Vignette #1

What Ought I Tell Him?

I feel very lucky to be alive and in good health. I have a genetic anomaly that is 95-99% lethal before birth. This anomaly is called Turner Syndrome. It is generally caused by the lack of one chromosome. This chromosome is one of the sex chromosomes. A normal human being has 46 chromosomes, 44 of which are called autosomes and two that are called sex chromosomes. Normally males are XY and females are XX, but I am what is called an XO or 45, XO. When people have fewer or more chromosomes than normal it is called an aneuploid. This has caused many problems in my past and is still causing problems.

I want to be very much like everyone else and in most ways I am. I have the needs and desires that everyone else has. My name is Linda and I am a senior in college. I am of average intelligence. Except for my math skills and space perception, I do okay in school. I have a boyfriend named Patrick.

When I was first born, the pediatrician noticed that my hands and feet were very puffy. I was a very short baby, had a very broad chest and my neck was webbed a little bit. As I began to grow older, my doctor recommended to my parents that before I start school, I have plastic surgery to change my appearance some. The doctors fixed my neck so that it didn't look so webbed and other parts of my physical appearance. One thing that they were not able to help me with was my shortness and slow development. I seem to have developed about 1/2 to 3/4 as fast as my friends. I am still fairly short at 140.8 cm. I never had a big growth spurt like my friends did. Another thing that the doctors told my parents was that I have a narrow aorta, and at sometime this needs to be corrected.

When my friends and I started entering puberty, it seemed that I was very far behind. When I entered the sixth grade, I went to a doctor and was told that I would never be able to produce children of my own because my ovaries will never develop to the point that they will be functional. I was also given estrogen shots, and I must continue to take medicine that will help replace the kind of chemicals that my ovaries would have produced if they had developed for the rest of my life. In appearance I look fairly normal for a short, overweight girl with a broad chest. I get along well with my friends, and they all accept who I am. In fact, most of them do not know that I have Turner Syndrome including Patrick.

In a Biology class, we studied human genetics, and I learned a lot about myself. I feel comfortable with the friends that I have grown up with that know about my condition, but I am not sure how my newer friends like Patrick would react if they knew. I was told that there is a cellular way to find out what has happened to my chromosomes called a Barr body test. This test shows that the second X most females have is mostly tightened into a dark staining spot (Barr body) on the nucleus. Most males do not have a Barr body. Also when sex cells go through division during meiosis mistakes can be made. If during separation of the X chromosomes the X chromosomes fail to separate then one sex cell will have no X chromosome and the other will have two. I learned that in 77% of the cases like mine, the one X that Turner females have came from their mothers. The failure of chromosomes to separate is called nondisjunction.

I have never had a menstrual cycle and will not be able to have children of my own. Which brings me to my present problem. Patrick is from a family that has 12 children, and he wants to have lots of children when he marries. I do not know whether to tell Patrick about my anomaly or if I do tell him, how much ought I tell him about my condition? What should I do?

Vignette #2

Access By Law Enforcement

Jerry Fox participated in a research study that sought to determine whether genetic factors contribute to Alzheimer's disease. Jerry has an older relative with this disease, and he wanted to participate in the study to help scientists discover more about its causes. A sample of Jerry's blood was drawn and sent to the lab at University Hospital where the study was conducted.

In order to keep track of each sample in the Alzheimer's study, the scientists utilized a method developed by law enforcement agencies to uniquely fingerprint the DNA, instead of using ordinary bar code numbers. The researchers in the Alzheimer's study also kept paper records that linked each DNA profile to the name of the person from whom that profile came - again, as a double-check to protect against mix-ups in the lab.

Several years after Jerry Fox provided his sample for the Alzheimer's study, a dozen university professors throughout the country are very seriously injured by letter bombs. The police have few concrete leads, but suspect the same perpetrator committed all or most of the crimes, based on the DNA profile of the biological evidence (saliva) found on the flaps of some of the envelopes. Following extensive investigation, law enforcement officials narrow the location from where the letter bombs were sent to a large metropolitan area – the same area where University Hospital is located. Police run the DNA profile of the suspect through all available criminal databases of DNA profiles, but they do not find a match. In the meantime, the letter bombs continue and another university professor is injured. As other leads dwindle and public pressure to arrest a suspect mounts, the police put out a court-approved warrant for "any person having a match" to the DNA profile of the suspect.

Shortly thereafter, law enforcement officials learn about the existence of the samples (>50,000) at University Hospital that were collected for the Alzheimer study and about the tracking system the researchers used, which makes it very easy for them to search for a match with the suspect. They request access to the DNA records from this study hoping that they will find one that matches the profile of the serial bomber. The researchers refuse to release their records, citing the promise of confidentiality they made to all participants at the time they collected their samples, but law enforcement officials obtain a subpoena. As a result, the researchers have no choice but to turn over their records to the police.

Jerry Fox is not a suspect in the case, and he is not the perpetrator of these crimes. Nevertheless, when he later learns (inadvertently) that the FBI has gotten access to his DNA identification profile, he is very upset.

* Is it fair to Jerry that the police were able to get his DNA identification profile from this research protocol without his knowledge, given that he was not a suspect with regard to these crimes (or with regard to any crimes)?
* If Jerry is truly innocent and a law-abiding person, what difference should it make to him whether or not the police have his DNA profile?
* If you were Jerry, would you have agreed to participate in the Alzheimer's genetic study if you had known his DNA identification profile would someday be given to the police for a crime in which he was not a suspect? Would the way that the scientists created unique identifiers for each sample influence your decision?
* Should law enforcement's interest in solving these serial crimes override Jerry's right to privacy?

In the past few years, a number of convicted criminals-including some on death row-have been exonerated of crimes they spent years in prison for, through the power of DNA identification evidence. Some people think this shows how imperfect our system of criminal justice is, and strengthens the arguments against capital punishment. Other people think DNA identification technology strengthens the arguments for capital punishment, because when DNA evidence found at a crime scene matches the DNA of a suspect, there can no longer be any doubt that the suspect is really guilty. What do you think?

Vignette #3

Interpreting Risk - The Impact Of Life Experiences

Background:

Approximately 5-10% of women with breast cancer have an inherited form of the disease. Scientists discovered several genes involved in breast cancer in the mid to late 1990's. Two such genes are BRCA1 and BRCA2. While every woman has a 12% lifetime risk of developing breast cancer, women with a mutation in one of those genes has significantly higher risk.

At this time there is no cure for breast cancer. While some studies have suggested that there are medications and surgeries that may reduce a woman's risk of getting breast cancer, the results are indecisive. It is clear, however, that early diagnosis and treatment greatly improves a woman's chance for survival. Women who have close family members with breast or ovarian cancer (especially early-onset) or who are known to have genetic changes in one of the breast cancer genes are encouraged to have earlier and more frequent screening mammograms and clinical breast exams, in addition to performing breast self-exams. In this way, it is hoped that any evidence of breast cancer will be found early and result in early treatment and long-term survival.

Vignette:

Lisa and Tonya are college roommates. They agree on almost everything but reacted very differently to an advertisement they saw in the local newspaper: A company was offering a simple blood test to screen for mutations in the BRCA1 and BRCA2 genes.

Tonya's 45-year-old mom was just recently diagnosed with breast cancer. Fortunately, the disease was diagnosed early and successfully treated by removing the small lump from her breast. Tonya's mom is now feeling great and has fully recovered from the surgery. Lisa, on the other hand, has had a very different experience with the disease.

When her mom was diagnosed with breast cancer at age 45, the cancer had already spread to other parts of her body. After two agonizing years of chemo and radiation therapy, Lisa's mom died. It was a month before Lisa's high-school graduation. Lisa desperately wants the genetic test. At this time, she feels convinced it will be just a matter of time before she will get breast cancer and die from it. She also says, that although she wants to have children some day, she won't if she is found to have one of the mutations in BRCA1 or BRCA2. She says she would rather adopt children than risk passing it on to her children.

Tonya thinks the test is unnecessary. She says it costs too much and has heard how hard it is to get health insurance if you don't 'pass' a genetic test. Tonya believes that having a genetic test is just not worth the risks involved. She decides she will do self-breast exams regularly, since that is how her mom initially found her lump. She will also start having regular mammograms sometime in her 20s. She feels that while she may one day get breast cancer, she will catch it early and do as well as her mother did.

Discussion points:

* Why do you suppose Tonya and Lisa have had very different reactions to the availability of the genetic test for breast cancer?
* How important do you think knowing this information is to Tonya? To Lisa?
* What scientific, medical and societal information should each consider before deciding whether or not to take the gene test?
* What are the potential benefits of the test?
* For Tonya? For Lisa?
* What risks are associated with genetic testing?
* Do you think there are risks to family members of Tonya or Lisa?
* Do you think the identification of a BRCA1 or BRCA2 mutation would have an impact on how people monitor themselves for breast cancer?
* How would the identification of a genetic mutation change your surveillance?
* If Tonya or Lisa decide to go ahead and have a genetic test, but no mutation is found, can they safely assume that there is NO chance that they will ever develop breast cancer?

Vignette #4

Genetic Screening & Discrimination - Relevance of historical experience

Background:

Sickle cell anemia is an autosomal recessive blood disease–a person has to have two copies of the sickle cell version of the beta-hemoglobin gene (HbS) to develop the disease. People who have inherited only one copy of the "sickle" variant do not suffer from the disease. However, they can pass the variant on to their children; they are referred to as carrying the sickle cell "trait". Sickle cell anemia affects people of all races but it affects people of African-American descent with the greatest frequency. During the 1970s, sickle cell screening programs were developed to identify individuals with the sickle cell trait in an effort to reduce the incidence of sickle cell disease. Due to inadequate education and counseling, however, there was confusion about the differences between sickle cell trait and sickle cell disease. This often resulted in stigmatization and unfair discrimination.

Vignette:

James Sr. was the class valedictorian at Central High School in 1975 and earned an aerospace degree from Georgia Institute of Technology. He became an accomplished pilot for Skyway Airlines, but his dream of flying ended when his employer restricted him from being a pilot even though he had outstanding scores on all his physical tests, because a blood test revealed the sickle cell trait.

James Jr. has his father's ambition to become a pilot. He has just graduated from college with an aerospace engineering degree and is looking for a job as a pilot. His father has read recent newspaper stories about how most employees are not protected from genetic discrimination in the workplace. He is very worried that his son might also have sickle cell trait and be denied a job. James Jr. thinks his dad is overreacting and does not want to discuss these issues with him. He dismisses his father's concerns and is ready to interview with a number of airlines and is willing to give a blood sample if necessary.

Discussion points:

* Are James, Sr.'s concerns justified?
* What lessons can be learned from the sickle cell screening programs of the 1970's?
* Is it reasonable for any employer to collect a blood sample from prospective employees and screen them for genetic variations?
* What additional concerns are there when a genetic test is available for diseases with a higher prevalence in minority populations?

Vignette #5

Genetic Discrimination: Inherited vs. Acquired Disease

John Jones has been working in a nuclear power plant for 15 years and has applied for a medical discharge because of the emergence of tumors in different parts of his body, all of which are attributed to a genetic condition called Von Hippel Lindau syndrome. In spite of these cancers, John has been an exemplary employee, having earned numerous awards throughout his career.

John learns that some workers who developed cancer were eligible for a substantial compensation from the employer. The company regulations governing qualification for the compensation package exempt diseases that are "hereditary and/or genetic" on the assumption that these diseases were incurred prior to employment with the company. Without this additional compensation, John's medical benefits do not cover a majority of his medical expenses and he is very concerned about how he will support his family.

John is denied the compensation package. However, John appealed his employer's decision and won a medical discharge when he was able to present evidence that environmental, as well as genetic, factors may have played a role in the development of his tumors.

Discussion points:

* Is it a fair for employers to differentiate between employees who have a condition with a known genetic basis and those with a medical condition not identified as genetic?
* Should employers hire individuals with a known medical condition? Should they be required to cover their medical benefits?
* Is it fair for employers to use genetic information under any circumstances?
* As more and more diseases are discovered to have a genetic component, will this mean that more and more employees will be denied medical benefits?
* Would your answers be different if John knew he was at risk for developing these cancers and sought employment in a potentially hazardous work environment?

Vignette #6

Genetic Determinism - Endurance Athletes

Background:

In 1999, scientists reported that a particular variant of the angiotens in-converting enzyme (ACE) gene was associated with superior physical endurance. The variant, known as the ACE insertion polymorphism, was found to be present in a higher proportion of elite endurance athletes, especially long distance runners, than in the general population.

Vignette:

Marathon University is offering full-tuition scholarships and a guaranteed spot on the varsity track team to high-school sophomores who "pass" a genetic test. If the parents sign a consent form, University doctors will screen them for the ACE insertion polymorphism.

Discussion points:

* Is it likely that a single gene determines physical endurance?
* Are other genes likely to be important?
* What other factors determine whether a child will grow up to be an elite endurance athlete?
* Should parents be allowed to use genetic information to make decisions that will limit their children's choices in adulthood?
* What else should MU do if it is concerned about the well being of the future student-athlete?

Vignette #7

Nature Vs. Nurture in the Criminal Justice System

Background:

The pace of research into genetic factors that may influence how we think and act has increased drastically in the last few years. For example, scientists are trying to determine how genetic factors make some people more susceptible to disorders like schizophrenia, depression and alcoholism. They also are exploring the contributions of genes to certain personality traits, like shyness and impulsiveness.

Although some forms of mental illness have a strong hereditary component, scientists currently believe that they, like the vast majority of human behaviors and traits, reflect a complex mix of genetics and the environment. It is unlikely that they will discover single genetic mutations that determine such characteristics as intelligence or that fully account for why some people become aggressive or violent.

Vignette:

It is 2010, and Joe Schmoe has been charged with assault. The evidence supporting his guilt is overwhelming and he pleads guilty. In preparation for his sentencing hearing, Joe's lawyer asks him to undergo a series of genetic tests to determine whether he carries any of four genetic mutations that scientists have associated with violent behavior. The tests, while controversial, show that Joe's DNA does, in fact, contain all four mutations. Based on these results, Joe's lawyer will argue that Joe should be sent to a psychiatric facility rather than to state prison. He claims that because Joe's genetic status predisposed him to this violent act, it would be unfair to sentence him as a criminal for behavior over which he had essentially no control.

Discussion Questions:

* If you were the judge at Joe's sentencing hearing, how, if at all, would the results of this controversial genetic test influence your decision?
* How would your decision be influenced if Joe had only 1 of the 4 mutations associated with violent behavior?
* Would your answer be the same or different if Joe was shown to suffer from a mental illness such as schizophrenia?
* What about family history? If a defendant's mother or father has a history of criminal offenses, should the judge consider that?
* If Joe gets sent to prison and tries to get released on parole fifteen years later, should the fact that he may have a genetic predisposition to violent behavior be used to keep him in prison, even if his behavior has been consistently good during his incarceration?
* In the future, should society screen all newborn babies for genetic mutations that could be linked to violent behavior? What if a medication became available to treat people with these mutations?