



# **Developing European Quality Standards for Patient Information on Genetic Testing**

**Interim Report for Eurogentest Project Unit 6.1**

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## **Chapter 1: Report Overview, Findings and Recommendations**

**‘Eurogentest will develop the necessary infrastructure, tools, resources, guidelines and procedures that will structure, harmonize and improve the overall quality of all EU genetic services – molecular, cytogenetic, biochemical and clinical’.** Jean-Jaques Cassiman, coordinator of Eurogentest Network of Excellence<sup>1</sup>.

### **Report Overview**

This report contains the findings of work done during the first eighteen months of Unit 6, work package 1 of the Eurogentest project. The focus of the work package relates to the current state of written patient information concerning genetic testing, across Europe. The main objective of the first eighteen months was to assess the current availability and quality of written information given to patients and families considering genetic testing. Where there are gaps in availability, or where information quality is poor, recommendations will be made to bridge these gaps and raise quality standards. This report will be of use to genetic healthcare professionals and health information providers. It will also be relevant to patient support organisations and policy makers.

### **Key Objectives**

1. To map the range of publicly available information for patients and the public, across Europe, relating to the provision of genetic testing.
2. To undertake the systematic evaluation of this selected information, and to identify common (cross EU) elements, and those which are specific to individual member states.

### **Key Findings and Outcomes**

- The organisation of genetic services varies across Europe with services reflecting the differing size and structure of the population served and resources available.
- Genetic services tend to have either grown out of other clinical departments, been set up in collaboration with academic departments, or run as independent enterprises. A number have been set up as part of multidisciplinary regional centres.
- Regulations relating to the provision of genetic services differ across Europe. Legal frameworks exist in those countries where genetics is well established.
- A number of genetic centres in different countries (Belgium, the Netherlands, UK) work as multidisciplinary teams consisting of geneticists, genetic counsellors, genetic nurses, psychologists and social workers.
- The availability of written patient information varies considerably across Europe, with those countries with better resources and well developed service networks being more active in developing patient information. Most material collected was from the UK, followed by the Netherlands; the least amount was collected from Poland.
- Most of the material collected related to hereditary breast cancer, the most prevalent of the five conditions focused on. The least amount of material gathered related to tuberous sclerosis, one of the rarer conditions of the five.

Very little information was collected for the 22q11 deletion even though this is one of the more common chromosome disorders. This highlights that information availability does not necessarily reflect need.

- Most of the materials collected came in the form of personal letters. This was followed by booklets and leaflets, and lastly standard letters, of which only a small number were collected. The majority of booklets and leaflets had been developed by patient organisations, and only a small number had been developed by genetic clinics themselves. The greatest number of booklets and leaflets were collected from the UK.
- A selection of information was taken, translated where necessary, and assessed for the inclusion or omission of fourteen key issues considered necessary to make informed decisions about genetic testing. In total four pieces discussed all fourteen key issues. All four of these related to hereditary breast cancer, and all four were pre-written leaflets or booklets.
- The quality of written patient information varied across conditions. Information on the more prevalent genetic conditions (i.e. hereditary breast cancer) was found to be of a higher quality, and discussed a greater number of the key issues, than information on rarer conditions.
- Overall, there was very little discussion in the written material concerning psychological and social aspects of genetic testing.
- Under half of the material collected discussed both the potential benefits and harms of genetic testing, considered to be an important aspect in making informed decisions about genetic testing. Material far more frequently discussed the benefits than the harms.

## **Recommendations**

### **1. Informed Consent**

All patients have the right to make decisions in their healthcare that relate to their own personal beliefs. Therefore any genetic testing should be subject to the patients own free and informed consent. Healthcare professionals have a duty to provide information that is unbiased and well informed so that it allows patients and families to make decisions that are right for them.

### **2. Healthcare Provider Education**

With genetic testing frequently provided by specialist outside of the genetic clinic, it is important that these specialists are adequately trained in knowing when genetic testing might be appropriate, the use of and access to genetic tests, counselling techniques, genetics and genomics. Genetics should be considered a core component in the training of all healthcare specialists.

### **3. Psychological and Social Information**

Very little information (outside of hereditary breast cancer) was available for patients and families relating to the psychological and social repercussions of receiving a diagnosis. Service providers need to develop strategies to either provide this information themselves, or signpost patients and families to where they can receive this information. More research needs to be done to establish what the psychological and social repercussions are for rare genetic disorders.

### **4. Provision of Written Information**

Whilst informative and accessible oral information is fundamental to the delivery of good genetic communication, it is equally important that patients and families are provided with good quality written information, at a time that is appropriate and useful to them. Practitioners have a duty to ensure that written information achieves the

minimum standard set by key stakeholders. Prewritten leaflets related to genetic conditions are a useful resource and should be available in genetic clinics and provided alongside personalised information. This information should be developed with the help of patients, families and professionals to insure the needs of patients and families are met. Information, given in both the written form and orally, should be available in the patient's own language.

## **5. Public Education**

Generic information relating to genetics, genetic tests, inheritance patterns and risk should be widely available to the public through government, hospitals, genetic department, support organisations and other appropriate public information sources. Governments and the media have a role to play in educating the public so that misunderstandings and misrepresentations of genetics is avoided. Greater public awareness can support informed decision making, understanding and appropriate use of genetic services.

## **6. Patient Support Organisations**

Patients and families should be given information about existing patient support groups relevant to them from the outset, to contact as and when they wish. Information relating to genetic conditions and genetic tests should be readily available to patients and families through these organisations.

## **Recommendations for Eurogentest Project**

A number of the gaps identified above can be addressed directly by Unit 6.1. Over the next three and a half years we hope to achieve the following:

1. Develop and refine key elements for written patient information. This will be an extension of the work undertaken during the assessment stage. Included in this work will be identification of:

- The key pieces of information that should be provided to patients
- The way in which this information is presented to maximise accessibility and utility

This work will be undertaken with the help of patient groups and healthcare professionals from a number of different European states.

2. Develop generic information leaflets for patients and families. These will cover key issues related to genetics including; the basic biological function of genes, chromosomes etc; inheritance patterns and risk; information about the various types of genetic tests available and their potential benefits, limitations and risks. This information will be developed with the help of professionals, patients and families to ensure that it is accessible, informative and discusses those issues that are important to all concerned. Where appropriate, this project will utilise existing information that has already been developed.

3. A 'Frequently Asked and Useful Questions' leaflet will also be developed to support patients and families that are going to speak to a healthcare professional about genetic testing for the first time. The list of relevant questions will be devised with the input of patients and families that have experienced the genetic testing process.

4. This information will be translated into a number of European languages where we have found there to be significant gaps at present. We are in the process of surveying professionals across 27 member states to find out where these gaps

currently exist. Translated information will be reviewed by both professionals and the public to ensure that the translation quality is high.

5. This information will then be disseminated, both in print and on-line, through genetic clinics, other relevant hospital departments (e.g. paediatrics, maternity, general practice), government, patient support groups and other appropriate public information sources. The information will also be available on the Eurogentest website ([www.eurogentest.org](http://www.eurogentest.org)).

6. There needs to be a commitment to update and maintain the information developed, otherwise it will soon become outdated. It is essential to secure further investment for this after the current Eurogentest project has been completed.

7. The research of the 18 months highlighted that significant gaps exist in service provision related to genetic testing in a range of European countries, in particular the lack of attention paid to discussion of the psychosocial impact on patients and families of genetic testing for the rare inherited conditions. Consensus around which health or social care professionals should provide this information is needed and discussion will take place on this specific issue with both patient groups and health professionals from across Europe.

8. WP 6.1 will work with WP 6.2 to identify the minimum set of skills required by any health professional who provides genetic counselling in the context of genetic testing. The elements of this set of skills will be discussed by both patient groups and professionals.

## Chapter 2 - Introduction

Across Europe great developments in genetics have taken place in recent years. Accompanied by these developments have been increasing demands for diagnoses, therapies, treatments, genetic services and public awareness. Yet these advancements have also been accompanied by high levels of technical errors and poor reporting due to a lack of structuring and synchronization at the European level<sup>2</sup>. With healthcare systems being nationally organised, and not falling under the responsibility of the EU, potential barriers may exist in harmonizing the quality of services. Coupled with this is the lack of a common arrangement across Europe to deliver quality services to consumers both at present and in the future. Varying standards of quality accompanied by lack of reference systems and differing Member State regulations, have added to the overall disorganization and fragmentation of services<sup>2</sup>.

### Project Objectives<sup>3</sup>

- The **EuroGentest NoE** aims at developing the necessary infrastructure, tools, resources, guidelines and procedures leading to the establishment of harmonized, quality genetic testing services in Europe, which can interact with, stand as a model for, or help to achieve similar services on other continents.
- This will be achieved by bringing together, in a real long-term partnership, experts and expert centers available in Europe engaged on different aspects of testing, including researchers, small and medium enterprises (SMEs), testing laboratories, quality management and public health experts, ethicists, lawyers, sociologists, educational authorities and patient groups.

The tools used to achieve these goals will be:

- 1) the promotion of research, proper utilization, quality assurance and control, and adequate management of genetic tests,
- 2) the creation of quality information resources that can be readily accessed, ultimately facilitating the emergence of new tests (e.g. for rare diseases for which tests are not available) and the improvement of existing ones (higher accessibility and improvement of genotyping techniques in terms of lower costs, higher efficiency and specificity),
- 3) agreement on best practice guidelines and protocols, strengthening, unification and organization of quality assessment schemes and accreditation of laboratories to achieve harmonization and high quality of the services offered across the EU,
- 4) establishing a knowledge and technology platform to facilitate impact assessment and information flow, and
- 5) joining resources in the area of rare disease services, where uniform patient access and quality standards are even more limited by the low number of affected individuals.

- The **EuroGentest NoE** will focus on the quality of genetic services, measured against existing European and international or developing quality norms for laboratory bench tests in the cytogenetics, molecular and biochemical genetics sectors. In addition the network will consider the quality of informatic systems, which are or will be available to stakeholders including researchers, medical and non-medical clinical and laboratory personnel, consumers, authorities and legislators.
- In addition, ethical, legal, intellectual property and societal issues related to these services will be evaluated and recommendations for their appropriate management will be formulated.

- A network of Health Technology Assessment units, from a number of different member states (MS) will evaluate the impact of genetic testing services on public health policies and economics. Recommendations to address potential problems will be drafted.
- An assessment of educational material relevant to genetic testing will be performed if necessary new educational materials will be developed and made available to key member state representatives.
- Through interactions with SMEs a network will be created which should allow the appropriate, quality controlled and focused development of new technologies for testing.

### **Work Package 6.1**

In order to achieve the project objectives, the project has been split up into 6 Units. Unit 6 is concerned with 'Education and Communication' for both patients (WP6.1) and professionals (WP6.2).

The main objectives for the first 18 months of WP6.1 are as follows:

- 1. To map the range of publicly available information for patients and the public relating to the provision of genetic testing.**
- 2. To assess the quality of a selection of information.**

These objectives will be achieved in a number of different stages:

**Chapter 3** will provide an overview of the current state of genetic services across Europe. This will provide a context for the work done during the first eighteen months.

**Chapter 4** will tackle the first main objective, the mapping of a range of publicly available information. This will be achieved through the collection of information materials from genetic services in an initial group of seven member states, chosen because of their diverse healthcare systems, and differing levels of development within genetic services.

**Chapter 5** will begin addressing the second objective, through the review of current approaches to assessing material related to genetic testing. The next stage will then be to develop either a new or existing strategy to assess written material.

**Chapter 6** will address the last stage of the second objective whereby we use the developed tool to assess a representative range of material from across the seven states.

**Chapter 7** will provide a general discussion of the findings.

### **The Role of Written Information within Patient Education**

It is integral to the delivery of good quality healthcare that patients are provided with information that is accurate, accessible and well informed. This is especially true in an age when patients are taking greater interest than ever before in managing their health<sup>4</sup>. Whilst great progress has been made on the research front within the field of genetics, there has been recent concern regarding the quality of information patients receive, especially in the written form.<sup>5,6,7,8</sup> Written information is an essential part of the genetic counselling process. Used alongside verbal communication, it has been shown to improve patient satisfaction<sup>9</sup> and their knowledge<sup>10</sup> of what are often complex and sensitive issues. Written information also has a role in allaying patient anxiety<sup>11</sup> and other negative psychological impacts of the encounter.<sup>12</sup> Leaflets and other material can therefore play an important part in supplementing and reinforcing information provided by clinicians. However it is not only through contact with clinicians that patients can obtain written information. Studies indicate that patients want as much information as possible<sup>13</sup>, and the internet provides an easy access route to such information. In the European Union an average of 23% of the population use the internet to find information about health issues<sup>14</sup>.

There is however growing concern about the quality of internet based information, with much of it inaccurate and biased<sup>15</sup>. Coupled with this is the fact that the majority of information available on-line is in English. This therefore makes it difficult for those whose first language is not English, and the information is inaccessible to those who do not speak any English. As genetics is an emerging field within a healthcare, genetic literacy amongst the general population is relatively low<sup>16</sup> making it imperative that oral information is supported by written material. In order that written information serves its purpose in aiding the decision making process, it is essential that it is available in the reader's first language, and covers a spectrum of issues that patients and families believe to be important. Not only must it do so in an accurate, user friendly and educational manner, it must also be informed by the very persons that have and will use the information. It is therefore vital that patients' views are taken into account when developing written information so that it provides information which reflects what patients need and want. Through assessing the quality and availability of information currently available to patients and families, we will gain a greater understanding of whether it does actually achieve this.

### **Chapter 3: Genetic Services in Europe**

It was decided during the initial stages of the project that we would consider 'genetic testing' to include those tests usually provided by a genetic department, i.e. carrier tests, diagnostic tests, predictive tests and prenatal tests. These tests were considered most relevant within the context of the Eurogentest project.

Seven countries were focused on that were considered collectively to be representative of the current state of genetic services in Europe. The seven were: the UK, Belgium, Sweden, the Netherlands, Italy, Germany and Poland. These seven have very different healthcare systems and their genetic services are at varying stages of development. It was also feasible to focus on seven in the time available.

#### **An Overview of the Organisation of Genetic Services across Europe**

The aim of genetic services is to respond to the needs of patients and families who are threatened by genetic disease, particularly their wish to know whether or not they are at risk of developing or transmitting a disorder with a genetic component. A primary responsibility of genetic counselling is to provide information, as accurately as possible, on diagnosis and chance of recurrence within the family. Genetic services may comprise of multidisciplinary groups of healthcare professionals in the clinical setting, and at the community level should include education for the general public through patient organisations and government organisations<sup>17</sup>.

Regulations on the provision of genetic services vary across Europe, due to organisational differences within healthcare services, and because genetic services are all at varying stages of development. In a comparative study of 31 countries<sup>18</sup> it was found that in European countries where genetics is well established, legal frameworks exist within which the services operate. On an international level, in 2001 the European Society of Human genetics recommended a formal recognition of medical genetics as a medical speciality in Europe in order 'to aid the provision and development of genetic services for individuals and families in Europe'. WHO also put forward recommendations to help develop and strengthen medical genetics services, and establish educational programs for teaching healthcare professionals<sup>19</sup>.

On a national level numerous regulations, guidelines, reports, statements and directives exist. Belgium developed legalisation in 1987 to restrict genetic counselling and diagnostic testing to genetic centres located in university hospitals<sup>20</sup>. In Germany, professional organisations such as The German Bundestag and The German Society of Human Genetics have issued comments and guidelines regarding the application of genetic testing in relation to issues such as postnatal predictive testing<sup>21</sup>, preimplantation genetic diagnosis<sup>22</sup> and BRCA 1 testing<sup>23</sup>. In Italy, National Guidelines for Genetic testing were prepared by a Task Force in 1998 to ensure the safety and effectiveness of both existing and newly introduced genetic tests, and to define criteria for quality assurance of laboratories performing genetic tests<sup>24</sup>. The Swedish Society for Medical Genetics issued guidelines for clinical genetic units for molecular routines as well as for genetic counselling and this document has since been adopted by all university clinical genetic departments in Sweden as a minimum standard for quality<sup>25</sup>. In the Netherlands regulatory frameworks exist for the licensing of clinical genetics centres, and for testing and counselling for prenatal and postnatal chromosome, biochemical, and DNA testing<sup>26</sup>. In the UK a number of committees have been set up to advise UK Health ministers on developments in genetic testing. These include the Advisory Committee on Genetic Testing (which has now been incorporated into the Human Genetics Commission), the House of Commons Select Committee on Science and Technology (which advises on any number of issues, one

of which is genetics), and the Genetics Research Advisory Group. These committees have looked at and advised on numerous issues relating to genetic testing, including genetic testing for late onset disorders<sup>27</sup>, the current state of clinical genetics services<sup>28</sup> and human genetic testing services supplied direct to the public<sup>29</sup>. These organisations tend to have specific remits, and may only be in existence for a limited amount of time. The UK Genetic Testing Network (UKGTN) and Genetics Commissioning Advisory Group (GenCAG) also act as commissioning mechanisms for England. The UKGTN advises on the clinical validity and reliability of new genetic tests, and GenCAG advises on the commissioning of service development.

The organisation of genetic services across Europe varies. Some services have been set up in collaboration with university departments or have evolved out of other hospital departments; some are organised as multidisciplinary regional centres, others are private clinics offering genetic testing and counselling. The locations of genetic centres vary in each country depending on the local population served or facilities and resources available. In Italy for instance, the North has a greater number of clinical genetic centres than the South (67 in the North, compared to only 13 in the South<sup>30</sup>). This is due to variations in funding and capabilities across each region, as well as perceived local 'need'. This has however resulted in regions in the South being unable to cope with demand, and patients seeking diagnosis or treatment might choose to travel to regions outside of where they live.

For some of the rarer diseases national centres have been established where counselling and genetic testing takes place, (e.g. connexin 26 alteration testing in Tübingen, Germany). Specialist laboratories may also receive samples from abroad. The number of genetic centres that offer both counselling and testing varies considerably across Europe. Of the seven countries specifically looked at, the following information was recorded. In the UK there are 24 regional genetic centres offering testing and counselling<sup>31</sup>, in the Netherlands 8<sup>32</sup>, in Sweden 6<sup>33</sup>, in Belgium 8<sup>34</sup> in Germany 97 (including private clinics)<sup>35</sup> and in Poland 21<sup>36</sup>. At least 88 genetic centres exist in Italy<sup>37</sup>, 16% of which are private.

In theory everyone should have access to genetic services. However in practice there are obstacles that can hamper access, the most serious obstacle being the lack of professional education in genetics by primary and secondary care physicians. This leads to patients being misdiagnosed and not referred on appropriately. In addition, it is believed that higher levels of education and urban living are all linked to better access to information and services. Those that live in rural areas, those with a low social economic status and immigrants or minority groups who do not speak the national language are less likely to access genetic services<sup>38</sup>. Another serious problem is that the workload of clinical genetic services has increased dramatically over recent years, and it is not always possible for resources and staffing to keep up with demand. Patients and families often have to wait months to see a specialist, and even longer to receive their test results. Governments will have to increase the amount of money allocated to genetic services, expand the capabilities of genetic laboratories, increase the intake of medical graduates into genetics, and train other healthcare specialists in genetics if they are to keep up with this increasing workload.

Genetic services are generally paid for by the public healthcare system, health insurance or other means used in the country concerned. If patients want tests that might be considered 'extra services', such as genetic tests for conditions where there appears to be no increased risk to the patient (e.g. no family history of the condition the patient wants to be tested for), services may be available through private healthcare systems at the patients own expense (as is the case in Germany, Poland and Italy).

Traditionally, genetic counselling was only performed by clinical geneticists as it was considered part of the clinical assessment. In more recent years however, an American system has been adopted in a number of European countries (such as in the UK, the Netherlands, Belgium and Sweden) enabling genetic nurses and genetic counsellors with master's degrees to provide genetic counselling. These healthcare professionals are considered integral members of a multidisciplinary genetic team and are trained to collect and confirm medical and family histories, perform risk assessment, offer patient education regarding genetics, provide supportive counselling and in some cases organise genetic tests. Genetic services are also frequently provided through a number of different healthcare departments including paediatrics, neurology, obstetrics and general practice. Face to face interviews in each of the seven member states indicated that patients may be referred to genetic services at various stages of the genetic testing process. They may be referred before genetic testing is performed, or when a diagnosis has been made in order to discuss the condition in further detail and discuss issues relating to heredity and risk.

Clinical genetic services provide a number of different types of genetic tests including pre and post natal diagnostic and carrier testing, and predictive testing. Preimplantation genetic diagnosis is another procedure available, although this is fairly costly and may be subject to legislative constraints (such as in Germany) hence is not routinely offered in genetic centres across Europe. Clinical genetic services may also have a role to play in providing education and screening for conditions that are considered 'highly prevalent' for certain population groups (e.g. thalassaemia screening in Mediterranean populations, Tay-Sachs screening in Ashkenazi Jewish populations). Yet medical services for ethnic minority groups are targets for improvement. In the UK an inquiry found marked regional inequalities of access to genetic services for beta thalassaemia major, a disease predominantly of ethnic minority groups<sup>39</sup>. The Genetic Interest Group also investigated this issue. In a pilot study of the efficacy of ethnic monitoring in clinical genetics, results showed a significant under representation of ethnic minorities in cancer referrals<sup>40</sup>.

Throughout Europe various attitudes and beliefs exist concerning human reproduction and genetic testing. It is commonly agreed that this diversity must be respected, and that genetic testing must only be done on a voluntary basis.

Partnerships with patient organisations are particularly well organised in a number of European countries, with patient organisations providing education and support to patients and families. In a number of countries umbrella organisations exist (The Genetic Interest Group in the UK, VSOP in the Netherlands, Belgium Rare Disorders, Sallsynta in Sweden). These organisations provide a platform for patients' views and are therefore an important means of informing policy, and cooperation between them and genetic centres have resulted in good information tools being developed.

## **Chapter 4: To Map the Range of Publicly Available Information for Patients and Public Relating to the Provision of Genetic Testing**

Due to the number of countries in Europe, and the practical limitations of the project, it was not feasible to collect data from every European country. Seven countries were identified with an aim to achieving maximum variation in terms of geographical location, health and political systems, and genetic service development. The seven were: the United Kingdom (UK), Belgium, Sweden, the Netherlands, Italy, Germany and Poland. Across these seven countries information was gathered concerning genetic testing for five conditions. These were; hereditary breast cancer, Duchenne muscular dystrophy, tuberous sclerosis, 22q11 deletion, and the connexin 26 alteration. This panel of conditions were chosen because they cover both the rare and more common genetic conditions. Hereditary breast cancer is one of the most prevalent and commonly tested for of the five, 1 in 9 women will develop it in their lifetime and 5-10% of these are thought to be caused by a genetic predisposition<sup>41</sup>. Duchenne muscular dystrophy is another frequently tested for condition across Europe (appendix Tables 4 & 5), with the condition affecting 1 in every 3500 male births<sup>42</sup>. Tuberous sclerosis is a much rarer genetic condition, thought to only affect 1 in 7000 people. Each condition is thought to be equally prevalent across the selected states. In addition the conditions cover a broad range of hereditary patterns (i.e. adult onset, autosomal dominant, autosomal recessive, sex linked, and chromosomal disorder).

### **Method**

- 1) A questionnaire was developed and sent out to genetic departments across the selected seven member states.
- 2) Service providers were identified through a network of key informants in each country. A letter was sent to each service provider asking them to participate in the study. Face to face interviews were organised with two or three service providers in each state who responded positively to the invitation.

The questionnaire and face to face interview was designed to gather information about four key issues which were:

- whether patients are provided with written material in general
- whether patients are provided with written material relating to the five conditions
- whether translated material exists
- whether patients are provided with details of relevant support organisations

Written information was also collected in a number of formats from the same service providers.

### **Results**

- 1) Response rates to the questionnaire were as follows:

UK	11/23 (48%)	Belgium	3/8 (38%)
The Netherlands	5/8 (63%)	Poland	4/24 (17%)
Italy	6/20 (30%)	Germany	11/54 (20%)
Sweden	3/6 (50%)	TOTAL	= 43

2) In the questionnaire we asked genetic services about the provision of written information in the context of genetic testing. 98% of respondents said that they generally do provide written information. When we asked specifically about written information relating to the five conditions, again professionals said they provided written information routinely:

- hereditary breast cancer: 88%
- Duchenne muscular dystrophy: 88%
- 22q11 deletion: 83%
- about tuberous sclerosis: 83%
- connexion 26 alteration: 82%

3) Relating to the provision of translated material, under a third of respondents (30%) said that this existed. In fact we were told no translated material was available in Sweden, Belgium or Germany. Translated material was most frequently available from the Netherlands.

4) Over two thirds of respondents (74%) said that they provided patients and families with information about relevant support organisations. The UK and Germany were the only countries who did not always provide patients and families with information about support organisations.

(appendix Figure 7 & Table 3 for a copy of the questionnaire questions and results)

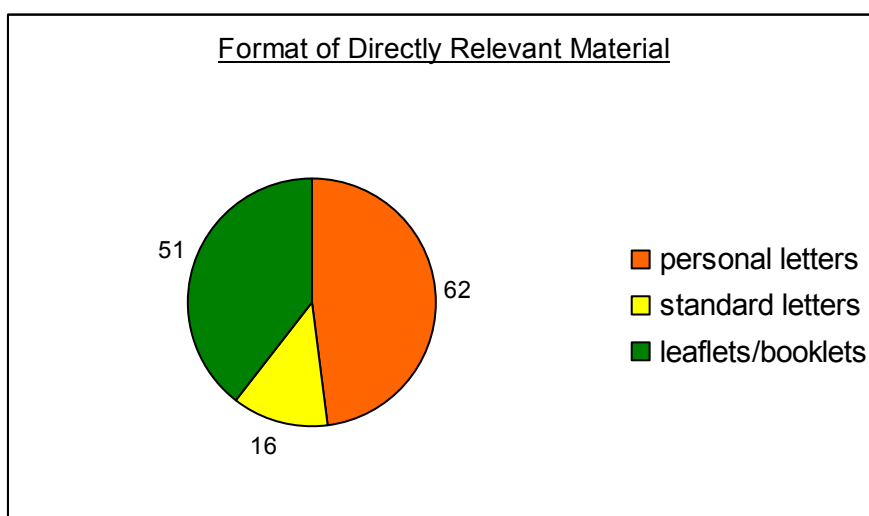
5) In total **194** pieces of written material were collected between March 2005 and March 2006. These can be grouped into two main categories:

**A)** Information directly related to the 5 conditions – **129** pieces. A third of this material had not been produced by the genetic clinic, but was either available from it, or patients could be directed to it.

**B)** Information of a more general nature – **65** pieces (e.g. a booklet about prenatal testing, a general leaflet about genetic counselling, a booklet explaining chromosome disorders).

6) The directly relevant material came in the following formats:

**Figure1**

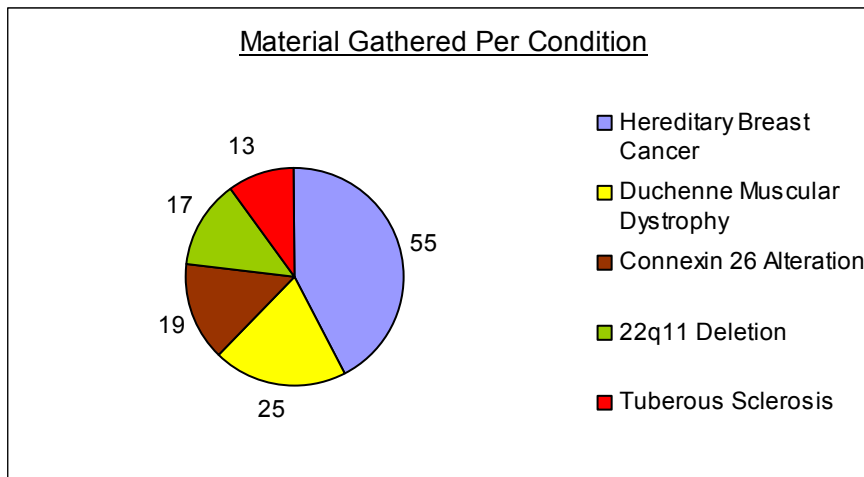


Material was either collected directly from the genetic clinic, or we were directed to a website, which patients would also be directed towards, by the clinician. Nearly half the material gathered (48%) came in the form of a personal letter. 40% of material

came in the form of a leaflet or booklet. The majority of these had been developed by patient organisations. Only a small number of these had been developed by the clinic itself, and these mostly related to hereditary breast cancer. A small percentage of material (12%) came in the form of a standard letter. These related to the more common conditions, hereditary breast cancer and Duchenne muscular dystrophy.

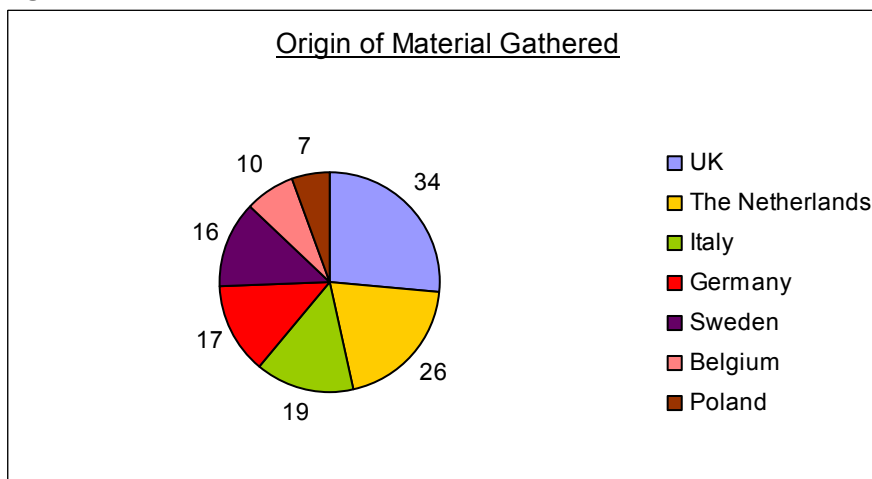
7) Most of the material gathered related to hereditary breast cancer. This was then followed by Duchenne muscular dystrophy, the connexin 26 alteration, 22q11 deletion and lastly tuberous sclerosis.

**Figure 2**



8) The majority of material was collected from the UK. Almost two thirds of this had been produced outside of the genetic clinic but was either available from it, or patients could be directed to it. The least amount of material was collected from Poland.

**Figure 3**



**Discussion**

- The results imply that the most common method of communication between physician and patient is through personalised letters. There were however a large number of leaflets and booklets developed by patient organisations that

patients and families could pick up directly at the clinic, or would be directed to. Only a few genetic clinics (in the UK, the Netherlands, Germany and Belgium) had developed their own leaflets and booklets, and these mainly covered hereditary breast cancer, prenatal testing, and general information about heredity and risk. The UK was the only country in which pre-written leaflets were available for all five conditions focused on. Guy's and St Thomas' genetic department in London have developed a whole series of leaflets<sup>43</sup> on a variety of genetic conditions, and these have been adopted by numerous genetic clinics across the UK.

- The questionnaire results imply that written information is routinely provided to patients and families across the seven member states. However when we went on to gather examples of this written information, we found it hard to gather information for each condition, from every country. In Poland and Belgium we were unable to gather written material on tuberous sclerosis, and in the Netherlands and Sweden we were unable to gather material on the connexin 26 alteration.
- There was a large amount of variation across the seven countries concerning how much information we collected. We were either given or directed towards nearly five times the amount in the UK as we were in Poland. This finding highlights that perhaps there is less material readily available for patients and families in Eastern Europe, as there is in Western Europe.
- Prevalence rates for the five conditions are similar across the seven countries focused on<sup>44</sup>. Therefore it is not surprising that there was more material gathered relating to hereditary breast cancer than for the other four conditions. This condition is the most prevalent of the five, (1 in 9 women will develop it in their lifetime and 5-10% are thought to be caused by a genetic predisposition)<sup>45</sup>. It is also one of the most frequently tested for conditions (appendix Table 4). A number of high profile cancer charities also exist throughout Europe which provide written information on genetics and cancer.
- Duchenne muscular dystrophy is another condition that is frequently tested for across Europe. It is the fourth most frequently performed prenatal test in the UK<sup>46</sup> and the second most frequent performed prenatal test in Italy<sup>47</sup> (appendix Tables 4 & 5). It is not surprising therefore that the second greatest amount of material collected related to this condition.
- The least amount of material gathered related to tuberous sclerosis. This is also one of the rarest of the five conditions (occurrence is 1 in 7000<sup>48</sup>). Discussion with geneticists highlighted that it is a condition that is often diagnosed symptomatically because the genetic test is both complex and expensive. In addition the condition is not often tested for prenatally because it is highly variable.
- 22q11 deletion is one of the more common chromosome conditions (1 in 3000-4000 births<sup>49</sup>) The fact that very little information was collected for it is therefore surprising, perhaps reflecting that the availability of patient information does not necessarily reflect need.
- Both the face to face interviews and the literature search highlight that genetic counselling and testing are not exclusively provided through genetic clinics. Often other clinicians such as paediatricians, oncologists, neurologists and

general practitioners perform these services. It is important therefore that key standards for written patient information be disseminated amongst these healthcare professionals. It would also be useful if pre-written leaflets about genetic conditions, and more general information related to genetics, was available from these departments.

## **Strengths and Limitations**

### **Strengths**

- The seven countries chosen provide maximum variation in terms of geographical location, health and political systems, and genetic service development.
- The conditions chosen cover a broad range of both rare and more common genetic conditions, a broad range of hereditary patterns, and a range of conditions thought to be equally prevalent across the selected states.

### **Limitations**

- The findings present a crude overview of the availability of written patient information across Europe as we could only look at a limited number of countries and conditions.
- Due to time and resource constraints, it was not possible to visit every genetic clinic in each of the seven countries. There may have been variations within each country that were not picked up.
- Conclusions can only be drawn from material collected. The quality and availability of material may differ considerably from clinic to clinic. In addition, diagnoses are often given in other departments which were not visited.
- In Italy and Germany we could only survey a proportion of genetic clinics because of the high number in existence (there are numerous private clinics). This could have potentially caused gaps in our method as there may have been variation between centres that were not picked up.

## **Chapter 5: Development of Evaluation Tool**

### **Introduction**

We crudely mapped the availability of written material that related to genetic testing for five conditions, in seven European countries. The following section of this report discusses the second objective of the project, the assessment of a selection of this material.

The second objective was performed in three stages:

1. Through a review of current approaches that exist to assess and write material related to genetic testing. This was done through internet and bibliographic database searches (e.g. Google, Ovid, Medline) and through discussion with researchers and geneticists working in patient education.
2. Through the assessment of these existing strategies to choose either the most appropriate, or as the basis to develop a new one.
3. This strategy was then used to assess a representative range of patient information across the seven selected states.

### **1. Review of Current Approaches to Assessing and Writing Material**

#### **Method**

Extensive searches of the bibliographic databases (Medline, Ovid, CINAHL) and search engines (Google) were conducted, using search terms including: 'health information evaluation', 'health evaluation tools', 'judge genetic information', 'assessing genetic information' 'genetic information development' 'develop genetic information' to find any existing evaluation tools, in the public domain, related to genetic testing. A number of genetic educationalists were also contacted at centres including the National Genetics Education and Development Centre in Birmingham and Oxford Department of Public Health.

#### **Results**

Fourteen different information assessment strategies were located from Europe and the USA. The majority of these had been developed to judge or help develop patient information in general, and very few related specifically to genetics or genetic testing. However four tools were identified that were created specifically to assess or help develop patient information relating to genetics. These were:

**-'DISCERN Genetics: A Validated Appraisal Tool for Judging the Quality of Information for the Public on Genetic Testing and Screening.'** Shepperd S, Farndon P, Grainge V, Oliver S, Parker M, Perera R, Bedford H, Elliman D, Kent A, Rose P in European Journal of Human Genetics, July 2006

<http://www.nature.com/doifinder/10.1038/sj.ejhg.5201701>

<http://www.discern-genetics.org/>

DISCERN Genetics is a tool that guides the production and appraisal of information resources produced for the public on genetic testing. It consists of 19 questions plus an overall quality rating. The questions relate to information resources, information on the condition, the test procedure and results, decision making, and the reliability of the information. It was developed with the help of information producers, providers and members of self-help groups (not restricted to the UK) in order to help ensure that the criteria chosen was both 'user friendly' and reflected the concern of patients.

**-Erfocentrum Manual for Writing Patient Information for the Website.**

Developed by Mies Wits-Douw at the Erfocentrum centre in The Netherlands (Vredehofstraat 31, 3761 HA Soestdijk, The Netherlands). Erfocentrum is dedicated to providing public information about genetic conditions. The descriptions on the website include information about causes, prognosis, treatment and heredity. The website was launched in 1999 and at present contains over 750 items.

<http://www.erfelijkheid.nl/>

The text also contains links and references that point to networks, patient organisation, treatments and other information. Erfocentrum have also developed a site providing information about genetics and genetic conditions that is specifically for children (<http://www.bogi.nl/>) as well as one for couples who are considering starting a family and want to know about their genetic risk (<http://www.zwangernu.nl/>).

### **'Education Material About Genetic Tests; Does it Provide Key Information for Patients and Practitioners?'**

M.K. Cho, M. Arruda and N.A. Holtzman, in *American Journal of Medical Genetics* 73:314-320 (1997) An appraisal tool that was developed to analyse pamphlets containing information about genetic testing. The tool has a list of 10 critical elements, recommended by a number of policy groups, which are considered necessary to evaluate the appropriateness and performance of the tests. These included issues that relate to the condition being tested for, the test itself, risks, limitations and benefits.

### **-'Your Child Has a Genetic Disorder, What do you Need to Know and From Whom?'**

Produced by the Genetic Interest Group, Angelman Syndrome Support Group and Cri Du Chat Syndrome Support Group in 2000.

A pamphlet that provides a list of useful questions that patients and families may want to ask during a genetic counselling session. Questions relate to the condition itself, the accuracy of the diagnosis, and further services and support networks available.

## **2. Assessment of Existing Strategies**

### **Method**

Each tool presented a number of key issues as being of importance when developing or assessing material relating to genetic testing. Each key issue was identified, given a suitable title, and tabulated. Care was taken so that there should be no overlap. We identified 14 key issues in total, 11 of these were taken from the 19 item DISCERN Genetics questionnaire. For the most part, titles were taken directly from the Discern tool as it was the most comprehensive of the four. Each key issue was correlated against each tool. This table appears below.

## Results

### KEY

Discern = Discern tool

Erfocentrum = Erfocentrum tool

AJMG = American Journal of Medical Genetics tool

GIG = Genetic Interest Group tool

**Table 1**

Correlation of Key Issues across the Four Tools

KEY THEME	Discern	Erfocentrum	AJMG	GIG
Background and Effect of Condition	/	/	/	/
Treatment and Management	/	/	/	/
Heredity and Risk	/	/	/	/
Patient Rights	/		/	
Type of Test	/	/	/	/
Test Procedure	/	/	/	
Accuracy of Test	/		/	/
What Happens after the Test	/			
Shared Decision Making	/			
Psychosocial Consequences	/		/	/
Consequences for Relatives and Partner	/		/	/
Benefits and Risks	/		/	
Date and Sources	/	/		
Additional Support and Information	/	/		/

Apart from two issues ('what happens after the test' and 'shared decision making'), each issue had been raised by two or more of the tools. This showed that there was a high agreement rate amongst the tools concerning the key issues surrounding genetic testing.

A table of issues was created with a title of the issue and a supporting description of each issue. Each description provides examples of the way in which the issue might be presented to the reader. Again, because many statements referred to the same issue, albeit in a slightly different way, care was taken to avoid overlap. However because each key issue had a fairly wide remit, it was also important that a variety of possible descriptions were used in order to ensure as full an explanation as possible. Again, because the Discern tool was the most comprehensive of the four tools, many of the descriptions used were taken directly from this tool. However, we did identify a number of additional descriptions from other sources.

Table 2

**Criteria from DISCERN Genetics<sup>50</sup>**  
 (Copyright University of Oxford 2005) [www.discern-genetics.org](http://www.discern-genetics.org)

**Additional Descriptions (from AJMG<sup>51</sup> GIG<sup>52</sup>,  
 Erfocentrum<sup>53</sup> and independent research<sup>55</sup>)**

<b>Background and Effect of Condition</b>	Symptoms, occurrence. A description between being a carrier and having the condition.	Cause (at genetic, chromosome, cellular, organ level <sup>53</sup> ), development <sup>53</sup> , characteristics <sup>52</sup> , prognosis <sup>52</sup> , diagnosis <sup>52,53</sup> . Any other name the condition might be known as <sup>53</sup> .
<b>Treatment and Management</b>	How condition is treated and how well treatment works. Procedure for referral to specialists.	Strategies for prevention <sup>51</sup> . Whether there is a cure <sup>51</sup> . Other medical management options <sup>52,51</sup>
<b>Heredity and Risk (modified from the original question 'Is risk explained in simple terms?')</b>	A reason as to why the reader might be at specific risk. The risk of developing, carrying or passing on the condition. Risk of developing the condition with the faulty gene compared with the risk if one does not have the faulty gene. Chance that the condition will not develop.	Why the reader might be appropriate for testing <sup>51</sup> . Reproductive risk <sup>52,51</sup> . Risk estimation without genetic testing <sup>54</sup> .
<b>Test Procedure</b>	Safety/risk of procedure. If it hurts.	Where and how it is performed <sup>55</sup> . Whether it has to be paid for privately or if it is free <sup>51</sup> .
<b>Accuracy of Test</b>	An acknowledgement of any limitations of testing such as an explanation of how tests fail due to either human or laboratory error. An explanation of false positive and false negative test result. Any evidence of local variations in lab result. An explanation that a repeat test may be needed and why.	Specificity <sup>51</sup> . An explanation of false positive and false negative test result <sup>51</sup> .
<b>What Happens after the Test</b>	Follow up procedures. Who gives results and how they are received.	Where the sample is sent <sup>55</sup> . Waiting time for results <sup>55</sup> .
<b>Shared Decision Making</b>	Things to discuss with family, friends, and health professionals.	
<b>Psychosocial Consequences.</b>	The various emotional and social consequences that might be experienced, both positive and negative (such as relief or increased anxiety). That a range of emotions are possible and normal. Possible discrimination arising from test results e.g.	Whether any benefits can be claimed <sup>52</sup> .

	employment and insurance (taken from the Discern question 'Are issues of discrimination discussed?')	
<b>Consequences for Relatives and Partner</b>	What an increased risk means to person being tested and their family. That different people have different reactions. That misattributed paternity may be discovered.	That the family may experience a range of emotional consequences <sup>55</sup>
<b>Date and Sources</b>	Name and date of publication and any revision.	References of experts or organisations quoted, a reference to a current guideline on which the information is based <sup>55</sup> .
<b>Additional Support and Information</b>	Whether any geographical differences in service provision are outlined e.g. test availability (taken from the Discern question 'Is information provided on local availability of services and test performance?')	Such as details of local services, support organisations other sources of information, other relevant health professionals <sup>52</sup> .

**Additional Criteria (taken from AMJG, GIG, Erfocentrum, March of Dimes<sup>56</sup> and Independent Research)**

<b>Type of Test</b> <sup>51</sup>	The type of test i.e. carrier, diagnostic, prenatal, predictive, newborn screening <sup>51</sup> .
<b>Patient Rights</b> <sup>55</sup>	That testing is voluntary, about the need for informed consent, that results are confidential, that the patient can specify to whom results can be disclosed <sup>51</sup>
<b>Benefits and Risks</b> <sup>56</sup>	Whether the information presents statements that discuss both the risks/limitations of genetic testing, as well as the benefits. <u>Benefits:</u> e.g. may lead to (early) diagnosis, may lead to disease prevention or treatment, can help guide reproductive decision-making, may provide psychological benefits, may be able to claim societal benefits after confirmation of a diagnosis <sup>56</sup> <u>Risks and Limitations:</u> e.g. may cause emotional and psychological difficulties, such as anger, guilt, anxiety, strained relationships etc, may put one in a position where difficult decisions have to be made, may identify other 'at risk' family members without their consent, may cause discrimination in insurance, expense, may not be able to reach a diagnosis, there may be no intervention or treatment available, may not be able to provide exact risk assessment, negative test results may not guarantee patient will not develop condition <sup>56</sup> .

The tool was then checked by applying it to a small number of leaflets/letters to see that it encompassed all the various issues discussed.

### **Strengths and Limitations**

There are a number of obvious benefits and limitations of using a tool such as this to assess the quality of written information. These include:

#### **Strengths**

- It provides us with a useful and valid means of assessing the quality of information as it allows us to check material against a number of important issues that have been identified by key stakeholders.
- The results provide useful insights into which issues are repeatedly being omitted within written information, and whether the quality of information is consistent across both conditions and countries.

#### **Limitations**

- It cannot tell us quite how detailed each piece of information is. Two pieces of information might score equally even though one piece is far more detailed than the other.
- Even though we are able to assess content, we cannot assess the accuracy of each statement, nor the style or readability of it.

## Chapter 6: Assessment of Selected Information

### Method

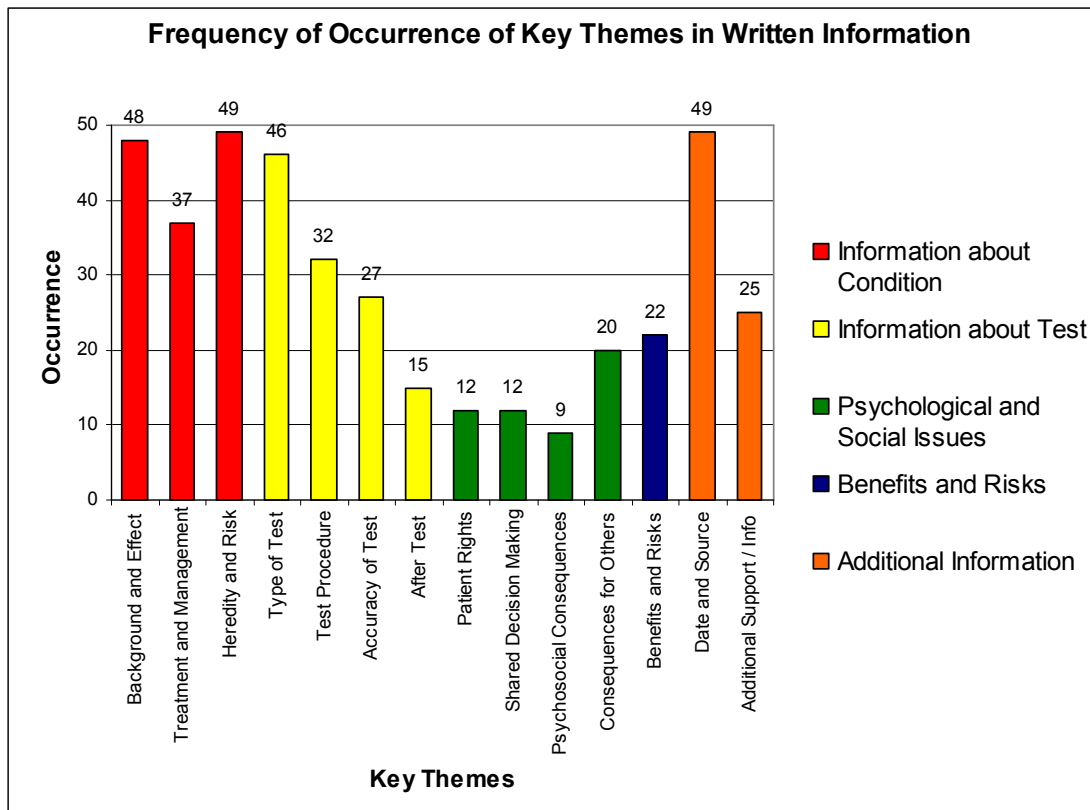
The last stage of the second objective was to assess a representative range of patient information across the seven selected states. When the material was collected, those items that were extremely brief were excluded from the assessment. Where possible two pieces of written material for each condition, from each country, were randomly chosen and those that were not in English were translated by a professional translation service experienced in working with health related materials. Where we were unable to collect two pieces, we assessed only one. Unfortunately in a few cases, we were unable to collect material for a particular condition, from a particular country.

Fifty piece of information were assessed in total. Of these fifty pieces twenty five were personal letters, two were standard letters, and twenty three were pre-written leaflets or booklets (appendix Table 15 for full breakdown of leaflets and letters per condition). Each piece was assessed for the presence or absence of each of the fourteen key issues. A statement fitting any part of the description was counted as a presence of the key issue, and the results were tabulated. In addition to scoring content of the written material, the level of detail and type of language used were taken into account.

### Results

For full breakdown of results see appendix Tables 6 to 12.

**Figure 4**



Whilst the majority of material discussed issues relating to the condition, including background and effect (n=48, 96%), treatment and management (n=37, 74%)

heredity and risk (n=49, 98%) and the type of test (n=46, 92%), less than half discussed key issues such as what happens after the test (n=15, 30%), shared decision making (n=12, 24%) patient rights (n=12, 24%) and a discussion of the benefits as well as potential risks (n=22, 44%). Benefits of genetic testing were more likely to be included (n=41, 82%) than any risks involved (n=24, 48%) The area that was covered by the least number of pieces of material was the possible psychological and social effects of genetic testing (n=9, 18%).

### Letters versus Leaflets

Figure 5

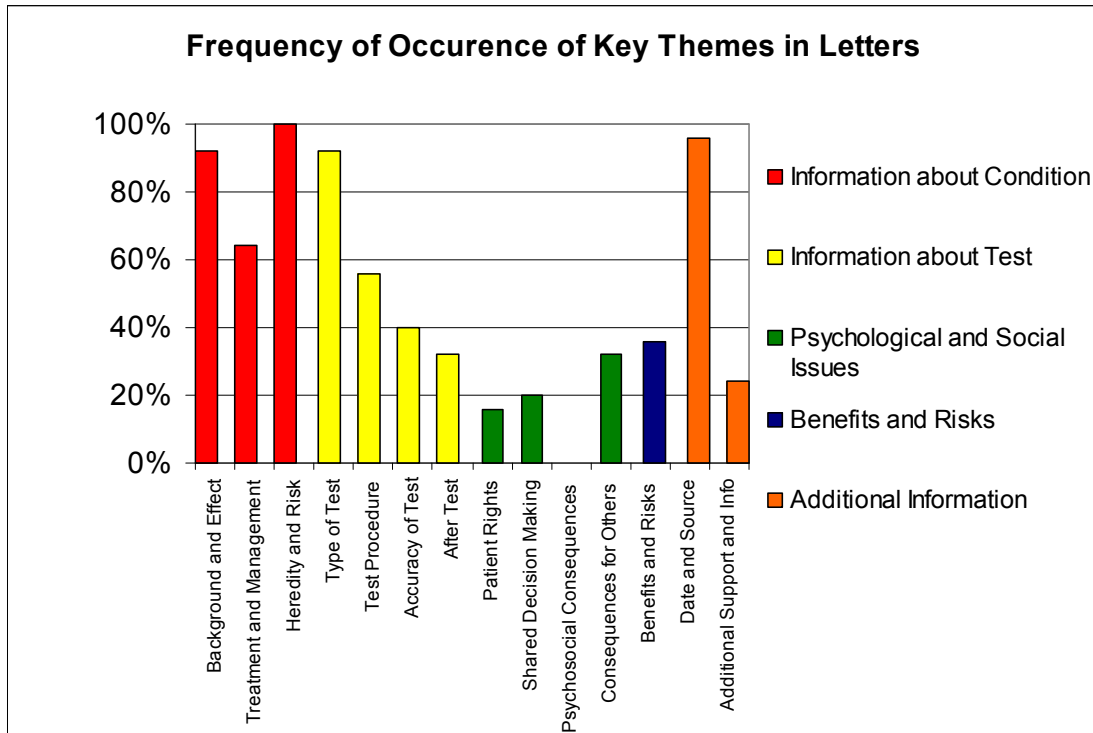
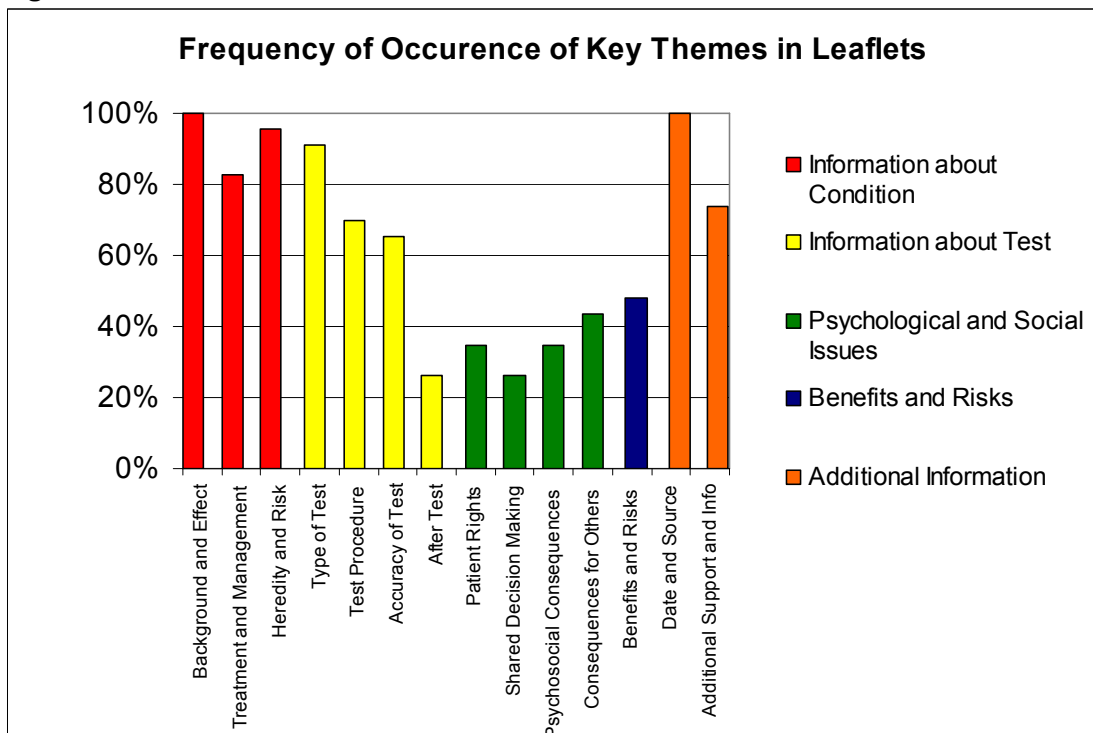


Figure 6



When the content of personal letters (Figure 5) was compared with the content of pre-written leaflets (Figure 6) it was found that a greater number of key topics were discussed in the leaflets. In particular, pre-written leaflets tended to include more information about the accuracy of the test and information related to patient rights. Pre-written leaflets were also more likely to discuss both the benefits and risks of genetic testing. There was no discussion of the possible psychosocial effects of genetic testing in any of the personal letters assessed. Only a small number of personal letters provided information about additional support organisations and information services.

### **Psychological and Social Issues**

Psychological and social issues were more readily discussed in information relating to hereditary breast cancer than the other four conditions. Of the twelve pieces that discussed patient rights, eight of them related to breast cancer. Of the twelve pieces that discussed shared decision making, seven of them related to breast cancer. Of the nine pieces that discussed psychosocial consequences, eight of them related to breast cancer. In fact of the four pieces that scored on every key issue, all of them related to hereditary breast cancer, and all were either in leaflet or brochure form, (two were developed by genetic departments, and two were developed by patient organisations).

### **Patient Rights**

Informed choice and non-directiveness are the cornerstones of good practice in genetic counselling. It is only through providing patients and families with relevant information about the condition and the test, in a way that is informative, accessible, and done without coercion, that patients and families can make decisions that are right for them<sup>57,58</sup>. The frequency of discussion around 'patient rights', i.e. the discussion of issues including that testing is voluntary, and that decisions should be consistent with the decision-maker's values, varied considerably across the five conditions. It was discussed in 67% of the material about hereditary breast cancer, in 17% of the material about the connexion 26 alteration, and in only 8% of the information about Duchenne muscular dystrophy. When the issue was discussed, it was done far more overtly in certain cases than in others. For example in one booklet assessed, the issue that testing is voluntary was made very explicit:

*'This is your decision to make and can be very difficult. Your genetic counsellor can talk through all the issues with you, and other people can help you to deal with the emotional issues that may arise'* (UK breast cancer booklet, CancerBACUP).

In another piece the fact that testing is voluntary was implied far more implicitly:

*'If the mutation is found, a predictive test can be carried out on Mrs X and other family members, to see whether they have inherited the mutation'* (German letter about hereditary breast cancer).

### **Benefits and Risks**

Of those pieces that did discuss both a benefit and a risk of genetic testing, the majority (10/22) related to hereditary breast cancer. The following Belgian leaflet is one example of a leaflet that presents both a benefit and a risk of genetic testing:

*'Women that are a carrier of an abnormality in the BRCA1 or BRCA2 gene and therefore have a strongly increased risk of developing breast and/or ovarian cancer must be kept under very close medical observation. An early discovery increases the chances of recovery [benefit].....The decision to have a predictive test carried out can have large consequences and be very emotional [risk]. It is very important to have people who ask to have these test carried out, treated and supported by a*

*multidisciplinary team. This team must pay special attention to all the decisions etc that a possible confrontation with hereditary breast cancer can involve.'* (KU Leuven genetic department)

Frequently cited benefits included that genetic testing could lead to a diagnosis, disease prevention or treatment. Frequently discussed risks and limitations were that testing could be very emotional, may be a time consuming process, and that not all the mutations that cause the condition are known.

### **Conditions**

The quality of written patient information varied across conditions. Information on the more prevalent genetic conditions (i.e. hereditary breast cancer) was found to be of a higher quality (i.e. discussed a greater number of key issues), than information on the rarer conditions. Information on hereditary breast cancer discussed key issues on average in 81% of the material. Information on the connexion 26 alteration discussed the key issues least frequently; on average in 44% of material. Some of the most notable findings were that none of the material on the connexion 26 alteration, 22q11 deletion or tuberous sclerosis discussed psychosocial consequences, tuberous sclerosis only discussed both the benefits and risks of genetic testing in 13% of the material collected, and none of the material on the connexion 26 alteration discussed where to access additional support and information from (Table 13).

### **Countries**

Overall, material from the UK discussed the key issues most frequently, and information from Belgium and Italy least frequently. On the whole the frequency of discussion for each of the key themes was the same across the seven countries, apart from in a few particular cases which include the following: Treatment and management was discussed in more than half the material from every country apart from Poland where it was discussed in only 40% the material collected. Test accuracy was discussed in all the information from the UK but in less than half the information from the Netherlands, Belgium, Sweden and Italy. Shared decision making was discussed in 60% of material from Germany, but in none of the information from Belgium, Italy and Poland. Benefits and risks were discussed in 75% of material from the UK, but in only 14% of material from Sweden, and additional support and information was discussed in all the material from the Netherlands but in none of the information from Italy (Table 14). There is no obvious explanation as to why these results might have occurred. It would nevertheless be interesting to explore this further.

### **Risk**

Some interesting stylistic variations were found when assessing the material, especially in the discussion concerning risk as the following two examples show:

*'It has been observed that women carrying BRCA1 mutations have increased risk of developing breast and ovarian cancer with respect to other women from the population at large.....This assertion is not the same as saying that carriers of a hereditary BRCA1 mutation are sick, or destined to become sick: considering one's entire lifespan a large proportion of the women carrying these mutations will not develop breast tumours....From available research data, it may be said that out of 100 women with the BRCA1 mutation, 15-40 will not develop breast tumours within their expected lifetime (70 years) and that 50-70 women will not develop breast tumours prior to the age of 50. (Italian letter, hereditary breast cancer)*

*'Presence of the mutation in the BRCA1 and/or BRCA2 gene indicates a significant predisposition to breast and ovarian cancer.....Women up to 70 years old with a mutation in these genes have a more that 80% risk of falling ill with breast cancer,*

*and 40% risk of developing an ovarian tumour.'* (Polish leaflet about hereditary breast cancer, Amazonki association)

The first piece, translated from Italian, explains risk in terms of the chance the condition will not develop. The Polish piece on the other hand, discusses risk in terms of the chance the condition will develop. Presenting a negative risk as opposed to a positive risk might significantly alter the way in which the reader interprets their risk, as might presenting a twenty year risk estimate (which the Italian letter does) in addition to a lifetime estimate<sup>59</sup>. Furthermore, the tones within the two pieces are quite different. The Italian letter comes across as quite reassuring and friendly, whereas the language in the Polish leaflet is more scientific ('significant predisposition'), matter of fact, and hence potentially alarming.

The level of detail provided within the material varied tremendously, especially around issues relating to the test. Whilst a few pieces went into great detail about the test procedure, others merely mentioned that a blood sample would be taken, and a 'genetic examination' performed. This can be seen from the two examples below:

*'In about 25% of patients the 22q11 deletion is visible when the chromosomes are looked at under the microscope. However, in about 75% of patients the deletion is submicroscopic, which means that although you cannot see the missing material under a microscope, you can prove that the piece is missing by using a special DNA test called FISH (fluorescent in situ hybridisation). If someone has a 22q11 deletion then when FISH studies are carried out rather than seeing two fluorescent signals (one on each chromosome 22q11 region) only one fluorescent signal is seen.'* (22q11 leaflet, North West Thames Regional Genetics Service, UK)

*'If there is a suspicion of the diagnosis [22q11 deletion] then a genetic examination is needed to confirm or exclude the diagnosis. This requires a tube of blood from the child. The genetic examination can demonstrate whether there is a micro deletion on chromosome 22.'* (Dutch 22q11 booklet, Federatie van Ouderverenigenen)

The amount of information provided to patients and families varied tremendously with information ranging from half an A4 side to a small booklet fifty pages long.

## **Discussion**

Overall, every piece discussed some of the issues considered important when discussing genetic testing. Information was however far more likely to discuss hard, factual information related to the condition and the test, than the more qualitative, experience based information related to the psychological and social implications of genetic testing.

### **Letters versus Leaflets**

The reason that personal letters were found to be less comprehensive than pre-written leaflets is likely to stem from a number of causes. The first is the severe shortage of resources and time experienced by genetic services. Writing personal letters is a time consuming process and clinicians most likely only to have time to write about those issues they consider most important. Secondly, pre-written leaflets can be assessed by patients and professionals during the development stage to ensure they cover all the key issues. Thirdly, pre-written leaflets are often prepared by patient groups and are hence patient driven. They are therefore more likely to tackle the issues important to patients and families.

### **Hereditary Breast Cancer**

Information that related to hereditary breast cancer scored far better during the assessment than information concerning the other four conditions; in particular they

discussed far more frequently those issues related to psychological and social consequences of genetic testing. There are various reasons as to why these leaflets might have scored so highly. First, there has been much research done on the information needs of hereditary breast cancer patients due to its high prevalence in the European population<sup>60,61,62,63</sup>. A number of genetic clinics were found to have developed pre-written leaflets due to the high number of people counselled about this condition<sup>64</sup>. In addition a number of high profile cancer charities are key players in developing patient information and consulting with patients during the development stages. Hence the information they produce is likely to reflect the issues that are of concern to patients and families.

### **Psychological and Social Issues**

Psychological and social issues were not readily discussed outside of information related to hereditary breast cancer. Whilst much research has been concentrated on cancer because it is such a major health issue, there is also a body of research relating to the social and psychological problems experienced when receiving a diagnosis for the rarer genetic conditions<sup>65,66,67,68</sup>. Testing positive as a carrier may result in feelings of guilt or anger<sup>69,70</sup>. Positive prenatal test results will undoubtedly cause a plethora of mixed emotions, with the added difficulty of having to make extremely tough decisions quickly. Genetic testing might also inadvertently disclose information about relatives or parentage, or cause complications when buying health and life insurance. A number of studies have also been conducted on the psychosocial effects of testing specifically for the rarer conditions looked at in this study including Duchenne muscular dystrophy<sup>71,72</sup>, 22q11 deletion<sup>73</sup>, tuberous sclerosis<sup>74</sup> and deafness<sup>75</sup>. With the lack of psychosocial information provided in written material concerning these conditions, it appears that much of this research has not yet been translated into practice.

Clinicians may also not believe it to be within their remit to provide information about social and psychological issues. They might not have access to, or keep up to date with 'non-medical' or social issues relating to genetic testing such as insurance, benefits, and specialist education services. These issues might be considered within the remit of patient organisations or social services. Yet the International Society of Nurses in Genetics (ISONG) states that one of the roles of genetic nurses is to 'provide genetic information and psychosocial support to individuals and families'<sup>76</sup>. The National Coalition for Health Professional Education in Genetics states that each healthcare professional should at a minimum be able to 'understand the social and psychological implications of genetic services'<sup>77</sup>. And the Fit for Practice in the Genetics Era document states that the competent practitioner should 'demonstrate a knowledge and understanding of the utility and limitations of genetic testing and information, including the potential physical and/or psychosocial consequences of genetic information for individuals, family members, and communities'<sup>78</sup>. Hence it is in fact one of the required competences of genetic professionals to be able to provide information about psychological and social issues.

### **Benefits and Risks**

Less than half of the material discussed both a benefit and a risk of genetic testing. Whilst many leaflets and letters discussed at least one potential benefit of genetic testing, information was far less forthcoming about the possible risks and limitations. There are a number of reasons as to why this might be the case. First, the general trend relating to patient information has always been to improve healthcare through early diagnosis and treatment, hence practitioners might be more inclined to present the benefits as opposed to the potential risks<sup>79</sup>. Secondly, there might be a desire not to 'worry' patients through the discussion of potential risks. Lastly, it may be argued that informed choice is a relatively new phenomenon<sup>80,81</sup> and therefore written information may not yet routinely reflect this trend. Yet the benefits, limitations and

risks of genetic testing are well documented. Benefits include providing certainty for patients<sup>82</sup>, providing information for relatives<sup>83</sup> and informing clinical management. Risks and limitations are cited as including mental distress<sup>84</sup>, inappropriate reassurance if negative<sup>85</sup>, and the fact that interventions may not be available<sup>86</sup>.

### **Additional Support and Information**

Only half the information assessed discussed where to obtain additional information, and how to contact support services and patient groups. This might be because additional information (such as complementary leaflets or patient information websites) may not exist in the patient's own language, or the clinician might be unaware of it. Alternatively the clinician may feel that he or she have given the patient and family all the information and support that is necessary. In addition it may be unclear exactly whose role it is to provide the patient and family with additional support, and hence the clinician might not signpost the patient and/or family towards any. There might also not be an additional support service or patient organisation in existence for the family to be referred to.

### **Conclusion**

The results of the assessment provide some interesting insights into the current quality of written patient information; in particular it highlights those issues that are being repeatedly omitted by information providers. The key findings are as follows: First, pre-written leaflets tend to provide a more comprehensive discussion of the issues surrounding genetic testing than personal letters do. Secondly, written information often fails to portray a balanced discussion of genetic testing, with far more discussion dedicated to the benefits than the potential harms. Thirdly, written information often fails to discuss the psychological and social aspects of genetic testing, especially in information outside of hereditary breast cancer.

Other studies have been carried out to evaluate existing patient information, with similar findings. A recent study looking at patient information on newborn bloodspot screening found that the majority of leaflets supported the public health agenda by informing parents of the benefits of screening, including the significance of early detection and treatment for these conditions. Few leaflets however supported the informed choice agenda by mentioning either the limitations of screening or choice<sup>79</sup>. Another study looking at educational material for patients and practitioners about genetic testing concluded that most materials did not contain basic information about the test itself, such as the purpose or accuracy of the test, as well as information about the risks and benefits<sup>87</sup>.

There has however been increasing pressure in recent years to improve the quality of written patient information. We live in an increasingly legalistic climate where clinicians are under increasing pressure to ensure realistic expectations of medical procedures and treatments. Patients and families take more interest in their healthcare than ever before, thus there is increasing pressure on clinicians and other information sources to provide patients with increasingly detailed and informative information. With these factors in mind, we are hopefully moving towards a climate in which good quality information becomes more necessary, and more readily available.

### **Strengths and Limitations**

#### **Strengths**

- A study comparing written information from across Europe for a range of genetic conditions had not been done before this one, making this research unique in its field.

- Every effort was made to ensure that language limitations were kept to a minimum by using a professional translation service with experience translating health information.

### **Limitations**

- Due to limitations of time and money, only a certain amount of information gathered could be translated and assessed. The information translated was selected at random. This might have given a slight advantage to information that was already in English. We are also unable to comment on the quality of information that was not translated.
- Information was translated by various translators. The quality of the translation might therefore not be uniform. We did however point out to all translators the key issues we were looking for within the material.
- Because any statement fitting any part of the description of the key themes was counted as a presence of that theme, the results do not indicate how complete or accurate the information actually is.
- We cannot determine the total content of information provided to patients by practitioners because there will be much information that is given orally. Therefore issues that are lacking in written information might have been discussed with the patient during the consultation. However it is important to remember that much oral information will be forgotten, and patients and families cannot be expected to retain what is often complicated and upsetting information, given under stressful circumstances. In addition, by only providing information orally, there is a risk that other relatives will be misinformed when information is passed on.

## **Chapter 7: General Discussion**

Communication around those issues that are of concern to patients and families is a fundamental part of good genetic service delivery. The development and provision of written information is integral to this process, as long as it is accurate, informative, and provided at an appropriate time.

The findings from this report however suggest that there are gaps across both conditions and countries, in the availability of good quality written information. This is especially the case in those countries where genetic services are less well developed. Information, especially for the rarer genetic conditions, often fails to portray a balanced view of genetic testing, and the potential psychological and social consequences are infrequently discussed.

The pre-written booklets and leaflets assessed during this study were generally more comprehensive than the personal letters in that they discussed a greater number of key issues relevant to genetic testing. Yet pre-written leaflets were not routinely available in genetic clinics across Europe. In addition, only a handful of generic information leaflets were available from genetic clinics. The development of pre-written materials, available through both the genetic clinic and the public domain, would be a good first step in ensuring that good quality written information is readily available to all. Clinicians, health authorities and patient support groups have a role to play in making sure that this target is met.

With this in mind, a number of recommendations that relate to the findings from this report have been developed. Their implementation would go some way to improving the overall standard of patient information across the EU.

### **Recommendations**

#### **1. Informed Consent**

All patients have the right to make decisions in their healthcare that relate to their own personal beliefs. Therefore any genetic testing should be subject to the patients own free and informed consent. Healthcare professionals have a duty to provide information that is unbiased and well informed so that it allows patients and families to make decisions that are right for them.

#### **2. Healthcare Provider Education**

With genetic testing frequently provided by specialist outside of the genetic clinic, it is important that these specialists are adequately trained in knowing when genetic testing might be appropriate, the use of and access to genetic tests, counselling techniques, genetics and genomics. Genetics should be considered a core component in the training of all healthcare specialists.

#### **3. Psychological and Social Information**

Very little information (outside of hereditary breast cancer) was available for patients and families relating to the psychological and social repercussions of receiving a diagnosis. Service providers need to develop strategies to either provide this information themselves, or signpost patients and families to where they can receive this information. More research needs to be done to establish what the psychological and social repercussions are for rare genetic disorders.

#### **4. Provision of Written Information**

Whilst informative and accessible oral information is fundamental to the delivery of good genetic communication, it is equally important that patients and families are provided with good quality written information, at a time that is appropriate and useful to them. Practitioners have a duty to ensure that written information achieves the minimum standard set by key stakeholders. Prewritten leaflets related to genetic conditions are a useful resource and should be available in genetic clinics and provided alongside personalised information. This information should be developed with the help of patients, families and professionals to ensure the needs of patients and families are met. Information, given in both the written form and orally, should be available in the patient's own language.

## **5. Public Education**

Generic information relating to genetics, genetic tests, inheritance patterns and risk should be widely available to the public through government, hospitals, genetic department, support organisations and other appropriate public information sources. Governments and the media have a role to play in educating the public so that misunderstandings and misrepresentations of genetics is avoided. Greater public awareness can support informed decision making, understanding and appropriate use of genetic services.

## **6. Patient Support Organisations**

Patients and families should be given information about existing patient support groups relevant to them from the outset, to contact as and when they wish. Information relating to genetic conditions and genetic tests should be readily available to patients and families through these organisations.

## **Recommendations for Eurogentest Project**

A number of the gaps identified above can be addressed directly by Unit 6.1. Over the next three and a half years we hope to achieve the following:

1. Develop and refine key elements for written patient information. This will be an extension of the work undertaken during the assessment stage. Included in this work will be identification of:

- The key pieces of information that should be provided to patients
- The way in which this information is presented to maximise accessibility and utility

This work will be undertaken with the help of patient groups and healthcare professionals from a number of different European states.

2. Develop generic information leaflets for patients and families. These will cover key issues related to genetics including; the basic biological function of genes, chromosomes etc; inheritance patterns and risk; information about the various types of genetic tests available and their potential benefits, limitations and risks. This information will be developed with the help of professionals, patients and families to ensure that it is accessible, informative and discusses those issues that are important to all concerned. Where appropriate, this project will utilise existing information that has already been developed.

3. A 'Frequently Asked and Useful Questions' leaflet will also be developed to support patients and families that are going to speak to a healthcare professional about genetic testing for the first time. The list of relevant questions will be devised with the input of patients and families that have experienced the genetic testing process.

4. This information will be translated into a number of European languages where we have found there to be significant gaps at present. We are in the process of surveying professionals across 27 member states to find out where these gaps currently exist. Translated information will be reviewed by both professionals and the public to ensure that the translation quality is high.

5. This information will then be disseminated, both in print and on-line, through genetic clinics, other relevant hospital departments (e.g. paediatrics, maternity, general practice), government, patient support groups and other appropriate public information sources. The information will also be available on the Eurogentest website ([www.eurogentest.org](http://www.eurogentest.org)).

6. There needs to be a commitment to update and maintain the information developed, otherwise it will soon become outdated. It is essential to secure further investment for this after the current Eurogentest project has been completed.

7. The research of the 18 months highlighted that significant gaps exist in service provision related to genetic testing in a range of European countries, in particular the lack of attention paid to discussion of the psychosocial impact on patients and families of genetic testing for the rare inherited conditions. Consensus around which health or social care professionals should provide this information is needed and discussion will take place on this specific issue with both patient groups and health professionals from across Europe.

8. WP 6.1 will work with WP 6.2 to identify the minimum set of skills required by any health professional who provides genetic counselling in the context of genetic testing. The elements of this set of skills will be discussed by both patient groups and professionals.

### **Similar Recommendations**

The work proposed for Unit 6.1 of the Eurogentest project will also be tackling a number of recommendations that were proposed by the European Commission's Independent Expert Group in 2004. In the recent '25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing', it was proposed that:

'materials and resources be developed and made available at the EU, national, and local level to provide information about genetic testing, genetic screening, and pharmacogenetics through a variety of media'; (recommendation 4a) and that

'genetic testing be accompanied by the provision of key information.....The provision of simple, printed information that can be consulted by the individual after counselling has been shown to be extremely valuable, so that such materials should always be available.' (recommendation 9).

## Appendix

### Figure 7

#### Questionnaire

- 1) Are patients provided with written information before the clinic appointment?
- 2) Are patients provided with written information prior to genetic testing?
- 3) Are patients provided with written information with the test results?
- 4) Are patients provided with written information at the end of the consultation episode?
- 5) Are translated materials available?
- 6) Are patients provided with details of relevant support groups?
- 7) Are patients provided with written information concerning genetic testing for hereditary breast cancer?
- 8) Are patients provided with written information concerning genetic testing for Duchenne muscular dystrophy?
- 9) Are patients provided with written information concerning genetic testing for 22q11 deletion?
- 10) Are patients provided with written information concerning genetic testing for tuberous sclerosis?
- 11) Are patients provided with written information concerning genetic testing for the connexion 26 alteration?

**Table 3**

#### Results of Questionnaire

	<u>UK</u>	<u>Netherlands</u>	<u>Italy</u>	<u>Sweden</u>	<u>Belgium</u>	<u>Germany</u>	<u>Poland</u>
<u>Q1</u>	11/11	5/5	1/6	1/3	0/3	4/11	1/4
<u>Q2</u>	10/11	2/5	3/6	2/3	1/3	8/11	2/4
<u>Q3</u>	10/11	4/5	5/6	2/3	3/3	9/11	4/4
<u>Q4</u>	11/11	5/5	6/6	2/3	3/3	11/11	4/4
<u>Q5</u>	6/11	4/5	2/6	0/3	0/3	0/11	1/4
<u>Q6</u>	7/11	5/5	6/6	3/3	3/3	4/11	4/4
<u>Q7</u>	10/10	5/5	2/2	3/3	1/3	6/8	1/1
<u>Q8</u>	10/10	5/5	5/5	2/3	1/3	8/10	4/4
<u>Q9</u>	10/10	4/5	3/3	2/3	1/3	6/8	4/4
<u>Q10</u>	9/10	5/5	3/3	1/2	1/3	6/8	4/4
<u>Q11</u>	10/11	4/5	5/5	1/2	1/3	6/8	4/4

**Table 4**

#### Frequency of Genetic Tests in the UK, 2004-2005

<u>Prenatal</u>		<u>Postnatal</u>	
Cystic Fibrosis	32%	Cystic Fibrosis	21%
Sickle Cell	21%	Fragile X	19%
b-globin	9%	BRCA	9%
Duchenne Muscular Dystrophy	8%	Misc	8%
ID/sexing/Maternal cell contamination	7%	Hemochromatosis	7%
Fragile X	4%	Factor 5 Leiden	4%
Abnormal haemaglobins	4%	Hereditary nonpolyposis	4%
Myatonic Dystrophy 1	3%	Spinocerebellar ataxia's	4%
Spinal muscular atrophy 1	3%	Thalassaemia	4%
Achondroplasia	3%	Prader-willi syndrome & Angelman syndrome	3%
Huntingtons disease	2%	Coagulation factor II	3%
Uniparental disomy 14	2%	Charcot-Marie-Tooth disease 1A	3%

Adrenal Hyperplasia	2%	Duchenne muscular dystrophy	2%
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Results supplied by the Clinical Molecular Genetics Society:

[http://www.cmgs.org/new\\_cmgs/Laboratory%20Information/Audit.htm](http://www.cmgs.org/new_cmgs/Laboratory%20Information/Audit.htm)

**Table 5**

Frequency of Genetic Tests in Italy, 2004

Prenatal		Postnatal	
Cystic Fibrosis	29%	Cystic Fibrosis	23%
Duchenne muscular dystrophy	18%	Factor 5 Leiden	25%
Connexion 26 alteration	18%	Factor 2	
Fragile X	12%	Thrombosis MTHFR	
Chromosome testing for numerical changes		HLA (for transplantation)	9%
Fraxe	5%	Leukaemia (non specific)	
Thalassaemia		Fragile x	3%
Uniparental disomy		Microdeletions Y chromosome	
Acondroplasia (dwarfism)		Hemacromatosis	
Spinal muscular atrophy		Connexin 26 alteration	1.8%

(note: BRCA1&2 data cannot be found above because this test is performed in oncology and not molecular genetics).

Censimento 2004 'Strutture di Genetica Medica in Italia', B Dallapiccola, I Torrente, A Morena, R Mingarelli.

**Table 6**

Key

I = letter

le = leaflet

sl = standard letter

BC = breast cancer

DMD = Duchenne muscular dystrophy

C26 = connexin 26 alteration

22q = 22q11 deletion

TS = tuberous sclerosis

Results of Evaluation of Material from Poland

	P1/BC/le	P2/DMD/I	P3/DMD/le	P4/C26/I	P5/22q11/I	
Background and Effect	1		1	1	1	4
Treatment and Management	1		1			2
Heredity and Risk	1	1	1	1	1	5
Patient Rights	1					1
Type of Test	1	1	1		1	4
Test Procedure	1	1	1		1	4
Accuracy of Test	1	1		1	1	4
After Test	1			1		2
Shared Decision Making						0
Psychosocial Consequences	1					1
Consequences for Others	1			1		2
Benefits and Risks	1					1
Date and Source	1	1	1	1		4
Additional Support and Info			1			1

**Table 7**  
Results of Evaluation of Material from Germany

	G1/BC/le	G4/BC/l	G5/22q11/l	G6/22q11/l	G7/TS/l	G8/TS/l	G9/DMD/l	G10/DMD/l	G11/C26/l	G12/C26/l	
Background and Effect	1	1	1	1	1	1	1	1	1	1	10
Treatment and Management	1	1	1		1	1	1			1	7
Heredity and Risk	1	1	1	1	1	1	1	1	1	1	10
Patient Rights	1	1			1				1		4
Type of Test	1	1	1	1	1	1	1	1	1	1	10
Test Procedure	1		1		1	1	1	1		1	7
Accuracy of Test	1		1			1		1	1		5
After Test	1	1									2
Shared Decision Making	1	1			1		1		1	1	6
Psychosocial Consequences	1										1
Consequences for Others	1	1	1	1							4
Benefits and Risks	1	1	1			1		1			5
Date and Source	1	1	1	1	1	1	1	1	1	1	10
Additional Support and Info	1	1		1		1	1				5

**Table 8**  
Results of Evaluation of Material from the UK

	UK1/BC/le	UK3/BC/le	UK4/DMD/le	5/DMD/sl	UK6/22q11/le	UK7/22q11/le	UK8/TS/le	UK9/C26/le	
Background and Effect	1	1	1	1	1	1	1	1	8
Treatment and Management	1	1	1	1	1	1	1		7
Heredity and Risk	1	1	1	1	1	1	1	1	8
Patient Rights	1	1	1						3

Type of Test	1	1	1	1	1	1	1	1	8
Test Procedure	1	1	1	1	1	1	1		7
Accuracy of Test	1	1	1	1	1	1	1	1	8
After Test	1	1		1					3
Shared Decision Making	1	1						1	3
Psychosocial Consequences	1	1							2
Consequences for Others	1	1		1	1				4
Benefits and Risks	1	1		1	1	1		1	6
Date and Source	1	1	1	1	1	1	1	1	8
Additional Support and Info	1			1	1			1	4

**Table 9**  
Results of Evaluation of Material from Belgium

	B1/BC/le	B2/BC/l	B3/DMD/l	B4/22q11/le	B5/C26/l	
Background and Effect	1		1	1	1	4
Treatment and Management	1	1	1			3
Heredity and Risk	1	1	1	1	1	5
Patient Rights	1					1
Type of Test	1		1	1	1	4
Test Procedure	1		1			2
Accuracy of Test	1			1		2
After Test			1			1
Shared Decision Making						0
Psychosocial Consequences	1					1
Consequences for Others	1		1			2
Benefits and Risks	1		1			2

Date and Source	1	1	1	1	1	5
Additional Support and Info	1			1		2

**Table 10**  
Results of Evaluation of Material from Italy

	I1/BC/I	I2/DMD/I	I3/DMD/I	I4/22q11/I	I5/22q11/I	I6/TS/le	I7/C26/I	
Background and Effect	1	1	1	1	1	1	1	7
Treatment and Management	1	1		1	1	1	1	6
Heredity and Risk	1	1	1	1	1	1	1	7
Patient Rights				1				1
Type of Test	1	1	1	1	1	1	1	7
Test Procedure	1		1			1		3
Accuracy of Test	1	1				1		3
After Test	1	1					1	3
Shared Decision Making								0
Psychosocial Consequences								0
Consequences for Others						1		1
Benefits and Risks	1	1	1					3
Date and Source	1	1	1	1	1	1	1	7
Additional Support and Info								0

**Table 11**  
Results of Evaluation of Material from Holland

	H1/BC/le	H2/BC/sI	H3/DMD/I	H4/DMD/le	H5/22q11/le	H6/22q11/le	H7/TS/le	H8/TS/le
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Background and Effect	1	1	1	1	1	1	1	1	8
Treatment and Management	1	1	1		1	1	1		6
Heredity and Risk	1	1	1	1	1	1	1		7
Patient Rights	1								1
Type of Test	1	1	1	1	1	1	1		7
Test Procedure	1	1	1		1				4
Accuracy of Test	1	1			1				3
After Test	1								1
Shared Decision Making	1	1							2
Psychosocial Consequences	1	1							2
Consequences for Others	1	1	1						3
Benefits and Risks	1	1	1		1				4
Date and Source	1	1	1	1	1	1	1	1	8
Additional Support and Info	1	1	1	1	1	1	1	1	8

**Table 12**  
Results of Evaluation of Material from Sweden

	S1/BC/I	S2/BC/le	S3/DMD/I	S4/DMD/le	S5/22q11/le	S6/TS/le	S7/TS/I	
Background and Effect	1	1	1	1	1	1	1	7
Treatment and Management	1	1	1	1	1	1		6
Heredity and Risk	1	1	1	1	1	1	1	7
Patient Rights		1						1
Type of Test	1	1	1	1	1		1	6
Test Procedure	1	1	1	1	1			5
Accuracy of Test		1	1					2
After Test		1	1				1	3

Shared Decision Making		1						1
Psychosocial Consequences		1		1				2
Consequences for Others	1	1			1		1	4
Benefits and Risks		1						1
Date and Source	1	1	1	1	1	1	1	7
Additional Support and Info	1	1		1	1	1	1	5

**Table 13**  
Frequency of Key Themes per Condition

	HBC	DMD	C26	22q11	TS
Background and Effect Treatment and Management	92%	92%	100%	100%	100%
Heredity and Risk	100%	69%	33%	73%	75%
Patient Rights	100%	100%	100%	100%	88%
Type of Test	67%	8%	17%	9%	13%
Test Procedure	92%	100%	83%	100%	75%
Accuracy of Test	83%	85%	17%	55%	50%
After Test	75%	46%	50%	55%	38%
Shared Decision Making	67%	31%	33%	0%	13%
Psychosocial Consequences	58%	8%	50%	0%	13%
Consequences for Others	67%	8%	0%	0%	0%
Benefits and Risks	83%	23%	17%	36%	25%
Date and Source	83%	46%	17%	36%	13%
Additional Support and Info	100%	100%	100%	91%	100%
	67%	46%	0%	55%	63%

**Table 14**  
Frequency of Key Themes per Country

	UK	the Netherlands	Belgium	Sweden	Italy	Germany	Poland
Background and Effect Treatment and Management	100%	100%	80%	100%	100%	100%	80%
Heredity and Risk	88%	75%	60%	86%	86%	70%	40%
Patient Rights	100%	88%	100%	100%	100%	100%	100%
Type of Test	38%	13%	20%	14%	14%	40%	20%
Test Procedure	100%	88%	80%	86%	100%	100%	80%
Accuracy of Test	88%	50%	40%	71%	43%	70%	80%
After Test	100%	38%	40%	29%	43%	50%	80%
Shared Decision Making	38%	13%	20%	43%	43%	20%	40%
Psychosocial Consequences	38%	25%	0%	14%	0%	60%	0%
Consequences for Others	25%	25%	20%	29%	0%	10%	20%
Benefits and Risks	50%	38%	40%	57%	14%	40%	40%
Date and Source	75%	50%	40%	14%	43%	50%	20%
Additional Support and Info	100%	100%	100%	100%	100%	100%	80%
	50%	100%	40%	71%	0%	50%	20%

**Table 15**  
Breakdown of Leaflets and Letters Analysed per Condition

	Leaflets	Letters
Hereditary breast cancer	7	4 personal, 2 standard
Duchenne muscular dystrophy	3	8
Connexin 26	1	5
22q1 deletion	6	5
tuberous sclerosis	5	3

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