

Research Programme >

Genetic Testing and Common Disorders

Background document, Recommendations

ESHG website: open for comments!

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 CENTRE FOR
Medical Systems Biology

Society & GENOMICS 
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Genetic health care in the previous century

- Monogenic disorders
 - Severe
 - High recurrence risk
 - Difficult reproductive decision making
- Chromosomal disorders
- Cases of MR/MCA asking for a diagnosis

Genetic health care in the future

- Monogenic disorders
- Chromosomal disorders
- Cases of MR/MCA asking for a diagnosis
- Cancer!

- Common disorders, complex etiology, monogenic subsets, multifactorial etiology, susceptibility genes?

Genetic health care in the future

- Complex etiology, monogenic subsets, multifactorial etiology, susceptibility genes?
 - New health care structures?
 - *Cardiogenetics*
 - *Cascade screening, pro-actively informing family members?*
 - *Consumer genetics; Direct To Consumer*
 - New tasks for geneticists?

Genetic health care in the future

- The promise of personalized medicine, tailored prevention advice
 - Hypes or hopes
 - Analytic validity, clinical validity, clinical utility
 - Quality of testing and information
 - *Genetic counseling?*

Regulation

- Direct-to-consumer
- In vitro diagnostics
- CE marking

- Information, communication, education, public awareness

The challenges for geneticists

- Engage in a responsible way in valorisation, translation, education, communication, multidisciplinary collaboration
- Quality of care
- Strengths in severe disorders, high risks, difficult decision making
- Coach other health care workers in FVL, HLA B27, HLA DQ 2-8, TCF7L2, α -1-AT deficiency
- Involve in policy making on lower-risk-genes