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. 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 an 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

Genetics is very important in medical practice its allow to diagnose the disease more easier . it takes a best way to find a solution or treatment toa disease.. It's flexibility comes from the ability to deal with genome at different levels from chromosomal to single-base alternation.

Paul Berg, Frederick Sanger and Walter Gilbert made possible several progresses in DNA sequencing field making technology named sanger sequencing . Throug 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 but still there are companies and labs used illumine , Ion _Torrent sequencing , Pac Bio and Exford Nano Pore.

Next generation sequencing can produce massive quantities of molecules (PCR) but first generation sequencing produce single sequencing without clonal amplifications .