

Information for Patients: Quality and Communication

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CAPABILITY 2008



Importance of written information

- Genetic literacy amongst the general population is relatively low.
- Written info has been shown to improve patient satisfaction and knowledge of complex issues.
- Good quality information can allay anxiety.
- Can improve decision-making process necessary for giving informed consent to genetic testing.
- Visual aids (e.g. for inheritance patterns) are important part of the understanding process.

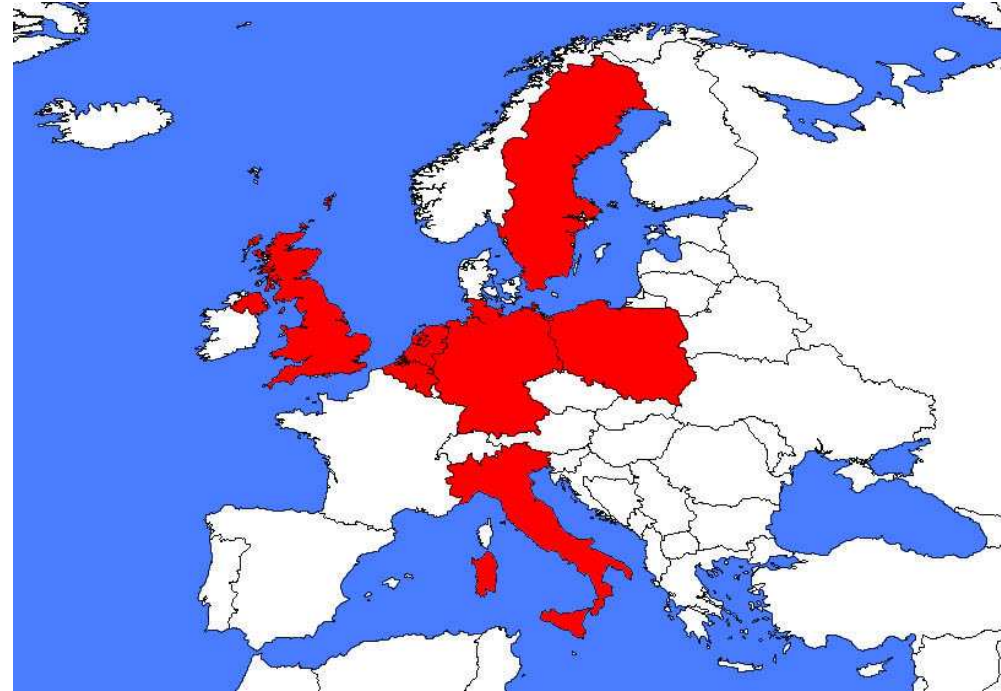
EUGT survey 2005-6

Assessed the quality of written patient information provided at the genetic clinic and relating to genetic testing in seven European countries.



Methods

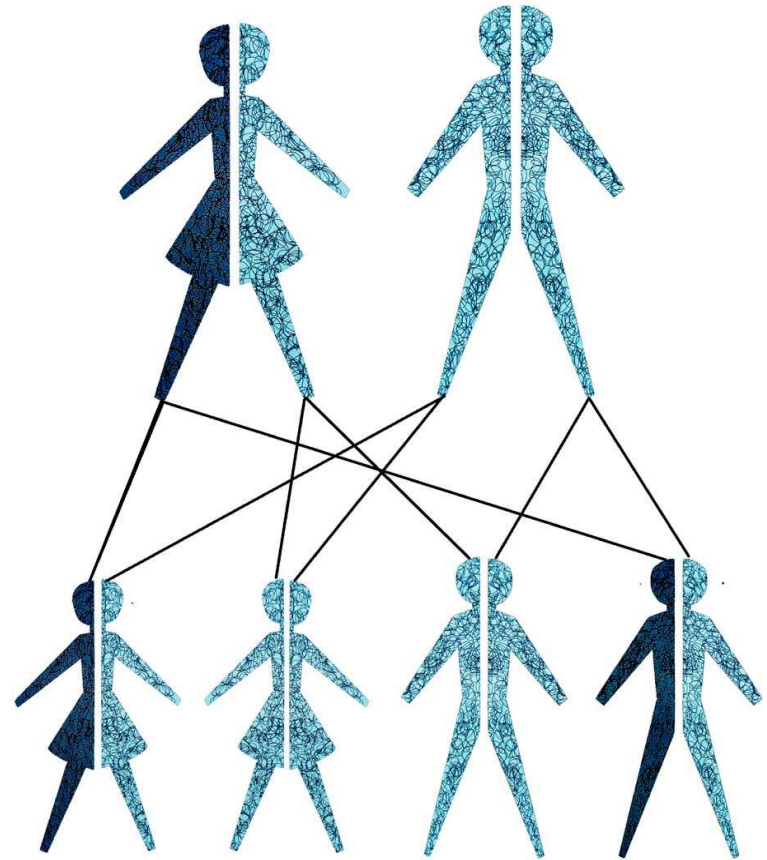
- 7 countries chosen :
UK
Netherlands
Belgium
Sweden
Germany
Poland



- Countries chosen due to variation in **geographical location, health and political systems, and genetic service development.**
- Contacted 2-3 genetic clinics in each country.

Methods

- Collected written info on genetic testing for 5 conditions:
hereditary breast cancer
Duchenne muscular dystrophy
22q11 deletion
tuberous sclerosis
connexin 26 alteration
- Range of conditions in terms of:
inheritance patterns
age of diagnosis
common and rare conditions



Methods

- Tried to gather 2 pieces of written information for each condition, from each country.
- 50 pieces assessed in total made up of **personal letters** and **pre-written leaflets**.
- Used number of tools to identify key issues that should be discussed in genetic consultation, in particular Discern Genetics (Oxford). **14 key issues** identified.

Written information assessed for the inclusion or omission of 14 key issues

Information about the condition:

- Background and Effect of Condition
- Treatment and Management
- Heredity and Risk

Information about the test:

- Type of Test
- Test Procedure
- Accuracy of Test

Psychosocial Issues:

- Psychological consequences both +ve and -ve e.g. relief, guilt, anxiety
- Consequences for relatives and partner e.g. what increased risk means for other family members

Key Findings:

- Written information more likely to discuss hard, factual information related to the condition and test, than more qualitative, experience based information related to the psychological and social implications of genetic testing.
- risks and limitations of genetic testing were infrequently discussed.
- pre-written leaflets provided a far more comprehensive discussion of key issues than personal letters.
- Pre-written information was more readily available in Western Europe than in Central and Eastern Europe.

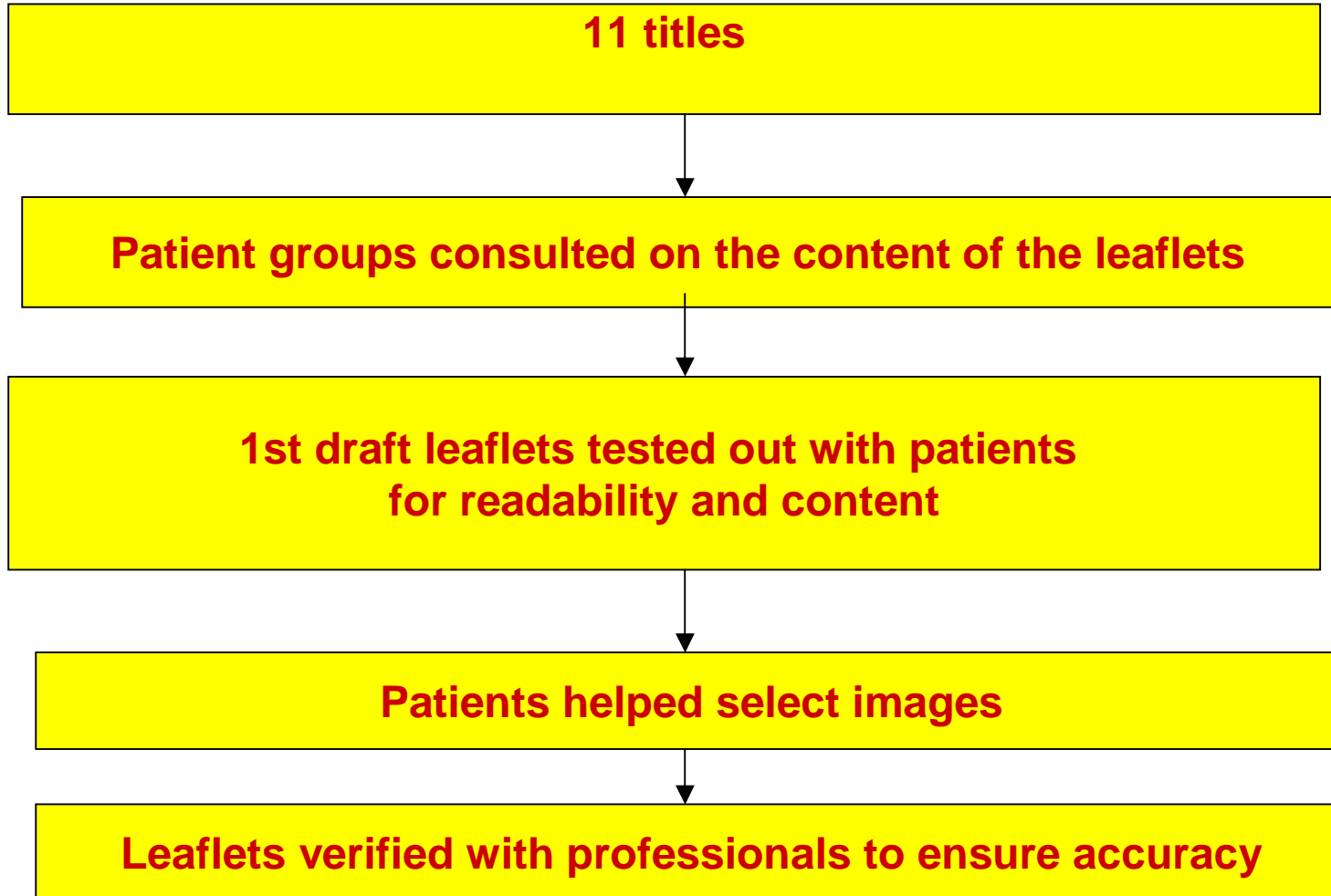
In Response.....

- 1) Develop pre-written information leaflets for patients and families about issues related to genetic testing.
- 2) Develop these with the help of patients and families to ensure they are 'patient focused'.
- 3) Ensure they address issues that were repeatedly omitted in information assessed.
- 4) Translate leaflets and make them freely available in genetic clinics across Europe.

Patient Leaflets

- Survey of genetic specialists across the E.U.
- Information clinicians indicated most useful:
 - What is a genetic test?
 - Inheritance patterns
 - Information about chromosomes
 - Predictive and prenatal testing
 - Genetic glossary
- Patient Group representatives:
- Frequently Asked Questions About Genetic Testing
- Benefits, Risks and Limitations of Genetic Testing

Development of Leaflets



Some Information About Your Genetic Appointment



Information for Patients and Families

What is a Genetic Test?



Information for Patients and Families

Chromosome Changes



Information for Patients and Families

Recessive Inheritance



Information for Patients and Families

Recessive Inheritance

The following will give you information about what recessive inheritance means and how recessive conditions are inherited. To understand recessive inheritance, it is first helpful to know about genes and chromosomes.

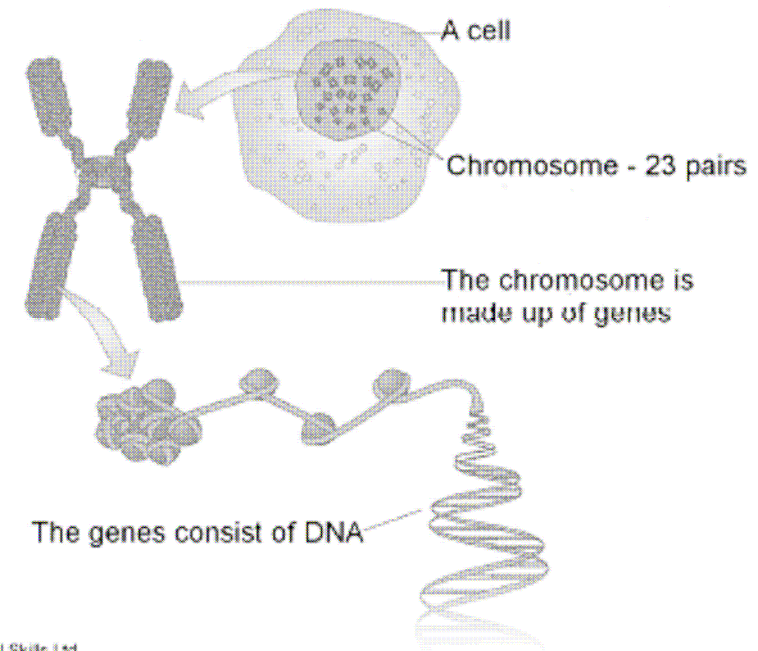
Genes and Chromosomes

Our bodies are made up of millions of cells. Most cells contains a complete set of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are also responsible for many of our characteristics, such as our eye colour, blood type and height. We have thousands of genes. We each inherit two copies of most genes, one copy from our mother and one copy from our father. That is why we often have similar characteristics to both of them.

Genes are located on small thread-like structures called chromosomes. Usually we have 46 chromosomes in most cells. We inherit one set of 23 chromosomes from our mother and one set of 23 chromosomes from our father. So we have two sets of 23 chromosomes, or 23 pairs.

Sometimes, there is a change (mutation) in one copy of a gene which stops it from working properly. If this occurs in only one recessive gene, and the person has another normal copy, this will not usually cause a genetic condition.

Picture 1: Genes, chromosomes and DNA



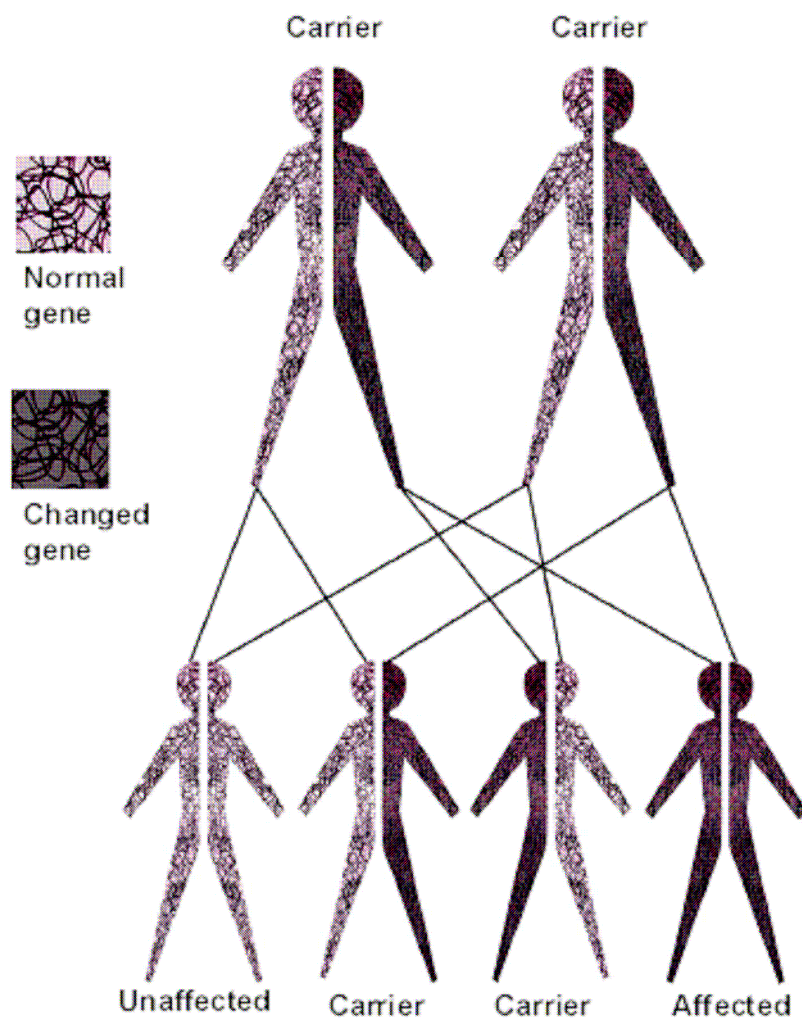
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What is autosomal recessive inheritance?

Some conditions are inherited as recessive conditions. This means that a person must inherit two changed copies of the same gene (one changed copy from each parent) in order to have the condition. If a person inherits one changed copy and one normal copy, then in most cases that person will be a healthy carrier because the normal copy compensates for the changed copy. Being a carrier means that you do not have the condition, but carry a changed copy of the gene on one of a pair of genes. Examples of autosomal recessive conditions include cystic fibrosis and sickle-cell anaemia.

How are recessive conditions inherited?

Picture 2: How recessive conditions are passed on from parent to child



If both partners are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child. This occurs randomly.

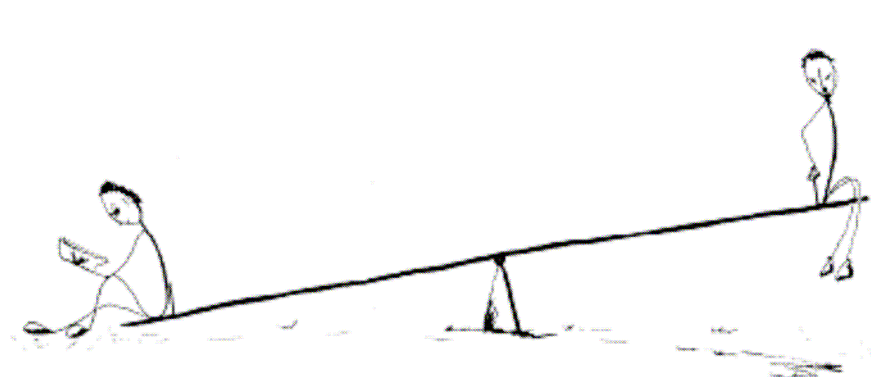
Each child of parents who both carry the same changed gene therefore has a 25% (1 in 4) chance of inheriting a changed gene from both parents and being affected by the condition.

This also means that there is a 75% (3 in 4) chance that a child will not be affected by the condition. This chance remains the same in every pregnancy and is the same for boys or girls.

There is also a 50% (2 in 4) chance that the child will inherit just one copy of the changed gene from a parent. If this happens, then they will be healthy carriers like their parents.

Lastly, there is a 25% (1 in 4) chance that the child will inherit both normal copies of the gene. In this case the child will not have the condition, and will not be a carrier.

These possible outcomes occur randomly. The chance remains the same in every pregnancy and is the same for boys and girls.





Carrier Testing and Tests in Pregnancy

A number of options may be available for people who have a family history of a recessive genetic condition. Carrier testing may be available to see if the couple are both carriers of the changed gene. This information may be useful when planning pregnancies. For some recessive conditions, it is possible to have a test in pregnancy to see if the baby has inherited the condition (more information about these test are available in the CVS and

amniocentesis leaflets). This is something you should discuss with your doctor or healthcare professional.

Other Family Members

If someone in the family has a recessive condition or is a carrier, you may wish to discuss this with other family members. This gives other family members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. This might be particularly important to family members who already have children, or are likely to have children in the future.

Some people find it difficult to tell other members of the family about a genetic condition. They may be worried about causing anxiety in the family. In some families, people have lost touch with relatives and may feel it is difficult to contact them. Genetic specialists often have a lot of experience with families in these situations and may be able to offer you help in discussing the situation with other family members.

Points to remember

- A person must inherit two copies of a changed gene, one from each parent, in order to be affected by the condition (25% chance). If a person inherits only one changed gene then they will be a carrier (50% chance). These outcomes occur randomly. They remain the same in every pregnancy and are the same for boys and girls.
- A changed gene cannot be corrected – it is present for life.
- A changed gene is not something that can be caught from other people. They can still be a blood donor, for example.
- People often feel guilty about a genetic condition which runs in the family. It is important to remember that it is no-one's fault and no-one has done anything to cause it to happen.



This is only a brief guide to recessive inheritance. More information can be obtained from your local regional genetics centre (www.gig.org.uk/services.htm) or from these addresses:

The Genetic Interest Group

Unit 4D, Leroy House,
436 Essex Rd.,
London, N1 3QP
Telephone: 0207704 3141
Provides information about
specific genetic conditions and
contact details of support
organisations.
Email: mail@gig.org.uk
Web: www.gig.org.uk

Contact a Family

209-211 City Rd.,
London,
EC1V 1JN
Helpline 0808 808 3555 or
Textphone 0808 808 3556
(Freephone for parents and
families, 10am-4pm Mon-Fri,
4.30pm-7.30pm Mon)
Email: info@cafamilv.org.uk
Web: www.cafamilv.org.uk

Orphanet

Free-access website providing
information on rare diseases
and orphan drugs, and links to
support groups across Europe.
Web: www.orpha.net

EuroGentest

Free-access website providing
information about genetic
testing and links to support
groups across Europe.
Web: www.eurogentest.org

Modified from leaflets produced by Guy's and St Thomas' Hospital, London; and the London IDEAS Genetic Knowledge Park, according to their quality standards.

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Translations

Adapt information to ensure it is correct for the country it will be used in (change information about waiting times for results, contact details of support organisations etc)



Translations done through bilingual genetic specialist (PHD student or genetic professional)



Translations formatted into leaflets. Diagrams added



Final check by translator and another genetic specialist

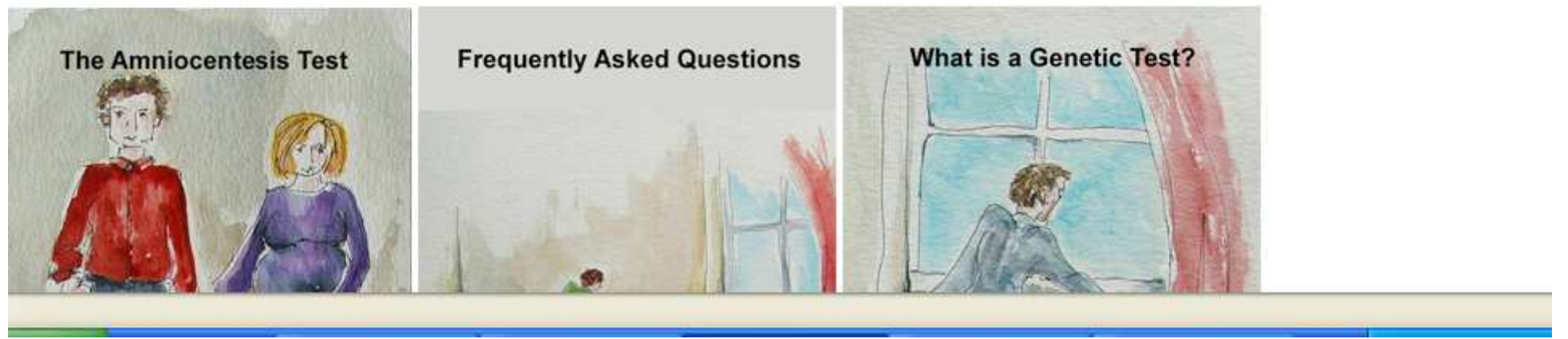


Disseminate leaflets through genetic clinics, national genetics societies and EuroGentest website

INFORMATION FOR PATIENTS AND FAMILIES

Select your language

English	Polski	българин	Türkçe	Român	Islenska	Eesti	Nederlands	Deutsch
								
Finnish								
								



HASTALAR VE AILELER İÇİN BİLGİLER



Aşağıdaki PDF dosyalarından bazıları 1 MB büyüklüğündedir ve açılması biraz zaman alabilir.

Eğer dosyanın açılmasında bir sorunla karşılaşırsanız, linki farenin sağ tuşu ile işaretleyiniz ve "Farklı kaydet" seçeneği ile dosyayı kendi bilgisayarınıza kayıt ediniz. Sonra dosyayı kendi bilgisayarınızdan açabilirsiniz.

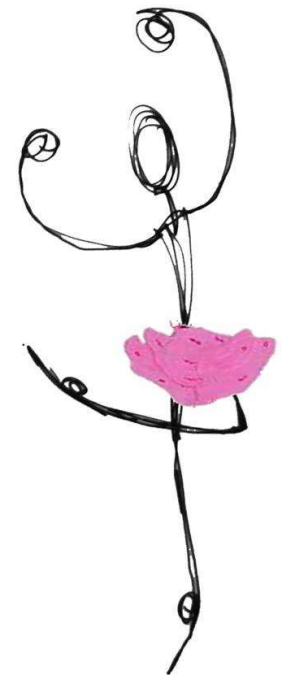
Leaflets	Download as:
Amniyosentez	PDF HTML
Kromozom Translokasyonları	PDF
Kromozom Değişimleri	PDF
Koryonik Villus Biyopsi (CVS) Testi	PDF
Dominant (Baskın) Kalıtım	PDF
Genetik Test Hakkında Sıklıkla Sorulan Sorular	PDF
Genetik Sözlük	PDF
Resesif (Çekinik) Kalıtım	PDF
Randevunuz Hakkında Bazı Bilgiler	PDF
Genetik Test Nedir?	PDF HTML
X'e Bağlı Kalıtım	PDF

Languages

1. BULGARIAN
2. CZECH
3. DUTCH
4. ENGLISH
5. ESTONIAN
6. FINNISH
7. GERMAN
8. ICELANDIC
9. POLISH
10. ROMANIAN
11. SPANISH
12. SWEDISH
13. TURKISH

• **Coming soon.....**

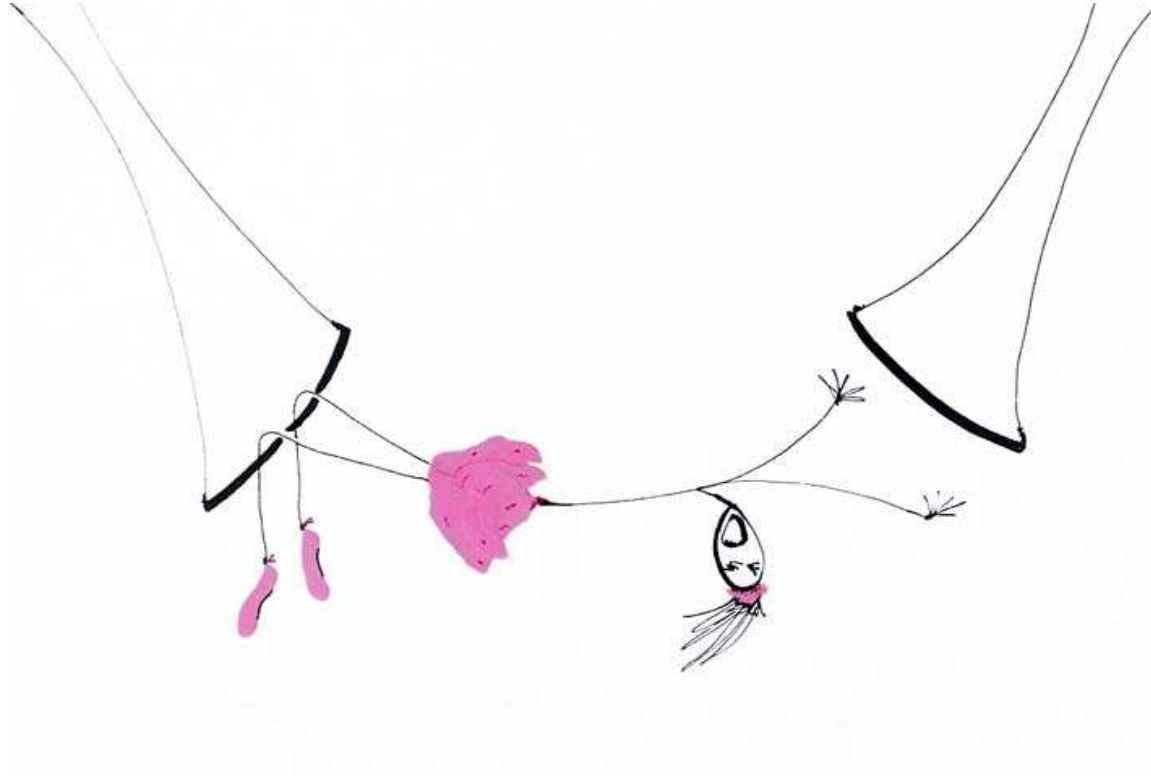
14. ARABIC
15. FRENCH
16. GREEK
17. HUNGARIAN
18. ITALIAN
19. PORTUGUESE
20. SLOVAKIAN



New for 2008

- What happens in the Genetics Lab?
- Predictive Testing
- Carrier Testing
- Non-Diagnosis after Genetic Testing





www.eurogentest.org/patients