Journal of Clinical Epigenetics
ISSN: 2472-1158

Open access Perspective

DNA Sequence from Diploid Individuals in Methylase Transferase

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INTRODUCTION

DNA methylation patterns can be viewed as biomarkers that can be used to study and treat several human diseases. A detailed protocol of the experimental approach is provided. Rapid advances in high-throughput DNA sequencing technology have enabled variant discovery from Whole-Genome Sequencing (WGS) datasets. However, linking chromosomal variants to haplotypes, also known as haplotype phasing, remains challenging. The human genome is diploid, and haplotype phasing is critical for the complete interpretation and analysis of genetic variation. Independently reconstruct the sequences corresponding to the two copies of each chromosome. Unfortunately, it has been difficult to generate the allelic linkage information required to perform stepwise genome assembly. Most current genome assemblies are therefore a haploid mix of her two underlying chromosomal copies present in the individual being sequenced the ultimate goal of *de novo* assembly of sequenced reads from diploid individuals is to. Sequencing techniques that provide long accurate reads are fundamental to the generation of step-by-step genome assemblies.

DESCRIPTION

This chapter provides a brief overview of major milestones in conventional genome assembly and focuses on bioinformatics techniques developed to generate haplotype information from a variety of specialized protocols. Using these techniques as background knowledge, this chapter reviews current algorithms for generating step-by-step assemblies from long reads with low error rates. Current techniques perform haplotype-aware error-correction procedures to improve the quality of raw reads. Additionally, a variation of the traditional Overlap Layout Consensus (OLC) diagram was developed to eliminate edges between reads sequenced from different chromosomal copies. In this way, large presence/absence variants between chromosomal copies can be taken into account. The development of these algorithms and improved sequencing techniques has been critical to completing the assembly of complex genomes at the chromosomal level. Haplo-typing of single full-length transcripts can be important for the diagnosis and treatment of certain genetic diseases. Many diseases that are repeat expansions of simple tandem repeats are at the root of over 40 neurological disorders. In many of these conditions, expanding polymorphic repeats beyond a certain threshold is strongly associated with disease onset and severity. Because most repeat expansions are inherited in an autosomal dominant manner, repeat expansion disorders are usually characterized by heterozygous expansion loci associated with a single haplotype. Precise genetic drugs can be used to selectively target sequences containing expansions in a haplotype-specific manner.

CONCLUSION

However, the length of repeat expansion often exceeds the capacity of Next-Generation Sequencing (NGS) reads. Determining the exact length and haplotype of repeat expansion therefore requires special consideration and the development of custom methods. It can be adapted for use with other full-length transcripts and other repeat expansion disorders. Pyro sequencing is a synthetic DNA sequencing technology that can quantitatively detect Single Nucleotide Polymorphisms (SNPS). Pyrosequencing can calculate the extent of DNA methylation from the ratio of artificial cytosine/thymine SNPS generated by bisulfite conversion at each CPG site. This method of analysis enables reproducible and accurate determination of methylation levels at CPG sites near sequencing primers with high quantitative resolution. DNA methylation plays an important role in mammalian development and cell physiology. Alterations in DNA methylation patterns are associated not only with cancer and imprinting disorders, but also with several common diseases. Assessment of DNA methylation levels by pyro sequencing can help identify biomarkers useful in diagnosis, prognosis, treatment selection, and risk assessment for the development of various diseases.

Received:	03-October-2022	**Manuscript No:**	ipce-22-15155
Editor assigned:	05-October-2022	**PreQC No:**	ipce-22-15155 (PQ)
Reviewed:	19-October-2022	**QC No:**	ipce-22-15155
Revised:	24-October-2022	**Manuscript No:**	ipce-22-15155 (R)
Published:	31-October-2022	**DOI:**	10.21767/2472-1158-22.8.49

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Citation Renart J (2022) DNA Sequence from Diploid Individuals in Methylase Transferase. J Clin Epigen. 8:49.