11. Population Health Genomics

Genomics provides a link between clinical practice, academic research and policy development. Screening and awareness of risk allows preventive measures and surveillance measures to be instituted before damage occurs. The identification of individuals who are susceptible to developing inherited or complex diseases is an important public health challenge through research on:

- Newborn bloodspot screening for early identification of children with conditions that can be treated and avoid disabilities or premature death. The expansion of screening to include untreatable or later-onset conditions raises a number of ethical and practical policy issues.
- Antenatal screening for Down syndrome and fetal anomalies based on maternal blood. Technological advances in genetic sequencing enable the detection of a large range of fetal and pregnancy-associated abnormalities that necessitate economic, ethical and policy analysis.
- Targeted familial cancer and mutation-carrier testing to identify individuals at high risk (i.e. carrying the genetic defect) of developing cancer or a chronic disease provides pathways to prevent disease or for earlier detection and treatment.

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