

# GENE90002 Clinical Genetics

<b>Credit Points:</b>	25
<b>Level:</b>	9 (Graduate/Postgraduate)
<b>Dates &amp; Locations:</b>	2010, Parkville This subject commences in the following study period/s: Semester 2, Parkville - Taught on campus.
<b>Time Commitment:</b>	Contact Hours: 12 weeks, 2x3 hours per week Total Time Commitment: Not available
<b>Prerequisites:</b>	Completion of Human Genetics.
<b>Corequisites:</b>	.
<b>Recommended Background Knowledge:</b>	.
<b>Non Allowed Subjects:</b>	.
<b>Core Participation Requirements:</b>	For the purposes of considering request 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 Description, Subject Objectives, Generic Skills and Assessment Requirements of this entry. The University is dedicated to provide support to those with special requirements. Further details on the disability support scheme can be found at the 3 Disability Liaison Unit website : 4 <a href="http://www.services.unimelb.edu.au/disability/">http://www.services.unimelb.edu.au/disability/</a>
<b>Coordinator:</b>	Ms Margaret Sahhar
<b>Contact:</b>	Ms Margaret Sahhar, <a href="mailto:margaret.sahhar@ghsv.org.au">margaret.sahhar@ghsv.org.au</a>
<b>Subject Overview:</b>	<p>The curriculum is based on the genetic knowledge requirements of the Human Genetics Society of Australasia Board of Census for Genetic Counselling, which governs the certification of genetic counsellors in Australia. Clinical and laboratory concepts relating to genetic disorders 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.</p> <p><b>Subject objectives:</b></p> <p>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:</p> <ul style="list-style-type: none"> <li># Dysmorphology and approaches to syndrome diagnosis</li> <li># Reproductive genetics, including prenatal screening and testing options</li> <li># Reproductive technologies, including ultrasounds and prenatal procedures such as amniocentesis, chorionic villus sampling, foetal biopsy and cord blood analysis, together with a consideration of the options available when a fetal abnormality is detected</li> <li># Inborn errors of metabolism and their clinical manifestations</li> <li># Common genetic conditions: clinical features, modes of inheritance and genetics, for example cystic fibrosis, muscular dystrophies, haemoglobinopathies</li> <li># Cancer genetics, including the role of genetics in the cause and/or predisposition of individuals to cancer</li> <li># Neurogenetics, including the role of genetics in the cause and/or predisposition of individuals to various neurological conditions such as Huntington disease, spinocerebellar ataxias and dementia</li> <li># Genetic causes of intellectual disability, including Fragile X syndrome</li> <li># Genetic conditions associated with various body organs/systems, for example skeletal, skin, eye, blood, immune, kidney, gastrointestinal tract, oral and craniofacial, endocrine and hormonal, respiratory, ear and cardiovascular system</li> <li># Treatment of genetic disease</li> </ul>

	<ul style="list-style-type: none"> <li># laboratory approaches to predictive, diagnostic and carrier genetic testing</li> <li># Australian health care system: organisation and economic aspects</li> <li># Working with other professionals and identifying community resources</li> <li># Ethical and legal systems in relation to the practice of clinical genetics, for example issues relating to privacy and confidentiality, professional guidelines and codes of conduct.</li> </ul>
<b>Objectives:</b>	<p>On completing this subject students should be able to:</p> <ul style="list-style-type: none"> <li># Elicit and document a family history</li> <li># Document a detailed family pedigree</li> <li># Understand more complex clinical genetic risk assessments for patients and families</li> <li># Convey more complex genetic information and discuss risk</li> <li># Have an appreciation for the range of molecular, cytogenetic and biochemical laboratory tests utilised in clinical genetic practice</li> <li># Understand the genetic testing approach taken for specific genetic disorders</li> <li># Understand the treatment approach taken for specific genetic disorders</li> <li># Understand the role of prenatal screening and testing in pregnancy management and care and the options available when fetal abnormality is detected</li> <li># Understand the role of genetics as the underlying cause of various disorders of the human body</li> <li># Understand the role of genetics in cancer</li> <li># Understand the organisational and economic aspects of health care in Australia</li> <li># Understand the role of the genetic counsellor in the context of the multidisciplinary approach to clinical genetic health care</li> <li># Understand the principles of the legal and professional duties and responsibilities of genetic counsellors as health professionals and members of a health care team</li> </ul>
<b>Assessment:</b>	End of semester 3 hr exam (50%), experiential and 2000 word written assignment (30%), one class presentation (10%), and class participation/in class assessments (10%). Students are expected to complete all in class assessments.
<b>Prescribed Texts:</b>	.
<b>Recommended Texts:</b>	<ul style="list-style-type: none"> <li># Thompson MW, McInnes RR, Willard HF (2001) Thompson and Thompson Genetics in Medicine (6th Edition), WB Saunders</li> <li># Jorde LB, Carey JC, White RL (1999) Medical Genetics (2nd Edition), Mosby Press</li> <li># Gelherter TD, Collins FS, Ginsburg D (1998) Principles of Medical Genetics (2nd Edition) USA: Williams and Wilkins</li> </ul> <p>Other references:</p> <ul style="list-style-type: none"> <li># Young ID (1999) Introduction to Risk Calculation in Genetic Counselling (2nd Edition), Oxford University Press</li> <li># Korf BR (2000) Human Genetics: A Problem-Based Approach (2nd Edition), Carlton: Blackwell Science</li> <li># Gardner RJM and Sutherland GR (2004) Chromosome Abnormalities and Genetic Counselling (3rd Edition), Oxford Monographs, New York: Oxford University Press</li> <li># Harper PS (2004) Practical Genetic Counselling (6th Edition), John Wright</li> <li># Trent RJ (1997) Molecular Medicine: An Introductory Text (2nd Edition), New York: Churchill Livingstone</li> <li># Strachan T and Read AP (2004) Human Molecular Genetics (3rd Edition), Thomson Publishing Services</li> </ul> <p>A subject reference list will be provided. Library available with selected texts within GHSV.</p>
<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># Elicit and document a family history</li> </ul>

	<ul style="list-style-type: none"><li># Document a detailed family pedigree</li><li># Understand more complex clinical genetic risk assessments for patients and families</li><li># Convey more complex genetic information and discuss risk</li><li># Have an appreciation for the range of molecular, cytogenetic and biochemical laboratory tests utilised in clinical genetic practice</li><li># Understand the genetic testing approach taken for specific genetic disorders</li><li># Understand the treatment approach taken for specific genetic disorders</li><li># Understand the role of prenatal screening and testing in pregnancy management and care and the options available when fetal abnormality is detected</li><li># Understand the role of genetics as the underlying cause of various disorders of the human body</li><li># Understand the role of genetics in cancer</li><li># Understand the organisational and economic aspects of health care in Australia</li><li># Understand the role of the genetic counsellor in the context of the multidisciplinary approach to clinical genetic health care</li><li># Understand the principles of the legal and professional duties and responsibilities of genetic counsellors as health professionals and members of a health care team</li></ul>
<b>Related Course(s):</b>	Master of Genetic Counselling