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NEWSBYTES

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About Xcelris

Xcelris Labs Limited is a life sciences based innovative research organization delivering solutions across domains such as Agriculture, Environmental Genetics, Animal science, Nutrition and Human health. We operate through two key divisions i.e Genomics and Medical Genetics. At Xcelris Genomics Division, we partner with more than 1000 research institutions across India and world to provide key services like Sanger Sequencing, Next Generation Sequencing, Molecular Biology kits and Oligo synthesis. In a span of 10 years, we have completed 15,000+ sequencing projects and 1000+ Next Generation Sequencing projects. Through our Xcelris Medical Genetics (XMDx) division we offer genetic testing service based on Sequencing for cancer diagnosis and therapy selection, infectious diseases and inherited genetic disorders in humans.

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EMERGING TECHNOLOGIES IN THE MANAGEMENT OF COMPLEX HEREDITARY DISEASES

by Dr. Prashanth Bagali

Hereditary diseases are complex, heterogeneous and caused single gene or polygenes, multiple alleles, quantitative trait loci in the genome, which are heritable (transmitted from parents to offsprings, germline or congenital cells) or non heritable (somatic cells). You will find number of books to understand inheritance patterns (autosomal or sex linked, dominant or recessive). Medical research is focused on investigation of susceptibility and onset of hereditary diseases. Most of the genetic disorders are quite rare and affect one person in several thousands or millions (rare genetic diseases). Therefore, it is a great challenge to humanity to understand genetic basis of hereditary diseases for early diagnosis and timely treatment.

In the past, conventional PCR (polymerase chain reaction) and quantitative PCR (qPCR) technologies were used in diagnostics of monogenic diseases. PCR method was used in the assessment of BCR-ABL in chronic myeloid leukaemia, SNP analysis, mitochondrial DNA studies, DNA methylation studies, microsatellite instability, chimerism studies following haemopoeitic progenitor cell transplantation, and rapid detection of fetal aneuploidy following amniocentesis. Microarray and it's related technologies (DNA arrays, Tissues arrays, protein arrays, SNP arrays and others) are extensively used in medical research for analysis of cancer tissue, analysis of multiple genes implicated in the metabolism of or response to a specific medication.

Medical genomics and clinicogenomics are powered by nucleotide sequencing (chain termination sequencing or targeted sequencing or Sanger sequencing, named after Dr.Frederick Sanger) have revolutionized medical practice. This technology is being used in our laboratory to sequence short pieces of DNA in the identification of multiple allelic mutations and SNPs of monogenic genetic disorders. Sanger sequencing is used in the identification of disease causing genes, namely, *CHEK2, TP53, EGFR, ALK, BRAF, HRAS, KRAS, MYBPC3, SLCO1B1, CYP2C9 and CYP2C19*. Sanger sequencing is used in the pharmacogenomics, such as selection of drugs for cancer treatment. Sanger sequencing is cost effective in the detection of hereditary diseases, namely, beta thalassemia, sickle cell anemia, lactose intolerance, cystic fibrosis and biotinidase deficiency mutations.

Recent developments in the selection and isolation of targeted sequences of interest from a genomic DNA sample, have given raise to the high-throughput method of detection of thousands of genes in a single experiment. Massively parallel DNA-sequencing technologies are referred as next-generation sequencing (NGS or next-gen sequencing).

NGS technologies have revolutionized our understanding in medical genetics, accelerating health-improvement projects, and ushering to a fully understood personalized medicine in near future. With next-generation sequencing, it is now feasible to sequence whole genome and whole exome (complete set of all exons) of an individual to identify possible disease-causing mutations in the genome. Identified genetic variants may or may not associated with the clinical condition of interest. Therefore, novel clinical bioinformatics tools are required for hereditary disease management including counseling, prevention and treatment / therapies and drug responses. Xcelris Labs is champion in the clinical and diagnostic applications of NGS technologies in a healthcare settings, especially for early diagnosis of hereditary diseases in adults, children and fetus caused by numerous genes.

Next article

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Genomics Industry Trends

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