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RECOGNISING ACHIEVEMENT

ADVANCED SUBSIDIARY GCE

HUMAN BIOLOGY

Growth, Development and Disease

F222/ADVANCE NOTICE

For issue on or after:
17 NOVEMBER 2010

Tuesday 18 January 2011
Afternoon

Duration: 1 hour 45 minutes

OCE/32227

NOTES FOR GUIDANCE (CANDIDATES)

1 This document contains two case studies, which are needed in preparation for questions 1 and 2 in the externally assessed examination **F222**.

2 You will need to read the case studies carefully and also have covered the learning outcomes for Unit F222 (Growth, Development and Disease). The examination paper will contain questions on the two case studies. You will be expected to apply your knowledge and understanding of the work covered in F222 to answer these questions. There are 100 marks available on the paper.

3 You can seek advice from your teacher about the content of the case studies and you can discuss them with others in your class. You may also investigate the topics yourself using any resources available to you.

4 You will **not** be able to take your copy of the case studies, or other materials, into the examination. The examination paper will contain fresh copies of the two case studies as an insert.

5 You will not have time to read the case studies for the first time in the examination if you are to complete the examination paper within the specified time. However, you should refer to the case studies when answering the questions.

- This document consists of **8** pages. Any blank pages are indicated.

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Case Study 1

BRCA-1 FREE AT BIRTH

A baby girl 'designed' to be free of a breast cancer gene was born in January 2009 at a London hospital. Her parents had opted for genetic screening tests after three generations of her father's family developed an inherited form of breast cancer. The father is a carrier and those diagnosed include his mother, sister, grandmother and cousin.

Tests had shown that the father carried a faulty copy of the *BRCA-1* 'high risk' breast cancer gene. Although *BRCA-1* mutations are rare in the population as a whole, out of ten carriers of this faulty gene, between five and eight will go on to develop breast cancer, often at a young age (compared to one out of nine in the general population).

Geneticists, Douglas Easton and Paul Pharoah, have co-ordinated the collection of data from 15 investigators from around the world who have carried out studies of *BRCA-1* mutation carrier frequency in breast cancer cases. The main aim of their study was to estimate cancer risks associated with mutations in *BRCA-1*. The data collected was compared to data collected from families with no cases of breast cancer, sampled at random.

Fig. 1.1 shows the cumulative risk (%) of developing breast cancer in *BRCA-1* mutation carriers.

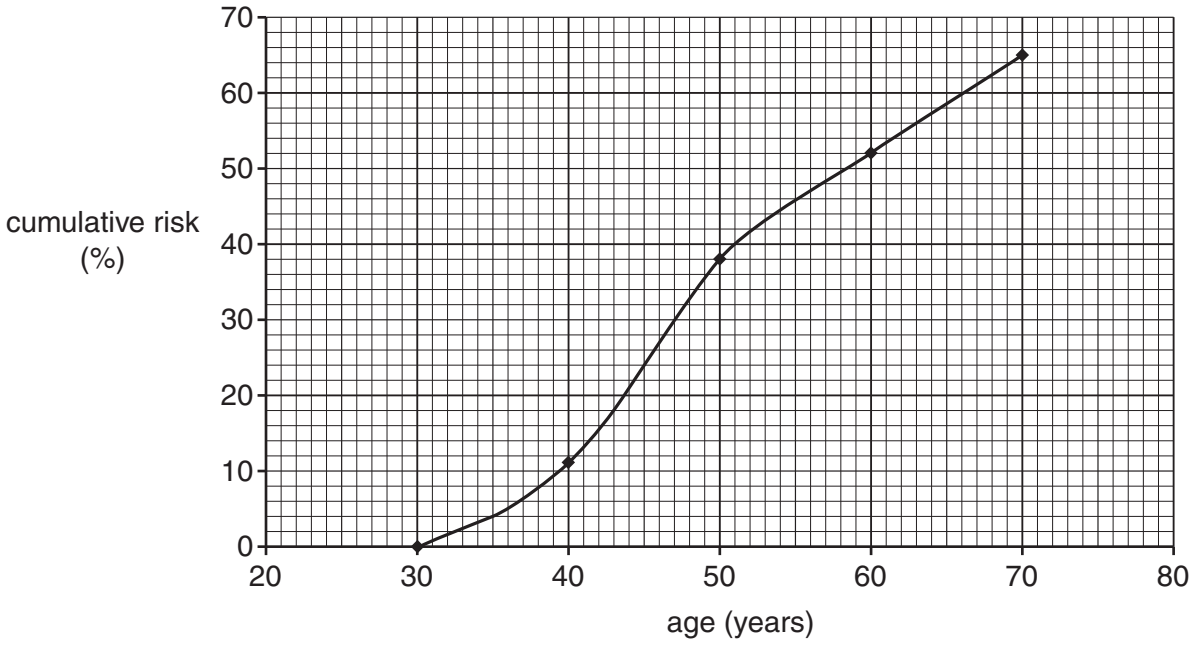

Fig. 1.1

The *BRCA-1* human gene belongs to a class of genes known as tumour suppressors. Mutations of this gene have been linked to the development of hereditary breast cancer. The test for *BRCA-1* is only offered to people with a strong family history of breast cancer and who have a living relative with breast cancer. This is because looking for a gene fault is a bit like looking for a single spelling mistake in a long book. The living relative with breast cancer has to be tested first to try and find out which fault in the breast cancer genes might run in the family. This is called the mutation search and may take a few months or more. If a faulty gene is found, the same gene fault can be investigated in other family members. This is called predictive testing.

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Most cases of breast cancer are sporadic, not familial, and are caused by gene damage (somatic mutations) acquired by breast cells during a woman's lifetime. A wide variety of genes that are commonly mutated or incorrectly regulated in sporadic breast cancers have been implicated in the development and progression of the disease. These include genes that code for:

- growth factors
- receptors
- intracellular signalling molecules
- cell cycle regulators
- apoptosis regulators.

Treatment is available for breast cancer and survival rates have been increasing. Despite these advances, it can still be a debilitating disease and successful treatment is not guaranteed. The prognosis has not been increasing at the same rate for all types of breast cancer. Some inherited forms of breast cancer develop earlier in life and are more aggressive and in general are more difficult to treat. These aggressive types of breast cancer are more serious and screening for them will prevent many women living much of their life fearing that they may develop breast cancer.

References:

1. *BRCA-1* free birth
http://scienceblog.cancerresearchuk.org/2009/01/30/brca1-free-birth-isnt-a-slippery-slope-to-designer-babies/

2. Breast cancer genes
http://www.cancerhelp.org.uk/type/breast-cancer/about/risks/breast-cancer-genes

3. Genetic epidemiological projects using population based case collections at Strangeways
http://www.oncology.cam.ac.uk/research/groupleaders/pharoah.html

All web references correct at time of production.

Other references should also be researched.

Case Study 2

RHESUS INCOMPATIBILITY

Moira has Rhesus negative blood and is expecting her second child in approximately six months. She has read conflicting information about the subject and would like to find out more about any problems that might arise from Rhesus incompatibility disease. Here is a transcript of a question and answer session that Moira had with 'ASK A DOCTOR ONLINE'.

Moira	I'm Rhesus negative and expecting my second child in six months. I'm very unclear on exactly how much of a potential problem there is with Rhesus incompatibility.
ASK A DOCTOR ONLINE	Rhesus incompatibility problems can only arise when a woman's blood is Rhesus negative and the baby's blood is Rhesus positive. This in turn can only happen if the baby's father's blood is also Rhesus positive.
Moira	My first baby was Rhesus positive and there were no problems. Why is that?
ASK A DOCTOR ONLINE	The problem arises if the woman's blood has previously come into contact with Rhesus positive blood, which usually only occurs if she has previously given birth to a Rhesus positive baby.
Moira	Does the blood mix during birth or can it mix during pregnancy?
ASK A DOCTOR ONLINE	The baby's blood does not normally mix with the mother's blood during the pregnancy, unless there has been a procedure, such as amniocentesis, or vaginal bleeding.
Moira	If there has been mixing of the baby's and the mother's blood, how does this put the pregnancy at risk?
ASK A DOCTOR ONLINE	The mother's body will recognise the baby's red blood cells as foreign and as a result will produce antibodies against the baby's red blood cells. This is called an immune response. In a subsequent pregnancy, these antibodies may cross the placenta and destroy the baby's red blood cells. This can lead to problems of anaemia and oedema while the baby is still in the womb and to severe jaundice of the baby after birth. This can be prevented by anti-D injections.
Moira	So, how do anti-D injections prevent this from happening?
ASK A DOCTOR ONLINE	Problems are prevented by giving Rhesus negative mothers an injection of anti-D immediately after the delivery of the baby. This destroys any Rhesus positive cells from the baby that are present in the mother's blood stream. As a result, the mother does not produce an immune response. Antibodies do not develop and the subsequent pregnancy is free from the problem.

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Moira	Are these injections dangerous?
ASK A DOCTOR ONLINE	Anti-D is manufactured from the plasma of human blood, and as with all blood products, there is a small possibility of viruses being transmitted from donor to woman..... but this is extremely unlikely to happen today in the UK.

References:

1. NHS Direct 2008 Rhesus Disease
http://www.nhs.uk/Conditions/Rhesus-disease/Pages/Causes.aspx

2. Haemolytic disease of the Newborn
http://www.patient.co.uk/doctor/Haemolytic-Disease-of-the-Newborn.htm

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