Bioinformatics in Biomedical Analysis and its impact on Personalized Medicine [

2024

text (article)

Analítica

Biomedical analysis represents a crucial process for understanding and diagnosing various diseases, including those of genetic origin. In this context, bioinformatics plays a fundamental role in facilitating the interpretation of genomic data and providing essential information that can guide diagnosis and clinical decisions. Bioinformatics is an area that promotes the use of computers to address questions of biological nature, tackling the massive analysis of available biological data. On the other hand, one of the most revolutionary techniques in the clinical field is Next-Generation Sequencing (NGS), which allows for the rapid and precise reading of the DNA of any patient suspected of having a genetic condition. With NGS, it is possible to identify mutations, genomic variants, and other changes that may be related to medical conditions. Other diagnostic techniques, such as Multiplex Ligation-dependent Probe Amplification (MLPA) and DNA arrays, also excel in detecting genetic changes. MLPA detects changes in the number of copies of DNA segments (known as CNVs), while arrays analyze multiple markers simultaneously, identifying CNVs and point mutations. When used together, these techniques generate a significant amount of information that can be crucial for approaching and understanding a pathology. Thus, bioinformatics is present in all stages of biomedical analysis. It begins with the alignment of sequencing reads against the Human Reference Genome using various software tools like BWA or Bowtie. Reads undergo variant filtering, which identifies discrepancies between the patient's genome and the reference genome. In this phase, tools like GATK and SAMtools are essential. Beyond all intermediate steps, variant interpretation is the decisive stage of the entire process. In this phase, factors such as heritability, penetrance, clinical impact, and population frequency are evaluated, determining whether variants are pathogenic, benign, or of uncertain significance, ac
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