What will a step-by-step approach to public health genomics in Portugal require?

João Lavinha

Departamento de Genética Humana

Instituto Nacional de Saúde Ricardo Jorge, Lisboa, Portugal

joao.lavinha@insa.min-saude.pt

www.insa.pt

Public health genomics:

The responsible and effective translation of genome-based knowledge and technology for the benefit of population* health.

*** individual/family/community/society**

Genome-based Research and Population Health, Bellagio 2005.

Genome-Based Information* and Technology (GBIT)

- **Opportunity:** GBIT will enable detailed risk profiling as the basis for targeted interventions, potentially improving health outcomes.
- **Threat:** premature technology and market driven applications will inundate physicians and patients with meaningless or uninterpretable data.
- **Gap** between our ability to generate 'more data for less money' and our ability to understand them or validate their clinical utility.
- **Drive:** Clinical utility, not technological ability!

* Knowledge >> Information?

Policy issues and options (*steps*)

1. Issues related to developments in medical genomics **research**
   a) Data sharing and intellectual property
   b) 'Big data' security and privacy
   c) Quality Assessment
2. From research to **clinical practice**: current issues and future challenges
   a) What to screen for and when?
   b) Patients' rights and professional responsibilities
   c) Informed consent and service provision
3. **Governance** in public health genomics

Medical research: Data sharing (IP) and privacy

- Closely monitor the public governance of g
  biobanks.
- Pay attention to the specific rules which g
  care and research as two different dom
  - Consider the need for harmonization of legislation governing these two domains.
- Support the development of an appropriate ethical and governance framework for data sharing across the EU and emerging players in e.g. China, India and Latin America.
- Rethink the current IP regime in order t
  obstacle to innovation and making (pub
  expensive.
  - Making the IP regime more flexible, while acknowledging
    flexible regime might undermine European competitivenes
- Consider special forms of data protection for genomic data s part of the current revision of the EU Data Protection Regulation.

Foster collaboration between health professionals and researchers.

Allow indefinite storage with consent. Paramount in familial conditions!

Medical research: Quality assessment

- Support the development and implementation of guidelines for quality assessment that are relevant for genome-wide sequencing tests, and which include assessment of the value of unsolicited findings.
- Invest in well-designed studies in populations that are representative of an intended healthcare application of genome-wide
  - Multiple smaller studies tailored to the intended ap
    value than one analysis using data from a large gen
- Consider the need for stricter regulation to protect the c umer, control societal healthcare costs and allow commercial DNA testing, all at the same time.
  - The EC has recently published a new draft for a revised in vitro diagnostic medical device (IVD) which proposes a ban on DTC marketing of genetic tests without the involvement of genetic counsellors.
- Make participation in quality assessment schemes (including assessment of clinical utility) mandatory for genetic testing laboratories.

There is no real need for DTC health-related DNA testing.

Clinical practice: What to screen for and when?
I. The scope of screening options

- Define a clear and guiding role for public health authorities in e introduction of whole genome
  f reproductive and
  orn screening can tal
  organized public grams, but may also be i
  on a commercial basis.
- Limit the range of screening options to a well-defined stand
  offer when introducing whole genome sequencing in carrier
  prenatal screening.
  - Broader tests might be available for parents only on an opt-in basis.
- When introducing genome-wide newborn screening, keep the data that are provided to parents limited to information that is actionable and of immediate clinical utility for the child.
- Preserv e right of a child to an 'open' future in defini he scop nome-wide screening.

Screenings tend to be universal and free of charge (access equity).

Risk of access inequity, e.g., to downstream interventions.

Train professionals to clearly define which data are clinically actionable or of potential relevance for a patient and/or sample donor.

But store all the collected data to allow future re-evaluation.

Clinical practice: What to screen for and when? II. Informed consent

- Stimulate empirical research among potential sample donors to find out whether there is large support for either specific, broad of presumed informed consent.
- Examine what kind of counselling framework needs to be created for whole genome screening, given the volume of data and the type of information generated.
- Distinguish different settings, such as research, clinical care and screening, in designing procedures for informed consent.
- Limit unsolicited findings by the use of filters that restrict the amount of data generated to particular clinical purposes.
- Establish clinical genetics as a profession in every country, stimulate collaboration between geneticists and other medical specialists.
  - Integrate genetic services into primary care in order to secure a proper framework for informed consent.

Governance

- Organize pilot experiments in different contexts and countries as support of a step-by-step approach to the introduction of GBIT in healthcare systems.
- Engage stakeholders, including patient advocacy and civil groups concerned with genome sequencing issues, in experimentation, assessment and decision-making.
- Use the best practice guidelines and legislation that is already available for genetic testing services to inform the development of HTA practices all over Europe.

HTA in Portugal has focussed mainly on economic (reimbursement) issues rather than on the medical, social, organizational and ethical issues.