S4-5

Genomic Tools for Large-Scale Genetic Analysis

Mostafa RONAGHI
Stanford Genome Technology Center, Stanford University, USA

During the last two decades, genomics has emerged to a strong scientific field and has revolutionized medicine. More than 100 genomes have been sequenced and we are now entering a new era in biology. In order to expand the field and analyze more genomes, the analysis cost shall be reduced by at least three orders of magnitude. Here, three technologies will be presented including Pyrosequencing (Ronaghi et al. Science 1998), Molecular Inversion Probe Assay (Hardenbal et al. Nature Biotechnology 2003), and In vivo Mismatch Repair Detection (Faham et al. 2001). Pyrosequencing, a DNA sequencing technique, will be reviewed. Recent advances, applications and future development will be discussed. Molecular Inversion Probe Assay, allows highly multiplex genotyping. Multiplexing of 13,000 reactions in a single tube will be presented. In vivo Mismatch Repair Detection technology will be reviewed and the use of this technology for large scale, accurate SNP and mutation discoveries will be presented. We will also discuss our plan in cost reduction efforts using these technologies for genome-wide analysis.