



## Genetic Testing in Europe - FP6 Network of Excellence

Network for test development harmonization, validation and standardization of services. [www.eurogentest.org](http://www.eurogentest.org)

UNIT 4-6. "Patient and Professional issues: education, ethics and patient rights

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WP 6.2 Leader Domenico Coviello, IT

WP 6.3 Leader Kris Dierickx, BE

WP 6.4 Leader Herman Nys, BE

### Minutes of the Eurogentest Unit 4-6 Workshop Building Towards Professional Competence and Patient Confidence

Milan, October 27-28, 2007

#### Venue of the meeting:

Aula Sforza

Fondazione IRCCS, Ospedale Maggiore Policlinico, Mangiagalli e Regina Elena  
Viale F. Sforza 35, 20122 Milan, Italy

The meeting was attended by:

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**All presentations from this meeting are available on the EUGT members website as powerpoint files.**

### **Meeting business report**

#### **Saturday 27<sup>th</sup> October 2007**

##### **Session 1.**

All those present were welcomed and introduced.

A brief summary was given of the work carried out during the first 2-3 years of the project.

##### **WP6.1**

An assessment of written patient materials about genetic testing for patients and families across Europe. This can be viewed at:

[<http://en.eurogentest.org/files/public/unit6/InterimReport.pdf>].

Development of a set of 11 patient information leaflets which provide information about genetic testing and inheritance patterns. These were developed with the help of both patients and professionals. All can be downloaded from:

[<http://www.eurogentest.org/web/info/public/unit6/patients.xhtml>].

Leaflets currently being translated into 13 European languages with 7 more to follow in 2008. Topics for psychosocial leaflets to be discussed during meeting. Initial ideas by participants included: Utility of genetic testing in multifactorial conditions, genetic tests on the market – whether to buy them or not, genetic testing and family – screening, communicating with relatives etc.

## **WP 6.2**

### Output of the first year

An overview of existing educational initiatives on genetics for health professionals in different countries. This can be viewed at:

[[http://en.eurogentest.org/files/public/unit6/EUGT\\_6-2\\_Document\\_1\\_Final\\_versions.pdf](http://en.eurogentest.org/files/public/unit6/EUGT_6-2_Document_1_Final_versions.pdf)].

### Output of the second year

A draft framework of core competences in genetics for a range of health professionals, available at: .

[[http://en.eurogentest.org/files/public/unit6/Core\\_competence\\_EUGT6\\_2.pdf](http://en.eurogentest.org/files/public/unit6/Core_competence_EUGT6_2.pdf)].

In the past year we have published a report of the work done in Year One in EJHG. The core competences have been disseminated and discussed with a range of stakeholders, including:

- All Unit 6 participants, professional and patient group experts.
- All other EUGT unit teams
- ESHG Board and membership
- Presidents/leaders of national professional genetic societies across Europe.

We have received and incorporated feedback into the document and compared the core competence document with all publications of ESHG Public Policy Committee for compatibility. A paper on the core competences has been prepared for publication.

## **WP6.4**

### Output of the work until now:

- Based on the answers from a questionnaire sent to at least two contact persons in each EU Member State and an advanced literature research, a country report for every Member State was produced with detailed information on the patient rights legislation and on the rights the users of genetic services.
- An article was written on the Patient Rights legislation in the EU Member States which already have ratified the Convention on Human Rights and Biomedicine and also one on the reasons why eight EU Member States signed, but not yet ratified the Convention on Human Rights and Biomedicine (both published in *Health Policy*).
- Booklets were produced in the European Ethical-Legal Papers – series,

on patient rights in the EU. There is already a booklet on Patient rights in the Czech Republic, Denmark, Estonia, Greece, Cyprus, Lithuania and Hungary (these last two are in press). (all booklets will be available after publication as a PDF-file on the EuroGentest-website)

Output in the months/ years to come:

- An article on the patient rights protection in the EU Member States that did not sign nor ratify the Convention on Human Rights and Biomedicine.
- Production of more booklets on patient rights in the EU.
- Production of leaflets on the rights of users of genetic services.
- In the end, all booklets on patient rights in the EU will be brought together in one book.

Objectives of the meeting were agreed with all those present as:

1. To inform participants of progress within Unit 6 to date and to obtain input on future directions and directives.
2. To define the knowledge, skills and attitudes required by health professionals to become competent in terms of genetic healthcare.
3. To plan how to involve member states in updating their health professional curricula appropriately to include genetics
4. To seek ideas and suggestions as to how to raise awareness of public, policy makers and politicians of the importance of genetic education
5. To begin developing a set of patient resources focusing on psychosocial issues of relevance to genetic testing.
6. To update participants on relevant initiatives (e.g. Family Route Maps Project).
7. To update and develop the EuroGentest Unit 6 website.

### **Session 2 and 3.**

#### ***Patient expert group***

Where next?

Discussion of Psychosocial Booklets.

There was an agreement that there is a need to develop psychosocial information for patients, parents and families related to genetic testing.

We brainstormed different possible themes that we could develop information about. These were as follows:

- 1) Information for parents who have lost a child.
- 2) Information for parents when an abnormality has been found through prenatal testing.
- 3) Information for carriers/carrier testing.
- 4) Implications of having a child with a genetic condition: implications for parents, siblings and grandparents.

- 5) Signposts to services when a genetic condition has been found in the family.
- 6) Information for children whose parents have been diagnosed with a late onset disorder.
- 7) Information for families when no diagnosis can be found, or results are inconclusive
- 8) Practical legal information relating to patient rights for patients and families.

We then voted on which three we thought should be prioritised. The most popular were 3, 4 and 7. It was felt WP6.4 could work on an a general EU based information leaflets about patient rights.

Participants were divided into 2 groups to think about the content of the three leaflets. After half an hour groups gave feedback.

### **Implications of having a child with a genetic condition**

#### **Parents**

You are not alone.

All conditions can be improved with correct management

Couples/families need to work together as a team. Can be difficult and you will need help.

Importance of communication. Communicate with each other, communicate with support groups.

Don't be afraid to ask for help – from support groups, other family members

Develop new friendships with people in a similar situation to yourself. It is often helpful to speak to other people about their experiences. Share ideas and feelings.

Look after yourself and your own needs. If you are in poor health yourself you are less likely to be able to care productively for someone else.

Try to lead as normal a life as possible.

You must not feel guilty or ashamed.

Try to retain your sense of humour.

If you are thinking about having another child – be aware of the risks, but don't be afraid.

Look for support from patient groups and additional services.

#### **Siblings**

Don't be ashamed

Be aware that your involvement will be important and helpful to your parents

Parents may need to spend more time with affected child – this is normal, try not to feel isolated or angry.

Your help helps!

#### **Grandparents**

'double whamy' syndrome – feel upset for your child as well as your grandchild.

Help is possible through providing respite care, finding out information, financial help if possible.

Is it of any help to the family if you get tested?

### **Carrier Testing**

What does it mean to be a carrier?

Usually not affected by a condition if you are a carrier, although sometimes show mild symptoms

Can be a carrier with or without an affected person in the family. Need to distinguish between the two and how each would affect the person.

Consequence for the rest of the family, e.g. other/future children.

Being a carrier does not mean you cannot have children, what are the options if you want to have children?

Need to distinguish between those who have children already, those who don't have children and don't want children, and those that don't have children but want children.

Carrier of a late onset condition

De novo mutations

Tendency to 'imagine' symptoms when you are a carrier – constantly looking for a sign of being affected.

Emotional feelings e.g. guilt, stigmatisation – normal to have such feelings.

However, everyone 'carries' genetic mutations. Therefore must not feel 'defective' or ashamed.

Communication with partner very important

When and should the partner be tested?

Communication with family members very important, might prevent condition in future children, other family members may want to know about their risk.

Should not interfere with social and family life – ways of coping if it does.

### **Non-Diagnosis**

For some parents it may be important not to give up looking for a diagnosis.

Others may not want to keep looking.

For those that want to keep looking - may be other specialists in other cities/countries that might be able to help.

Geneticists want to solve problem. Help them by keeping them updated.

Parents as experts – parents need to take on role of informing the professional about the child's progress.

Keep a diary of child's progress.

Are there any support services that can help?

Look for umbrella organisations at national and international level.

Try to live day by day. Try to lead as normal a life as possible.

No diagnosis can mean there is no specific treatment. Therefore treatment can have side effects due to the interaction with the condition, although all treatment can have side effects.

Can often take years to find the correct diagnosis. Might be improved scientific

techniques in the future.

### **Professional expert group**

Discussion of knowledge, skills and attitudes required by health professionals to be competent.

We started with several questions:

- What is the main purpose of the document?
- How can we gain approval from relevant stakeholders?
- How can we disseminate it?
- Do we need to add in Knowledge, Skills and Attitudes as an extra level of detail?

We also discussed whether it would be helpful to insert more descriptions of genetics as they are in each country.

It was felt that the strength of the document lay in that it provides general principles for professionals in different countries, rather than focusing on specifics. This enables it to be used flexibly. Also, rather than describing the current situation across Europe, it is a roadmap to support future development of services. It is important that the document can be used by professionals to argue for educational development in their own countries. We learnt of a new EC document on rare diseases that will soon be posted for public consultation for 8 weeks on the web; this may be of relevance and could be used to underpin the need for genetics education for professionals. We are waiting (from Rumen Stefanov) the correct name of document and the web address.

Other feedback included:

- The need for clear, concise messages at the start of the document to capture attention
- Relate to education, get educators on board
- Disconnect from licensing
- Knowledge, skills and attitudes may be too deep a level of detail
- Need to simplify, create an Executive Summary.

**Sunday 28<sup>th</sup> October 2007**

**Session 4.**

Presentation by Melissa Winter, GIG  
'Family Route Maps Project'

**Session 5.**

Presentation 'Telling Stories, Understanding Real Life Genetics' Interactive Website

**Session 6.**

Presentation 'Patient Rights and Genetic Testing' Unit 4

**Session 7.**

Planning of strategy

WP6.1

Discussion with professional group highlighted a number of issues related to the titles and content of psychosocial leaflets:

What about information for late onset disorders?

Leaflet on psychosocial impact of predictive testing and PND and PGD?

Communication in families – what is the attitude of professionals?

What if parent becomes too ill to care?

Be careful not to send contradictory messages, but also need to show both sides of story and be balanced.

Difficulty in defining risk e.g. for carriers (penetrance of certain conditions)

Families might not want to seek extra information from support groups – what then? Might however look at website even if they don't become members of the support group.

- 29 feb rare disease day - prepare materials for people to translate

- Turkish website - doc translated and circulated.

- www. Tamgen.org

WP6.2

Based on all the discussions, the plan is to change the format and create 4 more versatile documents, as described below:

Document 1: Background information

- Make into separate document - 'Background to the development of core competences'

- Put key messages up front

- Use as appendix.

Document 2: Executive summary

- Short, sharp document with clear messages
- Relate core competences to pre-registration and undergraduate education
- Label as suggested guidance, not directives.

Document 3: Core competences for specialist practitioners in genetics

- Reduce categories (eliminate psychologists for example)
- Obtain approval from ESHG and disseminate via national genetics societies.

Document 4: competences for non- specialists

- Make it clear these were derived from the professional groups concerned (not prescribed by genetics specialists)
- Get support from professional groups concerned
- Label as suggested guidance to inform education, not directive
- ?reduce number of professional groups included.

### **Session 8.**

Patient rights and genetic testing (Presentation by Tom Goffin).  
Discussion on how to exercise your legal rights.

Discussion with *Professional Expert Group* and *Patient Expert Group*

On the question 'what rights should users of genetic services be informed about?'  
The following remarks were made:

- General information about their patient rights is better than a detailed list. This way information can cross boundaries. General information should be provided before testing and specific information during and after testing.
- Patients and practitioners should be provided with the same list of patient rights
- Maybe it is better not to think in terms of 'rights', but what is correct procedure, e.g. guidelines. "If you are getting good medical treatment, it would involve ..." This good medical treatment includes the more general patient rights
- There is a need to a list of sources where one can find more information about (specific patient rights)
- The leaflets for users of genetic services should be directed towards the rights in/to genetic testing.

### **Session 9.**

The new website format was demonstrated and feedback invited.  
The meeting closed at 3pm.