Genetic Testing and Common Disorders
Background document, Recommendations

ESHG website: open for comments!

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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?
• 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, $\alpha$-1-AT deficiency
- Involve in policy making on lower-risk-genes