Ethical Issues in Genetic Testing: the Duty to Warn At-Risk Relatives

Nathaniel H. Robin, MD
Professor, Departments of Genetics and Pediatrics
ELSI research in clinical genetics

• No shortage of questions, and the list is only growing – new disease genes, new tests (WES)
• My research questions have been generated from my clinical experience
  – How patients, families, and non-genetic healthcare providers view and utilize genetic testing
  – Confidentiality of genetic information, and the duty to warn at-risk relatives
How patients, families, and non-genetic healthcare providers view and utilize genetic testing and genetic information

• Model: genetic testing for deafness*

• General conclusions
  – Genetic testing must be accompanied by pre- and post-test genetic counseling
  – Genetics v. non-genetics healthcare providers

*Brunger et al, 2000 (a,b); Robin et al, 2001; Duncan et al 2007
Genetics v. non-genetics professionals

- Patients/families tested by non-genetics healthcare providers have poor level of understanding about their test results
  - Comparable to group that was never tested
  - Far less than those tested by genetics professionals
  - High level of understanding
  - Low level of effective communication

-> lack training in genetic counseling
Future

• This question is becoming more relevant, more complex

• New tests in new specialties
  – Cardiology: cardiomyopathy, arrhythmia
  – Forensic pathology: arrhythmia
  – Neurology: ataxia, dementia
  – Hematology: clotting
Genetic testing is a family affair

- Questions also came from clinical experiences
- Genetic testing and genetic information is different from other medical tests
  - Genetic exceptionalism
- Familial nature of genetic information
- Confidentiality of genetic information and the duty to warn at-risk relatives
- Disclosure without consent
Genetic exceptionalism*

• Belief that genetic information is special and therefore should be treated differently from other types of medical information
• Not a good idea *in theory*, as it imbues a magical aura to genetic information that is neither accurate nor helpful
• In the real world, however, genetic test information *is* different, at least for now

Genetic testing is a family affair

- Genetic information is not personal
  - Positive tests create risks for biologic relatives
    - Grandchild & grandmother test positive for BRCA1, intervening relative is too

- What is obligation to at-risk relatives
Divulging information about relatives...

• ...is not unique to genetic testing
  – E.g., TB, STDs
• But is different in that it is only biologic relatives
  – TB: ALL contacts, household & otherwise
  – STDs: more intimate contacts, maybe less well-known
Betty tests positive for a BRCA1 mutation. She refuses to tell her sister Sue.

Q1: Does Sue have a right to know?
Q2: Would you tell Sue?
Confidentiality

• Confidentiality of medical information is a primary principle of medicine

• But, does the familial aspect of genetic information alter the right and/or expectation of confidentiality?
  – do relatives have a right to this information?
So...?

• Does daughter’s right to know overcome patients right to privacy?
  – Autonomy v beneficence v nonmaleficence

• Does severity of disease matter?
  – BRCA1? Huntington Dis? Color blindness?
Duty to warn

• First established in context of contagious disease
  – disclosing did not conflict with confidentiality - party was aware of illness

• Direct conflict in *Tarasoff v. Regents of Univ. California*
  – U.S. Supreme Ct: psychiatrist had duty to take reasonable steps to protect 3rd party of danger from patient
  – this duty overruled confidentiality
Duty to warn, genetic information & the law

• Uncertainty over what circumstances create duty to warn
• Decided on case-by-case basis
• *Pate, 1994; Safer, 1996*: judgements against physicians who did not warn family
  – what satisfies ‘warned’?
• No legislation addresses this
  – Professional guidelines? HIPPA?
Studies

• Present theoretical scenarios to medical geneticists, genetic counselors, and non-genetic physicians
• MG more likely to disclose than GC, but both GC & MG less likely to disclose than non-genetics MD*
• More recent studies focus on cancer and sudden death risks, still theoretical

Disclosure of genetic test results without consent

• What is the actual clinical experience with this dilemma?
  – different than theoretical-case studies?

• How common?

• Do genetics professionals follow their society guidelines (ASHG, NSGC)?
  – do they know they exist?
Two studies

• Questionnaire-based studies examined genetic counselor’s¹ and medical geneticists² experience
• Descriptive data analysis
• Pre-HIPPA, pre-GINA

Results

• Genetic professionals face the issue of disclosure commonly (GC: 46%; MG: 66%)

• Seldom seriously consider disclosure w/o consent (1GC, 2 MD)

• Both used colleagues as primary resource
  – few (12/15%) consulted ethics committees
  – GC and MG cited similar but not identical reasons
    • Not surprising, as their training is different
Conclusions

• Relevance of these studies can be questioned, as many things have changed
  – HIPPA
  – GINA
  – Whole Genome Sequencing
• Studies should be repeated