MENDEL

- Pisum Sativum → pea flower both female and male sex organs; large number of individuals within a short growing season; clear-cut alternative traits
- SELF-FERTILIZATION: both egg and pollen come from the same plant
  CROSS-FERTILIZE two plants: remove male sex organs from a flower to prevent selfing and brush pollen from another plant onto its female organs
- DISCRETE TRAITS
  CONTINUOUS TRAITS
- P_1 GENERATION → pure-breeding lines with specific characteristics that remain constant from generation to generation
  INBRED because mated to each other for many generations
  ↓ Mendel selects 7 ANTAGONISTIC PAIRS of mutually exclusive alternative traits
  CROSS FERTILIZE and RECIPROCAL CROSSES (reverse the characteristics of male and female parents → the two parents contribute equally to inheritance)
  ⇓ PURE BREEDING LINES
  F_1 GENERATION → hybrids with the characteristics of one parent (DOMINANT)
  ⇓
  SELF FERTILIZE (monohybrid crosses: involving hybrid for a single trait)
  ⇓
  F_2 GENERATION → 3 (dominant) : 1 (recessive)
- GENES: for each trait, every plant carries two copies of a unit of inheritance, receiving one from its maternal parent and the other from the paternal one
  ALLELES: alternative forms of a single gene
- MONOHYBRIDS: individual having two different alleles of a single gene
  DIHYBRIDS: individual heterozygous for two genes at the same time
- GAMETES: specialized cells (eggs within the ovules and sperm cells) that carry genes between generations
  SOMATIC CELLS: have two copies of each gene
  ZYGOTE: fertilized egg
- 1) LAW OF SEGREGATION: the two alleles of each gene segregate during gamete formation, and then unite at random, one from each parent, at fertilization
- PRODUCT RULE → probability of two or more events occurring together is PROBABILITY OF EVENT 1 x (and) PROBABILITY OF EVENT 2
  SUM RULE → probability of either of two mutually exclusive events occurring is PROBABILITY OF EVENT 1 + (or) PROBABILITY OF EVENT 2
- PHENOTYPE: an observable characteristic
  GENOTYPE: the pair of alleles present in an individual
  - HETEROZYGOUS: a genotype with two different alleles for a trait → HETEROZYGOTE
  - HOMOZYGOUS: a genotype with the same two alleles for a trait → HOMOZYGOTE

  (Individual)
  TEST CROSS: a mating between an individual with the DOMINANT PHENOTYPE and an individual with RECESSIVE PHENOTYPE (yy) (Y-)
- 2) LAW OF INDIPENDENT ASSORTMENT: during gamete formation, different pairs of alleles segregate indipendentely of each other

  $AaBb \times AaBb$

  $9(A-,B-) : 3(A-,bb) : 3(aa,B-) : 1(aa,bb)$
- most often the DOMINANT ALLELE specifies a PROTEIN, while the RECESSIVE ALLELE determines either a nonfunctional version of the protein or no protein at all
  DOMINANT DISEASE ALLELES (es. Huntington) disease: dominant allele specifies a deleterious version of the protein
  RECESSIVE DISEASE ALLELES (es. cystic fibrosis): recessive "

COMPLETE DOMINANCE when the hybrids resemble one of the two pure breeding parents

□ x ■ → □ or ■

INCOMPLETE DOMINANCE when the hybrid exhibits an intermediate phenotype between those of the pure-breeding parents

□ x ■ → ▩

CODOMINANCE when both traits show up equally in the heterozygote's phenotype

□ x ■ → ▨

ALLELES FREQUENCY
- greater than 1% : WILD TIPE ALLELES
- rare : MUTANT ALLELE

- POLIMORPHIC GENES : have more than one common allele (wied type) — MONOMORPHIC GENES : have only one wied-type allele — es. ABO blood type system alleles : I^A, I^B, i
- PLEIOTROPY : a single gene determining a number of distinct traits
- SYNDROME : a group of problems usually seen togheter

- ADDITIVE INTERACTION between two genes — 9:3:3:1
  es. seed color in lentils
  P) AA bb (Ab) × aa BB (aB)
  F_1) AaBb × AaBb
  F_2) 9 A-B- : brown; 3 A-bb : tan; 3 aaB- : grey; 1 aa bb : green

SICKLE-CELL DISEASE :
(gene Hbα → α-globin)
gene Hbβ → β-globin
- normal wied-type allele $Hb\beta^A$
- mutant allele $Hb\beta^S$ (affects different traits)

↓

- NORMAL $Hb\beta^A$ $Hb\beta^A$
- CARRIER $Hb\beta^A$ $Hb\beta^S$
- DISEASED $Hb\beta^S$ $Hb\beta^S$

P) AGOUTI MICE (AA) × YELLOW MICE (A^YA)
F_1) YELLOW MICE ½ ; AGOUTI MICE ½ } 1 : 1 ratio

P) YELLOW MICE (A^YA) × YELLOW MICE (A^YA)
F_1) A^YA^Y (RECESSIVE LETHAL ALLELE PLEIOTROPIC) dies ¼
A^YA ½ ; AA ¼ } 2 : 1

- EPISTASIS : a gene interaction between alleles of differ. genes in which an allele of one gene masks the effects of another gene's alleles
  EPISTATIC gene : masks
  HYPOSTATIC : is masked

es. Bombay phenotype in humans :
homozygosity for h Bombay allele is epistatic to the I gene determining ABO blood types hh individuals do not produce substance H needed for the addition of A or B sugars on red blood cells

genotype	phenotype
I^AI^BH-	AB
ii H-	0
hh	Bombay (appears 0)

P) hhI^AI^A × HHii
↓
HhI^Ai genotype
↓
type A

9 : 7 reciprocal RECESSIVE EPISTASIS
in sweet pea color :
enzymes specified by the dominant alleles of two genes are both necessary to produce purple pigment
recessive alleles of both genes specify no proteins

P) AA bb (white) × aa BB (white)
F_1) AaBb × AaBb (both purple)
F_2) 9 A-B- (purple)
7 = 3 A-bb, 3 aaB-, 1 aa bb (white)

9 : 3 : 6
es. coat color of labrador retrivers is controlled by two genes E and B : recessive ee homozygous genotype is epistatic to any allelic combination at the second hypostatic gene B

P) BBEE (black) × bbee (yellow)
F_1) BbEe × BbEe
F_2) 9 B-E- black; 3 bb E- chocolate; 3 B-ee, 1 bb ee } yellow

13 : 3 or 12 : 3 : 1
DOMINANT EPISTASIS → for white color
es. dominant B allele masks any A and a combination
A- and aa color is seen only in bb individuals
A- yellow; aa green

P) AABB (white) × aabb (green)
F_1) AaBb × AaBb white
F_2) 12 = 9 A-B-, 3 aa-B- white; 3 A-bb yellow; 1 aa bb green

REDUNDANCY : one or more genes in a path way are superfluous

es. normal maize ear (AA BB) ♀
leaf lacking both dominant alleles (aa bb) ♀ skinny
either dominant allele A or B is sufficient for normal leaf development

P) AA bb × aa BB
♀ ↓ ♀
F1) Aa Bb × Aa Bb
♀ ↓ ♀
F2) 15 = 9 A-B- / 3 A-bb / 3 aa B-] normal ♀
1 aa bb skinny ♀

LOCUS HETEROGENEITY
a heterogeneous trait: a mutation at any one of a number of genes can give rise to the same phenotype

es. DEAFNESS

If mutations in two DIFFEREN GENES: AA bb × aa BB (pure-breeding parents, with the same mutant phenotype, homozygous for recessive non functional alleles of different genes)
↓
Aa Bb ♫ (complementation occurs)

If mutations in the same gene: AA bb × AA bb
↓
AA bb (non complementation)

COMPLEX TRAITS: determined by several different genes and/or by the interaction of genes with the environment

PENETRANCE
means the proportion of individuals with a particular genotype who show the expected phenotype
↳ COMPLETE (100%)
↳ INCOMPLETE

EXPRESSIVITY
refers to the degree or intensity with which a particular genotype is expressed in a phenotype
↳ VARIABLE
↳ UNVARYING

MAJOR GENES have a large influence on phenotype
MODIFIER GENES have a secondary effect
GENETIC BACKGROUND: set of unknown modifier genes that influence the action of known genes

TEMPERATURE is an environmental element with a visible effect on phenotype
→ ALLELE is temperature sensitive:
siamese cat: at warmer temp → enzyme non functional → no melanin → light fur
at colder temp → enzyme functional → melanin → dark fur
can affect survivability ⇒ CONDITIONAL LETHAL: causes death only under certain conditions
- range of temperatures under which the insects remain alive are PERMISSIVE CONDITIONS
- lethal temperatures above that are RESTRICTIVE CONDITIONS

PHENOCOPY
change in phenotype caused by chemical or environmental agents

CONTINUOUS VARIATION is caused by increasing the number of genes contributing to a trait (so many similar phenotypic classes) → POLYGENIC

COMPLEX TRAIT are influenced by many alleles and by the environment

SEX CHROMOSOMES → two sexes are not distinguishable by any other pair of chromosomes

[women XX (matching pair)
men XY (unmatching pair)

HUMAN ANEUPLOIDS → males with KLINEFELTER SYNDROME (XXY)
- tall, thin, sterile, with cognitive disabilities
→ females with TURNER SYNDROME (XO)
- sterile, short stature, webbed neck

SRY determines MALENESS → 6 weeks after fertilization SRY protein activates testes development in XY embryos, in the absence of SRY protein ovaries develop

Sex reversal:
XX males (carry a portion of Y)
XY females (lack a portion of Y)

MSY region { SRY, 3 male fertility genes, 8 essential genes shared with X

PAR2 → pseudoautosomal regions pair with each other during MEIOSIS I

HETEROGAMETIC SEX has two different sex chromosomes (gives rise to two different types of gametes)

HOMOGAMETIC SEX has two similar sex chromosomes

XX cells inactivate one of their two X chromosome → Barr Body → to have the same amounts of protein in male and female cells

GAMETOGENESIS

OOGENESIS:

oogonia —MITOSIS→ PRIMARY OOCYTE —ASYMMETRICAL MEIOSIS I→ FIRST POLAR BODY / SECONDARY OOCYTE —ASYMMETR. MEIOSIS II→ MATURE OVUM / SECOND POLAR BODY

Fertilized (penetrated by a sperm nucleus) / exit during MESTRUAL CYCLE

SPERMATOGENESIS:

spermatogonia —MITOSIS→ PRIMARY SPERMATOCYTE —MEIOSIS I→ SECONDARY SPERMATOCYTE —MEIOSIS II→ SPERMATIDS —↓ differentiation→ SPERM

SEX LINKAGE

Morgan → Drosophila Melanogaster (prolific and short generation time)

NORMAL WILD-TYPE ALLELE w^+ specifies RED EYES → dominant
MUTANT ALLELE w specifies WHITE EYES → recessive
} X-linked

♀ × ♂ → $X^{w+}X^{w}$ / $X^{w+}y$ (red) (red)

1) $X^{w+}X^{w+}$ (red) $X^{w}y$ (white) → $X^{w+}X^{w}$ / $X^{w+}y$ (red) (red)

2) $X^{w+}X^{w}$ (red) $X^{w+}y$ (red) → $X^{w+}X^{w+}$ (red), $X^{w}X^{w+}$ (red) / $X^{w+}y$ (red), $X^{w}y$ (white)

3) $X^{w+}X^{w}$ (red) $X^{w}y$ (white) → $X^{w+}X^{w}$ (red), $X^{w}X^{w}$ (white) / $X^{w+}y$ (red), $X^{w}y$ (white)

4) $X^{w}X^{w}$ (white) $X^{w+}y$ (red) → $X^{w}X^{w+}$ (red) / $X^{w}y$ (white) (crisscross inheritance)

Males are HEMIZYGOUS for X-linked genes because they have half the number of alleles carried by females on XX

SEX LIMITED TRAITS are found in one sex but not the other

SEX INFLUENCED TRAITS show up in both sexes, but may differ because of hormonal differences

NONDISJUNCTION during MEIOSIS I or II → XXY, XXX, XO, OY

X LINKED RECESSIVE TRAIT	≠	X LINKED DOMINANT TRAIT
- more males		- more females
- never from father to son		- seen in every generation
- affected male passes to all daughters, whose son have $\frac{1}{2}$ chance (carriers) to inherit		- all daughters and none of the sons of an affected male will be affected
- often skip a generation (grandfath → carrier daughter → son)		- both sons and daughters of an affected female have $\frac{1}{2}$ chance of being affected
- in successive generation when a sister of an affected male is a carrier		Y LINKED TRAIT only in males