Duty to Warn Relatives at Risk of Genetic Disease

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Introduction

Breast cancer is a significant universal health concern within the female population. The American Cancer Society (ACS) (2008) reports that one in eight (12%) women will develop breast cancer in their lifetime. It is the second leading cause of cancer death in women, being the source of one in thirty-five of their deaths (American Cancer Society, 2008). In 2008, approximately 182,000 new invasive breast cancers will be diagnosed and 40,400 women will lose their lives due to this disease in the United States alone (ACS, 2008).

While most breast cancers do not follow a familial pattern, a genetic component of breast cancer has been identified. Breast cancer 1 (BRCA1) and breast cancer 2 (BRCA2) mutations are believed to account for the majority of hereditary breast and ovarian cancers (Allain, 2008). The BRCA1 gene was discovered in 1990 and tends to be associated with more aggressive tumors that are often estrogen and progesterone receptor negative (Domchek & Weber, 2004). The BRCA2 gene was identified in 1995, and mutations may contribute to fewer cases of early-onset breast cancer; however, a correlation with an increased risk of ovarian, prostate, and pancreatic cancers has been established (Domchek & Weber, 2004).

Cancers stemming from alterations in the BRCA genes have distinctive features. Early age of diagnosis, the presence of bilateral versus unilateral disease, vertical transmission from mother or father, and associated tumors that originate in the ovaries, endometrium as well as other locations are characteristic of these particular genetic aberrations (Domchek & Weber, 2004). Approximately 5-10% of women who are diagnosed with breast cancer will have an alteration within these specific genes (National Cancer Institute, 2002). Women positive for the BRCA mutation are estimated to have a 55-85% increased risk of breast cancer and a 15-60%
increased risk of ovarian cancer as compared to 13.2% and 1.4% of the general population respectively (National Cancer Institute, 2002; Halbert, 2004).

General Ethical Dilemma

Now that susceptibility genes for breast and ovarian cancer have been identified, it is possible for evaluation and testing to be done to detect those with the BRCA mutation. While the ability to perform these genetic assessments is of substantial benefit to providers as well as patients and family members, it brings to the forefront new ethical issues. One of these concerns is referred to as the “duty to warn.” Professional groups such as the American Society of Clinical Oncology (ASCO) and the American Society of Human Genetics (ASHG) have communicated their policy statements concerning the health care professional’s obligations and responsibilities when the subject of disclosing familial genetic information arises (i.e. duty to warn). These will be explored further later in this paper.

This matter presents challenges to providers that extend to both the patient and their family. Patterson, Robinson, Naftalis, Haley, and Tomlinson (2005) from the University of Texas Southwestern Medical Center in Dallas, Texas raise the following questions that surround the duty to warn topic: (1) Do providers/clinicians have the obligation to inform family members of a positive genetic test result if the patient dies before the results are available?; (2) If providers/clinicians do not inform members of the family, should the action, or lack there of, be considered negligent?; (3) If providers/clinicians are forthcoming with positive test results, is the responsibility of confidentiality then violated? The duty to warn relatives at risk of genetic disease is a multifaceted, complex process that demands further attention and has the capacity to make a mark on the lives of many individuals.
Summary of the Case

In the case that will be reviewed in this paper, there are both primary and secondary stakeholders involved or affected by this ethical dilemma. The primary stakeholders include the provider/clinician (Emily and Sarah), the individual with breast cancer (Mrs. Romero), and the immediate family members (Mr. Romero and the children) (See Appendix A). Secondary stakeholders are comprised of those healthcare professionals and members of society for which breast cancer and associated genetic analysis might impact their daily personal or professional lives. Mrs. Romero, a 42-year old Hispanic female with end-stage breast cancer is admitted to the hospital for pain control. She has made the decision to discontinue chemotherapy, and once her pain control has been optimized, she will go home with hospice care. She is requesting a BRCA test be done due to her strong family history of breast and ovarian cancer. She has a daughter who is twenty-one years old and 20 weeks pregnant with her first child as well as a son sixteen years of age. Sarah, the women’s health nurse practitioner participating in Mrs. Romero’s care, consults the genetic counselor, Emily, regarding the patient’s request for genetic testing.

Mrs. Romero did not have the BRCA test performed at diagnosis three years prior because her husband was against the evaluation. While he remains uncertain about the real necessity of this test, he is now supporting his wife with her decision. Within the Hispanic culture the male and female relationship is considered to follow a more traditional gender specific pathway (Galanti, 2003). Patriarchal power distinguishes the male from the more maternal role of the female (Galanti, 2003). Women usually do not make decisions independently, but are encouraged to defer to the ability and influence of their husbands (Galanti, 2003). Mrs. Romero realizes that her condition is terminal and is very worried about her daughter’s future health. Unfortunately Mrs. Romero dies before she receives the BRCA results,
and her husband, the next of kin, refuses to inform their daughter even after Emily and Sarah discuss the implications to her health with him. The right of individuals who are directly affected by the results of genetic assessments to be informed of the outcomes as well as the provider’s duty, right, or privilege to disclose the results to these individuals becomes the dilemma that will be explored through the context of various ethical frameworks and principles within this paper.

Clarification and Expansion of the General Ethical Dilemma

A significant issue representing the foundation within this ethical situation includes the question of who is ultimately considered the “patient” when matters of DNA are at hand. The patient used to constitute only the individual who presented with symptoms of a disease which was subsequently diagnosed, treated, and resulted in cure, maintenance of the illness, or death (Patterson et al., 2003). Due to the initiation of genetic evaluation for hereditary cancers, implications reach far beyond that of the person being treated for the illness. Mrs. Romero died unaware of her genetic status, and the only family member designated to be contacted was her husband. The future, life-long health status of her daughter is of great importance and concern given that the individual in charge of the results now, Mr. Romero, is declining to discuss the conclusion of the BRCA test with her or meet with the medical providers regarding implications of the outcomes at this time. Do providers have an obligation of disclosure to the daughter and family in order for negligence of care to be avoided?

The entity of patient confidentiality must be a paramount consideration as well. Now that Mrs. Romero is deceased, do health care professionals have the right to notify relatives whose well-being could be affected by results of the genetic test (Patterson et al., 2003)? Mrs. Romero’s healthcare providers must bear in mind the statues set forth in the Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule. The goal of this regulation is to ensure that
personal health information remains private and that the health and welfare of the public is protected (United States Department of Health and Human Services, 2003). These rules place a legal duty on the provider to protect a patient’s privacy. They describe a personal representative as “…a person legally authorized to make health care decisions on an individual’s behalf or to act for a deceased individual or the estate (United States Department of Health and Human Services, 2003, p. 16). Mr. Romero is in this case the next of kin and the person now in charge of his wife’s estate.

The principles of autonomy, nonmaleficence, and beneficence are standards surrounding this issue that are referred to frequently throughout the literature pertaining to this topic. The deontological and utilitarian ethical theories will also be evaluated within the realm of the duty to warn dilemma. Arguments substantiating and against the duty to warn at risk relatives considering these values and frameworks will now be explored.

Arguments Substantiating the Duty to Warn At Risk Relatives

Utilitarianism as described by Beauchamp and Childress (2009) refers to the ultimate standard of right and wrong. The justification of an act is measured by its consequences (Lo, 2005). Ultimately do the benefits of the action outweigh the harm that could arise if the action was not carried out? The correct act is the one that will result in the most favorable overall outcome (Beauchamp & Childress, 2009). Utilitarianism concentrates on “the value of well-being” and asserts that we should supply “…intrinsic goods such as happiness, freedom, and health that every rational person values” (Beauchamp & Champ, 2009, p. 337).

Within the context of the scenario being examined, the theory of utilitarianism would require that the daughter be told of her mother’s BRCA results thus fulfilling the provider’s duty to warn and “the greatest good for the greatest number” (Beauchamp & Champ, 2009, p. 337).
By disclosing Mrs. Romero’s results to her daughter she then will be able to undergo genetic counseling and testing for the mutation. If she is positive, she will then be informed of her options and begin screening processes that are appropriate for those with the BRCA mutation which include: beginning at the age of twenty, a clinical breast exam should be performed by a provider every six months (Burke, et al., 1997); at the age of twenty-five, mammogram ultrasound, and magnetic resonance imaging (MRI) should be done every year, rotating each of these modalities every six months (Burke, et al., 1997; Warner, et al., 2001).

The issues of confidentiality and Mr. Romero’s autonomous wishes regarding the handling of his wife’s test results will be not be considered in this case. Within the utilitarian approach these actions are justified because of the belief that the benefit of disclosing the BRCA results to the daughter outweighs the harm that would occur if she was not informed. Mr. Romero’s desires will not be taken into account as they are viewed as a detriment to the health and well-being of the daughter and therefore excluded as an influential factor within this case.

Utilitarianism looks at the probable consequences of particular courses of action (Beauchamp & Champ, 2009). Always seeking the greatest good, Utilitarians attempt to balance the concerns of all people involved. Mr. Romero has the legal right to the results of his wife’s BRCA test and is able to inform or not inform his daughter or any others that could potentially benefit from the information (i.e. perhaps Mrs. Romero’s brothers, especially if they have daughters) as he chooses. The fact that he may eventually decide to disclose the results does not supersede the elevated potential risk for the daughter to develop early onset, aggressive breast or ovarian cancer. The probability that the daughter will experience harm is greater than the potential negative effect on her father. Rules of confidentiality are set aside and the principle of

The principle of utility is also based on beneficence (Beauchamp & Champ, 2009). Beneficence requires that the provider proceed with actions that are in the best interest of the patient and promotes general safety and welfare (Lo, 2005). As noted earlier in this paper, the matter of who is considered the “patient” when the dissemination of genetic information is being considered is not concretely defined. Does the duty to promote others’ well-being outweigh the primary patient-provider affiliation (Godard, Hurlimann, Letendre, & Egalite, 2006)? This feeling of obligation is a powerful force in the minds of healthcare providers and thus leads to great discord in controversial situations that arise within the realm of genetic information disclosure. The fiduciary nature of the provider-patient relationship is the foundation for an association based on trust. If one regards a broader definition of the “patient” then informing those within this unit is validated.

Promotion of the future health and well-being of those relatives directly affected by the results of Mrs. Romero’s BRCA test (i.e. primarily the daughter) is certainly maintained within the literal context of the principle of beneficence. Promoting good and preventing harm is the core basis behind the duty to warn. The belief in mutuality and the support of the notion that the “patient is the family” substantiates the duty to warn family members of genetic risk when patients refuse to do so (Lacroix, Nycum, Godard, & Knoppers, 2008). At the heart of rendering medical care is upholding the value and worth of existing in a positive state of physical and psychological wellness. The conviction that Mrs. Romero’s daughter’s best interest is served through disclosure of the BRCA results is supported through the principle of beneficence. The
focus of providers is to elevate those they care for to their highest health potential, ideally acting at all times in their best interest.

Autonomy or “self rule” allows one the ability to act freely and without restriction by others (Lo, 2005, p. 10). In considering this principle inside the milieu of this dilemma, respect for the autonomy of the patient must be weighed against the respect for the autonomy of the family. Arguments supporting the duty to warn would assert that the family (particularly the daughter in this case) would be deprived of the ability of autonomous decision-making if they were not provided with the BRCA gene results.

How can one make informed healthcare choices without vital information that might very likely impact his/her ability to actively formulate these choices? There are many potentially preventive options and medical recommendations that the Romero’s daughter has available to her if she chooses them (i.e. more intensive screening regimens, surgical interventions, and future family planning considerations). According to Beauchamp and Childress (2009), in order for an action to meet the criteria of an autonomous action, an extensive level of comprehension and a “freedom of constraint” must be in existence (p. 101). If the Romero’s daughter is never informed of the study results, the future health decisions she will make will not meet the above mentioned qualifications of autonomy. The right to self-determination is not available to her if the BRCA results are withheld.

One must also consider the autonomous wishes of Mrs. Romero. While she has since passed away, her reasoning for obtaining the BRCA analysis was her concern for her daughter’s future health. Her verbalized feelings of apprehension and anxiety lead her to confide in her healthcare provider and eventually undergo testing. Mrs. Romero’s final wishes and desires would be regarded within this point of view.
Respect for the principle of autonomy in this case can be viewed from the standpoint of autonomy as a positive obligation. This standard proposes that autonomous choices should be fostered by considerately disclosing information and performing subsequent actions that promote self-directed assembly of personal healthcare decisions (Beauchamp & Childress, 2009). While Mr. Romero does not choose to inform his daughter of her mother’s BRCA results, this approach supports disclosing this information to family members. Without this data, the ability to act autonomously would be compromised and future health-related choices would be suboptimal and formulated without all necessary facts.

Arguments Against the Duty to Warn At Risk Relatives

The deontological ethical theory as depicted by Lo (2005) regards right and wrong as going above and beyond consequences of actions as a single concern. The inherent wrongness or rightness of a particular action is based on moral reasoning (Beauchamp & Childress, 2009). Moral judgments should not be based solely on a single situation and its consequences but on those that are relevant to all persons in similar circumstances (Beauchamp & Childress, 2009). Morality as perceived through this framework “…is grounded in reason, rather than in tradition, intuition, or attitudes such as sympathy” as in Utilitarianism (Beauchamp & Childress, 2009, p. 344).

Acting within the structure of this belief, the duty to warn is not supported. Breaching the HIPAA statutes of confidentiality is not condoned. Mr. Romero, as Mrs. Romero’s next of kin and legal “owner” of the test results, has the right to disclose the BRCA results as he sees appropriate. Confidentiality is the core of the provider-patient relationship. There are potential negative repercussions if the provider decides to circumvent Mr. Romero’s decision for non-disclosure of the outcomes of the test. An irreparable compromise could occur within the
relationship that had been established with Mr. Romero while caring for his wife. If rapport and subsequent trust is lost, the possibility of eventually making contact with Mr. Romero and fostering a therapeutic relationship that might allow him to make a more informed decision to tell his daughter of the results would be less likely.

The framework of deontology encourages a moral obligation to do what is “right” in a general sense regardless of the impact or consequences within individual situations. The accepted action coincides with the legal precedent set forth within written laws and standards. A universal duty to warn relatives of results would place the provider-patient relationship “…subservient to a more diffuse public health obligation…” (Offit, Groeger, Turner, Wadsworth, & Weiser, 2004, p. 1472). Providers should make every effort to counsel, educate, inform and persuade patients to share test results with relatives that are at potential risk; however, in order to uphold the views evident within this theory, a duty to warn is not mandated in this case.

The principle of nonmaleficence, first and foremost doing no harm, has long been the basis for traditional medical ethics (Beauchamp & Childress, 2009). The healthcare professional’s obligation as a trusted advisor to avoid injury or damage to human welfare is the standard that guides the care of those in need. Nonmaleficence differs from beneficence in that the former focuses on avoiding the actual infliction of harm versus the latter’s focus of preventing/removing harm or promoting good (Beauchamp & Childress, 2009). According to Lo (2005), avoiding harm is usually believed to be a greater responsibility than promoting well-being.

Not only is the obligation to avoid imposing harm of significance, but the duty to prevent risk of harm is also included beneath the umbrella of this standard (Beauchamp & Childress,
In the case being evaluated, this aspect of nonmaleficence is perceivable. A complication of the duty to warn encompasses the lack of the desire of certain relatives to know their genetic risk (Godard et al., 2006). One must consider the principle of nonmaleficence when information of this nature is being divulged.

Contained within the stipulations of this belief is the obligatory need to balance the risks of disclosure against the potential harm that the knowledge of the information may cause. The adverse psychological reactions possible when considering potential positive BRCA results have been demonstrated in the form of anxiety and depressive symptoms (Halbert, 2004). The Romero’s daughter is currently pregnant and just suffered the loss of her mother. Now may not be the most appropriate time for her to be informed of her mother’s BRCA results or effectively make decisions regarding her need for testing and subsequent possible implications of the evaluation. Mr. Romero might also find it more painless to discuss this matter with his daughter once he knows she is in a less vulnerable state.

As discussed previously, respect for autonomy can be viewed from an individual or family standpoint. The duty to warn potentially compromises respect for Mr. Romero’s autonomous decision making capability as well as the autonomy of the patient’s relatives who may not desire to know their risk of genetic disease. These aspects of the case must be of paramount concern when deciding how to handle situations such as these. If confidentiality is not breached, and the decision to disclose information is afforded to Mr. Romero, the principle of autonomy of the individual as well as the family will be upheld. If he resolves to inform his daughter and other potentially affected family members, he will have the option to do so in person or with the assistance of the genetic counselor by sending “Dear Family” letters (see
Empowering him with alternatives and means of disclosure will further promote his right of self determination.

Autonomy as a negative obligation can be regarded in the situation where the duty to warn is not advocated. The respect for autonomy from this aspect should not be controlled or constrained by others (Beauchamp & Childress, 2009). The rights of privacy, confidentiality, truthfulness, and informed consent are considered under this approach (Beauchamp & Childress, 2009). Confidentiality of Mrs. Romero’s BRCA test results will be kept private, providers will maintain the individual patient-provider relationship with Mr. Romero, and he will be provided with necessary information vital in making the decision to disclose or not disclose the BRCA results to his daughter and extended family.

Other Ways of Reasoning Through the Case

Guidelines set forth by professional organizations are adhered to by genetic counselors and providers within certain hospital organizations when issues of genetic information disclosure are at hand (L. Robinson, personal communication, September 19, 2008). ASCO (2003) and ASHG (1998) have submitted statements regarding the disclosure, confidentiality, and communication of familial risk.

Because genetic information is both individual and familial, conflict between the responsibility to maintain confidentiality of individuals and the duty to warn families occur (i.e. fidelity) (ASHG, 1998). ASCO (2003) supports the idea that providers should uphold and protect the confidentiality of their patient’s genetic information (i.e. veracity). They also recognize the ethical and legal implications created when efforts are made to notify family members at risk of genetic disease (ASCO, 2003). ASCO (2003) feels that the medical benefits of genetic testing in adult hereditary cancer syndromes are still being identified and characterized, “Therefore, we do
not believe that the federal requirements to justify a breach of confidentiality are currently met by genetic syndromes of cancer predisposition” (p. 2403). Both ASCO (2003) and ASHG (1998) assert that healthcare professionals should communicate with the patient, the person undergoing evaluation of the familial risk, or the family member in charge of the test results of the particular genetic disorder and the importance of sharing the results with his/her family members.

Legal Cases

Several legal cases concerning the duty to warn have transpired. A precedent setting duty to warn case occurred in the 1976 decision of Tarasoff v the Regents of the University of California. This case contended with a psychotherapist’s failure to warn the plaintiff’s deceased daughter of his patient’s verbalized intent to kill her (Suter, 2008). The court ruled in the favor of the plaintiff stating that the physician should breach patient confidentiality and take action to warn third parties if the patient creates an impending risk (Offitt, 2004).

Pate v Threlkel occurred in Florida in 1996 (Suter, 2008). In this case a patient was treated for medullary thyroid carcinoma and her daughter eventually developed the same disease. She alleged that the physician was liable for not warning her mother of the risk to her children and thus disease could have been prevented (Suter, 2008). The court ruled that the physician had a duty to warn the patient of the genetically transferable nature of the condition (Suter, 2008). This case established the existence of the duty to warn the patient of the genetic transmission of his/her disease but did not find it reasonable for the physician to seek out and warn members of the family (Suter, 2008).

Safer v Estate of Pack took place in New Jersey in 1996 (Suter, 2008). The plaintiff’s father had been previously diagnosed with malignant multiple polyposis of the colon and eventually died due to the illness. He was not warned of the genetic component of his disease.
His daughter (the plaintiff) subsequently developed this hereditary form of colon cancer. The court ruled that the physician had the duty to warn the patient and the immediate family of the heritability of the cancer (Suter, 2008). The court found that providers should take “reasonable steps” in order to notify the family, but the ramifications of the breach of patient-physician confidentiality were not addressed.

Thus far there has been an ill-defined legal precedent regarding a provider’s duty to warn relatives not under their direct care (ASCO, 2003). Currently the Texas legislature states that physicians owe no duties toward third parties (Kelso, 1999). The onus of responsibility of information dissemination to relatives falls on the patient while education and documentation of the hereditary nature of the illness is the duty of the provider.

Personal Decision

I support the autonomous right that individual patient’s or family members of deceased patients possess in deciding when, if, and who to inform of genetic test results. As a provider, I strongly believe that potentially affected relatives should be informed, giving them options to consider when making their future healthcare choices; however, I also stand by the right to privacy, confidentiality, and the foundations of the patient-provider relationship. The duty to warn could potentially compromise this basic premise.

The guidelines set forth by professional organizations such as ASCO uphold the provider’s duty to the patient. While this obligation is principle in fostering a trusting association, there are also actions such as thorough education and inclusion of all individuals of which the patient desires notified within the chart, which the provider must carry out in order to maximize informed decision making, minimize ambiguity of disclosure, and thus cultivate the most beneficial outcome for all those involved.
Summary

The duty to warn dilemma raises many issues that must be considered by providers caring for patients with heritable illnesses. The arguments for and against this subject are most assuredly valid and thus cause the large area of gray that is apparent within this matter. The particular case that was presented in this paper was thankfully resolved in a manner that hopefully provided the most optimal healthcare choices for all involved. Mr. Romero eventually made contact with his wife’s healthcare team and was open to discussing the results with providers with the inclusion of his daughter. He was given time to grieve and eventually came to the conclusion that most anticipated he would.

Providers are many times placed in arduous situations, and a tremendous ethical burden is produced when it comes to a patient’s unwillingness to share results with relatives at risk. Should the medical model of privacy within the HIPAA regulations be modified? Should set guidelines be addressed within professional societies (Patterson et al., 2005)? Research has shown that out of 206 geneticists surveyed, 69% felt they had a duty to warn patient’s relatives of their disease risk (Falk, Dugan, O’Riordan, Matthews, & Robin, 2003). Twenty-five percent who had been faced with the dilemma of patients refusing to disclose results to their family members considered informing at risk relatives without consent, but only four actually did so (Falk et al., 2003). The reasons for nondisclosure that were cited by 76% of medical geneticists were confidentiality, eventual resolution of the case, and legal liability (Falk et al., 2003). Given these facts, further evaluation of genetic information disclosure processes must take place in order to establish a more concrete set of standards within this DNA related dilemma. Until then, “…the informed consent process preceding testing presents the best opportunity to achieve the dual
goals of providing optimal care for hereditary cancer families, while avoiding unreasonable expansion of legal liability” (Patterson et al., 2005, p. 2103).
References


Appendix A

Complete Case Study
Case Study

A genetic counselor, Emily, working at a large teaching hospital in the South received a referral from the Women’s Health Nurse Practitioner, Sarah, on the medical oncology team for evaluation of a 42-year old Hispanic female currently admitted on the Oncology floor. The referral reports that the patient is in the end stages of breast cancer and is now requesting genetic testing because of a strong family history of cancer. Counseling and assessment is requested by the primary care team regarding possible BRCA gene evaluation of this patient prior to her being discharged home.

Emily arrives on the floor and reviews the patient’s chart prior to visiting with her. The patient, Mrs. Romero, is admitted for pain management (she has known focal areas of metastasis in her spine as evidenced by a bone scan indicating lytic lesions at T10-T11 completed 4 months ago after presenting with a humerus fracture with no known trauma/pathologic in nature) and generalized weakness. Salvage chemotherapy regimen of Zometa and Cisplatin was initiated at the time of recurrence with minimal response. Her last chemotherapy treatment was 6 weeks ago (has been delayed due to patient requesting a chemotherapy holiday). This was her second day in the hospital. She notices the admission note stating the patient is declining further chemotherapy and is now requesting hospice and palliative care. A recent history and physical assessment performed and documented by Sarah was present in the chart:

Physical Exam: Alert and oriented X 3, NKDA; No longer walking on her own because of weakness and back pain; Eating and drinking small to moderate amounts of food and water; Vital signs: Respiration 20, pulse 92, temp. 99.5, wt 110 lbs (150 at diagnosis), pulse oximeter 100% on 5L by nasal canula; Neck: Full ROM, no palpable LAD; Lungs: CTA bilat; CV: RRR, no murmurs; Right breast is surgically absent/no masses or overlying skin changes, no masses in
left breast/bilateral axilla; Abd soft/NT, bowel sounds present X 4 quadrants; Ext: moves all extremities although limited by back pain/pain to palpation over the region of the thoracic spine. Pain scale now at 4 on morphine PCA and fentanyl patch 50mcg.

**Tests:** (the following tests were performed on admission) chest x-ray pa/lat: lung fields clear, normal cardiac silhouette, normal mediastinum without pneumothorax or effusion, new T10-T11 “wedge-shaped” compression deformity of the vertebral bodies now present, not seen on previous x-ray done one month ago; UA negative; Labs: CBC-H/H 8/24 (declines transfusion), Plt-100, CMP-Na 137, K 3.8, HCO3 27 (25-30), Albumin 2.7, Ca 8.0, Bun 19, Creat 0.9, AST 34, ALT 30, Alk phos 120 (20-40); Hemocult negative.

**PMH:** negative; **PSH:** significant for BTL in 1992; **GYN history:** menarche age 13, on OCP’s from age 18-25, birth of first child by vaginal delivery at age 21 (1987) and second child by vaginal delivery at age 26 (1992), no complications; **FH:** mother diagnosed at the age of thirty-two with breast cancer and lived until the age of forty, mother’s sister (maternal aunt) died at the age of 35 from ovarian cancer, she never knew her maternal grandmother because she died at a young age of some type of cancer, but she was unsure what it was, father alive with hypertension, two brothers ages 45 (hypertension, hypercholesterolemia) and 46 (alive and well).

**HPI:** Initial diagnosis (II B Infiltrating Ductal Carcinoma) had been made when the patient presented at age thirty-nine (October 2005) with a three centimeter palpable mass in the upper outer quadrant of her right breast. A bilateral diagnostic mammogram revealed an irregular mass in the area of concern. Upon further evaluation with sonography, this area was again appreciated along with an abnormal appearing lymph node in the right axilla. An ultrasound guided core biopsy of the mass and lymph node was subsequently performed with a pathology diagnosis of a grade 3 infiltrating ductal carcinoma with findings in the axilla consistent with metastatic breast
cancer. The tumor profile showed an estrogen and progesterone negative tumor. A right modified radical mastectomy with right axillary lymph node dissection was performed. Five of twenty-three nodes were positive. Mrs. Romero received subsequent chemotherapy (Adriamycin, Cytoxan, and Taxol) and radiation.

Emily finds Mrs. Romero resting in her room. She welcomes Emily inside. Mrs. Romero speaks a moderate amount of English (she moved the United States from Mexico in 1992), but Emily is bilingual. Mrs. Romero tells her that her husband had to leave for work, her son is at school, and her daughter has not been able to make it to the hospital yet because she lives two hours away in East Texas. Her husband is not handling things well and does not agree with her decision to stop chemotherapy, but she feels like the treatments make her tired and sluggish. She desperately wants the opportunity to enjoy her time remaining with her family.

Mrs. Romero tells Emily of her family history of which Emily had noticed from the nurse practitioner’s note. She had initially declined genetic testing because her husband was against it, but given her terminal condition, she is now very concerned about the well-being of her daughter who just turned twenty-one and is pregnant with her first child. Her second child is a son who is sixteen. Emily extensively counsels Mrs. Romero regarding BRCA testing, and Emily completes a pedigree (see Appendix C). Mrs. Romero signs the consent form (see Appendix D), and the blood sample is obtained and sent to the lab. She lists her husband as the next of kin and point of contact. He is aware that she is going ahead with the test, and while still not in complete agreement that she should do so, he is supportive of her decision. Emily informed Sarah that the test usually takes two weeks to be completed and that she would contact her as well as Mrs. Romero with the results. Accurate contact information for Mrs. Romero was recorded in the chart. Two days later, once her pain had been controlled, Mrs. Romero was discharged home.
Two weeks to the day, Emily received the test results. Mrs. Romero was positive for the BRCA1 mutation. She notified Sarah and then made the phone call to Mrs. Romero. Much to Emily’s dismay, her husband informed her that his wife had passed away two days ago in her bed with her family at her side. Since Mrs. Romero had listed her husband as a contact and her next of kin, the results were able to be given to him. Emily had informed Mr. Romero about the test, why it had been done, the results, and their importance and their implications in his daughter’s future health. He was very angry and told Emily that breast cancer had caused his family great pain, and he would not burden his daughter with this information. She asked if he would like to come in with his daughter for a face to face meeting, and they could talk with her together. He adamantly refused, asked that she not call the house again right now, and hung up the phone.

**Plan:** Discussion of this issue with the oncology and genetic healthcare teams then ensued. Sarah had located a telephone number for Mrs. Romero’s daughter in the emergency contact section of her medical record; however, it was decided to give Mr. Romero some time to cope with his wife’s death. He might be more open to this information once some time has passed. Another attempt at contact will be made in a few weeks. Documentation of Emily’s initial discussion with Mr. Romero as well as the team’s plan for follow up contact was completed. Patient confidentiality is of great concern at this point as well as the health and well-being of the Romero’s daughter. Respect for Mr. Romero’s decision is also warranted.

**Result:** After 3 weeks had passed Sarah attempted another contact. Mr. Romero did not answer, but she left a message. Two weeks later, Mr. Romero made contact with Emily. He was ready to sit down and discuss the results. He was planning to bring his daughter in with him.
Appendix B

Dear Family Letter
Appendix C

Pedigree
Family Pedigree

- Breast Cancer
- Ovarian Cancer
- Deceased

Male
Female

Age 30
Car Accident

Age 55
A&W

Age 63
Diabetes

Age 65
HTN

Age 32
A&W

Age 42
A&W

Age 46
A&W

Age 45
HTN, High Cholesterol

Age 16
A&W

Age 21
A&W

Age 22
A&W

Age 42
A&W

Age 35
A&W

Age 45
A&W

Age 46
A&W

Age 45
HTN, High Cholesterol
Appendix D

Consent for Hereditary Cancer Testing
Appendix E

Case Consultation Worksheet A
## Case Consultation

### Worksheet A

**Step 1:** Personal Responses: I am torn between a patient’s/individual’s right to privacy and confidentiality and a provider’s continuous drive to help others. One always hopes that people make decisions that are in the best interest of others. I do not think that Mr. Romero has any negative intentions toward his daughter. He has just suffered a tremendous loss and probably wants to protect his daughter from any further pain. He is asserting his autonomy in making this decision. Maybe in time he will be more open to discussion and will find that informing his daughter could greatly impact her future health and choices she might make considering the positive BRCA results of her mother (if she chooses to be tested).

**Step 2:** Facts of the Case: (1) Nurse Practitioner (Sarah) consults the genetic counselor (Emily) to evaluate a patient (Mrs. Romero) for BRCA testing (2) Mrs. Romero is terminally ill, (3) She requests BRCA evaluation because she has a daughter, and she realized the impact on her future health and decisions, (4) Her husband, Mr. Romero, does not agree with the test being performed, but is now supporting her decision to have it done, (5) Mrs. Romero receives counseling regarding BRCA analysis from Emily, consent is signed, and blood is sent, (6) Two weeks later the test is resulted, (7) Emily and Sarah attempt to contact the patient, but she has since passed away, (8) Her next of kin is her husband, (9) He is informed of the results and potential implications to his daughter’s health, (10) He is angry, refuses to tell his daughter, and is unwilling to come in to the clinic alone or with her to discuss options, implications, etc.

**Step 3a:** Clinical/Psychosocial Issues Influencing Decision: Mr. Romero has just experienced a substantial loss at the passing of his wife. Emily and Sarah attempt to discuss the results, implications, and general pathophysiology of the disease and why the findings are important given Mrs. Romero’s family history and the fact that they have a daughter. He is not in the most optimal frame of mind for hearing, processing, and affectively acting on information at this time. Timing is an extremely important consideration in this case.

**Step 3b:** Initial Plan: Attempt to inform Mr. Romero of his wife’s BRCA results, implications, and options. Offer assistance in disclosing the information to the daughter and perform testing if she desires once she is counseled. Mr. Romero is currently adamant about not wanting to tell his daughter of her mother’s BRCA results because he feels breast cancer has caused his family enough pain. He also asks that the providers not call back at this time. I think it would be prudent and respectful to give him some time to digest his current situation given the recent loss of his wife. Attempting another contact in a couple of weeks might give Mr. Romero time to think about the initial discussion, begin coping with his wife’s death, and potentially be able to look at the different aspects of the situation in a more objective manner, giving thought to the effect on his daughter’s future.
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<th>Step 4: Policy &amp; Ethical Code Directive: The American Society of Human Genetics and the American Society of Clinical Oncology - obligations to relatives and family members are best fulfilled by informing the person undergoing the test of the importance of communicating results with family members if they are to be able to get testing themselves or make other changes necessary within their lives. Texas legislation.</th>
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<td>Step 5: Ethical Principles Analysis: Autonomy (as a positive and negative obligation). Beneficence and non-maleficence. Utilitarian and deontological approaches. For the Duty to Warn: Utilitarianism, beneficence, autonomy as a positive obligation; Against the Duty to Warn: Deontological approach, nonmaleficence, autonomy as a negative obligation</td>
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<td>Step 6: Possible Legal Issues: (1) Negligence/failure to warn the Romero’s daughter/family of Mrs. Romero’s BRCA results and future implications to their personal health, (2) Violation of patient confidentiality if the providers make the decision to locate and disclose Mrs. Romero’s BRCA test results after Mr. Romero voiced his desires about doing so; psychological harm to the Romero’s daughter if the providers do tell her, and she preferred not to know.</td>
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Appendix F

Case Consultation Worksheet B
Case Consultation

Worksheet B

Plan & Implementation Strategy: Attempt contact in a few weeks, giving Mr. Romero and his family time to deal with the loss of their wife/mother/etc. If/Once successful contact is made with Mr. Romero, validating his ill feelings toward this disease and actively listening to his concerns is very important in order to develop a trusting relationship. Education is a powerful tool, and once people are given accurate facts and information about a condition, anxiety and anger can potentially lessen. Individuals can then feel empowered to make informed choices regarding future critical healthcare decisions.

Write down how your plan:

Advances Clinical/Psychosocial Interests: By allowing Mr. Romero time to cope with his current situation, the eventual open and hopefully more trusting communication that has the potential to occur will ultimately be more beneficial for the Romero’s daughter. Discussion with Mr. Romero as well as with the daughter can occur in a way which would allow each member to take part in informed decisions concerning individual health choices. The need for more definite standards of disclosure are evident in this case. The nurse practitioner and other providers could become active in outlining guidelines, meeting with professional organizations, etc.

Adheres to agency policies and professional ethics codes: The guidelines and professional statements set forth by the American Society of Human Genetics and the American Society of Clinical Oncology are followed and upheld.

Minimizes harm and maximizes other ethical principles to the extent possible for the client and relevant others: Explored the principles of nonmaleficence and beneficence as well as autonomy. Guidelines proposed by ASCO and ASHG are reviewed. Texas legislature ruling.

Allows you to operate within the law: HIPAA laws and regulations regarding patient confidentiality are not violated. Case law is reviewed and considered:  
Tarasoff v the Regents of the University of California
Pate v Threlkel
Safer v the Estate of Pack