

LENScience Senior Biology Seminar Series Human Aneuploidy and Related Biotechnologies Questions and Discussion

Pre-seminar School Discussion

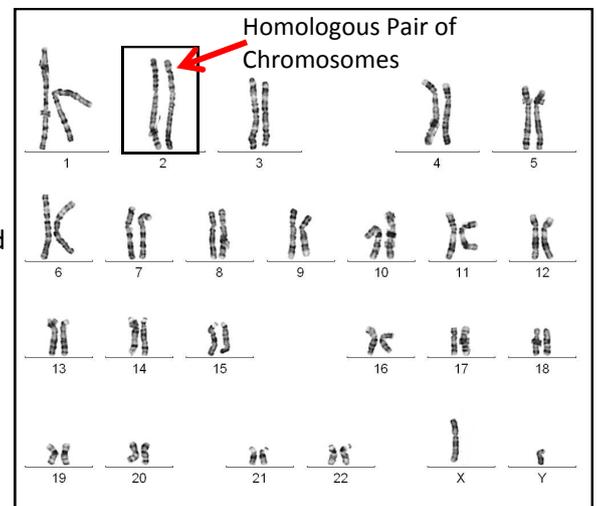
This seminar focuses on the role of mutations in the inheritance of genetic conditions, and the biotechnologies that can be used to screen and diagnose for these conditions before and during pregnancy. While the focus is on aneuploidy, it will link to heritable gene mutations. Your understanding from Year 11 and 12 of the concepts of cell division, gene expression and mutations will provide you with a starting point. The seminar will link to concepts in the Year 13 Biology programme and develop your understanding of how knowledge of these concepts can be applied in human situations. A list of the Year 13 concepts linked to the seminar can be found on page 4 of this handout. Don't worry if you have not covered these in class yet. It is recommended that students review this seminar again later in the year once you have completed molecular genetics and biotechnology topics.



A Review of Cell Basics

Use your knowledge of Y12 Biology and the information in the seminar paper to discuss the following questions.

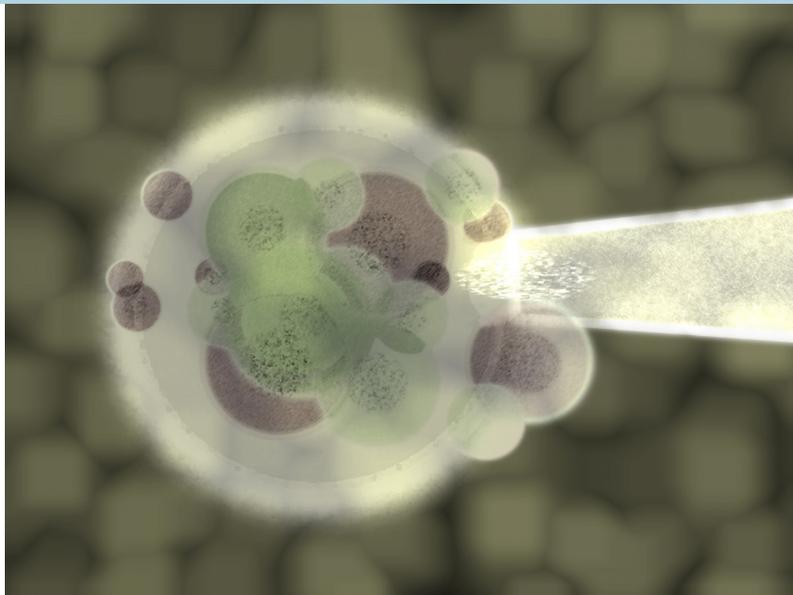
1. The diagram on the right is a human karyotype, containing 22 pairs of homologous chromosomes and the sex chromosomes.
 - a. Is this a male or a female karyotype?
 - b. Explain what homologous chromosomes are and what information they contain.
 - c. Define the term chromatin.
 - d. Describe the structure of a chromosome and explain the relationship between chromatids and chromosomes.



2. Explain the difference between a gene and a chromosome mutation.
3. Compare and contrast the purpose, processes and products of mitosis and meiosis.
4. Discuss the different mechanisms within meiosis which contribute to the genetic variation found in gametes.

Vocabulary

Gamete	Allele	Ultrasound scan
Oocyte	Gene Mutation	Chorionic Villus Testing
Spermatozoa	Chromosome Mutation	Amniocentesis
Semen	Autosomal Dominant	PGD—Pre-Implantation Genetic
Zygote	Autosomal Recessive	Diagnosis
Blastocyst	Mitosis	Byopsy
Embryo	Meiosis	<i>In vitro</i> fertilisation
Fetus	Aneuploidy	Fluorescence <i>in situ</i> hybridisation
Placenta	Polyploidy	Comparative Genomic Hybridisation
Chromosome	Non-disjunction	Preimplantation Genetic Haplotyping
Chromatid	Monosomy	Restriction Enzymes
Chromatin	Trisomy	Ligation
Autosome	Translocation	Anneal
Sex Chromosome	Pre-natal / Antenatal	Polymerase Chain Reaction
Karyotype	Screening	Gel Electrophoresis
Homologous	Diagnosis	Saviour Siblings
Gene	Trimester	Genetic Counselling



Bio 3.2 AS 90714 vs 2 Research a Contemporary Biological Issue

Aspects of reproductive technologies offer good potential as topics for this achievement standard. When selecting a topic you are looking for an issue which will meet the following criteria:

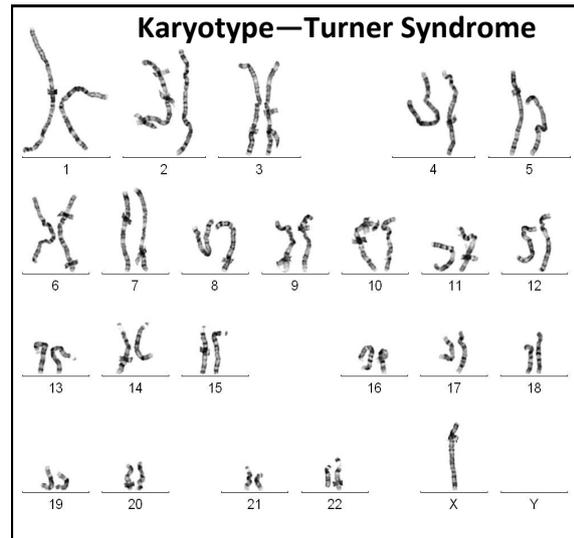


- Is it a contemporary issue? This is defined as an issue for which people hold different opinions or viewpoints.
- Can you define the biological concepts and processes relating to the issue?
- Can you define the implications of the issue—biological, social, ethical, economic or environmental?

While the use of reproductive technologies such as IVF are generally accepted in society, advances in reproductive technologies that have led to the availability of **Pre-implantation Genetic Diagnosis**. This technology offers good potential as a topic for this achievement standard. Issues around decisions relating to **when PGD should be used** and **how it should be made available** would offer a good starting point for your writing. A discussion around identifying where use of this technology is appropriate would be very relevant. The use of PGD to create **Saviour Siblings** is one particular aspect of the issue that would make an interesting topic to explore for this standard. There are extensive links provided on LENSscience Wiki Site designed to support your research if you choose this as a topic for Bio 3.2.

Post Seminar Challenge Questions

1. Aneuploidy resulting in the loss of an entire chromosome usually results in a non-viable embryo. However, if the chromosome concerned is the X-chromosomes the embryo may live. Explain why the loss of an entire autosome is almost always lethal but the loss of the X-chromosome may not be lethal.



2. Compare and contrast the **three** possible mechanisms by which Trisomy 21 can arise.
3. Discuss how the use of named **biotechnologies** have enabled scientists to develop effective methods of **diagnosing genetic abnormalities in embryos** and how these technologies have met a human need or demand.
4. Pre-natal diagnosis occurs **during** pregnancy and involves either Chorionic Villus Testing or Amniocentesis. Pre-implantation diagnosis occurs **before** a pregnancy is established and uses a combination of IVF technologies and genetic testing technologies such as Fluorescence *in situ* hybridization. These technologies have been established to meet a human need and demand.
 - (a) Define the human need and demand that has led to the development of these technologies and discuss the biological issues that underpin this need.
 - (b) Discuss the ethical issues that arise from the use of these reproductive technologies. Consider whether there are any differences between the issues related to the two types of diagnosis testing (pre-natal vs. PGD) from the perspective of a range of identified groups within New Zealand society.



Login and contribute your answers at

http://lens.auckland.ac.nz/index.php/Seminar_3_2011_Discussion_Page



Level 3 Achievement Standards linking to this seminar:

AS 90714 Biology 3.2 Research a contemporary biological issue

AS 90715 Biology 3.3 Describe the role of DNA in relation to gene expression

AS 90718 Biology 3.6 Describe applications of biotechnological techniques



Key Concepts from Level 3 Biology that link to this seminar:

Below are selected objectives from the Year 13 biology programme that link to this seminar. THESE ARE NOT A FULL LIST OF THE CONCEPTS IN YOUR COURSE. You may wish to review these concepts before the seminar.

Molecular Genetics / Mutations

Please remember these are only the objectives linking to this seminar—refer to your unit hand out at school for a full list

- Describe the principles of simple dominant / recessive monohybrid and dihybrid inheritance patterns and use these to predict the outcome of simple dominant / recessive monohybrid and dihybrid inheritance problems.
- Determine the outcome of inheritance crosses involving linked and sex-linked genes.
- Describe the structure of a chromosome.
- Describe the process of mitosis and meiosis.
- Define the term mutation and identify ways in which mutations may affect an organism.
- Identify the causes of mutations.
- Differentiate between somatic and gametic mutations and identify the potential effect of each of these.
- Describe the effect of gene mutations; identify types of gene mutations and explain the range of potential consequences of these on the expression of the gene.
- Describe the effect of chromosome mutations; identify types of chromosome mutations and the range of potential consequences of these on the expression of the gene.
- Describe the process that leads to aneuploidy and polyploidy and explain the range of potential consequences of these types of mutations.
- Show understanding of molecular genetics by using the core knowledge to give reasons for events that happen in the cell and linking ideas.
- Show understanding of the cause and effect of mutations by using the core knowledge to link ideas.

Biotechnology

Please remember these are only the objectives linking to this seminar—refer to your unit hand out at school for a full list

- Describe the techniques involved in gene cloning and how gene cloning meets human needs and demands.
- Describe the techniques involved in DNA profiling and how DNA profiling meets human needs and demands.
- Describe the techniques involved in genome analysis and how genome analysis meets human needs and demands.
- Be aware of the differing viewpoints of the use of biotechnological applications.
- Show understanding of applications of biotechnological techniques by using core knowledge to link ideas.

Contemporary Biological Issue

- Identify a contemporary issue - defined as one for which people hold different opinions or viewpoints.
- Define the biological concepts and processes relating to the issue.
- Define the implications of the issue—biological, social, ethical, economic or environmental.