The future role of professonal geneticists

The changing Landscape in testing
Gothenburg June 11, 2010

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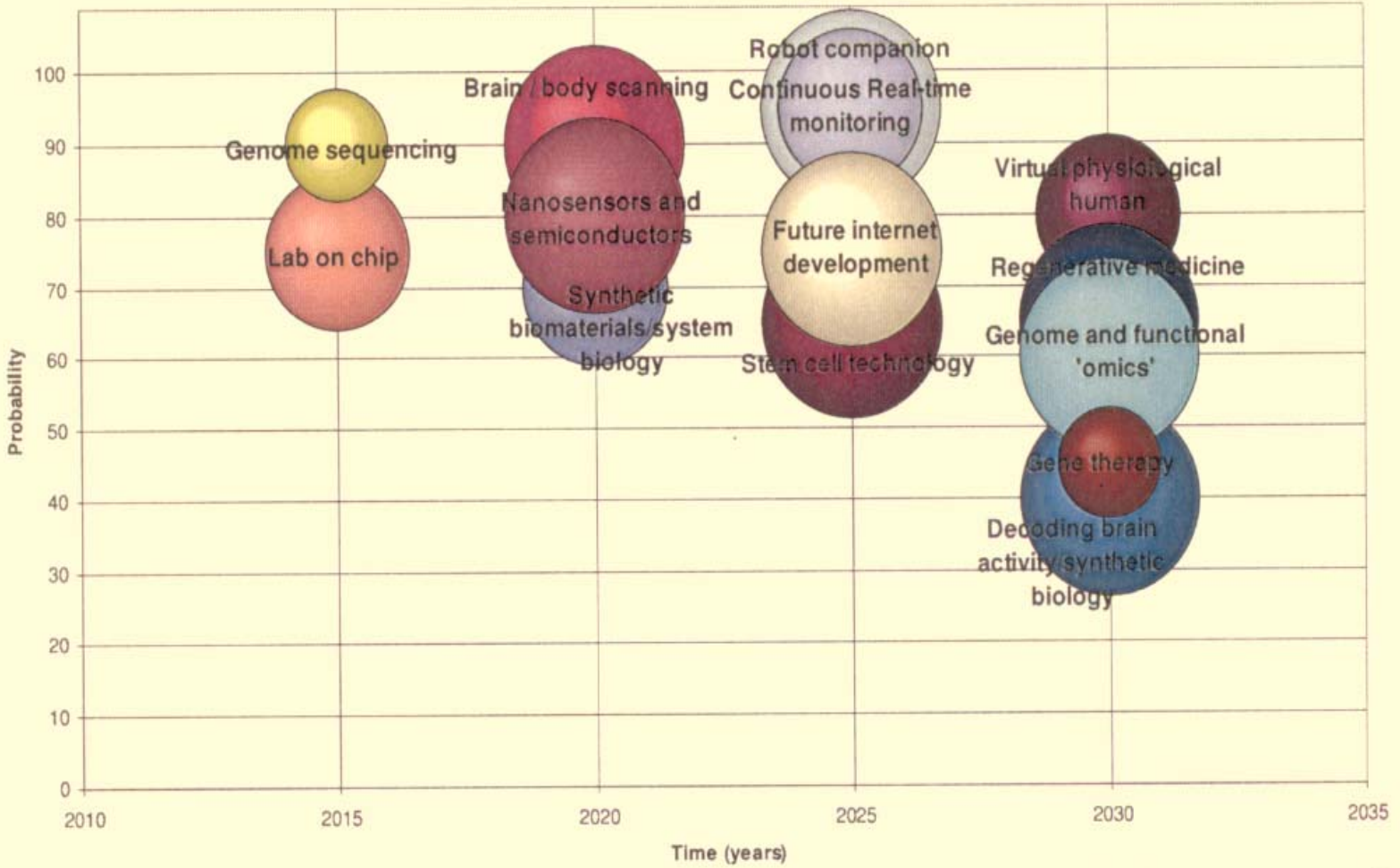


Figure 3 – This roadmap predicts which technologies will become available from now until 2030.

Cost Foresight 2030: Report on Future healthcare and biomedical research

Sources of Genetic testing results

Human:

- Non-medical (Forensic, ancestry)
- Medical applications:
  1. Research
  2. Public health:
     a) population screening
     b) clinical genetics:
        i. heritable:
           Diagnostic: (confirmation, exclusion)
           Counseling: presymptomatic
           familial cancer
           susceptibility
           PND
           PGD
        ii. other applications:
           PGD (aneuploidy screening)
           Drug treatment: pharmacogenetic testing
           Somatic: Cancer identification, prognosis

What is being tested?

- 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)

Who generates testing results?

Clinical services:

- *Clinical and molecular genetic services in academic or private setting*
- *DTC facilities*
- *In time any medical department in hospital setting*
- *Family physician*

Research facilities:

- *Academic, private, commercial (pharma) labs*

Non Medical facilities:

- *Forensics academic , police or private*
- *Ancestry: academic, private, DTC*

Will Medical Genetics Evolve to a Consumer Business?

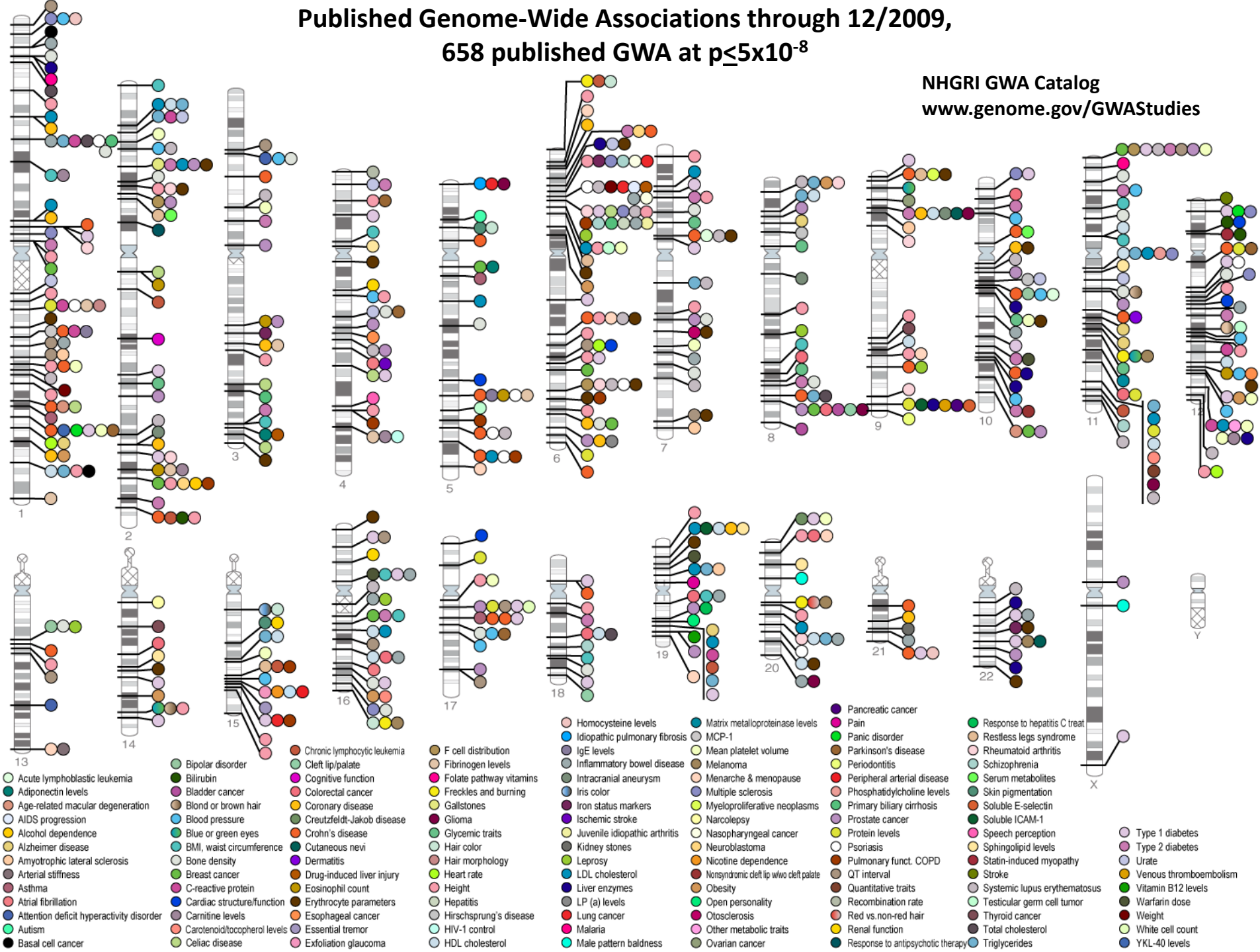
Certainly, medical genetics will not completely evolve to a consumer business; but, thanks to the confluence of two transformational technologies - the Internet, and the delineation and measurement of the human genome - and the creativity of some entrepreneurs, the availability of some genetic tests has migrated from the scientist- or health professional-controlled domains of research, hospital, and clinical laboratories to the wild, wild world of cyberspace, available to anyone, almost anywhere, with a click of the mouse.

Laurie Sullivan

published in CHI's Insight Pharma Report, Direct-to-Consumer Genetic Testing: Business Prospects in the United States.

Published Genome-Wide Associations through 12/2009, 658 published GWA at $p \leq 5 \times 10^{-8}$

NHGRI GWA Catalog
www.genome.gov/GWAStudies



Systems Biology

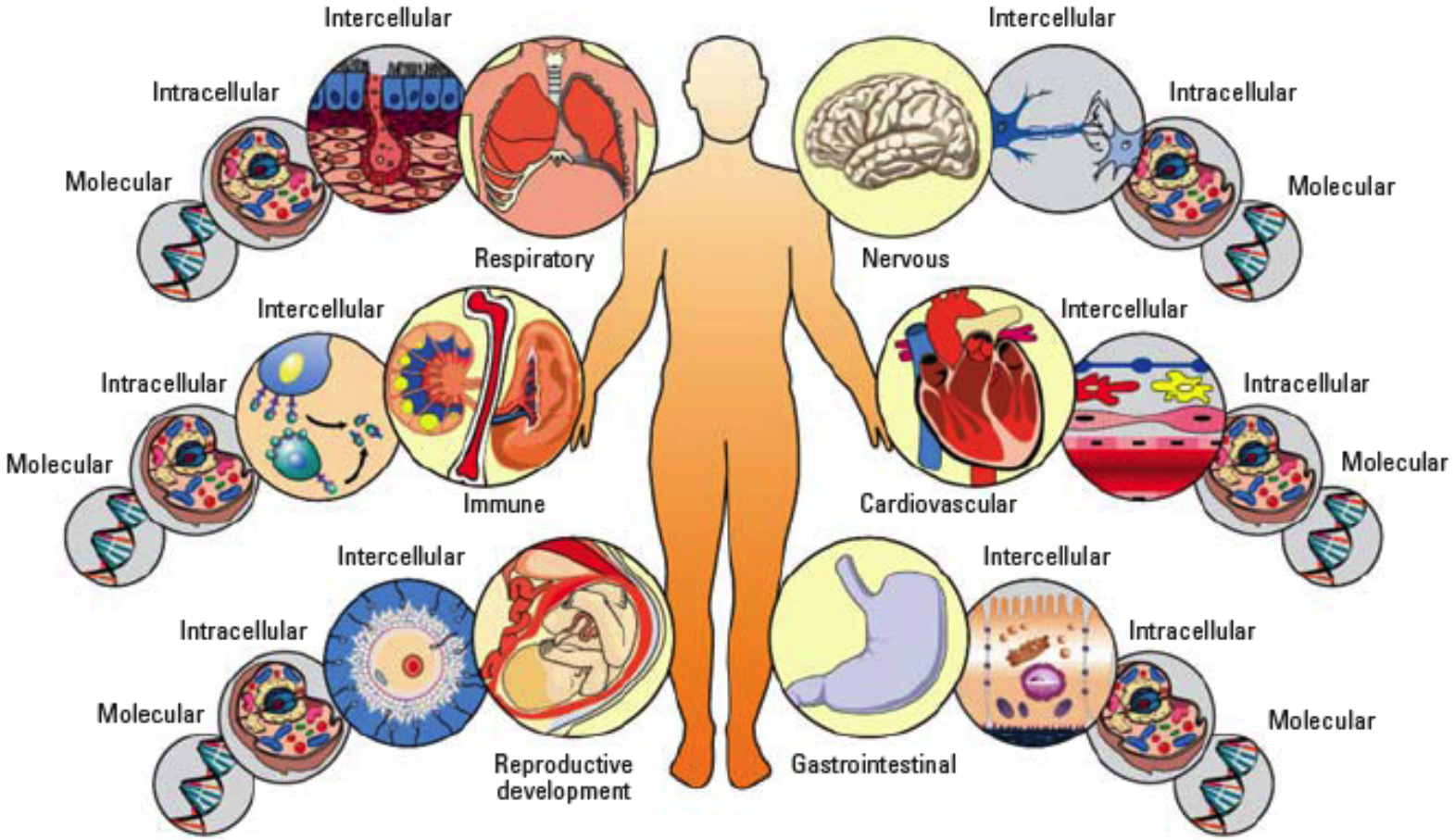
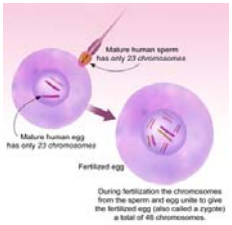
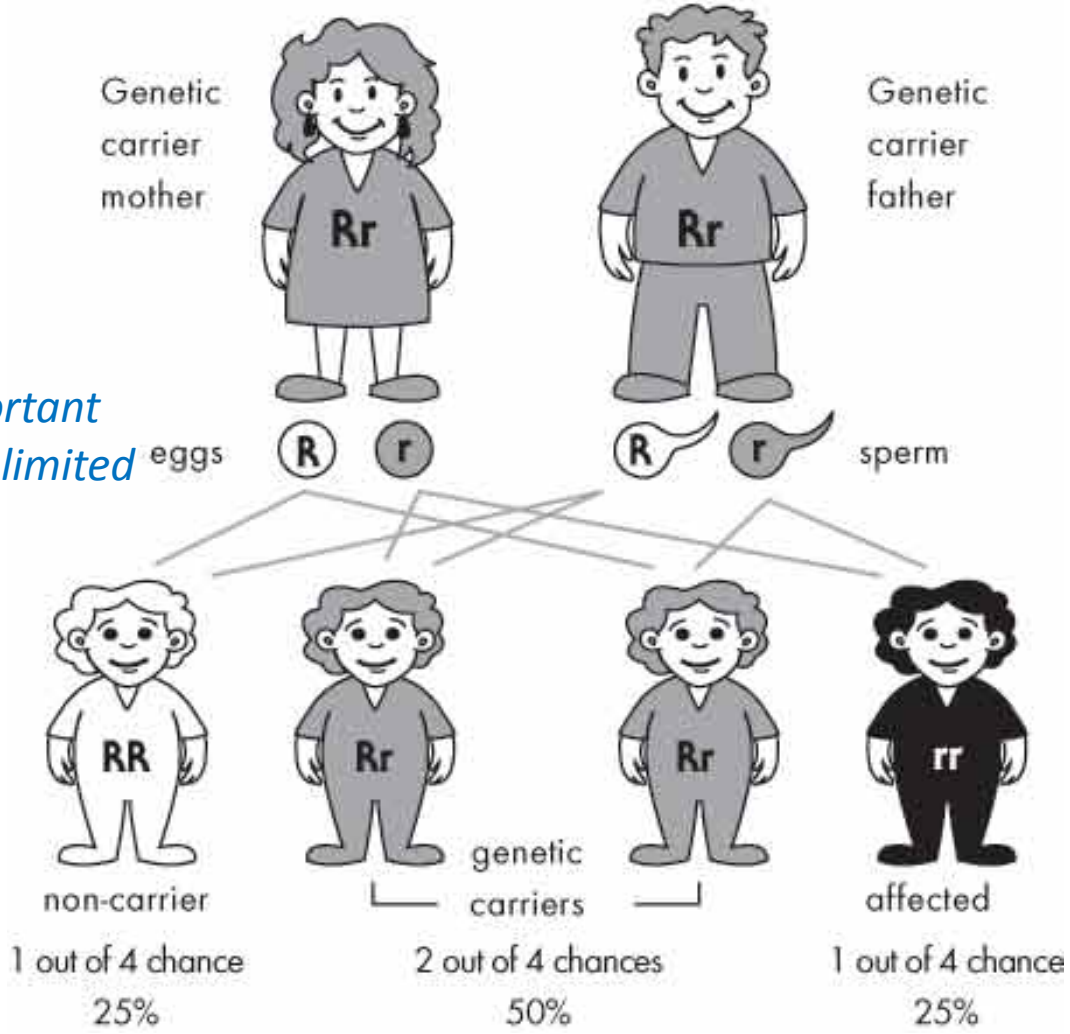


Figure 1. Systems biology framework for the individual. Current systems biology methodologies take advantage of high-throughput data generated at the molecular level in the hope of one day translating these maps of molecular interactions into cellular-level responses, then intercellular responses, and finally to an organ-level response. The interconnections between organ systems will need to be elucidated to understand an organism-level system.

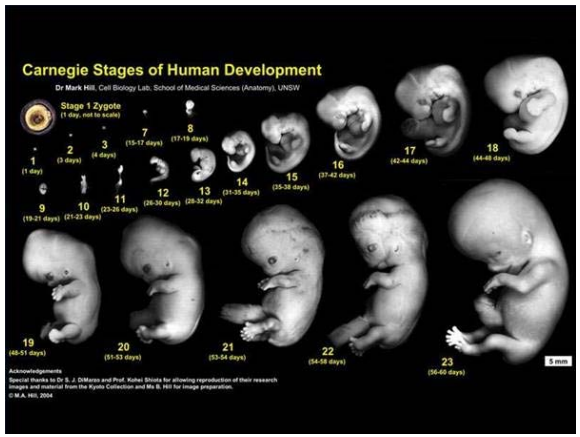
Transgenerational aspects

- *inheritance is important*
- *the focus is on germ cells*
- *reproductive fitness is important*
- *nature-nurture interactions limited*



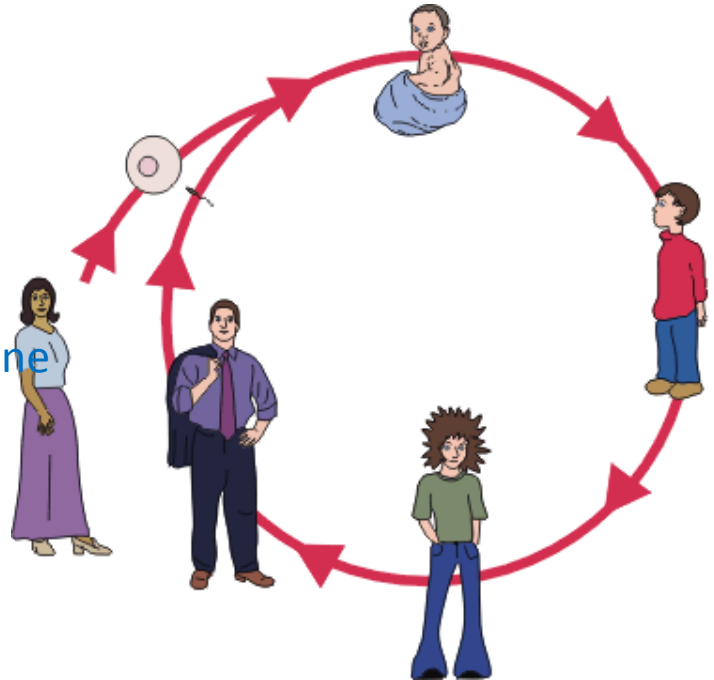


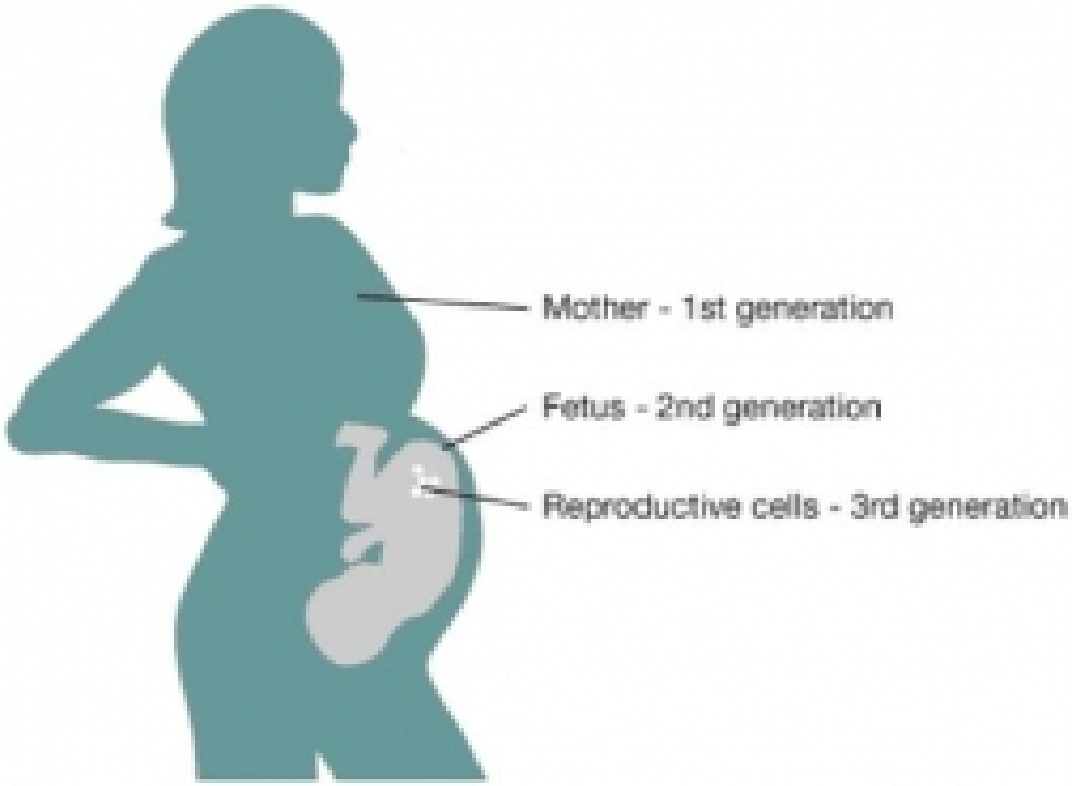
Developmental aspects



- the genome is gradually expressed
- the focus is on somatic cells
- nature - nurture interactions (epigenetics)
- possble appliocations for personalized medicine
- gene therapy is acceptable

→ role in susceptiblility/ predispostion




Mother - 1st generation
Fetus - 2nd generation
Reproductive cells - 3rd generation


Vita quidem cursu surgit mortalis amoeno
Cum vita vires ingeniumque abeunt
Masculа dum juvenis virtus contemperat æstum,
Prudenti gressu cana senecta venit.
So fangt das Leben an, in diſem Menſchenlauf,
Hört Witz, Verſtand u. Muth, auch endlich wieder auf,
Circulo humanæ vitæ et ætatis.
Circkel des menſchlichen Alters.
Der Jugend Feüer wird zu einer Mannes Glut,
Diß daß der Alters Eis zuletzt verächtlich thut.
Cum Priv. Sac. Cae. Maj.
Nº 210.
Martin Engelbrecht excudit A.V.

Expertise of the geneticists

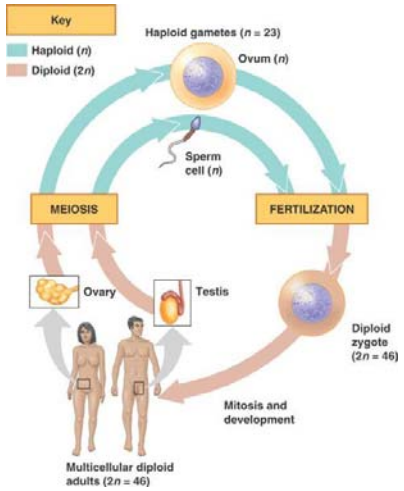
Transgenerational

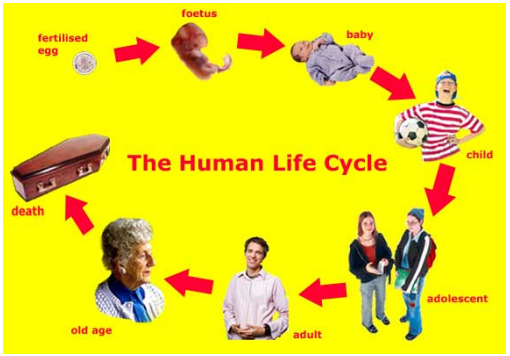
Mainly expertise of the ***geneticists***

Developmental

Joint expertise of specialists and ***geneticists***

"Multidisciplinarity"





The ACCE model

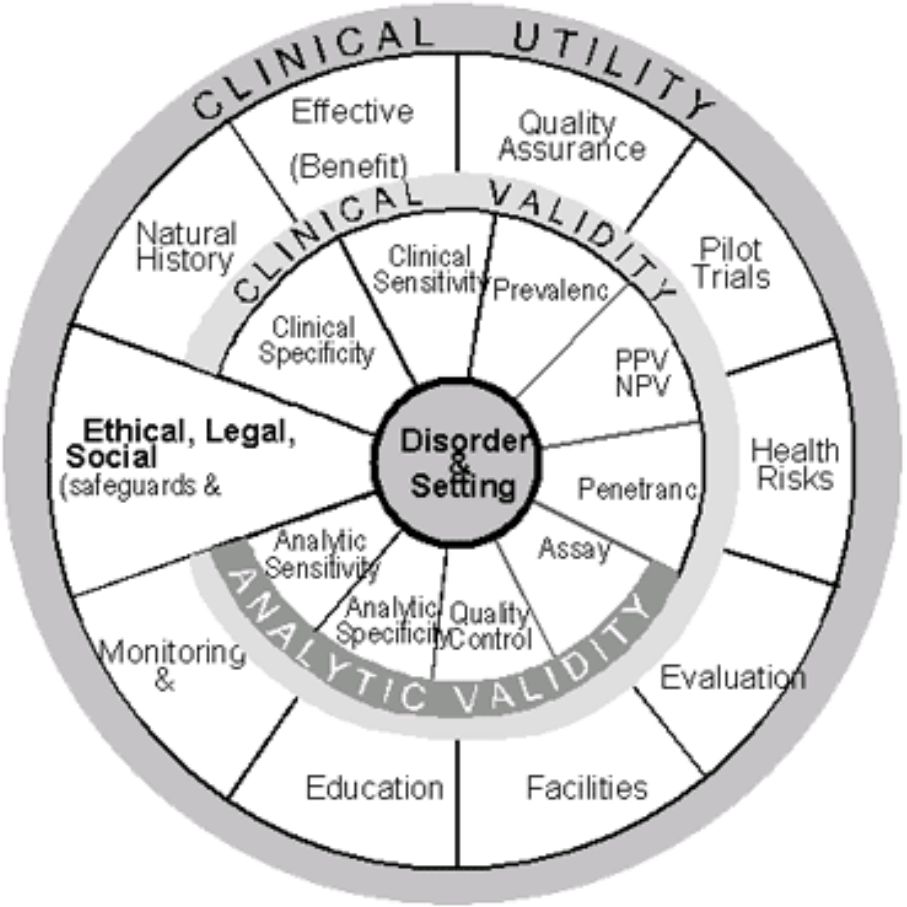


Figure 1. The ACCE evaluation process for genetic testing

CDC-US

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About EGAPP

Evaluation of Genomic Applications in Practice and Prevention (EGAPP): Implementation and Evaluation of a Model Approach

The EGAPP initiative was launched in 2004 by the CDC National Office of Public Health Genomics in the fall of 2004. The intiative's goal is to establish and evaluate a systematic, evidence-based process for assessing genetic tests and other applications of genomic technology in transition from research to clinical and public health practice. EGAPP also aims to integrate:

- existing recommendations on implementation of genetic tests from professional organizations and advisory committees.[1,2,3,4]
- knowledge and experience gained from existing processes for evaluation and appraisal (e.g., US Preventive Services Task Force, CDC's Task Force on Community Preventive Services), previous CDC initiatives (e.g., the ACCE process for assembling and analyzing data on genetic tests; http://www.cdc.gov/genomics/gtesting/ACCE/FBR/index.htm) 5, and the international health technology assessment experience.

Why is genetic testing a public health issue?

The success of the Human Genome Project has led to increasingly rapid translation of genomic information into clinical applications. Genetic tests for more than 1,200 diseases have been developed, with more than 1,000 currently available

Practical Clinical Utility

Component	Definition	Relevance to clinical utility
Analytic validity	Accuracy of test assay compared to gold standard measure	Determines whether test meets acceptable analytic standard
Clinical validity	Association of genetic characteristic with specified health condition or risk, and sensitivity, specificity and predictive value in the population groups and clinical setting proposed for testing	Determines potential clinical uses of test
Test setting and purpose	Clinical and/or demographic description of group(s) to be offered testing, health condition tested for, and specific clinical goal of testing, including any associated services	Determines outcomes sought from testing
Societal legitimacy	Consistency of testing and associated services with ethical principles, values, norms, mores, laws and regulations	Determines whether test is compatible with societal expectations
Efficacy and effectiveness	The ability of the test and any associated services to achieve the intended health purpose under the most favorable circumstances (efficacy) and under routine conditions (effectiveness)	Determines the potential for the test to achieve the health outcomes sought
Balance of outcomes	Assessment of negative relative to positive outcomes of testing and associated services for the person tested	Determines whether testing provides a net benefit to the person tested
Patient and family acceptability	Consistency of testing and associated services with the wishes, desires, and expectations of patients and their families	Determines whether testing is compatible with patient and family preferences
Economic measures	The ability of the test and associated services to lower the costs of care without diminishing benefits and/or provide an appropriate health value for the investment of resources	Determines benefits provided by testing relative to investment of resources
Equity	Access to test and associated services among patients who can benefit	Determines whether testing is compatible with equitable health care delivery

Adapted from Burke et al., 2007 [12]; Burke and Zimmern, 2007 [13]; and Zimmern and Kroese, 2007 [14].

Debating Clinical Utility; W. Burke, A.-M. Laberge, N. Press
PHG 13, 215-223, 2010

CORE COMPETENCES IN GENETICS FOR HEALTH PROFESSIONALS IN EUROPE

The final version of the documents on "Core Competences in Genetics for Health Professionals in Europe" is now ready. After one year of consultation on the ESHG website, EuroGentest website and the EuroGentest workshop in Milan (27-27 October, 2007) the documents are now in a form that we feel can be shared with other professionals, patients and the public. As a result of feedback, the original single document has been completely revised and divided into four separate documents:

1. The Executive Summary
2. Background document (updated)
3. Suggested core competences for health professionals who are generalists or specialising in a field other than genetics
4. Suggested core competences for genetic specialists

The overall aim of this work is to enable national societies for health professionals to use the core competences as a basis to guide curricula and new initiatives in professional education. We are aware that for patients and the public these documents may be too detailed, but we can announce that another document addressing this topic is being produced as part of a parallel project titled "EuroGenGuide". Patient associations are playing the major role in producing this document.

You can download the documents from this page and we welcome any further final feedback.
You can email any comments to Domenico Coviello and Heather Skirton

Thank you for your support.

Domenico Coviello and Heather Skirton
ESHG Education Committee

EuroGentest

Recommendations for genetic counselling related to genetic testing

1. Introduction:

The main goal of the EuroGentest Network of Excellence (www.eurogentest.org) is to improve the quality of genetic testing. As patients' understanding of the results and consequences of the test is an integral part of genetic testing, EuroGentest also aims at improving the quality of genetic counselling services associated with genetic testing, across Europe. One of the goals is to establish recommendations for genetic counselling in connection with different testing situations.

2. Method:

To achieve this, the group assigned by EuroGentest for this task has collected and analysed international and European non-national guidelines and policies related to genetic counselling, as well as some relevant national recommendations and other documents. In addition, legislation related to genetic counselling in EU countries has been collected. Three workshops attended by experts on genetic counselling have been organized (May 2005, September 2006, December 2007). In addition, data has been collected with the help of surveys. The draft of the recommendations was distributed to European clinical geneticists and genetic counsellors and European National Human Genetic Societies for comments. The final version was written taking these comments into consideration.

What can the geneticists do?

- *More research and correct validation*
- *Make sure the guidelines developed by ESHG, EuroGentest, OECD etc… are being implemented to improve our services to the highest quality*
- *Participate in multidisciplinary approach in the services – bring genetic expertise in the approach*
- *Strive for efficient and affordable treatments*
- *Inform the public*

- *Involve patient organisations*