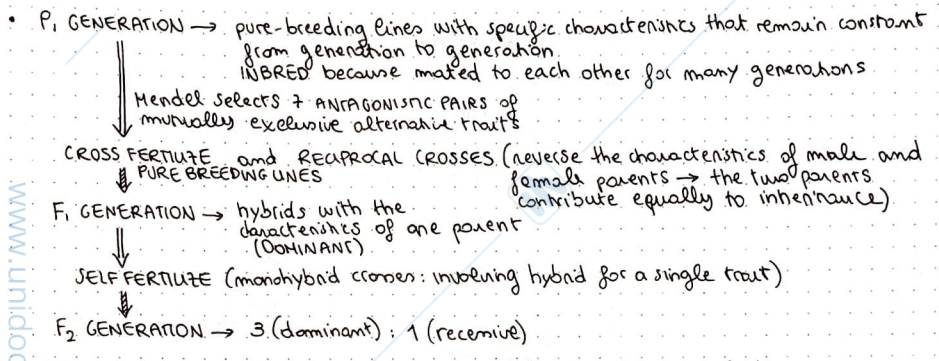


MENDEL

peo flower both female and male sex organs
large number of individuals within a short growing season
clear-cut alternative traits

- Pisum Sativum
- 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



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

TEST CROSS: a mating between an individual with the DOMINANT PHENOTYPE and an individual with RECESSIVE PHENOTYPE (yy) (Y-)

2) LAW OF INDEPENDENT ASSORTMENT: during gamete formation, different pairs of alleles segregate independently of each other

AaBb x AaBb

9 (A-, B-) : 3 (A-, bb) : 3 (aa, B-) : 1 (aa, bb)

most often the DOMINANT ALLELE specifies a PROTEIN, with the RECESSIVE ALLELE determines either a non-functional 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 "

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COMPLETE DOMINANCE when the hybrid resembles one of the two pure breeding parents



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



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



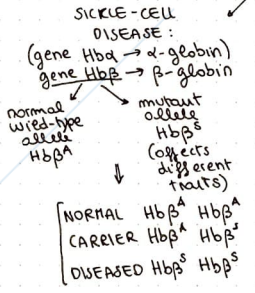
ALLELES FREQUENCY
 greater than 1% : WILD TYPE ALLELES
 rare : MUTANT ALLELE

- POLYMORPHIC GENES**: have more than one common allele (wild type)
- HOMOMORPHIC GENES**: have only one wild-type allele
- PLEIOTROPY**: a single gene determining a number of distinct traits

SYNDROME: a group of problems usually seen together

ADDITIVE INTERACTION between two genes

es. seed color in lentils
 P) AA bb x aa BB
 F1) Aa Bb x Aa Bb
 F2) 9 A-B- : brown
 3 A-bb : tan
 3 aa B- : grey
 1 aa bb : green



es. ABO blood type system alleles: I^A, I^B, i

P) AGOUTI MICE (AA) x YELLOW MICE (A^YA)

F1) YELLOW MICE (1/2)
 AGOUTI MICE (1/2) } 1:1 ratio

P) YELLOW MICE (A^YA) x YELLOW MICE (A^YA)

F1) A^YA^Y (recessive lethal allele) 1/4
 A^YA 1/2 } 2:1
 AA 1/4

EPISTASIS: a gene interaction in which an allele of one gene masks the effects of another gene's alleles
EPISTATIC gene: masks
HYPOMATIC: is masked

es. coat color of Labrador retrievers 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) x bbee (yellow)
 F1) BbEe x BbEe
 F2) 9 B-E- black
 3 bb E- chocolate
 3 B- ee yellow
 1 bb ee yellow

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- → O
 hh → Bombay (appears O)
 P) hh I^AI^A x HH ii
 F1) Hh I^Ai genotype
 F2) type A 13:3 or 12:3:1

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) x aa BB (white) (both purple)
 F1) Aa Bb x Aa Bb (purple)
 F2) 9 A-B- (purple)
 7 3 A-bb (white)
 3 aa B- (white)
 1 aa bb (white)

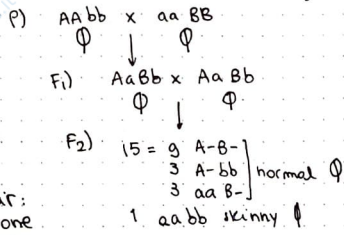
DOMINANT EPISTASIS for white color
 es. dominant B allele masks any A and a combination
 A- and aa color is seen only in bb individuals
 P) AABB (white) x aabb (green)
 F1) Aa Bb x Aa Bb (white)
 F2) 12 9 A-B- (white)
 3 aa B- (white)
 3 A-bb (yellow)
 1 aa bb (green)

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REDUNDANCY : one or more genes in a pathway are superfluous

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

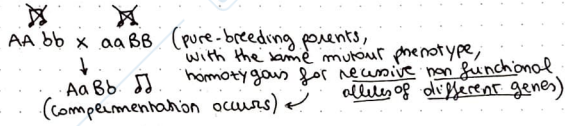


LOCUS HETEROGENEITY

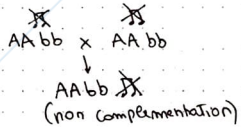
a heterogeneous trait: a mutation at any one of a number of genes can give rise to the same phenotype.

es. DEAFNESS

If mutations in two DIFFERENT GENES



If mutations in the same gene



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:
- dominant rat: 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

PHENOECOLOGY

change in phenotype caused by chemical or environmental agents

- range of temperatures under which the insects remain alive are **PERMISSIVE CONDITIONS**
- lethal temperatures above that are **RESTRICTIVE CONDITIONS**

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

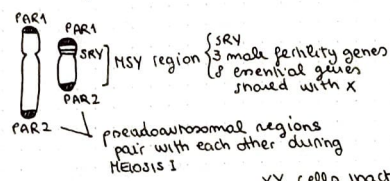
Women XX (matching pair)
men XY (unmatching pair)

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 fertilisation SRY protein activates testes development in XY embryos, in the absence of SRY protein ovaries develop

Sex reversal:
XX Males (copy a portion of Y)
XY females (lack a portion of Y)



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

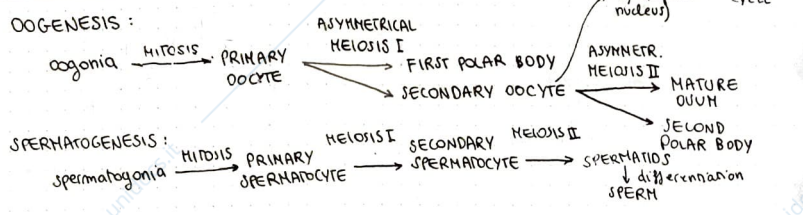
HOMOGAMETIC SEX has two similar sex chromosomes

to have the same amounts of protein in male and female cells

XX cells inactivate one of their two X chromosomes → Barr Body

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

GAMETOGENESIS



SEX LINKAGE

Organism → 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 ♂
- $X^{w^+}X^{w^+}$ (red) × X^wY (white) → $X^{w^+}X^w$ (red), $X^{w^+}Y$ (red)
 - $X^{w^+}X^w$ (red) × $X^{w^+}Y$ (red) → $X^{w^+}X^{w^+}$ (red), $X^wX^{w^+}$ (red), $X^{w^+}Y$ (red), X^wY (white)
 - $X^{w^+}X^w$ (red) × X^wY (white) → $X^{w^+}X^w$ (red), X^wX^w (white), $X^{w^+}Y$ (red), X^wY (white)
 - X^wX^w (white) × $X^{w^+}Y$ (red) → $X^wX^{w^+}$ (red), X^wY (white) (cis/trans 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

NON DISJUNCTION during MEIOSIS I or II → XXY, XXX, XO, OY

- X LINKED RECESSIVE TRAIT** ≠
- more males
 - never from father to son
 - affected male passes to all daughters, whose son have 1/2 chance to inherit
 - often skip a generation (grandfather → carrier daughter → son)
 - in successive generation when a sister of an affected male is a carrier

- X LINKED DOMINANT TRAIT**
- more females
 - seen in every generation
 - all daughters and none of the sons of an affected male will be affected
 - both sons and daughters of an affected female have 1/2 chance of being affected
- Y LINKED TRAIT** only in males

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