MedGenome Offers State-of-the-Art Genomics Services to Academic Research Institutions

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Enabling Deeper Insights Into the Genetics of Human Diseases

Genomics has been instrumental in basic disease mechanism research, and has helped to identify the causes of several diseases. Although single gene sequencing and cloning techniques have resulted in the identification of the genetic causes for several diseases, a number of rare as well as familial diseases cannot be fully understood with these techniques alone. Recent developments in Next Generation Sequencing (NGS) technologies are rapidly changing the scope of genetics, genomics and medicine. The application of NGS platforms to selected regions of the human genome would be of great interest to the research community.

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The exome (the protein-coding region of the human genome) represents less than 2% of the genetic code, but contains ~85% of known disease-related variants. Exome sequencing can efficiently identify coding variants across a wide range of applications, including population genetics, genetic disease, and cancer studies.

Exome sequencing utilizes parallel nucleotide sequencing to identify all of the protein-coding variants in the genome. By limiting the sequencing to the protein-coding sequences, only about 5% of the human genome is sequenced. Aided by the growing public databases of genetic variants, exome sequencing enables identification of genetic mutations and risk factors in samples that were deemed insufficiently informative for previous genetic studies. It is a cost-effective approach when whole-genome sequencing is not necessary. Sequencing only the coding regions of the genome allows pointed focus on the genes most likely to affect phenotype, and facilitates quicker turnaround time and lower price.

Targeted exome sequencing has helped identify a variety of rare diseases in a number of patients who were referred to MedGenome. With its high-throughput state-of-the-art laboratory and latest NGS platforms (Illumina HiSeq 4000 & 2500), MedGenome provides end-to-end NGS services for all DNA and RNA applications to researchers in a time-bound and cost-effective manner, without compromising the quality of project deliverables. It is also an experienced microarray service provider, leveraging Illumina, Affymetrix and Fluidigm microarray technologies for myriad applications, including services for whole genome and custom genotyping, gene expression, methylation, exome, cytogenetics and pharmacogenomics. Processing of samples on best-in-class instruments combined with the informed technical and bioinformatics expertise guarantees quality outcome to sequencing and array projects.