Ethical issues in genetic counselling in Hereditary Cancer

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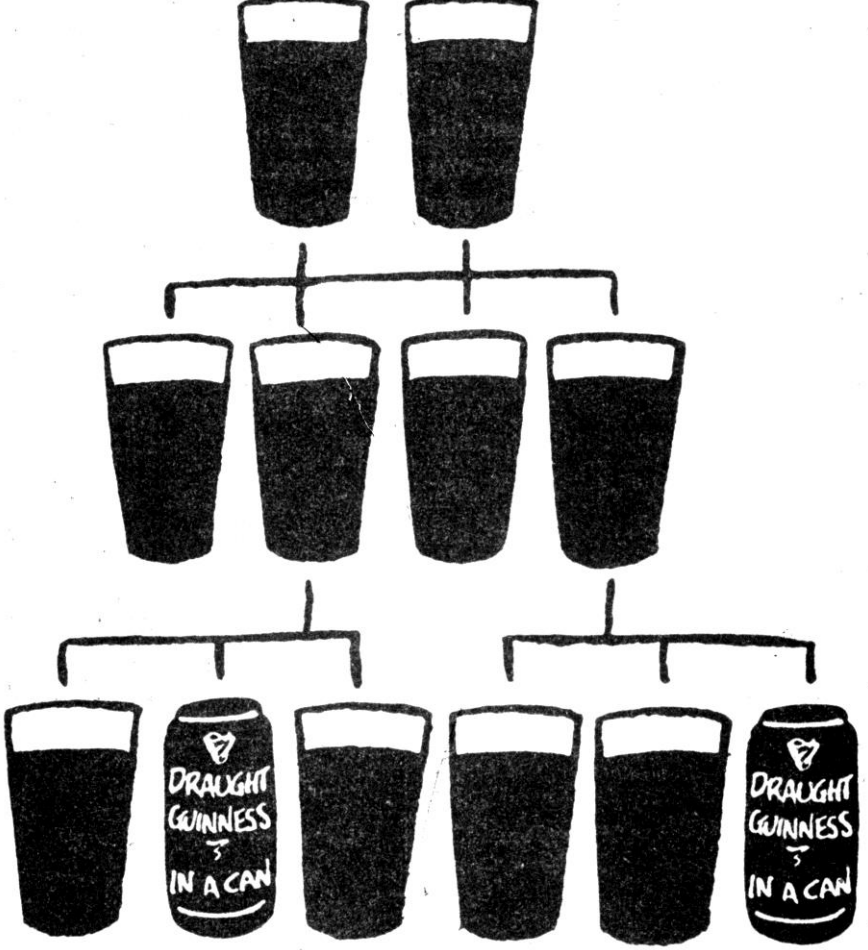


Taste - Some people are just born with it.

Ethical Issues

- Not unique to genetics
- Genetics is unique in that ***ethical issues rarely only relate to one person in a family.***
- Work with families, not just individuals
- Decisions can affect other family members
- Family members do not always get on with each other!

4 medical ethics principles

1. **Respect for autonomy:** enabling individuals to make their own reasoned informed choice
2. **Beneficence:** acting in the best interest for the patient
3. **Non-maleficence:** Avoiding unnecessary harm to the patient
4. **Justice:** Acting in fairness, equal care for all

Beauchamp and Childress (2008) 6th Ed. Principles of Biomedical Ethics, OUP

Autonomy

- Self determination
- Reflect on own life, act intentionally
- Free from coercion
- Competent to make decisions

Beneficence

- Obligation to prevent harm
- To do good
- Further the patient's welfare and interests

Non-maleficence

- The obligation not to inflict harm
- Minimise risk to the patient

Justice

- Healthcare provided in fairest possible way
- Equity of service and access
- Respect for individuals and their rights

Common Issues in Practice

- Childhood testing
- Non disclosure
- Informed choices in prenatal testing
- Right to know/ or not know
- Sharing of information/ confidentiality
- Coercion
- Relevance of testing for low penetrance mutations

Common ethical scenarios

- Testing young child (e.g.FAP) before the age at which screening would begin
- Testing (e.g.MEN2) young child prior to adoption
- Testing grandchild of affected person with untested intervening relative
- Affected individual unwilling to divulge results to extended family
- Testing in pregnancy
- Unknown or low penetrance of mutation

Example 1
Testing 2nd. degree relative

- Known 65y old carrier of VHL mutation, with severely affected siblings
- Her son aged 40y doesn't want to be tested, but his estranged daughter aged 21y wishes to be tested.
- If she has the mutation you have inadvertently tested her father

Suggested strategy

- Discuss the issues with the individual, explain benefit to him of the test, explore reasons for denial and non-disclosure
- Explain the benefits of testing for his daughter
- Should you offer the test to the daughter if her father is adamant he doesn't wish to release the information to her?

Example 2
A woman with breast cancer and a *BRCA1* mutation

- She refuses to allow you to contact her estranged daughter aged 26y to arrange genetic counselling for her

Issues

- Confidentiality
  - Why is proband not sharing information with her daughter?
    - acting maliciously?
    - Protecting?
  - Should we break confidentiality?
    - Autonomy and privacy of proband
    - Right of others to know

Reasons for Non-Disclosure

- Limited understanding of the genetic diagnosis
- Denial or guilt
- Not wishing to be the messenger of bad news/ desire to protect
- Judgement about the person's ability to cope
- Conflict, or difficult family relationships
- Not able to find the right time/ way to give the information

Confidentiality

- Enshrined in law
- Can confidentiality be breached?

Genetic information

- Who owns genetic information?
  - Individual?
  - Should families own genetic information?
- Who has the right to release/withhold genetic information?

Confidentiality issues in genetic counselling

Debate on ownership of genetic information

Individual ownership

V

Family ownership

Should families own genetic information?

BMJ 7 July 2007, V335, 22-23

- Genetics involves families - genetic information about one person has implications for relatives
- Patient confidentiality and autonomy: cornerstones
- Tension between maintaining duty of confidentiality, and disseminating genetic information to others, if intervention could decrease morbidity and mortality

The arguments for and against

‘Joint account model’

Genetic information should be available to all “account holders” (family members)

- “if anyone should own genetic information it should be all those at risk”
- “Modern medicine must facilitate sharing information”

- Arguments against ‘Joint account model’:
- “Harm from failure to disclose will usually not entail immediate/grave damage”
- “Genetic information should be private and personal”
- “Specific disease mutation should belong to health service, not individual/family”

New Reform to Australian law

- Amendment in 2006 to the Privacy Act 1988 introduces a new clause allowing health practitioners to disclose genetic information to genetic relatives without the patient's consent where there is a serious threat to life, health or safety of the genetic relative, and disclosure is necessary to lessen or prevent that threat

What to consider when making an ethically sound choice

- Questions:

  -Is the patient your only concern?

  -Do we always know what is good for the patient? (Conflict between autonomy and beneficence)

- Finding a balance & minimising harm

Example 3, testing children

- A woman with FAP has had difficult surgery after colectomy due to extensive abdominal desmoids, and is seriously ill.

- She has two children aged 3y and 5y. She wants to arrange predictive tests for them now. Her husband does not.

Genetic testing in children

- Children are usually only tested if the result will benefit the health of the child before they reach adulthood
  - Childhood onset inherited cancer e.g. von Hippel-Lindau disease or FAP. Some screening starts at 5 yrs
- For adult-onset hereditary cancers testing is usually deferred until the person can give informed consent
  - e.g. Lynch syndrome, Hereditary breast and ovarian cancer

Issues

- Benefit of testing children before medical intervention appropriate
  - Reduce/relieve anxiety (may also increase anxiety)
  - Child/parent relationship
- Benefit of waiting until child is able to make informed decision
  - Maintain autonomy of child – child's right not to know
  - Avoid child being labelled and/or feeling a burden

Example 4: A couple asks for a test for *BRCA1* mutation in pregnancy

- 35y old pregnant woman is affected and has had bilateral breast cancer.
- She is tested and found to have a female baby with the mutation
- The couple decides to continue the pregnancy

Genetic testing in pregnancy

- Decision-making at an emotional time with a time-limit
- Couples with different views/experiences of cancer
- Prenatal testing or Pre-implantation diagnosis?

Testing in Pregnancy

- For which conditions should this be offered? Adult onset hereditary cancer? Consider the potential for improved management in future
- Implications if couple decides not to terminate an affected pregnancy with late-onset hereditary cancer; predictive test on baby without it's consent
- *BRCA* mutation test - keep male carrier pregnancy?

Example 5: Carney-Stratakis syndrome

- (Carney dyad: paragangliomas and gastric stromal tumours)
- Phaeochromocytoma at 46y
- Benign low grade gastric stromal tumor (CD34 +ive) derived from smooth muscle
- 43y fundic gland polyps
- 48y uterine fibroids

Surveillance

- Annual 24 h. urinary catecholamines and MRI 2 yrly for paragangliomas/phaeochromocytoma, endoscopic ultrasound 6 monthly for gastric tumors.

- Family screening? 2 daughters 9y and 17y Parents healthy, in late 70's, paternal uncle died of stroke aged 58y

Mutation analysis

- No mutation in *SDHA, SDHC, SDHD, VHL, PTEN, PTCH*, Ret, Max, *TMEM12-7* (variant)
- *SDHB* revealed 6bp intronic duplication.
- Uncertain pathogenicity. Requires family segregation studies. Parents tested, father carried the variant. 24h urine normal.
- Surveillance for children?

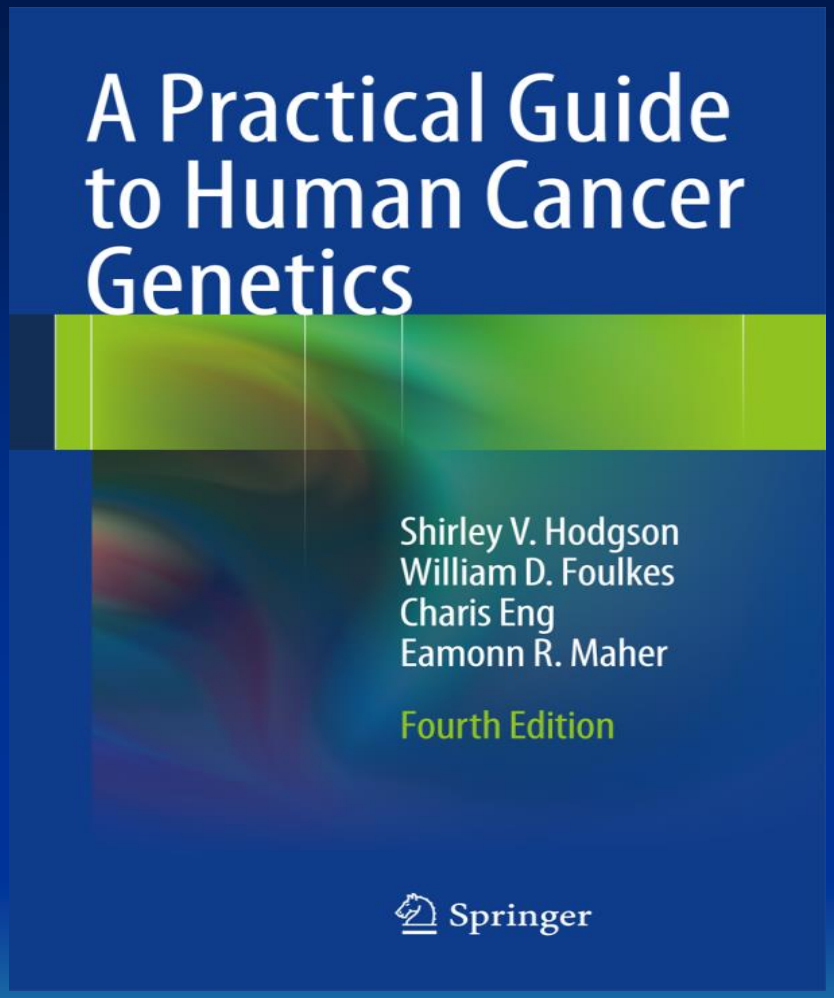
Clinical Ethics Committee

- What is the ethical question considered?
- Has this question been discussed in the department?
- What difficulties arose out of these discussions?
- Cases would only be considered by CEC if clinicians could identify a clear ‘ethical question’, and had been unable to resolve that question within their department.

Summary

- Genetics can be ethically complex as it involves families and not just individuals
- Help identify barriers to disclosure and develop strategies to overcome these
- When faced with a difficult ethical dilemma consider the ethical principles, assess situation in discussion with other team members
- Contact clinical ethics committee if necessary


what's my risk
for cancer?


A Practical Guide to Human Cancer Genetics
Shirley V. Hodgson
William D. Foulkes
Charis Eng
Eamonn R. Maher
Fourth Edition
Springer