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Massoud Houshmand

National Institute for Genetic Engineering and Biotechnology, Iran

Genetic diagnostic methods

The identify cation and characterization of the genetic basis of disease is often fundamental to diagnosis. Detection of cause of the disease in a blood or other tissues sample can lead to a diagnosis, possible prognosis, and prospective therapy treatments. Over the years, a variety of cytogenetic (Karyotyping) Molecular Cytogenetic (FISH-CGHarray-Cish) and molecular biology techniques (PCR-RFLP-ARMS-Dot Blot-Sequencing-MLPA-Western, Norden and southern blot-Real Time PCR, PGD) have been utilized in clinical diagnostic laboratories in the analysis of patient samples. The recent development of next-generation sequencing (NGS) techniques has revolutionized the field of clinical molecular diagnostics. Here, we review the development of molecular diagnostic approaches and some of the most commonly used assays prior to different kind of techniques in this area and mention advantage and disadvantage of each technique. PCR-based testing methodologies still currently predominate most clinical molecular diagnostic laboratories. The choice of detection method used in the analysis of gene mutations depends on a variety of factors and can range from laboratory to laboratory. Sample volume, the spectrum of mutations in a given gene of interest, and equipment investment required can all play a role in what type of assays a molecular diagnostic laboratory chooses to perform.

Biography

Massoud Houshmand has completed his PhD in Medical Molecular Genetic from Gothenburg University, Gothenburg, Sweden. He is the Head of the Genetic Diagnostic Laboratory, Faculty Member of National Institute for Genetic Engineering and Biotechnology and Responsible Director of Personalized Medicine journal. He has organized about 22 workshops and seminars and has published more than 220 papers and 17 books. He is the Winner of Best Iranian Researcher in Medical Genetic 2010, Winner of ISESCO prizes in Science & Technology 2014 and winner of Best Iranian Researcher 2015.

massoudh@nigeb.ac.ir

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