Genetic Testing for At-Risk Individuals

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Pre-symptomatic Gene Testing for HD

- Genetic Counseling
- Neurological exam
- Psychological interview
- Discussion of results
- Follow-up

Pros of Testing

- Reasons people elect pre-symptomatic testing for HD
  - Reproductive planning (for themselves or their kids)
  - Reduce uncertainty (need or want to know)

Cons of Testing

- Reasons people decline pre-symptomatic testing for HD
  - Cost
  - Desire uncertainty (not knowing is *not* a problem)
  - Absence of an effective treatment or cure
  - Potential discrimination (loss of insurance)
  - Fear of learning their children are at-risk
  - Not prepared to know gene status and the impact of this information

Goals of Genetic Counseling

- Facilitate decision making by providing a non-directive counseling approach
- Determine the client's needs and concerns
- Provide up-to-date information
- Explain the risks, benefits, and limitations of testing

Goals of Genetic Counseling

- Help client understand the implications of testing
- Provide pretest informed consent
- Provide result notification and follow-up
- Address psychosocial issues

Information from Family

- Review patient/family questions
  - reason for referral
  - knowledge base
  - perception of disease status and/or risk
  - what diagnoses have been considered
  - perceived notion about causation

- Obtain family history
  - neurological diseases, uncontrolled movements, mental illness, age of onset of symptoms, ages and causes of death
  - Medical records

Counseling Session

- Assess family functioning and use of community resources
- Assess possible ethical concerns
  - confidentiality, insurability, discrimination, prenatal diagnosis
    - genetic counseling for any additional family members being tested concurrently; evaluation of individuals at 25% risk; prenatal testing

Counseling Session

- Summarize information obtained and discuss with patient/family the (possible) diagnosis and the degree of certainty of the diagnosis based on available information
  - relationships with significant partner, children, extended family, friends
  - career decisions, and telling colleagues at work
  - impact on financial planning, including insurance (life, medical, disability)
  - "survival guilt" (i.e. perceive increased burden of taking care of affected family members)
  - optimal timing of testing

Informing Patient

- Discuss natural history:
  - prognosis
  - developmental outcome/intellectual functioning
  - anticipated possible medical complications
  - including pregnancy related risks for affected women if indicated, and preventive measures
    - a positive HD DNA test result does not give information about age of onset of symptoms, severity of symptoms nor prognosis.

Informing Patient

- Review inheritance pattern (including penetrance and expressivity)
  - autosomal dominant
  - trinucleotide repeat
  - age related penetrance

Informing Patient

- Discuss reproductive options (e.g. assisted reproductive technologies, adoption, taking risk and no additional pregnancies) when appropriate

- Review management recommendations/options including screening protocols
  - Neurologist
  - Professional psychological support

Discussion with Patient

- Address psychosocial issues
  - anticipatory guidance (the what ifs)
  - patient and family reaction to diagnosis
  - need for community support services

- Pre-symtomatic testing usually requires more that one visit
  - Visit 1: information and counseling
  - Visit 2: neurological examination with additional counseling
  - Visit 3: results
  - Many centers now combine visits 1 &2.

Disclosure and Follow-up

- Disclosure of the test result
- Implications for patient and family
- Coping strategies

- Follow-up in short and long term
- Make resources available.

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Huntington’s Disease Society of America

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