## Molecular Diagnostics 631
### Unit Outline

### Area
BioMedical Sciences

### Credit points:
25 credit points

### Study Mode:
Internal

### Study Package:
310599

### Unit Index No:
310168 (v.1)

### Contact Hours
2 hours weekly (2 h lecture, 1 h tutorial)

### Unit significance
Core

### Result Type
Grade/Mark

### Ancillary charges
None

### Web based unit materials
Blackboard

### Pre-requisite Units:
Entry to Master of Biomedical Science course (310599), or as agreed by the Unit Coordinator dependent on appropriate undergraduate qualifications

### Syllabus
The nature of molecular testing for disease and the available technology. Specificity and sensitivity of diagnostic testing and relative cost-benefit comparisons. Molecular diagnosis of infectious agents; examples from bacteria, viruses or parasites. Mutation detection and the diagnosis of inherited disease. Oncogenomics and pharmacogenomics.

### Unit Coordinators
Dr Cyril Mamotte and Dr Brian Brestovac

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### Assessment

<table>
<thead>
<tr>
<th>Assessment</th>
<th>Mark Allocation</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>Quizzes</td>
<td>30%</td>
<td>See program</td>
</tr>
<tr>
<td>Assignment</td>
<td>20%</td>
<td>Week of 12th May</td>
</tr>
<tr>
<td>2 h written examination</td>
<td>50%</td>
<td>University examination period</td>
</tr>
<tr>
<td>TOTAL</td>
<td>100%</td>
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</tbody>
</table>
Introduction
Welcome to Molecular Diagnostics 631. Molecular diagnostic is that area of laboratory science which specializes in the analysis of various body fluids, cells or tissue using molecular genetic techniques. The techniques are applied in numerous different contexts including the diagnosis or confirmation of genetic disorders, disease management, prediction of disease, pathogen detection/quantification, tissue typing and identity testing (e.g. in forensic investigations) to name a few. The intended target for this unit are postgraduate students with experience or an interest in life, medical science or pharmaceutical sciences or in biotechnology. Please note that Molecular Diagnostics 631 is a Core unit in this course. This means that if you fail the unit twice, your course of study can be terminated.

Unit Learning Outcomes (ULO)

On successful completion of this unit, you will be able to

1. Critically appraise different approaches and methods for:
   1.1. Scanning and typing of genetic mutations and polymorphisms
   1.2. Pathogen detection and quantification
   1.3. Chromosomal abnormalities

2. Summarize key concepts, methodological principles and issues in the application of molecular diagnostics in numerous fields including diagnosis of genetic disorders, cancer, infectious diseases, cytogenetics, and transplantation.

3. Retrieve and analyze data for the determination of diagnostic sensitivity and specificity of molecular diagnostic tests.

4. Analyze and critically evaluate the molecular diagnostic literature and produce a report on your findings.

5. Compare molecular diagnostics to more traditional approaches and justify its importance to patient care.

Tuition Pattern

3 hours per week allocated as follows:
Lecture: 2x1 hour
Tutorial: 1 x 1 h

Learning opportunities
While the emphasis should be on self-directed learning, particularly at a post-graduate level, the lectures, tutorials, and quizzes will all help to achieve the desired outcomes.
<table>
<thead>
<tr>
<th>Week (Curtin No.)</th>
<th>Date (Week Starting..)</th>
<th>LECTURE TOPIC 2 h duration</th>
<th>Lecturer</th>
<th>Tutorial 1 h duration</th>
</tr>
</thead>
</table>
| 1                 | 1/3                    | Introduction to Molecular Diagnostics  
                      |                       | C Mamotte | Tutorial on 1st 2h Lecture |
|                   |                        | Scope of Molecular Diagnostics  
                      |                       |          |                      |
|                   |                        | Cystic Fibrosis as an example  
                      |                       |          |                      |
| 2                 |                        | Familial Hypercholesterolaemia  
                      |                       | C Mamotte | Tutorial on 2nd Lecture and review of quizz. |
|                   |                        | + Quiz on First 2h Lecture  
                      |                       |          |                      |
| 3                 |                        | Molecular Virology  
                      | Brian Brestovac |          | Molecular Virology Tutorial |
| 4                 |                        | HLA typing & Quiz on Mol Virology Lecture  
                      | Brian Brestovac |          | Tutorial on HLA Typing & review of quiz |
| 5                 |                        | Diagnostic sensitivity and specificity of genetic tests  
                      | C Mamotte |          | Tutorial on diagnostic sensitivity and specificity |
| 6                 |                        | Mutation detection/scanning technologies  
                      |          |          | Mutation detection/scanning technologies |
| 7                 |                        | Thalassaemia & Quiz on Mutation Detection Techniques  
                      | C Mamotte |          | Tutorial on Thalassemia and review of quizz |
| 8                 |                        | Molecular methods in forensic science  
                      | Gavin Turbett |          | Tutorial & Discussion of assignment questions |
| 9                 |                        | Pharmacogenetics  
                      | C          |          | Tutorial & Discussion of assignment questions |
| 10                |                        | Duchenne Muscular Dystrophy  
                      |          |          | Tutorial and Student discussions |
|                   |                        | Clinical and Technical Aspects  
                      |          |          | |
| 11                |                        | Fragile X syndrome  
                      |          |          | Tutorial and Student discussions |
|                   |                        | Clinical and Technical Aspects  
                      |          |          | |
| 12                |                        | Breast Cancer  
                      |          |          | Tutorial and Student discussions |
|                   |                        | The future of molecular diagnostics  
                      |          |          | Tutorial and Student discussions |
| 13                |                        | STUDY WEEK  
                      |          |          | |
| 14                |                        | EXAM PERIOD  
                      |          |          | |

* topics which are formally reviewed during the semester (each worth 10% of the final mark (3x10 =30%)).
Unit Materials

Blackboard
For on-line course materials, electronic submissions and some quizzes.

Note on Attendance of Lectures & Blackboard Content
Not all of the material covered will available via Blackboard. Furthermore, context and emphasis of important areas is important. Therefore attendance at all lectures is strongly recommended.

Electronic Journals.
Access the library through the Curtin homepage and follow the prompts for the electronic journals or use the following URL and follow the prompts. http://library.curtin.edu.au/htbin/erl_menu. Having gained access you can select abstracts or full text. In the abstracts section Current Contents is particularly useful for the most recent information and Science Direct, Swetsnet and Wiley Interscience are good for relevant full text information. Journals also can be accessed via the web using a search engine (eg Google) and accessing the home page of the journal of interest.

Textbooks and References (these are available on-line or through the library).

There are no set texts for the unit, but the following may be useful:


Assessment Details

There are several components which are assessed and students must pass all components to pass the unit.

Module quizzes
These will consist of a mix of short and long answer questions on the topics (the format may differ depending on the topic). Three of these will be formally assessed (each worth 10% of your final mark; in total, the module quizzes are worth 30% of your final mark).

Assignment
There will be one written assignment. It involves a considerable amount of work including research of the literature. Therefore, it is important that you develop a plan for completion of the assignments at the earliest opportunity. Organization, planning, scheduling and meeting deadlines is critical to success at the postgraduate level. If you experience any difficulties, please ask for help as soon as possible. The assignment is worth 20% of your final mark.

Final Exam
There will be a 2 hour test on the material covered in assignments, lectures and tutorials, in the examination period at the end of the semester.
Assignment topics. Differences in the molecular diagnosis of 2 of the following conditions (one from list a) and one from list b)

a) Duchenne Muscular Dystrophy, adult onset polycystic kidney disease
   • See Strachan & Read 488-504 for an introduction.

b) Huntington disease, Fragile X, MELAS,

In discussion of these conditions, you must demonstrate your understanding of the following:

a) What the disease is
b) Mode of inheritance for the condition/disease
c) Penetrance
d) Expressivity
e) Mutational spectrum (including the differing types of structural mutations that can cause disease)
f) At least three different techniques suitable for molecular diagnosis or confirmation of the disorder (and their relative advantages/disadvantages)
g) A case example from the literature
h) How mutations result in disease (pathophysiology)
i) Particular difficulties/issues in molecular testing.

The submitted assignment will comprise of:

a) A hard copy version comprised of:
   i) A signed assignment cover page.
   ii) Title page (E.g. Molecular Diagnostics 631 Assignment, Name/Student number)
   iii) Contents page
   iv) The main body of the assignment
   v) Reference section: Use appropriate referencing (Chicago, Vancouver, or Harvard style). Textbooks and journal articles that are indexed in PubMed are the only references that are permissible in the reference list. Use of Wikipedia or other similar sources will incur an automatic 10% deduction from your mark.

b) Submission of an electronic version of the above, without the cover page, as a MS Word or PDF file. A second file containing the abstracts (where relevant) to all references that have been used for the assignment. The electronic version will be subject to analysis using Turnitin software.

Marks will be allocated approximately as described below:

<table>
<thead>
<tr>
<th>Marking Area</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Introduction</td>
<td>15%</td>
</tr>
<tr>
<td>Ensure the scope of the assignment is clearly defined.</td>
<td></td>
</tr>
<tr>
<td>2. Referencing (including appropriateness)</td>
<td>10%</td>
</tr>
<tr>
<td>3. Presentation (including grammar, absence of typographical errors, correct</td>
<td>20%</td>
</tr>
<tr>
<td>use of abbreviations, organization, and keeping within the word limit</td>
<td></td>
</tr>
<tr>
<td>(2,500 words total, excluding reference section)).</td>
<td></td>
</tr>
<tr>
<td>4. Critical analysis (evaluation/interpretation/synthesis of information)</td>
<td>40%</td>
</tr>
<tr>
<td>5. Conclusion (rational, and in context of the preceding discussion and the</td>
<td>15%</td>
</tr>
<tr>
<td>Introduction)</td>
<td></td>
</tr>
</tbody>
</table>

Referencing Style
Chicago, Harvard or Vancouver. The use of EndNote is strongly encouraged.
Criteria for successful completion of the unit

Students should note that it is necessary to pass all components of the unit and that failure of any component may result in an overall failure in this unit regardless of the total marks accrued. That is, a pass in the practical component but failure in the theory component (or vice versa) may lead to a fail grade for the unit, even though the total mark for the unit exceeds 50%.

The components for which a mandatory pass mark is required include:

- Final Exam
- The Assignment

Plagiarism monitoring

Some or all of the assessments in this unit may be monitored for plagiarism using the Turnitin plagiarism detection service (http://turnitin.com). Students who do not want their assignments retained in the Turnitin database (e.g. because of intellectual property implications) must lodge a special request prior to the submission date. See www.academicintegrity.curtin.edu.au/studentsturnitin.html for additional information.

Avoid regurgitating or paraphrasing text or other materials by other authors, from any source (including your fellow students). If you use direct quotes, you must acknowledge that source by putting the direct quote in quotation marks and citing the authors name(s) and the relevant reference. All graphs, illustrations, tables or data from other sources must also be acknowledged and referenced. The reference is for these is usually cited below the graphic or table. While cooperation between students is encouraged, collusion is an offence.

Turnitin at Curtin University of Technology

This document briefly explains how Turnitin works and why it is currently being trialed at Curtin University.

What is Turnitin?

Turnitin (www.turnitin.com) is an electronic text matching system that compares text in a student assignment against electronic text on the Internet, in published works, on commercial databases, and in assignments previously submitted to Turnitin by students in universities all over the world, including assignments obtained from ‘paper mills’ (Internet sites which sell papers). The Turnitin system operates through a web site and is accessed using standard web browsers

Turnitin supports the implementation of the University’s mission and values (strategic.curtin.edu.au/vmv.html) and its policy on plagiarism (www.policies.curtin.edu.au/documents/plagiarism.doc). It is one of many resources that can assist in ensuring academic integrity is maintained

What if I do not want my work retained by Turnitin?

The effectiveness of Turnitin depends on its ability to store copies of all documents submitted to it. This has the benefit of protecting a student’s rights and intellectual property in the future. If for any reason you do not want your assignment to be retained on the Turnitin database after it has been checked, you are required to lodge the ‘non-retention’ form available at http://academicintegrity.curtin.edu.au/local/docs/nonretentionform.doc and submit it along with your assignment to your Lecturer/Tutor.

Supplementary Information

Late submission penalties

There are penalties for late submission of assessable components. Students will have 10% of the total assessment mark deducted for each day (Including weekends) the assessment is late without prior negotiation with the Unit Coordinator. Assessments will not be marked (nor feedback given) if work is submitted more than 10 days after the due date. See the Division of Health Sciences Web site for further details.

Legitimate grounds for extensions include:

- Health Issues (medical certificate required).
- Psychological grounds (medical certificate from a registered health professional or University Counselling).
- Equity considerations (as requested by a University Disability Counsellor)
- Compassionate grounds.