

Medical Genomics

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Whole genome sequencing is the process of determining the sequence of complete DNA at a single time of an organism's genome. This sequencing all of an organism's chromosomal DNA as well as DNA contained in the mitochondria and, for plants DNA contained in the chloroplast.

Sequencing of genome also called as WGS, complete genome sequencing, full genome sequencing, or entire genome sequencing.

Whole genome sequencing has been used largely as a research tool, but it was introduced in 2014 to clinics. In future personalized medicine, whole genome sequence data is also an important tool to for therapeutic intervention to guide. The tool of gene sequencing at SNP level is also used to pinpoint functional variants from association studies and improves the knowledge which is available for researchers who are interested in the evolutionary biology, and hence it lays the foundation for predicting the disease, susceptibility and drug response.

Whole genome sequencing should not be confused with the DNA profiling, which will determine the likelihood of that genetic material from a particular individual or group, which does not contain any additional information on origin, genetic relationships, or susceptibility to specific diseases. Whole genome sequencing should also not be confused with the methods of sequence specific subsets of the genome - such methods include whole exome sequencing (1-2% of the genome) or SNP genotyping (<0.1% of the genome). Between 4% to 9% of the human genome, mostly satellite DNA, had not been sequenced.

A biological sample containing DNA—even a very small amount of DNA or ancient DNA—can provide the genetic material necessary for full genome sequencing. Such samples may include saliva, epithelial cells, bone marrow, hair (hair contains a hair follicle), seeds, plant

leaves, or anything else that has DNA-containing cells.

The genome sequence of a single cell selected from a mixed population of cells can be determined using techniques of single cell genome sequencing. Single cell genome sequencing has important advantages in environmental microbiology in cases where a single cell of a particular microorganism species can be isolated from a mixed population by microscopy on the basis of its morphological and other distinguishing characteristics.

Single cell genome sequencing is being tested as a method of pre-implantation of genetic diagnosis, wherein a cell from the embryo created by in vitro fertilization is taken and analysed before embryo transfer into the uterus. After implantation, cell-free foetal DNA can be taken by simple venipuncture from the mother and used for whole genome sequencing of the foetus. In such cases the normally necessary steps of isolation and growth of the organism in culture may be omitted, thus allowing the sequencing of a much greater spectrum of organism genomes.

Currently using techniques are capillary sequencing to sequence full human genome successfully which was the first approach in sequencing the full human genome. This method is too expensive and will take a long for commercial purposes. Since 2005, capillary sequencing has been displaced by high-throughput (next-generation) sequencing technologies such as Illumina SMRT sequencing, dye sequencing, and pyrosequencing. All of these technologies continue to employ the basic shotgun strategy, namely, parallelization and template generation via genome fragmentation.

Other technologies are emerging, including nanopore technology. Though nanopore sequencing technology is still being refined, its portability and potential capability of generating long reads are of relevance to whole-genome sequencing applications.

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