

Research Programme >

# Susceptibility screening

Background document and  
recommendations

Mw.prof.dr. M.C. Cornel

Care and Prevention

Department of Clinical Genetics

Section Community Genetics



# 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?*
  - 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

- 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