Putting Together the Pieces of the Genome Puzzle: Steps towards a Clear Picture

Editorial

DNA is the basic molecule and 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. Cancer genetics and prenatal diagnosis can be considered to be instrumental in popularizing genetics amongst clinicians. The article on cancer genetics in this issue illustrates the extensive use of cytogenetic and molecular genetic techniques in cancer diagnostics, prognostication and treatment. The fetal medicine conference held in Kolkota on 9th to 11th September 2016 also proved that the importance of genetic testing is accepted by clinicians. The Fetal medicine specialty takes care of birth defects and genetic disorders where genetic diagnosis is of prime importance. The scientific program of the *FetalMed2016* had given a stress on genetic diagnosis and all fetal medicine specialists appeared enthusiastic towards the 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 as illustrated in the article on exome sequencing for neurogenetic disorders in this article. The cost of this latest technological marvel is within the reach of many families and the costs are likely to come down further. The GenExpress in this issue covers some interesting issues about next generation sequencing in medicine. The article about the ExAC database in GenExpress highlights the 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 a greater degree of 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 other article on the use of next generation sequencing for the diagnosis of genetic metabolic disorders is also a guiding star for all clinicians especially neonatologists struggling to reach an etiological diagnosis. As is shown in the article by Tarailo- Graovac et al., the diagnosis of a disorder where a therapeutic intervention is possible is a golden opportunity of translating research to patient care which is what everyone dreams of. The use of next generation sequencing is also of tremendous use in identifying variant clinical presentations, mild forms or late onset variants of genetic metabolic disorders as clinical suspicion in such clinical situations is very, very difficult.

The whole objective of diagnosis is finding a path towards curative treatment. Understanding the pathogenesis is the first step towards research for treatment. The other form of treatment is correction of the basic genetic defect by gene therapy which is still elusive to genetic scientists. Now CRISPR technology is showing a great hope for gene therapy. So at this juncture of the twenty first century, 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. Though a complete understanding of basic biology appears too complex, additional information will show a clearer picture of some parts of the puzzle and medicine will definitely take some big leaps in the next decade or two. Genetic Clinics continues to bring the brilliant and clinically applicable advancements to the readers in simple language. Enjoy the marvels of genetics!

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