Hereditary Nonpolyposis Colon Cancer: Disclosure of genetic information

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Genetics/Ethics Presentation
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Case Study

- Mr. Miralax – 42 year old white male presents to his PCP with change in bowel habits and RUQ abdominal pain.
- CT- mass in the hepatic flexure and lymphadenopathy anterior to superior mesenteric vessels
- Referred to GI Specialist
Case Study - Colonoscopy
Hereditary Nonpolyposis Colon Cancer (HNPCC)

• AKA- Lynch Syndrome
• Originally called- Cancer Family Syndrome
• Lynch Syndrome I- familial colon cancer
• Lynch Syndrome II- HNPCC associated with other cancers of the reproductive system
• Prevalence 2 to 5 per 1000
• Accounts for 3-8 % of colorectal cancers

Nussbaum, McInnes & Willard, 2001
HNPCC

- Other cancers associated with HNPCC
  - Endometrial cancer
  - Ovarian cancer
  - Gastric cancer
  - Transitional cell carcinoma
  - Adenoca of small bowel
  - Glioblastoma- Turcot syndrome

Munoz & Lambiase, 2009
HNPCC - Genetic Inheritance

- Heterogeneous autosomal dominant
- DNA Mismatch repair gene mutation

Nussbaum, McInnes & Willard, 2001
HNPCC - Molecular Nature

- Gene
  - **MSH2**
  - **MLH1**
  - **PMSL1**
  - **PMSL2**
  - **MSH6**
  - **MSH3**
  - **EX01**

- Chromosomal location
  - 2p22-p21
  - 3p21
  - 2q31.1
  - 7p22
  - 2p16
  - 5q14.1
  - 1q43

Nussbaum, McInnes & Willard, 2001
Munoz & Lambiase, 2009
HNPCC- Diagnosis

- 3 stage process
  - Review of family history
  - Tumor testing
  - Genetic testing
HNPCC- Family History

• Diagnostic Guidelines
  – Amsterdam Criteria- families that should be classified as having HNPCC
    • At least 3 members with colon cancer- 1 first degree relative
    • 2 generations
    • One case before age 50
  – Amsterdam II Criteria- 1998
    • Include extracolonic HNPCC associated cancer
    • 2 first degree relatives
    • 2 generations
    • One case before age 55

Umar, Risinger, Hawk & Barrett, 2004
**HNPCC- Family History**

- Bethesda Guidelines
  - Recommend genetic testing for:
    - those diagnosed before age 50
    - presence of synchronous or metachronous colorectal or other HNPC C associated cancer regardless of age
    - colorectal cancer with the MSI-H histology in a patient under the age of 60
    - One or more first degree relatives with colorectal cancer or other HNPCC-related tumor with one before age 50
    - Colorectal cancer diagnosed in 2 or more first or second degree relatives with HNPCC related tumor regardless of age

Umar, Risinger, Hawk & Barrett, 2004
HNPCC - Tumor testing

- Immunohistochemistry (IHC) testing
- Microsatellite Instability (MSI) testing - seen in 90% of patients with HNPCC

Munoz & Lambiase, 2009
HNPCC- Genetic testing

Munoz & Lambiase, 2009
HNPCC- Genetic testing

- Provides definitive diagnosis
- RISKS
- BENEFITS

Munoz & Lambiase, 2009
HNPCC- Treatment

• Early recognition and screening
• Surveillance colonoscopy
  – age 25 then every 2-3 years thereafter
  – after age 40 every 1-2 years
HNPCC- Treatment

• Surveillance for extracolonic cancers
  – Pelvic exams, ultrasounds, endometrial biopsies

• Surgical treatment
  – Prophylactic colectomy
  – Total colectomy
  – Subtotal colectomy

Munoz & Lambiase, 2009
Case Study

• Mr. Miralax undergoes right hemicolecotomy and returns to GI Specialist for follow-up visit
• Referred to cancer genetics clinic
• FH- Paternal grandfather died at age 70 with metastatic colon cancer. Paternal great grandmother- colon cancer. Sister- “female” cancer at age 30.
• SH- Married, 1 daughter (15 years old)
Case study

• Genetic testing- + HNPCC
• Returns to GI Specialist 2 weeks after obtaining results of genetic testing and counseling
• Mr. Miralax- tells the APRN that he has a 24 year old son from a previous relationship and asks if he is a risk for HNPCC
• Mr. Miralax refuses to contact him regarding the results
Ethical Dilemma

• The ethical dilemma in this case is: Should the health care provider notify the son who is at risk for HNPCC?
The rights, wellbeing, and safety of the individual patient should be the primary factors in arriving at any professional judgment should be the primary factors in concerning the disposition of confidential information received from or about the patient, whether oral, written or electronic.

ANA, 2001
Institutional policies/Professional ethical code

- Institute of Medicine Committee on Assessing Genetic Risk recommends breaching confidentiality about genetic risk when:
  - Attempts to obtain voluntary disclosure fail
  - High probability of harm to the relative at risk
  - Disclosure will prevent harm
  - Disclosure is limited to information that is needed for diagnosis and treatment
  - No other way to prevent the harm

Beauchamp & Childress, 2009
Institutional policies/Professional ethical code

- American Medical Association- Council on Ethical and Judicial Affairs
  - “Genetic Miranda Warning”

- American Society of Human Genetics

Offit, Groeger, Turner, Wadsworth & Weiser, 2004
Analysis of Ethical Principles

- Autonomy
- Beneficence- Duty to warn
- Non-malificience
- Justice

Beauchamp & Childress, 2009
Other ways of reasoning

• Genetic information
  – Property of the proband or the family?

de Wert, 2005
Lo, 2009
Legal issues

• HIPAA
  – Privacy Rule
    “Covered entities may disclose protected health information that they believe is necessary to prevent or lessen a serious and imminent threat to a person or the public, when such disclosure is made to someone they believe can prevent or lessen the threat (including the target of the threat)”

Offit, Groeger, Turner, Wadsworth & Weiser, 2004
United States Department of Health & Human Services, 2003
Legal Issues

"I'M SORRY BUT DUE TO NEW HIPAA REGULATIONS ALL PATIENTS MUST WEAR MASKS."
Legal Issues/Cases

- Pate vs. Threlkel
- Safer vs. Estate of Pack
Plan/Implementation

• Discuss disclosure of genetics testing
• Informing relatives by letter
  – Study by Suthers, Armstrong, McCormick & Trott
    • Genetic status was clarified in 40% of the relatives that received a letter compared to 23% that were notified by the patient
Summary

• When considering the Duty to Warn relatives of genetic risks the APRN must consider the:
  – risk of the disease
  – effectiveness of preventative interventions
  – emerging legal considerations and liabilities

Offit, Groeger, Turner, Wadsworth & Weiser, 2004
Reference


Reference


