The changing landscape of genetic screening II
Governing the balance between ‘duty to protect’ and ‘right to test’

Amsterdam
27-28 May 2010

Ten years after – Mapping the societal genomics landscape

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Quality of Care

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Screening involves the clinical and laboratory examination of individuals who exhibit no health problems with the aim of detecting disease, predisposition to disease, or risk factors that can increase the risk of disease.

*(Health Council of the Netherlands, 2008)*

*Note: “systematic offer” not in this definition*
Genetic testing

- Clients ask about potentially increased risk of serious disorder
- Clinical geneticist diagnoses disorder
- Discuss diagnosis, recurrence risk, reproductive choices including possibilities for prenatal diagnosis/PGD
- (Broader definitions exist)
Who governs?

- Screening programs: public health authorities
- Testing: medical professionals
What was available?

- Hundreds of diagnostic tests for rare monogenic conditions
- Few predictive and screening tests (related to genomics)
- The best predictive tests: age, gender, BMI, waist circumference, family history
Company plans to sell genetic testing kit at drugstores

By Rob Stein
Washington Post Staff Writer
Tuesday, May 11, 2010

Beginning Friday, shoppers in search of toothpaste, deodorant and laxatives at more than 6,000 drugstores across the nation will be able to pick up something new: a test to scan their genes for a propensity for Alzheimer's disease, breast cancer, diabetes and other ailments.

The test also claims to offer a window into the chances of becoming obese, developing psoriasis and going blind. For those thinking of starting a family, it could alert them to their risk of having a baby with cystic fibrosis, Tay-Sachs and other genetic disorders. The test also promises information on the likelihood of cancer, heart disease and other conditions.

Jarrod Morgan tests DNA samples at Pathway Genomics in San Diego. The company plans to sell genetic testing kits at drugstores. (Sandy Huffaker/Bloomberg)
the rate at which useful new screening opportunities become available is not as rapid as reports in the media might sometimes indicate.

cultural, social and economic factors contribute to a situation in which various types of screening (including self-testing kits) are placed on the market without any proper investigation having been conducted to ascertain whether the benefits for those affected outweigh the disadvantages that always also exist.
Social developments relevant for screening

- Health care moving from a government-regulated health care sector to one which is driven to a greater or lesser extent by market forces.

- Blurring distinction between collective prevention and individual client-focused care.
  - Clinical genetic family testing vs cascade screening for FH

- Need for reassurance: people increasingly receptive to anything that promises to eliminate risk.
Genome information for all

People want to make an autonomous choice

People are increasingly held responsible for their own health

Risk of DTC

Lack of expertise

Lack of counseling

Incorrect results
  • analytic validity
  • clinical validity
  • clinical utility

Free fall. As with computer technology, the plunging cost of DNA sequencing has opened new applications in science and medicine.
Screening in new contexts.

National population screening programme (see chapter 5):

- population screening
- systematic family screening (cascade screening)

Public

Other forms of screening in the healthcare/public healthcare sector:

- preventive screening of at-risk groups in standard treatment practice
- screening in the context of scientific research

Private provision by doctors/general practitioners:

- health checks

Private

Uncontrolled screening, usually in a commercial context:

- general medical check-ups and body scans
- home-collection lab tests

DIY self-testing kits available for purchase
New technological possibilities

- Attunement between parties

The role of the government *(Health Council 2008)*

- **Duty of care: ensure worthwhile screening**
  - National population screening programme: provide facility itself
  - Available in basic healthcare package
  - Reproductive screening: special position: provide worthwhile options and guarantee both quality and informed decision making

- **Duty of protection against unsound screening**
  - Guard citizens against health damage from risky or unsound forms of screening
Balance pros and cons: role of government?

- A fresh approach is needed to encourage sensible screening and to protect individuals against the risks of unsound screening.
- Extending regulations??????? Not..most suitable
- Independent body... nat screening committee UK
- Establish a quality-mark for responsible screening, based on scientific assessments of new developments and aimed at promoting responsible provision and responsible choices.

www.gr.nl Screening between hope and hype. Presentation of report
Three categories of screening *(Health Council 2008)*

- Useful and cost effective
  - Potentially part of national program
  - Responsible screening should be available and accessible

- Utility less clear, no harm foreseen
  - Up to the customers?

- Potentially harmful or misleading
  - Protect!
Self testing: European IVD directive

- Risk classes: high, medium, low
- Low: assessed by the manufacturer

- NEW is often LOW

- NL: Marketing channel regulations: High risk only sold via professional intermediary
• Population screening act: arbitrary categories for licencing, inflexible. Why are some intensively evaluated while others are not?

  – Cardiovascular disease vs. cancer

• Enforcement: prosecution only for parties who carry out screening, not those who offer it and carry it out (*in Germany, USA or China...*)

• Do it yourself testing kits easily obtained from Internet or pharmacist

• Ban=restriction on freedom?
Protection – Self regulation?

- Quality control
- Accreditation/certification
- Standards
- Recognition of competence

www.epbs.net/brussels/
NIH Launching Genetic Test Registry

Bridget M. Kuehn

In the face of increasing calls for more transparency about and regulation of genetic testing in the United States, the National Institutes of Health (NIH) announced in March that it is creating a new voluntary genetic test registry that may soon help physicians and patients select and interpret genetic tests.

Test makers can voluntarily submit information about the availability, validity, and clinical relevance of their products to the registry, which is expected to be available in 2011. The registry will be publicly searchable, allowing physicians and patients access to an array of information about genetic tests.

Currently, genetic tests are available for more than 1,700 conditions. But critical information about the tests and how to use them in clinical practice is not always easy to find. Joan Scott, director of Johns Hopkins University’s Genetics and Public Policy Center in Washington, DC, said that clinicians and patients need information about which tests are available, what variants the tests detect, and what the results mean in the context of an individual’s medical or genetic history. For example, although some genetic tests (such as that for the variant that causes Huntington disease) may be used to determine whether an individual will develop a particular condition, others may indicate only that an individual has a higher risk of developing a condition. Additionally, these consumers need information about the evidence supporting a particular test and the reliability of the laboratory conducting the testing.

The Center and an array of other stakeholders have urged the NIH to create a genetic test registry that is mandatory rather than voluntary. But Scott called the creation of the voluntary registry a significant step. “We think it is important to do, to increase transparency about genetic tests—what is known and not known,” she said.

The NIH Office of the Director is overseeing the project, while the National Library of Medicine (NLM) National Center for Biotechnology Information will develop the registry and will cross-link it with other NLM databases on genetic and medical information. The NIH is seeking input from stakeholders on the database’s design and content and will coordinate with the US Food and Drug Administration and Centers for Medicare & Medicaid Services, which already collect some information about genetic tests and/or testing laboratories.

To obtain more information or to comment on the design of the registry, visit http://www.ncbi.nlm.nih.gov/gtr/.
Concluding remarks

- Fast increase of testing options for healthy people
- New actors (commercial parties, prevention services, autonomous citizens)
- Autonomy > right to test vs. tests that lack validity > duty to protect
- Tension between individual needs and interests and collective needs and interests
- Need for transparency
Main source of presentation: