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HUMAN GENETICS AND GENETIC DISORDERS

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Putting together the pieces of the genome puzzle

Genetics is the basic science for biology and medicine. This truth is getting wider acceptance by the medical community as nowadays any disease of any system, monogenic to multifactorial and infectious disease to cancers, needs a molecular diagnosis. There is extensive use of cytogenetic and molecular genetic techniques in cancer diagnostics, prognostication and treatment. The importance of genetic testing is accepted by clinicians as has been well proved. There is a great need of better understanding of the genetic aspects of birth defects and principles of genetic techniques so that genetic testing can be appropriately used for the benefit of evaluation of fetal anomalies and providing genetic counseling and prenatal diagnosis to the families. Genomic techniques which can analyze the whole genome in one go have made genetic testing easier and the techniques of microarray and exome/genome sequencing are being applied in clinical situations more and more frequently. It has become practically the first-tier test for most of the genetic disorders by exome sequencing for *neurogenetic disorders*. The cost of this latest technological marvel is within the reach of many families and the costs are likely to come down further. There are some interesting issues about next generation sequencing in medicine. Also, there is importance of knowing the significance of each nucleotide in the genome, so that the exome sequencing data can be analyzed in a more meaningful manner and with more confidence. As more and more exomes are sequenced, more and more data about pathogenic and polymorphic sequence variations is getting accumulated and these comprehensive databases will ease the challenging task of genome/exome analysis to some extent. The whole objective of diagnosis is finding a path towards curative treatment. So, at this juncture, when the clinicians are gradually getting prepared for molecular medicine, the scientific community is putting together some more pieces of the extremely complex puzzle of human physiology and pathology using genomic techniques. Medicine will take some big leaps in the next decade or two.

Biography

Manjeet S Mehta is renowned Medical Geneticist from India par excellence. She has a rich experience of more than 3 decades in Human Genetics. She has worked with top hospitals in Bombay including Kokilaben Hospital, Jaslok Hospital, Lilavati Hospital, etc. She has set up the Genetic Department at the country's leading chain of referral labs. She has worked with Indian Council for Medical Research (ICMR), India's pioneer institute for biomedical research. After completing PhD, she has also gained advanced training at North York General Hospital, Toronto, and St. George's Hospital, London. She is in the Review Board for several journals. She is an invited speaker at several conferences worldwide including the World Congress of Perinatal Medicine to be held in Belgrade this year.

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