International Journal of Genomics

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Special Issue on

From Genome Sequencing to Cytogenomics: How Long-Read Sequencing Will Advance Clinical Practice?

CALL FOR PAPERS

The recent advances enabled by high-throughput sequencing have the potential to revolutionize clinical practice. Indeed, the past decade has seen next-generation sequencing (NGS) contributing to the identification of the molecular basis of many genetic diseases. While these impacts from massively parallel NGS have primarily been restricted to the so-called short-read platforms (i.e., Illumina), recent developments in long-read sequencing, mainly from Pac Bio and Oxford Nanopore, have expanded the scope of genetic variation beyond SNPs to structural rearrangements and copy number variants. In addition to true long-read technologies, a number of platforms have been developed to generate the so-called 'linked-reads' using short-read techniques to generate synthetic long-reads. Collectively these 'long-read platforms (LRP)' allow one to identify structural alterations in the genome, overcoming limitations posed by short-read sequencing.

In the clinical context, microarray comparative genomic hybridization (CGH) has become the principal clinical diagnostic test for patients with developmental disabilities and congenital anomalies and for certain cancers. Although powerful, array CGH has several limitations. For example, CGH is not suitable for identifying balanced chromosomal events and small-size rearrangements as well as variants present in a rare cell population within the individual (low-level mosaicism). Because LRP can identify balanced rearrangements such as inversions and translocations, it has the potential to augment and eventually supplant standard cytogenetics methods to identify disease-related changes ranging from chromosomal rearrangements to CNVs (as well as SNPs) on a single analytical platform.

In this special issue, we aim to highlight how LRP can be applied to detect variants whose identification is currently problematic, such as balanced events, small indels, and low-level mosaic events, all of which can impact clinical practice. Moreover, since LRP still suffer from a number of limitations such as the relatively high quantity of gDNA required and the error rate of such methods compared with Illumina, we also aim at reviewing these aspects and considering possible solutions. We encourage contributions, from the development of computational approaches for facilitating the data analysis to practical applications of LRP in a clinical context, as well as review articles describing the current state of the art.

Potential topics include but are not limited to the following:

- LRP data collection, analysis, management, and visualization
- Approaches to managing the error rate of LRP and improving its predictive power
- Genotyping and haplotyping of variants to identify associations with diseases
- Identification of structural variations, such as tandem/interspersed repeats or balanced/unbalanced rearrangements
- Benchmarking the performance of LRP with respect to NGS in the identification of different genetic variations
- Characterization and implications of epigenetic variation using LRP
- *De novo* assembly and reference-free approaches to studying complex genomic rearrangements such as those resulting from chromothripsis

Authors can submit their manuscripts through the Manuscript Tracking System at https://mts.hindawi.com/submit/journals/ijg/gscil/.

Papers are published upon acceptance, regardless of the Special Issue publication date.

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