Thursday 16 January 2020

Supervised hours: 3 hours | Paper Reference **21327L**

Applied Human Biology

Unit 3: Human Biology and Health Issues

You do not need any other materials. | Total Marks

Instructions

- Use **black** ink or ball-point pen.
- **Fill in the boxes** at the top of this page with your name, centre number and learner registration number.
- Answer **all** activities.
- Answer the activities in the spaces provided
  – *there may be more space than you need*.
- This booklet contains materials for the completion of the set task under supervised conditions.
- This booklet is specific to each series and this material must be issued only to learners who have been entered to undertake the set task in the relevant series.
- This booklet should be kept securely until the start of the 3 hour supervised assessment period.
- This set task should be undertaken in a session timetabled by Pearson.

Information

- The total mark for this paper is 60.
- The marks for **each** activity are shown in brackets
  – *use this as a guide as to how much time to spend on each activity*.

Advice

- Read each activity carefully before you start to answer it.
- Try to answer every activity.
- Check your answers if you have time at the end.

Turn over ▶

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Pearson

Instructions for teachers/tutors and/or invigilators

Centres must issue this booklet at the appropriate time and advise learners of the timetabled session.

All learner work must be completed independently and authenticated before being submitted to Pearson by the teacher/tutor or invigilator.

Learners are advised to spend 30–45 minutes reading through the stimulus material in the set task and annotate the material if they wish to do so.

Learners are required to use the stimulus material to complete the activities.

Learners must complete the set task using this task booklet.

Teachers/tutors and/or invigilators should note that:

- learners should not be given any direct guidance or prepared materials
- all work must be completed independently by the learner
- learners must not bring anything into the supervised environment or take anything out without your approval.

Centres are responsible for putting in place appropriate checks to ensure that only permitted material is introduced into the supervised environment.

Maintaining security

- During supervised assessment sessions, the assessment areas must only be accessible to the individual learners and to named members of staff.
- Learners can only access their work under supervision.
- Any work that learners produce under supervision must be kept securely.
- Only permitted materials for the set task can be brought into the supervised environment.
- During any permitted break, and at the end of the session, materials must be kept securely and no items removed from the supervised environment.
- Learners are not permitted to have access to the internet or other resources during the supervised assessment period.

After the session, the teacher/tutor or invigilator will confirm that all learner work has been completed independently as part of the authentication submitted to Pearson.

The set task is a formal external assessment and must be conducted with reference to the instructions in this task booklet and to the *BTEC Nationals Instructions for Conducting External Assessments (ICEA)* document.

Outcomes for submission

This task and answer booklet should be submitted to Pearson.

Instructions for learners

Read the set task information carefully.

You are advised to spend 30–45 minutes carefully reading the article in the set task.

You may add notes to the article before you start the activities.

Complete all your work in this booklet in the spaces provided.

This session is three hours. Your teacher/tutor or invigilator will tell you if there is a supervised break. Plan your time carefully.

In your response to the activities, you should consider information from the article and use your knowledge and understanding of applied human biology principles, procedures and techniques to support your answers.

You will complete this set task under supervision and your work will be kept securely during any breaks taken.

You must work independently throughout the supervised assessment period and you should not share your work with other learners.

You may ask your teacher/tutor or invigilator to explain any words or sentences you do not understand in the article or the activities. Your teacher/tutor or invigilator cannot help you complete the set task.

Outcome for submission

This task and answer booklet should be submitted to Pearson.

Set task information

You are given the following article:

Preimplantation Genetic Diagnosis: Avoiding genetic diseases, but at what cost?

You need to read the article so that you get an understanding of the health issue involved.

You should be able to interpret, analyse and evaluate the article.

At the end of the article there is a glossary giving the meanings of some of the words in bold used in the article.

Pre-implantation Genetic Diagnosis: Avoiding genetic diseases, but at what cost?

Pre-implantation genetic diagnosis (PGD) is a technique used to identify genetic defects in **embryos** created for *in vitro* fertilisation (IVF). PGD can be offered when one or both genetic parents have, or are carriers of, a known genetic abnormality[1]. PGD has been licensed by the Human Fertilisation and Embryology Authority (HFEA) to detect almost 400 genetic conditions[2], such as Huntington disease, Tay Sachs, etc.

During PGD, a single cell is removed from an embryo three days after fertilisation. Removing the cell does not have any long-term effects on the embryo, which will continue to grow normally. The removed cell is then tested to find out if the embryo contains normal chromosomes[3]. Only those embryos without the genetic markers for specific diseases are selected for IVF treatment. Figure 1 shows an outline of a single PGD cycle.

A cell biopsy is taken from the early stage embryo before implantation

DNA is extracted from the biopsy, amplified, and **genotyped**

Embryo is selected for implantation into the mother's uterus based on suitable genotype
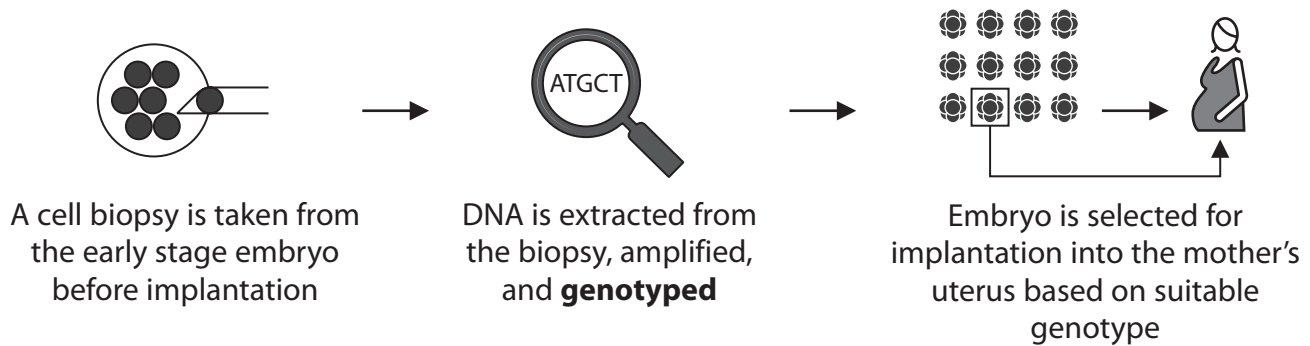
Figure 1

Why is PGD used?

PGD reduces the risk of producing a child affected by specific genetic diseases. While PGD has an extremely high success rate of identifying genetic diseases and chromosomal abnormalities, it is not yet 100% accurate. **Prenatal diagnosis** is still strongly advised following PGD and could identify an undetected issue with the **fetus**.

As a woman gets older, she is more likely to have a child with a chromosomal defect due to decreased egg quality[4], as shown in Figure 2. PGD can be used to make sure that only the embryos without chromosomal defects are implanted in the womb. This reduces the risk of genetic disease and improves the chance of a live birth following IVF treatment.

Figure 2

The Cost of PGD

PGD requires highly skilled technicians and precise medical equipment, meaning it is significantly more expensive than other prenatal dia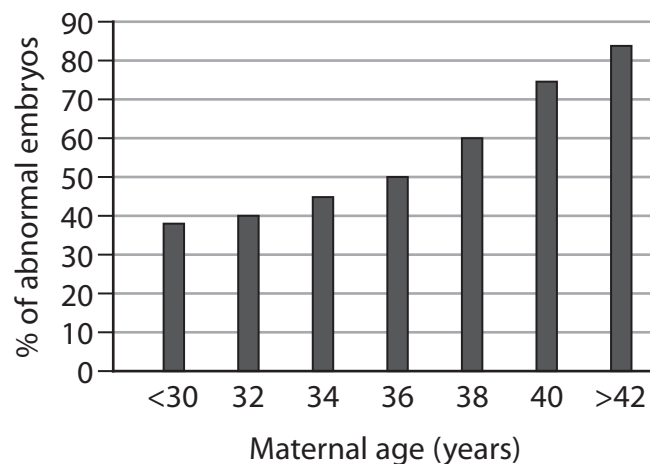gnosis options. It is a long and complex process, which can involve travelling to one of the few specialist centres that deliver the service.

PGD is available privately at a cost of about £8,000 per cycle and, in England, patients must be referred by a genetic counsellor[5]. PGD is also available through the NHS, but not if a couple already has a child who is affected by the genetic condition they want to check for. This is due to the concern that the PGD-tested baby might be created primarily as a donor to treat their sibling.

There are several conditions that must be met to get a referral. To be considered for a PGD cycle a couple must meet the following criteria[6]:

- be living together in a stable relationship
- the woman must be under 40 years of age at the start of treatment
- the woman's hormone levels must be within a range that suggests her ovaries will respond to treatment
- an accurate test for the genetic abnormality is available and there is a licence from the HFEA
- the PGD team agrees that the couple is suitable for treatment
- there are no concerns about the welfare of any child conceived using the treatment
- funding is available – either from the NHS or the couple if they choose to pay for their own treatment.

According to the NHS, in 2011 there were approximately 5000 couples eligible for PGD in the UK. Figure 3 shows the number of PGD cycles carried out each year and the number of cycles carried out per million people in the UK. This suggests that although the number of PGD cycles is increasing, the overall number of PGD referrals and cycles represents a very small part of the population[1].

Calendar year	Number of PGD cycles carried out	PGD cycles per million UK population
2004	95	1.6
2005	134	2.2
2006	184	3.0
2007	198	3.2
2008	214	3.5
2009	288	4.7
2010	373	6.0
2011	435	7.7

Figure 3

PGD Controversy

PGD has raised ethical and legal issues, opening the potential for the procedure to be used to select for the sex of the fetus, as well as genetic features such as intelligence and beauty. There is concern that PGD will be used to select against perceived negative traits such as certain disabilities, which may not prevent individuals from living a full and happy life. There are fears that this could lead to **'designer babies'** and a resurgence of the **eugenics** movement[7]. The problem with this belief is the concern about who decides which traits are desirable and what effects this would have on society and genetic diversity. The availability of PGD could raise the question to parents that, if they are able to select for disease, should they also be able to select for other features that will give their future child the best opportunity for a better life, such as beauty and intelligence, etc[8].

Another concern is what happens to the embryos that are not selected for IVF. At present they are either discarded or donated to science. Statistics collected by the HFEA, and published in the Daily Telegraph newspaper, stated that between August 1991 and December 2012 more than 3.5 million human embryos had been created through IVF procedures, including those involved in PGD. Of these, almost 1.4 million embryos were implanted in the hope of beginning pregnancies, with fewer than one in six resulting in a pregnancy. Nearly 1.7 million were discarded unused and a further 23,480 were discarded after being taken out of storage[9]. This could be seen as the destruction of human life and is actively opposed by many religious organisations[10].

Public opinion on PGD

In 2002, John Hopkins University, USA, surveyed 1,211 people. The survey showed that people were both optimistic and fearful about the rapidly advancing ability to manipulate human reproduction. Figure 4 shows the results of a survey[11] about the public's general preference for the medical applications of PGD rather than the ability to select for socially advantageous genetic features.
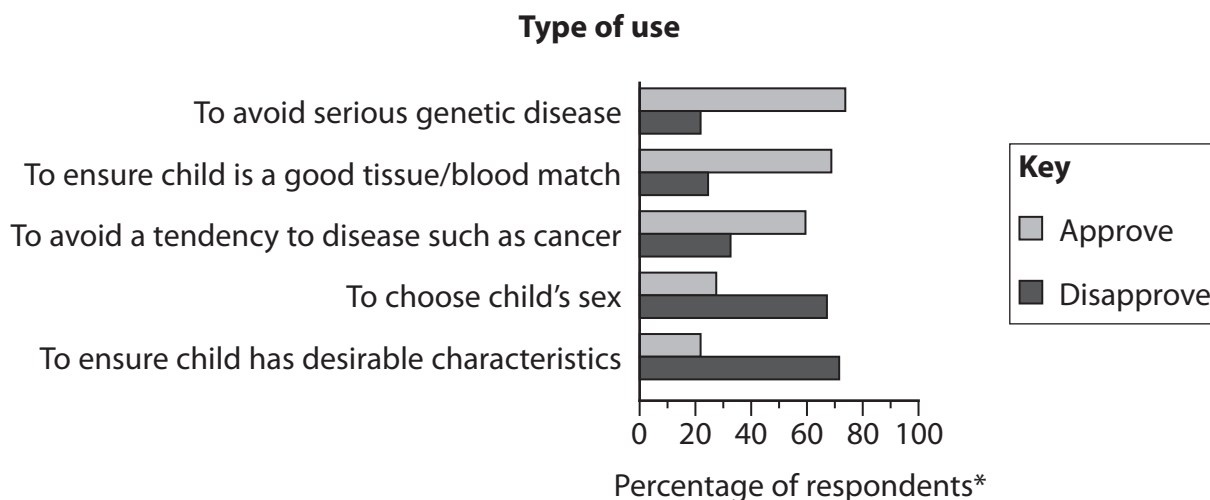
Type of use

Figure 4

*Figures may not add up to 100% as respondents may not fit into any category

Turn over ▶

Conclusion

PGD is a powerful tool that can be used to reduce genetic abnormalities and their linked health conditions. It can increase the chance of live births, be used to produce an offspring whose cells can be used to help a terminally ill sibling (not available on the NHS), and determine whether a potential child will be predisposed to certain diseases. It gives doctors tremendous power to combat genetic diseases and potentially eradicate them from the **gene pool**.

Care should be taken to consider the ethical implications of PGD. If we can select embryos that are free from disease then we can select for other traits, such as sex, hair colour, etc. Even the seemingly helpful use of PGD in selecting against disabilities, such as deafness or short-sightedness, can be extremely problematic. This could suggest to people that their conditions, a large part of who they are, are 'undesirable' and something to be removed from society. Such non-medical selection can have severe implications that could last for generations and completely alter the human race.

Glossary

Designer babies – the genetic code of an embryo is selected so that the children have traits the parents find desirable

Embryo – an unborn offspring in the first eight weeks of development

Eugenics – a widely-opposed philosophy that the human race can be 'improved' by selecting for 'desirable' traits

Fetus – an unborn offspring after eight weeks of development

Gene pool – the total genetic information of a population

Genotype – genes in DNA that are responsible for a particular characteristic

***In Vitro* Fertilisation (IVF)** – a medical procedure whereby an egg is fertilised by sperm in a Petri dish outside the body

Prenatal diagnosis – tests carried out for genetic disorders or other abnormalities, while the embryo is in the uterus

References

All references correct at time of writing.

1. NHS Commissioning Board Clinical Reference Group for Genetics, NHS [internet], London UK, April 2013; cited 11/2018. Available from https://www.england.nhs.uk/wp-content/uploads/2013/04/e01-p-a.pdf

2. HFEA.gov.uk, Human Fertilisation and Embryology Authority [internet], UK, updated 11/2018; cited 11/2018. Available from https://www.hfea.gov.uk/treatments/embryo-testing-and-treatments-for-disease/approved-pgd-and-ptt-conditions/

3. Dr John Zhang, drjohnzhang.com [internet], New York USA, December 2015; cited 11/2018. Available from https://drjohnzhang.com/pros-cons-pgd/

4. Fertilite, Hospital Angeles Tijuana [internet], Tijuana Mexico, 2018 ; cited 11/2018. Available from http://pgd-pgs.fertilitecenter.com/

5. Genetic Disorders UK, UK Genetic Disorders Partnership Network [internet], Leatherhead UK, 2017; cited 11/2018. Available from http://www.geneticdisordersuk.org/aboutgeneticdisorders/genetictesting

6. NHS Guy's and St Thomas', NHS Foundation Trust [internet], London, UK 2019; Available from https://www.guysandstthomas.nhs.uk/our-services/pgd/treatment/starting-pgd.aspx#na

7. Robertson JA, Human Reproduction. 18 (3): 465–71, March 2003, "Extending preimplantation genetic diagnosis: the ethical debate. Ethical issues in new uses of preimplantation genetic diagnosis"

8. Hens K, Dondorp W, Handyside AH, Harper J, Newson AJ, Pennings G, Rehmann-Sutter C, de Wert G (2013). Human Reproduction Update. 19 (4): 366–75 "Dynamics and ethics of comprehensive preimplantation genetic testing: a review of the challenges".

9. Andrew Hough, Telegraph Media Group Ltd. [internet], London, updated 12/2012; cited 11/2018. Available from https://www.telegraph.co.uk/news/health/news/9772233/1.7-million-human-embryos-created-for-IVF-thrown-away.html

10. Sozos J. Fasouliotis and Joseph G. Schenker. Human Reproduction vol.13 no.8 pp.2238–2245, Jerusalem 1998 Preimplantation genetic diagnosis principles and ethics

11. Brian Vastag, JAMA [internet], Chicago USA, updated 2/2004; cited 11/2018. Available from https://jamanetwork.com/journals/jama/article-abstract/198258

Set Task

Answer all activities in the spaces provided.

The activities in this task are based on the article.

Use the article and your knowledge and understanding of applied human biology principles, procedures and techniques to support your answers.

1 Discuss how the article uses scientific information to present the pre-implantation genetic diagnosis (PGD) issue.

In your answer, you should consider:

- how the article has interpreted and analysed scientific information to support the conclusions/judgements being made
- validity and reliability.

(12)

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(Total for Activity 1 = 12 marks)

2 Discuss the key factors affecting the pre-implantation genetic diagnosis (PGD) issue.

(16)

(Total for Activity 2 = 16 marks)

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DO NOT WRITE IN THIS AREA

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3 Explain how different organisations/individuals influence the pre-implantation genetic diagnosis (PGD) issue.

In your answer, you should consider:

- research
- health initiatives.

(10)

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(Total for Activity 3 = 10 marks)

4 Suggest potential areas for further development and/or research of the pre-implantation genetic diagnosis (PGD) issue.

(6)

(Total for Activity 4 = 6 marks)

5 A recent headline in a national newspaper said:

PGD: order your Einstein baby from the genetic menu

You have been asked to write a letter to the editor of this national newspaper to raise awareness of the benefits and limitations of pre-implantation genetic diagnosis (PGD).

When writing your letter, you must consider:

- who is likely to read your letter
- what you would like the reader to learn from your letter.

(16)

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(Total for Activity 5 = 16 marks)

TOTAL FOR PAPER = 60 MARKS

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