WELCOME to 2006 and the second year of EuroGentest. We are now in full reporting mode as the various working parties have scoped out their tasks and are now drawing up and even starting to disseminate recommendations in their fields of expertise. In this issue we focus on two of the new EU member states – the Czech Republic and Poland – both of which are enthusiastic supporters of the project as they strive to strengthen their genetic services. We also hear from Unit 6 on their initial survey of public education tools available – and the surprising findings. Technology wise we are in a period of great activity – and expectation – as you will see in the press watch section. Therefore I am also pleased to announce EuroGentest's "Call for new genome technologies" initiative in which we will endeavour to help bring useful new testing methods to the public faster. As usual we welcome all feedback and look forward to your comments.

Jean-Jacques Cassiman

EUROGENTEST NEWSLETTER3/2006

Czech Republic welcomes EuroGentest

As one of the new member states of the EU, the Czech Republic has been an enthusiastic backer of EuroGentest seeing major opportunities to learn and benefit as it seeks to restructure and develop its health service in general and genetic service in particular. In this interview, Milan Macek Jr, (MM) Professor of Medical and Molecular Genetics at Charles University in Prague explains the reasons for this enthusiasm and the fascinating background and heritage of genetic testing in the Czech Republic.

EuroGentest (EG): Genetic testing has a long history in the Czech Republic does it not?

MM: It's true there are strong roots. Mendel's pioneering work was performed in Brno in the 1860s. This had a strong influence on Czech biologists with Ruzicka publishing a very modern sounding monograph on "Human Inheritance in Health and Disease" as early as 1917. Sekla established the first genetic counselling unit in Prague in 1937 and in 1946 was one of the first to condemn the Nazi misuse of genetics and ethnic cleansing. Sekla then bravely continued to criticize Lysenkism during the Soviet era. Despite scientific and political oppression, Czech immunogenetics reached world-leading status with the work of the Hasek's group which formulated the laws of immunological tolerance at the same time as Nobel Laureate Medwar but published in a Czech journal. Progress accelerated in the sixties and then there was a further setback with the Soviet invasion of 1968 which led many well-known geneticists to emigrate. However, medical genetics was recognised as an important discipline by the Ministry of Health and in 1980 the rather modern concept of genetics care was established. A network of medical genetics departments was formed across the country mostly comprising counselling and cytogenetics units, with associated specialised laboratories for the detection and screening of metabolic disorders. The Czech Republic was the first ▶

to introduce prenatal diagnostics in 1970 and again helped increase international recognition. Indeed the first International Conference on Prenatal Diagnostics was held in Prague in 1990. Medical Genetics was also recognised in 1970 as a medical speciality. Following the Velvet Revolution in 1989 in common with other medical specialities a large number of geneticists went abroad to study and work. However, just a few returned back home - in other words we have suffered a substantial "brain drain".

EG: It seems healthcare in the Republic has gone through a rapid period of change recently?

MM: Yes. The political situation has become more stable since the early to mid 90s and this has the effect of people becoming more interested in quality of life, leading to renewed interest in healthcare and science. The brain drain seems to have stopped with students looking to study in Europe and then commit themselves long term here. There is also a commitment to a public health service. Following the Velvet Revolution there was a rush to privatisation and a healthcare insurance based system. What other countries have experienced in say 15-20 years, we compressed in 4-5. In medicine in general we saw the sudden proliferation and duplication of private service providers concentrated around the main centres of population such as Prague. In genetics in particular we had the arrival of labs offering OTC paternity tests. Now however there is a reaction and agreement that there should be an NHS style service with access to services not based solely on insurance qualification and ability to pay. In genetics there is also recognition that the old communist system of a regional network was in fact quite good. The challenge of course is how to fund such a system. Currently the Czech economy is doing rather well. Our country has reached the status of eg. Portugal (in general we are at 70% of the EU GDP average), while the Prague region has even surpassed it by approximately 30%! Nevertheless, benefits from such recent growth have a natural delay for us in the academia. A 13% national insurance tax sounds a lot, but has to spread a long way, from an average salary of about 600,- EUR. Simply, we purchase at the general EU market at "Western" prices, yet collect proportionally much lower insurance tax given the overall lower wage range of our population.

In genetics we have now to justify ourselves – why should there be investment in genetics which "only" offers long term prevention as opposed to other sectors of healthcare which provide "immediate" treatment or as the politicians say "immediate returns". We have a lot to do convincing both the politicians and public that investment will enable us to provide faster outcomes and thus predict risk better and that this is just as vital as acute healthcare.

EG: How are services currently organised?

MM: We have a network of 39 institutes/departments of medical genetics providing services for our 10.3 million population. This is a private/state owned mixture. Within the academic institutes, which are responsible for undergraduate and some even for post graduate training, there are centres for national competence and quality testing. These range from the Cystic Fibrosis Centre to the Centre of Molecular Biology and Gene Therapy. Broad-based international research is performed at the Institute of Inherited Diseases and Laboratory of Mitochondrial Diseases. 67 full-time geneticists are employed throughout the network, with a general ratio of geneticist per head of population of 1-156,000.

Professor Milan Macek Jr with his team from the Molecular Genetics Unit in Charles University Prague.

EG: Why did the Czech Republic get involved in EuroGentest?

MM: Since our genetics services are at the more or less European "standard of operations", we have the privilege that we can address and develop all issues put forward by this NoE. Already when EuroGentest was being discussed we were looking to share experience and learn on a European and international basis. Genetically the Czech Republic is very "European" thanks to our history at the centre of the continent providing a mixing pot for different populations. We had strong links with Belgium and as stated previously many of us had studied and worked abroad. Moreover, we have participated within the EU FP4 Inco Biomed activities and the FP5 Cysic Fibrosis Thematic Network. We follow international ethical recommendations for medical genetics (several step counselling during testing, informed consent, right to privacy and protection against misuse of information gained). Internal and external quality assurance schemes were also being developed in close collaboration with for example EMQN, ECA and the already mentioned CF Thematic Network. Now with EuroGentest, we have a forum not only for further developing this work, but also to help convince politicians here domestically about the potential benefits of investing in genetics. We will be able to show them the results in other member states which lends credence to our arguments.

EG: So you are an enthusiastic supporter?

MM: Indeed, at all levels, scientific, economic, political and most importantly patient, the project will bring great benefits.

Childrens Centre in the University Hospital, Motol where part of the Medical Genetics unit is located.

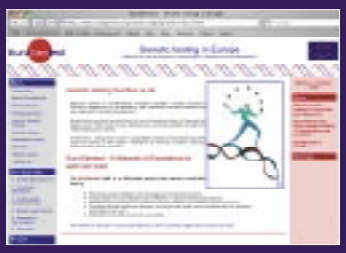
Poland struggles for recognition

Poland is again a strong supporter of EuroGentest, but as Professor Michal Witt of the Institute of Human Genetics in the Polish Academy of Sciences in Poznan reports the struggle for recognition and increased resources is hard.

Poznan is home to one of the leading Polish genetics laboratories.

The situation is difficult since genetic testing started to be offered by various medical centers (mainly private) which don't even have trained clinical geneticists on board! This is all allowed by current legislature in Poland, although it is fiercely criticized by professional geneticists. On the other hand, real genetic testing is based mainly on state funding which is limited and not always properly allocated. OB-GYN specialists in particular use up funds which should be for genetic testing in genetic laboratories. Nevertheless, there are already quite a number of molecular labs performing diagnostic testing properly, e.g. cystic fibrosis is being tested in 7 centers: some of them have international certificates, some Polish ones issued by the Polish Society of Human Genetics. Personally I'm involved in one of the internationally certified labs of the highest reference (there are two like this in the country). In addition, those locally certified were assessed within the framework of the European CF Genetic Network run in Poland by myself in the past, before EuroGentest started its activities. Unfortunately health authorities do not seem to care much about such marginal problems as genetic testing; at least I don't see any physical proof of such interest. All the actions aimed at improving the level of genetic testing in Poland are definitely based on an intrinsic feeling of responsibility of Polish geneticists. EuroGentest can be instrumental in sorting out priorities for such activities in full accordance to European regulations. Creating such regulations for the Polish legislature should also be one of the most important targets.

WEB CORNER



14000

hits a month demonstrates website's popularity

The popularity of EuroGentest's website continues to grow. According to Olivia Wilcocks there are now over 14,000 hits a month: "Not just from existing users, but new ones from all over Europe. We are now aiming to make the site more accessible and outward looking than before with new designs for the unit pages under discussion and of course more items such as the newsletter downloadable for the general public."

www.eurogentest.org

Microarray technology should lead to faster and more complex diagnosis of genetic disorders.

EuroGentest calls for new technologies to enhance genetic testing

Unit 5 has just announced an urgent 'Call for new Innovative Techniques in Genome Diagnostics'. This is because one of the key objectives of EuroGentest is to bridge the gap observed in the chain of new technology transfer from research into routine use in genetic diagnostics. To address this, Unit 5 aims to coordinate all the activities required for accelerated technical evaluation, validation and subsequent implementation of new technologies into clinical practice. Responses to the call will be used to help build a web-based inventory of new techniques and also discussed in an open satellite workshop at the ESHG meeting in May in Amsterdam.

Bert Bakker and Nienke van der Stoep, of the Center for Human and Clinical Genetics at the Leiden University Medical Center who are involved in Unit 5, say the timing of the call is no accident: "At present, the field of genetics is witnessing an impressive expansion of new technologies, including not only new advanced high throughput and micro-array approaches, but also straightforward yet highly sensitive detection-assays for DNA mutations and genome copy-number changes. As geneticists we see that several of these technical developments could have real potential to improve genetic testing." With this in mind, EuroGentest Unit 5 intends to acquire a comprehensive inventory of all the latest technological developments in genetic testing and to coordinate their technical evaluation through a network of accredited Diagnostic Centres all over Europe using well-characterized clinical samples. Bakker and van der Stoep believe that introducing such a coordinated introduction and evaluation of various new techniques in different Diagnostic Centres in place of the adhoc informal system that exists currently will not only improve the technical evaluation process, but will also speed up subsequent validation procedures and implementation of new genetic tests.

"We would also hope such a transparent system would increase public acceptance and uptake of tests. Therefore we are now making a web-based call for "Innovative Techniques in Genome Diagnostics" at our Unit 5 homepage. We invite both academic and industrial research groups to introduce and assess their latest developments through expert diagnostic laboratories. We aim to build an inventory of the new techniques with open access to complement our list of existing techniques we are trialling currently with EuroGentest members," continue Bakker and van der Stoep.

As part of this initiative, EuroGentest will also be hosting a Satellite meeting on "Innovative Techniques in Genome Diagnostics" at the ESHG in May 2006. At this meeting EuroGentest will provide a platform where companies and academic research groups can interact and present their new techniques in genetic testing and receive feedback. More information and application details can be found on the EuroGentest Unit 5 homepage. (http://www.eurogentest.org/cocoon/egtorg/web/info/homepage/unit5v2.xhtml)

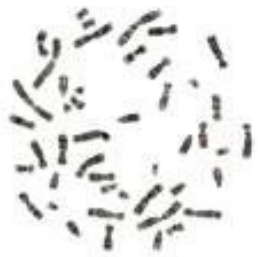
Understanding rare chromosome disorders

In a new section for non-specialist readers we look at the genetic conditions EuroGentest is helping to improve testing of. We start with rare chromosome disorders which get little general publicity but bring heartbreak to the families involved. To get an outside perspective, we invited a leading health journalist to interview Beverly Searle, member of EuroGentest and organizer of Unique, a UK patient support group and the mother of an affected child .

Unique

RARE CHROMOSOME DISORDER SUPPORT GROUP

A Registered Charity No. 1024624



Unique is a UK support group for families with rare chromosome disorders. To contact or donate to Unique, log on to www.rarechromo.org.

Inside every human cell's nucleus are found the chromosomes, which contain most of our genes – the blueprint of how our bodies developed in the womb, grew, and how they work now.

There are usually 23 pairs of chromosomes, one of the pair inherited from your mother and the other inherited from your father. The genes are strung along the chromosomes like beads on a necklace. We know there are about 30,000 genes in every cell.

Approximately one in 200 children are born with a rare chromosome disorder including material missing, duplicated or re-arranged.

Sometimes, these disorders are not picked up by standard genetic testing at amniocentesis such as the test that diagnoses Down Syndrome – characterised by an extra copy of chromosome 21 – and Edward's Syndrome, the presence of three instead of two copies of chromosome 18. Diagnosis before birth of the some smaller chromosome disorders might be possible within few years as microarray technology is introduced. Microarrays can detect more subtle chromosomal problems than the more conventional tests currently used at amniocentesis – their use in this field is still experimental.

People with any loss or gain of material from chromosomes 1 to 22 will usually have learning disability or developmental delay. This is because there are so many genes located across these chromosomes for normal development of the brain.

EuroGentest is working to help parents of children with rare chromosome disorders and other types of genetic disorder get better counselling, more accurate diagnosis and access to the best and latest genetic testing technology.

Dr Beverly Searle, Chief Executive Officer of Unique, a help group for families affected by rare chromosome disorders, says: This is important because families need to be able to understand what implications the child's disorder has for their development and health. By having the best information possible, families and the professionals who work with them are empowered to provide their children with the most appropriate and timely therapies and treatments to enable them to reach their full potential.

CASE STUDY

'I've been waiting four years for a diagnosis for my son'

Eleanor Fiske, 35, from Suffolk, UK, has an only son, George, aged four. George can neither walk or communicate and has a limited life expectancy. Doctors believe he has a rare genetic disorder but due to lack of funds for testing, they still haven't given Eleanor a diagnosis - and she still doesn't know if she or George's father passed on the disease to him

"After I got pregnant with George, I had a routine test at 12 weeks called the Alpha Foetal Protein (AFP) test.

The results showed abnormally high levels of proteins – which can be a sign of a genetic abnormality.

My obstetrician recommended an amniocentesis - which tests for the most common genetic disorders like Down's and Edwards Syndromes – but the results of this came back as normal.

But when George was born, he failed to thrive, could not feed properly, had facial abnormalities and a significant heart defect. Tests at three weeks showed he was deaf.

The doctors thought he would not survive – which was devastating news.

Genetic counsellors at the hospital I was being treated at offered tests but they explained that looking for a defect in DNA is like looking for a needle in a haystack – there are 27,000 genes, and 300-400 possible variations or deletions of each gene.

What doctors normally do is to look for specific signs of the more common chromosomal disorders and then do specific tests to try to find what has caused the symptoms.

My son shows classic signs of a relatively common genetic disorder affecting one in 3,000 babies believed to be a problem with a gene on chromosome 17 called the fibromatosis gene.

It took two years to be offered a gene test to see if I or George's father carry the same variation (it could just be de novo - ie the variation wasn't passed on genetically but happened spontaneously). Unfortunately, the first test sample was accidentally destroyed and I have recently had a second one – I am now waiting for my results. But in the meantime I don't want to risk having another child like George.

I feel the public is completely ignorant of the rarer genetic disorders – when I am out with George, often people stop in the street to ask me what's wrong with him and when I say I don't know because he hasn't been conclusively diagnosed, they blame me - and say it must have been a virus or that I probably partied too much before I knew I was pregnant. I find this very isolating."

Initial survey shows pressing need for improved patient education materials

EuroGentest aims to structure, harmonize and improve the overall quality of all aspects of genetic services in the EU, by paying substantial attention to the important issues raised by genetic testing. A central component of this work undertaken by Unit 6 concerns 'patient education'.

According to Celine Lewis from GIG: "Genetic counselling and the provision of written patient information plays an important part in informing patients about the various issues that surround genetic testing. Due to the complex nature of the information being given, and the fact that it is often given during a particularly stressful time, it is important that patients and families can go back and read over information relating to issues concerning genetic testing. Written information therefore allows people to go away and have time to think about the main issues. Without this, psychological harm to family members and decisions that may latter be regretted are more likely to occur, as well as misunderstanding complex information. The aim of work package 6.1 is to assess the range of information available to the public relating to genetic testing, and to evaluate it using a specifically designed EuroGentest evaluation tool. This will help determine what patients and families want to know, and whether clinicians and other health professionals are meeting these needs in the different health care systems across the EU. The tool will also, we hope, be useful for creating new education materials. "

It was decided that during the first 18 months five European countries would be focused on, and patient information in those countries would be mapped and assessed. The countries chosen were: the UK, The Netherlands, Belgium, Italy and Sweden. because of the close links Unit 6 and The Genetic Interest Group had with both patient organisations and genetic centres in those countries. Two methods – a questionnaire and on-site interviews - were used to map the range of written information available to patients about genetic testing. Five conditions were chosen to ensure a representative spread of both rare and common conditions, and hereditary patterns. These were - hereditary breast cancer, Duchenne muscular dystrophy, 22q11 deletion, tuberous sclerosis, alteration of the connexin 26 gene.

Celine Lewis busy analysing the patient education material she has gathered from five countries.

A wide and mixed bag collected

A variety of information was gathered. These included:

- Leaflets (either produced by genetic departments, national health services, or patient support organisations)
- Personal letters (produced by genetic departments)
- Standard letters (produced by genetic departments)
- Booklets (either produced by genetic departments, national health services, or patient support organisations)
- Print outs from web pages (either from genetic department websites, national health service websites, or patient support organisation websites)

The information collected fell into three categories.

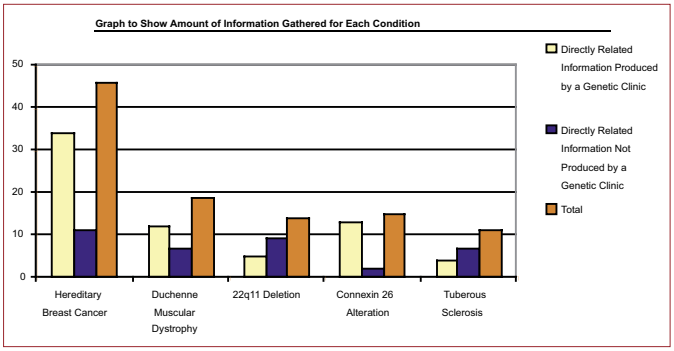
- Information that was directly relevant to the questions asked in the questionnaire, and was produced by the genetic department (e.g. a personal letter written to a patient who was thinking of taking a predictive test for hereditary breast cancer).
- Information that was directly relevant to the questions in the questionnaire, but was not produced by the genetic department (i.e. information produced by support organisations, the national health service etc)
- Information that was indirectly relevant to the questions asked in the questionnaire, produced by either the genetic department or other sources. These included:

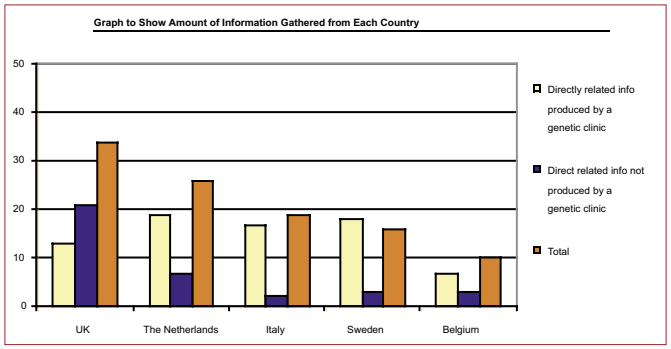
  -a booklet about prenatal testing

  -a general leaflet about genetic counselling

  -a general booklet about chromosome disorders

  -a consent form for a predictive test





According to Celine, some general conclusions can be drawn from the information gathered. "It seems that patients are not always provided with written information before deciding whether or not to take a genetic test. Our questionnaire revealed that only 68% of respondents provided written information before genetic testing. It is difficult to know therefore whether patients are really given the opportunity to go away and think about the main issues concerning genetic testing. Nor are patients considering genetic testing always seen within the genetic department. Often consultants from other specialisations such as paediatrics, obstetrics or neurology will order genetic tests and we do not know what information is given out in these cases."

Turning to the kind of information collected, there was almost twice the amount of information available for hereditary breast cancer than for any of the other conditions, both from genetic clinics, and from patient organisations. This is not particularly surprising considering the condition is also the most prevalent of all five conditions, in all five countries. In addition it was found that numerous charities exist in each country that provide the public with information about genetic testing for breast cancer. This contrasts with the least amount of literature collected overall which was from a genetic clinic for tuberous sclerosis, and then the 22q11 deletion. "This is surprising since prevalence of 22q11 deletion ranges from 1 in 3000 to 1 in 5000, so it occurs more frequently than both Duchenne and the connexion 26 alteration, where there was more information available. This highlights that the amount of patient information available does not necessarily reflect need," continues Celine.

Assessing Patient Information

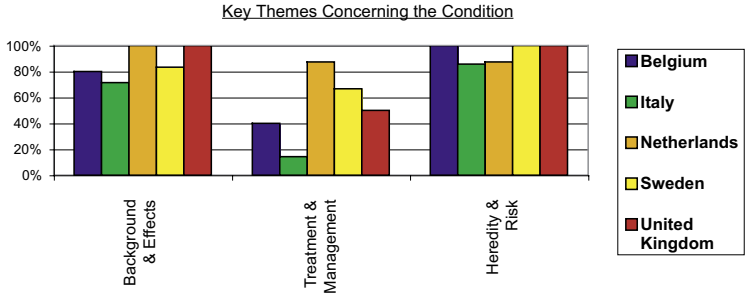
Overall, only three existing tools were found that specifically evaluated patient information about genetic testing and genetic conditions and could be used as starting points:

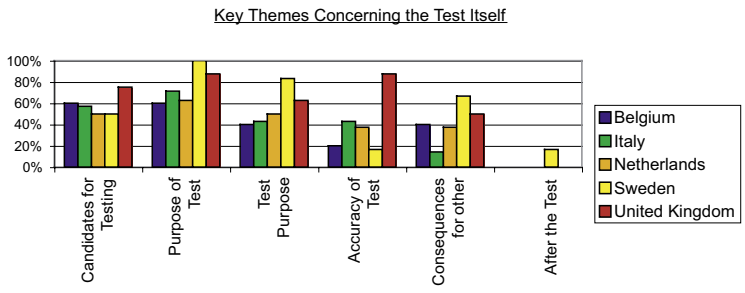
1) DISCERN Genetics (UK): A Validated Appraisal Tool for Judging the Quality of Information for the Public on Genetic Testing and Screening (awaiting publication)

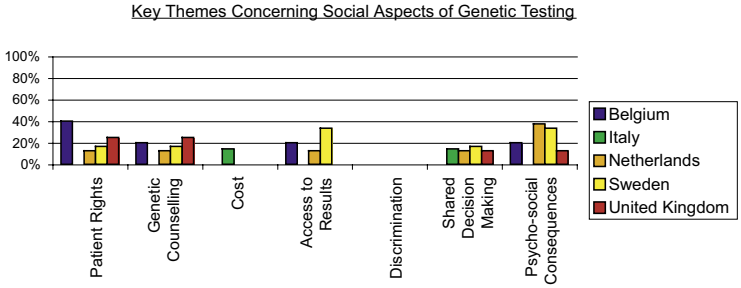
2) Erfocentrum (The Netherlands): Manual for Writing Patient Information for the Website

3) American Journal of Medical Genetics: 'Education Material About Genetic Tests; Does it Provide Key Information for Patients and Practitioners?' 73:314-320 (1997)

The majority of themes used to analalyse the written material had been identified by the Discern tool. There were however a few additional themes (namely 'candidates for testing', 'patient rights' and 'genetic counselling') that had been identified in the American Journal of Medical Genetics. What follows are three graphs that show the frequency of each of the themes within the written material.'







Evaluation of Information

Breast cancer material scores highest

Overall, every piece contained at least some information that would aid patients in deciding whether to take a genetic test or not. Most of the pieces contained information about the condition and some information about the test itself. None of the pieces contained information on all the themes, but the two highest scoring pieces contained information on 16 of the 21 themes. Both these pieces were on hereditary breast cancer. One of the pieces was from the UK, the other from The Netherlands.

"Purpose of the test" usually well covered

The findings can be categorised into three major groups. At the top are a group of four issues that are mentioned the most, between 85%-94% of the time. These are issues that are mainly concerned with the condition itself. "This is to be expected. If one is going to agree to take a genetic test, it is obviously important to know what one is being tested for. Without an understanding of the condition itself (the risk of passing and carrying, the development, the prognosis etc) one would not be able to make an informed decision as to whether it would be beneficial to take the test or not." The issue 'purpose of the test' can also be found in this group. This is the most common issue addressed concerning genetic testing, and can be found in 71% of the information. "It is important information as it informs the patient as to what type of test is being offered (e.g. carrier, diagnostic, prenatal, predictive etc). Only by knowing the issues that surround the exact nature of the test, (e.g. what the test results of a predictive test will tell you, what it does and doesn't mean to be a 'carrier' of a particular genetic mutation, that a prenatal test will be able to tell if your unborn child has a particular genetic condition etc), can you give informed consent to undertake such a test in full knowledge of the implications of the results. Without understanding what the test results might tell you, one could not know whether it was in one's best interests to go ahead with genetic testing," comments Celine.

Surrounding issues less so

The next seven issues are mentioned 41%-59% of the time, i.e. (approximately half the time). This group is mainly concerned with issues concerning the genetic test (bar the purpose of the test). These include: candidates for testing, test procedure, accuracy of the test, and consequences for others of genetic testing. "These issues are again important to consider when making an informed decision about genetic testing. For example, one would be far more inclined to take a genetic test in the knowledge that it will be 100% accurate than if the test is only 50% accurate, as otherwise the results would not be particularly conclusive. Again, one might be far more inclined to take a genetic test if there is no health risk attached, than if a slight risk exists, (as is the case with an amniocentesis). Hence these are all issues that are important to be informed about when considering genetic testing," continues Celine.

Social aspects mainly lacking

The last nine issues are mentioned fairly infrequently, in 0%-21% of the time. This group of less frequently mentioned issues are mainly concerned with the social aspects of genetic testing, e.g. the psychosocial consequences of genetic testing, patient rights, genetic counselling, shared decision making, discrimination. It is interesting to note that in this bottom group, virtually all of the statements describing what can be considered 'social aspects of genetic testing' are from leaflets/letters on hereditary breast cancer. "There are various reasons as to why this might be so. Information concerning hereditary breast cancer might be better informed than for the other conditions, due to its prevalence in the population. In addition numerous studies have been done assessing what key issues hereditary breast cancer patients want to be informed of. Another reason might be because for certain genetic tests, (such as a diagnostic test for 22q11 deletion), the test itself might not pose any grave social consequences for the child or parents. A parents' first priority will most probably be to get a diagnosis confirmed, and therefore the issue that for example, testing is voluntary, might be insignificant. It is important to be aware therefore that every issue is not necessarily relevant in every case. The issue of 'access to test results' i.e. to whom test results can be disclosed, will probably be a far more significant issue to someone who might be taking a predictive test for hereditary breast cancer, than for someone taking a diagnostic test for the Connexin 26 alteration. A family that already have a child affected with a condition such as Duchenne muscular dystrophy will be extremely familiar, undoubtedly experts, in the social aspects of the condition (issues to do with schooling, health insurance etc) hence it might not be appropriate to reiterate this kind of information to them. The question of which issues are relevant to address is very much dependant on the particular case in hand. It is important to remember therefore that any guidelines used to assess patient information can only be used as 'guidelines', and the circumstances of each case need to assessed individually".

"Nethertheless, social and psychological repercussions are likely to occur for tests other than predictive ones," Celine believes. "Testing positive as a carrier for any condition may result in feelings of guilt or anger. Positive prenatal test results will undoubtedly cause a plethora of mixed emotions, with the added difficulty of having to make extremely tough decisions. Genetic testing might also inadvertently disclose information about relatives or parentage, or cause complications when buying health and life insurance. For these reasons and more, it is important that the social and psychological aspects for all kinds of genetic testing are brought to light and discussed before any definite decisions are reached. It will be important to find out from patients themselves in the next phase of research, just how important this group of issues are to patients, because what the results do show is that these issues are mentioned infrequently, especially outside of information about hereditary breast cancer."

Another consideration is that certain issues may only be relevant in certain countries. The issue of cost is one such example. In the UK it is standard procedure that a genetic test within the NHS will be free, and hence will not be an important issue to consider. This will however not necessarily be the case for a patient in Italy who is being seen privately. Here, cost will be an important consideration to take into account when considering genetic testing.

Celine suspects that practitioners might also not have information concerning certain issues. The issue of 'discrimination' is one such example, and might be the reason that it was not mentioned in any of the pieces analysed (it might also be because it is not considered an important issue to address by practitioners). Practitioners might not be aware of the particular employment and insurance issues that surround particular genetic conditions or the results of genetic tests. It might be considered the role of patient organisations or social service networks to address these issues.

Work to be done

Overall, Celine believes the findings show that there is a great disparity within the quality of information patients are provided with when considering genetic testing. "This varies from both country to country, and condition to condition. In addition, procedures differ across the countries concerning when written information is provided about genetic testing, who provides it (if any is provided), and in what forms it is available in. The next phase of research will help refine the evaluation tool that has been created, so that recommendations can be made as to the key issues patients want to be informed about when considering genetic testing," she concludes.

Letter from America

EuroGentest has strong links with both the US and Japan with for example joint attendance at meetings and sharing of information. Here we start a regular series of features on both areas of common interest and current issues in genetics in the US as reported by our contacts in the US Centre for Disease Control.

QC Material Development Efforts in the United States

The U.S. Centers for Disease Control and Prevention (CDC) has been involved for many years in efforts to develop appropriate and verified quality control materials for use by the genetic testing community. Recently, a new CDC-based program, the Genetic Testing Quality Control Materials Program (GTQC), has been established in partnership with the U.S. genetics community. This program coordinates a self-sustaining community-based process to improve the availability of appropriate and verified materials for quality control, proficiency testing, test development/validation, and research purposes. The GTQC program also maintains a website to facilitate and coordinate information exchange between users and providers of QC materials. This program is coordinated by the CDC, but all of the actual work, including decisions about QC material priorities, validation schemes, specimen collection, and material development and verification, occurs through voluntary collaboration of CDC with laboratories in the genetics community.

The GTQC Program is currently developing verified genomic DNA based QC materials for fragile X, disorders on the Ashkenazi Jewish Panel (Bloom syndrome, Canavan disease, Fanconi anemia, Familial dysautonomia, Gaucher disease, mucolipidosis IV, Neimann-Pick disease and Tay-Sachs disease), cystic fibrosis, and Huntington disease. These materials will be publicly available from Coriell Cell Repositories (http://locus.umdnj.edu/ccr/) as an international resource.

The first GTQC Expert Panel Meeting, organized by the CDC, was held on November 29, 2005, in Turnhout, Belgium, in conjunction with the First International Symposium on Reference Materials for Genetic Testing organized by EuroGentest and the Institute for Reference Materials and Measurements (IRMM). The 14 meeting participants included U.S. experts in genetics and genomic testing from professional organizations, government agencies, industry, commercial and academic clinical laboratories, and cell repositories. The main goals of the meeting were to assess the progress of the GTQC Program, discuss plans for the coming year and explore ideas for potential interactions and collaborations with international QC material development efforts such as EuroGentest.

The participants expressed positive comments on the progress of the GTQC Program in its first year of efforts. They felt that the program should continue with its current projects and discussed new areas of focus for the coming year. The group suggested tests they felt are in immediate need of QC material development. These include: pharmacogenetic tests, disorders included on The College of American Pathologists (CAP) proficiency surveys, disorders on newborn screening panels, and other genetic disorders (including: hereditary breast/ovarian cancer, spinal muscular atrophy, conexin 26, associated polyposis conditions, multiple endocrine neoplasia type 2, and Rhett syndrome). The participants discussed the importance of developing collaborations between GTQC, EuroGentest and other international groups. Such partnerships will allow better use of available resources and prevent duplication of effort. Suggested international collaborative efforts include:

- Sharing of QC material resources, such as cell lines
- Sharing of proficiency testing data and needs analysis survey results
- Cooperation on joint QC material development projects
- Development of consensus/agreement documents

Information about the GTQC Program and currently available materials (as well as other QC info) can be found on the GTQC Program website: http://www.phppo.cdc.gov/dls/genetics/qcmaterials/default.aspx .

EuroGentest recently held its second Annual General Assembly in the historic setting of Leuven. Pictured here are the steering committee.

The Catholic University of Leuven (K.U.Leuven) will confer an honorary doctoral degree on Prof. Mary-Claire King from Seattle (Washington, US). This happens, as usual, on the occasion of the anniversary of the University, on February 2.

Professor Mary-Claire King.

Prof. King has been presented as a candidate for this honorary degree by the Center for Human Genetics in Leuven.

Mary-Claire King has a degree in mathematics and obtained her PhD in genetics in 1973 at the University of California in Berkeley. After a post-doc in San Francisco and a short stay in Chili, she returned to Berkeley in 1976 to become a professor in genetics and epidemiology. In 1995, she moved to Seattle, where she became the American Cancer Society Research Professor at the Departments of Medicine (Medical Genetics) and of Genome Sciences of the University of Washington, and an affiliate member of the Fred Hutchinson Cancer Research Center.

She worked initially in population genetics and evolution, and she published original work on the genetic difference between humans and chimpanzees. In the early seventies, she initiated epidemiological research on cancer, breast cancer in particular. In 1990, her laboratory located the first genetic defect for familial breast cancer on chromosome 17. With this discovery, she led the way to the positional cloning of the BRCA1 gene. The gene was eventually cloned in 1994. She is still active in the field of breast cancer, but she has also taken up interest in other scientific topics among which deafness and susceptibility to HIV infection and AIDS. She became famous for her forensic work in different parts of the world, and most particularly in Argentina, where she assisted the 'grandmothers of the Plaza de Mayo' to reunite them with their grand-children.

Mary-Claire King has also expressed her explicit support to her European colleagues who opposed against the monopoly on the genetic test for breast cancer, which resulted from the patenting of the BRCA1 and BRCA2 genes.

EUROGENTEST PEOPLE

EuroGentest has attracted many of the leading names in genetics in Europe. Equally exciting though are the number of enthusiastic young scientists – from recent graduates to PhDs - who have joined reflecting the growing popularity of genetics as a discipline.

NAME
Jana Camajova

NATIONALITY
Slovak

QUALIFICATION
PhD in Molecular Biology

LANGUAGES
Slovak, Czech, English, German

FAVOURITE MUSIC
Rock music, for example a Czech band Lucie. And I am also fond of listening to organ and guitar instrumental compositions.

HOBBIES
I love mountain hiking and photography.

LAST BOOKS READ
James Herriot's "It shouldn't happen to a vet" series and Dr Zhivago.

Q: Why did you get involved in Genetics?

A: I had been keen on natural sciences since being a little girl. I was so fascinated by how everything can function on the base level that I decided to study Molecular Biology and Genetics – in Commenius University in Bratislava. First I worked in the basic/ fundamental research field, then my interest shifted more to human disease diagnostics to be closer to the patients. I attended to nephrogenous diseases in Germany. At last I came to Prague to work in Milan Macek's department.

Q: What do you see the benefits of EuroGentest being?

A: Genetics is exciting and fast moving. By pulling our resources together we can respond better and benefit faster from new ideas and experiences. EuroGentest is a perfect way of doing this since there are so many experts from all over Europe involved and such a good spirit of cooperation.

Press Watch

Avian flu and the stem cell scandal in South Korea have dominated science coverage over Christmas, but there were also some interesting articles in which experts were invited to make their predictions for the year ahead.

In an article for the UK Independent Newspaper for example it was predicted that microarray technology would come up with genetic-based tests for "common" diseases such as cardiovascular and cancer already this year. There was also a great deal of excitement about biobanks and the possibilities of understanding the genetic basis of behaviourial disorders such as autism. Of course experts are entitled to their opinion, but raising public hopes in this way is not always wise

December 28 2005 The Washington Post, US

Act now to end genetic discrimination

This opinion column by Susanne B Haga congratulates the computer giant IBM for agreeing not to use genetic information in its hiring practices or in deciding eligibility for health insurance coverage for its 300,000 employees. Such action highlights the gap in federal legislation to adequately protect job candidates and employees from discrimination based on their personal genetic information, argues Haga. She points out that at least 20 bills have been introduced in the US Congress to prohibit genetic discrimination in the past 10 years, but only one has passed. "National legislation to prohibit genetic discrimination by employers and health insurers is way overdue", Haga concludes.

December 30 2005 The Financial Times, London, UK

Disgraced stem cell scientist further discredited

Hwang Woo-suk, South Korea's disgraced scientist, suffered another devastating blow to his reputation yesterday, says the FT, when an investigative panel at Seoul National University said there was no evidence for his research team's claims to have derived stem cells from cloned human embryos.

The latest findings will depress scientists worldwide, reports the newspaper, who had cited Professor Hwang's research to support the idea that therapeutic cloning could produce "patient-specific" stem cells to treat degenerative diseases such as Parkinson's and diabetes, without the risk of rejection by the immune system. No researchers elsewhere have successfully produced stem cells from cloned embryos.

Given the strong financial and moral support previously given to Prof Hwang by the South Korean government and people, his disgrace will "inevitably damage the country's credentials as a location for world-class biomedical science".

December 30 2005 The Baltic News Service

Napoleon's Troops in Lithuania Died of Infection

One-third of several thousand of Napoleonic soldiers whose remains have been discovered at a mass grave site in Vilnius might have died of infectious diseases spread by lice, reports the Baltic News Service.

Rimantas Jankauskas, senior lecturer at the Department of Anatomy, Histology and Anthropology of the Medicine Department of Vilnius University, said that French and Lithuanian scientists had discovered traces of vectors of spotted typhus and the so-called trench typhus - also widespread during World War I.

January 17 2006 Glasgow Herald, UK

Breakthrough as team find crucial diabetes gene

The gene for Type II diabetes - one of the holy grails of genetic science - has been discovered by the deCODE project in Iceland.

The gene is involved in the regulation of glucagon, a hormone that raises blood sugar levels and raises the risk of developing Type II diabetes by between 45 to 140 per cent.

January 17 2006 The New York Times, US

Genetic Testing Creates New Versions of Ancient Dilemmas

This feature, written by Robert Klitzman from the Columbia University Center for Bioethics discusses the tensions that arise when there's a disagreement among family members over whether one of them should be tested for a genetic predisposition to disease.

January 13 2006 The National Post, Toronto, Canada

Hospital pays out after misdiagnosing toddler

The newspaper reports that Toronto's Hospital for Sick Children has paid just under seven thousand Canadian dollars $7,000 (3,993) to the parents of a child who was misdiagnosed with a hereditary mutation for a rare eye cancer (retinoblastoma). The award was made because of the number of times the child was anaesthetised so his retinas could be examined for tumours. The false diagnosis also prompted doctors to stop screening Travis's two older sisters, both of whom should have been checked regularly for retinoblastoma. A positive test for the mutation makes eye cancer in siblings unlikely, says The National Post.