

Chance Mating or Panmixis

The zygotic combinations predicted in a randomly mating population may be represented by

$$p^2:2pq:q^2$$

where p^2 represents the AA genotype,

2pq the Aa

q^2 the aa genotype

$p^2+2pq+q^2 = 1$.

When only two alleles are involved, and, therefore, p and q represent the frequencies of all of the alleles concerned, p+q=1.

Hardy-Weinberg Law

- This is also known as the Hardy Weinberg principle, equilibrium, model, theorem, or law.

- This states that **allele and genotype frequencies in a population will remain constant from generation to generation in the absence of other evolutionary influences.**

- The $(p+q)^2 = p^2 + 2pq + q^2$ expresses the genotypic expectations of progeny in terms of gametic or allelic frequencies of the parental gene pool.

Assumptions of Hardy-Weinberg

Random mating – the first assumption is random mating. Individuals in the population mate randomly with respect to the locus in question. Deviations from random mating alter genotypic frequencies but not allelic frequencies.

Large population size – the second assumption is that the population size is infinitely large. The larger the sample of successful gametes, the greater the probability that the allelic frequencies of the offspring will accurately represent allelic frequencies in the parental population.

No mutation or migration – the third and fourth assumptions are that neither mutation (the origin of new alleles) nor migration (the movement of individuals and their genes into or out of the population) is introducing new alleles into the population.

No natural selection – the final assumption is that no individual will have a reproductive advantage over another individual because of its genotype.

Example 1

Obviously, some of these assumptions will not hold in real biological situations.

For example, consider a sample of 100 individuals with the following genotype frequencies:

	Observed Genotype Frequencies	Allele count	Allele frequency	Expected genotype frequencies under H-W
BB	0.71	142 B	p = 156/200 = 0.78	$p^2 = (.78)^2 = 0.61$
Bb	0.14	14 B, 14 b		$2pq = 2(.78)(.22) = 0.34$
bb	0.15	30 b	q=44/200=0.22	$q^2 = (.22)^2 = 0.05$

*Note that the observed are different from expected, thus some force must be at work to change frequencies.

Example 2

Albinism is a rare genetically inherited trait that is only expressed in the phenotype of homozygous recessive individuals (aa). The average human frequency of albinism in North America is 1 in 20,000. Calculate the frequencies for each of the three genotypes for this trait in the population.

Using Hardy-Weinberg equation,

$p^2 + 2pq + q^2 = 1$ where q^2 is the frequency of homozygous recessive individuals (aa).

$q^2 = 1/20000 = 0.00005$

$q = 0.007$ i.e. the frequency of the recessive albinism allele (a) is 0.00707 or about 1 in 140.

$p = 1-q$

Example 2

p = 1 – 0.007

p = 0.993 i.e. the frequency of the dominant normal allele is 0.99293

Substitute into Hardy Weinberg equation:

$p^2 + 2pq + q^2 = 1$

$(0.993)^2 + 2(0.993)(0.007) + (0.007)^2 = 1$

0.986 + 0.014 + 0.00005 = 1

p^2 = predicted frequency of homozygous dominant individuals = 0.986 = 98.6%

2pq = predicted frequency of heterozygous individuals = 0.014 = 1.4%

q^2 = predicted frequency of homozygous recessive individuals = 0.00005 = 0.005%

Factors influencing allele frequency or deviations from Hardy-Weinberg equilibrium

Selection - Selection occurs when individuals of different genotype leave different numbers of progeny because they differ in their probability to survive to reproductive age (viability), in their mating success, or in their average number of produced offspring (fertility).

Mutation - Mutations are the ultimate source of genetic variability, and form the raw material upon which selection can act. Typically, spontaneous mutation rates per locus per generation are of the order of 10^{-4} to 10^{-6}, and genomic mutation rates summed over all loci may be on the order of one per generation, but can vary substantially between species.

Meiotic drive - crossing over between two homologous chromosomes may occur. This process is called recombination and has the potential to combine favourable alleles of different ancestry in one gamete and to break up combinations of deleterious alleles.

Factors influencing allele frequency or deviations from Hardy-Weinberg equilibrium

Migration – It is similar to mutation in that it adds or removes alleles and thereby changes allelic frequencies.

Random genetic drift – random fluctuation in allele frequencies or gene variants. The effect of genetic drift is negligible in large populations but in small breeding populations all the limited number of progeny might be of the same type.

Founder principle – when a few individuals or a small group migrate from a main population, only a limited portion of the parental gene pool is carried away. In the small migrant group, some genes may be absent or occur in such low frequency that they may be easily lost. For example, North American Indian tribes, for the most part, lack the gene IB that controls type B blood. However, in Asia, the ancestral home of the American Indians, the IB gene is widespread.