

# **Core Competences in Genetics for Health Professionals in Europe**

## **04 – Suggested core competences for health professionals who are specialists in genetics**

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# Core competences for health professionals who are specialists in genetics

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## General introduction

The following information is provided as a background to the core competences for specialist in genetics:

- Clinical genetics, a medical specialty as well as an extensive field of knowledge, provides a unique health care service to people.
- Although clinical genetics interacts with every medical discipline during service delivery, one of its distinctive objectives is the inclusion of care of future generations through genetic counselling and prenatal diagnosis (i.e. transgenerational care). This service is based on interdisciplinary consultations, wide-ranging diagnostic investigations, communication processes and emotional support for the patient and family. The competencies, therefore, should be determined by focusing on the genetic counselling, genetic diagnosis and management of uncertainty when a diagnosis is not made.
- In the provision of genetic services, the interventions of the health professionals may be grouped into the following main categories:
  - initiation of genetic examinations
  - diagnosis
  - presentation of options for testing/management/prevention
  - counselling with respect to decision-making
  - support after decision-making
  - performing genetic tests
  - post test counselling.
- For practical reasons, it is worth distinguishing prenatal counselling and predictive counselling as special interventions undertaken by genetics specialists.
- Although the skills and attitudes are basically not professional specific, there are some which needs special knowledge and training. Hence, in the preparation of professionals to operate in the specialist genetic services there are distinct competencies which should be bound to the relevant system of registration and/or of licensing of practitioners in the country concerned.
- Increasingly, medical specialist in areas outside traditional medical genetics may develop specific areas of expertise in genetics and practice as a geneticist specialising in a particular disease or patient group. The foremost example of this is the oncology or cancer geneticist. However, it is envisaged that specialists in other fields (such as cardiovascular genetics) will seek to practice as genetic specialists. As at present these are few, we have included only the competences required by an oncology geneticist. This may, however,

be used as a model for defining competences in other genetic specialties.

- There is some discussion about the use of the terms 'patient', 'client' and 'counselee' in reference to those who use genetic services. We acknowledge that a person who uses genetic services may not always be a patient in the traditional sense of the word, as he or she may not have any medical condition. However, for simplicity, we have used that term in this document.

Further information on this topic is available on the EuroGentest website under Unit 6. [<http://www.eurogentest.org/unit6/>]. Other documents include an *Executive Summary*, *Background document* and *Core competences for generalists and those who specialise in a field other than genetics*.

## 1. Clinical geneticist

- **Professional background**

The Clinical or Medical Geneticist is a qualified medical practitioner who has undertaken specific training in medical genetics, as specified in the country of practice and is licensed to offer services as a specialist in genetics according to the law in the country of practice.

- **Setting for practice**

In general, the Clinical Geneticist will function as part of a multi-disciplinary team within a specialist genetic service, often operating as a tertiary care facility. In some cases, the Clinical Geneticist will work within a team that offers services to a particular part of the population, such as children or pregnant women or to patients who are affected with a particular type of disease, such as cancer, neurological or metabolic conditions.

Competence	Some proposed learning outcomes
1. Identify individuals and families whose disorder or condition is determined, partly or fully, by a genetic component.	1.1 Is aware of the potential genetic component to disease.  1.2 Takes an accurate family and medical history.
2. Determine the accuracy of the clinical diagnosis, and initiate, if needed, additional clinical examination to make an exact diagnosis.	2.1 Ascertains sufficient medical, family and personal information from the client to make appropriate genetic risk assessment.  2.2 Ascertains medical information from other sources to confirm family information and diagnosis.  2.3 Performs clinical examinations as appropriate.  2.4 Orders clinical investigations as appropriate.  2.5 Understands all potential patterns of inheritance and the underlying mechanisms by which genetic disease may occur.
3. Be familiar with the feasibility and accessibility of genetic services that could help persons and families through genetic counselling.	3.1 Seeks to raise awareness of available services and resources related to genetic healthcare.  3.2 Acts as a resource for other professionals and lay groups.

<p>4. Provide an accurate speciality-focused view on the nature of a genetic disorder.</p>	<p>4.1 Actively seeks opportunities to update knowledge and skills, and reflects on the implications of these for own practice and that of professional colleagues.</p> <p>4.2 Collects, evaluates and uses relevant information about the genetic disorder in question.</p> <p>4.3 Critically appraises current evidence to inform practice and professional development.</p> <p>4.4 Disseminates evidence of good practice and service improvement through verbal and written media.</p> <p>4.5 Facilitates understanding of how genetics impacts on affected individuals, their families, partners and carers.</p>
<p>5. Determine the need for and utility of genetic tests relating to a disease or special condition.</p>	<p>5.1 Is aware of current approaches and limitations of tests.</p>
<p>6. Understand the meaning of genetic test results and translate those results into practical disease-specific information.</p>	<p>6.1 Maintains own knowledge base.</p> <p>6.2 Provides information for persons on the benefit and risk of a genetic test prior to obtaining consent for the test.</p>
<p>7. Help individuals and families to understand the information provided during genetic counselling.</p>	<p>7.1 Provides information about the genetic disorder appropriate to the client's assessed needs, reflecting their values, religious and cultural beliefs and preferences.</p> <p>7.2 Provides information based upon appropriate interpretation of genetic and clinical knowledge.</p> <p>7.3. Communicates with respect to the genetic risk assessment and possible options.</p>
<p>8. Facilitate understanding between individuals, families, their family doctors and specialists about genetic disorders, test results, and inheritance patterns.</p>	<p>8.1 Communicates information about the specific genetic disorder, test results, and inheritance pattern relevant to the condition to the affected or at risk individual.</p> <p>8.2 Promotes dissemination of information to family members as appropriate.</p> <p>8.3 Conveys the implications of test results to individuals in terms of potential prognosis, screening options and management options.</p>

	<p>8.4 Participates in education programs to promote understanding of genetic conditions amongst health professionals.</p> <p>8.5 Promotes public understanding of genetic conditions, tests and inheritance patterns through appropriate means.</p>
<p>9. Determine the risk of occurrence or recurrence of a disease or condition.</p>	<p>9.1 Makes an accurate genetic risk assessment based on diagnosis and published data.</p>
<p>10. Understand the genetic and environmental components of common diseases.</p>	<p>10.1 Understands the epidemiological relevance of genetic markers (e.g. DNA variants and polymorphisms) in common diseases.</p> <p>10.2 Contributes to health policy planning.</p> <p>10.3 Understands the potential benefits and disadvantages of the use of biobanks.</p>
<p>11. Provide genetic information that helps persons or couples make informed reproductive decisions.</p>	<p>11.1 Enables clients to make informed choices about the implications of their family history.</p> <p>11.2 Provides information on a range of reproductive options.</p>
<p>12. Work within the boundaries of ethical practice.</p>	<p>12.1 Recognises practice limitations and demonstrates referrals to other health professionals when appropriate.</p> <p>12.2 Consults other health professionals when the client's needs fall outside the scope of genetic practice.</p> <p>12.3 Upholds professional standards of safe and ethical practice at all times.</p> <p>12.4 Uses professional standards of practice to evaluate own and others' performance.</p> <p>12.5 Contributes to the debate on ethical challenges in genetic practice.</p>

## 2. Oncology geneticist

- **Professional background**

The oncology geneticist or cancer geneticist is a qualified medical practitioner who is trained in clinical genetics and cancer genetics.

- **Setting for practice**

The oncology geneticist may work as part of a multi-disciplinary team within a specialist genetic service, often operating as a tertiary care facility. However, he or she may also work within a team that offers services to those individuals or families who have been diagnosed with cancer or have a family history of cancer in a department affiliated with oncology or cancer services. The oncology geneticist is expected to have the basic competences applicable to the clinical geneticist.

<b>Competence</b>	<b>Some proposed learning outcomes</b>
1. Make an assessment of the risk associated with inherited cancer for individuals and families.	1.1 Elicits an accurate family history of cancer.  1.2 Verifies details of cancer family history.  1.3 Recognises inherited conditions which predispose to specific cancers.  1.4 Understands the various models for risk estimation dependant on family history, and their accuracy.  1.5 Estimates the likelihood that the individual counselled will develop specific cancers.  1.6 Understands the implications of specific tumour morphology in relation to cancer susceptibility.
2. Communicate cancer genetic risk information to individuals, families and other healthcare providers.	2.1 Elicits the individual's concerns and responds appropriately.  2.2 Clearly communicates the principles of oncology and genetics as required to inform individuals as to their inherited cancer risk.  2.3 Discusses the options available to those family members who are assessed as being at increased risk of cancer.  2.4 Facilitates discussion of risk or potential risk to appropriate family members.



<p>3. Utilise genetic and other appropriate laboratory tests to support genetic healthcare of individuals and families at risk of inherited cancer.</p>	<p>3.1 Recognises when genetic testing is appropriate in a family.</p> <p>3.2 Administers such tests, and discusses the communication of the results of genetic tests to at-risk relatives with appropriate consent.</p> <p>3.1. Arranges predictive testing for at-risk relatives where the mutation is known, adhering (as appropriate) to a predictive testing protocol.</p> <p>3.4 Understands the sensitivity, specificity and significance of tests on tumour samples.</p> <p>3.5 Understands the implications of the detection of a variant of unknown significance in a cancer predisposing gene, and the strategies that can be employed to elucidate the pathogenicity further.</p> <p>3.6 Understands the frequency of specific ancestral mutations in certain populations, and the implications for testing for such mutations.</p>
<p>4. Identify, communicate and arrange appropriate surveillance protocols, with discussion of the benefits and limitations of such surveillance.</p>	<p>4.1 Determines the level of cancer risk for which surveillance is cost-effective and appropriate for individuals with a family history of common cancers.</p> <p>4.2 Discusses options for surveillance with the individuals concerned.</p> <p>4.3 Liaises with other medical disciplines to provide appropriate surveillance and management for individuals at increased genetic risk of specific cancers.</p>
<p>5. Act to help prevent occurrence or recurrence of cancer where possible.</p>	<p>5.1 Understands the relevant and appropriate general health and lifestyle advice which may be appropriate to give.</p> <p>5.2 Understands any modification of management or treatment that may be appropriate for individuals carrying cancer predisposing mutations, as distinct from that appropriate for people in the general population.</p>
<p>6. Utilise the healthcare system for promotion of cancer genetics services.</p>	<p>6.1 Understands the organisation of cancer genetics services, and how health professionals such as trained genetic counsellors and nurses may contribute to these services.</p> <p>6.2 Understands the training that such healthcare professionals require in order to be competent to run cancer genetics clinics.</p>

## References

American Society of Clinical Oncology (1997): 1997 **Resource document for curriculum development in cancer genetics education**. J.Clin.Oncol. 15: 2157-69.

### 3. Specialist genetic nurse and genetic counsellor

While the work undertaken by genetic nurses and genetic counsellors is highly comparable, they may have different professional backgrounds and therefore while one set of competences is presented, the professional backgrounds are described separately. Professional roles vary across the countries of Europe, therefore this is presented as a suggested framework which may need to be adapted to local contexts.

#### a. Specialist genetic nurse

- **Professional background**

The specialist genetic nurse is a registered nurse who has undertaken specific education and training to enable him or her to develop the knowledge, attitudes and skills required to offer a specialist service to clients or patients. Many specialist genetic nurses have additional qualifications in either midwifery or community nursing. In some countries a registration system exists to ensure the nurse has achieved appropriate standards of competence before working in an autonomous manner within the team.

#### b. Genetic counsellor

- **Professional background**

The genetic counsellor has typically completed a relevant first degree (e.g. in nursing, genetics, biology, psychology or social work), followed by a Master's degree in genetic counselling which includes human genetics and counselling skills training.

In some countries a registration system exists to ensure the nurse has achieved appropriate standards of competence before working in an autonomous manner within the team.

- **Setting for practice**

The specialist genetic nurse or genetic counsellor may be employed as part of a multi-disciplinary team providing generic genetic services. The setting in which the nurse or counsellor works may be focussed on a specific disease or genetic setting (such as oncology or prenatal). In general, genetic nurses and counsellors will work with families with a pre-established diagnosis or in a general setting under the supervision of a clinical doctor. The genetic nurse or counsellor will not be responsible for making diagnoses.

\* As the work undertaken by specialist genetic nurses and genetic counsellors are highly comparable, one set of competences is presented here for both types of practitioners.

<b>Competence</b>	<b>Learning outcomes</b>
1. Establish relationship and clarify clients' concerns and expectations.	<p>1.1 Establishes an environment which facilitates client to expression of feelings, anxieties, beliefs, and expectations and considers clients' experiences.</p> <p>1.2 Identifies client needs.</p> <p>1.3 Enables clients to make informed choices about the implications of their family history.</p> <p>1.4 Takes appropriate action to meet identified needs with the agreement of the client.</p>
2. Make appropriate and accurate genetic risk assessment.	<p>2.1 Ascertains sufficient medical, family and personal information from the client to make appropriate genetic risk assessment.</p> <p>2.2 Ascertains medical information from other sources to confirm family information and diagnosis.</p> <p>2.3 Understands the patterns of inheritance and the underlying mechanisms by which genetic disease may occur.</p>
<p>3. a. Convey clinical and genetic information to clients, appropriate to their individual needs.</p> <p>3. b. Explain options available to the client, including the risks, benefits and limitations.</p> <p>3. c. Evaluate the understanding of the individual related to the topics being discussed.</p> <p>3. d. Acknowledge the implications of individual and family experiences, beliefs, values and culture for the genetic counselling process.</p>	<p>3.1 Provides information about the genetic disorder appropriate to the client's assessed needs, reflecting their values, religious and cultural beliefs and preferences.</p> <p>3.2 Provides information based upon appropriate interpretation of genetic and clinical knowledge.</p> <p>3.3. Communicates with respect to the genetic risk assessment and possible options.</p> <p>3.4 Supports dissemination of information about the genetic disorder to at risk relatives by the client.</p>

<p>4. Make an assessment of clients' needs and resources and provide support, ensuring referral to other agencies as appropriate.</p>	<p>4.1 Ascertains psychological needs of the individual or family.</p> <p>4.2 Respecting clients' preferences, provides support and makes referrals to other agencies (such as psychologist or patient support groups).</p> <p>4.3 Identify and support clients' access to local, regional and national resources and services.</p>
<p>5. Use of a range of counselling skills to facilitate clients' adjustment and decision-making.</p>	<p>5.1 Uses safe, effective and appropriate counselling skills to support clients to make adjustments and decisions.</p>
<p>6. Document information including case notes and correspondence in an appropriate manner.</p>	<p>6.1 Uses a systematic approach to collecting and maintaining comprehensive and accurate records that detail the rationale underpinning any interventions.</p> <p>6.2 Maintains confidentiality and security of written and verbal information.</p>
<p>7. Find and utilise relevant medical and genetic information for use in genetic counselling.</p>	<p>7.1 Collects, evaluates and uses relevant information about the genetic disorder in question.</p> <p>7.2 Critically appraises current evidence to inform practice and professional development.</p> <p>7.3 Disseminates evidence of good practice and service improvement through verbal and written media.</p>
<p>8. Demonstrate ability to organise and prioritise a case load.</p>	<p>8.1 Addresses client needs in a sensitive and fair manner making best use of resources available</p> <p>8.2 Prioritises according to patient need.</p>
<p>9. Plan, organise and deliver professional and public education</p>	<p>9.1 Facilitates understanding of how genetics impacts on affected individuals, their families, partners and carers.</p> <p>9.2 Seeks to raise awareness of available services and resources related to genetic healthcare.</p> <p>9.3 Acts as a resource for other professionals and lay groups.</p>
<p>10. Establish effective working relationships to function within a multi-disciplinary team and as part of the wider health and social care network.</p>	<p>10.1 Promotes patient-centred care in partnership with the client, their family, and appropriate care providers.</p> <p>10.2 Facilitates communication via a strong multidisciplinary network of professional and lay colleagues.</p> <p>10.3 As appropriate, co-ordinates patient and family care.</p>

<p>11. Contribute to the development and organisation of genetic services.</p>	<p>11.1 Evaluates own practice and that of others in the light of new evidence and modifies practice appropriately.</p> <p>11.2 Uses skills of critical appraisal to consider how new evidence may contribute to the improvement of service organisation and delivery.</p> <p>11.3 Actively seeks opportunities to meet with colleagues to discuss professional issues and innovations in care, in order to disseminate best practice and improve standards of care.</p> <p>11.4 Actively seeks opportunities to collaborate with colleagues in audit and research that has the ultimate aim of improving client care</p>
<p>12. Practice in accordance with an appropriate code of ethical conduct.</p>	<p>12.1 Upholds professional standards of safe and ethical practice at all times.</p> <p>12.2 Uses professional standards of practice to evaluate own and others' performance.</p> <p>12.3 Recognises the duty to seek professional advice if standards of care are threatened.</p> <p>12.4 Contributes to the debate on ethical challenges in genetic practice.</p> <p>12.5 In normal circumstances discloses information about individuals to appropriate third parties only with the client's permission.</p>
<p>13. Recognise and maintain professional boundaries and limitations of own practice.</p>	<p>13.1 Recognises practice limitations and demonstrates referrals to other health professionals when appropriate.</p> <p>13.2 Consults other health professionals when the client's needs fall outside the scope of genetic practice.</p> <p>13.3 Refers clients to colleagues when necessary.</p>
<p>14. Demonstrate reflective skills and personal awareness for the safety of individuals and families.</p>	<p>14.1 Demonstrates reflective practice, which informs future clinical interactions.</p> <p>14.2 Maintains a portfolio recording reflection on practice.</p> <p>14.3 Accesses counselling and/or clinical supervision to underpin and enhance practice.</p>

15. Present opportunities for clients to participate in research projects in a manner that facilitates informed choice.	15.1 Enables clients to make an informed choice on whether to participate in a research project or not.
16. Demonstrate continuing professional development as an individual practitioner and for the development of the profession.	16.1 Actively seeks opportunities to update knowledge and skills, and reflects on the implications of these for own practice and that of professional colleagues.

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#### 4. Molecular geneticist (general or specialising in a particular field such as haemophilia)

- **Professional background**

The molecular geneticist is a qualified laboratory specialist who is trained in molecular genetic methodology and its usage in diagnostic, prognostic/predictive purposes, ascertainment of carrier status in patients and their family members. In some cases the molecular geneticist will have a medical qualification. He/she is able to interpret results and their clinical consequences in collaboration with the clinical geneticist.

- **Setting for practice**

Molecular genetics laboratory, publicly or privately funded, often linked to genetics service or academic department of medical genetics.

<b>Competence</b>	<b>Some proposed learning outcomes</b>
1. Work independently in the molecular genetics testing laboratory.	1.1 Has an expert knowledge of the principles of molecular genetics.  1.2 Is aware of the significance of molecular genetics in medicine and healthcare.  1.3 Demonstrates ability to manage workload effectively. 1.4 Appropriately prioritises ordered tests.  1.5 Understands safe codes of practice in the laboratory and adheres to these.  1.6 Supervises laboratory technicians.  1.7 Implements and follows up programs of internal and external quality assessment.  1.8 Ensures maintenance and calibration of laboratory equipment and devices is performed.
2. Perform a range of genetic tests for the purposes of diagnosis, ascertainment of carrier status and predictive testing.	2.1 Isolates nucleic acids from human samples and other organisms.  2.2 Chooses a suitable technique for DNA and RNA examination.  2.3 Performs tests using appropriate molecular genetic methodology, including restriction endonucleases, gel electrophoresis, PCR and its modification, mutation and polymorphism analyses, direct and indirect DNA diagnostics, sequencing and high throughput techniques.  2.4 Keeps accurate and contemporaneous records of the analyses.



<p>3. Interpret the results of genetic tests that are performed.</p>	<p>3.1 Understands principles of inheritance to determine the risk of occurrence or recurrence of a disease or special condition in the family, according to results of laboratory investigation.</p> <p>3.2 Is familiar with current scientific literature to inform interpretation of genetic test results.</p> <p>3.3 Participates in interpretation of the analyses.</p> <p>3.4 Evaluates the results of tests.</p> <p>3.4 Uses molecular genetics analysis software and computer systems appropriately.</p>
<p>4. Provide information to health care professionals based on both the results and interpretation of the results.</p>	<p>4.1 Effectively communicates results and analyses and implications for the patient and/or family to clinical staff who are providing genetic counselling for the family.</p> <p>4.2 Provides clear written reports on the results of the analyses.</p> <p>4.3 Uses recognised scientific nomenclature according to the latest international recommendations.</p>
<p>5. Participate in clinical research and introduction of new methods.</p>	

## 5. Cytogeneticist

- **Professional background**

The clinical cytogeneticist is a qualified laboratory specialist who is trained in the cultivation of cells for both prenatal and postnatal chromosomal analyses, processing of cells, methods of classical and molecular cytogenetic methods and karyotyping. He/she is able to interpret results and their clinical consequences in collaboration with clinical colleagues. In some cases the cytogeneticist will have a medical qualification.

- **Setting for practice**

Cytogenetics laboratory, publicly or privately funded, often linked to a clinical genetics service or academic institution of medical genetics.

Competence	Learning outcomes
1. Work independently in the cytogenetic laboratory.	1.1 Has an expert knowledge of the principles of meiosis, mitosis, chromosome segregation, chromosome inactivation and the causes of chromosomal aberration.  1.2 Demonstrates ability to manage workload effectively.  1.3 Appropriately prioritises ordered tests.  1.4 Understands safe codes of practice in the laboratory and adheres to these.  1.5 Supervises laboratory technicians.  1.6 Implements and follows up programs of internal and external quality assessment.  1.7 Ensures maintenance and calibration of laboratory equipment and devices is performed.
2. Work proficiently in the cultivation of cells for both prenatal and postnatal chromosomal examination, processing of cells, preparation of slides and karyotyping.	2.1 Deals with human lymphocytes, amniocytes or other human cells for cytogenetic examination.  2.2 Performs analyses by the classical cytogenetic methods or other methods including molecular cytogenetic methods.  2.3 Calculates and estimates recurrent risk of the disease in the family according to the results of laboratory investigations.  2.4 Utilises cytogenetic analysis software and computer programs appropriately.  2.5 Keeps accurate and contemporaneous records of performed analyses.
3. Interpret the	3.1 Is familiar with current scientific literature to inform

<p>results of cytogenetic and molecular cytogenetic findings.</p>	<p>interpretation of genetic test results.</p> <p>3.2 Calculates and estimates recurrent risk of the disease in the family according to the results of laboratory investigations.</p> <p>3.3 Participates in clinical interpretation of the analyses.</p> <p>3.4 Formulates advice for follow-up investigations for attending clinicians.</p>
<p>4. Provide information to health care professionals based on both the results and interpretation of the results.</p>	<p>4.1 Effectively communicates results and analyses and implications for the patient and/or family to relevant health professionals.</p> <p>4.2 Uses recognised scientific nomenclature according to the latest international recommendations.</p> <p>4.3 Provides clear written reports on the results of the analyses.</p>
<p>5. Participate in clinical research and in the introduction of new methods.</p>	

## 6. Biochemist geneticist/ biomedical scientist

- **Professional background**

The biochemical geneticist is a qualified laboratory specialist who is trained in providing a special service for the laboratory diagnosis and monitoring tests for example, for patients with inborn errors of metabolism or disorders of the clotting mechanism.

- **Setting for practice**

Biochemical laboratory, often within a healthcare institution, may be privately or publicly funded.

<b>Competence</b>	<b>Some proposed learning outcomes</b>
1. Work independently in the biochemical laboratory.	1.1 Has expert knowledge of pathological and biochemical changes, clinical symptoms, investigation and management of metabolic disease.  1.2 Demonstrates ability to manage workload effectively.  1.2 Appropriately prioritises ordered tests.  1.3 Understands safe codes of practice in the laboratory and adheres to these.  1.4 Implements and follows up programs of internal and external quality assessment.  1.5 Ensures maintenance and calibration of laboratory equipment and devices is performed.
2. Carry out biochemical testing related to diagnosis and management of genetic disease and carrier states, especially related to inborn errors of metabolism.	2.1 Performs a range of specific biochemical tests for the purpose of diagnosis of inborn errors of metabolism.  2.2 Adheres to national and other professional guidelines for biochemical testing of samples.  2.3 Controls and supervises the work of laboratory technicians.  2.4 Keeps records of performed analyses.
3. Interpret the results of biochemical tests.	3.1 Understands principles of inheritance, determination of risk of occurrence or recurrence of a disease or specific condition.  3.2 Is familiar with current scientific literature to inform interpretation of genetic test results.  3.3 Evaluates test results accurately. 3.3 Participates in clinical interpretation of the analyses

	with staff providing clinical care.
4. Provide information to health care professionals based on both the results and interpretation of the results.	4.1 Effectively communicates results and analyses and implications for the patient and/or family to relevant health professionals. 4.2 Makes recommendations for follow-up investigations.
5. Participate in clinical research and in the introduction of new methods.	

Prepared in conjunction with the Society for the Study of Inborn Errors of Metabolism (SSIEM) and the UK Haemophilia Centre Doctors Organisation Genetics Working Group.

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### ***Acknowledgements:***

*The authors wish to thank all members of the Unit 6 Experts Group who contributed at workshops in Porto, Portugal in 2006 and Milan, Italy in 2007. We are also grateful for all comments received from members of the European Society for Human Genetics and the Eurogentest project teams.*