Informing Family Members About Their Genetic Risk:

Is it Prohibited, Required, or Permitted?

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Session Objectives

• Understand the ethical, legal and social issues involved with duty to inform

• Improve comprehension of ethics principles by actively engaging participants
Audience participation exercise

Dr. Janice Jones, a pediatrician, has treated the Smith family for years. The three Smith daughters, Alice, Betsy, and Carol, all were patients of Dr. Jones. Now that each of the Smith girls is married with children of her own, Dr. Jones takes care of their children. One day, Alice arrives for an appointment with her 10-year-old daughter, Andrea. Alice tells Dr. Jones that, on a lark (and to take advantage of a 2 for 1 special from a mail order genomics company), both Alice and Andrea have undergone genomic analysis.
Both Alice and Andrea have tested positive for a BRCA1 mutation. Alice indicates that she will consult with her own primary care doctor, but she wants to know what Dr. Jones recommends for Andrea. Dr. Jones asks whether Alice has told Betsy (who has a daughter, Barbara) and Carol (who has a daughter, Cathy). Alice says that she can't bring herself to tell them and does not want Dr. Jones to do so.
Dr. Jones is concerned that, as the physician for Barbara and Cathy, she has a duty to inform Betsy and Carol. But she also has a duty of confidentiality. Dr. Jones wants to tell Betsy and Carol, but she isn't sure what she should do.

Based on your understanding of legal and ethical considerations, can Dr. Jones disclose this information?
Can Dr. Jones disclose this information?

A. Yes
B. No

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CONSIDERATIONS

1. Protected health information cannot be disclosed to a third party without the consent of the patient.

2. The fact that the third party is also a patient of the physician (or the parent of a minor patient) does not provide an exception.

3. The finding of a BRCA1 mutation does not require immediate action, and it might be more appropriate (and have a different result) if the issue of disclosure is raised again at a later time.
QUESTION TO ADDRESS:

Based on the results of genetic testing of a child, does a pediatrician have a duty to inform other family members about their genetic risk?
KEY ILLUSTRATIONS

Prohibited disclosures: HIPAA Privacy Rule

Required disclosures: Wrongful birth; Continuing duty to notify

Permitted disclosures: American College of Medical Genetics and Genomics
1. PROHIBITED DISCLOSURES: HIPAA PRIVACY RULE

45 C.F.R. § 164.502

(g)(1) Standard: personal representatives.

(3) Implementation specification: unemancipated minors.

(i) If under applicable law a parent, guardian, or other person acting in loco parentis has authority to act on behalf of an individual who is an unemancipated minor in making decisions related to health care, a covered entity must treat such person as a personal representative under this subchapter, with respect to protected health information relevant to such personal representation, except that such person may not be a personal representative of an unemancipated minor, and the minor has the authority to act as an individual, with respect to protected health information pertaining to a health care service, if:
(A) The minor consents to such health care service [and no other consent is required by law];

(B) The minor may lawfully obtain such health care service without the consent of a parent, guardian, or other person acting in loco parentis; or

(C) A parent, guardian, or other person acting in loco parentis assents to an agreement of confidentiality between a covered health care provider and the minor with respect to health care service.

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EMANCIPATION OF MINORS

The HIPAA Privacy Rule defers to state law on the issue of when a minor is emancipated.

Generally, the legal age of majority is 18, but a younger minor may be considered emancipated under the following circumstances:

1. Self supporting and/or not living at home
2. Married
3. Pregnant or a parent
4. In the military
5. Declared emancipated by a court

“Mature minors” may be given the right to consent to medical treatment without parental involvement for certain conditions (e.g., STD, substance abuse, pregnancy).
ABUSE, NEGLECT, OR ENDANGERMENT

(5) Implementation specification: abuse, neglect, endangerment situations.

Notwithstanding a State law or any requirement of this paragraph to the contrary, a covered entity may elect not to treat a person as a personal representative of an individual if:

(i) The covered entity has a reasonable belief that:
   (A) The individual has been or may be subjected to domestic violence, abuse, or neglect by such person; or
   (B) Treating such person as the personal representative could endanger the individual; and

(ii) The covered entity, in the exercise of professional judgment, decides that it is not in the best interest of the individual to treat the person as the individual’s personal representative.
TO SUMMARIZE (SO FAR)

- In general, a physician may provide a parent or guardian with a child’s health information, including the results of genetic testing.

- A physician may not disclose health information, including the results of genetic testing, to relatives or other third persons unless authorized by the parent or guardian.
2013 INTERPRETATION OF THE HIPAA PRIVACY RULE


“[A] health care provider may share genetic information about an individual with providers treating family members of the individual who are seeking to identify their own genetic risks, provided the individual has not requested and the health care provider has not agreed to a restriction on such disclosure.”
This interpretation is deeply troublesome for the following reasons:

1. It puts the burden on the patient to seek to restrict disclosure of his or her health information, which runs counter to the medical ethics presumption of nondisclosure.

2. It provides the physician with a veto power to deny the patient’s request for a restriction, with the result that it authorizes physicians to disclose sensitive information over the objection of their patients.
3. It directly contravenes Section 2.131 of the AMA Code of Medical Ethics, Disclosure of Familial Risk in Genetic Testing, which provides that “Physicians have a professional duty to protect the confidentiality of their patients’ medical information, including genetic information.”
4. It directly contravenes the following statement from the American Society of Human Genetics.

“Disclosure without the consent of the patient is only permissible when “attempts to encourage disclosure on the part of the patient have failed; the harm is likely to occur and is serious, imminent, and foreseeable; the at-risk relative(s) is identifiable; and the disease is preventable, treatable, or medically accepted standards indicate that early monitoring will reduce the genetic risk.”
WHAT IS THE EFFECT OF THE HIPAA INTERPRETATION?

• It does not require disclosure. It permits a health care provider to disclose genetic information without violating the HIPAA Privacy Rule.

• It does not address whether disclosing the information contravenes ethical guidelines.
• The 2013 interpretation permitting disclosure of genetic information is questionable.

• The Privacy Rule provision applicable to disclosure of a minor's health information prohibits any disclosures without the consent of the parent or guardian, except in rare circumstances (not present here).
2. REQUIRED DISCLOSURES

• This means a physician who fails to disclose certain health information could be found liable for medical malpractice.
Wrongful birth

Wrongful birth is a type of medical malpractice case in which parents of a child with severe congenital anomalies claim that their physician failed to warn them of the risk of conceiving or giving birth to a child.

The parents seek to recover the extraordinary costs of treating and raising a child with serious health problems.

These lawsuits are permitted in 29 jurisdictions, prohibited in 8 jurisdictions, and the others have not yet addressed the issue.

Typically, the cases are brought against OB/GYNs.
Wrongful birth cases against pediatricians

Pediatrician negligently fails to diagnose a genetic disorder in a child or fails to inform the parents of the genetic nature of the disorder.

The parents have a second child who also is born with the disorder.

The parents assert that if they were informed by the pediatrician, they would not have had a second child, they would have used PGD, or they would have adopted.
Leading cases:


Lininger ex rel. Lininger v. Eisenbaum, 764 P.2d 1202 (Colo. 1988) (Leber’s congenital amaurosis)
Continuing notification

Traditionally, physicians’ duties were limited to the clinical encounter or episode of care.

They had no duty to inform the patient of new medical information or discoveries because the burdens of doing so outweighed the potential benefits.

With new HIT, the burdens are minimal (e.g., Cleveland Clinic and the recall of rofecoxib in 2004).
Proposed guidelines on notification

1. Only applies to patients seen within the last 3 years (based on Medicare billing rule on “established” patient).

2. For multi-specialty groups, HMOs, clinics, etc., the institution would have the duty to provide the notice.

3. Patients differ in their desire for updates, and patient preferences could be noted in the medical record.
4. Potential reasons for notification:
   (a) Drug interactions, adverse events, market withdrawals
   (b) Medical device recalls and warnings
   (c) Changes in important lifestyle recommendations (e.g., diet, exposures)
   (d) New treatments with enhanced safety and efficacy

5. When, if at all, would new evidence on genetic risk rise to this level?
3. PERMISSIBLE DISCLOSURES:

AMERICAN COLLEGE OF MEDICAL GENETICS AND GENOMICS

With cheaper WGS, there will be pressure (from payers, parents, commercial entities) to perform WGS, even if the main purpose is to answer a single diagnostic question.

Incidental findings are inevitable, with estimates of 100-313 “abnormal” findings with every WGS.
In March 2013, the American College of Medical Genetics and Genomics issued a statement proposing that whenever genome sequencing is ordered in the clinical setting, laboratories have a mandatory duty to analyze 56 genes and to report the results to the clinicians and patients, regardless of the patient’s age, medical condition, or desire to obtain the information.

The theory is that some conditions are serious and treatable and that it would be irresponsible not to return these results.
This proposal has been very controversial.
Among the reasons for disagreeing with this recommendation:

1. Who is going to pay for the extra cost of interpretation?
2. It overrides the patient’s autonomy.
3. It overrides the clinical judgment of the ordering physician.
4. More information is not always beneficial.
A PROBLEM-SOLVING GUIDE FOR DISCLOSURES OF PROTECTED HEALTH INFORMATION

1. Is disclosure prohibited by law, such as HIPAA or FERPA, or a provision in a code of ethics?

2. Is disclosure required by law, such as a court order or common law duty, or a provision in a code of ethics?
3. If the answers to #1 and #2 are no, then disclosure is permissive or optional. What considerations should be used?
   a. What do non-binding ethical provisions suggest?
   b. Are there any instructive analogs?
   c. What are the likely consequences of a decision either way?
d. Will either decision set a bad precedent or constitute a slippery slope?

e. Are there any likely, unintended consequences?

f. Is your decision something you would be comfortable defending to other patients, your colleagues, or the public?

g. Have you consulted with experts?
1. Lainie Friedman Ross, Mark A. Rothstein, & Ellen Wright Clayton, Mandatory Extended Searches in All Genome Sequencing: “Incidental Findings,” Patient Autonomy, and Shared Decision Making, JAMA 310(4) (2013): 367-368 (criticizing the ACMG recommendations on mandatory return of all results)

2. Mark A. Rothstein, HIPAA Privacy Rule 2.0, Journal of Law, Medicine & Ethics 41(2) (2013): 525-528 (criticizing amendment permitting disclosure or genetic test results to relatives without the consent of the individual)
