Ministry Curriculum Expectations

D1.1 analyse, on the basis of research, some of the social and ethical implications of research in genetics and genomics (e.g., genetic screening, gene therapy, in vitro fertilization) [IP, PR, AI, C]

Initiating and Planning [IP]
A1.3 identify and locate a variety of print and electronic sources that enable them to address research topics fully and appropriately

Performing and Recording [PR]
A1.6 compile accurate data from laboratory and other sources, and organize and record the data, using appropriate formats, including tables, flow charts, graphs, and/or diagrams

Analysing and Interpreting [AI]
A1.10 draw conclusions based on inquiry results and research findings, and justify their conclusions with reference to scientific knowledge

Communicating [C]
A1.11 communicate ideas, plans, procedures, results, and conclusions orally, in writing, and/or in electronic presentations, using appropriate language and a variety of formats (e.g., data tables, laboratory reports, presentations, debates, simulations, models)

D3.1 explain the phases in the process of meiosis in terms of cell division, the movement of chromosomes, and crossing over of genetic material

D3.4 describe some genetic disorders caused by chromosomal abnormalities (e.g., non-disjunction of chromosomes during meiosis) or other genetic mutations in terms of chromosomes affected, physical effects, and treatments

Sources of Ideas and References


**Notes Regarding Implementation**

- This dry lab may be too wordy for some students since it has a lot of reading and writing (this is a limitation); however the teacher could modify and decrease the amount of text on the first page depending on how much background knowledge the students have before this assignment
- If required, the teacher could give some students a copy of the definition sheet from the teacher notes (if some students don’t have enough background knowledge) or direct them to the course textbook to look these up
- Teacher can scaffold this lab by directing students to the appropriate pages in the textbook for some relevant information about the different disorders
- Students could be given time in the computer lab to complete the dry lab so that they are able to perform research on the topics addressed
Karyotyping and Chromosomal Disorders Lab

As a student in Grade 11 University Biology, you have been granted an honorary position in the Canadian College of Medical Geneticists or CCMG. Due to a shortage of doctors, the CCMG needs your help in helping to diagnose one of their patients, Patient A, who is suffering from a chromosomal disorder. The CCMG knows that Patient A is suffering from 1 of 4 possible chromosomal disorders:

1. Down syndrome
2. Klinefelter syndrome
3. Turner syndrome
4. Trisomy X

To help you in this task, the CCMG has provided you with data in the form of a karyotype to help study Patient A’s chromosomes. In general, to create a karyotype, cell samples from patients (usually white blood cells are used but theoretically, any cell could be used) are exposed to chemicals that induce mitosis and halt cell division during metaphase. As a result, genes are essentially frozen in time. Homologous pairs of chromosomes are identified by their size (length), shape, and banding pattern. To help visualize the cells, they are stained using special dyes to allow their chromosomes to be seen and then photographed.

To create a karyotype, the chromosome pairs are arranged by length from longest to shortest (with the exception of sex chromosomes which always appear last), and each pair is assigned a number between 1 and 23. The longest chromosome pair is designated number 1 and the longest is designated 22. The number 23 is reserved for the sex chromosomes, which allow for the determination of a person’s sex. Most human cells have 23 pairs of chromosomes. A normal male’s karyotype has 44 autosomes, one X chromosome, and one Y chromosome (XY) whereas a normal female’s karyotype has 44 autosomes and then two X chromosomes (XX).

Sometimes, chromosomal material is lost or rearranged during the formation of gametes or during cell division. These changes are usually the result of non-disjunction or translocation. Some of the abnormalities associated with chromosome structure and number can be detected by using a karyotype. After a karyotype has been assembled, scientists can examine the karyotype for signs of a chromosomal disorder. Scientists use karyotypes for many different purposes, including studying the relationships between species and diagnosing genetic disorders.

Your job is to examine the karyotype given below and use the knowledge you have gained in this unit so far to make a diagnosis. The corresponding questions will help you with this task. Good luck!
Karyotype and Chromosomal Disorders Lab: Questions

Please complete the following questions and hand them in.

1. What point in the life cycle of the cell would be best for creating a karyotype and why? (K/U: 3 marks)

2. How many chromosomes does a normal human being have? How many of these are autosomes and how many of these are sex chromosomes? (K/U: 3 marks)

3. How many chromosome pairs are shown in any normal karyotype? (K/U: 1 mark)

4. What is the sex of Patient A and how did you determine this? (APP: 2 marks)

5. Complete the following chart using your notes, textbook, or any other resource: (K/U: 12 marks)

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Description (Chromosomes)</th>
<th>Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down syndrome</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Klinefelter syndrome</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Turner syndrome</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Trisomy X</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
6. How many chromosomes does Patient A have and how does this compare to the number of chromosomes found in a normal human being? (APP: 2 marks)

7. Based on your findings, and using the information from Question 5, what diagnosis would you give Patient A? Justify your answer using your own knowledge as well as information provided by the karyotype. (APP: 2 marks)

8. What types of symptoms could Patient A expect based on your diagnosis? (APP: 2 marks)

9. Is there any treatment available for this chromosomal disorder? If so, briefly describe the treatment options. (APP: 3 marks)

10. The CCMG would like to thank you for your support in diagnosing Patient A. Since you did such a good job, they would like your help one last time to help them diagnose Patient B, Patient C, and Patient D (karyotypes on the next few pages).
My Diagnosis for Patient B (APP: 3 marks)

Sex:

Chromosomal Disorder:

Justification:
Karyotype of Patient C

My Diagnosis for Patient C (APP: 3 marks)

Sex:

Chromosomal Disorder:

Justification:
My Diagnosis for Patient D (APP: 3 marks)

Sex:

Chromosomal Disorder:

Justification:
11. Give two reasons why a pregnant woman **would** want a karyotype of her baby. 
   (APP: 2 marks, COMM: 1 mark)

12. Give two reasons why a pregnant woman **would not** want a karyotype of her baby. (APP: 2 marks, COMM: 1 mark)
Karyotype and Chromosomal Disorders Lab: Answers

Please look at the suggested marking scheme below.

1. What point in the life cycle of the cell would be best for creating a karyotype and why? (K/U: 3 marks)

   During the metaphase stage of cell division, the chromosomes can be seen most clearly because they have been replicated and condense into tightly coiled structures. For this reason, scientists use images of cells undergoing metaphase to study chromosome shape and structure and create karyotypes because they are clearly visible under a microscope.

2. How many chromosomes does a normal human being have? How many of these are autosomes and how many of these are sex chromosomes? (K/U: 3 marks)

   A normal human being has a total of 46 chromosomes. 44 of these are autosomes whereas the other 2 are sex chromosomes (XX or XY)

3. How many chromosome pairs are shown in any normal karyotype? (K/U: 1 mark)

   23 chromosome pairs are seen in any normal karyotype.

4. What is the sex of Patient A and how did you determine this? (APP: 2 marks)

   Patient A is a male because he has one X chromosome and one Y chromosome.

5. Complete the following chart by using your notes, textbook, or any other resource: (K/U: 12 marks)

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Description (Chromosomes)</th>
<th>Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down syndrome</td>
<td>- extra copy of chromosome 21</td>
<td>- poor muscle tone&lt;br&gt;- slanting eyes with folds of skin at the inner corners&lt;br&gt;- short and broad hands and feet&lt;br&gt;- small head&lt;br&gt;- respiratory problems&lt;br&gt;- speech problems&lt;br&gt;- visual problems&lt;br&gt;- hearing loss&lt;br&gt;- heart defects and gastrointestinal problems&lt;br&gt;- slow intellectual development</td>
</tr>
<tr>
<td>Klinefelter syndrome</td>
<td>- extra X chromosome in males (XXY)</td>
<td>- affects only males&lt;br&gt;- secondary sex characteristics fail to develop properly&lt;br&gt;- testicular changes take place in puberty, which typically results in infertility</td>
</tr>
<tr>
<td>Turner</td>
<td>- missing X</td>
<td>- affects only females</td>
</tr>
</tbody>
</table>
6. How many chromosomes does Patient A have and how does this compare to the number of chromosomes found in a normal human being? (APP: 2 marks)

Patient A has 47 chromosomes (3 copies of chromosome 21) whereas a normal human being has 46 chromosomes. Thus, Patient A has one extra chromosome.

7. Based on your findings, and using the information from Question 5, what diagnosis would you give Patient A? Justify your answer using your own knowledge as well as information provided by the karyotype. (APP: 2 marks)

Since Patient A has an extra copy of chromosome 21, I would say that he has Down syndrome. I say this because when I look at the karyotype, I can see 3 copies of chromosome 21 so this makes a total of 47 chromosomes instead of 46.

8. What types of symptoms could Patient A expect based on your diagnosis? (APP: 2 marks)

Since I think that Patient A has Down syndrome, he could experience any of the following symptoms:
- poor muscle tone
- asymmetrical or oddly shaped skull (or round head with flat area at the back of the head)
- small head
- slanting eyes with folds of skin at the inner corners
- flattened nose
- protruding tongue
- short and broad hands and feet
- single crease at the palm
- problems with growth and development
- slow intellectual development
- delayed mental and social skills (mental retardation)
- respiratory problems
- speech problems
- visual problems
- hearing loss
- heart defects and gastrointestinal problems
9. Is there any treatment available for this chromosomal disorder? If so, briefly describe the treatment options. (APP: 3 marks)

While there is no cure for Down syndrome, the major goal is to control symptoms and manage any resulting medical conditions. This includes regular checkups and screenings, medications, and surgery. Counseling and support groups are also aspects of treatment for those who need help in coping with the emotional and practical aspects of Down syndrome. Because of these medical conditions and their associated complications, a person with Down syndrome is at increased risk of premature death. Regular checkups are important. These visits help ensure that any changes in health are noted and treated if necessary. The doctor may also schedule certain screening tests to look for other medical problems before symptoms occur. This is important, given the increased risk of vision and hearing problems, infections, and cancer in people with Down syndrome. Furthermore, special education and training is offered in most communities and schools for mentally handicapped children.

10. The CCMG would like to thank you for your support in diagnosing Patient A. Since you did such a good job, they would like your help one last time to help them diagnose Patient B, Patient C, and Patient D (karyotypes on the next page).

Please note that if students are having trouble identifying the sex of the person’s karyotype, remind them that the presence or absence of the Y chromosome determines a person’s sex. Presence of the Y chromosome means that the person is a male, whereas absence of the Y chromosome means that the person is a female.

<table>
<thead>
<tr>
<th>My Diagnosis for Patient B (APP: 3 marks)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex: <strong>Female</strong></td>
</tr>
<tr>
<td>Chromosomal Disorder: <strong>Trisomy X</strong></td>
</tr>
<tr>
<td>Justification: <em>3 copies of the X chromosome are seen in the karyotype (XXX instead of XX).</em></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>My Diagnosis for Patient C (APP: 3 marks)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex: <strong>Male</strong></td>
</tr>
<tr>
<td>Chromosomal Disorder: <strong>Klinefelter syndrome</strong></td>
</tr>
<tr>
<td>Justification: <em>Extra copy of chromosome X in the karyotype (XXY instead of XY).</em></td>
</tr>
</tbody>
</table>
11. Give two reasons why a pregnant woman would want a karyotype of her baby. (APP: 2 marks, COMM: 1 mark)

- allows parents to determine the baby’s health and sex
- helps them to determine if any infertility problems they may be having are due to a genetic defect in the baby
- allows them to become mentally, emotionally, and financially prepared to handle the medical condition of the child (if any)
- allows parents to make informed decisions about the continuation or termination of a pregnancy

12. Give two reasons of why a pregnant woman would not want a karyotype of her baby. (APP: 2 marks, COMM: 1 mark)

- may be emotionally difficult to handle if there is a problem with the baby
- may lead to selective abortions where parents choose to abort the baby if he/she is not genetically perfect (designer babies)
- may lead to sex selection if their baby is not the sex they wanted (typically a problem in countries like India and China where females are selectively aborted because males are preferred)
- small possibility of false positives and false positives and abortion based on incorrect test results

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**My Diagnosis for Patient D** (APP: 3 marks)

**Sex:** Female

**Chromosomal Disorder:** Turner syndrome

**Justification:** Missing X chromosome in the karyotype (X instead of XX).
Some of the terms in the lab are bolded and it is assumed that the students have prior knowledge of these terms. However, the following is a list of the bolded words from the lab and some brief definitions (arranged in alphabetical order):

**Autosome:** a chromosome that does not carry the genes that determine gender (chromosomes 1-22); humans have 22 pairs of autosomes or 44 in total

**Chromosomal Disorder:** a type of genetic disorder that occurs when a person’s cells do not have the correct number of chromosomes; involves missing or extra copies of chromosomes or a change in chromosome structure; are typically caused when an error occurs during cell division and the chromosomes do not separate properly

**Chromosome:** rod-shaped structure composed of protein and DNA within a cell’s nucleus; every human cell contains two sets of 23 chromosomes (one set from the mother and one from the father) for a total of 46 chromosomes - chromosomes are passed from one generation to the next

**Karyotype:** a picture of a cell's complete set of chromosomes grouped together in pairs and arranged in order of decreasing size; used to detect chromosomal disorders and to study the relationship between different species

**Metaphase:** the second stage of cell division (following prophase and preceding anaphase) in which condensed & highly coiled chromosomes, carrying genetic information, align in the middle of the cell along the metaphase plate before being separated into each of the two daughter cells

**Non-disjunction:** an error that can occur during mitosis or meiosis in which both homologous chromosomes move to the same side of the dividing cell; one daughter cell receives two copies of the chromosome and the other daughter cell does not receive any copies

**Sex Chromosome:** chromosome carrying the genes that determine a person’s sex (chromosome pair 23); females have two copies of the X chromosome while males have one X chromosome and one Y chromosome

**Translocation:** a type of mutation in which a segment of a chromosome moves to a new position either within the same chromosome or onto a non-homologous chromosome