Genetic Counselling Programme
Division Human Genetics
University of Cape Town

2020/2021

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Introduction

As this is a professional programme, the courses in the first year provide the foundation of medical genetic knowledge, genetic counselling skills and research methodology. These courses will be applied in genetic counselling practice in first and second year. Medical Genetics, Principles of Genetic Counselling and Genetic Counselling Practices 1 are written off at the end of the first year.

Courses in the second year require application of the above knowledge and skills in the clinical situation by accurate assessment, planning and formulation of counselling sessions independently; and conducting research and writing a research dissertation, under supervision. Not everything will be covered in lectures over the 2 years and there is a significant amount of self-directed learning which will be required of you.

Purpose of the programme

The first purpose of the qualification is to provide students with a wide range of master-level knowledge, attitudes and skills unique to genetic counselling which will equip you for life-long learning and competent clinical practice. Following a complementary internship it will enable you to register with the Health Professions Council of South Africa (HPCSA) and to work independently in the public or private sectors in communities, hospitals, clinics, special schools and laboratories in southern Africa as well as internationally. There is an explicit commitment to a Primary Health Care approach of client and family management throughout the programme.

The second purpose of the qualification is to equip students with management, leadership and research skills which will enable you to participate in national health care development and policy making and contribute to the international body of knowledge in genetic counselling.

The third purpose of the qualification is to provide South Africa with a group of competent genetic counsellors to provide effective, efficient and accessible genetic counselling services while continuing to improve their knowledge and skills.

Aims

The general aims of the programme are that by the end of two years the student will be able to:

- Demonstrate a level of professional excellence;
- Demonstrate comprehensive knowledge of basic medical genetics and its relevance to genetic counselling;
- Manage time effectively;
- Problem-solve autonomously and co-operatively with other disciplines;
- Communicate effectively and courteously;
- Have a keen interest in research and continuing education; and
- Demonstrate competency in professional life-skills.

At the conclusion of the programme, the student should be able to demonstrate the objectives listed under each course.

Content of programme

The prescribed courses include:

PTY5003F	Principles of Genetic Counselling: Course work
PTY5004S	Principles of Genetic Counselling: Applied learning
PTY5005F	Medical Genetics 1
PTY5006S	Medical Genetics 2
PTY5009W	Genetic Counselling Practice 1
PTY5008W	Genetic Counselling Practice 2
PTY5001W	Genetic Counselling Minor Dissertation

Mark allocation:

Principles of Genetic Counselling (80 hours)

Module 1 Principles of Genetic Counselling: Course work	10 credits
Module 2 Principles of Genetic Counselling: Applied learning	10 credits

Medical Genetics (80 hours)

Module 1 Medical Genetics 1	12 credits
Module 2 Medical Genetics 2	12 credits

Genetic Counselling Practice (1720 hours)

Module 1 Genetic Counselling Practice 1	40 credits
Module 2 Genetic Counselling Practice 1	40 credits

Genetic Counselling Minor Dissertation (60 hours)

Module	Minor dissertation	60 credits

Progression and readmission

Students may be allowed to repeat a course they have failed, at the convener's discretion. No course may be repeated more than once. A student failing a course twice will be asked to withdraw from the programme. No supplementary examinations will be offered. A deferred examination may be granted where applicable, e.g. on medical grounds as per Faculty rules. Students must pass all of the courses in the first year before progressing to the second year. Faculty rules relating to attendance and leave of absence apply. All cases will be considered on an individual bases in consultation with the convener.

Any student whose performance is not satisfactory may be required to withdraw from the programme.

In addition, it is highly recommended that a research proposal must be approved by the Department of Pathology Research Committee (DRC) and the Faculty of Health Sciences Ethics Committee (HREC) prior to progression into the second year of the programme to enable timeous submission of the research report to allow for graduation.

Distinction requirements

The degree which is by coursework and dissertation may be awarded with distinction where a student:

(a) Obtains an average overall mark of 75%; and
(b) Obtains at least 70% for each individual component.

Duly Performed requirements

In order to qualify for the Genetic Counselling Practice examinations the student must:

a) attend 80% of all classroom activities;
   Attend all formal lectures, seminars, tuts, journal clubs, discussion groups and supervision sessions
   Spend a minimum of 20 hours a week in the office
b) attend 90% of clinical counselling sessions;
   Minimum of 10 clinics a month
   See a minimum of 8 patients a month with a least 2 under direct supervision
c) Clinical Admin
   All patient admin to be completed within 2 weeks
d) achieve 50% for Module 1 Principles of Genetic Counselling
e) achieve 50% for Module 2 Principles of Genetic Counselling: Applied learning
f) achieve 50% for Module 1 Medical Genetics
g) achieve 50% for Module 2 Medical Genetics
h) achieve 50% for Genetic Counselling Practice;
   - achieve 50% for the end-of-block performance evaluations;
   - achieve 50% for the continuous clinical case assessments;
   - achieve 50% for the case reports

When, due to illness or other situations, an extended period of absence is required, the student has to discuss this with the course convener. Each case will be assessed on an individual basis within the Division.

Confidentiality and ethical conduct

As with all staff and students in the Faculty of Health Sciences it is important to preserve and protect all confidential patient information and to conduct oneself in an ethical manner at all times. Confidentiality includes patients' as well as their family members' medical records (including computerised data and verbal information provided by or about them), diagnoses, test results, conversations, discussions,

research records and financial information. Ethical conduct refers to clinical as well as research matters.

Students will be required to sign the Division's Confidentiality Agreement and the Laboratory Safety Guidelines and Procedures at the beginning of the first year.

Registration with the Health Professions Counsel of South Africa (HPCSA)

Students, who are required to register with the HPCSA, should do so in January of the first year of study and register as a "Student". In March of the second year of study students are required to register as an "Intern Genetic Counsellor".

Internship

Any person who wishes to register with HPCSA as a Genetic Counsellor needs to have completed two years of internship in an accredited training facility within three years of graduating. The second year of the MSc Genetic Counselling programme is recognised as one year of internship. The second year of internship needs to be completed after graduation. The Division of Human Genetics does not guarantee that there will be funded/unfunded internship posts available to students when they have completed the MMedSc in Genetic Counselling degree.

Part-requirement for satisfactory completion of the internship is for the student to maintain a log of all patients/clients observed, counselled under supervision and counselled independently during clinical exposure over the three-year period. The following information should be recorded in an index system:

Patient's name, hospital, patient's file number, medical condition, and whether the counselling aspect involved observation, counselling under supervision or counselling independently. A brief description and comments of the case should also be documented (See logbook).

There is a HPCSA guideline document, The Standards of Practice Document, on how this portfolio should be completed and it is suggested that you consult this document early in your training so that you can align your work with these requirements. It is advisable that you log/record all your activities (including special seminar attendance, workshops etc.) as this will all be required in your final portfolio for assessment. This information is also required for your block assessments.

Medical Genetics

Medical Genetics is taught over a 12-month period as two semester courses:

PTY5005F Module 1 Medical Genetics 12 credits

PTY5006S Module 2 Medical Genetics 12 credits
Hours: 80 hours

Outline of the course

Medical Genetics is taught as two semester courses. It aims to inform genetic counsellors of the principles of Medical Genetics. Medical Genetics is the specialty of medicine involved with diagnosis, care and prevention of birth defects (may or may not be genetic in origin), and human diseases that are, at least, partially genetic in origin. It deals with hereditary, the mechanisms of hereditary transmission and the variation of inherited characteristics among individuals with the same disorders. Although the course is assessed independently it is also assessed in Genetic Counselling Practice by application of this knowledge in the clinical arena.

Objectives

The objectives of the course are to enable the student to:

- Describe the epidemiology, aetiology, phenotypes, medical management and special investigations of well-known genetic disorders;
- Apply the scientific and medical aspects specific to individual patients/families with genetic disorders;
- Interpret special investigations associated with a genetic condition;
- Assess the risk status of an individual and family (risk assessment);
- Apply theoretical knowledge scientifically;
- Source genetic data from appropriate internet databases and websites;
- Discuss current and developing technologies in all areas of genetics;

Topics

Medical Genetics 1

Chromosome, DNA, basic patterns of inheritance, prenatal diagnosis.

Embryology
Teratogens
Approach to dysmorphic examination
Chromosomes
Phenotypes and management of common chromosomal abnormalities
Testing for chromosome abnormalities
Pedigree drawing and family history
Patterns of inheritance
Common Mendelian conditions in SA (CF Albinism etc)
Genetic Testing
Screening principles
Prenatal screening for aneuploidy
Prenatal diagnosis

Risk calculation

Medical Genetics 2

Multifactorial inheritance, complex patterns of inheritance, clinical focus
Genomics, Science and Society: HGP and beyond
Epigenetics overview (UPD, X-inactivation, mitochondrial, imprinting)
X-inactivation, Fragile X and XLMR
Triplets repeat disorders
Mitochondrial disorders
Multifactorial disease - Neural Tube Defects
Inherited metabolic diseases
Deafness
Haemoglobinopathies
Inherited cardiomyopathies
Neuromuscular disorders
Connective tissue disorders
Neurodegenerative disorders
Cancer - Breast and Colorectal

Assessment

Assessment is by assignments and examination. The examination contributes 50% of the coursework marks, while the assignments and test accounts for 50%. A pass mark of 50% is required for the exam, with a 45% subminimum for the assignments. Total contribution of the course to the final mark at the end of the programme is 17%.

Reference materials

1. Firth Helen V and Hurst Jane A (2005) ***Oxford Desk Reference Clinical Genetics***. Oxford University Press Inc: London

This book is a good resource for quick assess in clinics. It is a good reference manual for clinical practice after graduation – Highly recommended.

2. Harper, PS (2010) ***Practical Genetic Counselling (7th ed)***. London : Hodder Arnold
3. Mendelow B, Ramsay M, Chetty N, Stevens W (2009) ***Molecular Medicine for clinicians.*** Wits University Press: Johannesburg.
4. Brookes/Cole- Korf, Bruce R (2000) ***Human Genetics - a problem-based approach (2nd ed).*** Blackwell Science: Malden, Mass.
5. Lewis, Ricki (1997-2010) ***Human Genetics: concepts and applications (2nd to 10th ed).*** Wm C Brown: Dubuque, IA.
6. Pritchard, D and Korf, B (2008) ***Medical Genetics at a glance (2nd ed).*** Blackwell Publishers: Oxford.
7. Thompson, James S (2007) ***Thompson and Thompson genetics in medicine (7th ed).*** Saunders/Elsevier: Philadelphia.

8. Turnpenny, P.D. and Ellard, S., (2011) ***Emery's elements of medical genetics.*** Elsevier Health Sciences.
9. Sandler, T.W (2015) ***Langman's medical embryology (13th Ed).*** Wolters Kluwer: Philedelphia

All books are available in the Health Sciences Library (some are available as e-resource). Some of them are available in the Division.

In addition to the prescribed and recommended texts there are several excellent web sites to which to refer:

Gene Reviews
National Organization for Rare Disorders (NORD)
The Rare Chromosome Disorder Support Group (Unique)
Clinical Key (UCT online resource)
UpToDate (UCT online resource)

Principles of Genetic Counselling

Principles of Genetic Counselling are taught over a 12-month period as two semester courses:

PTY5003F Module 1 Principles of Genetic Counselling: Course work 10 credits
PTY5004S Module 2 Principles of Genetic Counselling: Applied learning 10 credits
Hours: 80 hours

Outline of the course

Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. It provides individuals and their families with information about genetic conditions, diagnostic testing, and risks to other family members within a framework of nondirective counselling and ethical principles. Although the course is assessed independently it is also assessed in Genetic Counselling Practice by application of this knowledge in the clinical arena.

Objectives

The objectives of the course are to enable the student to:

- Describe the purpose of genetic counselling;
- Describe the structure of a genetic counselling session;
- Describe and critically evaluate the range of genetic counselling techniques used in practice;
- Describe the scope of genetic counselling;
- Risk communication;
- Pedigree drawing;
- Communication skills;

- Discuss ethical considerations in the field of Genetics;
- Discuss different settings in which Genetic Counsellors practice.

Assessment

Assessment is by assignments and an examination. The examination contributes 50% of the coursework marks, while the assignments accounts for 50%. A pass mark of 50% is required for the exam, with a 45% subminimum for the assignments. Total contribution of the course to the final mark at the end of the programme is 13%.

Topics

Principles of Genetic Counselling: Basic Concepts

Introduction to Genetic Counselling theories and processes
- Role and scope of Genetic Counselling
- Basic communication
- Structure of a session
- Family history taking as a psychosocial tool
- Information communication
- Non-directiveness, risk communication and decision making
- Family functioning
- Client styles, defenses, resistance
- Guilt and Shame
- Ethics
- Patient experience

Principles of Genetic Counselling: Applied learning

Advanced theories in Genetic Counselling
- Psychology theory/skills
- Family communication
- Anger
- Cultural competence
- Genetic testing in children
- Counselling children and adolescents
- Ethics
- The psychology of predictive testing
- Grief
- Breaking bad news
- Genetic counselling in the genomics era

Reference materials

1. Ulhmann, Wendy R; Scheutte Jane L and Yashor, Beverly M (2009) ***A guide to genetic counselling***. Wiley-Blackwell: New Jersey
2. Weil, Jon (2000) ***Psychosocial Genetic Counselling.*** Oxford University Press Inc: New York.

3. Evans, Christine (2006) ***Genetic counselling: a psychological approach.*** Cambridge university press: New York
4. Veach, Patricia McCarthy, LeRoy, Bonnie S and Bartels Dianne M (2003) ***Facilitating the Genetic Counseling Process: A Practice Manual.*** Springer-Verlag: New York.
5. LeRoy, Bonnie; McCarthy Veach, Patricia; Bartels, Dianne M (2010) ***Genetic counseling practice: advanced concepts and skills***. Wiley-Blackwell: New Jersey
6. Egan Gerard (2002) *The* ***Skilled Helper: a problem-management and opportunity--development approach to helping (7th ed).***
7. Harper, PS (2010) ***Practical Genetic Counselling (7th ed)***. London : Hodder Arnold

All recommended books are available in the Health Sciences Library. Some of them are available in the Division.

Genetic Counselling Practice

Genetic Counselling Practice is taught over a 24-month period as two modules:

PTY5009W Genetic counselling practices 1 40 credits
PTY5008W Genetic Counselling Practices 2 40 credits
Hours: 2072

Outline of the course

This course addresses the theory and practical application of genetic counselling to genetic conditions. Students spend a portion of each week in various clinics, counselling patients/clients and their families under supervision and participating in clinical management discussions. Counselling practice starts from the beginning of year one, initially simulated sessions and later in clinics. It involves attending clinics at Groote Schuur Hospital, Red Cross Hospital, schools and outreach visits.

Objectives

The objectives of the course are to enable the student to:

- Elicit a relevant family and medical history;
- Construct an accurate pedigree;
- Calculate risk status of the individual and family;
- Elicit a relevant social and psychosocial history of the individual and family;
- Writing case reports and letters
- Interpret special investigations associated with the genetic condition;
- Apply theoretical knowledge learned in Medical Genetics and Principles of Genetic Counselling scientifically;
- Plan and execute a range of interviewing and counselling techniques;

- Manage the clients, patients and families with empathy; caring; compassion; sensitivity; acceptance of diversity; respect for their dignity, privacy and confidentiality;
- Assessing and counselling clients effectively in a realistic time.
- Critically evaluate genetic counselling techniques used in practice;
- Identify personal limitations and the need to seek help when necessary;
- Identify the limitations of the scope of genetic counselling;
- Refer to other health practitioners appropriately and timeously;
- Identify relevant ethical and legal aspects associated with clients and their families;
- Refer to specific genetic support groups and Non-Government Organisations (NGOs) where relevant;
- Adapt to change and tolerate uncertainty;
- Apply knowledge of human rights as referred to in the constitution;
- Recognise the cultural, social, religious and historical context of the clients and families;
- Demonstrate responsibility to clients and families;
- Discuss the impact of economic imperatives of business, industry and government on genetic counselling;
- Discuss legislation guiding genetic counselling practice and to apply it in genetic counselling practice;
- Observe and interpret interaction between family members and counsellors; and
- Inform patients of local and international resources and services.

Students will attend all clinics over the two-year period of the programme. Rotations will involve an initial period of observation only before progressing to taking a medical history and drawing a pedigree, counselling under guidance and finally counselling independently. Goal 1, 2 and 3 provide guidance as to the focus of the experiential training. The most frequently presented medical conditions are available. Students should prepare for cases prior to attending the clinics in order to ensure that patients and their families receive optimum counselling, minimise the stress of the clinical environment and maximise the learning experience.

Each student will receive regular clinical facilitation/supervision. It is the student's responsibility to feedback to the clinical facilitator any problems experienced during and/or between facilitation sessions. It is recommended that once comfortable, students record their sessions (with the patients consent) to reflect on the session and identify areas of improvement.

Attendance at all clinics is compulsory.

In addition to clinic attendance, each student will be allowed the opportunity to go on "outreach", circumstances permitting. Students should also take turns observing and performing a predictive test for adult onset neurological conditions. Students are encouraged to do clinical rotations in other centers, both locally and internationally.

Referral to parent/patient support groups is an essential part of comprehensive patient care. Genetic counsellors therefore interact regularly with the support groups. Students are therefore required to join a support and participate in their activities for the duration of the course. A report (1000-1500 words) of the lessons learnt should be submitted for the final block assessment and needs to be included in the intern portfolio.

Case discussions

The aim of the case discussions and role play sessions is to direct your learning in terms of the medical and psychosocial aspects of the case and basic understanding and broad overview to enable you to understand the medical and/or anatomy and/or physiology and/or embryology of the topic.

Assessment

Assessment is by End-of-block performance evaluations, continuous clinical case assessments and case reports during the year. Students will have counselling examinations at the end of each semester. The exam at the end of the second year will be externally moderated. In addition, the student's performance during the clinical rotations will be assessed by means of an end-of-block performance evaluation. Observation guidelines, clinical case assessment and end-of-block evaluation forms are available. A formal 3 hour written exam will be written at the end of each year.

The examinations contribute 50% of the course marks. An average of 50% is required to pass the exam. The block evaluations and continuous counselling assessments account for the remaining 50%. The student will be required to obtain 50% for the end-of-block performance evaluations, clinical case tests and case reports in order to qualify for admission to the final examination. The student will be required to obtain an average of 50% for the exams in order to pass. Total contribution of course to final mark is 40%.

All students are required to log all the cases that they have observed or counselled and to make notes on each case to allow for reflection and learning. Cases will be discussed in clinical facilitation sessions and should be signed by your supervisor. In addition, it is expected that students discuss difficult or interesting cases in clinical meeting with the entire team. All notes and letters should be co-signed by a fully registered HPCSA Medical Geneticist or Genetic Counsellor. Students will be expected to follow-up and discuss all their patients (to be discussed with relevant registered supervisor).

Continuous (50%):
- Continuous clinical case assessments (40%)
- End-of-block performance evaluation (40%)
- 2(Y1) and 3(Y2) Case reports (20%)

Examination (50%):
- Clinical case exams midyear (33%)

Clinical case exams end of year (33%)
Written exam (33%)

Genetic Counselling Minor Dissertation

It is taught over a 12 month period as:
PTY5001W Genetic Counselling Minor Dissertation

Outline of the course

This module involves an introduction to Research Methodology (focus is qualitative research methods) to prepare students for writing a proposal in the first year and conducting research for a minor dissertation (maximum of 20 000 words from introduction to end of conclusion). The research should have relevance to the field of genetic counselling and involve issues related to patient perceptions and experience.

The majority of the course content will be covered in a short course. Additional lectures may be arranged as required. It is also the onus of the student should they feel that they require extra training.

The general aims of the course are to enable the student to:
- critically appraise the literature;
- participate in the generation of new knowledge in genetic counselling through research activities; and
- Continually subject his/her counselling practice to critical reflective review.

Objectives

The objectives of the module are to enable the student to:
- Define research;
- Discuss the importance of research to genetic counselling practice;
- Explain the difference between quantitative and qualitative research;
- Discuss the importance of a clearly articulated research question;
- Design research aims and objectives;
- List the elements of a well-formulated research objective;
- Describe the different research designs;
- Explain the purpose of each design;
- Explain different methods of data collection;
- Explain different sampling methods;
- Explain inclusion and exclusion criteria;
- Identify different types of data generate;
- Understand different types of measuring instruments;
- Discuss the issues of type of questions, construction of questions, cultural sensitivities and sequence of questions as related to questionnaire design;

- Understand the principles of quality in research;
- Discuss the underlying principles of ethical research;
- Identify the aims and objectives of published research papers and critically analyse the objectives;
- Conduct a comprehensive literature review;
- Formulate a research proposal by applying the principles of research methodology;
- Conduct and write up a scholarly research project which is relevant to genetic health issues.

Topics

Overview of the research process
- What is qualitative research?
- Qualitative research designs.
- Paradigms, theories and approaches

The research question
- Good research question
- Rigour – Triangulation, Credibility, Dependability, Confirmability, Transferability

Data collection
- Interviewing
- Questions
- Data collection methods

Data analysis
- Transcribing
- Coding
- Deriving themes

Assessment

The proposal has to be submitted to the Faculty Research Ethics Committee (HREC) by the first year and the dissertation by August of the second year to enable the student to graduate in December. The dissertation will be marked by two external international examiners. A pass mark of 50% is required. The student will present a critical analysis of two journal articles (one in year one and one in year two) and a seminar presentation at the end of second year.

Reference materials

1. Barbour, Rosaline (2008) ***Introducing qualitative research: A student guide to the craft of doing qualitative research.*** SAGE Publications: London.
2. MacFarlane, I. M., Veach, P. M., & LeRoy, B. (2014). *Genetic counseling research: A practical guide*. Oxford University Press, USA.
3. Neville, Colin (2007) The ***complete guide to referencing and avoiding plagiarism.*** Open University Press, McGraw-Hill: Maidenhead, UK

4. Polgar, Stephen and Thomas, Shane A (2008) ***Introduction to research in the health sciences (5th ed).*** Churchill Livingstone: London.
5. Silverman, David (2006) ***Interpreting qualitative data (3rd ed).*** SAGE Publications: London.
6. Smith, Jonathan A (2008) ***Qualitative psychology: A practical guide to research methods (2nd ed).*** SAGE Publications: Los Angeles, CA.
7. Holloway, I., Galvin, K. (2017). ***Qualitative research in nursing and healthcare (4th Ed)***. John Wiley & Sons: Iowa
8. Savin-baden, M., Howell Major, C. (2013) ***Qualitative Research: The essential guide to theory and practice*****.** Routledge: London

These books are available in the Health Sciences Library.

In addition to the recommended book there are several excellent web sites on research. The student is encouraged to read around the lecture topic as not everything of importance can be covered during the module.