

# GENE90001 Human Genetics and Genetic Counselling 1

<b>Credit Points:</b>	12.5
<b>Level:</b>	9 (Graduate/Postgraduate)
<b>Dates &amp; Locations:</b>	2016, Parkville This subject commences in the following study period/s: Semester 1, Parkville - Taught on campus.
<b>Time Commitment:</b>	Contact Hours: 2 x 2 hour sessions per week Total Time Commitment: 170 hours
<b>Prerequisites:</b>	None
<b>Corequisites:</b>	None
<b>Recommended Background Knowledge:</b>	
<b>Non Allowed Subjects:</b>	None
<b>Core Participation Requirements:</b>	For the purposes of considering requests for Reasonable Adjustments under the Disability Standards for Education (Cwth 2005), and Students Experiencing Academic Disadvantage Policy, academic requirements for this subject are articulated in the Subject Overview, Objectives, Assessment and Generic Skills sections of this entry. It is University policy to take all reasonable steps to minimise the impact of disability upon academic study, and reasonable adjustments will be made to enhance a student's participation in the University's programs. Students who feel their disability may impact on meeting the requirements of this subject are encouraged to discuss this matter with a Faculty Student Adviser and Student Equity and Disability Support: <a href="http://services.unimelb.edu.au/disability/">http://services.unimelb.edu.au/disability/</a>
<b>Coordinator:</b>	Ms Lisette Curnow
<b>Contact:</b>	<b><a href="mailto:lissette.curnow@ghsv.org.au">lissette.curnow@ghsv.org.au</a> (mailto:lissette.curnow@ghsv.org.au)</b> Phone: 03 8341 6250 <b>Administrative Contact:</b> Vicki Hirt <b><a href="mailto:vhirt@unimelb.edu.au">vhirt@unimelb.edu.au</a> (mailto:vhirt@unimelb.edu.au)</b> Phone: 03 8341 6336
<b>Subject Overview:</b>	The curriculum is based on the genetic knowledge requirements of the Human Genetics Society of Australasia Board of Censors for Genetic Counselling, which governs the certification of genetic counsellors in Australia. Basic biological and genetic concepts will be taught via a combination of didactic and problem based learning (PBL) methods. Various human disorders will be discussed to illustrate basic concepts. PBL will be used to facilitate the sharing of information between students and to support the development of group work - which models the work of a genetic counsellor, who works as part of a multidisciplinary team. Tutors will have experience working in the areas of clinical genetics, laboratory genetics, and genetic education / research.
<b>Learning Outcomes:</b>	This subject is designed to enable students to acquire knowledge in the following areas as well as an appreciation of the application of this knowledge to the practice of clinical genetics: <ul style="list-style-type: none"> <li># Cell biology</li> <li># Meiosis, mitosis</li> <li># Principles of cytogenetics, including molecular approaches</li> <li># Human embryology and the effects of major human teratogens</li> <li># Gene structure and regulation</li> <li># Transcription, translation</li> <li># Protein structure and function</li> </ul>

	<ul style="list-style-type: none"> <li># Mutations and polymorphisms</li> <li># Developments in gene technology</li> <li># Principles of molecular genetic testing</li> <li># Patterns of inheritance, e.g. mendelian, mitochondrial, polygenic, multifactorial</li> <li># Pedigree drawing</li> <li># Pedigree analysis and risk calculation</li> <li># Biostatistics including population genetics and consanguinity, linkage analysis, Bayes theorem analysis, assumptions in risk assessments</li> <li># Principles of population screening. Examples to include newborn screening and 1st and 2nd trimester maternal serum screening.</li> </ul>
<b>Assessment:</b>	2.5 hour exam (60%) End of Semester approximately 1500 word written assignment (30%) Mid Semester class participation/in class assessments (10%) Throughout Semester Hurdle Requirement: Students must achieve a 'pass' on the 2.5 hour exam Students are expected to complete all in class assessments.
<b>Prescribed Texts:</b>	None
<b>Recommended Texts:</b>	<p>1 Gardner, R.J.M., Sutherland, G.R. &amp; Shafer, L.G. (2011). Chromosome Abnormalities and Genetic Counselling (4th edition), Oxford Monographs, New York: Oxford University Press.</p> <p>2 Harper, P.S. (2010). Practical Genetic Counselling (7th edition), Hodder Arnold</p> <p>3 Medical Genetix CD-ROM by Sylvia Metcalfe (University of Melbourne/Murdoch Childrens Research Institute)</p> <p><b>PLUS ONE OR MORE OF THE FOLLOWING:</b></p> <ul style="list-style-type: none"> <li># Turnpenny, P.D. &amp; Ellard, S. (2011). Emery's Elements of Medical Genetics (14th edition). Elsevier</li> <li># Strachan, T. &amp; Read, A.P. (2010). Human Molecular Genetics (4th edition), New York: John Wiley &amp; Sons.</li> <li># Korf, B.R. &amp; Irons, M.B. (2013). Human Genetics and Genomics (4th edition), Carlton: Blackwell Science.</li> <li># Trent, R.J. (2012). Molecular medicine: Fourth Edition: Genomics to Personalized Healthcare. Academic Press</li> <li># Sudbery, P. &amp; Sudbery, I. (2010). Human Molecular Genetics (3rd Edition) (Cell and Molecular Biology in Action). Benjamin Cummings</li> <li># Young, I.D. (2006). Introduction to Risk Calculation in Genetic Counselling (3rd edition), Oxford University Press.</li> <li># Jorde, L.B., Carey, J.C. &amp; Bamshad, M.J. (2009). Medical Genetics (4th edition), Mosby Press.</li> <li># Nussbaum, R.L., McInnes, R.R. &amp; Willard, H.F. (2007). Thompson and Thompson Genetics in Medicine (7th edition), WB Saunders (3rd edition), Elsevier Academic Press</li> </ul>
<b>Breadth Options:</b>	This subject is not available as a breadth subject.
<b>Fees Information:</b>	Subject EFTSL, Level, Discipline & Census Date, <a href="http://enrolment.unimelb.edu.au/fees">http://enrolment.unimelb.edu.au/fees</a>
<b>Generic Skills:</b>	<p>On completing this subject students should be able to:</p> <ul style="list-style-type: none"> <li># Understand the principles of inheritance</li> <li># Understand chromosomal disorders and the genetic basis of disease</li> <li># Understand the normal stages of human embryo development and have an awareness of how this can be disrupted</li> <li># Document a family tree</li> <li># Understand clinical genetic risk assessments for patients and families</li> <li># Convey genetic information and discuss risk in a clinical context</li> <li># Have an appreciation for and a basic understanding of the range of molecular and cytogenetic laboratory tests utilised in clinical genetic practice</li> <li># Understand the issues relating to population based screening</li> </ul>
<b>Related Course(s):</b>	Graduate Diploma in Genetic Counselling Master of Genetic Counselling