Ethical, Legal, and Social Issues in Genetics Research Case Studies

1. You are studying the gene mutations in the *NF1* gene in the hope of establishing genotype-phenotype correlations. One of the participants in the study contacts her physician to see if her mutation has been identified. She has just learned that she is pregnant and she is interested in having prenatal diagnosis to determine if the fetus will be affected by this autosomal dominant disorder. You checks your records and find that the mutation was indeed found and is a stop mutation that leads to premature termination of translation of the *NF1* gene product.

2. You are studying the impact of genes involved in the etiology of colon cancer. In one person with nonpolyposis colon cancer a missense mutation is found in one of the mismatch repair genes. The mutation has never been seen before, either in the general population or in affected individuals. The study participant has two siblings who are interested in being tested, and he therefore requests that the laboratory provide such testing, since no commercial laboratory is so far prepared to offer testing for this particular gene.

3. You are conducting a study in which participants are asked to provide family history of depression. Blood samples are also requested from relatives. One relative is approached and expresses anger at having his diagnosis revealed. He refuses to give a blood sample, and requests that information about him be removed from the study.

4. You are involved in a study of a native American tribe known to have a high incidence of type 2 diabetes. You gain their consent to do a genetic study intended to identify possible contributors to that risk. You learn that there is also a high frequency of alcoholism in the tribe and now wish to conduct a study seeking genetic association with this trait.

5. You are conducting a whole exome sequencing project aimed at identification of a gene associated with a rare phenotype in a young child. In the course of data analysis, you discover that the child is a carrier for cystic fibrosis and also is heterozygous for a stop mutation in the BRCA1 gene.