

BMS2042

Lecture 1

1. Define Genetics
2. Understand the punnet square
3. Know terms

Genetics: study of genes

Geneticist: how genes work together

- Traits
- Variation
- Genotype
- Phenotype
- Laws of inheritance (defined by Mendel)

Approach: study gene function via deliberate mutation

Transmission: how traits are passed from parents to offspring

Molecular: traits affected by heritage

Population: variation and role in evolution

Common models:

- Yeast
- Drosophila (vinegar fly)
- Worm
- Zebra fish
- Mice
- Arabidopsis thaliana (plant)

Models require:

- Cheap
- Easy access
- Short reproductive process
- Large amount of progeny (offspring)

Molecular level: function of enzyme or transcription factor

Cellular level: increase/decrease in trait (eg. increased pigmentation)

Organism level: phenotype (light/dark butterfly)

Population level: environmental factors (dark butterfly is only found in forested regions due to its colour it can camouflage better)

Albinism: autosomal/recessive/one gene alters

Colour is on a spectrum

Mendel

Pangenesis: all skills were transferred to the baby

Homunculus: a little man was in the head of the sperm

Blending theory: law of inheritance- physical traits from both parents were passed onto the offspring

Autosomal inheritance was discovered by Mendel due to his model organism:

- Thousands of crosses
- Short reproductive period
- Simple phenotypical traits
- Rigorous testing of a hypothesis
- Tested only one trait
  - o Round or wrinkled
  - o Yellow or green
  - o Short or tall

- Inflated or pinched pod

Phenotype: controlled by a gene

Different version of the gene = allele

1. Law of Segregation: two alleles segregate into gametes and each gamete has 50% chance of inheriting each allele
  - Segregation is random

T = purple

t = white

parents = TT × tt

|   |    |    |                     |
|---|----|----|---------------------|
|   | T  | T  |                     |
| t | Tt | Tt | = 100% purple<br>F1 |
| t | Tt | Tt |                     |

F1 = Tt × Tt

F2

To test whether F2 generation is homozygous purple (TT) or heterozygous purple (Tt), a testcross with a purely recessive plant must occur.

50% purple =  
50% white

|   |    |    |
|---|----|----|
|   | T  | t  |
| T | TT | Tt |
| t | Tt | tt |
| t | Tt | tt |

|   |        |       |                  |
|---|--------|-------|------------------|
|   | T      | t     |                  |
| t | purple | white | = 100%<br>purple |
| t | Tt     | Tt    |                  |

Two traits at a time: dihybrid cross  
9:3:3:1 ratio

2. Law of Independent Assortment: independent segregation of allele

Y = yellow  
y = green

R = round

r = wrinkled

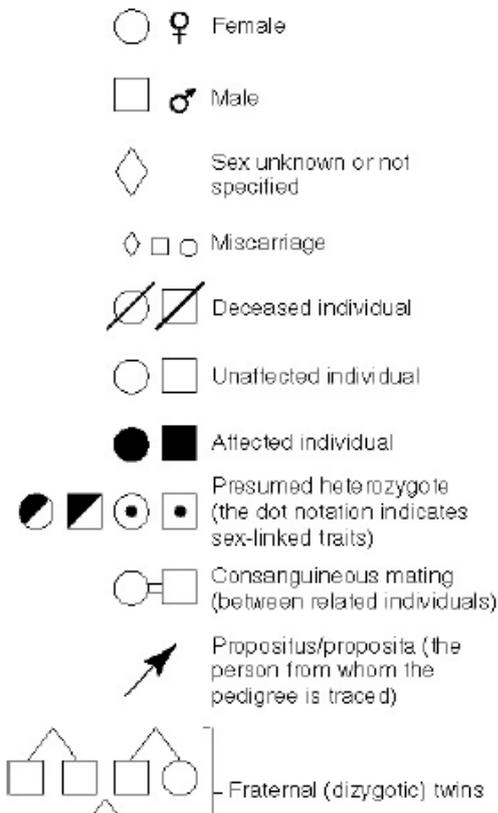
Parents = YYRR × yyrr

|    |      |    |    |    |
|----|------|----|----|----|
|    | YR   | YR | YR | YR |
| yr | YyRr |    |    | →  |
| yr |      |    |    |    |

F1 = YyRr

## Autosomal Inheritance

- Requires Pedigrees
- Limited data
  - o Autosomal or X-linked
  - o Dominant or recessive
  - o Heterozygous for a recessive mutation involves carriers



### Autosomal recessive:

- Males and females equally affected
- Skips generations
- Incest

### Autosomal dominant:

- Males and females equally affected
- Affected individuals in multiple generations
- Transmission from both sexes to both sexes

Mendel laws apply to all eukaryotic organisms

← Pedigree key

## GENETIC TERMS

|              |                                                                                 |
|--------------|---------------------------------------------------------------------------------|
| trait        | Characteristic of an organism, e.g. seed colour                                 |
| phenotype    | Appearance of an organism e.g. yellow seed coat or green seed coat              |
| genotype     | Genetic composition of the individual e.g. YY, Yy or yy                         |
| gene         | Unit of heredity (region of DNA) influencing a trait, e.g. gene for seed colour |
| allele       | Alternate version of the same gene; Y or y                                      |
| locus        | Specific place on a chromosome occupied by a gene                               |
| homozygote   | An organism possessing two of the same alleles at a locus                       |
| heterozygote | An organism possessing two different alleles at a locus                         |

### Lecture 2

1. Sex-linked Inheritance
2. Chromosome theory

### 3. Meiosis

Thomas Hunt: white-eye mutant *Drosophila*

Sex-linked inheritance: different results from reciprocal crosses

- Eg. Female wildtype x male white-eye = all female wildtype and all male wildtype
- Female white-eye x male wildtype = female wildtype x male white-eye

Therefore the white gene is on the X chromosome (females have 2 copies, males have 1 copy- hemizygous)

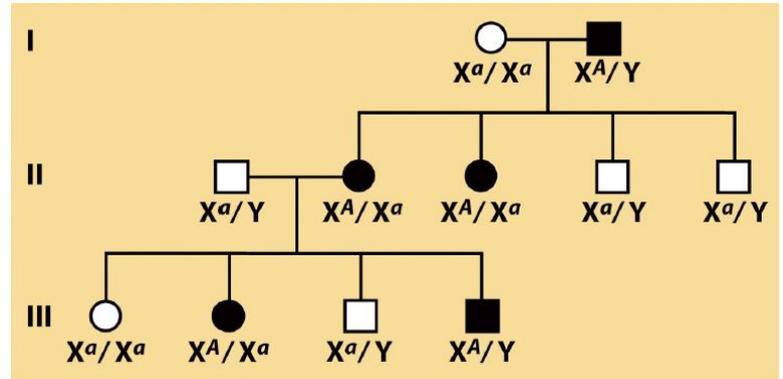
X-linked recessive:

- Mostly males usually affected
- Carrier females
- No male to male transmission (as mutation is on the X chromosome)
- Eg. Haemophilia

Red-green colour blindness

X-linked dominant:

- Rare
- Both sexes affected but females often in excess
- Females less severely affected (due to X inactivation)
- No male to male transmission
- Eg. Hypophosphatemia (rickets due to Vitamin D resistance)  
Rett Syndrome (neurological disorder- lethal in males)  
Incontinent pigment (skin abnormality- lethal in males)



Y-linked inheritance:

- SRY

Chromosome Theory: genes are on chromosomes

1. Chromosomes during meiosis divide parallel to allele assortment into gametes
  - a. Genes are in pairs as are chromosomes. The same allele exists on two homologous chromosomes.
  - b. Alleles segregate equally into gametes as do homologous chromosomes (1<sup>st</sup> law)
  - c. Segregation is random/independent (2<sup>nd</sup> law)
2. Sex-linked genes: traits follow inheritance of a particular chromosome
  - a. If you have a particular trait, you will have the linked chromosome which contains the gene causing expression
  - b. Eg. SRY on Y chromosome = male

AND = x

OR/EITHER = +

Birds/reptiles/butterflies:

- Male is homogametic (XX)
- Female is heterogametic (XY)

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