

## Preimplantation genetic diagnosis – exam style question

### Introduction

This activity is similar to Unit 1 exam questions. It tests understanding of PGD and some of the ethical issues involved.

The question is adapted from the Science for Public Understanding SPU 1 exam in 2007

a		<ul style="list-style-type: none"> <li>all cells carry the <u>whole</u> genome/all genes/all chromosomes/all DNA</li> <li>genome/DNA copied when cells divide</li> </ul>	any 1 for 1 mark	1
b	i	<ul style="list-style-type: none"> <li>carrier</li> <li>1 normal and 1 CF</li> </ul>	for 1 mark each	2
	ii	2 CF genes	for 1 mark	1
c	i	<ul style="list-style-type: none"> <li>Amniocentesis</li> <li>removal of fluid from around baby</li> <li>CVS</li> <li>removal of cells from placenta</li> </ul>	any 1 for 1 mark	1
	ii	<ul style="list-style-type: none"> <li>PGD may be wrong/false negative</li> <li>need to repeat any test</li> <li><b>Not</b> 'further mutation may have happened'</li> </ul>	for 1 mark	1
d		antenatal <ul style="list-style-type: none"> <li>testing only after woman is actually pregnant</li> <li>advantage - natural conception, no need for hormone treatments unlike IVF</li> <li>disadvantage - abortion necessary if fetus found to carry disorder</li> </ul> PGD <ul style="list-style-type: none"> <li>testing on embryo in dish, before pregnancy</li> <li>advantage - can enjoy pregnancy with some confidence that child is unaffected /avoids risk of abortion</li> <li>disadvantage - low conception rate/ IVF unpleasant process</li> </ul>	any 4 for 1 mark each	4

(e) The marking scheme for this section includes an overall assessment for the quality of written communication. There are no discrete marks for the assessment of written communication but quality of written communication will be one of the criteria used to assign the answer to one of the three levels.

## Examples of the sort of information or ideas that might be used to support an argument

## No

- ethics of discarding potential humans/ only acceptable if risk of death in early life is 100%
- only eliminates small part of cancer risk
- new cancer treatments means less likely to die from cancer
- could lead to other tests for more minor problems/'designer babies'
- IVF low success rate
- 

## Yes

- for risk greater than.....%
- right to use technology to prevent suffering
- these cancers strike younger people
- anxiety of child or family throughout their life
- suffering of families where several members have died of inherited cancer
- may be cost effective for health service

In marking this question make sure that students are clear that gene carriers will only develop cancer as adults and that it is not certain that they will develop cancer at all. This makes the arguments somewhat different from sickle cell or cystic fibrosis.

Level	Descriptor	Mark range	Level guidance for this question
3	<b>Good</b> An answer will meet most of the criteria given in the level descriptor Claims supported by an appropriate range of evidence Good use of information or ideas about science going beyond those given in the question Argument well structured with minimal repetition or irrelevant points Accurate and clear expression of ideas with only minor errors of grammar, punctuation and spelling	5 - 6	usually 4 specific points going beyond general 'prevent suffering' type  may include a scenario or example clear reasons, not just criteria for decision  usually have both sides with a clear conclusion
2	<b>Modest</b> Claims partially supported by evidence Good use of information or ideas about science given in the question but limited beyond this The argument shows some attempt at structure The ideas are expressed with reasonable clarity but with a few errors of grammar, punctuation and spelling	3 - 4	usually 3 points some of which are reasons or an example, not just criteria  may make good points but have no conclusion
1	<b>Limited</b> Valid points but not clearly linked to an argument structure Limited use of new information or ideas about science Unstructured Errors in grammar, punctuation and spelling or lack of fluency	1 - 2	only 2 points which may be very general or criteria for decisions, not argued reasons
0	<b>Incorrect or no response</b>	0	

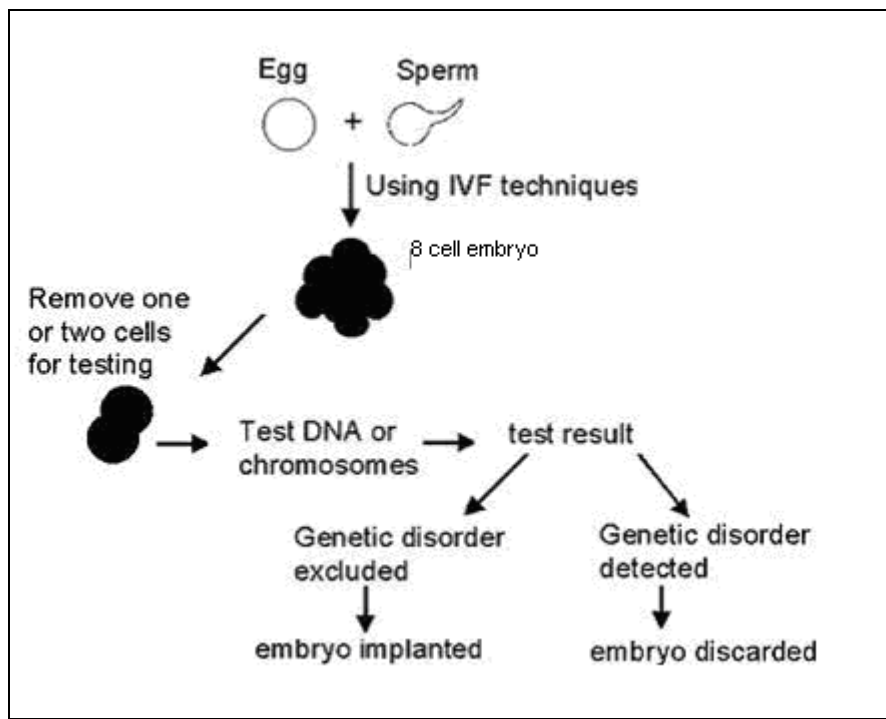
Total marks 16

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Figure 1 shows the process of Preimplantation Genetic Diagnosis, PGD, which can be used to prevent the birth of babies carrying serious genetic diseases such as cystic fibrosis or sickle cell anaemia.

**Figure 1 Preimplantation Genetic Diagnosis**



(a) (i) A child has about 10 000 billion cells. Explain how a single cell removed from the eight cell embryo can contain information relevant to the future health of the whole child.

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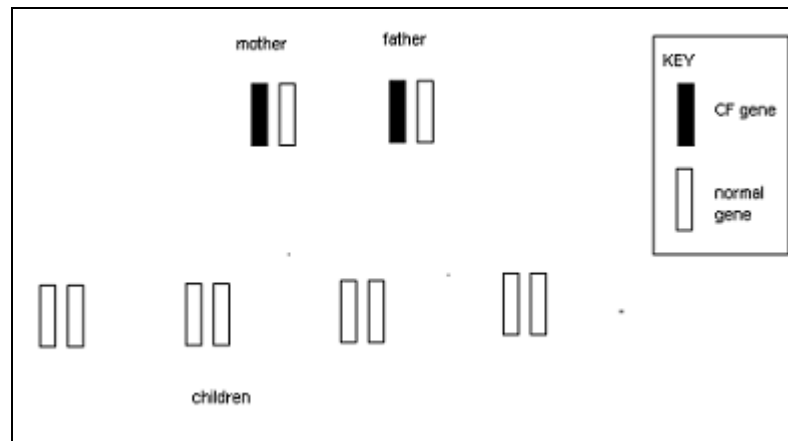
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(1 mark)

(b) Figure 2 represents the genes for cystic fibrosis in a healthy couple who are both carriers of the defective CF gene. A person with two defective CF genes will develop the disease.

**Figure 2**



(i) Complete the diagram, Figure 2, to show all the possible combinations of CF genes in their 4 children. (2 marks)

(ii) Identify on your diagram which of these children will develop cystic fibrosis. (1 mark)

(c) Doctors normally test the genes of the developing fetus again during the early part of the pregnancy.

(i) Name or describe a technique they would use to do this.  
 .....  
 ..... (1 mark)

(ii) Why would they need to do this further test?  
 .....  
 ..... (1 mark)

(d) Before PGD was available antenatal testing of cells from the fetus was the main way of preventing the birth of a child carrying a genetic disorder.

Explain the key differences between these two approaches in a way that would be relevant to a couple at risk of having a child with a genetic disorder.  
 .....  
 .....  
 .....

