Core competences in genetics for health professionals in Europe

Background document: development of a set of core competences in genetics for health professionals

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Background information to the development of sets of core competences in genetics for health professionals

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Background document: development of a set of core competences in genetics for health professionals

1. Rationale and scope of the core competence documents

The science of genomics is increasingly important to healthcare provision in Europe as an estimated 30 million people now suffer from genetic diseases within the enlarged European community (Cassiman, 2005). Genomics is having an increasing impact on the diagnosis, prevention and treatment of common disease (Collins, 2001). The number of tests being performed is changing rapidly (Constantin et al, 2005), with a concurrent increase in the number of individuals that have to make decisions about testing that may profoundly influence not only their own lives, but the lives of their family members. More professionals will be involved in testing, either directly or in dealing with the impact of testing on healthcare provision (Guttmacher et al, 2007). It is therefore essential that health professionals are appropriately prepared to deliver such patient care.

Status of clinical genetics as a specialty in Europe

The status of clinical genetics as a specialty in Europe varies from country to country. In several countries, clinical genetics is officially recognised as a specialty, with a specific and prescribed medical training in clinical genetics in departments of genetics or genetic laboratories. However, at present it is not identified as a medical specialty at European level. The European Society of Human Genetics has established an ad hoc Board to pursue the aim of establishing medical genetics as a European specialty to facilitate development of genetic healthcare in the European community. This group has issued guidelines which have been endorsed by the ESHG Board and are being used as a basis for discussions with the European Union of Medical Specialists (UEMS). The ad hoc Board has drafted a core curriculum on genetics for medical education but these have not yet been endorsed by UEMS.

Placing the recommendations for core competences into the context of the EuroGentest project

EuroGentest [www.eurogentest.org] is a project funded by the European Union for 5 years, the aim of which is to create a European Network of Excellence (NoE) in genetic testing (Hayhurst & Cassiman, 2006). It aims to develop the necessary infrastructure, tools, resources, guidelines and procedures that will structure, harmonize and improve the overall quality of all EU genetic services at the molecular, cytogenetic, biochemical and clinical level (Cassiman, 2005). While some of the units are naturally focused upon the practical issues related to developments in laboratory testing (Unit 5), the quality of genetic tests (Unit 1) and the use of data generated by tests (Unit 2), there are other important aspects of the project that aim to facilitate the safe use of genetic tests in a social context within Europe. These are addressed in Units that relate to the provision of appropriate counselling to
accompany genetic testing (Unit 3) and the ethical, legal and social issues surrounding testing (Unit 4). Unit 4 is now closely connected at an operational level with Unit 6, which is concerned with ensuring that patients are informed of the relevant information associated with any test (Work Package 6.1) and the education of professionals who offer, arrange and perform such tests (Work Package 6.2). In this Unit, patients and professionals are working in partnership at every level of the project, each informing the priorities and work of the other.

An extensive literature review was undertaken as a foundation for the project. This is described in the Year One Report of Work Package 6.2. In addition, several databases of direct relevance to professional education were compiled and these are available at the EuroGentest website [www.eurogentest.org]. At the meeting of the project team and experts in 2005, a decision was made to focus work on the development of a set of core competences in genetics for health professionals in Europe. The discussion that led to this decision centred on the need for a common minimum standard of education and training for all health professionals, to equip them to practise appropriately. It was obvious that the level of knowledge, skills and attitudes related to genetic healthcare varied according to the profession of the practitioner, the setting in which he or she worked and the relevance of genetics to his or her area of practice.

It is acknowledged that there are significant differences in the way in which professional education is delivered and practice is regulated across the countries of Europe. While a minimum standard for practice is required, setting curricula is not viewed as a practical way of ensuring those standards are met, given the differences in systems. The establishment of core competences is currently being used as a basis for health professional education in many other fields and settings (Walton & Elliott, 2006; Wold et al, 2006; Smith, 2005). It was therefore agreed that a pragmatic and workable solution would be to describe and agree, by consensus, a set of core competences that could apply to health professionals in Europe, whatever their national setting. This could provide an appropriate framework for establishing minimum standards of preparation for health care professionals in genetics across national boundaries. While core competences in genetics have been described for some health professionals in some countries, a set of competences that could be applied across Europe to the range of health professionals involved in provision of genetic healthcare does not yet exist.

These core competences are presented in two separate documents: i) for those professionals whose specialisation is in genetics and ii) for those professionals who are generalists or who specialise in an area of health care other than genetics. It must be emphasised that in each case the recommendations have been based on work involving the particular group of professionals involved.

While the two core competence documents described above address the needs of existing professionals, and can be used as the basis for writing curricula, we are aware that it will be helpful to write a set of competences in
genetics that would directly inform the education of health professionals at the undergraduate level. Such documents exist in the United Kingdom but have not been developed in the European context. This will be the focus of future work by this project team.

2. Evidence provided for the need for further genetic education for health professionals

The term genomics relates to the interactions between genes and the environment that influence health and disease (Collins et al, 2003). While genetic healthcare has been offered for several decades by specialist doctors, nurses and counsellors working in the context of genetics clinics, increasingly it is clear that knowledge of genetics is needed by health professionals to practice in the current genomic era (Guttmacher et al, 2007). As understanding of the impact of gene variation on the complex or common diseases increases, the number of health professionals who require education in genetics also rises. To respond to the needs of patients, health care providers need a set of core skills and knowledge to evaluate family history and to recognize clinical findings that indicate increased genetic risk. Primary care provision is increasingly directed at proactive rather than reactive consultation and this is particularly relevant to genetic medicine. Many patients will approach a primary care practitioner in the first instance (Burke, 2005a) but the question of genetic contribution to a disease may also arise in the context of secondary care such as oncology. In recent years, dentists have also been identified as practitioners who have input into diagnosis and management of genetic disease and are therefore also required to develop competences in this area (Dudlick, 2004; Gettig & Hart, 2003). While specialist genetic services will continue to provide genetic diagnosis and counselling for a number of the rare inherited conditions, practitioners in both secondary and primary care will need to be equipped to deal with initial patient enquiries, to have an awareness of the potential implications of family history, to offer information related to their specific area of practice and refer individuals or families appropriately to other healthcare providers, possibly including specialist genetic services. Because of the dynamic nature of the field, practitioners at all levels of health care, primary, secondary and tertiary, require both pre-registration and continuing education in genetics to ensure early diagnosis and prevention of disease. Due to the different levels of expertise required by the range of practitioners involved in delivering acceptable genetic healthcare, a set of competences at each level is also required.
3. Existing frameworks of core competences in genetics for health professionals

A level of professional education in genetics for some specific health professionals exists in many countries in Europe. However, few countries have established sets of core competences. In this section, we will briefly describe these.

1. United Kingdom

a) Nurses and midwives

In the United Kingdom, the core competence work undertaken by Kirk et al (2003) focused on nurses and midwives who work in mainstream healthcare as opposed to specialist genetic services. A research-based approach was taken to the formulation of the set of competences, using an Expert Panel of stakeholders to come to a consensus about the requisite competence in knowledge, skills and attitudes required by nurses and midwives at the point at which they became professionals registered for practice [http://www.glam.ac.uk/socsschool/research/gpu/FinalReport.pdf]. The agreed set of genetic competences have been aligned to the general statements of competence that are required of each practitioner before registration with the Nursing and Midwifery Council, the statutory body responsible for standards of practice in nursing and midwifery.

b) Specialist genetic nurses and genetic counsellors

In preparation for the introduction of a formal registration system for genetic nurses and counsellors (Skirton et al, 2003), the Education Working Party of the Association of Genetic Nurses and Counsellors in the UK (2006) devised a set of 23 competencies for genetic nurses and counsellors (non-medical) [www.agnc.org/Registration/registration.htm]. These were prepared by a group of specialist practitioners and were informed by the competences for specialist nurses published by the precursor to the Nursing and Midwifery Council, (called the United Kingdom Central Council for Nursing, Midwifery and Health Visiting). These 23 competences are used as the basis for a portfolio of evidence that is assessed prior to the awarding of registration as a genetic counsellor.

c) Non-genetics healthcare professionals

In the United Kingdom, a competence framework describing genetic activities that may be carried out by non-genetics healthcare staff has been developed by the NHS National Genetics Education and Development Centre, Skills for Health and a wide range of healthcare professionals [http://www.geneticseducation.nhs.uk/develop/index.asp?id=44]. This competence framework describes how each genetics activity should be performed and the underpinning knowledge and understanding required to carry out the activity. The competence framework describes 9 activities:

- Understand genetics within your area of clinical practice
- Identify patients with or at risk of genetic conditions
- Gather multi-generational family history information
- Use multi-generational family history information to draw a pedigree
- Recognise a mode of inheritance in a family
• Assess genetic risk
• Refer individuals to specialist sources of assistance in meeting their healthcare needs
• Recognise the indicators for and the implications of ordering a molecular genetic test
• Communicate genetic information to patients, families and healthcare staff.

These competences will be incorporated into job descriptions of health professionals to describe a competence benchmark for any genetics activity they currently perform. Only those competences relevant to the healthcare professional’s role will be included in their job descriptions.

d) Medical professionals
Work has been undertaken in the UK to ensure the curricula and training for undergraduate medical students, general practice trainees, trainees in specialties other than genetics and trainee medical geneticists include the requisite genetics content. Additional information on curricula and learning outcomes for medical professionals in the UK is included in Appendix 1.

2. USA

a) Health professionals (across disciplines)
The National Coalition for Health Professions Education in Genetics (NCHPEG) established in 1997 is an interdisciplinary group comprising leaders from over 120 health professional organizations, consumer and voluntary groups across the USA. The NCHPEG prepared core competencies [http://www.nchpeg.org/core/Corecomps2005.pdf] recommending that all health professionals possess the core competencies in genetics to enable them to integrate genetics effectively and responsibly into their current practice. Competency in these areas represents the minimum knowledge/skills/attitudes necessary for health professionals from all disciplines (medicine, nursing, allied health, public health, dentistry, psychology, social work, etc.) to provide care to their patients that involve awareness of genetic issues and concerns.

According to NCHPEG, every health care professional should at a minimum be able to:
1. Appreciate the limitations of his or her genetic expertise
2. Understand the social and psychological implications of genetic services
3. Know how and when to make a referral to a genetics professional.

In addition other recommendations for a genetic curriculum have been developed in the USA for continuing education (Jenkins et al, 2001).

b) Nursing
A recent consensus document sponsored by NCHPEG detailing the core competencies in genetics for general nurses in the USA and guidelines for development of nursing curricula has now been ratified by a large number of nursing organisations, including the American nurses Association and the International Society of Nurses in Genetics
Previously, a study by Calzone et al (2002) was used as a basis for development of a set of core competencies in genetics for oncology nurses working in the USA.

The International Society of Nurses in Genetics has, in conjunction with the American Nurses Association, produced a detailed document on the scope and standards of practice for specialist genetic nurses (Greco et al, 2006).

3. **France**
   a) Dentistry

Dentists in France are expected to develop competences in genetics (Monteil, 2006). These have been included in the specific competences for dentists included in Section 2.

The systems described above have been useful in developing proposed sets of core competences for Europe.

4. **Categorization of the three professional groups and the need to describe different levels of competence**

Given the need to prepare a range of healthcare professionals across the primary, secondary and tertiary care spectrum and the need to set up a common framework for use in a European context, three groups of health professionals that require education and training in genetics were identified:

- Health care professional working in a generalist settings
- Specialist health professionals (non-genetic)
- Specialists in genetics.

It was agreed to use these groups as a framework for the work on development of a set of core competences. Each group requires a different level and depth of genetics education, and the classification has been adopted to enable the needs of each group to be addressed. In addition to setting for practice [mainstream healthcare, specialist (non-genetic) and specialist genetic], it was agreed to divide the groups by professional background, the main groups being medical, nursing/midwifery, dental and laboratory professionals.

For each group, the project team will agree a list of competences and the learning outcomes associated with each competence. The learning outcomes will therefore be available as guidance in the development of curricula that are appropriate to the national context, educational system and healthcare setting of the professional involved.

The main groups for the purposes of developing competences are:
1. **Health care professionals working in generalist settings**

This group includes all healthcare professionals who work in general settings, such as general practitioners, district nurses, physicians in general medicine or surgery, nurses working in general acute settings, general midwives and dentists. Evidence that additional education in genetics is required for nurses has been provided in recent studies (Barr & McConkey, 2006; Burke & Kirk, 2006) that demonstrated that while nurses and midwives perceive a need for genetics knowledge, they feel underprepared for practice in relation to genetics. Similarly, in a study of graduates of six allied health professional training programs, Christianson et al (2005) showed that allied health professionals were engaging in tasks related to genetic healthcare with their clients (such as discussing patterns of inheritance), but 78% of those studied did not believe they were sufficiently prepared during training to undertake those tasks. McCann et al (2004) undertook a study of over 500 general practitioners that indicated that the majority were keen to access more training in genetics. Family practitioners surveyed by Burke et al (2006) indicated they felt genetics was an important topic for practice but that they felt their educational preparation was lacking.

2. **Specialist health care professionals (non geneticist)**

This group includes practitioners who specialise in one area of healthcare and who require specific genetics education to enable them to provide care to the specialised group of patients. Practitioners in this group would include colorectal or breast surgeons, haematologists, fetal medicine midwives, obstetricians, sickle cell disease nurses, and specialist dentists. Recent work by Burke et al (2006) indicated that preparation of doctors in a number of specialist areas (neurology, cardiology and dermatology) is still inadequate in terms of knowledge of genetics that would be directly relevant to their field of practice. A Working Party on genetics in haemophilia care strongly suggested that nurses working in this speciality have relevant education in genetics to enable them to support patients and their families appropriately (Ludlam et al, 2005). The need for oncology nurses to develop competence in genetics has been suggested by Lewis et al (2006) and Skirton (1999).

3. **Specialists in genetics**

The clinical genetics specialist team may include many professionals. However, not all will be considered specialists in genetics and core competences for only those fulfilling the criteria of specialist professional in genetics will be described. The terminology is challenging because of the range of terms for professionals, for example a medical doctor offering genetic healthcare may be called a medical geneticist in some countries and a clinical geneticist in others. Laboratory staff without a medical qualification may be allowed to offer genetic counselling directly to patients in some countries, where they are called medical geneticists, but this is not permitted in others. The term genetic counsellor is used to describe anyone who offers genetic counselling in some countries, but is the title for non-MD counsellors with a nursing or master qualification in the UK. We have therefore tried to use
generic, descriptive terms to classify the professionals involved in giving specialist genetic healthcare. The core competences can then be applied to those professionals that are offering the applicable level and type of genetic healthcare activity in each country. In general, Geneticists [Medical Doctors (MD)], genetic nurse specialists and genetic counsellors (non MD) will be geneticists responsible for clinical care, while cytogeneticists and molecular geneticists will be responsible for laboratory services.

5. Obtaining consensus on a set of core competences

At present, there is neither a coherent policy nor a defined set of guidelines regarding core competence in genetics Europe. There are, however, curricula described for the education and training of genetic health professionals in many countries. Since the practice of medicine is firmly regulated, particularly for medical doctors and for laboratory staff participating in direct patient care, the existing curricula can be used to inform the development of core competences, although the genetic components of such curricula are still in many cases inadequate to the needs of current and future professionals. The examples given above in Section 1.4 demonstrate that it is possible to obtain consensus on the competences required by health professionals. It was also clear when collecting the data included in the Year One report that educational preparation has not been consistent with introduction of genetic testing and genetic counselling in Europe and it is felt that a framework of core competences would support the development of appropriate learning outcomes for the necessary courses and training. While the schemes devised by other organisations outside Europe are helpful when used as guidance, it is essential that a framework that is sensitive to the historical background of European countries and the current needs of European citizens is required. For example, in the UK a Knowledge and Skills Framework for health professionals who are not genetic specialists has been devised. However, the sets of competences, learning outcomes and curricula for professionals working in specialist genetics is now well-developed in the UK and this approach may be more useful when a framework for professional education in genetics already exists.

It should be noted that these core competences relate to clinical and laboratory health services, rather than public health. A Working Group of Public Health Genetics (PHG) on Policy Development for Education and Training in Genomics for Population Health is currently addressing the core competences required by Public Health professionals and these will be published by that group.
Table 1. Examples of the differences in professional titles used in European countries.

<table>
<thead>
<tr>
<th>Description</th>
<th>Terms used</th>
<th>Countries where used (examples)</th>
</tr>
</thead>
</table>
| Medical doctor trained in genetics and competent to offer specialised clinical genetic services | Medical geneticist  
Specialist geneticist  
Clinical geneticist | Italy, Portugal, Turkey, UK  
Belgium, Netherlands, Sweden, UK |
| Laboratory scientists trained in human genetics and competent to offer molecular genetic testing in a laboratory environment | Molecular geneticist  
Medical geneticist  
Clinical scientist  
Clinical Molecular Geneticist | Italy (if PhD), UK  
Italy (if MD)  
UK  
Netherlands (if PhD) |
| Laboratory scientists trained in human genetics and competent to offer cytogenetic testing in a laboratory environment | Cytogeneticist  
Medical geneticist  
Clinical scientist  
Clinical cytogeneticist | Italy (if PhD), UK  
Italy (if MD)  
UK  
Netherlands |
| Nurse trained in specialist genetics and competent to provide genetic counselling in a clinical setting. | Genetics nurse  
Genetic counsellor | Denmark, Netherlands, Norway, Sweden, UK  
UK |
| Individual trained through Master's degree in genetic counselling and competent to provide genetic counselling in a clinical setting. | Genetic counsellor | France, Sweden, UK |

It is also necessary to emphasise that the core competences relate to a specific health professional in a given setting, and are not a means for assessing the general quality of the service. The services should be evaluated using other means, such as the guidelines issued by the Public Policy Committee of the European Society of Human Genetics (available at http://www.eshg.org/).

The expert group for EuroGentest Work Package 6.2 has assembled two sets of core competences for health professionals. The goal of this work is not to unify the existing genetic services across national boundaries. However, by hopefully achieving consensus, the document will help countries to adjust their education and genetic service delivery systems for the future, according to a coherent set of standards. The consensus document will guide the states of Europe with regard to how the new generation of health professionals should be educated in the post-genomic era and how genetics training should
be built into the new European higher education system (the Bologna process) (Confederation of EU Rector’s Conferences and the Association of European Universities, 1999). National societies and professional and patient groups are invited to refine the competences, learning outcomes and curricula consistent with their own needs. We are actively seeking input and would be willing to meet with professional and patient groups in different countries to receive feedback and to support development of competences that are applicable to the local and regional situation.

All aspects of the core competences have been developed with consideration of the policies of the European Society of Human Genetics Public Policy Committee and support those policies.

Relevant definitions

There are many different models of competence suggested by authors in education and a range of different professional fields (Cheetham & Chivers, 1996). For the purpose of this document, competences are defined as the set of behaviours that are expected by independent professionals (Plasschaert et al, 2002). The competence therefore describes the behaviour expected by the professional in the particular setting in which he or she works. Learning outcomes are used to dissect the competence into more manageable units that can be the focus of learning. Each learning outcome encompasses particular knowledge, skills or attitudes. In turn the learning outcomes can be used as a basis to build a curriculum. The curriculum should include the information and opportunity for skill development to enable the student or trainee to acquire the necessary knowledge, skills and attitudes to work competently. We are aware that in many countries there are curricula that have been devised to educate and train health professionals in genetics. These curricula could be assessed against the competences and learning outcomes for particular groups of professionals to ensure that all the requisite areas are covered.

Before setting out the specific core competences for each type of health professional that may deliver genetic healthcare on the three levels, the main types of interventions used in genetic healthcare can be assigned in broad terms, according to the appropriate professional involved. Table 2 is presented as a general foundational structure for the specific core competences that follow in this section. It must be appreciated however, that each genetic counselling situation is context specific and the appropriate person to provide care may vary according to the precise family circumstances, type of disease and level of expertise of the professional. As a general rule, counselling accompanying predictive and pre-natal testing will be offered by trained genetics specialists (medical and non-medical). However, counselling for a range of other genetic/genomic tests may be undertaken by other health professionals.
Table 2. General foundational structure for genetic healthcare interventions, core competences and type of professional involved.

**Key for Table 2: Health care professionals**

<table>
<thead>
<tr>
<th>Professionals who should have competence</th>
<th>MD</th>
<th>DDS</th>
<th>DDM</th>
<th>MDs</th>
<th>DMDs</th>
<th>MDg</th>
<th>DMDg</th>
<th>N-MDg</th>
<th>C/Mg</th>
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<tbody>
<tr>
<td>Medical doctor working as generalist</td>
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<td></td>
<td></td>
<td>MD</td>
<td>DDM</td>
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<tr>
<td>Doctor of Dental Surgery</td>
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<td></td>
<td>DDS</td>
<td>DDM</td>
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<tr>
<td>Doctor of Dental Medicine</td>
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<td>Dentist working in specialty other than genetics</td>
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<td></td>
<td></td>
<td>MDs</td>
<td>DMDs</td>
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<tr>
<td>Dentist with clinical genetics specialty</td>
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<td></td>
<td></td>
<td>MDg</td>
<td>DMDg</td>
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<tr>
<td>Nurse/midwife/genetic counsellor with registered genetic licence</td>
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<td>N-MDg</td>
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<td>Cyto/molecular geneticist with registered laboratory licence</td>
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<td>C/Mg</td>
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**Main stages of interventions**

<table>
<thead>
<tr>
<th>Competences</th>
<th>MD</th>
<th>DDS</th>
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<th>MDs</th>
<th>DMDs</th>
<th>MDg</th>
<th>DMDg</th>
<th>N-MDg</th>
<th>C/Mg</th>
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<tr>
<td>Initiation of genetic examinations</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
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<td>X</td>
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<tr>
<td>➢ identify persons and families whose disorder or condition is determined, partly or fully, by genetic component;</td>
<td>X</td>
<td>X</td>
<td>X</td>
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<td>X</td>
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<td>➢ determine the accuracy of the clinical diagnosis, and to initiate, if needed, additional clinical examination for exact diagnosis;</td>
<td>X</td>
<td>X</td>
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<td>X</td>
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<td>➢ be familiar with the feasibility and accessibility of genetic services that could help persons and families through genetic counselling.</td>
<td>X</td>
<td></td>
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<td>X</td>
<td>X</td>
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<td></td>
<td>Pre-test genetic counselling</td>
<td>Performing genetic tests</td>
<td>Post-test genetic counselling</td>
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<td>➢ provide accurate speciality-focused view on the nature of a genetic disorder; ➢ determine the need for and utility of genetic tests relating to a disease or special condition; ➢ understand the meaning of genetic test results and translate those results into practical disease-specific information; ➢ provide information for persons on the benefit and risk of a genetic test prior to obtaining consent for the test.</td>
<td>X</td>
<td>X</td>
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<td>➢ perform a range of genetic tests for the purpose of diagnosis, and ascertainment of genetic susceptibility and carrier status; ➢ interpret the results of genetic tests that are performed; ➢ provide information to health care professionals based on both the results and interpretation of the results.</td>
<td></td>
<td>X</td>
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<td>➢ help individuals and families to understand the information provided during genetic counselling; ➢ determine the risk of occurrence or recurrence of a disease or special condition;</td>
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- facilitate understanding between individuals, families, their family doctors and specialists about genetic disorders, test results, and inheritance pattern;
- facilitate communication of risk or potential risk to appropriate family members.

| Predictive genetic counselling | translate and interpret genetic test results to persons who are at high risk of having genetically determined disease in their lifetime. | X | X |
| Pre-natal genetic counselling | provide genetic information that helps persons or couples make reproductive decision. | X | X |
References


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