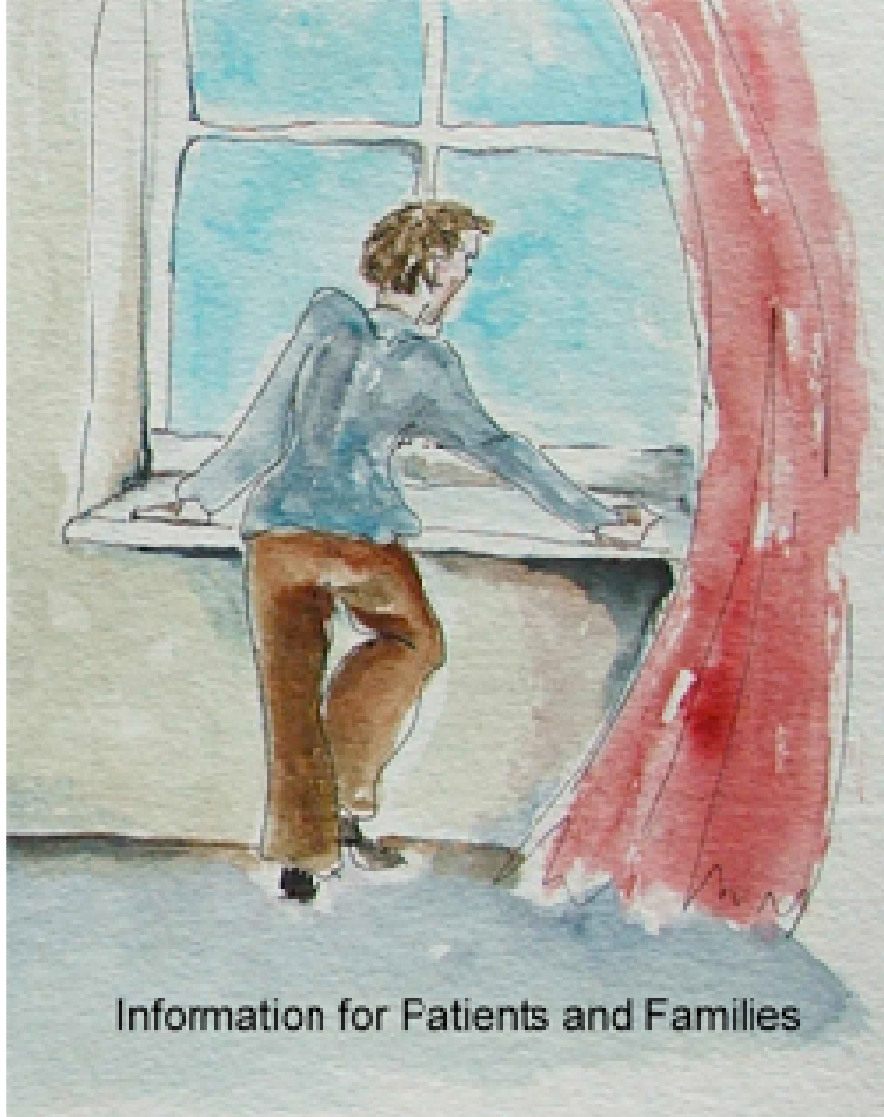


What is a Genetic Test?



Information for Patients and Families

Perspectives from Human Genetics for Public Health

EHF Gastein, October 5, 2007

Jean- Jacques Cassiman
Catholic University of Leuven, Belgium

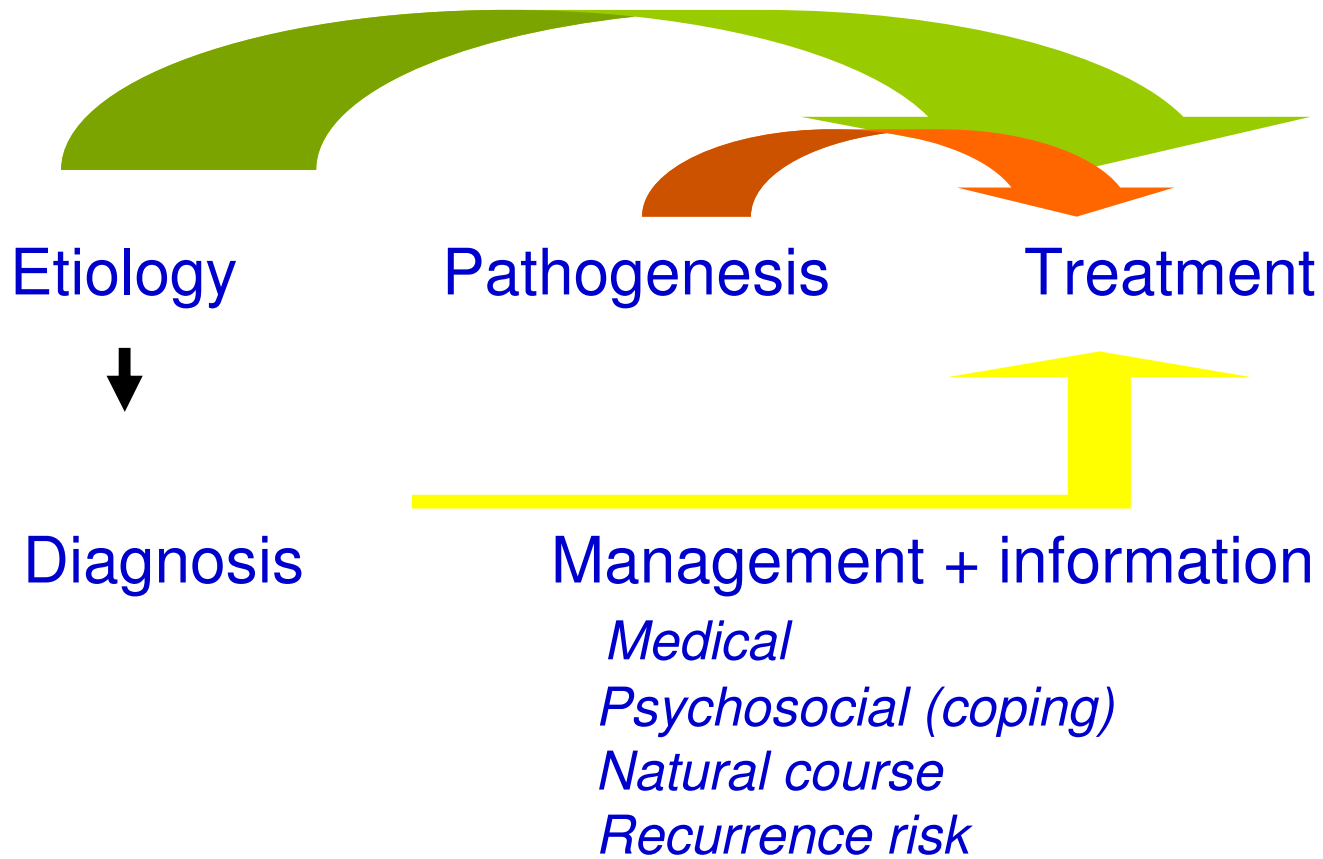


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What are the aims of the geneticists?

- to provide quality genetic information and services to everybody
- to integrate new approaches and technologies in an efficient and ethically responsible way into the genetic services
- to collaborate with colleagues and customers to integrate the knowledge acquired in human genetics into all medical specialties





Procedures in genetic services

- Intake: family history, clinical evaluation, procedure, risks, potential impact on the individual and family
- Sampling: blood, saliva, tissue, amniotic fluid, blastomere....
- Counseling: explain the meaning of the result, the consequences for management and for family; later follow up



Genetic testing

- For what purpose?

Diagnosis, prediction, susceptibility, paternity, forensics, lifestyle

- At what age?

Adult, child, fetus, embryo



GENETIC TESTS

Diagnostic tests : yes / no



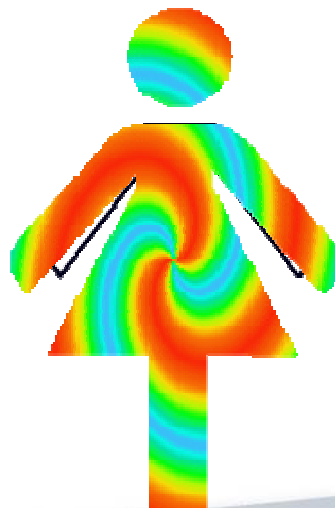
you are healthy
carrier



you are sick

Predictive tests

**Presymptomatic tests:
yes / no**



you will be sick

**Susceptibility tests:
possibly**



your risk of
becoming ill is high



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What is tested in Europe?

- 12,000 monogenic diseases listed in OMIM (Online Mendelian Inheritance in Man ; www3.ncbi.nlm.nih.gov/Omim/)
- 7000 disease loci localized on a particular chromosomal region
- ~ 2000 genes identified
- ~ 100 - 150 genes tested routinely; most others on a research basis



The most popular genetic tests

- Cystic Fibrosis
- Hemochromatosis
- Fragile X mental retardation
- Factor V leiden
- BRCA 1 & 2



Potential impact of genetic tests on European health budget

30 million potential ? customers

- 500 – 4000 molecular tests/ million citizens / year
- 4000 – 10.000 cytogenetic tests/million citizens/ year

200 – 2000 € per test

0.9 – 28 K € / million citizens/ year

! Counseling poorly funded



The ACCE model

- **Analytical validity:** accuracy of test identifying biomarker
- **Clinical validity:** relationship between biomarker and clinical status
- **Clinical utility:** likelihood that test will lead to improved outcome
- **Ethical, legal and societal implications**



What are the potential problems ?

Informed consent:

- concerning the procedures
- concerning the individual investigations
- concerning his/her family
- concerning minors and people unable to consent
- concerning the use of biological materials in the future



ACCESSIBILITY

- access for individuals, for referrals, in context of health problems
- non-discriminatory – financial, social (ethnic minorities)
- not leading to discrimination (social, insurance, employment)
- outreach services



UNRESOLVED ISSUES

diagnosis vs research

patents - licenses

DNA banking

networking

screening

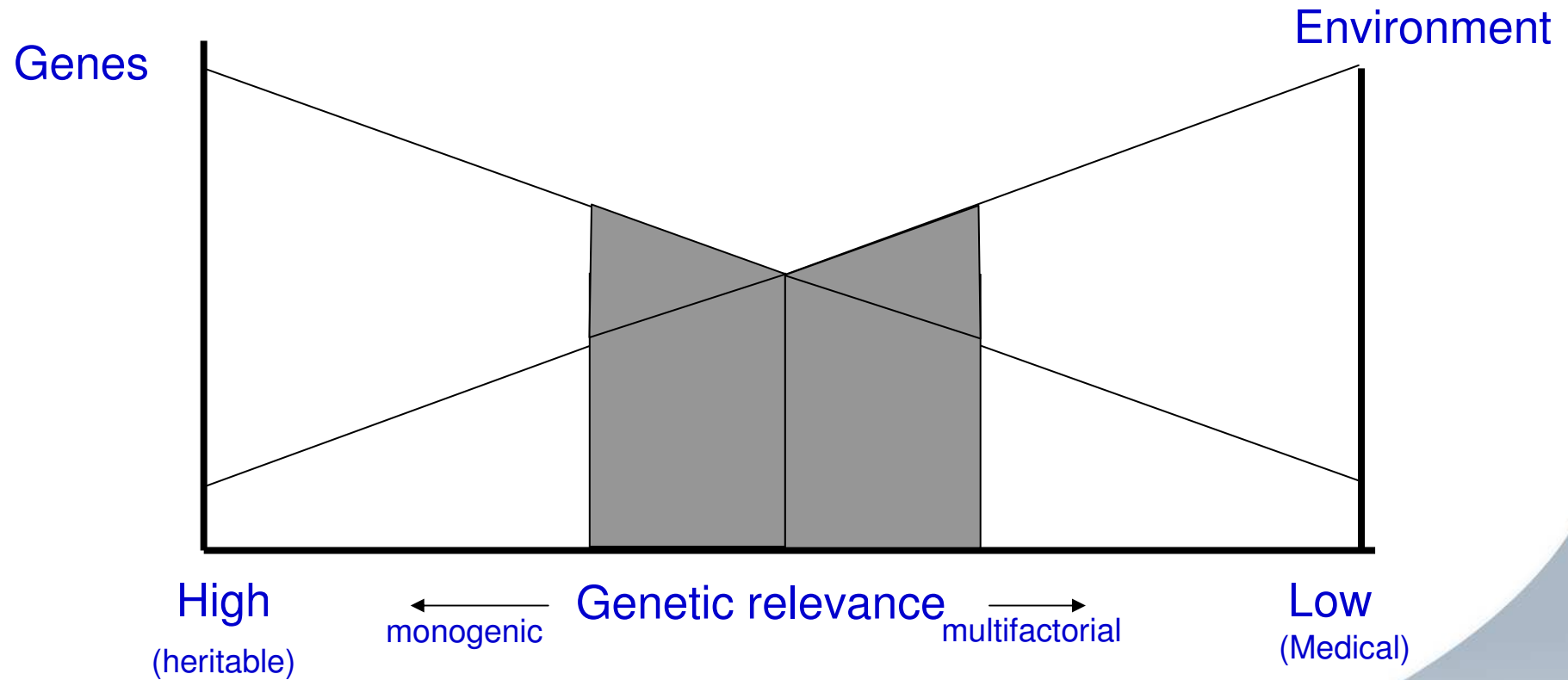


Genomic testing technologies

- Genome changes (multiple genes, mutations variants)
- Epigenetic changes (inhibition or release of inhibition of genes)
- Expression profiles (quantitative measure of transcription of active genes)
- Proteomics (quantitative/qualitative changes in protein profiles)
- Metabolomics (peptides and metabolites)



Information content of DNA test



Genetic tests = Medical test?



Is genetic information different from other medical information?

1. Implications for family
2. No change in time
3. Can determine fate of pregnancy
4. Often complex interpretation

But information content can be = other medical tests

Providing:

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Predictive testing is offered with the same care and information (counseling) as is provided in genetic testing

« Not the testing is unique, it is the approach »



Priorities for Future Action

3rd progress report on the implementation of the European Strategy on Life Science and Biotechnology 29.06.2005

The Commission will

- ▶ launch an initiative on the protection of workers' personal data in the employment context, taking account of the European Group on Ethics in Science and New Technologies Opinion No 18 “Ethical Aspects of Genetic Testing in the Workplace”. The initiative will also address the processing of genetic data,
- ▶ analyse the possibility of setting standards on genetic testing under Article 152 or 153 of the Treaty and the appropriate legal instrument
- ▶ analyse the Directive 98/79/EC on in vitro diagnostic medical devices in the context of genetic testing and in particular regarding quality and performance assurance of genetic test devices
- ▶ launch a mapping and networking exercise on public health aspects of genetic testing.

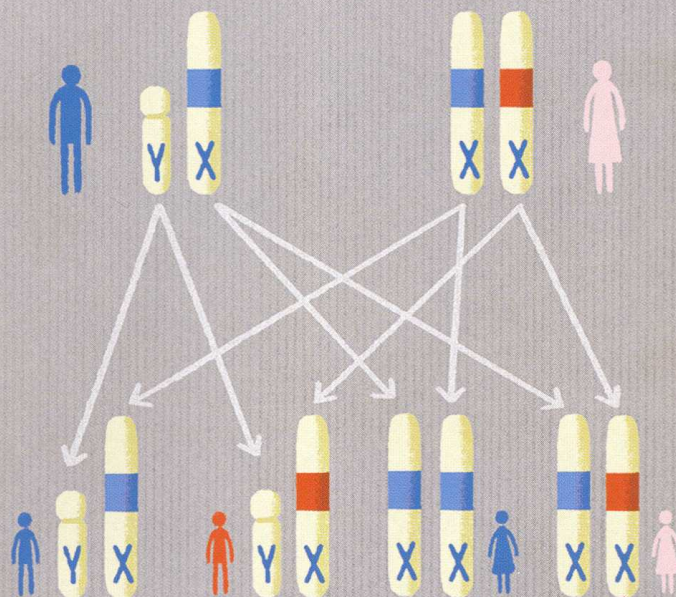




EUROPEAN
COMMISSION

Community research

25 Recommendations on the ethical, legal and social implications of genetic testing



SCIENCE AND SOCIETY



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STEERING COMMITTEE ON BIOETHICS (CDBI)

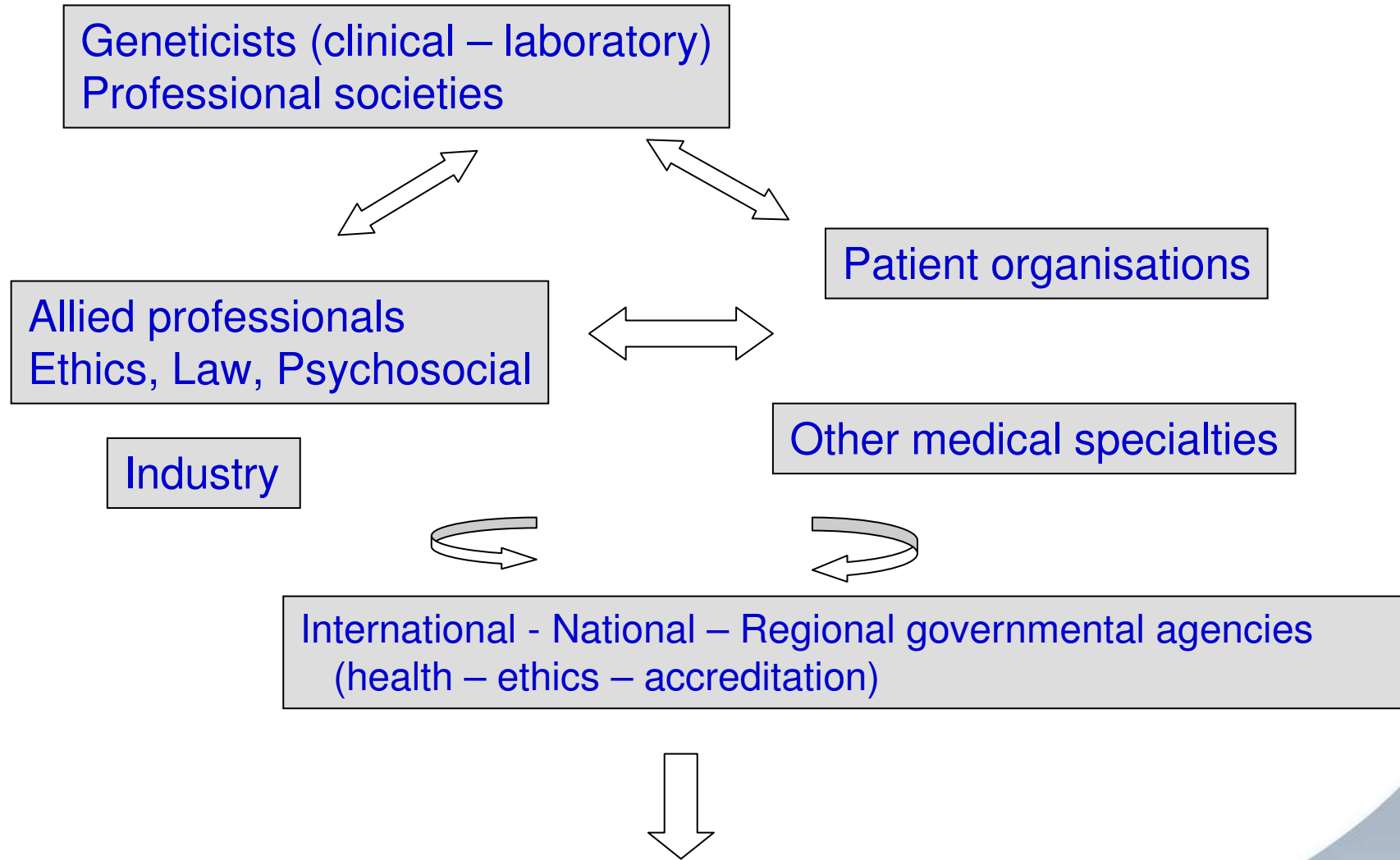
September 27 2007

Draft Additional Protocol to the Convention on Human Rights
and Biomedicine concerning genetic testing for health purposes

Draft Explanatory Report to the Additional Protocol to the
Convention on Human Rights and Biomedicine concerning
genetic testing for health purposes



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Quality local – Regional genetic services



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Genetic testing in Europe -
A network for test development,
harmonization, validation and
standardization of services

FP6 2005-2009

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