Biology 30 IB
Genetics

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Contents

(18.1) Gregor Mendel

- Created the Laws of Inheritance of Traits
- Studied inheritance of traits in pea plants (test question)
- Called DNA and chromosomes particles — he didn't know
- Father of Genetics

Peas

- Can be grown in a small area
- Produce lots of offspring
- Self-pollinate
- Can be artificially cross-pollinated (test question)

Introduction

General

- **Trait** = any characteristic that can be passed from parent to offspring
- **Heredity** = passing of traits from parent to offspring
- **Genetics** = study of heredity

Terms

- **Alleles** = two forms of a gene — dominant & recessive
- **Dominant** = stronger of two genes, represented with capital letter (R)
- **Recessive** = weaker of two genes, represented with lowercase letter (r)
- **Genotype** = gene combination for a trait (RR, Rr, rr) separated with colons
- **Phenotype** = physical feature resulting from genotype (red, white) separated with colons
- **Homozygous genotype** = genotype involving 2 dominant OR 2 recessive genes — pure, RR or rr
- **Heterozygous genotype** = genotype involving 1 dominant OR 1 recessive gene — hybrid, Rr

Dominance

- Dominant and recessive alleles can code for different things
- e.x. R = Brown eyes, r = blue eyes
- Any dominant alleles = dominant phenotype — RR, Rr in most cases
- Only recessive alleles = recessive phenotype — rr

Genotypes	RR	Rr	rr
Phenotypes	RED	RED	YELLOW

Crosses

- **Monohybrid cross** = cross involving a single trait
- **Dihybrid cross** = cross involving two traits
- **Test cross** = cross involving always involing a homozygous recessive (rr) parent crossed with an unknown genotype, in order to find the genotype

Generations

- **P1 Generation** = parental generation in a breeding experiment
- **F1 Generation** (1st filial gen) = first-generation offspring in a breeding experiment
- **F2 Generation** (2nd filial gen) = second-generation offspring, and so on...

Mendel's Laws

LAW	PARENT CROSS	OFFSPRING
DOMINANCE	TT x tt tall x short	100% Tt tall
SEGREGATION	Tt x Tt tall x tall	75% tall 25% short
INDEPENDENT ASSORTMENT	RrGg x RrGg round & green x round & green	9/16 round seeds & green pods 3/16 round seeds & yellow pods 3/16 wrinkled seeds & green pods 1/16 wrinkled seeds & yellow pods

(18.2) Punnett Squares

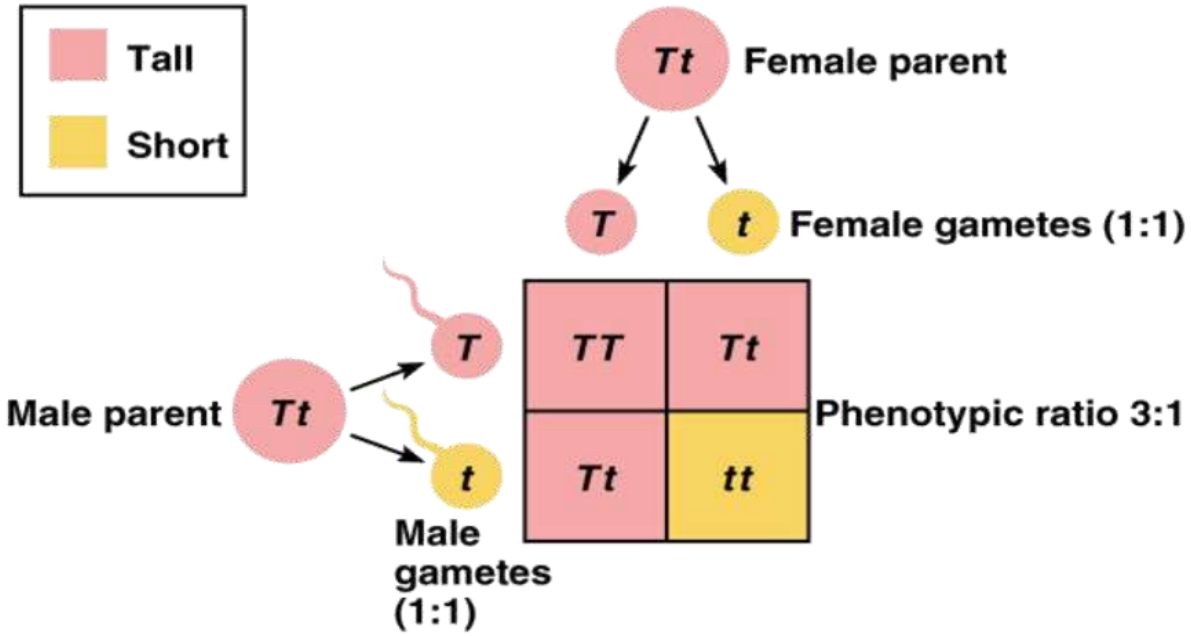


Figure 1: Punnett square demonstrating law of segregation, which occurs in anaphase I

- One gene from father, one gene from mother
- Dominant and recessive alleles compete in punnett squares
- **Phenotypic ratio** = ratio of dominant phenotypes to recessive phenotypes, in this diagram its 3 : 1

(18.3) Pedigree Charts

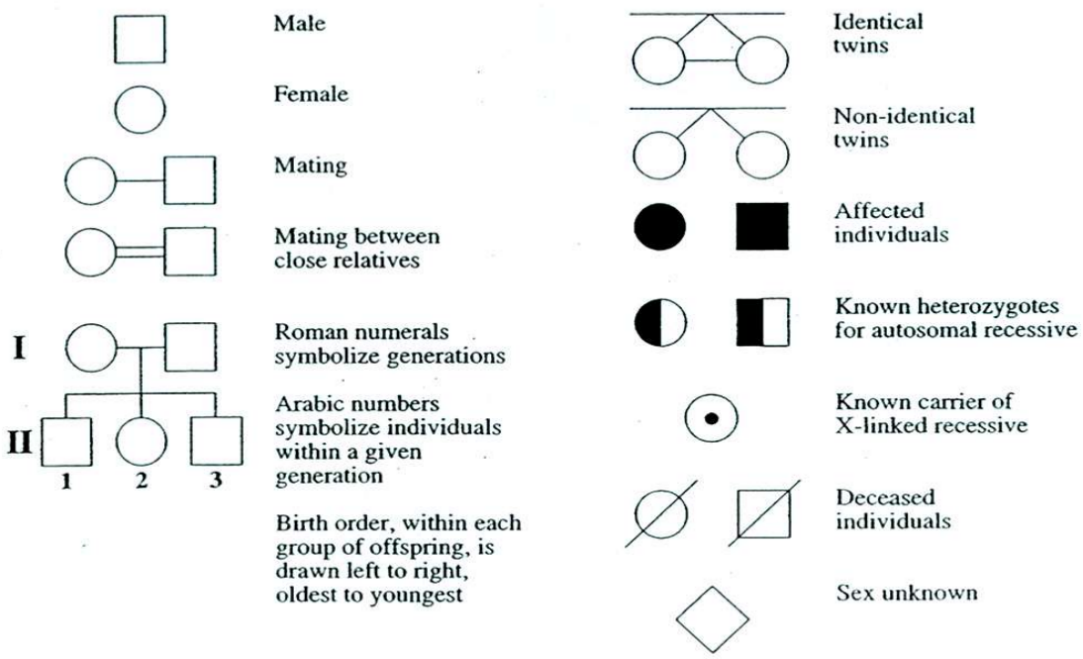


Figure 2: All in your data booklet, except known heterozygotes

- **Pedigree** = a family tree used to trace inherited traits from parents to offspring
- Roman numerals denote different generations (rows)
- Arabic numerals denote different individuals in a generation (columns)

Terms

- **Autosomal condition** = almost equal number of males and females are affected by a condition
- **Dominance condition** = condition appears in every generation
- **Recessive condition** = condition does not appear in every generation; skips generations

(18.4) Other Patterns of Inheritance

Pleiotropy

- **Pleiotropic gene** = one gene affects more than one phenotypic characteristic
- Examples of wide-ranging effects from a single gene include...
  - dwarfism (achondroplasia)
  - gigantism (acromegaly)

Multiple Alleles

- Possible to have more than two alleles for a trait
- Superscript identifies different alleles
- Different alleles are dominant to one another
  - e.g. $E^1 > E^2 > E^3 > E^4$

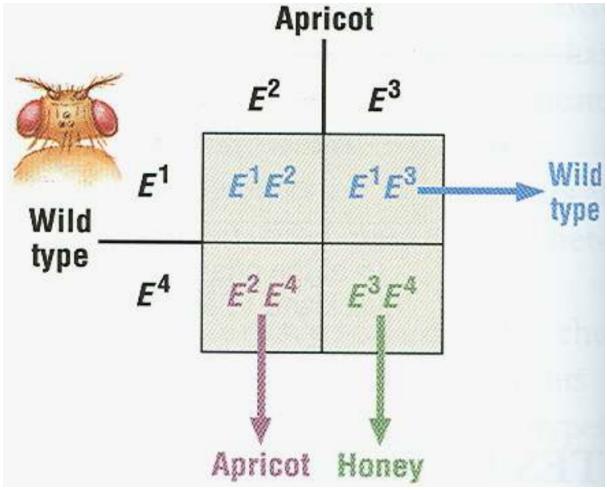
Example

Predict the genotypic and phenotypic outcomes of crossing $E^1E^4 \times E^2E^3$.

- E^1 = wild, E^2 = apricot, E^3 = honey, E^4 = white

Wild type	E^1E^1, E^1E^2, E^1E^3, E^1E^4
Apricot	E^2E^2, E^2E^3, E^2E^4
Honey	E^3E^3, E^3E^4
White	E^4E^4

Figure 3: This is typically given



- Genotype $= 1 \times E^1E^2 : 1 \times E^1E^3 : 1 \times E^2E^4 : 1 \times E^3E^4$
- Phenotype = 2 wild : 1 apricot : 1 honey

Incomplete Dominance

- A hybrid (Rr) appearance can sometimes be in between the phenotypes
- For example...
  - RR = red
  - rr = white
  - Rr = pink (normally red)

Codominance

- Both alleles expressed in heterozygous individuals

Figure 4: Red bull + white cow = red and white hair

Blood Type

Example of codominance and multiple alleles

I^A & $I^B > i$

- Type A = $I^A I^A$ or $I^A i$
- Type B = $I^B I^B$ or $I^B i$
- Type AB = $I^A I^B$
- Type O = ii

(18.5) Dihybrid Crosses

- A breeding experiment that tracks the inheritance of two traits
- **Law of Independent Assortment** = each pair of alleles segregate independently (metaphase)
- The two traits do not influence one another, they are independent
- These traits are on different locations — called **loci**, **locus** — of a chromosome

of Unique Gametes

$$2^n$$

$n = \#$ of heterozygotes of each parent

Examples...

- RrBb = 2 heterozygotes = $2^2 = 4$ gametes
- yypp = 0 heterozygotes = $2^0 = 1$ gamete
- AaBbCCDd = 3 heterozygotes = $2^3 = 8$ gametes
- MmNnOoPPQQRrssTtXx = 6 heterozygotes = $2^6 = 64$ gametes

Drawing a Dihybrid Cross

Determine how many different possible gametes/phenotypes with the formula.

- $2^2 = 4$
- There will be 4 different types of offspring from this cross
- If a question specifically asks for the adults, there may be a gene that prevents adulthood — i.e. a fatal gene — so do not include that combination in any percent calculations and ratios
- If you are doing a test cross — there is an unknown gene — make a dihybrid cross for each possible combination; this applies to monohybrid aswell

Prepare a cross for every possible combination of all heterozygotes.

1. Top header = FOIL between genes of the gene pair of the first parent
2. Left header = FOIL between genes of the gene pair of other parent
3. You can skip redundent combinations
4. Fill in the table like usual, including the top and left header, to list every possible combination
5. Make sure to order the alleles dominant to recessive, and same letters together

Using the cross, you can...

- Determine the number/ratio of all possible genotypes by counting
- Determine the number of phenotypes from all possible genotypes

It helps to mark each of the table cells with a symbol to keep track.

Example

- R_ = round, Y_ = yellow
- rr = wrinkled, yy = green

Question: Determine the genotype and phenotype of a RyYy x RyYy cross.

	RY	**Ry**	**rY**	**ry**
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

- Genotype
  $1 \times RRYY : 2 \times RRYy : 2 \times RrYY : 4 \times RrYy : 1 \times RRyy : 2 \times Rryy : 1 \times rrYY : 2 \times rrYy : 1 \times rryy$
- Phenotype
  - 9 round, yellow
  - 3 round, green
  - 3 wrinkled, yellow
  - 1 wrinkled, green

Probability

$$\text{probability} = \frac{\#\text{ of chances of an event}}{\#\text{ of possible combinations}}$$

- **Independent event** = individual event, random chance, previous events do not influence

Monohybrid Method

Alternative to creating a dihybrid cross.

1. Create a monohybrid cross for each of only the matching letter genes between both parents
   e.g. MmNnOo x MmNnOo = Mn x Mn, Nn x Nn, Oo x Oo

Combination (Monohybrid to Dihybrid)

- Similar to distributive property
- Multiply each gene in the first monohybrid by every gene in the other monohybrid
- This should result in the same answers as FOILing in a dihybrid cross

Calculating Probability

- Divide the number of a desired gene out of the number of possible combinations
- This applies to both methods — the entire dihybrid FOIL, and the monohybrids of the monohybrid method
- Multiply all probabilities together if looking for the probability of multiple specific independent genes together
  e.g. the probability of blue eyes and brown hair

Gene Interaction

- **Polygenic** = more than one genes influence a trait
  e.g. skin colour, eye colour, height
- **Epistasis** = one gene masks/epistatic to another, influences ratio

(19.1) Chromosomal Theory

- **Chromosomal Theory** = anything to do with chromosomes (diploma question)
- Sex-linked genes discovered by **Morgan**

Autosomal Traits

These are tested!!!

- Cystic fibrosis = mucus build up in many organs
  - normal alleles = Cc, CC
  - mutated alleles = cc
- Sickle cell anemia = sickling of blood cells, cannot carry oxygen
  - normal alleles = Hb^AHb^A
  - mutated alleles = Hb^SHb^S
  - carrier alleles = Hb^AHb^S (less extreme effects)

Sex-linked Genes

- **Autosomes** = chromosomes that do not code for proteins related to the sex of the individual
- **Autosomal trait** = traits not located on sex chromosomes
- **Sex-linked trait** = traits located on sex chromosomes

Carriers in X-linked

- Females cannot be affected by an irregular allele, since they have two X chromosomes in which one of them may dominate the other; they can be carriers — they carry a dormant recessive irregular allele, which can be given to their offspring
  (homozygous recessive females are rare)
  - **Barr body** = dormant, unexpressed X chromosome; chromosome shrinks into barr body; not in males, somatic cells only
  - **Barr bodies** = whichever allele is dormant is not consistent in all body cells; causes spots of dark skin and no sweat glands
- Males can be affected by an irregular allele, but cannot be carriers; they only have one X chromosome, which could be the irregular one from their mom
- A mother's recessive alleles are most likely to be given to their sons

X-Linked Conditions

These are tested!!!

- At this level, X chromsomes carry traits, while Y chromosomes do not
- **Hemophilia** = blood clotting disorder
  - normal allele = X^H
  - mutated allele = X^h
  - X^hY, X^hX^h
- **Colour-blindness** = typically red-green colourblind
  - normal allele = X^R
  - mutated allele = X^r
  - X^rY, X^rX^r

Testes Determing Factor (TDF)

- Gene located on the Y chromosome
- Develops male gonads while in embryo
- Produces testosterone when activated

Testicular Feminizing Syndrome (TFS)

- Gonad forming tissues ignore testosterone
- Produces female vaginal folds AND penis

Mitochondrial DNA

- From mother
- Can be defective, causing deafness

(19.2) Linked and Incomplete Genes

Discovered by Morgan.

Linked Genes

Linked genes aka. **linkage groups**

- Genes on the same chromosome tend to be transmitted and linked together
- close together
- inherited together
- move together in anaphase, do not segregate
- fewer combinations of alleles
- **Non-recombinants** aka. **parental types** = offspring from linked genes, since diversity/variety is reduced, looking very similar to parents

How to tell

- Determine all possible combinations like before — FOIL or multi monohybrids
- If the combinations do not match the genotypes given to you, there are linked genes

Incomplete Linkage

- Incomplete linkage, and crossing-over during prophase I, can provide new combinations
- **Recombinants** = offspring formed as a result of new combinations between alleles of parents

How to tell

- Determine all possible combinations like before — FOIL or multi monohybrids
- If the combinations do match the genotypes given to you, there are incompletely linked genes

Mapping

- **Cross-over value** = distance between genes, typically in % but can be treated as any unit
- You need to find the order that genes are on a chromosome when given only the cross-over values

Steps

There isn't really any strict tricks or methods, but these are some suggestions.

1. Start with the genes with the smallest distance between one another
2. The next genes to plot should share genes with the previous one plotted

Example

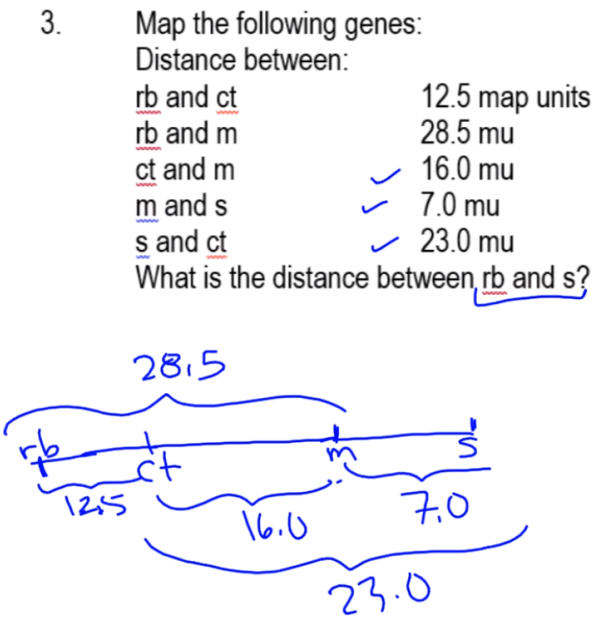


Figure 5: rb $\longrightarrow$ ct $\longrightarrow$ m $\longrightarrow$ s

Answer: 35.5 mu (28.5 mu + 7.0 mu)

Applications

You don't really need to memorize this.

Gene Insertion

- Normal gene inserted into position on chromosome of a diseased cell

Gene Modification

- Defective gene is modified chemically to recode genetic message

Gene Surgery

- Defective gene is extracted
- Replaced with a normal gene