Advances in portable DNA analysis bring us closer to a future with medical tricorders

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An international team of scientists led by Dr Niranjan Nagarajan, from A*STAR's Genome Institute of Singapore (GIS), has released an updated version of GraphMap, a software specifically designed to analyse data from nanopore sequencing, which has been lauded for its potential to revolutionise genomics in being rapid, cheap and portable, and able to provide results in real-time. Reported in the scientific journal *Nature Communications*, the updated software makes more than 90% of the information coming out of an Oxford Nanopore Technologies (ONT) system usable.

The analysis of the DNA code of all life forms has been rapidly advancing due to the availability of new sequencing technologies to read DNA. Applications are widespread – from monitoring the spread of deadly infectious diseases, to improving crops and livestock, to sequencing human DNA for precision medicine that could improve clinical-decision making and improve healthcare outcomes.

Until recently, DNA sequencing machines have been expensive, bulky and cumbersome, rooting their use to a few research centres around the world. With the arrival of low-cost, portable sequencing technologies in early 2014, the field is transforming rapidly and may soon go the way of the revolution in personal computing.

It may not be too far in the future before multifunction hand-held devices can be used to scan, analyse and record data, effectively allowing us to measure and monitor the genetic make-up of daily life. There are

currently ongoing efforts to develop such "tricorders", first described in the fictional universe of Star Trek, which can sense and intuitively visualise a diverse array of phenomena. For example, saliva and blood samples could be used to diagnose and prevent the spread of infections at home and at work.

However, while DNA analysis has become easier, it is still error-prone and could be made more robust. Analytical tools such as GraphMap help compensate for this using sophisticated algorithms. Furthermore, recent advances in DNA preparation techniques and sequencing hardware improvements are also rapidly moving us forward. A glimpse of the future was recently reported in a ground-breaking work, where the authors described the use of nanopore sequencers for real-time surveillance of Ebola in a field setting during the 2015 epidemic in Guinea.

Expressing his excitement for the future of genomics, Dr Nagarajan, lead author of the study and Principal Investigator of Computational & Systems Biology at the GIS noted, "Advances in DNA technologies have been truly mind-boggling and we are delighted to play a part in this revolution. GraphMap resulted from a wonderful trans-national collaboration with Ivan Sovic and Mile Sikic. Together, we hope that GraphMap will serve as a valuable addition to the toolbox for nanopore sequence analysis."

"GraphMap is a great contribution to the nanopore sequencing community. As MinION read lengths get longer and sequencing throughput increases, optimised tools like GraphMap will facilitate fast and accurate data analysis," added Prof Mark Akeson from the UC Santa Cruz Genomics Institute & Biomolecular Engineering Department, who was not involved in the research.

GIS Executive Director Prof Ng Huck Hui said, "I am delighted to learn

of this novel algorithm successfully developed by GIS and our scientific collaborators. In order to keep up with the rapid advances in the sequencing field, it is imperative that we constantly innovate to maximise the utility of genomic technologies available to us. I am positive that GraphMap's ability to mitigate sequencing errors will significantly advance our efforts in the field of consumer genomics."

More information: Ivan Sović et al. Fast and sensitive mapping of nanopore sequencing reads with GraphMap, *Nature Communications* (2016). DOI: 10.1038/ncomms11307

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