

# BIOL215 Summary Notes

## Lecture 1:

### History of genetic discoveries

#### **1928 – Fred Griffith (experiment #1)**

- Was studying the infection patterns of the pathogen *Streptococcus pneumoniae* in mice
- **S-strain**: 'smooth' appearance due to a polysaccharide capsule **virulent**
- **R-strain**: 'rough' in appearance; lacking polysaccharide capsule **avirulent**
- Several variants of each major strain (IIS, IIIS, IIR, IIIR).
- Occasionally IIS mutated to IIR, but never to IIIR. IIIS mutated to IIIR, but never IIR

#### **Experiment**

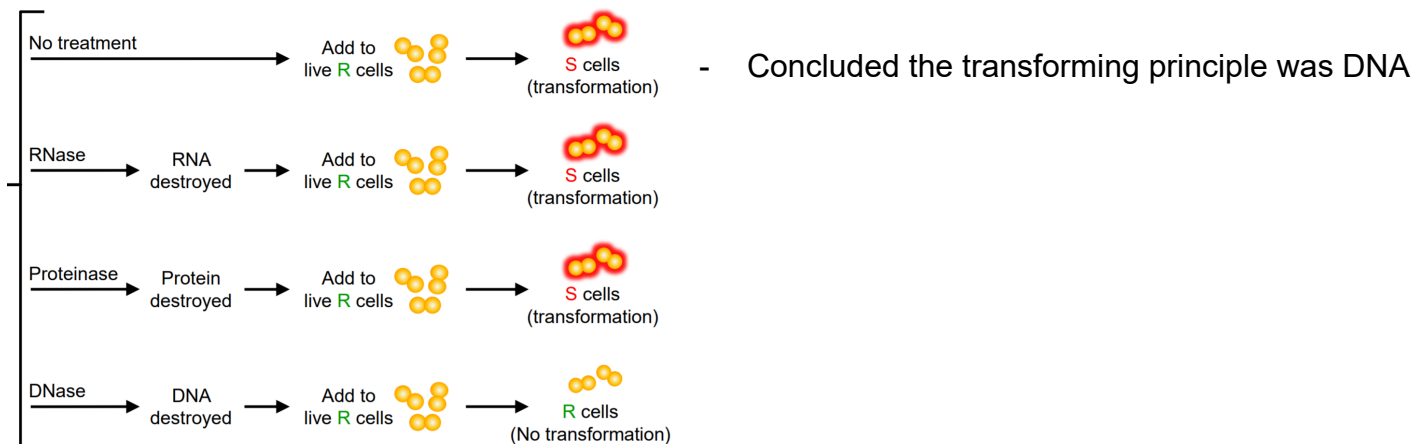
- Type IIR injected = mice survives
- Type IIIS injected = mice dies + recovered IIIS
- Type IIIS HEAT SHOCKED = mice survives
- Type IIIS HEAT SHOCKED + type IIR = mice dies + IIIS recovered

#### **Conclusion:**

- Some heat-stable component present in IIIS transformed IIR into IIIS and is inherited
- Termed the transmitted substance the 'transforming principle'

#### **1944 – Avery, MacLeod & McCarty (experiment #2)**

- Repeated the Griffith experiment, but fractionated the 'transforming principle'



#### **1953 – Hershey & Chase (experiment #3)**

- Experimented with the T2 bacteriophage (only protein and DNA)
- Cannot replicate on its own – relies on its host cell for everything (obligate parasite)
- used radioactive phosphorus (DNA) and sulphur (protein)
- After dna insertion into bacteria and empty vessel removed, the dna was seen in bacteria and protein was discarded → Confirms that the only material to enter the cells is DNA
- 2nd experiment allowed the phage to reproduce → small amounts of <sup>32</sup>P are found in the new phage, but no <sup>35</sup>S → DNA being passed on to progeny, not protein

## RNA

- RNA is the genetic material in some viruses
- RNA is generally very unstable, but viral protein coats protect from degradation by RNases
- Eg. Tobacco Mosaic Virus
- **1956 – Gierer and Schramm** Showed that the genetic material of TMV is RNA
- After purification it was discovered that RNase and not DNase degraded the viral genetic material
- Fraenkel-Conrat & Singer - 1957 found that One coat protein could encapsulate the other's RNA and still function
- But RNA determined the coat of the offspring (progeny)

## Nucleotides

- The phosphate group attached to the 5' carbon of one ribose and the 3' carbon of the next
- RNA has a OH group on the 2nd carbon, DNA only has a H atom
- Purines (2 rings) = Adenine and Guanine
- Pyrimidines (1 ring) = Cytosine, Thymine (DNA), and Uracil (RNA)
- 5' end has a free Phosphate
- 3' has a free OH group
- DNA synthesised in a 5' to 3' direction on the strand being synthesised
- 'Chargaff's rules' found the number of bases matched for base pairing

## Lecture 2 - Meiosis and segregation

**Meiosis** = halves the number of chromosomes in cells by going through 2 divisions with only one round of DNA replication

### Interphase I

- homologous chromatids duplicate, so that each chromosome has 2 arms

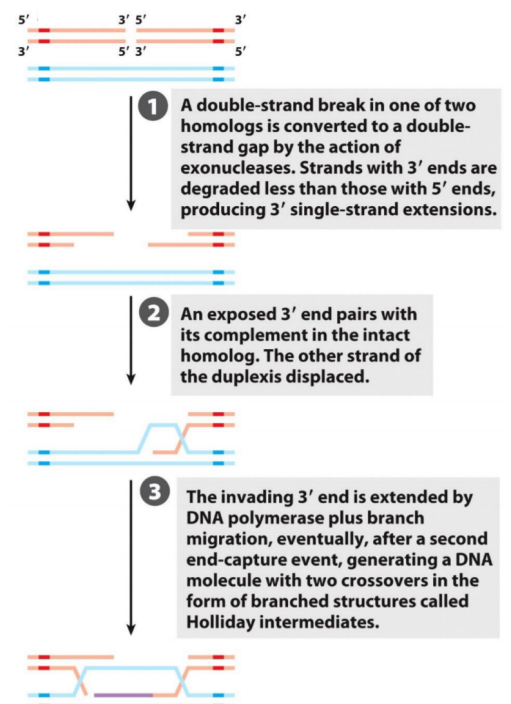
### Prophase I

- sets of homologous chromosomes align to form tetrads, held together by covalent links at homologous junctions (chiasmata)

- Very precise; corresponding nucleotide sequences line up with one another.

- This precise pairing allows 'crossing over' or 'non-sister chromatid exchange' to occur.

- Chromosomes align and exonuclease degrades dsb ends to make bigger gap
- 3'-ending strands of the gap "invade" Chromosome B they pair with a complementary region on displacing ne chromosome B strand
- The displaced region of Chromosome B serves as the template to replace missing DNA



## Metaphase I

- The nuclear envelope dissipates and spindle fibres attach to kinetochores
- Homologous chromosomes align on the metaphase plate with one sister chromatid of each orienting toward open pole.

## Anaphase I

- Homologous chromosomes separate and migrate to opposite poles
- Each chromosome sorted independently\*
- Note that one pole will contain the father's allele, the other the mother's allele

## Telophase I

- The cell divides, producing two cells each with just one set of chromosomes → Haploid cells
- The cells produced at telophase I are haploid, because they contain either the mother's or the father's chromosome, not both (excluding of course elements that crossed over)

## Meiosis II

- Same as meiosis I, however chromatids split, creating 4 daughter cells

## **Mendel's laws**

1. Segregation = alleles segregate during meiosis
2. Independent Assortment =

## Inheritance

### **Pedigree analysis**

Females ○

X-linked ○●

Males □

Autosomal carrier ○◐

Affected individuals ■ / ●

Deceased ◐

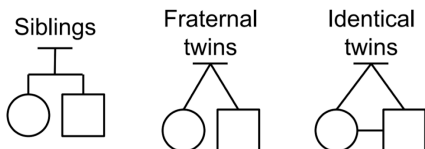
Paired couple □—○

Consanguineous pairing □=○

If two unaffected parents have an affected child the trait is recessive.

If two affected parents have an unaffected child the trait is dominant.

Recessive genes can skip a generation



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To determine genotype of an unaffected individual where a recessive disease is present;

- A **testcross** is a cross of an individual of unknown genotype with a homozygous recessive
- The phenotypes of the offspring indicate the genotype of the individual tested.

## Dihybrid crosses

Consider a cross between:  $Aa Bb \times Aa Bb$

Four possible phenotypes: AB, Ab, aB, and ab

The probability of A =  $3/4$ , a =  $1/4$

The probability of B =  $3/4$ , b =  $1/4$

The expected proportion of AB:Ab:aB:ab is 9:3:3:1