# Discussion:

Genetics is currently extremely important in medical practise because it allows for a conclusive diagnosis of a wide range of clinically heterogeneous diseases. As a result, it allows for a more precise disease prognosis and offers assistance in selecting the best available treatment choices for affected patients. Most of its current promise stems from its ability to probe human genom at various stages, from chromosomal to single-base changes.

so,We will discuss a few topics that show us the importance of addressing genetics issues and the interest in developing the next generation sequencing technology that has greatly contributed to the human genome project that exists in our time.

**NGS library :** A library is a series of DNA/RNA fragments that represents the entire genome/transcriptome or a target region in next-generation sequencing. Each NGS platform has its own quirks, but in general, the preparation of an NGS library begins with fragmentation of the starting content, followed by sequencing Fragments are linked to adaptors to enable enrichment of those fragments. The fragmentation of nucleic acid is the first step in most NGS workflows to prepare libraries Physical or enzymatic methods may be used to fragment materials.Adaptors are bound to the fragments of the starting DNA after it has been fragmented. The adaptors are used to give random sequences known beginnings and endings, enabling the sequencing process to proceed.A new technique was developed that combines fragmentation and adaptor ligation in a single step, making the process easier, quicker, and with less sample input Tagmentation is a transposon-based technology that is used in this process.

The fragments are chosen based on the desired library size after nucleic acid fragmentation.

**NGSPlatforms:** Illumina and Ion Torrent are two of the most well-known second-generation sequencing firms Illumina is a well-known American company that sells a variety of integrated systems for the study of genetic variation and

biological function in a variety of biological systems ranging from agriculture to medicine The Illumina sequencing method is based on the sequencing-by-synthesis (SBS) concept.

**NGS Bioinformatics and analysis:** To handle, analyze, and interpret the massive amount of NGS data, computational and bioinformatics skills are needed. As a result, significant progress in NGS (bio)informatics is being made, which can only be accelerated by growing computational capabilities (hardware) and algorithms and applications (software) to help with all of the necessary steps: from raw data collection to in-depth data analysis and variant interpretation in a clinical setting NGS bioinformatics is usually divided into three categories: principal, secondary, and tertiary research Regardless of the NGS platform, the end objective of each review is essentially the same; nevertheless, each platform has its own unique characteristics.

**PrimaryAnalysis**,QualityControl(ReadFilteringand Trimming),

**SecoundaryAnalysis**(SequenceAlignment,Post-AlignmentProcessing,Variant Calling),

**TertiaryAnalysis**(VariantAnnotation,VariantFiltering,Prioritizationand Visualization

**NGSPitfall:** It is undeniable that NGS provided a plethora of benefits and solutions for medicine as well as other fields such as agriculture, all of which helped to improve quality and productivity. It has, however, brought with it new challenges. The first issue is with the costs of sequencing. Although it is true that the total costs of When compared to the gene-by-gene sequence of Sanger sequencing, NGS is more expensive and not yet available to all laboratories. It has high upfront costs for the sequencing machine, which can range from thousands to hundreds of thousands of euros depending on the model depending on the computer, as well as consumables and reagents Expenses of experimental design It's also essential to consider sample selection and sequencing library preparation. Furthermore, costs associated with the construction of sequencing pipelines and bioinformatical methods are often underestimated The costs of data management, informatics equipment, and downstream data analysis, as well as the costs of improving such pipelines and performing downstream sequence analysis, are not factored in overall costs of NGS.