

# **Bioinformatics and computational tools for next generation sequencing analysis in clinical genetics**

## **Abstract**

Clinical genetics play an important role at discovering ambiguous and rare disease and also search for the best option of treatment for patients .And because of huge amount of sequencing and reference genome , Next generation sequencing make it easy to analysis hundreds of genes and chromosomes at lower price and perfect time(by using PCR technic. This paper aims to show how next generation sequencing do that operation and attached it to bioinformatics . it will focus on Illumina and Ion Torrent and show how they work by using some algorithms and some data analysis . although NGS has provided too much solves to many problems , further improvements in bioinformatic algorithms are still required to deal with complex and genetically heterogeneous disorders.

## **Introduction**

In medical practice to make it easy to diagnose a disease, researchers use genetics to help them discover it quickly. genetics takes a best way to find a solution or treatment to a disease. It's flexibility comes from the ability to deal with genome at different levels and forms from chromosomal to singlebase alternation.

Paul Berg, Frederick Sanger and Walter Gilbert made possible several progresses in DNA sequencing field making technology named sanger sequencing . Through sanger technology u can generate a huge amount of sequencing in one machine run in a fast and cost effective way . and they considered it as a Next generation sequencing .

In market sanger sequencing profit has reached to billions dollars by 2025 of course because of it's advantages from doing tasks fast in addition to costs little money compared to other machines but still there are companies and labs use illumine , Ion \_Torrent sequencing , Pac Bio and Exford Nano Pore.

Using (PCR) plays an integral role in targeted NGS . it produce multiple of targeted regions simultaneously. Next generation sequencing can produce massive quantities of molecules (PCR) but first generation sequencing produce single sequencing without clonal amplifications.