Advanced Genomics in Health Care?

Using TA to design a step-by-step approach

in EU member states

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German Gene Diagnostics Act

(Effective since 1 February 2010)

- Valid for genetic diagnostics, handling of samples and information for medical purposes
- Not valid for purposes of research
- Initiation of genetic diagnostics only by MDs
- Accreditation of genetic laboratories and participation in external quality assessment procedures
- Informed consent
- Limited examination of minors and mentally handycapped persons
- Predictive genetic diagnostics
- Prenatal genetic diagnostics
- Genetic screening of healthy persons: only treatable disorders

German Gene Diagnostics Act

(Effective since 1 February 2010)

Genetic counselling + testing:

- MDs: specialists for human genetics
- Clinicians with a special genetic training (field-limited)
- Genetic analysis only for a defined aim
- Predictive genetic diagnostics: offer of genetic counselling obligatory
- Result of genetic analysis: only tested person + personal physician
- Result of genetic analysis: to be stored only for 10 years
- After analysis: biological sample has to be discarded

The Difference:
Genetic Diagnostics versus Genetic Research

Targeted analyses of specific genes for medical/health purposes

- in heterogenous disorders: genetic panel diagnostics
- multifactorial disorders not yet suited for diagnostic analyses

Whole genome/exome sequencing

- presently only for research purposes
- informed consent necessary
- agreement how to deal with unpredictable findings
- 1st example: cancer genome project
- (cf. ethics of total genome analysis: Eurat project)

(http://www.uni-heidelberg.de/md/totalsequenzierung/informationen/mk_eurat_position_paper.pdf)

- 2nd example: Individualised Medicine

The Future

Whole genome sequencing of adults for health purposes

- rigorous informed consent necessary
- agreement how to deal with unpredictable findings
- agreement how data should be stored: central or decentral

but

Targeted genetic panel diagnostics may continue to play a role in the future e. g. for examining predispositions for multifactorial disorders

A Major Problem

Private laboratories use published genetic data for interpretation of their results,
but they do not make the results of their analyses accessible to the scientific community, although financed by the health system.

How can private laboratories be forced to enter mutations into public data banks?