Detection of Structural Variants in the Human Genome using Nanopore Sequencing

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Nanopore sequencing is a recent technology that allows direct real-time sequencing of DNA/RNA molecules with read lengths as long as the size of the original fragments. The characteristics of nanopore sequencing make it well suited for whole genome sequencing and as a preferential tool to identify structural variants (SV), namely those associated with tumours. In this work, we present a complete workflow to detect SV in tumour samples using nanopore sequencing. We used a tumour sample to prepare DNA libraries using the Rapid Sequencing kit (Oxford Nanopore Technologies-ONT), sequenced those on the MinION device (ONT) and implemented a pipeline for SV detection using multiple bioinformatics tools. The MinION generated a total of 2.34 Gb of sequence. Overall, 87.8% of reads had a quality (Q) value >7 (quality threshold). The longest read obtained had 74369 bases and the mean read length was of 4508 bases. A total of 3470 SV were identified, including deletions, duplications, translocations, insertions and inversions. Nanopore sequencing is a fast and sensitive approach of great potential for human genomics research, namely in the detection of SV in the human genome. (This work was supported by Centre for Toxicogenomics and Human Health - UID/BIM/00009/2019 - and GenomePT project – POCI-01-0145-FEDER-022184)