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Commentary

A brief note on DNA sequencing in humans and its applications

Mikhail Cameron*

Department of Biochemistry and Molecular Biology, Washington University Saint Louis, USA

***Corresponding Author's** Email: mikhail46ca@wustl.edu

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DESCRIPTION

The method of determining the nucleotide sequence of DNA is known as DNA sequencing (deoxyribonucleic acid). A gene's or genome's nucleotide sequence is the most basic level of information. It's the blueprint for creating an organism, and without it, no knowledge of genetic function or evolution would be complete.

DNA sequencing uses electrophoresis separation and autoradiography, which is a sensitive detection method but this is a time and labour-intensive method. As a result, a real-time detection approach should be used instead, in which a computer analyses measured data automatically. They used four types of fluorophores, corresponding to four DNA fragment families terminating in A, C, G, or T.

APPLICATION

New-born and paediatric disease

Many individuals with serious, often fatal disorders with a genetic foundation are seen in newborn critical care units and children's hospitals. Some of these are well-known genetic illnesses that have been accurately identified and verified by clinical genetic testing. However, a significant proportion match established diseases but affect people who have received negative genetic test findings. Exome sequencing is being used in a number of experimental initiatives, including the National Institutes of Health's Undiagnosed Disease Network. Exome sequencing finds a harmful mutation in 25%-30% of patients on average.

Whole-genome sequencing is the logical next step, as it can examine poorly captured exonic areas and find structural variations. Whole genome sequencing may now be the natural initial step using the X Ten system. It offers a shorter turnaround time, doesn't require hybridization, and can detect everything from single nucleotide variations to massive deletions. Sequencing should be done on the patient, both parents, and a sibling.

Drug trials and pharmacogenomics

Personalized medicine, which tailors illness treatments to an individual's genetic composition, is one of the great promises of genomic research. To get there, researchers will have to look at the genetic diversity that underpins illness prediction and drug response. Many pharmacogenomics initiatives are now underway, with the majority relying on SNP arrays or targeted sequencing. Whole genome sequencing would strengthen these attempts by capturing a far greater range of variation that might influence the response.WGS might potentially be employed as a front-end tool in clinical trials to stratify individuals depending on their likelihood of responding to the medicine under study.

Regulatory variation and eQTLs

One of the numerous benefits of the International HapMap Project was the identification of genetic diversity in fibroblast cell lines, which could then be obtained from Coriell for further research. Researchers could analyse gene expression first with microarrays, then with RNA-seq and then connect that with genetic variation after they had all of the SNP genotypes. Thousands of expression quantitative trait loci (eQTLs) were discovered as a result of these investigations, as well as new insights into how genetic variation regulates transcription.

Large cohorts with extensive phenotyping

Tests from enormous, well-phenotyped partners have consistently been popular for hereditary investigations. A significant number of them have been overviewed with SNP exhibits and all the more as of late exome sequencing. Over the long run, numerous companions become both in the quantity of members and the measure of aggregate information gathered. Enormous scope, longitudinal investigations of complicated attributes are fundamental for pinpointing the hidden hereditary qualities.

Rare tumour types

Huge scope malignant growth sequencing endeavors, for example, TCGA and ICGC have recorded physical transformations in an assortment of normal disease types. The greater part of these tasks had both exome sequencing and an entire genome sequencing part, yet because of the expense, most of cases got exome sequencing. All things being equal, these investigations have been inconceivably helpful for distinguishing repetitively changed qualities and pathways