

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 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% 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

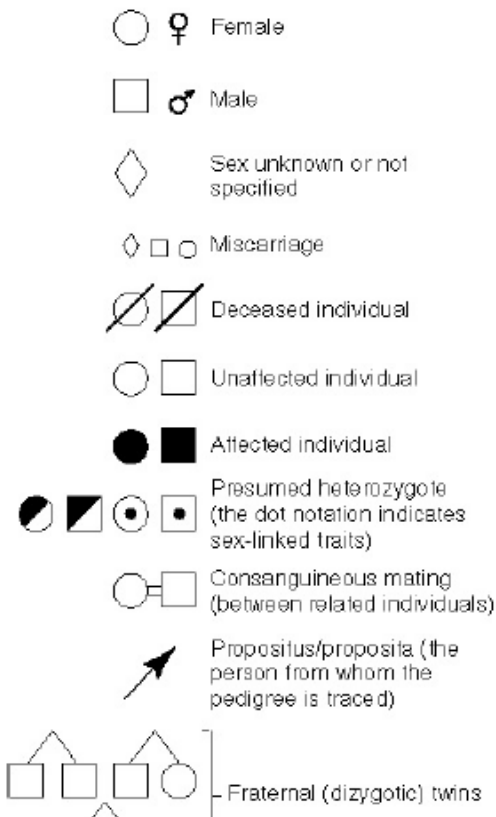
Parents = YYRR × yyrr

	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)

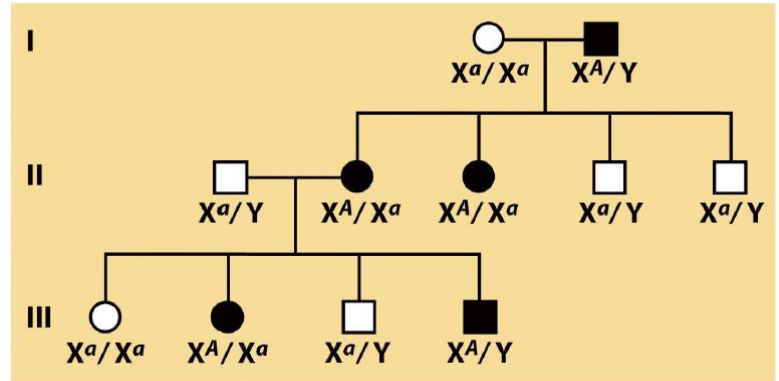
X-linked recessive:

- 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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