Gregor Mendel

An Austrian monk who first discovered the laws of genetics

• Mendel's symbols for crossing (breeding) experiments:

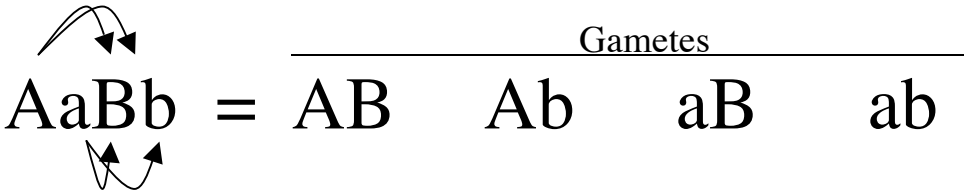
Symbol	Generation
P	parents
F_1	offspring from parents
F_2	offspring from F_1

Figs 14.1 and 14.3

Mendel's rules of genetics

- Each trait is controlled by a gene
- Each gene can exist in two forms (two alleles): Dominant or recessive
- Each organism has two of each gene: One maternal (inherited from mother) and one paternal (inherited from father)
- When an organism makes gametes, each gamete receives only one of each gene (The Law of Segregation)
    - √ Half the gametes receive the material gene and half receive the paternal gene
- Gametes with all possible combinations of maternal and paternal genes are made, and all combinations are made with equal frequencies (The Law of Independent Assortment of Genes)

Example: Diploid cell with AaBb genotype



Dominant allele

An allele that is expressed (controls the trait) whenever it is present.

- Symbol = An upper case letter

Recessive allele

An allele that only is expressed when the dominant allele is not present.

- Symbol = The lower case letter of the dominant allele letter

What makes alleles dominant or recessive?

- Dominant alleles usually encode functional proteins
- Recessive alleles are usually alleles that encode non-functioning proteins.

Genotype

The alleles present in an organism's genes

Phenotype

The physical traits of an organism.

Genotype	Phenotype
PP (Homozygous dominant)	Dominant
Pp (Heterozygous)	Dominant
pp (Homozygous recessive)	Recessive

Homozygous is also known as "true-breeding"
Heterozygous is also known as "hybrid"

Figs 14.3 and 14.6

Punnett square

A table used for diagramming genetic crosses

- All possible gamete genotypes from one parent go on top. All possible gamete genotypes from the other parent go on the left

- The squares in the table show the possible genotypes the offspring may have.

- The Punnett square is used to predict the chance that any one offspring will have a certain genotype:

$$\frac{\text{Number of squares with one genotype}}{\text{Total number of squares}} = \text{Chance offspring will have the genotype}$$

- Note: Multiply the chance of each individual genotype to find the chance of an offspring with all those genotypes

Some common Punnett squares:

Name:	Phenotype ratios:	Genotype ratios:
Monohybrid cross (Aa x Aa)	3 dom :1 rec	1AA : 2:Aa : 1aa
Test cross (AA x aa)	All dom	All Aa
…or (Aa x aa)	1 dom : 1 rec	1Aa : 1aa
Dihybrid cross (AaBb x AaBb)	9 domA domB: 3 domA recB : 3 recA domB : 1 recA recB	1AABB : 2AABb 1AAbb : 2AaBB 4AaBb : 2Aabb 1aaBB : 2aaBb 1aabb

For crosses with three or more genes, the chances of offspring with a certain genotype can also be calculated without a large Punnett square:

Example: Calculate chance of aaBbCc offspring from a AaBBCc x Aabbcc cross

1) Use small Punnett squares to find the odds of each genotype in the offspring, one gene at a time

aa (from Aa x Aa) = 1/4

Bb (from BB x bb) = 1

Cc (from Cc x cc) = 1/2

2) Multiply the odds of all genotypes together

1/4 x 1 x 1/2 = 1/8 chance of aaBbCc offspring

Common human dominant and recessive traits:

Dominant	Recessive
Brown eyes	Blue eyes
Unattached earlobes	Attached earlobes
Widows peak hairline	Straight hairline
Hair on fingers	No hair on fingers
Can roll tongue	Can't roll tongue

Genetic disorders ("Inborn errors of metabolism")

A medical problem caused by the genes.

- Most genetic disorders are recessive and are caused by inheriting two recessive (non-functioning) copies of a gene needed for proper health.

  √ Victims usually are born to healthy parents who are carriers (heterozygous) for the disorder

- Dominant disorders are extremely rare

  √ Because they are rare, individuals with the disorder are almost always heterozygous (not homozygous dominant)

Examples of recessive genetic disorders:

Phenylketonuria (PKU)

Lack of the enzyme to metabolize the amino acid Phenylalanine

- Causes newborns to develop mental retardation unless they are put on a special diet low in Phenylalanine

Cystic fibrosis

A lethal disease caused by lack of a membrane transport protein for Cl^- ions

- Causes a thick mucus to clog the respiratory and digestive systems of children. Most victims die before their 20's

Tay-Sachs

A lethal disease caused by the lack of an enzyme that metabolizes a certain lipid in nerve cells

- Causes seizures, paralysis, and death of the child in just a few years after birth

Example dominant genetic disorders:

Huntington's Disease

A lethal disorder that causes the nervous system to degenerate, beginning at middle age.

Polydactyly

A disorder that causes extra fingers or toes

Achondroplasia

A type of dwarfism

Fig 14.15

Pedigree analysis

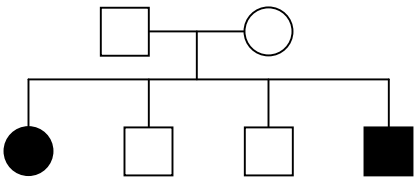
Constructing a chart of offspring and parent relationships in a family to determine the genotypes of family members.

• Some symbols used in pedigree analysis:

○ = Female without trait ● = Female with trait

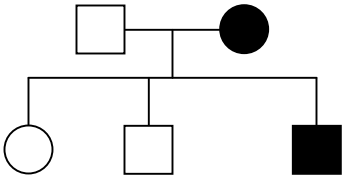
□ = Male without trait ■ = Male with trait

• Example:



Conclusion: The trait must be recessive. Both parents are Qq. Offspring with the trait are qq. Offspring without the trait are either QQ or Qq.

• Example:



Conclusions: If the trait is recessive, the father is Qq, the mother is qq. The offspring without the trait are Qq, and the offspring with the trait are qq.

If the trait is dominant, the father is qq, the mother is Qq. The offspring without the trait are qq, and the offspring with the trait are Qq.

Figs 14.14 and 14.16

Non-Mendelian genetics

Patterns of inheritance that do not follow Mendel's rules of genetics

- Co-dominance
- Incomplete dominance
- Continuous variation
- Sex-linked traits

Co-dominance

Two or more dominant alleles. Organisms having both dominant alleles fully show both traits

- Usually both co-dominant alleles encode differing but functional proteins

Examples of co-dominance

Blood type

I^A and I^B alleles (for blood cell antigens A and B) are both dominant alleles. The i allele (for no blood cell antigen) is recessive

Genotype	Phenotype (antigens on blood cells)
$I^A I^B$	AB
$I^A I^A$ or $I^A i$	A
$I^B I^B$ or $I^B i$	B
ii	O (no antigens)

Table 14.2

Sickle-cell anemia

The dominant allele Hb^A encodes normal hemoglobin protein (which carries O_2 inside red blood cells). The dominant allele Hb^S encodes sickle-cell hemoglobin, which causes blood cells to have a sickle shape

Genotype	Phenotype (blood cell shape)
$Hb^A Hb^A$	All normal shape
$Hb^A Hb^S$	Some normal shape, some sickled
$Hb^S Hb^S$	All sickled shape

Fig 23.13

Incomplete dominance

An allele that is not fully dominant. Organisms that are heterozygous show a weakened form of the dominant trait

- Usually incomplete dominance results when a single copy of the dominant allele makes too little of its protein for the full dominant phenotype.

<u>Examples of incomplete dominance</u>

Snapdragon flower color:

R allele causes red pigment. r allele is recessive (no pigment)

RR = Red
Rr = pink
rr = white

Fig 14.10

Hair shape:

W allele causes curling of hairs. w allele is recessive (no curling)

WW = Curly
Ww = Wavy
ww = straight

Skin color in human beings:

A allele causes melanin (brown pigment). a allele is recessive (no melanin)

AA = Dark skin
Aa = medium skin
aa = light skin

Continuous variation (Quantitative trait)

A trait that has a wide range of phenotypes

• Continuous variation usually results from many different genes contributing to a trait.

Examples of continuous variation

Human skin color

There are at least three genes (A, B, and C) for melanin. The total number of dominant alleles in all three genes determines the darkness of the skin

Number of Dominant alleles	Example	Skin color
6	AABBCC	Very very dark
5	AABbCC	Very dark
4	aaBBCC	Medium dark
3	AAbbCc	Medium
2	aaBbCc	Medium light
1	aabbCc	Very light
None	aabbcc	Very very light

Fig 14.12

Autosomes

The chromosomes that are the same in both sexes

Sex chromosomes

The chromosomes that differ between the sexes (The X and Y chromosomes)

• Women are XX, men are XY

Sex-linked genes

Genes located on the sex chromosomes (especially the X chromosome)

• Men are haploid for all genes on the X chromosome

• Men tend to show more recessive phenotypes for these genes

√ Men have no second X chromosome to mask recessive alleles

Fig 15.10

Examples of X-linked recessive genetic disorders:

Red-green color blindness

Inability to distinguish certain colors

- Caused by lack of a protein needed for perceiving certain colors

Hemophilia

Excessive bleeding from even minor cuts

- Caused by lack of a blood-clotting protein

Duchenne muscular dystrophy

A fatal muscle degenerative disease

- Caused by a defective Ca^{2+} transport protein in muscle cells

X-chromosome inactivation in females

Early in embryonic development, female embryos inactivate one of their two X chromosomes in each cell

- The inactive X chromosome becomes a small shrunken structure called a Barr Body

  √ Essentially no genes in the Barr body are expressed

- Which of the two X chromosomes is inactivated is random and differs from one embryonic cell to another

  √ Once an embryonic cell has inactivated one of its X chromosomes, all the cells that develop from that cell will inactivate the same X chromosome

- Therefore, adult female females are a "mosaic": Certain patches of tissue express one X chromosome and other patches of tissue express the other X chromosome

Fig 15.11

Karyotype

A display of chromosomes in a cell using a microscope

- Used to inspect for chromosomal abnormalities

Fig 13.3

Some genetic disorders are caused by aneuploidy (having the incorrect number of chromosomes)

- The offspring receive either one too many or one too few chromosomes from one parent

- Caused by non-disjunction of the chromosomes during gamete formation

Figs 15.12 and 15.15

Non-disjunction

When one daughter cell in meiosis receives two homologous chromosomes and the other daughter cell receives none

Fig 15.12

<u>Example of aneuploidy of an autosomal chromosome:</u>

Down syndrome (trisomey 21)

An abnormality characterized by mental retardation, heart defects, and other abnormalities

- Caused by inheriting an extra chromosome 21 from one parent
  - √ People with Down syndrome have 47 chromosomes (three chromosome 21)

- The older the mother is the larger her risk of Down syndrome children

Figs 15.15

Examples of aneuploidy of sex chromosomes

Turner syndrome (only one X chromosome, written "XO")

- People with Turner syndrome are female
  - √ Sterile because ovaries are missing
  - √ Female secondary sex characteristics (such as breast development at puberty) do not develop
  - √ Most have normal intelligence

Klinefelter syndrome (XXY)

- People with Klinefelter syndrome are male
  - √ Sterile because testes do not develop
  - √ Male secondary sex characteristics (such deep voice and male musculature at puberty) do not develop
  - √ Most have normal intelligence

XYY condition (Jacob syndrome)

- People with XYY condition are male
  - √ Normal intelligence, no significant abnormalities

Population genetics

The study of genes and alleles in entire populations

- Population = A group of organisms in one area that mate only with each other

Allele frequency

The portion of a particular allele compared to the total number of alleles for that gene within the population

- The sum of all allele frequencies for a gene equals 1
- If mating is random and both alleles of a gene are equally beneficial, the allele frequencies will stay the same from generation to generation

Example allele frequency calculation:

A certain population is made up of 100 individuals.

Each individual in the populations has two of each gene, therefore there are actually 200 alleles total in the population.

In this population of 100 individuals, there are 6 BB individuals, 38 Bb individuals, and 56 bb individuals.

- B_f (The allele frequency of the B allele) is 0.25
  - √ (12 B alleles from the 6BB individuals) + (38 B alleles from the 38Bb individuals) = 50 B alleles total in this population.
  - √ B_f = The portion of the B allele compared to the 200 alleles of the population = 50 B alleles/200 total alleles = 0.25.
- b_f (The allele frequency of the b allele) is 0.75
  - √ (112 b alleles from the 56BB individuals) + (38 b alleles from the 38Bb individuals) = 150 b alleles total in this population.
  - √ b_f = The portion of the b allele compared to the 200 alleles of the population = 150 b alleles/200 total alleles = 0.75.
- The sum of all allele frequencies for a gene equals 1
  - √ $B_f + b_f = 1$
  - √ $0.25 + 0.75 = 1$

Genotype frequencies

The portion of individuals having a particular genotype compared to the total number of individuals within the population

- The sum of all genotype frequencies equals 1

Example genotype frequency calculation:

A certain population is made up of 100 individuals.

In this population, there are 6 BB individuals, 38 Bb individuals, and 56 bb individuals.

- Homozygous Dominant frequency (HD) = 0.06
  √ 6 BB genotypes/100 total individuals = 0.06

- Heterozygous frequency (HT) = 0.38
  √ 38 Bb genotypes/100 total individuals = 0.38

- Homozygous recessive frequency (HR) = 0.56
  √ 56 bb genotypes/100 total individuals = 0.56

- The sum of all three genotype frequencies for a gene equals 1
  √ HD + HT + HR = 1

  √ 0.06 + 0.38 + 0.56 = 1

Hardy-Weinberg equilibrium

An equation that relates the allele frequencies to the genotype frequencies

- The Hardy-Weinberg equation is only valid if mating is random and both alleles of a gene are equally beneficial.

$HD = B_f^2$
$HT = 2\ B_f\ b_f$
$HR = b_f^2$

Therefore the genotype frequency equation…

$$HD + HT + HR = 1$$

…becomes the Hardy-Weinberg equation:

$$B_f^2 + 2\ B_f\ b_f + b_f^2 = 1$$

$$\| \qquad \| \qquad \|$$

$$HD \qquad HT \qquad HR$$

- This equation is usually shown in this form:

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

Where p = the allele frequency of the dominant allele
and q = The allele frequency of the recessive allele