Case study #1:

Susan, a 40 year old woman, embarked on a family research project. During the course of the research, she discovered other family members, hitherto unknown to her who live abroad. While she knew that her mother had been diagnosed with breast cancer at age 43, she was unaware that her mother's five sisters and Susan's three cousins had also been diagnosed with breast cancer, some at relatively early ages. Given this history, she consulted her GP who advised her about the availability of genetic counseling and possible pre-symptomatic genetic testing. She informed the genetic counselor that she did not want her mother or other family members to know she was having genetic testing. The result indicated that Susan had inherited a predisposition to develop breast cancer that meant that she had a 40%-80% chance of developing breast cancer during her lifetime. Being a private person and because there was some discord within the family, Susan did not wish to discuss this result with other family members. She did, however, tell her 18 year old daughter who now has a 50% chance of having inherited the same faulty gene. She also asked that the genetic counselor inform her GP of the test result. When it was suggested that her three sisters and three brothers may also want to have information about their chance of having the mutated gene for predisposition to breast cancer, Susan indicated that she would not be communicating with them. Two years later, Susan's older sister, Barbara, who lived interstate, was diagnosed with advanced breast cancer. She was unaware of her risk for developing breast cancer.

Just after Susan had received her genetic test result, her younger sister, Mary, applied for life insurance. Susan and Mary are both patients of the local GP. The GP was asked to provide relevant medical information for the insurance policy. When asked about family history of cancer, Mary answered honestly that her mother had breast cancer but that there was no other family history. She was not aware of the research undertaken by Susan. She is also of course unaware of the result of Susan's genetic test.

Case study #2:

Huntington disease (HD) is a neurological degenerative disease that has an onset in most people between the ages of 30 and 50. There is no cure for this condition and it is progressive. Features include deterioration in movement, cognition and generalized functioning. Death usually results from respiratory illness. HD is an inherited condition. A child of an affected person has a 50% chance of inheriting the faulty gene that causes the condition. Genetic predictive testing is now available for persons over the age of 18 who have an affected parent or relative which will tell them in almost all cases whether they will develop the disease at some stage in their life. Worldwide, of those eligible for the test, only around 15% of people have taken up the option of testing. Mr. H. is a 25 year old man whose grandfather died some 10 years ago from Huntington disease. Mr. H's mother has therefore a 50% chance of developing HD. She decided to have the genetic test and has been shown to have the faulty gene. She will definitely develop HD at some time and Mr. H is now at 50% risk of developing HD. Mr H. is an air traffic controller. He loves his job and he feels he could perform his duties most adequately for many years, irrespective of whether he carries the faulty gene for HD or not. He does not wish to have the genetic test. His employer is unaware of his family history.

Case Study #3:

A General Practitioner (GP) has three male patients. Peter and Karl are aged eight years. Both are short for their age. Peter's short stature is due to a congenital deficiency in human growth hormone. His parents are both over 170 cm tall. Karl, however, is simply short because his parents are both under 165 cm in height. He has normal growth hormone levels. A pharmaceutical company has used genetic technology to produce human growth hormone. It is an expensive treatment but is being used by many doctors to treat growth problems. Both Peter and Karl's parents want the GP to prescribe it for their sons. A child's genetic height potential is determined by the genes inherited from both parents as well as by environmental factors such as diet. Therefore the treatment should enable Peter to reach a height similar to that of his parents and attain a normal stature. Karl's potential height is likely to be similar to his parents, that is, on the short side of normal. He will grow taller with the treatment, but will always be on the short end of the normal range for height. The GP's third patient is Tom, an athlete aged 22. Tom aspires to the 2004 Olympic Games rowing team. Tom has also asked to be treated with human growth hormone. He believes that with the extra muscle bulk and strength that the treatment will give him, he may be able to achieve his Olympic ambitions.

Case Study #4:

Ms. AP, now 30 years old, has a rare genetic condition, affecting only about 25 people in Australia. In the course of the diagnosis of the condition, when she was a child, samples of skin and bone were taken for testing and subsequently stored in the pathology laboratory, as is common practice for such tissues. Consent for the tests was obtained from Ms. AP's parents. At that time it was not envisaged that these stored samples could be a source of DNA. Research on the condition has been undertaken over the last five years and DNA has been extracted from the stored samples of skin and bone. Consent was not sought from Ms. AP, or her parents, to conduct research on the samples and on the extracted DNA. The research results in the discovery of an important gene and the gene is patented, providing the researcher and the Institution where the research was undertaken, with considerable funding for on-going work.

Case Study #1 Questions:

- Does the genetic counseling team, or a GP, have an obligation to inform Susan's relatives with whom they have had no previous contact that they may also be at increased risk, regardless of Susan's wishes?
- What is the situation if Susan's sisters and brothers were also the GP's patients?
- Should stronger efforts have been made to encourage Susan to share the information with her family to minimize the harm that followed?
- Regardless of legal issues, is the GP morally obliged to be honest to the insurance company about his knowledge of the family history?
- If the GP does reveal the information about Susan to the insurance company, should the company use this information in the assessment of risk for Mary in underwriting any policies issued to her?

Case Study #2 Questions:

- Do employers in industries involving public safety have the right to demand family health history information? In cases where genetic predictive testing is available for conditions that may impact on public safety, do employers have a right to predictive testing information about an individual whose current health status is excellent?
- Who actually 'owns' this information and who should decide who can access it?

Case Study #3 Questions:

- Which of these three people should be entitled to use the genetically engineered product?
- Will the use of genetically engineered products such as human growth hormone alter social perceptions and acceptability of characteristics like height?
- Could people of 150 cm be seen as having a disability?

Case Study #4 Questions:

- Who has ownership of the stored sample, and the DNA extracted from it?
- Given the rarity of the condition, does Ms. AP have the right to refuse consent for the research on her DNA? That is, how should the balance be struck between individual rights and public health in this case?
- In the case of a child, a person not legally competent to act, or a deceased person, whose consent (if any) should be required for research, ex. a guardian or next of kin?
- Should the contribution of individuals or families, who have enabled research by contributing DNA samples, be acknowledged or reimbursed perhaps by some form of royalty?