Molecular diagnostics: nucleic acid and protein based diagnostic methods

Genomics- the branch of molecular biology concerned with the structure, function, evolution, and mapping of genomics using methods such as next generation sequencing and fluorescent in situ hybridization.

Transcriptomics- the study of the transcriptome, the complete set of RNA transcripts that are produced by the genome, under specific circumstances or in a specific cell, using high-throughput methods, such as microarray analysis and polymerase chain reaction.

Personalized medicine

Personalized medicine is based on using an individual's genetics profile to make the best therapeutic choice by facilitating predictions about whether that person will benefit from a particular medicine or suffer serious side effects. Drugs are generally tested on a large [p[ulation of people and the average response is reported. Medical decision making based on empirical data relies on the law of averages, whereas personalized medicine recognizes the differences between individual patients.

Maxam-Gilbert's method of DNA sequencing

Maxam and Gilberts developed a method for sequencing single-stranded DNA. In the first step, ourines will react with dimethyl sulfate and pyrimidines will react with hydrazine in such a way as to break the glycosidic bond between the ribose sugar and the base, displacing the base. In the second step, piperidine will catalyze phosphodiester bond cleavage where the base has been displaced. The use of these elective reactions to DNA sequencing then involved creating a single-stranded DNA substrate carrying a radioactive label on the 5' end. The labelled substrate is then subjected to four separate cleavage reactions, each of which creates a population of labelled cleavage products ending in known nucleotides. The reactions are then loaded on high percentage polyacrylamide gels and the fragments resolved by gel electrophoresis. The gel is then transferred to a light-proof x-ray film cassette, an x-ray film is placed over the gel, and the cassette is placed in a freezer. Wherever a labelled fragment stopped on the gel the radioactive tag would expose the film due to particle decay-autoradiography. The dark autoradiographic bands on the film represent the 5' to 3' DNA sequence when read from bottom to top. Limitations of the maxam-gilbert's method include the use of large amounts of radioactive material and the neurotoxin hydrazine.

Sanger method of DNA sequencing

In sanger sequencing, the DNA to be sequenced serves as a template for DNA synthesis. A DNA primer is designed to be a starting point for DNA synthesis on the strand of DNA to be sequenced, four individual DNA synthesis reactions are performed. The four reactions include normal A, G, C and T deoxynucleotide triphosphates (dNTPs), and each contains a low level of one of four dideoxynucleotide triphosphates (ddNTPs); ddATP, ddGTP, ddCTP, and ddTTP. The four reactions can be named A, G, C and T according to which of the four ddNTPs was included. When a ddNTP is incorporated into a chain of nucleotides, synthesis terminates. This is because the ddNTP molecule lacks a 3' hydroxyl group, which is required to form a link with the next nucleotide in the chain. Since ddNTPs are randomly incorporated,