Do you Really Want to Know?
Pros and Pitfalls of Genetic Testing

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*Presenter Disclosure:*

*(Nolan D’Souza)* has no potential for conflict of interest with this presentation
Definitions

**Confirmatory/Diagnostic Genetic Testing**
- Symptomatic individual – do they have specific genetic condition.
- Find genetic variant explaining individuals disease

**Pre-symptomatic Genetic Testing**
- Testing at-risk and **asymptomatic** individual
Objectives

- To discuss genetic test implications for patient privacy and confidentiality

- To review genetic discrimination/insurance implications of pre-symptomatic testing

- To review policy/exceptions in testing minors for adult onset conditions
Emotional Impact

Motives for proceeding with test:
- certainty of carrier/gene status
- decisions concerning marriage and children
- planning a career/education

Motives for declining testing:
- mainly psychological problems in coping with the test result + impact on relationships.

Canadians at risk for Huntington’s
- ~20% percent have pre-symptomatic testing
Privacy and Confidentiality

- Ethical guiding principles
  - Beneficence, Non-Maleficence, Autonomy, Justice, etc.

15 years later

Papillary RCC, 40

HPRCC

E+

Papillary RCC, 37

?mutation status
Privacy and Confidentiality

Declined testing

Ov Ca, 58
E^+ BRCA2
Privacy and Confidentiality

Dilated cardiomyopathy risk

??Insurance ??
Insurance Discrimination

- Canadian focus is primarily on life insurance
- Less discussed: disability, critical illness, long-term care
- GINA: U.S. Non-discrimination Act
- Insurance companies/employers cannot force genetic testing

Illustration by Daniel Pudles Photograph: Guardian
Genetic Testing in Minors

“We want to test her now and tell her if she is a carrier when she is older”
Genetic Testing in Minors

- Testing unaffected at-risk children younger than age 18 years for adult conditions is discouraged unless it impacts their management.

- Deferring predictive genetic testing allows individuals to choose for themselves as adults, taking into account their own circumstances, preferences, and beliefs.
Questions?