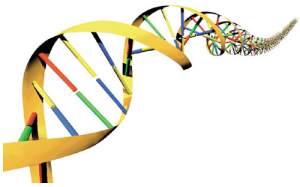


Year 10 Genetics Counselling Task

Introduction

In this task you will work in groups of 3 to research a genetic disease. The information you d
develop will be used by your group to counsel a couple who want to have children, but are
may inherit this disease. You must consider all of the information a couple may want or nee
make a decision on whether or not to have offspring.

To assist you in giving the consultation you are required to develop support materials. They will be used both to prompt you during the consultation and to help explain things to the client. These materials should take a variety of forms such as written, visual, auditory……can you think of anything else?

What will happen on presentation day?

- Half the groups in the class will set up a consultation room (these are the consultation groups)
- The remaining groups (the client groups) will be given a scenario. This scenario will direct them to a specific consultation area.
- Two people in the client group will be counselled, while the third films the consultation.
- The consultation group must help the couple based on the specific scenario **and** the information they have researched. All group members must contribute equally to the consultation and will be given **10 minutes** to complete their consultation.
- The clients should remember to ask questions when they do not understand to give the consultation group an opportunity to re-explain a concept.
- The consultation groups and client groups will swap and repeat the process above.
- Each group will evaluate the group they were counselled by using a rubric provided to them after all groups have presented.

How long will this task take?

The full task will take 5 periods. The following table shows what you will be expected to do in each of these periods. A planning sheet will also be provided to assist you.

Period	Work to complete
1	Decide what to research & begin research
2	Research & develop support materials
3	Research & develop support materials
4	Research & develop support materials
5	Complete consultation

How will we be assessed?

You will be assessed in 3 ways:

- *Teacher assessment:* video of the consultation.
- *Teacher assessment:* choose ONE piece of your support material for your teacher to look at in depth and submit this after the consultation, as well as any additional materials you create during the consultation to assist the understanding of your clients.
- *Peer assessment:* your clients will complete an evaluation form of your consultation.

Questions?

Planning Sheet

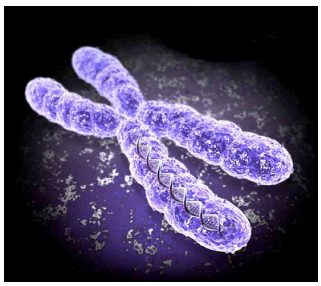
To perform your duties as a genetic counsellor effectively, you will first need to determine what a genetic counsellor actually does and what sort of information they provide to their clients. What would **YOU** want to know if you wanted to have a child who could potentially inherit a genetic disease? Remember your clients are not geneticists.

Research the role of a genetic counsellor and brainstorm what information you will need to find:

Role of a genetic counsellor:

What information will we need to research about our disease and how it is inherited?

Consider **ALL** the information we have covered so far in genetics. How can you apply this to your genetic counselling session? How will you work through a specific scenario and inform the parent both about the specific disease and whether or not their child will inherit it. What will you need to know and need to be able to do? Write your ideas here:



What materials do you need to create to help you complete the consultation? Remember this will be handed in for assessment. Will this involve audio, written material, visual material.....you MUST let your teacher know what you require so it can be booked in time to be used in your planning lessons.

On consultation day what materials might be useful to help you work through the specific scenario that you will be given? Will you need anything else other than your support materials? Will you use any props?

Who will be responsible for what? Divide your tasks evenly to ensure your group works productively.

Group Member	Responsible For

Peer Evaluation Sheet

Genetic counselling group (group being assessed): _____

Client group (group assessing): _____

Use the following rubric to rate how well your genetic counsellors covered the following criteria:

Criteria				
The disease and its symptoms	The group gave an overview of the disease and described in detail the symptoms that an individual with the disease may experience.	The group gave an overview of the disease and described some symptoms that an individual with the disease may experience.	The group didn't really explain the disease and only mentioned a few symptoms.	The consultation did not cover this section.
Life expectancy and quality of life	The group stated how long individuals with the disease typically live and explained in detail the quality of life expected.	The group stated how long individuals typically live and briefly explained the quality of life expected.	The group stated how long individuals typically live but did not mention quality of life.	The consultation did not cover this section.
Chances of the child inheriting the disease	The group explained the chance of a child of the couple inheriting the disease (either as a percentage, ratio or fraction) and showed the couple precisely how this was determined using Punnett squares **AND** pedigrees.	The group stated the chance of a child of the couple inheriting the disease (either as a percentage, ratio or fraction) and showed the couple how this was determined using Punnett squares **OR** a pedigree.	The group stated the chance of child of the couple inheriting the disease (either as a percentage, ratio or fraction) but did not show the couple how this was determined.	The consultation did not cover this section.
Treatment and management of the disease if the child should inherit it	The group explained the different options for treatment or management in detail.	The group explained some options for treatment or management of the disease.	The group briefly explained one treatment or management option.	The consultation did not cover this section.
Impact on parents and where they can get support	The group discussed in detail the impact the disease is likely to have on the parents and provided a number of places the couple can get support.	The group briefly discussed the impact the disease is likely to have on the parents and provided some places the couple can get support.	The group mentioned the impact the disease is likely to have on the parents **OR** provided some places the couple can get support.	The consultation did not cover this section.
Genetic testing	Explained whether it can be determined if the baby has the disease before it is born, how this might occur and what the risks are.	Explained whether it can be determined if the baby has the disease before it is born and briefly states how this will occur but does not discuss the risks.	States whether it can be determined if the baby has the disease before it is born but does not explain how this might occur or whether there are any risks.	The consultation did not cover this section.

PTO

Communication skills and use of genetic terminology	The information was very well communicated and easy to understand and genetic terminology was used where appropriate.	The information was mostly well communicated and easy to understand and genetic terminology was used where appropriate.	The information was not well communicated and hard to understand. Genetic terminology was not used correctly **OR** the group gave the clients their support material and did not offer any further explanations.	The consultation did not cover this section.
Use of support material	The group supported their explanation with other material such as audio or visual components. These components were relevant, creative and easy to understand.	The group supported their explanation with other material such as audio or visual components. These components were relevant and mostly easy to understand.	The group supported their explanation with other material such as audio or visual components but it was difficult to see their relevance or understand the content.	The consultation did not cover this section.
Group Work	All group members contributed equally during the consultation	The group members mostly contributed equally during the consultation.	One group member was mostly responsible for the consultation and received minimal assistance.	One group member completed the consultation and received no assistance.

Comments: _____

_____

_____

_____

_____

_____

_____

Disease Cards

HAEMOPHILIA	**HAEMOPHILIA**
DUCHENNE'S MUSCULAR DYSTROPHY	**DUCHENNE'S MUSCULAR DYSTROPHY**
TAY-SACHS DISEASE	**TAY-SACHS DISEASE**
CYSTIC FIBROSIS	**CYSTIC FIBROSIS**
HUNTINGTON'S DISEASE	**HUNTINGTON'S DISEASE**

Scenario Cards

Scenario 1: Haemophilia Susan and John want to have a child. John has haemophilia, so they are concerned that their child could inherit the disease. Susan does not have haemophilia and neither did her mother, but her father did. Counsel Susan and John based on the instructions you were provided and the materials you have developed.	***Scenario 2: Haemophilia*** Susan and John want to have a child. John has haemophilia, so they are concerned that their child could inherit the disease. Susan does not have haemophilia but knows she is a carrier. Counsel Susan and John based on the instructions you were provided and the materials you have developed.
Scenario 3: Duchenne's Muscular Dystrophy Sam and Evelyn want to have a second child. Their first son, James, had Duchenne's Muscular Dystrophy and are worried subsequent children could also inherit the disease. Neither of the parents are affected by the disease. Counsel Sam and Evelyn based on the instructions you were provided and the materials you have developed.	***Scenario 4: Duchenne's Muscular Dystrophy*** Sam and Evelyn want to have a child. Sam has the disease and they have recently discovered that Evelyn is a carrier. Counsel Sam and Evelyn based on the instructions you were provided and the materials you have developed.
Scenario 5: Tay Sachs Disease Christie and Robert want to try and have another child. Unfortunately their first two daughters died of Tay-Sachs disease and they are worried any subsequent children will also be affected by the disease. Counsel Christie and Robert based on the instructions you were provided and the materials you have developed.	***Scenario 6: Tay Sachs Disease*** Christie and Robert want to try and have another child. Christie has always known she is a carrier of Tay Sachs disease and suggested Robert get tested. It was found that he was also a carrier. Counsel Christie and Robert based on the instructions you were provided and the materials you have developed.
Scenario 7: Cystic Fibrosis Lynne and Ben want to have another child. Lynne has cystic fibrosis but Ben is not affected, and they already have a daughter, Alexandria, with the disease. Counsel Lynne and Ben based on the instructions you were provided and the materials you have developed.	***Scenario 8: Cystic Fibrosis*** Lynne and Ben want to have another child. Lynne has cystic fibrosis and Ben is a carrier. Counsel Lynne and Ben based on the instructions you were provided and the materials you have developed.

Scenario 9: Huntington's Disease Samantha and Joe want to have a child. Both parents have Huntington's disease. Their first child, Kate, was tested and it was determined that she did not have the disease. However, Samantha and Joe are worried about subsequent children being affected. Counsel Samantha and Joe based on the instructions you were provided and the materials you have developed.	***Scenario 10: Huntington's Disease*** Samantha and Joe want to have a child. Samantha knows she has Huntington's disease, which she inherited from her father – her mother was not affected. Joe does not suffer Huntington's but The couple are worried about their children inheriting Huntington's disease. Counsel Samantha and Joe based on the instructions you were provided and the materials you have developed.

Teacher Notes on Planning Sheet

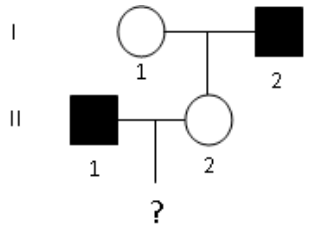
Please work through this sheet with students so they are aware of the range of things they will need to speak to their client about during the consultation. i.e. they will not just have to work with Punnett squares and pedigrees, but also explain about the disease, its symptoms etc (see peer assessment rubric).

Teacher Notes on Peer Evaluation Sheet

After all consultations have been completed, each client group should fill out a peer evaluation sheet to assess their consultation group. Students should not see this sheet before their consultation.

Teacher Notes on Scenarios

<table>
<tr><th>Scenario</th><th>Category</th><th>Chances of Child Inheriting the Disease</th></tr>
<tr><td>Scenario 1: Haemophilia
Susan and John want to have a child. John has haemophilia, so they are concerned that their child could inherit the disease. Susan does not have haemophilia and neither did her mother, but her father did. Counsel Susan and John based on the instructions you were provided and the materials you have developed.</td><td>Hard</td><td><u>Mode of inheritance:</u> X-linked recessive

<u>Pedigree</u>
 

<u>Genotypes of individuals concerned:</u>

I1 – Susan’s mother: X^RX^R or X^RX^r
- Doesn’t have the disease so must carry the dominant allele
- We have no information about her parents, so do not know if she is a carrier or not

I2 – Susan’s father: X^rY
- Has haemophilia so must carry the recessive allele

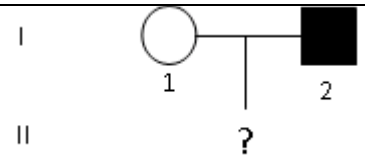
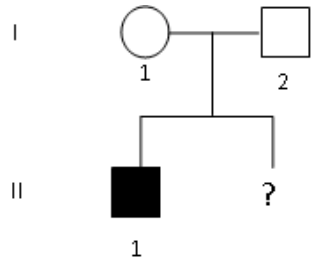
II1 – John: X^rY
- Has haemophilia so must carry the recessive allele

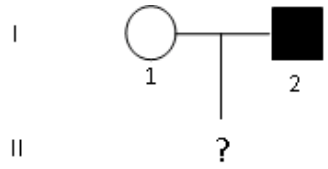
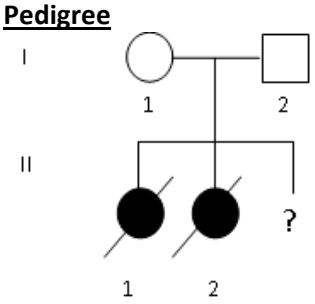
II2 – Susan: X^RX^r
- Doesn’t have the disease so must carry the dominant allele
- Can only inherit the recessive allele from her father

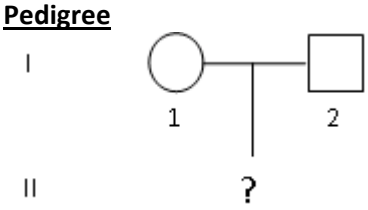
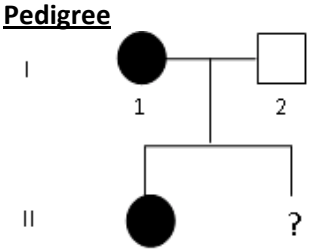
<u>Punnett square showing possible children of Susan and John:</u>

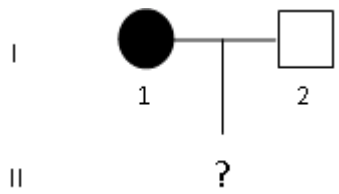
<table><tr><td></td><td>X^R</td><td>X^r</td></tr><tr><td>X^r</td><td>$X^R X^r$</td><td>$X^r X^r$</td></tr><tr><td>Y</td><td>$X^R Y$</td><td>$X^r Y$</td></tr></table>
<u>Chance of a child getting the disease:</u>
50%</td></tr>
<tr><td>Scenario 2: Haemophilia
Susan and John want to have a child. John has haemophilia, so they are concerned that their child could inherit the disease. Susan does not have</td><td>Easy</td><td><u>Mode of inheritance:</u> X-linked recessive

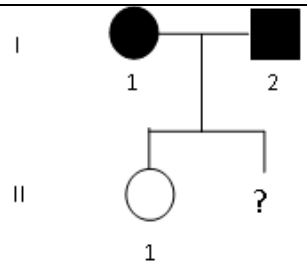
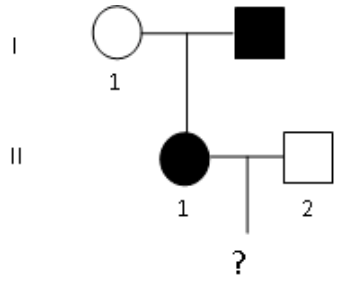
<u>Pedigree</u></td></tr>
</table>

haemophilia but knows she is a carrier. Counsel Susan and John based on the instructions you were provided and the materials you have developed.		 **Genotypes of individuals concerned:** I1 – John: X^rY - Has haemophilia so must carry the recessive allele I2 – Susan: X^RX^r - Doesn't have the disease so must carry the dominant allele - Is a carrier so must have the recessive allele **Punnett square showing possible children of Susan and John:** | | X^R | X^r | | X^r | $X^R X^r$ | $X^r X^r$ | | Y | X^R Y | X^r Y | **Chance of a child getting the disease:** 50%
Scenario 3: Duchenne's Muscular Dystrophy Sam and Evelyn want to have a second child. Their first son, James, had Duchenne's Muscular Dystrophy and are worried subsequent children could also inherit the disease. Neither of the parents are affected by the disease. Counsel Sam and Evelyn based on the instructions you were provided and the materials you have developed.	Hard	**Mode of inheritance:** X-linked recessive **Pedigree**  **Genotypes of individuals concerned:** I1 – Evelyn: X^RX^r - Doesn't have the disease so must carry the dominant allele - Has had a son with the disease – his affected allele must be inherited from his mother as he must inherit a Y chromosome from his father I2 – Sam: X^RY - Does not have the disease so must have the dominant allele II1 – James: X^rY - Has the disease so must have the recessive allele **Punnett square showing possible children of Sam and Evelyn:** | | X^R | X^r | | X^R | $X^R X^R$ | $X^R X^r$ | | Y | X^R Y | X^r Y |

		Chance of a child getting the disease: 25%
Scenario 4: Duchenne's Muscular Dystrophy Sam and Evelyn want to have a child. Sam has the disease and they have recently discovered that Evelyn is a carrier. Counsel Sam and Evelyn based on the instructions you were provided and the materials you have developed.	Easy	**Mode of inheritance:** X-linked recessive **Pedigree**  **Genotypes of individuals concerned:** I1 – Evelyn: X^RX^r - Doesn't have the disease so must carry the dominant allele - Is a carrier so must have the recessive allele I2 – Sam: X^rY - Has the disease so must carry the recessive allele **Punnett square showing possible children of Sam and Evelyn:** | | X^R | X^r | | X^r | $X^R X^r$ | $X^r X^r$ | | Y | X^R Y | X^r Y | **Chance of a child getting the disease:** 50%
Scenario 5: Tay Sachs Disease Christie and Robert want to try and have another child. Unfortunately their first two daughters died of Tay-Sachs disease and they are worried any subsequent children will also be affected by the disease. Counsel Christie and Robert based on the instructions you were provided and the materials you have developed.	Hard	**Mode of inheritance:** Autosomal recessive **Pedigree**  **Genotypes of individuals concerned:** I1 – Christie: Rr - Doesn't have the disease so must carry the dominant allele (Tay Sachs individuals die very young) - Had daughters with Tay Sachs therefore must carry the recessive allele I2 – Robert: Rr - Doesn't have the disease so must carry the dominant allele (Tay Sachs individuals die very young) - Had daughters with Tay Sachs therefore must carry the recessive allele

		II1 & 2 – Affected Daughters: rr - Must have two affected alleles to show the disease **Punnett square showing possible children of Christie and Robert:** | | R | r | | R | RR | Rr | | r | Rr | rr | **Chance of a child getting the disease:** 25%
Scenario 6: Tay Sachs Disease Christie and Robert want to try and have another child. Christie has always known she is a carrier of Tay Sachs disease and suggested Robert get tested. It was found that he was also a carrier. Counsel Christie and Robert based on the instructions you were provided and the materials you have developed.	Easy	**Mode of inheritance:** Autosomal recessive **Pedigree**  **Genotypes of individuals concerned:** I1 – Christie: Rr - Doesn't have the disease so must carry the dominant allele (Tay Sachs individuals die very young) - Is a carrier so must have the recessive allele I2 – Robert: Rr - Doesn't have the disease so must carry the dominant allele (Tay Sachs individuals die very young) - Is a carrier so must have the recessive allele **Punnett square showing possible children of Christie and Robert:** | | R | r | | R | RR | Rr | | r | Rr | rr | **Chance of a child getting the disease:** 25%
Scenario 7: Cystic Fibrosis Lynne and Ben want to have another child. Lynne has cystic fibrosis but Ben is not affected, and they already have a daughter, Alexandria, with the disease. Counsel Lynne and Ben based on the instructions you were provided and the materials you have developed.	Hard	**Mode of inheritance:** Autosomal recessive **Pedigree**  **Genotypes of individuals concerned:** I1 – Lynne: rr - Has the disease so must have two recessive alleles

		I2 – Ben: Rr - Doesn't have the disease so must carry the dominant allele - Has a child with the disease so must carry the recessive allele II1 – Alexandria: rr - Has the disease so must have two recessive alleles **Punnett square showing possible children of Lynne and Ben:** | | R | r | | r | Rr | rr | | r | Rr | rr | **Chance of a child getting the disease:** 50%
Scenario 8: Cystic Fibrosis Lynne and Ben want to have another child. Lynne has cystic fibrosis and Ben is a carrier. Counsel Lynne and Ben based on the instructions you were provided and the materials you have developed.	Easy	**Mode of inheritance:** Autosomal recessive **Pedigree**  **Genotypes of individuals concerned:** I1 – Lynne: rr - Has the disease so must have two recessive alleles I2 – Ben: Rr - Doesn't have the disease so must carry the dominant allele - Is a carrier so must have the recessive allele **Punnett square showing possible children of Lynne and Ben:** | | R | r | | r | Rr | rr | | r | Rr | rr | **Chance of a child getting the disease:** 50%
Scenario 9: Huntington's Disease Samantha and Joe want to have a child. Both parents have Huntington's disease. Their first child, Kate, was tested and it was determined that she did not have the disease. However, Samantha and Joe are worried about subsequent	Hard	**Mode of inheritance:** Autosomal dominant **Pedigree**

children being affected. Counsel Samantha and Joe based on the instructions you were provided and the materials you have developed.		 **Genotypes of individuals concerned:** I1 – Samantha: Rr - Has the disease so must have the dominant allele - Has a child without the disease so must have the recessive allele I2 – Joe: Rr - Has the disease so must have the dominant allele - Has a child without the disease so must have the recessive allele II1 – Kate: rr - Doesn't have the disease so must have two recessive alleles **Punnett square showing possible children of Samantha and Joe:** | | R | r | | R | RR | Rr | | r | Rr | rr | **Chance of a child getting the disease:** 25%
Scenario 10: Huntington's Disease Samantha and Joe want to have a child. Samantha knows she has Huntington's disease, which she inherited from her father – her mother was not affected. Joe does not suffer Huntington's but The couple are worried about their children inheriting Huntington's disease. Counsel Samantha and Joe based on the instructions you were provided and the materials you have developed.	Hard	**Mode of inheritance:** Autosomal dominant **Pedigree**  **Genotypes of individuals concerned:** I1 – Samantha's mother: rr - Doesn't have the disease so must have two recessive alleles I2 – Samantha's father: HH or Hh - Has the disease so must have the dominant allele - Unable to determine if he is homozygous or heterozygous II1 – Samantha: Rr - Has the disease so must have the dominant allele - If her mother was not affected, Samantha must have inherited a recessive allele from her

II2 – Joe: rr

- Doesn't have the disease so must have two recessive alleles

Punnett square showing possible children of Samantha and Joe:

	R	r
r	Rr	rr
r	Rr	rr

Chance of a child getting the disease:

50%