Genetic Testing and Duty to Warn vs. Patient Confidentiality

Rachel comes from a family with a history of breast cancer on her mother's side. Rachel's mother died of breast cancer when she was very young. Rachel has two sisters, Lisa and Kristin. Rachel has remained close to Lisa, but she no longer has a relationship with Kristin.

At a routine check-up, Rachel is told about the availability of genetic testing for identifying risk of breast cancer. Her doctor recommends that Rachel get tested for breast cancer risk (especially given her family history)

Rachel gets the genetic test and finds that she has a mutated BRCA1 gene, which puts her at risk for developing breast cancer. Rachel's doctor suggests that Rachel ask her sisters to be tested too (since her sisters may also test positive for the mutated BRCA1 gene). Rachel decides to tell Lisa to get tested. However, Rachel isn't close to her other sister Kristin, so Rachel decides not to tell Kristin to get tested.

The doctor feels confident that she can locate Kristin, but the doctor is worried about breaching patient confidentiality if the doctor goes against Rachel's wishes.

THE DILEMMA: As a doctor you have a duty to "do no harm". By not warning someone of a potential cancer risk, you are inflicting harm. On the other hand, Rachel is your patient, not your sister. Therefore your primary obligation is to treat Rachel. You cannot risk compromising Rachel's privacy by contacting Kristin.

If you were the doctor, what would you do?

Case Study: Gene Therapy for Enhancement Purposes

Dr. Anderson specializes in a particular type of gene therapy that targets Alzheimer's Disease (AD). Neural degeneration and synapse loss in the brain are characteristic of AD. Therefore, this gene therapy aims to protect neurons from degeneration and enhance the function of any neurons that are remaining. Dr. Anderson has two patients request her services. However, after an initial meeting with them, she is unsure whether she should treat them both.

Alexis is a 50 year-old woman who has a family history of AD and is already beginning to experience very mild symptoms. She tells Dr. Anderson that her mother was afflicted with AD. So, she knows first-hand the sadness and frustration the family of an AD patient has to experience. Alexis has a husband and three children and does not want to put them through the same difficult journey. Therefore, she is requesting the gene therapy to reverse the small-scale symptoms she already has and prevent the onset of the disease.

Kelly is a 21 year-old college student who is applying for medical school in the very near future. Her academic history is strong but not exceptional. For this reason, Kelly fears that she will not be accepted to the top medical schools. Kelly wants to attend medical school so she can help underserved populations and work in impoverished areas that lack good healthcare. She tells Dr. Anderson that she would like to receive the Alzheimer's gene therapy in hopes it will boost her memory and enhance neural function. Kelly believes a good score on the MCAT will strengthen her application and enable her to fulfill her dream of providing medical aid to the world's neediest people.

THE DILEMMA: Alexis is a patient already sick with Alzheimer's. Alexis wants the gene therapy to treat her disease. Kelly is a healthy patient who wants the gene therapy to enhance her performance as a student. Is it ethical for Dr. Anderson to give the gene therapy to Kelly?

Case Study: Reporting Incidental Findings

Barbara S., her husband, and her son participated in a study investigating the genetics of multiple sclerosis because her son is affected by the disease. While researchers compiled genetic information from many participants, Barbara and her family assumed their normal lives. Although she expected news about her son's multiple sclerosis, and perhaps even an answer to what side of his parentage the disease came from, Barbara was surprised when she received a phone call that some information had been uncovered that affected *her* specifically. Barbara went in to meet with the researchers and learned that, while doing their studies on MS, the doctors discovered an incidental connection between Barbara's genes and a greatly increased chance of Alzheimer's disease after 65.

Barbara is about to celebrate her 55th birthday, and is devastated by this news. There is not much medical action Barbara can take, and though she is a healthy and active adult, the news worries Barbara to no end.

THE DILEMMA: Barbara participated in a study to help her son. Barbara did not express any interest in learning about her own genetics. The researchers delivered grave news about a disease Barbara cannot prevent.

Was it ethical for the researchers to tell Barbara the news that her genes may lead to a disease that has no cure and no sure preventative measures?

Case Study: Huntington's Disease and Personal Autonomy

Scott, a 30 year-old male, has a family history of Huntington's disease. Huntington's disease causes neural degeneration, and eventually death. Affected individuals may experience mental and behavioral changes including paranoia, hallucinations and dementia, as well as physical symptoms such as difficulty walking and jerky movements. The disease has a late onset, which means symptoms don't show up until about 35-40 years of age. Most people live about 20 years after symptoms become apparent. Scott decides to be tested for the genetic mutation that causes Huntington's disease and finds out that he has it and will eventually get the disease.

Meanwhile, Scott's wife, Catherine, discovers she is pregnant. Together they decide that they should get genetic testing done to determine if their unborn child inherited the mutation and will also get Huntington's disease in adulthood. They will continue with the pregnancy regardless of the results.

Although there is no medical intervention possible to stop the disease, they feel strongly that they want to know about their child's future. At their next obstetric appointment, they inform their doctor of their wishes. The doctor hesitates because the parents are requesting information about a disease that will not affect their child until adulthood.

THE DILEMMA: The parents want to conduct genetic testing to see if their unborn child will have Huntington's disease. If the parents know their child will have Huntington's disease, they will be better able to support their child with appropriate emotional and psychological support. On the other hand, perhaps the child himself will NOT want to know if he will have Huntington's disease in the future. It should be up to the affected individual (the unborn child), and no one else, to decide.

If you were the doctor, would you conduct the genetic test?

Case Study: Preimplantation Genetic Diagnosis and "Modern Eugenics"

Phil and Jessica are young adults that want to start a family of their own. Phil was born with polydactyly, which caused him to have an extra finger at birth that was subsequently removed when he was a toddler. Because Phil has polydactyly he realizes that his children have a 50% chance of inheriting the condition. While polydactyly results in extra digits, it does not cause other side effects or health concerns. Usually the extra digits can be removed surgically.

Phil and Jessica pursue preimplantation genetic diagnosis (PGD) to ensure their children will not have polydactyly. The PGD procedure involves removing eggs from Jessica and sperm from Phil, combining them in a dish, allowing the resulting embryos to develop for a few days, genetically testing the embryos, and then implanting selected embryos into Jessica.

Six embryos result from the procedure.

After genetic screening the doctors determine that the embryos produced are all healthy, but all carry the mutation for polydactyly. Now Phil and Jessica are forced to make a decision. Do they implant one of their embryos knowing it has inherited polydactyly, or do they try again, and hope for an embryo without polydactyly? Additionally, it is unclear what will happen to the six embryos if Phil and Jessica decide not to implant them.

Is the PGD procedure ethical? Would you consider this to be a case of modern eugenics (attempting to improve the human race through genetics)?

Case Study: When Informed Consent is Unclear

A Native American tribe agreed to give blood samples to a local university research group in hopes that they could discover clues as to why the tribe suffered from high rates of diabetes.

In this particular tribe, blood has a significant spiritual importance. The tribe eventually learns that the samples they had provided were used for more than the intended diabetes study. When the researchers were unable to find any genetic link to diabetes in the tribe, they turned to other questions. In fact, the University researchers were using the samples to study the genetic underpinnings of mental illness and the tribe's geographical origins.

When the tribe found out how their samples were actually being used, they were outraged. Although members of the tribe had signed a consent allowing their samples to be used for "wider-ranging genetic studies", they maintain that they verbally agreed only to the diabetes research. In fact, the informed consent document was deliberately simpler than is typical because many in the tribe are barely literate.

The tribe asks the University for financial compensation. Their blood samples were used in ways they did not consent to. The University says it was an honest mistake, and technically they did nothing wrong based on the document the tribe signed.

THE DILEMMA: The tribe should have been told precisely how their samples would be used, so the consent obtained was not sufficient. On the other hand, the researchers were trying to understand a wide variety of genetic diseases, and they were doing their work in the best interest of the tribe. Should the University compensate the tribe?

Case Study: Preimplantation Genetic Diagnosis and "Designer Babies"

Martha and Robert, a young couple, are both 26 years old. Both of them are also extremely short, Robert at 5' 1" and Martha at 4' 7". They each earned their college degrees in fields that are not science related, although they do know that genetics plays a large role in determining height. They both know first-hand that being short has its disadvantages, especially when it comes to sports and being ridiculed by your peers at an early age. Robert especially was targeted by bullies at a young age, and he spent many years overcoming the anger he felt growing up. Martha and Robert are ready to start a family but do not want their children to experience what it is like to be extremely short.

They want to try pre-implantation genetic diagnosis (PGD) to ensure that their offspring have a high probability of being tall. To achieve this, they want to do *in vitro* fertilization and only embryos that are likely to be tall will be implanted.

Martha and Robert visit a fertility doctor and explain their intentions. The doctor tells them that PGD technology is usually only used to prevent life threatening diseases, not to ensure traits like height.

THE DILEMMA: PGD is intended for use in cases of disease-risk. Choosing embryos based on predicted height is unethical and is a troubling step towards "designer babies". On the other hand, PGD is safe, and there is no reason that a technology developed for one purpose can't be used for another. What would you do if you were the fertility doctor?

Case Study: Genetic Discrimination

Jim Sanders is a small business owner; he owns a bakery in the corner of town. The bakery currently has ten employees. Jim is interested in hiring one more employee. Two candidates apply for the position and both are highly qualified with similar work experience and skills. Even after Jim has interviewed both candidates, he isn't sure which one he will hire. However, he learns soon after, through a Facebook post, that Candidate #1 has a genetic risk for multiple sclerosis (MS). The candidate is healthy as of now, but it is *possible* that the disease will manifest in the future.

Jim decides that he is not willing to take the chance, and he ends up hiring Candidate #2, who is not genetically predisposed to any disease. .

If you were Jim, would you have hired Candidate #2 also?

Case Study: Ownership of Genetic Information

A patient named Doug Chambers went to his doctor in hopes of determining the cause of his illness. After a series of blood, bone marrow, and other tests, Chamber's physician, Dr. Richards, determined that Chambers had leukemia. In order to better Chamber's chances of survival, Dr. Richards suggested that he have a splenectomy to slow the progression of his disease. Following the advice of his doctor, Chambers decided to proceed with the operation and signed the necessary consent forms for the surgery. The consent form stated that the hospital could "dispose of any severed tissue or member by cremation", but at no time was it explained to Chambers either by his doctor or the hospital that his tissue could be utilized for research.

After the operation, Dr. Richards discovered that Chamber's spleen was producing a valuable protein sought after by pharmaceutical companies. He went on to sequence the genome of the cancer cells and discovered the DNA sequence encoding the protein. Dr. Richards marketed the sequence to several companies and obtained a patent for the DNA sequence and subsequent protein. Dr. Richards, and the company he partnered with, made a large amount of money based on the tissue, cells, and DNA procured from Chambers. Dr. Richards did not disclose any of his activities to Chambers.

Eventually Chambers discovered the full breadth of the situation with his tissue and filed a lawsuit to obtain a share of the profits.

THE DILEMMA: The tissue and DNA was Chambers' personal property and therefore Chambers is entitled to any derived profits. On the other hand, once the tissue leaves a body, it no longer belongs to its original owner. If you were the judge in the case, how would you decide?

Case Study: Using Fetal DNA for Gender Selection

Dexter and Susan are the parents of four healthy girls. During all four pregnancies, they had ultrasounds performed during the 20th week of pregnancy to determine the gender of the baby. Susan is pregnant again, and she and Dexter want to learn the gender of the newest addition to their family. They've heard about new technology that will determine the sex of the baby from a blood test based on tiny pieces of fetal DNA that are circulating in the maternal blood supply. The test can be performed as early as the seventh week of pregnancy. Dexter and Susan make an appointment with their obstetrician to ask about the test. Their OB recalls that the couple had been hoping for a boy during the last three pregnancies and is surprised that Susan is pregnant again. While asking about the gender test, Susan seems uncomfortable and looks at the floor a lot. Their doctor begins to suspect that they want the early gender test in order to terminate the pregnancy if it is another girl. When pressed, Dexter and Susan deny that this is their intention, but their doctor is not convinced.

THE DILEMMA: If you were the doctor, what would you do? You cannot be certain of the couple's plans, and it is not the role of a doctor to pass moral judgment. On the other hand, Dexter and Susan are within their rights to plan their family as they see fit.