

# **PERSONALIZED MEDICINE: An elusive concept, diversified practices**

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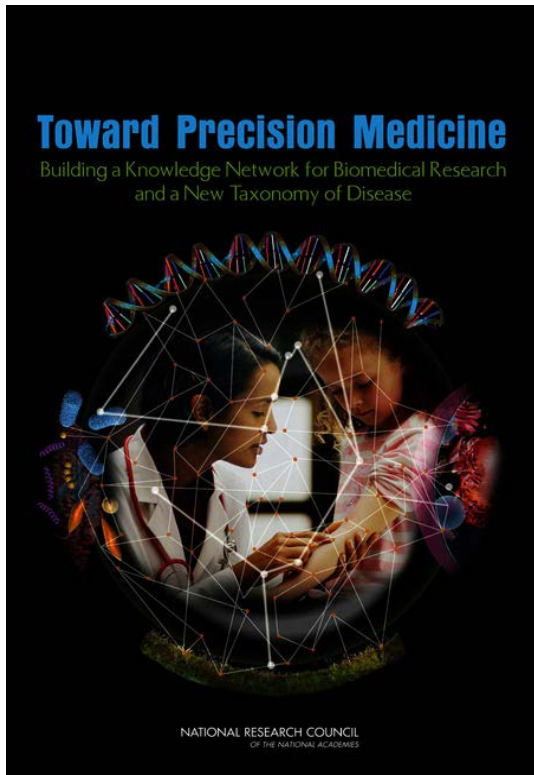
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# An Elusive Concept

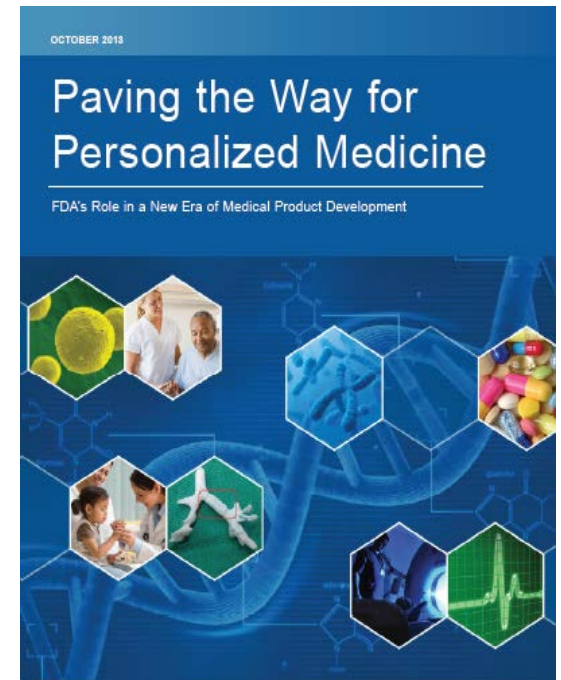
- What is personalized medicine?
  - Difficult to pinpoint a consensual definition
- So let us ask the question differently:  
*What is personalized medicine all about ?*



2011



2012



## *Reports on Personalized Medicine*

2013

## *No consensual definition of personalized medicine*

Common emphasis on tailoring/customisation to individual characteristics

Personalised medicine can be broadly described as a customisation of healthcare that accommodates individual differences as far as possible at all stages in the process, from prevention, through diagnosis and treatment, to post-treatment follow-up. In practice, however, what people understand the term to mean varies widely and no single definition has been agreed upon. Many other terms, such as *genomic medicine*, *stratified medicine* and *precision medicine* are frequently used synonymously with *personalised medicine*.<sup>1</sup>

*ESF Report, 2012*

Each of these examples demonstrates the promise of “personalized medicine,” which is the tailoring of medical treatment to the individual characteristics, needs and preferences of each patient.

*FDA Report , 2013*

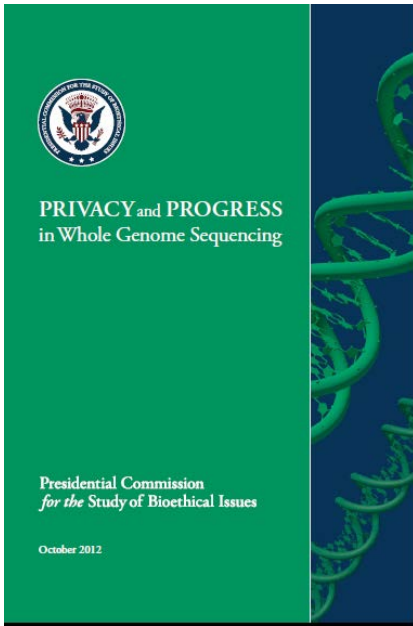
## *Personalized? Stratified? Precision ?*

*Absence of agreement on*

*the characterisation of this medicine*

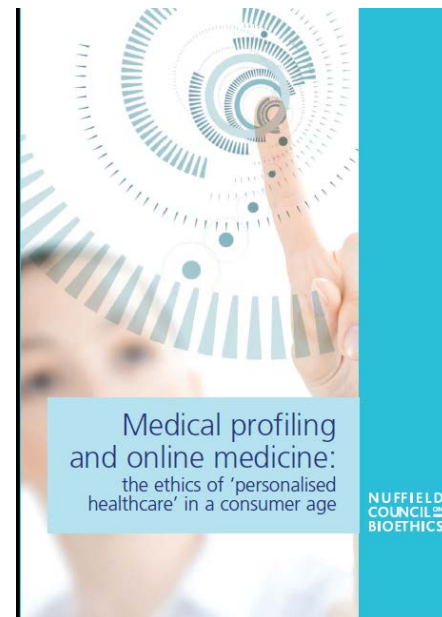
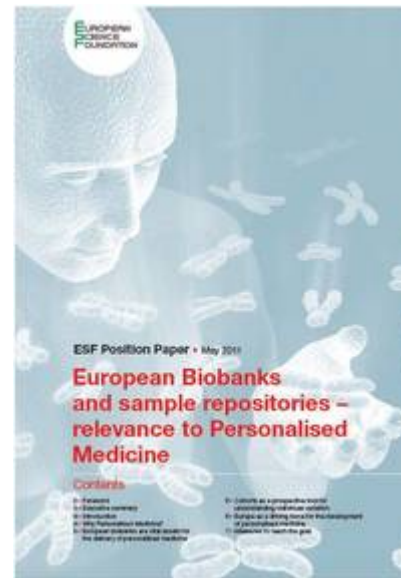
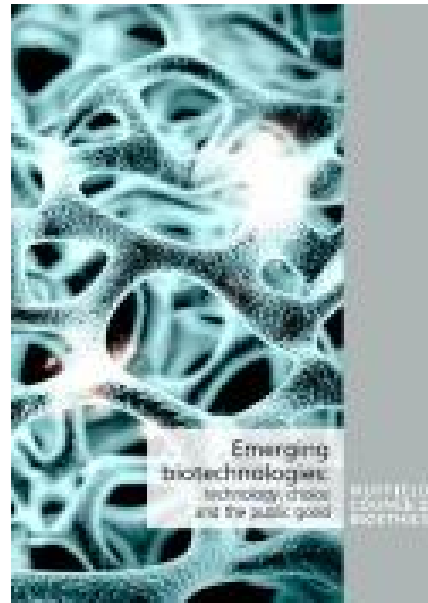
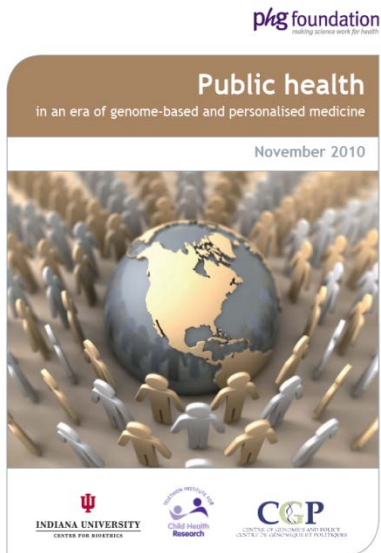
The vision for a New Taxonomy informed by the proposed “Knowledge Network” shares some similarities with the widely discussed concept of “Personalized Medicine,” recently defined by the President’s Council on Advisors on Science and Technology (PCAST) as “the tailoring of medical treatment to the individual characteristics of each patient . . . to classify individuals into subpopulations that differ in their susceptibility to a particular disease or their response to a specific treatment. Preventative or therapeutic interventions can then be concentrated on those who will benefit, sparing expense and side effects for those who will not” (PCAST 2008, p. 1).

*National Academy of Sciences Report, 2011*



# Approach through enabling technology

- Genomics
- Emerging biotechnologies
- Internet
- Biobanking



# Personalized **Genomic** Medicine

New sequencing methods

- Increased speed
- Production of greater quantities of data
- Better quality?

- ***Does this necessarily mean more useful information?***

Expectations heightened through combined use with other technologies

- Internet - Direct to consumer testing
- Biobanking - Preservation of biological samples
- Cloud computing – Data collection
- Etc.

# Importance of Genomics

High expectations invested in new technology:

- Aid in medical decision-making
- Improved outcomes in prevention and treatment
- Possible paradigm shifts in certain specialities

Classic example: Targeted therapies in cancer treatment

***Will this be a useful paradigm for other specialities?***

New options in genetic screening and diagnostic testing

- Adults, infants, prenatal (NIPT)...preimplantation?



SYMPOSIUM: FUTURES IN REPRODUCTION  
REVIEW

# Personalized reproductive medicine on the brink: progress, opportunities and challenges ahead

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*CELMATIX - biotechnology company (2009)*

« Harnessing the power of data and genomics to optimize medical  
decisions about fertility

- Moving toward a truly individualized predictive model »

# *Personalized medicine combined with alternative forms of postnatal screening*

National Human Genome Research Institute

National Institutes of Health  
U.S. Department of Health and Human Services



National Human Genome Research Institute  
[www.genome.gov](http://www.genome.gov)

*Eunice Kennedy Shriver* National Institute of Child Health and Human Development  
[www.nichd.nih.gov](http://www.nichd.nih.gov)

## **NIH program explores the use of genomic sequencing in newborn healthcare**



Bethesda, Md., Wed., Sept. 4, 2013 - Can sequencing of newborns' genomes provide useful medical information beyond what current newborn screening already provides? Pilot projects to examine this important question are being funded by the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) and the National Human Genome Research Institute (NHGRI), both parts of the National Institutes of Health. Awards of \$5 million to four grantees have been made in fiscal year 2013 under the Genomic Sequencing and Newborn Screening Disorders research program. The program will be funded at \$25 million over five years, as funds are made available.

# Paradox of personalized medicine

« Individualisation » requires collection of genomic data from *large populations*

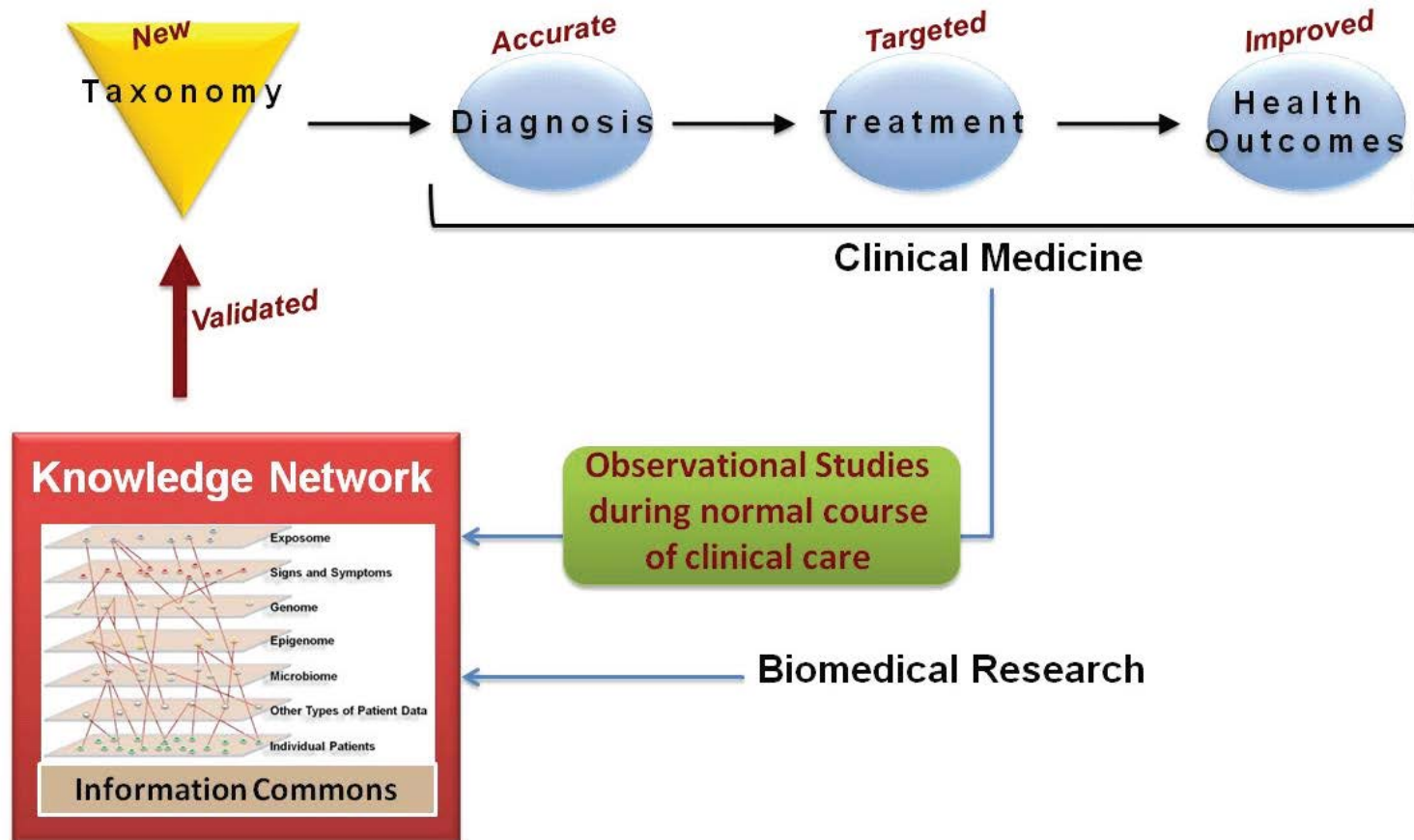
- GWAS
- PheWAS
  - Use of electronic medical records to search for clinical conditions associated with a variant
  - Use of blood samples for genetic studies
- Pertinence of such data as clinical tool remains controversial
- Serious concerns about privacy of data

Creation of Global Alliance

- Concern about responsible sharing of genomic and clinical data (June 2013)

# Toward Precision Medicine

Building a Knowledge Network for Biomedical Research  
and a New Taxonomy of Disease



# Challenge to therapeutic relationship

## High expectations regarding client/patient duties:

- Increased responsibility in prevention and treatment
  - Increased pressure to provide samples and share data?
- Indirect participation in research (even in direct-to-consumer testing )
  - Not always informed and voluntary
  - Blurring of research-therapy distinction?
- Genomic data for treatment may generate genetic counseling issues
  - Incidental findings : does right not to know still hold?

# In conclusion...

Personalized genomic medicine is about...

- the potential **uses** and **usefulness** of new tools in diagnosis, treatment, and prevention
- **redefining reciprocal expectations** in the therapeutic relationship
- **novel or renewed forms of collaborations** among diverse professionals
- at present, **more about research** than about medicine