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# Clinicogenomics: An Overview

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## Introduction

Clinicogenomics, also known as clinical genomics, is the investigation of clinical outcomes using genomic data. Clinical data are affected by genomic factors. Clinicogenomics is the use of a patient's entire genome to diagnose diseases or adjust medications specifically for that patient. Whole-genome sequencing can detect more mutations and structural abnormalities than targeted gene sequencing. Furthermore, targeted gene testing can only test for the diseases for which the doctor screens, whereas whole genome testing screens for all diseases with known markers at the same time.

## Description

Clinicogenomics, like pharmacogenomics and oncogenomics, is currently used in personalised medicine. A physician can create medical plans based on an individual patient's genome rather than generic plans for all patients with the same diagnosis by studying the entire genome. For example, in a study of renal tumours that were previously only diagnosed through morphological anomalies, researchers were able to identify the mutations that cause that cancer type by studying the genomes of many patients with that cancer type. Furthermore, researchers can pinpoint the medications and treatments that are most effective against specific cancer-causing mutations, which can then be used to treat future patients.

Clinicogenomics is the use of genome sequence data in diagnostic, therapeutic, and public health applications. The high-throughput DNA sequencing of genomes and metagenomes is central to this field. A recent symposium presented at the 115$^{th}$ general meeting of the American Society for Microbiology in New Orleans, LA, focused on the role of clinicogenomics in infectious disease diagnostics and public health microbiology. The following is a summary of the most important and promising aspects of each symposium presentation.

High-throughput DNA sequencing, also known as Next-generation Sequencing (NGS), technologies that allow the genomic content of entire microbial communities (bacterial, viral, and eukaryotic organisms) to be described, is driving the explosion of microbiome research. Although much of this research focuses on the structure of "commensal" communities, the methodology can also be used to identify pathogens in clinical samples. The key concept in using NGS methodology is that microbe detection is independent of culture and is not limited to PCR targets. Rather, it is the process of generating large-scale sequence data sets that adequately sample a specimen for microbial content, followed by the application of computational methods to resolve the sequences into individual species, genes, pathways, or other features [1-3].

Most microbiome analyses have concentrated on describing bacterial content, which is typically accomplished by sequencing the 16S rRNA gene. To amplify all or part of the 16S rRNA gene from a diverse range of species in the sample, PCR primers with degenerative sequences are used. The number of sequences found for each species' respective 16S rRNA gene is determined by the mix of amplicons generated from different organisms in the community. Although this is useful for defining communities, it also allows pathogens with unique 16S rRNA sequences to be identified [4,5].

## Conclusion

The NGS technology greatly influences the sensitivity and specificity of this method. Prior to NGS, the full-length 16S rRNA gene was sequenced with high-quality, 700-base-long Sanger sequencing (also known as "first-generation" sequencing technology). This was time-consuming and costly, and deep sampling was not possible. When NGS became available, the majority of work was done on the 454 Life Sciences FLX sequencing instrument (second-generation sequencing technology) (Roche Diagnostics, Indianapolis, IN, USA). Only 400-base-long sequencing reads were allowed, and only a portion of the 16S rRNA gene was sequenced. The 16S rRNA gene contains nine hypervariable regions that contribute significantly to species identification. Typically, only three of these regions can be sequenced using 454 sequencing. Nonetheless, most taxa were detected to the genus level. When compared to culture results, this methodology correctly identifies pathogens in stool samples from diarrhoea patients. Furthermore, using this NGS approach, an additional pathogen that had not been reported by the diagnostic laboratory was identified in 15% of the samples.

## Conflict Of Interest

None

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