2016 Suite

OCR
Oxford Cambridge and RSA

Cambridge **TECHNICALS LEVEL 3**

APPLIED SCIENCE

Cambridge TECHNICALS 2016

Unit 5

Genetics

H/507/6152

Guided learning hours: 60

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LEVEL 3

UNIT 5: Genetics

H/507/6152

Guided learning hours: 60

Essential resources required for this unit: Access to leading research such as the Human Genome Project.

This unit is internally assessed and externally moderated by OCR.

UNIT AIM

Genetics is the study of inheritance. We now know that in all organisms, genes control the characteristics that are passed on from generation to generation. Advances in DNA technologies have demonstrated the potential of this work in several areas of science, including our understanding of disease and screening for inherited conditions, epidemiology and disease control, forensic science and historical and archaeological investigations.

This unit looks at how characteristics are inherited using the long-established techniques of crossing plants and animals, and in humans, by looking at the inheritance of certain characteristics and clinical conditions.

By studying this unit, you will be able to apply techniques used in genetics crosses using mathematical techniques to determine probability of inheritance by producing genetic chromosome maps using data from crosses. You will understand how geneticists are now able to produce detailed chromosome maps using the science of genomics.

TEACHING CONTENT

The teaching content in every unit states what has to be taught to ensure that learners are able to access the highest grades.

Anything which follows an i.e. details what must be taught as part of that area of content. Anything which follows an e.g. is illustrative, it should be noted that where e.g. is used, learners must know and be able to apply relevant examples in their work, although these do not need to be the same ones specified in the unit content.

For internally assessed units you need to ensure that any assignments you create, or any modifications you make to an assignment, do not expect the learner to do more than they have been taught, but must enable them to access the full range of grades as described in the grading criteria.

Learning outcomes The Learner will:	Teaching content Learners must be taught:
1. Understand the importance of meiosis	1.1 Meiosis as a reduction division i.e.: • Stages of meiosis • The importance of meiosis (halving the chromosome number; genetic variation) in sexual reproduction
2. Be able to apply techniques used in genetics crosses	2.1 Monohybrid inheritance i.e.: • Use of Punnett square • Genotypes and phenotypes of a monohybrid cross (a normal trait, single gene disorder, codominance, incomplete inheritance; sex-linked trait) 2.2 Dihybrid inheritance i.e.: • For a dihybrid cross: o inheritance of two characteristics o predict genotypic and phenotypic ratios (for two non-linked autosomal genes) • Expected and observed data in a cross o chi-squared test (χ^2-squared test) o statistical significance of differences in data and probabilities • Gene linkage and epistasis • Genetic maps o simple genetic maps from recombinant data. o limitations of genetic maps of chromosomes
3. Understand the techniques of DNA mapping and genomics	3.1 Deoxyribonucleic acid (DNA)/whole genome sequencing i.e.: • Principles of DNA sequencing (DNA polymerase and Polymerase Chain Reaction (PCR); techniques using single strand analysis) • The Sanger sequencing process (chain-termination or dideoxy method)

Learning outcomes The Learner will:	Teaching content Learners must be taught:
	• Next-generation DNA sequencing (NGS, or high-throughput sequencing). Evaluating advantages: o sample size o speed o accuracy o cost • Principles of genetic profiling (PCR of short tandem repeats [STRs]). o statistical probabilities (Second Generation Multiplex Plus SGM*Plus*® and DNA-17) o limitations of genetic profiling techniques
4. Understand the impact of an innovation in an application of genomics	4.1 Assess the impact and implications of a DNA-sequencing project (e.g. The Human Genome Project, NHS 100 000 Genomics Project, ENCODE Project) • Impact on increasing understanding, i.e.: o understanding of genome o location of genes linked to medical conditions (e.g. cancers); non-medical conditions and complex human traits o understanding of functional elements of DNA determined by non-protein-encoding regions o ecological relationships o genetic variation; evolutionary development • Implications i.e.: o medical treatments o design, development and prediction of effects of drugs and biotechnological solutions to treat conditions; preventive, personalised and pre-emptive medicines o ecological applications o conservation applications • Ethical, Legal and Social Implications (ELSI)

GRADING CRITERIA

LO	Pass	Merit	Distinction
	The assessment criteria are the Pass requirements for this unit.	To achieve a Merit the evidence must show that, in addition to the Pass criteria, the candidate is able to:	To achieve a Distinction the evidence must show that, in addition to the pass and merit criteria, the candidate is able to:
1. Understand the importance of meiosis	*P1: Describe the stages of meiosis	M1: Explain the importance of meiosis	
2. Be able to apply techniques used in genetics crosses	*P2: Demonstrate the genotypes and phenotypes produced in monohybrid genetic crosses	M2: Demonstrate that statistics can be used to compare expected and observed data in a cross	
	*P3: Demonstrate the genotypes and phenotypes produced in dihybrid genetic crosses involving two non-linked autosomal genes		
	*P4: Use phenotypic ratios to identify gene linkage and epistasis	M3: Construct a simple genetic map of a chromosome using data on recombinants from dihybrid crosses	D1: Discuss the limitations of genetic maps of chromosomes
3. Understand the techniques of DNA mapping and genomics	*P5: Describe the principles of DNA sequencing	M4: Evaluate the advantages of next-generation DNA sequencing (NGS)	
	P6: Describe the principles of genetic profiling	M5: Discuss the statistical significance of genetic profiling matches	D2: Evaluate the significance and limitations of genetic profiling techniques
4. Understand the impact of an innovation in an application of genomics	*P7: Describe an application of genomics	M6: Evaluate the impact of the specified genomic application	D3: Assess the implications and future impact of the specified genomic technique

ASSESSMENT GUIDANCE

LO1 Understand the importance of meiosis

P1: Describe the stages of meiosis

Learners should understand that a reduction division needs to occur in producing gametes. Learners should describe the stages of meiosis, but the use of the old nomenclature for the events of prophase I is not recommended.

M1: Explain the importance of meiosis

Learners should appreciate its importance in halving the chromosome number in gamete formation and leading to genetic variation in sexual reproduction. The principles of random distribution of chromosomes and therefore independent assortment of genes in gamete formation should be addressed, along with crossing over of homologous chromosomes during prophase I of meiosis as sources of genetic variation.

These explanations will require the use of some extended prose to provide sufficient detail.

LO2 Be able to apply techniques used in genetics crosses

P2: Demonstrate the genotypes and phenotypes produced in monohybrid genetic crosses

Learners should appreciate the theory of Mendelian genetics to demonstrate the genotypes and phenotypes produced for a single trait, i.e. in monohybrid crosses. The Law of Segregation should be defined, that paired genes separate in such a way that each gamete is equally likely to contain either member of the pair. The crosses demonstrated must include at least one showing incomplete dominance, e.g. the snapdragon flower, *Antirrhinum,* and codominance, e.g. blood groups; roan coat colour in cattle, and the inheritance of a single gene disorder or a sex-linked trait, e.g. haemophilia, Duchenne and Becker muscular dystrophy.

Learners do not always consider the relationship between genotypes and phenotypes at a molecular or biochemical levels. This can be reinforced here, eg:

- a tall and dwarf pea plant is determined by an enzyme that is involved in the synthesis of a plant growth regulator (gibberellin oxidase and gibberellin respectively)
- round or wrinkled seeds in peas are related to an enzyme responsible for linking glucose residues in a branched fashion
- in humans, the gene involved in cystic fibrosis (CFTR) encodes a cell membrane protein that functions as a channel for the transport of chloride.

P3: Demonstrate the genotypes and phenotypes produced in a dihybrid genetic cross involving two non-linked autosomal genes

Learners should appreciate Mendel's Second Law, of Independent Assortment, i.e. that allele pairs separate independently during the formation of gametes, so two traits are transmitted to offspring independently of one another. Examples should involve two non-linked autosomal genes. A characteristic F_2 generation 9:3:3:1 ratio should be discussed, and then a cross using a different example must be demonstrated by the learner, and examples of 1:1:1:1 ratios in test crosses.

The learner could carry out some genetics crosses if time permits. Use *Drosophila melanogaster* or rapid cycling brassicas, *Brassica rapa*. In the latter, yellow-green and rosette mutants will give a 9:3:3:1 ratio in the F_2 generation.

M2: Demonstrate that statistics can be used to compare expected and observed data in a cross

Learners will be introduced to the chi-squared test (χ^2-squared test) to compare expected and observed progeny in a cross. It is important that they show understanding of statistical significance of differences in data, and probabilities of (apparent) differences having occurred by chance.

Calculations from secondary or primary data collected should be accompanied by the appropriate discussion.

P4: Use phenotypic ratios to identify gene linkage and epistasis

Learners should understand that historical data sometimes shows that the F_2 generations do not give the 9:3:3:1 ratio expected; some phenotypes occurred more frequently than Mendelian genetics would predict. This should lead to a discussion of genes influencing each other, or physically coupled or linked in some way on the chromosome.

Learners should also identify another type of gene interaction called epistasis, where two or more genes at different loci interact. This interaction could produce a new phenotype, cause one allele to mask the effects of another/others, or one allele to modify the effects of another/others. In a typical dihybrid cross, each gene locus has an independent effect on a single phenotype. This is best illustrated using examples of the principle. Typically, learners should identify that if epistasis is involved, crossing two dihybrids produce a modified Mendelian ratio, e.g. instead of a 9:3:3:3:1 ratio, a 9:7 ratio, 15:1 ratio, etc.

M3: Construct a simple genetic map of a chromosome using data on recombinants from dihybrid crosses

Initially, again, these principles could be considered by looking at historical evidence. To fulfil this criterion, learners should be conversant with DNA recombination – the exchange of genetic material that occurs between homologous chromosomes during meiosis. This leads to the production of recombinant gametes in addition to the parental gametes expected. Recombination can occur between any two genes on a chromosome.

The amount of crossing over will be related to how close the genes are to each other on the chromosome. Genes that are closer together undergo fewer crossing over events, so fewer recombinant gametes are produced, while for genes next to each other on a chromosome, crossover events will be rare.

By looking at observed recombinant offspring in cross, it is possible to estimate the genetic distance on a chromosome between the genes in question. Learners should be taken though this procedure, and the numerical components involved (the percent of recombinants is the distance in centimorgans [cM]), then fulfil the criterion using real or hypothetical recombination data from crosses to illustrate the principles of producing a simple genetic map of a chromosome.

D1: Discuss the limitations of genetic maps of chromosomes

Chromosome mapping by counting recombinant phenotypes produces a **genetic map** of the chromosome. The limitations of the technique should be discussed with learners, e.g. as the distance between two loci increases, the probability of a second crossover occurring between them also increases, perhaps nullifying the effect of the first and restoring the parental combination of alleles.

Scientists cannot, of course, perform controlled matings of humans to map our chromosomes (though it is possible to estimate map positions by examining linkage in several generations of relatives).

Learners will also be aware that with advances in DNA sequencing and having produced genomes for many species, it is now possible to produce **physical maps** of the genome.

LO3: Understand the techniques of DNA mapping and genomics

P5: Describe the principles of DNA sequencing

The techniques involved in DNA sequencing could be introduced through a timeline of events, with the significant stages being the Sanger-sequencing method of 1977, the development of the polymerase chain reaction (PCR) in 1983, enabling scientists to rapidly amplify DNA, the first fully automated system developed by Applied Biosystems in 1986, and a series of next-generation sequencing techniques (NGS, or high-throughput sequencing).

Learners could describe these principles in a display or a webpage.

M4: Evaluate the advantages of next-generation DNA sequencing (NGS)

Learners should research current techniques and evaluate their advantages over other techniques. Their evaluations could be added to the display or webpage produced for P7.

P6: Describe the principles of genetic profiling
Learners often confuse DNA sequencing with genetic profiling; the latter focuses on the analysis of short repeating units of DNA (short tandem repeats, STRs) at specific loci, rather than the whole genome. The nature and variability of STRs should be discussed.

In the process, DNA is extracted, the STRs amplified using PCR, labelled with fluorescent dyes using locus-specific primers, and then subjected to electrophoresis. The resulting plot of the electrophoresis is an electropherogram, and learners should be given the opportunity of interpreting examples of these.

The old system used in the UK, SGM*Plus*® used ten loci plus a gender identifier, but DNA 17 was introduced in the UK in July 2014, using 16 STR loci plus the gender identifier (cf. the CODIS loci, using 13, used in the USA).

Learners could describe these techniques in an information leaflet, e.g. for legal use, which could be supplemented when working on M7 and D2.

M5: Discuss the statistical significance of genetic profiling matches
Learners should discuss the probability of a match occurring at the 10/16 loci. Because of the number of loci used, the product rule for probabilities can be applied. This has resulted in the ability to generate match probabilities of 1 in a quintillion ($1x10^{18}$) or more. In reality, forensic scientists still tend to quote a match probability estimated in the order of 1 in a billion (i.e. one thousand million). When partial profiles are obtained from crime scene samples, with between 19 and 32 matching loci when compared with a full subject profile, the probability is still estimated in the order of 1 in a billion. These probabilities should also be discussed in terms of the population of the UK, Europe and the global population.

Learners could describe these techniques in an information leaflet, e.g. for legal use.

D2: Evaluate the significance and limitations of genetic profiling techniques
With an analysis based on 17 loci, learners should appreciate that DNA 17 offers improved discrimination between profiles, greatly reducing the probability of getting a chance match between any two unrelated individuals' DNA profiles, and improved sensitivity - making it possible to produce DNA profiles using standard methodologies from less DNA, or poor quality DNA samples, e.g. samples of DNA that have become degraded or are mixed with chemical inhibitors such as the dyes used on certain clothing.

This system also offers improved comparability between profiles on the national DNA profiles produced in other European Union countries and beyond.

Learners should consider that with DNA-17 profiling, there are now a number of companies offering different version of profiling methodology, and discuss compatibility.

Using the PCR process, there can sometimes be errors in amplification. The greater the amplification used, the more likely it is that errors will occur. Learners may have heard of 'Low copy number' (LCN) testing, a method used in the United Kingdom when little DNA (typically <100 pg or <17 diploid genomes) is available. The method uses an increased number of PCR cycles (for example, 34, rather than the usual 28), providing scope for error, and more scope for contaminant DNA. Contamination is always a possibility in any DNA collection and analysis.

Where the condition of recovered material is very poor, DNA may have degraded such that the DNA variations at loci are no longer preserved intact, or the amount of DNA present in the sample is extremely low, resulting in a partial DNA profile.

DNA profiles relating to crimes in England and Wales are held on the National DNA Database (NDNAD), managed by the National DNA Database Delivery Unit (NDU) at the Home Office.

Retention of DNA profiles has always been controversial, but has resulted in criminal prosecutions in many cold cases. Learners should discuss the current governmental policy when writing their reports, and might also consider the implications of other initiatives that have been suggested over the years, e.g. profiling of everyone at birth. Miscarriages of justice as a result of flawed DNA analyses are rare, but should be discussed.

Learners should also consider the benefits and drawbacks of using mitochondrial DNA, which is better preserved in old samples and is particularly useful in historical or archaeological cases.

Learners could describe these techniques in an information leaflet, e.g. for legal use.

LO4 Understand the impact of an innovation in an application of genomics

P7: Describe an application of genomics

Learners should assess the impact and implications of a DNA-sequencing project, e.g. the Human Genome Project, The 100,000 Genomes Project, ENCODE Project or other genome-sequencing application.

The case study generated as a product of this LO could be in the form of a report or website.

M6: Evaluate the impact of the specified genomic application

This could be an application with medical implications, or one involved with systematics and evolutionary development, or conservation.

Examples of implications include an understanding of:

- the genome, along with those of other organisms
- genetic variation (e.g. HapMap)
- evolutionary development
- location of genes linked to medical conditions, including cancer (through mutations and oncogenes) [The Cancer Genome Atlas]; The 100,000 Genomes Project
- location of genes linked to non-medical conditions and complex human traits, e.g. intelligence, cardiometabolic and musculoskeletal phenotypes; design, development and prediction of effects of drugs and biotechnological solutions to treat conditions; preventive, personalised and pre-emptive medicines
- functional elements of DNA determined by non-protein-encoding region
- ecological applications.

Along with Ethical, Legal and Social Implications (ELSI) of these initiatives.

D3: Assess the future impact of the specified genomic technique

The particular impacts here will depend on the application reported on, but could include aspects such as developing personalised medicines as our understanding of the genetic causes of various human conditions are determined, and possible manipulation of the human genome.

Feedback to learners: you can discuss work-in-progress towards summative assessment with learners to make sure it's being done in a planned and timely manner. It also provides an opportunity for you to check the authenticity of the work. You must intervene if you feel there's a health and safety risk.

Learners should use their own words when producing evidence of their knowledge and understanding. When learners use their own words it reduces the possibility of learners' work being identified as plagiarised. If a learner does use someone else's words and ideas in their work, they must acknowledge it, and this is done through referencing. Just quoting and referencing someone else's work will not show that the learner knows or understands it. It has to be clear in the work how the learner is using the material they have referenced to inform their thoughts, ideas or conclusions.

For more information about internal assessment, including feedback, authentication and plagiarism, see the centre handbook. Information about how to reference is in the OCR Guide to Referencing available on our website: http://www.ocr.org.uk/i-want-to/skills-guides/.

SYNOPTIC LEARNING AND ASSESSMENT

It will be possible for learners to make connections between other units over and above the unit containing the key tasks for synoptic assessment. Please see Section 6 of the Qualification Handbook for more details. We have indicated in the unit where these links are with an asterisk.

Name of other unit and related LO	This unit:
Unit 1 Science fundamentals LO3 Understand cell organisation and structures LO4 Understand the principles of carbon chemistry	LO1 Understand the importance of meiosis (P1) LO3. Understand the techniques of DNA mapping and genomics (P5, P6)
Unit 2 Laboratory techniques LO2 Understand the principles of carbon chemistry LO4 Be able to examine and record features of biological samples	LO2 Be able to apply techniques used in genetics crosses (P2, P3, P4)
Unit 3 Scientific analysis and reporting LO1 Be able to use mathematical techniques to analyse data LO2 Be able to use graphical techniques to analyse data LO5 Be able to draw justified conclusions from data LO7 Be able to record, report on and review scientific analyses	LO2 Be able to apply techniques used in genetics crosses (P2, P3, P4)
Unit 8 Cell biology LO3 Understand the cell cycle and the importance of mitosis LO4 Understand the process and significance of differentiation	LO1 Understand the importance of meiosis (P1)
Unit 18 Microbiology LO3 Understand the use of microorganisms in agriculture LO4 Understand the action of antimicrobials on microorganisms	LO4 Understand the impact of an innovation in an application of genomics (P7)
Unit 19 Crop production and soil science LO2 Understand factors affecting the growth of crops	LO4 Understand the impact of an innovation in an application of genomics (P7)

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