

LENScience Senior Biology Seminar Series

Insight into Blindness

Questions and Discussion

Pre-seminar School Discussion

This seminar will take you on a very special journey with a whanau who have themselves journeyed to discover the genetics behind the serious visual impairment that has plagued the family for 5 generations. As you read through the paper, take time to consider how you would feel if it was your family finding out the reason behind a serious condition like this.

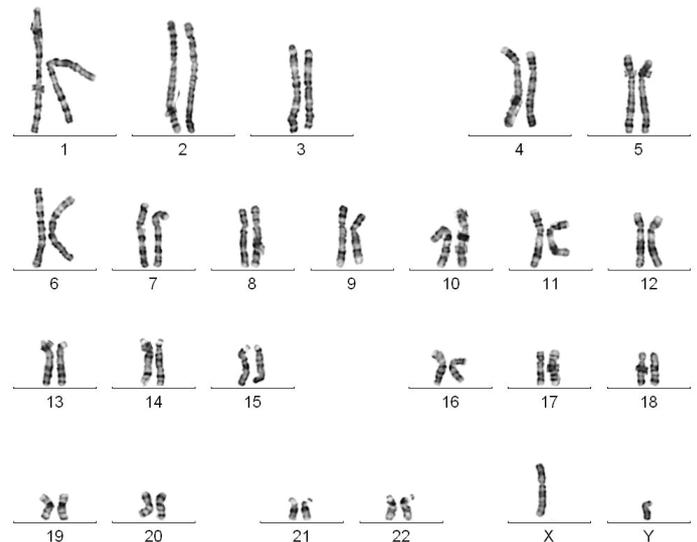


The genetics of this condition are very interesting. In the paper there are questions that will help you work your way through the family tree diagrams. During the seminar, members of the research team will help take you through these diagrams and explain what they tell us about the genetics of the condition.

A Review of Processes and Patterns of Evolution

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

1. Define the term sex-linked genetic condition and explain why a mutation with found on the x-chromosome will may affect females differently to males.
2. During meiosis, variation is increased through independent assortment (shuffling of chromosomes) and crossing over. Describe each of these processes and explain how they contribute to variation.
3. Describe what is meant by the term "linked genes". Explain why linked genes are less likely to be separated during crossing over.
4. Explain the difference between a DNA profiling and DNA sequencing.
5. Gene mutations can be caused by insertions, deletions or substitutions. Explain why only some mutations cause a change to the structure of the protein that is coded for by a particular gene.



Vocabulary

Allele	Gene	Pedigree
Anneal	Genetic Counselling	Phenotype
Autism	Genome	Photoreceptors
Autosome	Genotype	Plasmid
Chromatid	Hemizygous	Polymerase Chain Reaction
Chromosome	Heterozygous	Restriction Enzymes
Cone	Homologous	Retina
Crossing Over	Ion Channels	Rod
Gel Electrophoresis	Karyotype	Sex Chromosome
Gene Mutation	Ligation	Variable Tandem Repeats
	Linked gene	Vector
	Microsatellite Markers	X-linked

Post-Seminar Challenge Questions

- Explain what a Barr Body is and why they are only found in females.
 - How could the presence of Barr Bodies may affect the phenotype of a female who is:
 - Heterozygous for an x-linked gene
 - Homozygous for an x-linked gene
 - All the females in the family who are affected are heterozygous for the mutation. Within the females in the whanau, there is variation in the severity of the vision impairment. What biological explanation could account for this variation in the heterozygous female phenotype?

Use fig 2 on page 4 of the seminar paper to answer the following questions

- What evidence from the family tree suggests that this is an x-linked condition?

Use fig 5 on page 7 of the seminar paper to answer the following questions

- Compare person 16 in generation VII (affected) with person 17 in generation VI (unaffected). What does the difference between the microsatellite pattern for these two individuals tells us about where on the chromosome the mutation is? Look at the sequence **"2 2 6"**.
- Compare person 45 in generation VII (unaffected) with person 48 in generation VII (affected). The difference between these two individuals confirmed for the scientists that the mutation must be in the middle section of the chromosome where the satellite pattern **"1 1 3 2"** was found. What evidence did they use to come to this conclusion?



Level 3 Achievement Standards linking to this seminar:

AS 90718	Describe applications of biotechnological techniques
AS 90714	Research a Contemporary Biological Issue
AS 90715	Describe the role of DNA in relation to gene expression

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.
- Describe the techniques that would be used to insert a gene of interest into a cell using an expression vector such as a plasmid.
- 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, economic or environmental