

# EuroGentest

*Guidelines for quality genetic counselling*

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## *Why European recommendations?*

- Quality genetic counselling is an integral part of genetic testing.
- Testing is increasing; new guidelines are emerging.
- Samples are crossing borders: the laboratory should be able to trust that the patients are properly informed and counselled.
- Health care personnel are crossing borders: guidelines and education/training should be similar in all European countries.
- Patients are also moving: family members may be counselled in different countries which requires uniform guidelines



## *OECD guidelines for quality assurance in medical genetic testing*

- “Pre and post test counselling should be available. It should be proportional and appropriate to the characteristics of the test, the test limitations, the potential harm, and the relevance of the test results to individuals and their relatives”
  - More in Annotations
- Relevant government or professional authorities should recognise medical genetics as a discipline comprising both a clinical and a laboratory specialty.



## *Council of Europe*

- Council of Europe is preparing additional protocols for “Convention for the protection of Human Rights and dignity of the human being with regard to the application of biology and medicine: Convention on Human Rights and Biomedicine” in 1997.  
...which Poland has signed 1999 but not yet ratified
- Draft Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes is under preparation; the final adoption of the text could be expected in the second part of 2008.



## *European Council: Draft Additional Protocol ...*

### ● **Article 8 – Information and genetic counselling**

- When a genetic test is envisaged, the person concerned shall be provided with prior appropriate information in particular on the purpose and the nature of the test, as well as the implications of its results.
- For predictive genetic tests ... appropriate genetic counselling shall also be available for the person concerned.
- The form and extent of this genetic counselling shall be defined according to the implications of the results of the test and their significance for the person or the members of his or her family, including possible implications concerning procreation choices.
- Genetic counselling shall be given in a non-directive manner.



## *Eurogentest recommendations for genetic counselling*

- EuroGentest sees genetic testing as a process
- It consists of
  - Correct indication for the test with appropriate clinical validity and utility
  - Pre-test counselling
  - Informed consent
  - Taking and sending the sample
  - The test in the laboratory
  - Interpreting the test result to the clinician
  - Post-test counselling
  - Possible other post-test events, like organising surveillance, contacting family members etc.



## *EuroGentest method for developing the recommendations*

- Investigating the background
  - Forming an Expert Group
  - Drafting Guidelines for quality genetic counselling
  - Circulating the draft to ESHG members and National Human Genetic Societies for comments (and endorsement)
  - Finalizing the draft with the help of Expert Group
  - Having endorsement from ESHG Board
  - Publishing the Guidelines
  - Publishing the method for developing the guidelines
- We are now here!



## *Investigating the background*

- International guidelines: ideal genetic counselling
- National regulation, guidelines and practices
- What happens in real life
  - See [www.eurogentest.org](http://www.eurogentest.org)



## *Ideal genetic counselling*

- In international guidelines, an ideal counselling session is seen as consisting of
  - an appropriately trained professional, who understands genetics and its ethical implications well
  - relevant and objective information
  - ensuring of understanding of the patient
  - psychological support
  - informed consent
  - confidentiality of genetic information
  - dealing properly with the familial implications and potential discrimination caused by testing
  - assuring autonomous decision-making by the counsellee



## *Forming an Expert Group*

- Angus Clarke
- Andrew Fawcett
- Lauren Kerzin-Storarr
- Gerry Kiebooms
- Luis Perez-Jurado
- Claire Julian Reyner
- Heather Skirton
- Aad Tibben
- Gerhard Wolff

- Jorge Sequeiros
- Helena Kääriäinen
- Elina Rantanen
- Marja Hietala
- Joerg Schmidtke
- Ulf Kristoffersson
- Irma Nippert

EuroGentest  
Public Health Group



## *Drafting Guidelines for quality genetic counselling*

- Definition of genetic counselling
- Definition of different testing categories
- Defining need for counselling in each testing category



## *Need for genetic counselling*

Test	Pre-test genetic counselling	Post-test genetic counselling
Diagnostic test	Sometimes	Sometimes
Presymptomatic (or predictive) testing	Always	Always
Susceptibility testing	Depends on the possible implications	Depends on the possible implications
Carrier testing	Always	Always
Prenatal testing	Always	Always
PGD	Always	Always
Genetic screening programmes	Always information, option for counselling	Always if test is positive



## *General statements about counselling*

- The resources needed to perform genetic tests and to provide the appropriate pre- and post-test counselling should be developed and put in place together.
- Cannot be compulsory?
- Has to be provided or supervised by an appropriately trained health care professional.
- Has to be given in a language well understood by the counsellee (or an interpreter should be available).
- Prerequisite is free and informed consent.



## *Components of pre-test counselling*

- Guideline lists 9 points, for instance:
  - the purpose of the test
  - possible uncertainties due to present methods/lack of knowledge
  - the rights to know and to decide including the right not-to-know
  - need to offer assistance in decision making, and encouragement to take ample time for it, whenever possible.
  - a written summary, if the counsellee so wishes.
  - initiation of the discussion about the need to inform relatives, especially in conditions where early diagnosis may improve the prognosis.



## *Components of post-test counselling*

- Some points from pre-test counselling have to be repeated, in addition:
  - implications of the test result to the individual (including a follow-up plan, when relevant) and his/her near relatives should be discussed.
  - a strategy to inform relatives has to be discussed with the counsellee (or, if necessary, a decision to discuss this further, after time for reflection).
  - a written summary of the test result and issues discussed during the counselling should be, as a rule, given to the counsellee. Also, written material to help the counsellee to spread the information in the family may be offered.



## *Problems encountered when drafting the recommendations*

- Clinical genetics is not a medical specialty in all European countries, the other "specialties" like genetic counsellor or laboratory geneticist are even less well defined in Europe.
- Networking with top experts is an excellent way of working...except for that we are all too busy.
- National Human Genetic Societies are not quick in their comments, as their Boards meet only infrequently.
- There are details for which a consensus is difficult to reach (like presymptomatic versus predictive)



## *Will the recommendations be "final"?*

- Quality of genetic testing as well as of counselling will be in a process of developing in the coming years.
- EuroGentest is preparing tools and recommendations, knowing that the field will always evolve.
- After EuroGentest, ESHG together with National Human Genetic Societies will possibly continue this work.
- OECD guidelines and, most probably, the additional protocol to the Convention on Human Rights and Biomedicine, have many similar views as Eurogentest recommendations. Thus these documents will give support to each other.

