

1. From our knowledge gained from studies of a variety of genetic diseases, there are at least eleven ways of mutations that can abolish or reduce the function of gene products or subcellular location that lead to diseased phenotype. Please describe them, you may use specific examples to explain your points. (20%)
2. Comparative genome hybridization is one of the commonly used techniques in molecular pathology and genetic studies of human diseases. Please describe its principle and you may use example to illustrate your point. (15%)
3. One investigator is studying a human disease. From a preliminary cytogenetic study, it seems that it is a balanced translocation event. Also from Western blotting analysis of the diseased tissue, one kinase activity is abnormally upregulated. Please describe what is "balanced translocation" and provide two experiments to prove that it is indeed a balanced translocation involving two genes, and one of the gene encodes the kinase (15%).
4. Microsatellite is commonly used in cancer genetics and molecular genetics for mapping study. Please describe what is the microsatellite? Two phenomena are commonly seen in microsatellite study, loss of heterozygosity and microsatellite instability. Please explain them and describe their biological and clinical significance. (20 %)
5. One investigator studies a protein's function. From the deletion mapping analysis, he may have identified a nuclear translocation motif within the protein. From what we have known about the nuclear translocation sequence motifs, please describe the general characteristics of nuclear translocation motif and propose experiments to demonstrate that it indeed has a nuclear translocation function. These experiments should include proper control. (15%)
6. There are a number of techniques that have been used for detecting the DNA sequence mutation and/or polymorphisms. Please describe at least five different methods and their principles. (15%)