

Regulations and practices related to genetic counselling in 38 European countries

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| | |
|---|-----------|
| Introduction | 1 |
| Methods | 1 |
| Results and discussion | 2 |
| Legislation and guidelines..... | 2 |
| Effect of regulation..... | 3 |
| Generally applied practices in context of different clinical situations | 5 |
| Generally applied practices related to different areas of counselling | 6 |
| The present state of genetic counselling | 7 |
| Predictions of future development | 8 |
| Conclusions | 9 |
| Annex 1 | 11 |
| Annex 2 | 14 |

Introduction

One aim of the EuroGentest project is to improve and harmonize the practices of genetic counselling in the EU and beyond. In order to find out the current regulations and practices concerning counselling in the context of genetic testing and to get an overview of European genetic counselling, the EuroGentest work package 3.1 gathered information from the National Societies of Human Genetics (NSHG's) in Europe.

Methods

A survey on legislation, professional guidelines and generally applied practices within genetic counselling was performed among European countries between September 2005 and February 2006. An electronic questionnaire was sent to all European countries (except for Andorra, Liechtenstein, Monaco, San Marino and Vatican that were assumed to co-operate with the neighbouring countries in the field of genetic counselling). The questionnaire was sent to the president of the NSHG in 29 countries that were found to have such a society. All of these societies provided the answers that were written either by the board of the society, by the president or by a selected expert. An appropriate contact person was searched with the assistance of the EuroGentest network and the European Society of Human Genetics in the countries that were not found to have a NSHG. The questionnaire was sent to these contact persons in Albania, Belarus, Bosnia-Herzegovina, Bulgaria, Iceland, Luxembourg, Malta, Macedonia, Moldova, Slovakia and Ukraine. No appropriate contact person was reached in Belarus and Luxembourg, but all of the other countries provided the answers. Altogether 38 answers out of 40 requested were received. Within EuroGentest, the survey was performed in cooperation with Unit 4.

The questionnaire had two parts. The first part dealt with legislation and professional guidelines related to genetic counselling. The respondents were asked if there were legislation or written professional guidelines related to different clinical genetic testing situations and to different practices related to counselling. With legislation we refer to official laws passed by the Parliament and additions that go through a Ministry. There are, in some countries, governmental or other bodies that have the authority to oversee practices in the health care, for instance to see whether the health care provider has the training needed for a certain health care act, like genetic counselling. Such type of regulation was not specifically enquired in this Survey. With guidelines we mean professional and other best practice papers that cannot be called legislation. The respondents were also asked to describe how the existence or lack of such legislation and guidelines affected the practical genetic counselling work. In the second part of the questionnaire the respondents were asked about generally applied but not officially written practices in different clinical situations in their country. They were also asked to estimate how well genetic counselling in their opinion was organized in their country and how they predicted the situation to change in the coming years. The questionnaire can be seen at <http://en.eurogentest.org/files/participant/other/questionnaires/Unit3CE.pdf> . The answers are summarized in Annex 1.

Results and discussion

Legislation and guidelines

Legislation forms an important framework for practices and services related to genetic testing. Medical Law Researcher Sirpa Soini investigated legislation concerning genetic counselling in the EU countries and found out that there are only few countries that have national regulation concerning counselling at the level of written laws (see <http://www.eurogentest.org/web/info/public/unit3/regulations.xhtml> .) In spite of these results, legislation was enquired about in the survey, as it was thought that there might be some laws that affect counselling even though they do not specifically cover it. The guidelines were asked about, as they regulate the practice too, especially if they are produced by an official body in the field of medical genetics.

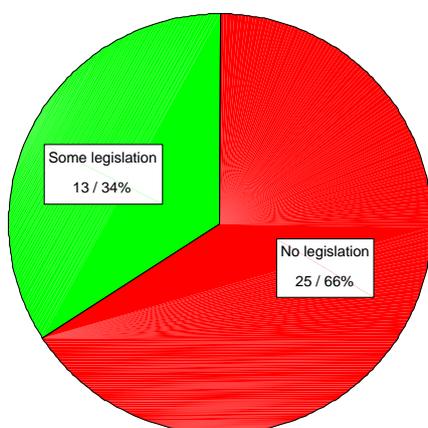


Fig.1: Existence of legislation related to genetic counselling in 38 European countries.

According to the survey, in 25 countries there is no national legislation related to genetic counselling, except for some general legislation that deals, e.g., with issues such as informed consent and confidentiality in the field of health care. In 13 countries there is legislation that specifically regulates genetic counselling (fig. 1).

In Sirpa Soini's study, legislation was searched in 26 EU countries, and some regulation related to counselling was found in five countries. In Austria, there is the Austrian Gene Technology Act – Gentechnikgesetz. In France, there are Code Civil and Code de la Santé Publique that regulate many bioethical issues, and also the Law on Bioethics. In Norway, there is the Lov om humanmedisinsk bruk av bioteknologi and, in Switzerland, the Federal Law on the

Genetic Testing of Humans. Also Portugal has a Law on Genetic Information. These came up also in our study.

In addition to these, four countries that were also involved in Soini's study – Belgium, Czech Republic, Italy and Lithuania – reported about their legislation in our study. The difference is explained by different phrasing of the question: in our survey the respondents were asked about all legally binding regulations that affect genetic counselling, while Soini looked only for laws specifically regulating counselling. Also, four countries that were not involved in Soini's study – Albania, Bulgaria, Moldova and Ukraine – reported to have legally binding regulations concerning genetic counselling.

The situations the reporters mentioned their national legislation to cover most often were counselling in the context of prenatal diagnosis, informed consent and confidentiality. These issues were also mentioned in some of the answers from the countries that do not have legislation specifically related to genetic counselling. This means that consent and confidentiality are covered in most of the European countries in other laws regulating health care. Prenatal diagnosis is also regulated in many countries, even though this does not always specifically relate to genetic counselling. There were also topics seldom covered in the national legislation: for instance, in only very few countries counselling persons from ethnic minorities or the duty to recontact the patient are mentioned in the legislation. (see Annex 2.)

According to the survey, there are some professional guidelines that deal with genetic testing issues in 21 countries. In 17 countries, there are either no guidelines or the guidelines relate more generally to health care (fig. 2). Topics that are most often covered in the national guidelines are counselling in the context of prenatal diagnosis, testing of children and adolescents, non-directiveness of counselling and who can perform genetic counselling. These topics are mentioned in the national guidelines of 10-15 countries. Topics that are covered in the guidelines of only two or three countries are counselling in the context of predisposition testing for multifactorial diseases, counselling persons from ethnic minorities and the duty to recontact the patient. (see Annex 2.)

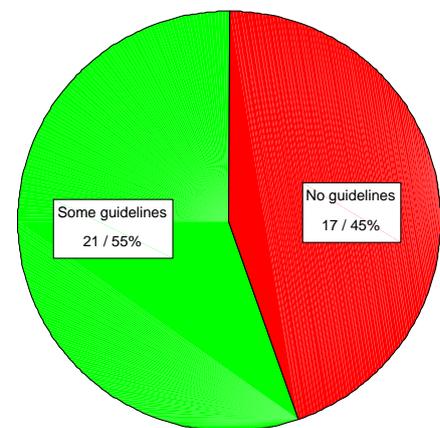


Fig.2: Existence of guidelines related to genetic counselling in 38 European countries.

Effect of regulation

The respondents were asked how they think that the existence or the lack of regulation affects the practice of genetic counselling. Most of the respondents thought that the effect of regulation was positive and that genetic counselling should be more regulated (fig. 3).

Respondents from nine countries were happy that they have legislation. “The existence of such legislation helps to standardize practical work”, mentioned one respondent, and another one thought that “Guidelines and specific laws are very important to guarantee the quality of treatment in respect to the patients and their families in all medical centres”.

Respondents from 18 countries wanted more or better regulation. In 10 of these countries there were already some regulations, but the respondents did not find them comprehensive enough (fig. 4). “The undeveloped specific legislation and lack of guidelines has a negative impact on practical work in this field”, remarked one respondent. It was also stated that “national legislation should be more detailed”. In 8 countries there is no regulation and the respondents wished to have improvements in this respect. One respondent thought that the lack of regulation has a negative effect “due to different considerations among specialists and patients as well”.

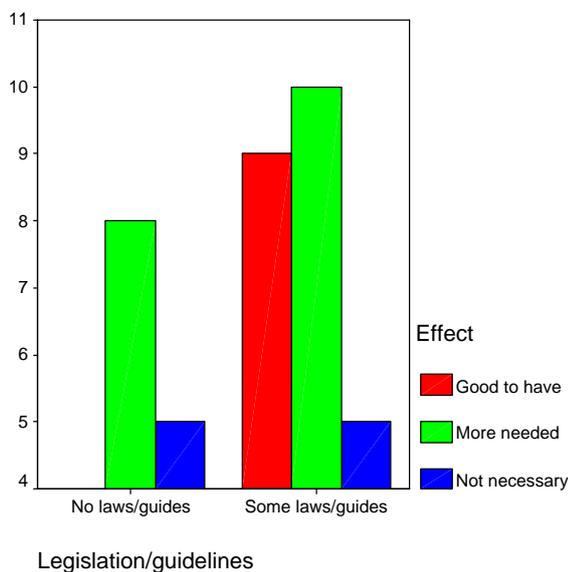


Fig. 4: Opinions of respondents from 38 European countries on the effect of regulating genetic counselling according to the existence of legislation and guidelines in the country.

recommendations. More than 70 % of the respondents thought, however, that it is either good to have regulations or that more regulations are needed at the national level. The neutral comments came mostly from the old EU member states, but also from Iceland, Romania, Serbia and Slovenia. Also most of the respondents who were satisfied with their existing legislation were from the old member states. This group included also Lithuania, Norway, Russia and Switzerland. The respondents who wanted more regulation came from all over Europe, seven of them belonging to new EU member states, four to candidate EU member states, four to other European countries, and three to old EU member states (fig. 5). In this respect, it can be stated that regulation is wanted especially in the countries where the development of genetic medicine may have been more recent and issues related to human genetics may be a newer area of consideration.

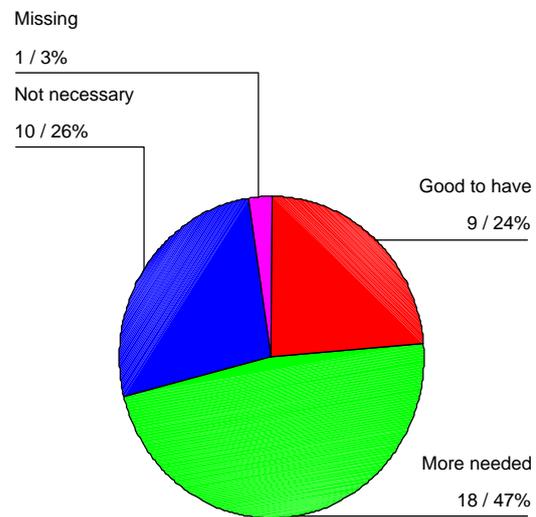
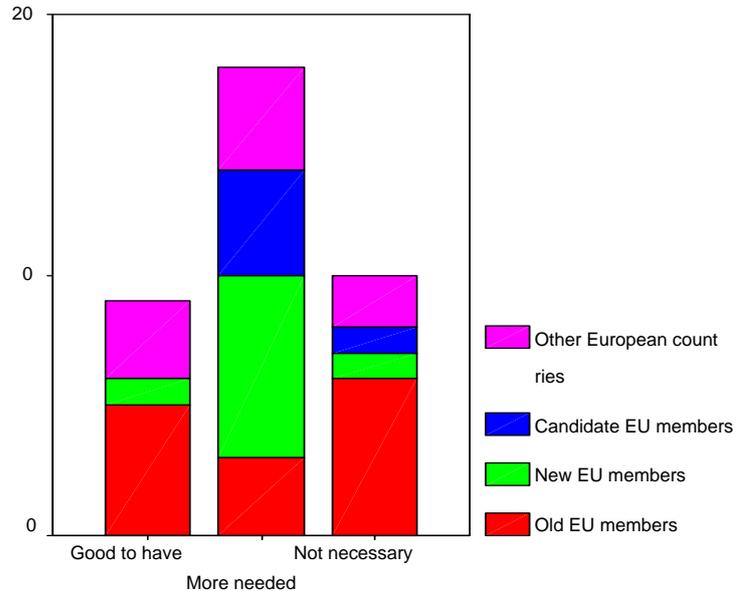


Fig. 3: Opinions of respondents from 38 European countries on the effect of regulating genetic counselling.

Respondents from 10 countries thought that national regulation is not necessary, even though five of them had legislation or some guidelines (fig. 4). “Most guidelines which exist are helpful, but lack of legislation allows flexible practice”, remarked one respondent. Another one did not find the lack of regulation to affect practical work, because “the existence of generally applied practices among clinical geneticists covers these clinical situations”. Some of the 10 respondents thought that there is no need for national guidelines, because “the professional staff follows international guidelines”.

Hence, regulations are seen as a positive factor in most European countries. Only one respondent questioned this view pointing out that the lack of legislation allows flexible practice. Some respondents were neutral about having guidelines and some did not miss national guidelines, but found it good to have international

Fig. 5: Opinions of respondents from 38 European countries on the effect of regulating genetic counselling according to the membership of the EU of the country.



Generally applied practices in context of different clinical situations

Even though there is no legislation related to genetic counselling in 25 European countries and no guidelines in 17 countries, the representatives of most of the countries, except for Bosnia-Herzegovina, Iceland and Malta, considered that they have generally applied practices of genetic counselling.

The respondents were first asked, if they had some generally applied practices *for different clinical situations*. They were also asked to describe what these practices were like. Half of the respondents described their practices in context of at least one clinical situation, whereas half of the respondents either did not have the practices or did not describe them (see Annex 2).

Respondents from more than 30 countries reported to have some generally applied practices within counselling in the context of *diagnostic, carrier and prenatal testing*. Most of the respondents either referred to the legislation and guidelines or stated that counselling takes place both before and after testing, or, on the contrary, only before or after a genetic test, as needed. Some respondents mentioned that counselling is performed in appropriate settings, e.g., in clinical genetics services.

According to the answers received, there are generally applied practices for counselling in the context of *predictive testing* in 29 countries and in the context of *preimplantation testing* in 19 countries. Only 11 respondents considered they have generally applied practices for counselling in the context of *susceptibility testing for multifactorial diseases*. Most of the answers again dealt with performing counselling both before and after testing. There were also referrals to legislation and guidelines, and appropriate clinical settings were mentioned as well. In the context of predictive genetic testing, offering psychological support was also mentioned, as well as not performing the test in children.

Generally applied practices related to different areas of counselling

As *informed consent* is generally considered to be an important part of genetic testing procedure, the representatives of the European countries were asked if they had uniform practices to ask for consent in different clinical situations (fig. 6). Half of the respondents answered that written consent is required for some cases while in other cases only verbal consent is asked for. Most often, written consent was required in the context of prenatal and predictive testing. In 12 countries, written consent was required in every case, and in 4 countries, verbal consent was always sufficient. According to representatives of 3 countries, there was no generally applied practice of asking for consent in their country.

There are some practices that most of the representatives reported to be generally applied in their countries (see Annex 2). More than 80 % of the respondents considered that it is generally agreed *who may perform genetic counselling*, and that *counselling should be non-directive*. Some of the respondents mentioned that these issues are regulated in written guidelines. Respondents from 19 countries described that only specialized medical geneticists or genetic counsellors perform genetic counselling, while 8 respondents said that sometimes other medical professionals provide counselling as well. Representatives from 6 countries thought that non-directiveness of counselling is a golden rule in their country, but 9 respondents felt that it is applied generally only among medical geneticists.

Respondents from 26 countries considered that there is a generally applied practice of *contacting relatives at risk*. Seven of them described that the relatives are contacted only through the index patient, while other seven respondents mentioned that it is possible that the professional contacts the relatives as well, with the consent of the index patient. Respondents from 22 countries considered *offering psychological support* in connection with counselling as a generally applied practice in their country, and respondents from 19 countries thought that the practices related to *counselling of minors or persons with diminished capacity*, are also generally applied in their country. In addition to referring to guidelines, altogether 6 respondents mentioned assistance of parents, appropriate level of counselling and counselling in clinical genetics settings as such practices.

The practices that were considered more rarely applied in a general way were the *duty to recontact the patient* in case of any further information, *need for breaching confidentiality*, and *counselling persons from minority ethnic groups*. However, nearly half of the respondents considered the duty to recontact the patient and need for breaching confidentiality as generally applied practices. It was mentioned that recontacting happens when ever possible. Only one representative mentioned that there are registers through which patients are contacted, if needed. Respondents from 4 countries said that breaching confidentiality is forbidden in the law or in the guidelines, and other 4 respondents mentioned that it is done only in exceptional cases and never

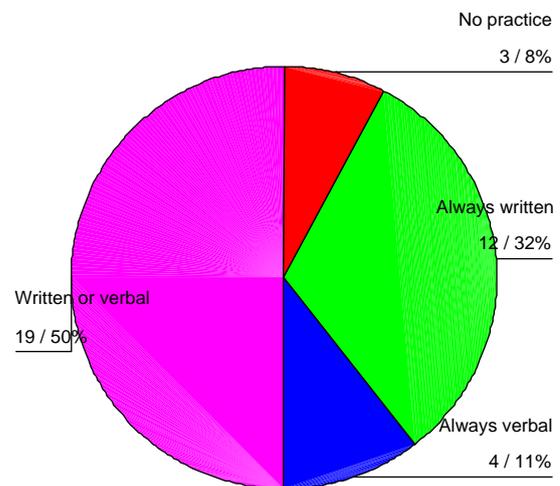


Fig. 6: Asking for consent in context of genetic testing in 38 European countries.

without the consent of the patient. Only 3 respondents mentioned having generally applied practices related to counselling persons from ethnic minorities. They described using interpreters and involving aspects of cultural differences in the training of professionals.

The present state of genetic counselling

The respondents were asked to describe how well genetic counselling was organised in their country in their opinion (fig. 7). Only respondents from Finland, the Netherlands and the UK were completely satisfied with the situation and thought that genetic counselling is well organised in their country. “There is a good access throughout the country and there is a sufficient number of well trained clinical geneticists”, was explained.

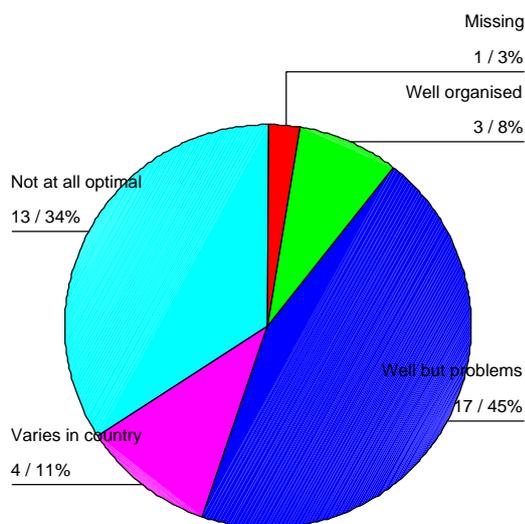


Fig. 7: Opinions of respondents from 38 European countries on the organisation of genetic counselling.

Thirteen respondents considered that genetic counselling is not at all optimal in their country. It was stated that genetic counselling is not organised at all or that a lot of improvement is still needed. It was also said that there is a lack of specialists, need for better education, failures in networking and lack of formal recognition of the field. In addition, representatives of four countries thought that organisation of counselling varies within the country. One of them explained that “genetic counselling is well organized only in the capital”.

The respondents that were either quite satisfied or not satisfied at all with the organisation of counselling came from all

Most of the respondents were quite satisfied with the situation, but thought that some improvements are needed. Positive sides that were mentioned in the answers were good access to and infrastructure of counselling services and sufficient amount of education and expertise. One respondent stated that “we have trained professionals and knowledge, but there is a lack of legislation and national guidelines”. The gaps that were mentioned also related to educational and service provision issues. One respondent said that genetic counselling is quite well organised, but “there is a lack of provision in relation to the demand on genetic counselling”.

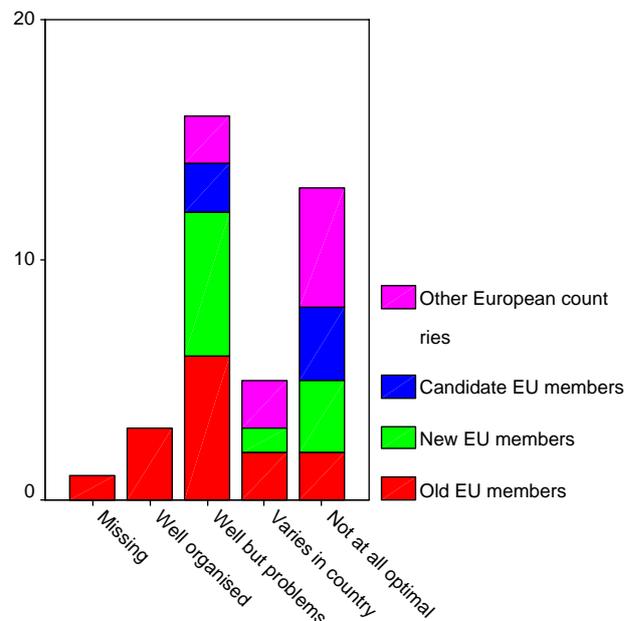


Fig. 8: Opinions of respondents from 38 European countries on the organisation of genetic counselling according to the membership of the EU of the country.

over Europe. Somewhat fewer of the old EU member countries belonged to the least satisfied group, and the only three respondents that were completely satisfied were from old member states. The respondents who were a little bit less satisfied with genetic counselling in their country came mostly from the new and candidate EU member countries, as well as from other European countries (fig. 8).

The respondents were provided a list of factors that may have an impact on the organisation of counselling. They were asked to mark the factors they consider to affect the situation in their country. More than 70 % of the respondents marked the lack of general genetic knowledge among healthcare professionals and prioritization of healthcare resources. More than 60 % of the respondents considered lack of legislation and guidelines and lack of trained professionals to be such factors. Representatives of 10 countries thought that unequal access to counselling as well as geographical distances were problems in their country. These respondents were partly the same; so, in some cases the long geographical distances may explain unequal access. Only representatives of 4 countries considered that there are language and cultural problems in counselling in their country. The respondents were also asked if there were any other reasons than those on the list; the ones that were mentioned included the poor quality control of practices, lacks of medical insurance, lack of corporate identity among geneticists, and problems specific to a small population.

Predictions of future development

Finally, respondents were asked to describe what their predictions were about changes that might happen within genetic counselling practices in their countries in the near future (fig. 9). Even though this was an open question, 17 respondents predicted or hoped for more legally binding regulations or written professional guidelines. Some of these respondents described why this development would be needed. “The new legislation I hope will make the provision of medical genetic services more uniform all over the country”, explained one respondent. In some countries changes were expected to formalize the position of medical genetics. “If genetics is finally recognized as an independent speciality, genetic counselling and the remaining activities related to human genetics will significantly change for better, since genetic services will have to be organized and run by trained and experienced professionals in the field of human genetics”, stated one respondent.

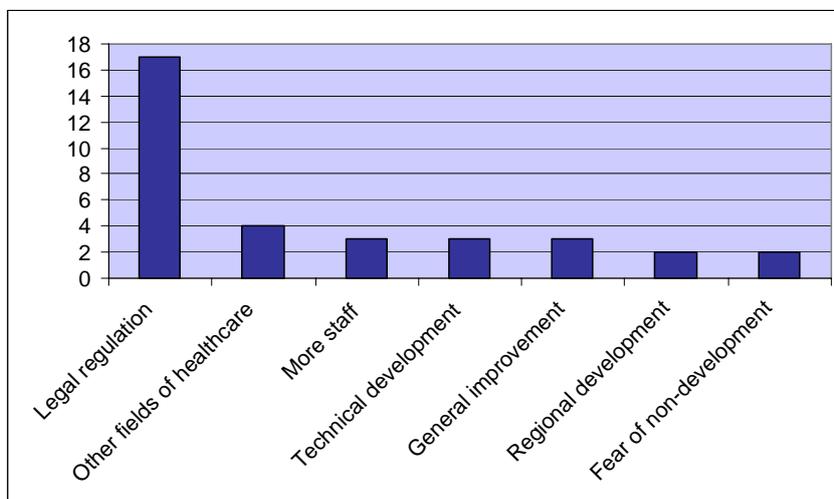


Fig. 9: Predictions of respondents from 38 European countries about future development in genetic counselling.

It was also thought that genetics would be more integrated into other fields of healthcare, both into primary care, and into other specialities such as oncology. “Medical genetics will tend to be performed more and more by non-geneticists, with widely variable competence in the discipline”, predicted one of the respondents.

Increase in the amount of professionals in the field was also brought out. Three respondents mentioned technical development, which mainly referred to the quality of the laboratories. Three respondent predicted general improvements in the field of genetic counselling. Regional development was also mentioned: “We hope that in the near future the regional organization of genetic services will be improved with the employment of more specialists in genetics”, said one respondent. Two respondents were afraid that there would be no development in genetic counselling in their country for the near future.

Issues that only one respondent mentioned as a future development were improvement in the definition of cases that need counselling, increase in continuous training, and that development would take place only through private sector or when the next generation of physicians is on the field. It was also predicted that the patient journey through the genetic service system would get more difficult.

Conclusions

As the aim of the EuroGentest work package 3.1 is to improve and harmonize practices of genetic counselling in the EU and beyond, a survey about current regulations and practices of counselling was very much needed. The answers that the NSHG’s provided highlighted some of the key issues of counselling that need improvement and harmonization.

The survey was performed in order to have an overall picture of genetic counselling practices in Europe, and to get a perspective of national experts to help in the writing of European recommendations for counselling in the context of genetic testing, as well as tools for assessing the quality of genetic counselling. Both legislation and guidelines were asked about, as national experts could best describe their content knowing the language and the legislative system of their country. The legal framework as well as professional guidelines in different countries is important to know, as they strongly affect counselling practices.

It was known beforehand that most countries do not have regulation specifically related to genetic counselling. Therefore, generally applied practices were also asked about. It can be assumed that, at least in the small countries, it is generally known or even agreed, how issues related to counselling, e.g. consent procedure and non-directiveness, are handled.

Questions about legislation, guidelines and generally applied practices were difficult both to formulate and to answer. Some of the respondents may not have known about every guideline in their country. There may be legislation on different levels, and some of the respondents may have included general health care legislation as applied to genetic counselling, while others may have focused more on legislation specifically mentioning genetics. Some of the respondents answered more generally about genetic testing, and did not focus on counselling. There may also have been language problems. Therefore, the answers cannot be considered as completely comparable. They also reflect the perspective of a group or only a single respondent in each country. The survey was performed between September 2005 and February 2006, so the legal framework may have changed and some new guidelines may be in preparation.

The answers, however, give information on the main lines of legislation, guidelines and practices. They show which topics are more often discussed and regulated, and which ones need

more attention in the future. The answers also bring out that more regulation is hoped for and expected, and that in order to improve genetic counselling, more uniform practices of education and organisation of genetic counselling within the healthcare system are needed.

Annex 1

Regulations and practices related to genetic counselling in 38 European countries

| Country | Existence of legislation related to genetic counselling (+ = yes / - = no) | Existence of professional guidelines related to genetic counselling (+ = yes / - = no) | Effect of regulation on practical work (+ = good to have / would be necessary, - = not necessary) | Existence of generally applied practices related to genetic counselling (+ = all practices, + / - = some practices, - = no generally applied practices) | Existence of generally applied practice of informed consent (+ = in every case, +/- = in some cases, - = no generally applied practice) | Organisation of genetic counselling (+ = well organised, +/- = well organised, but some problems, - = not well organised, * = varies within country) | Predictions of future development |
|---------------------------|---|---|--|--|--|---|--|
| Albania | + | + | + | +/- | +/- | * | Regional development |
| Austria | + | - | + | +/- | + | +/- | National guidelines |
| Belgium | + | + | - | +/- | + | +/- | More non-geneticists and genetic education |
| Bosnia-Herzegovina | - | - | + | - | - | - | Development through private sector |
| Bulgaria | + | + | + | +/- | + | +/- | Written guidelines, medical insurance |
| Cyprus | - | - | + | + | + | +/- | Improvement of services, awareness raising |
| Croatia | - | - | + | +/- | + | +/- | National legislation |

| | | | | | | | |
|-----------------------|---|---|---|-----|-----|-----|---|
| Czech Republic | + | - | + | +/- | +/- | +/- | More counsellors |
| Denmark | - | + | + | + | + | +/- | More professionals |
| Estonia | - | - | + | +/- | + | +/- | Official recognition, written guidelines |
| Finland | - | - | - | +/- | - | + | Written guidelines |
| France | + | + | + | | + | | |
| Germany | - | + | + | +/- | + | +/- | Law in preparation |
| Greece | - | - | + | +/- | + | +/- | Legislation for medical genetics specialty |
| Hungary | - | + | + | +/- | + | +/- | Legislation, guidelines |
| Iceland | - | - | - | - | - | - | Improvement of practices |
| Ireland | - | - | - | +/- | +/- | - | More professionals, more guidelines, better recognition |
| Italy | + | + | + | +/- | + | * | Regional development |
| Latvia | - | + | | +/- | +/- | - | |
| Lithuania | + | + | + | +/- | + | +/- | Electronic medicine, distance education |
| Macedonia | - | + | + | +/- | +/- | +/- | Legislation |
| Malta | - | - | + | - | +/- | - | Changes with next generation |
| Moldova | + | - | + | +/- | + | +/- | Legislation |
| Netherlands | - | + | - | +/- | +/- | + | Common cases into regional hospitals |
| Norway | + | + | + | +/- | +/- | +/- | Better defined cases |

| | | | | | | | |
|------------------------------|---|---|---|-----|-----|-----|---|
| Poland | - | - | + | +/- | +/- | +/- | More complex disorders to counselling |
| Portugal | + | - | + | +/- | +/- | +/- | Legislation |
| Romania | - | - | - | +/- | +/- | - | Improvement of practices |
| Russia | - | + | + | +/- | + | - | Pessimistic predictions |
| Serbia and Montenegro | - | - | - | +/- | +/- | * | Quality assessment, national databases |
| Slovakia | - | + | + | +/- | + | +/- | Legislation |
| Slovenia | - | + | - | +/- | + | * | Legal and professional regulation |
| Spain | - | - | + | +/- | +/- | - | Formal recognition |
| Sweden | - | + | - | +/- | + | +/- | Legal regulation |
| Switzerland | + | + | + | +/- | + | - | Hope for non-deterioration |
| Turkey | - | + | + | +/- | + | - | Legislation for medical genetics specialty |
| UK | - | + | - | +/- | + | + | Complication of patient journey through genetic service |
| Ukraine | + | + | + | +/- | + | +/- | Improvement of methods and guidelines |

Annex 2

Different clinical situations and topics related to genetic counselling in legislation, guidelines and practices of 38 European countries

| | Mentioned in legislation of N countries | Mentioned in professional guidelines of N countries | Considered as a generally applied practice in N countries |
|---|--|--|--|
| Diagnostic testing | 15 | 10 | 31 |
| Carrier testing | 12 | 5 | 32 |
| Predictive testing | 9 | 9 | 29 |
| Susceptibility testing for multifactorial diseases | 7 | 3 | 11 |
| Prenatal diagnosis | 21 | 13 | 32 |
| Preimplantation diagnosis | 8 | 6 | 19 |
| Testing of children and adolescents | 11 | 12 | |
| Consent of the patient | 24 | 5 | 22 |
| By whom can genetic counselling be performed | 14 | 10 | 33 |
| Non-directiveness | 11 | 11 | 31 |
| Counselling persons from minority ethnic groups | 2 | 2 | 8 |
| Counselling minors or persons with diminished capacity | 8 | 6 | 19 |
| Psychological support during counselling | 9 | 9 | 22 |
| Informing at-risk relatives | 9 | 7 | 26 |
| Confidentiality (and need to breach it) | 19 | 10 | 17 |
| Duty to recontact the patient | 4 | 3 | 18 |