Traditional and Novel Approaches to DNA Sequencing and Genotyping

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The number of available methods for analyzing DNA sequence has grown quickly in recent years, reflecting the dramatic increase in the use of molecular diagnostics in medicine. This lecture reviewed traditional and more recent approaches to the analysis of DNA sequence, including full gene sequencing (by dideoxy and Affymetrix GeneChip Resequencing technology) and individual base genotyping [RFLP, ASO (Nanogen, Affymetrix), ARMS, primer extension (Sequenom, TrimGen), DNA repair-based methods (TaqMan, Third Wave Invader), oligo-ligation (ABI SNPlex) and DNA synthesis (Pyrosequencing). Benefits and drawbacks of the various approaches were discussed and examples of their uses were presented. Practical issues concerning integrating these approaches, particularly direct sequencing, into a clinical laboratory were discussed. Helpful details about sequencing assay development and data analysis approaches were also presented.

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