Deviations from Mendel's laws

Contents: Mendel's 1st law: The law of segregation; Deviations from the law of segregation: Examples; Mendel's 2nd law: The law of independent assortment; Deviations from the law of independent assortment: Examples; Conclusions; Suggested reading.

Mendel's 1st Law: The law of segregation

During the formation of gametes, the unit 'factors' (= genes) are separated so that each gamete receives only one 'factor' for a given pair.

Examples:

1. Monohybrid cross	2. Dihybrid cross	3. Trihybrid cross
P: TT × tt	TTRR × ttrr	TTRRYY × ttrryy
G: T, t	TR, tr	TRY, try
F_1: Tt	TtRr	TtRrYy
$F_1 \times F_1$: Tt × Tt	TtRr × TtRr	TtRrYy × TtRrYy
G: T, t	TR, Tr, tR, tr	TRY, Try, TrY, Try, tRY, trY, tRy, try

Conclusion:

(a) Mendel's 1st law is nothing but the process of gametogenesis;

(b) The law is universally applicable to all sexually reproducing diploid (2n) organisms except where non-disjunction and meiotic drive take place.

Deviations from Mendel's 1st law

1. Non-disjunction

The failure of homologous chromosomes to separate at meiosis (usually at anaphase of meiosis I or meiosis II) is called non-disjunction. It may occur in both autosomes and sex chromosomes.

Types of non-disjunction:

Primary non-disjunction: *e.g.* non-disjunction occurring in XX♀ or XY♂; and Secondary non-disjunction: *e.g.* non-disjunction in XXX♀ or XXY♂.

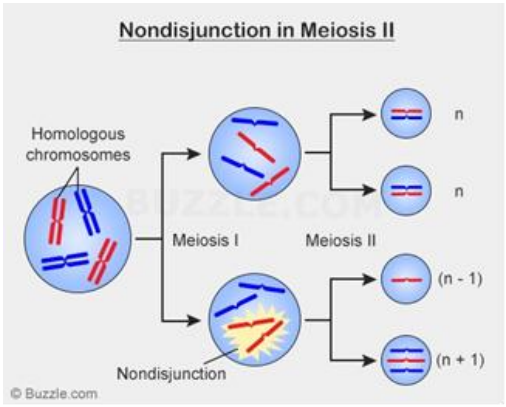

Fig. 4.1 showing the processes of non-disjunction in meiosis I and meiosis II

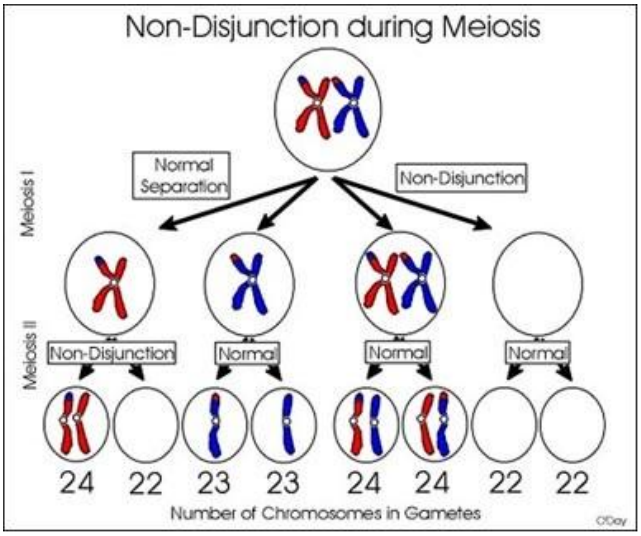

Fig. 4.2 showing the processes of non-disjunction in meiosis I and meiosis II.

Some examples of the consequences of non-disjunction in man

Examples of autosomal non-disjunction:

D(13) trisomy or Patau's syndrome; E(18) trisomy or Edwards' syndrome; G(21) trisomy or Down's syndrome etc.

Examples of sex-chromosomal non-disjunction:

47, XXX trisomy or triplo-X female; 47, XXY trisomy or Klinefelter's syndrome; 45, XO monosomy or Turner's syndrome etc.

2. Meiotic drive

Unequal recovery of homologous chromosomes at meiosis is known as meiotic drive.

Examples include:

(a) Segregation distorter (SD) gene in *Aedes aegypti;*

(b) Recovery disrupter (RD) gene in *Drosophila melanogaster*

Segregation distorter (SD) gene in *Aedes aegypti*

SD gene in *A. aegypti* gives rise to a male biased sex-ratio where >95% males and <5% females are produced, although the normal sex-ratio is 50: 50.

Fig. 4.3 Yellow fever mosquito *Aedes aegypti* in which SD gene brings about sex-ratio distorsion by producing >95% males and <5% females

Recovery disrupter (RD) gene in *Drosophila melanogaster*

RD gene in *D. melanogaster* also gives rise to male biased sex-ratio where >65% males and <35% females are produced although the normal sex-ratio is 50: 50.

Fig. 4.4 The fruit fly *Drosophila melanogaster* in which RD gene brings about sex-ratio distorsion where >65% males and <35% females are produced

Recent reports suggest that meiotic drive of Y chromosome leads to male-biased sex ratio while meiotic drive of X chromosme leads to female-biased sex ratio in many insects such a Diptera. However, the X chromosme drive is more common than the Y chromosome drive.

Deviations from Mendel's 2nd law

Mendel's 2nd Law: The law of independent assortment

The law states that when two or more pairs of 'factors' are involved, they assort or combine at random during the formation of gametes.

Limitation of Mendel's 2nd law

Mendel's 2nd law is only applicable to cases where genes are located on separate pairs of homologous chromosomes. Examples are shown below:

1. Dihybrid crosses:
e.g. TTRR ×ttrr (T/R and t/r genes are located on separate chromosomes)
F_2 ratio: (3: 1) × (3: 1) = 9: 3: 3: 1

2. Trihybrid crosses:
e.g. TTRRYY ×ttrryy (T/R/Y and t/r/y genes are located on separate chromosomes)
F_2 ratio: (3: 1) × (3: 1) × (3: 1) = 27: 9: 9: 3: 9: 3: 3: 1

3. Polyhybrid crosses:
e.g. n × (Dominant genes) × n × (recessive genes)
F_2 ratio: n × (3: 1)

However, the following cases DO NOT follow Mendel's 2nd law:

1. **Linkage and crossing-over** (when genes are located on the same chromosome);
2. **Polygenic inheritance** (when more than two genes control a trait);
3. **Pleiotropic effects or pleiotropism** (when a single gene has more than one phenotypic effect)

Examples of linkage and crossing-over

Example 1 Dihybrid cross in sweet peas, *Lathyrus odoratus* by Bateson and Punnett (1906).

P: Purple flower-Long pollen grain × red flower-round pollen grain
(PPLL) (ppll)

F_1: All Purple-Long (PpLl)

$F_1 \times F_1$: PpLl × PpLl

F_2:	P-L-	=	7/16	(Purple-Long)
	P-ll	=	1/16	(Purple-round)
	ppL-	=	1/16	(red-Long)
	ppll	=	7/16	(red-round)

Note: Instead of 9: 3: 3: 1, Bateson and Punnett (1906) observed 7: 1: 1: 7 ratios in F_2 generation in the sweet peas.

Fig. 4.5 The sweet pea plant *Lathyrus odoratus*

Bateson and Punnett (1906) proposed the terms '**coupling**' for the parental types (P-L- and ppll) and '**repulsion**' for the recombinant types (P-ll and ppL-).

Example 2 Dihybrid cross in fruit fly, *Drosophila melanogaster* by T. H. Morgan (1910).

P: Grey body-Long wing × black body-vestigial wing
(GGLL) (ggll)

F_1: All Grey-Long (GgLl)

$F_1 \times F_1$: GgLl × GgLl

F_2:	G-L-	=	41%	(Grey-Long)
	G-ll	=	9%	(Grey-vestigial)
	ggL-	=	9%	(black-Long)
	ggll	=	41%	(black-vestigial)

Note: Instead of 9: 3: 3: 1, Morgan (1910) observed 6.56: 1.44: 1.44; 6.56 ratio, which are approximately 7: 1: 1: 7.

Fig. 4.6 Gray-bodied and long-winged (bottom) and black-bodied and vestigial-winged *Drosophila melanogaster*

Morgan (1910) proposed the terms '**linkage**' for the parental types (G-L- and ggll) and '**crossing-over**' for the recombinant types (G-ll and ggL-), replacing the terms 'coupling' and 'repulsion' proposed earlier by Bateson & Punnett (2006. Later in 1942, however, J. B. S. Haldane proposed *cis*-arrangement and *trans*-arrangement for such alternative configurations at DNA level.

An example of polygenic inheritance in man

Polygenes: When more than two pairs of genes control a single trait like height, weight and skin colour, the genes are referred to as polygenes. The term was proposed by Kenneth Mather (1941) by supplanting the older terms 'multiple factors' or 'multiple genes'. Here, inheritance of the skin colour in man, a polygenic trait, is explained. Suppose, skin colour in man is controlled by 3 pairs of genes:

P: White (Caucasian) man (○○○ ○○○) × black (Negroid) woman (● ● ● ● ● ●)

F_1: All Mulatto or brown-skinned offspring (○○○ ● ● ●)

$F_1 \times F_1$: Brown-skinned (○○○ ● ● ●) × Brown-skinned (○○○ ● ● ●)

F_2:

○○○ ○○○	● ○○ ○○○	● ● ○ ○○○	● ● ● ○○○	● ● ● ● ○○	● ● ● ● ● ○	● ● ● ● ● ●
1/64	6/64	15/64	20/64	15/64	6/64	1/64

Phenotypic ratio: 1: 6: 15: 20: 15: 6: 1

White Light… Brown … Dark Black

(Instead of classical trihybrid ratio 27: 9: 9: 3: 9: 3: 3: 1)

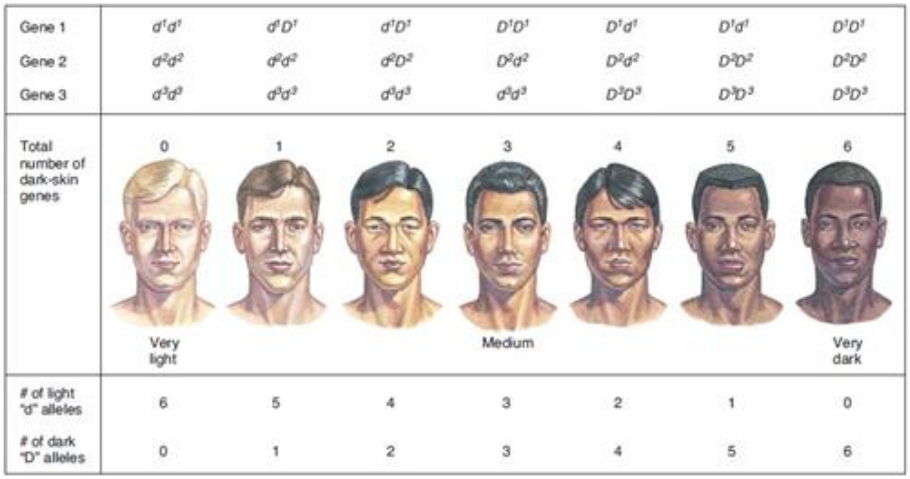

FIGURE 10.7 Polygenic Inheritance
Skin color in humans is an example of polygenic inheritance. The dark "D" alleles are found in several different genes and have an additive effect on skin color. The top portion of the figure shows examples of genotypes that can produce the different skin colors. The number of dark "D" alleles is more important than how the "D" alleles are distributed in the different genes.

Fig. 4.7 showing the polygenic inheritance of skin colour in man involving 3 pairs of genes (=6 alleles) where the F_2 ratio is 1: 6: 15: 20: 15: 6: 1, ranging from white (very light) through medium (mulatto or brown) to black (very dark) phenotypes

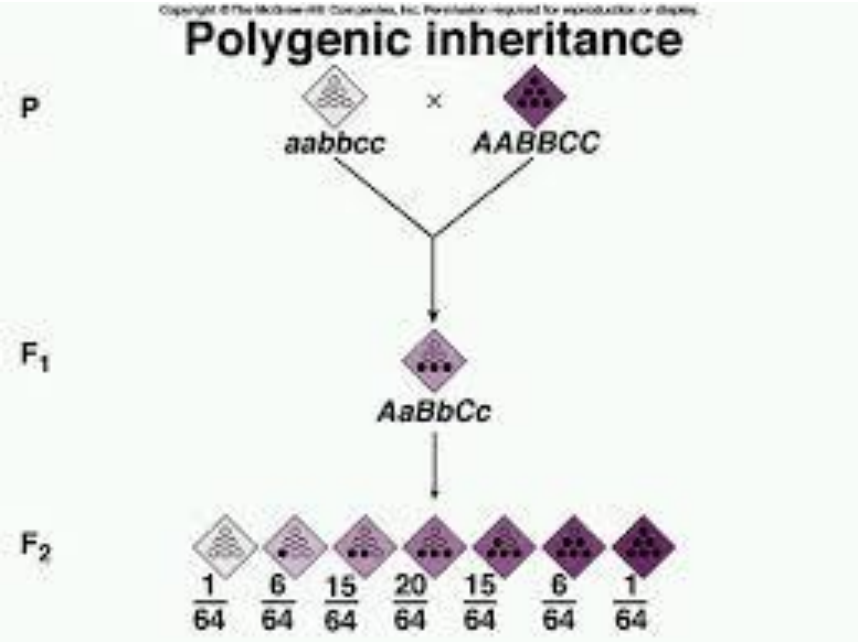

Fig. 4.8 Diagram showing the F_2 ratio (1: 6: 15: 20: 15: 6: 1) of a trihybrid cross involving polygenes, which is different from a similar cross involving major genes, where the F_2 ratio would have been 27: 9: 9: 3: 9: 3: 3: 1

Examples of pleiotropic effects or pleiotropism

Pleiotropic effect or pleiotropism refers to a case where a single gene has more than one phenotypic effect.

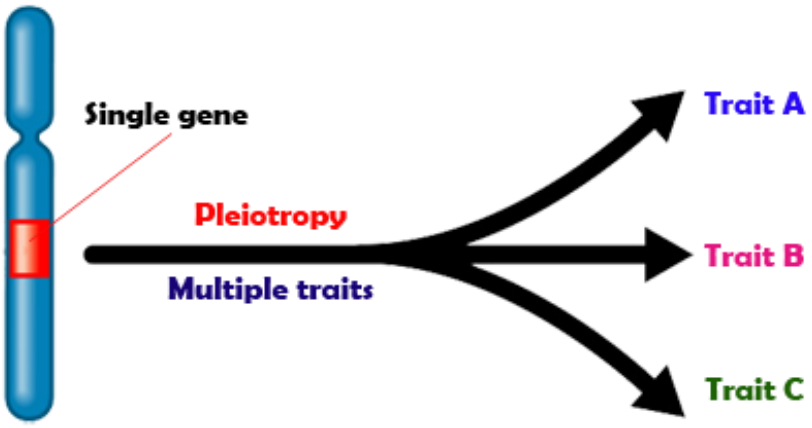

Fig. 4.9 Diagram showing a single pleiotropic gene affecting multiple traits

Examples of pleiotropic effects

Example 1 Vestigial wing (*vg* gene) in *D. melanogaster*

Note: vg gene is located on the 2nd chromosome at 67.0 locus.

The main effect of *vg* is small and wrinkled wings in both sexes of the fly. Other effects of the *vg* gene are:

(a) Modification of balancers (halteres);
(b) Reduced number of bristles;
(c) Reduced number of eggs in ovaries;
(d) Reduced size of the spermatheca in females; and
(e) Reduced longevity.

Example 2 Hb^S gene in man

Note: Mutation of Hb^A gene gives rise to Hb^S gene, resulting in sickle-shaped RBC and sickle-cell anaemia in man (see Fig 4.9).

The main effect of the Hb^S gene is sickle-shaped RBC and sickle-cell anaemia in man. Other effects of the Hb^S gene are:

(a) Defective heart, bones, lungs, kidneys and spleen;
(b) Defective vision;
(c) Resistance to falciparum malaria; and
(d) Inflammation of the blood vessels etc.

Other examples of pleiotropism in man:

1. Alkaptonuria in man: This is due to mutation of a single gene on chromosome 3, that blocks the breakdown of tyrosine (an essential amino acid), resulting in black urine in the newborn babies. In addition to this defect, alkaptonuria also produces darkened skin and darkened sclera of the eyes in the affected babies.
2. Phenylketonuria (PKU) in man: It is caused by a single gene mutation that fails to code for an enzyme (phenylalanine hydroxylase) which converts phenylalanine (another essential amino acid) to tyrosine. As a result, phenylalanine concentrations in the blood and urine increase to toxic levels, causing damage at several locations in the body. The pleiotropic effects of the gene include: mental retardation, reduced hair and skin pigmentation, blonde hairs and weakened ankle bones.

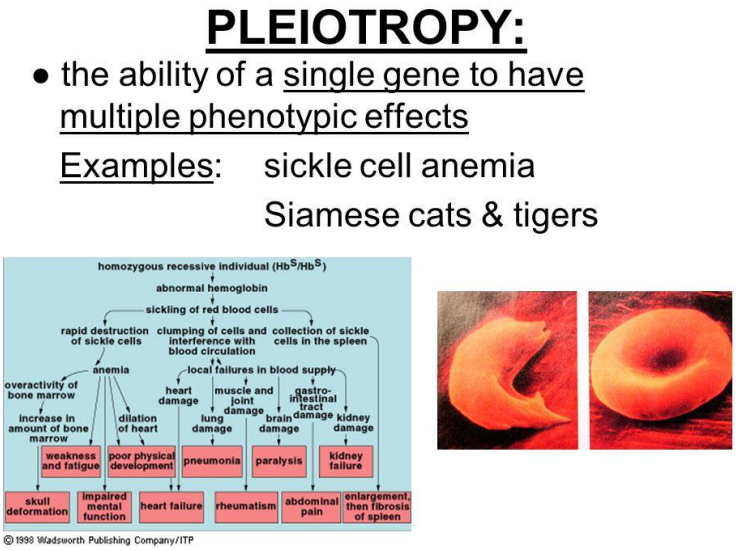

Fig. 4.10 Diagram showing the pleiotropic effects of the Hb^S gene (left), and the sickling of RBC compared to the normal RBC (right) in man. Sickle-cell anaemia, however, is also found in siamese (identical twins) cats and tigers

Conclusions on deviations from Mendel's laws

1. Mendel's 1st law refers to gametogenesis and it is applicable to all sexually reproducing diploid organisms;

2. Deviations from Mendel's 1st law may occur due to abnormalities like non-disjunction and meiotic drive;

3. Mendel's 2nd law is only applicable to cases where genes are located on separate chromosomes; and

4. Deviations from Mendel's 2nd law may take place where genes are located on the same chromosomes showing linkage, or many genes (polygenes) that control a single trait, or a single gene (*i.e.* pleiotropic gene) that controls more than one phenotypic trait.

Suggested reading:

Ayala & Kiger, 1980.
Burns, GW. 1980.
Gardner *et al*. 1991.
Islam, MS. 2018.
Sinnott *et al*. 1973.
Winchester, AM. 1966.
Internet sources, 2018.
ইসলাম, ম.সা., খান, হা.সা. ও রানা, ম.হা.তা. ২০১৭।