Cambridge TECHNICALS

Accredited

# OCR LEVEL 3 CAMBRIDGE TECHNICAL

CERTIFICATE/DIPLOMA IN

# HEALTH AND SOCIAL CARE

## HUMAN INHERITANCE FOR HEALTH AND SOCIAL CARE

Y/600/8991

LEVEL 3 UNIT 23

GUIDED LEARNING HOURS: 60

UNIT CREDIT VALUE: 10

OCR

# HUMAN INHERITANCE FOR HEALTH AND SOCIAL CARE

## Y/600/8991

## LEVEL 3 UNIT 23

## AIM OF THE UNIT

We are the product of our genes. They control our looks, our physiology and even our personality. Cystic fibrosis is a terminal condition caused by possessing a pair of rogue genes. About one in twenty five people in the United Kingdom carry a single copy of this gene. Each week, five babies are born with Cystic Fibrosis. Each week, two young lives are lost to Cystic Fibrosis. It is Europe's most common genetic condition. This unit will enable learners to discover how individuals acquire this condition amongst others. In order to do this they will study both the production of sex cells prior to the reproductive process and the structure and function of genetic material. The learners will investigate the events of fertilisation and how science can intervene to facilitate this process. They will also discover how the human race can, if it wants, alter and select the genetic traits of individuals to remove and control certain conditions including cystic fibrosis. Learners will also investigate the regulatory bodies and legislation that is in place to prevent the inappropriate manipulation of human tissue and genes.

## PURPOSE OF THE UNIT

Working in health and social care will often involve dealing with individuals whose lives are affected by genetic conditions and diseases ranging from mild disorders to terminal conditions that often cut short people's lifespan. Relatives and friends of people with health issues will often require clarification and information as well as support. This unit will enable learners to gain a basic understanding of the complexities of human genetics and inheritance. They will start by gaining an understanding of how eggs and sperm are formed and what precisely occurs as the genetic information in the form of chromosomes are shuffled around during these processes. They will investigate a range of factors that can affect the formation of eggs and sperm and the possible resulting event of fertilisation.

Learners will study the nature of genetic material and how it determines both the anatomy and physiology of individuals and even possibly our personality and behaviour. By exploring a range of genetic conditions learners will discover how what appear to be minor mistakes can have catastrophic results. They will study patterns of inheritance as well as how mistakes in the formation of sex cells can result in life changing disorders such as Down's syndrome.

Modern science is rapidly discovering ways of detecting conditions and manipulating human genes and synthesising artificial ones. Modern medicine has removed barriers to fertilisation and the first 'test tube ' baby is now in her thirties and women in their sixties are being enabled to conceive. Learners will be given an opportunity to understand some of these techniques and to question the morality of such advances. They will investigate the barriers and measures societies impose to try and prevent scientific advances from getting out of hand and from individuals deciding who should live and who should die so creating ideal or 'perfect' people. This unit will provide a core understanding and knowledge of human genetics that will allow an understanding of other specialised physiology units. Standing alone it allows learners to develop a rudimentary comprehension of how genes work and why everyone is so individual.

## ASSESSMENT AND GRADING CRITERIA

| **Learning Outcome (LO)**<br>The learner will: | **Pass**<br>The assessment criteria are the pass requirements for this unit.<br>The learner can: | **Merit**<br>To achieve a merit the evidence must show that, in addition to the pass criteria, the learner is able to: | **Distinction**<br>To achieve a distinction the evidence must show that, in addition to the pass and merit criteria, the learner is able to: |
|---|---|---|---|
| 1 Understand human reproduction | P1 Explain the process of meiosis in relation to the production of sperm and ova | | D1 analyse the significance of meiosis |
| | P2 Explain factors that may affect the reproductive process in humans | M1 assess the possible impact of lifestyle choices on the reproductive process in humans | |
| 2 Understand patterns of inheritance | P3 Explain the structure of chromosomes and their role in reproduction | | |
| | P4 Explain key principles of genetics using examples relating to inheritance in humans | M2 assess the impact on individuals of genotypes that impair normal body functioning | D2 summarise factors that may increase the likelihood of atypical genetic effects occurring in a population |
| 3 Know about reproductive and gene technologies | P5 Outline principles of reproductive and gene technologies and state their implications for individuals, society and the environment | | |
| 4 Understand ethical dilemmas in relation to reproductive and gene technologies. | P6 Explain ethical dilemmas that may arise from reproductive and gene technologies | M3 explain the need for regulatory bodies in relation to reproductive and gene technologies | |

# TEACHING CONTENT

## 1 Understand human reproduction

- *Reproduction:* anatomy of male and female reproductive systems
- *Gametogenesis:* spermatogenesis in testes, oogenesis in ovaries; significance of meiosis – reduction division, independent assortment of chromosomes, phases of meiosis; chromosome number – diploid, haploid
- *Conception:* events of fertilisation, chromosomal behaviour during fertilisation, mitosis
- *Influences:* role of reproductive hormones; other biological factors, (e.g. parental genotype, age, diet, health; environmental, for example, temperature, pollution (for example, oestrogens in the food chain, PCBs); chemicals (for example, nutrients, alcohol); radiation, for example from X-rays, environmental disasters)
- *Chromosomes:* structure, DNA, histones; autosomes, sex chromosomes; behaviour during meiosis, chromatids, chiasmata.

## 2 Understand patterns of inheritance

- *Genetic code:* codons, genes, homologous chromosomes, alleles, genome, messenger RNA and transfer RNA, protein synthesis
- *Variation:* discontinuous, continuous
- *Discontinuous variation:* Mendelian inheritance, genotype, phenotype; homozygous, heterozygous; dominant and recessive alleles; autosomal recessive inheritance, for example, cystic fibrosis, phenylketonuria; autosomal dominant inheritance, for example, Huntington's disease; co-dominance, for example, inheritance of blood groups, inheritance of sickle cell disorders; genetic determination of sex; sex-linked inheritance, for example, haemophilia, colour blindness, Duchene's muscular dystrophy; genetic susceptibility ,for example, breast cancer, heart disease
- *Continuous variation:* polygenic inheritance for example, height
- *Biological basis of personality:* Eysenck, Cattell
- *Mutations:* genes, for example, sickle cell anaemia/ trait; chromosomes, for example, Down's syndrome, Kleinfelter's and Turner's syndromes; mutagenic substances or events.
- *Gene pools;* out breeding; inbreeding.

## 3 Know about reproductive and gene technologies

- *Reproductive technologies:* screening techniques; assisted reproduction – in vitro fertilisation, artificial insemination
- *Gene technologies:* genetic engineering, for example, recombinant DNA, Human Genome Project, gene therapy
- *Implications:* genetic screening, diagnosis, , genetic counselling; tissue matching for transplantation, for example.

## 4 Understand ethical dilemmas in relation to reproductive and gene technologies

- *Regulation:* in home country, European Union, United Nations; legislation, adjudicators, adapting to emergent technologies, changing society
- *Individuals:* rights, responsibilities, confidentiality, quality of life, ownership, advocacy; others, for example, scientists, parents/family members, carers, professionals, pressure groups, commerce; rights, responsibilities, beliefs
- *Society:* values, benefits versus risks, support systems, resources, individual versus wider perspective, eugenics.
- *Wellbeing:* developmental delay, disability, susceptibility to ill health, behavioural difficulty.

# DELIVERY GUIDANCE

**LO1 Understand human reproduction**

Learners need to develop an understanding of the production of sperm and eggs and the role of meiosis This unit does not specifically require access to science laboratories. Prepared or self-made microscope slides can be used to investigate cell structure within the gonads and the stages of meiosis. Alternatively, images from slide packs, DVDs, books, websites could be used to illustrate these. Provided images could be identified and annotated by the learners. Models of cell division and meiosis can be made easily using a variety of media –either individually or on a larger scale as a group activity.

There could be teacher led research on gametogenesis and meiosis with group discussion/ Blockbuster quiz/ class mind maps to reinforce knowledge and understanding. Whilst it is realised that learners may have little to no previous knowledge of the scientific principles addressed in this unit it would be useful if they understand human anatomy, particularly the reproductive organs, and the physiology of reproduction.

There could be group or individual research followed by presentations to the whole group on a range of factors that can affect human fertility. These should cover those that affect gametogenesis and those that may make either fertilisation or implantation less likely. An appreciation of lifestyle factors or choices on human fertility needs to be developed if higher grades are to be attained. This could be covered in a group discussion following the suggested presentations.

Learners require a basic knowledge of the structure of chromosomes and why their number varies during gametogenesis. This could be followed by a delivery of the concepts of chromosome numbers and the role of meiosis in reducing diploid to haploid. The relationship of chromosomes to DNA needs to be understood and a simple overview of protein synthesis understood. The learners need to appreciate the link between DNA, RNA and the order of amino acids in proteins. This in turn requires an understanding of the importance of proteins within the body. Models can easily be constructed to show how the order of bases in DNA can be read at the ribosomes into sequences of amino acids.

**LO2 Understand patterns of inheritance**

Once the importance and significance of the genetic code has been established various examples of genetic conditions and patterns can be interpreted. It is recommended that cystic fibrosis is used as the prime example to show cause and effect of the condition and how it is inherited. Research can be made using case studies on those with the condition and how they are affected across PIES.

Further examples can then be taken and explored to cover the various types of inheritance outlined in the teaching content and the principles involved. Only one example needs to be used to illustrate each of these and this could be chosen either by the learner or the tutor. The concepts of genetic and chromosomal mutations can be covered at the same time and possible causative agents or events. Posters, reports, PowerPoint presentations could all be made. The work on protein synthesis should allow learners to understand the significance of even slight changes in the genetic code. Again cystic fibrosis could be used to illustrate this.

Learners need to appreciate that not all genes act according to predictable patterns, nor that all recessive traits are necessarily harmful or disadvantageous. Polygenic inheritance such as height can be used to discuss the former and effects such as tongue rolling or Gilbert's syndrome can be used to illustrate the latter. Case studies of families with histories of such conditions as breast or bowel cancer and heart disease can be used to explain genetic susceptibility.

A brief exploration of either Eysenck's or Cattell's personality or trait theories would allow learners to grasp the far reaching consequences of gene action in moulding an individual. They would need to only understand the biological basis of the traits and not specific details of the psychology involved.

A group discussion could be used to explain the term gene pool when applied to a population and the advantages of out breeding over the disadvantages of inbreeding emphasising the consequences of the latter. Examples drawn from existing isolated populations can be used. Learners need to be sensitively guided to the difference between inbreeding within a population and incestuous activities by individuals.

**LO3 Know about reproductive and gene technologies**

Learners should be introduced to both reproductive and genetic engineering principles and an overview of some of the techniques involved could be given. Only one type of IVF treatment needs to be covered. Genetic engineering could be illustrated through reference to treatment for diabetes and cystic fibrosis. The dangers of using viral vectors need to be established. This could be done through research, use of programmes or possibly by guest speakers if any are available locally, possibly from a local university. Case studies of those who have undergone fertility treatment, or visiting speakers who have had IVF, would be beneficial. The learners would need to be suitably prepared with questions. How genetic technologies are applied and their range of uses should be included and can provide a useful introduction to a discussion on the implications of these technologies.

**LO4 Understand ethical dilemmas in relation to reproductive and gene technologies**

Teacher input and discussion can be used to highlight ethical issues and high profile cases of where children of specific gender have been produced to help disadvantaged siblings. Discussion could also be used to consider frivolous reasons for such technologies as well as the more sinister intentions of shaping populations and the principle of eugenics. There needs to be either group research or teacher input on some examples of ethical guidelines, legislation and regulatory bodies involved in maintaining appropriate use as well as those that protect the rights of individuals with genetic conditions. Learners might like to consider the merits of using diagnostic tests in establishing genotype in conditions such as Huntington's disease. Discussion could include formal debates as a means of raising awareness of different perspectives on the issues involved. Videos and documentaries can be useful to provide a focus for exploring the issues raised by reproductive and gene technologies. Consideration needs to be given to the potential challenge to learners' values and beliefs which may be raised by such debates.

## SUGGESTED ASSESSMENT SCENARIOS AND TASK PLUS GUIDANCE ON ASSESSING THE SUGGESTED TASKS

Assessment is likely to be learning outcome based and written in nature. Display work or posters may be used as evidence, as can photographic evidence. It is advisable to have all evidence to hand when an external visit by a moderator is planned.

P1 – This could be met by a written report or as a poster or slide presentation showing the events of gametogenesis and how and when meiosis occurs during the formation of both eggs and sperm.

D1 – Learners would need to analyse the significance of meiosis as both a means of a reduction division and as a method of introducing genetic variation through independent assortment and crossing over. This is likely to be evidenced through a written report although a slide presentation or posters could be used as long as depth of knowledge and understanding was shown.

P2 – A second assignment could meet the grading criteria for P2 and M1 and could be approached as for assignment one. Learners will need to explain a range of factors that may affect both fertility and genetic material as covered in the teaching content. For M1 the learners need to assess the degree to which lifestyle choices affect reproductive processes. This should include dietary aspects, smoking, alcohol, recreational drugs and sexual promiscuity leading to STIs. This is likely to be as a written report although it could be covered as a guidance leaflet designed for those hoping to get pregnant.

P3 – Assignment three requires an explanation of the structure of chromosomes which could be in the form of a model or annotated poster. The role of chromosomes in reproduction can be cross referenced partially to assignment one relating to meiosis and then related to what happens to the gametes' chromosomes at the point of fertilisation. The subsequent division of the zygote by mitosis needs to be included. This is likely to be in the form of a written report supported by diagrams. Learners could design a series of annotated diagrams or cardboard models to illustrate the process of protein synthesis with a small sequence of DNA reproduced and a corresponding section of mRNA together with a ribosome and appropriate RNA and amino acids. Some written explanation of the process would be required.

P4 – Assignment four is likely to be an illustrated written report although it could include a slide presentation. To meet the pass criteria the learner needs to provide evidence of a number of genetic conditions that illustrate the principles, patterns and forms of inheritance identified in the teaching contents section. For this level of assessment it is only the mechanism of inheritance that needs to be explained and not the nature of the condition/dysfunction. This aspect can be explored if attempting M2. For this, a variety of conditions need to be described and their impact on the quality of life or everyday life of individuals assessed. This could be related to PIES. A condition for each of the mechanisms identified for the pass criterion is not necessary as long as a range of distinct types is covered. For example the effects of cystic fibrosis, Huntington's, Duchene's muscular dystrophy, Down's and susceptibility to breast cancer would be more than sufficient. To meet the requirements of D2 learners would need to investigate how factors such as isolation and inbreeding as opposed to out breeding are likely to affect the frequency of genetic disorders within a population. Case studies of environmental or cultural isolation could be cited but this must be done sensitively and without prejudice. Types and effects of a number of mutagens could also be discussed. These could include references to damage to DNA as a result of over exposure to UV light, heavy metals for example, mercury, X –rays or radioactive disasters such as Hiroshima, Chernobyl and Fukushima.

P5, P6 – A fifth assignment could be multi-part. Initially learners are required only to outline the principles of reproductive and gene technologies and so this could be done simply through annotated diagrams on posters. Alternatively two leaflets could be produced covering either of the two areas. There needs to be evidence of the implications of these technologies to both individuals and society. This could be an individual record of a group discussion on the subject. There needs to be a brief reference made to environmental concerns when genetic manipulation is carried beyond the human genome and into plant and animal food sources. This should cover LO3 but a further written report on the ethical dilemmas resulting from the use of these technologies should then cover LO4. Potential conflicts between individual choices and rights, benefits versus risk, and the impact on societies need to be explained. If this is then expanded to explain how these technologies

are to be policed or regulated and why, learners should be able to meet the requirements of M3. This is likely again to be a written report or a write up of a group discussion. Assessors will need to carefully record this evidence ensuring that each record is sufficiently individualised.

## SUGGESTED ASSIGNMENTS

The table below shows suggested assignments that cover the pass, merit and distinction criteria in the assessment and grading grid. This is for guidance and it is recommended that centres either write their own assignments or adapt any Cambridge Assessment assignments to meet local needs and resources.

| Criteria | Assignment title | Scenario | Assessment |
|---|---|---|---|
| P1, D1 | An explanation of the purpose of meiosis during the formation of gametes. | The learner has to provide non science specialists with an explanation of the process of meiosis and its significance. | An illustrated written leaflet or poster on meiosis. |
| P2, M1 | An investigation of factors that may contribute to infertility. | Advice to be given to a couple considering starting a family and who are concerned about levels of infertility and their ability to conceive. | Leaflets to offer advice on strategies on increasing the likelihood of conception. |
| P3 | An explanation of how genetic information controls human form and function. | An exhibition for non science specialists on the process of protein synthesis. | Posters or models accompanied by appropriate explanations. |
| P4, M2, D2 | Human genetic conditions –possible causes, effects and inheritance. | The use of information leaflets and/or posters to explain to a non –specialist audience/ reader a range of genetic conditions, their possible effects on individuals and factors that may increase their frequency. | A written report/leaflets/ posters together with illustrations/images. |
| P5, P6, M3 | An exploration of reproductive and genetic technologies. | A couple have been trying to conceive for six years. The man has a history of Huntington's disease in his family. | An information pack that explains what technological options are available to them and what regulations govern reproductive research and technology together with some ethical considerations regarding such a conception. |

# RESOURCES

## Text books


Clancy J and McVicar A – *Physiology and Anatomy: A Homeostatic Approach* (Hodder Arnold, 2002) ISBN 9780340762394

Gilbert P – *A-Z of Syndromes and Inherited Disorders* (Nelson Thornes, 2000) ISBN 9780748745296

Jenkins M – *Human Physiology and Health* (Hodder and Stoughton, 2000) ISBN 9780340658529

Mader S – *Understanding Human Anatomy and Physiology* (McGraw, 2004) ISBN 9780071111607

Minett P, Wayne D, Rubenstein D – *Human Form and Function* (Hyman, 1989) ISBN 9780713527148

Seamons S *Applied Health & Social Care: A2 Student Book OCR* (Folens, 2007) ISBN: 1850082480

Stretch B and Whitehouse M – *BTEC Level 3 Nationals in Health and Social Care Student Book 1* (Pearson, 2010) ISBN 9781846907663

Stretch B A2 GCE *Health and Social Care Student Book for OCR* (Heinemann,2006) ISBN 97804353529

Toole A and S – *Understanding Biology for Advanced Level* (Nelson Thornes Ltd, 2000) ISBN 9780748739646

Tortora G – *Principles of Anatomy and Physiology* (John Wiley and Sons, 2005) ISBN 9780471718710


## Journals

*'Inside the human Body'* Bright Star Publishing
*Biological Science Review*
*National Geographic Magazine*

## Websites

www.bbc.co.uk/health BBC health

www.humangeneticsdisorders.com

www.ornl.gov/sci/techresources/Human

www.innerbody.com/htm/body.html Human anatomy online

www.nhsdirect.nhs.uk NHS Direct patient information leaflets

www.womens-health.health-cares.net/female-fertilitytests.

www.nhs.uk/conditions

## DVDs

Inside the Human Body [DVD] (DVD - 2011)
The Human Body [DVD] [1998]

## MAPPING WITHIN THE QUALIFICATION TO THE OTHER UNITS

**Unit 4:** Development through the life stages

**Unit 5:** Anatomy and physiology for health and social care

**Unit 7:** Psychological perspectives for health and social care

## CONTACT US

Staff at the OCR Customer Contact Centre are available to take your call between 8am and 5.30pm, Monday to Friday.

We're always happy to answer questions and give advice.

Telephone 02476 851509
Email cambridgetechnicals@ocr.org.uk
**www.ocr.org.uk**