

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?

