

## MEDIA RELEASE FOR IMMEDIATE RELEASE

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# ADVANCES IN PORTABLE DNA ANALYSIS BRING US CLOSER TO A FUTURE WITH MEDICAL TRICORDERS

New software analysing data from portable DNA sequencers enables applications such as pathogen surveillance in remote parts of the world

**SINGAPORE** – 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<sup>1</sup>, 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<sup>2</sup>. 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<sup>3</sup> 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.

<sup>&</sup>lt;sup>1</sup> GraphMap was released in December 2014 by the same team of researchers. The development describes the algorithmic innovations under the hood, and the new DNA analysis applications that it enables on the ultra-portable MinION sequencer.

<sup>&</sup>lt;sup>2</sup> http://biorxiv.org/content/early/2016/01/27/038042

<sup>&</sup>lt;sup>3</sup> In spring 2014, ONT released the thumb drive-sized DNA sequencing technology, which was cheap, portable and easy to use. However, it has been a challenge to interpret the resulting information as the characteristics of the data produced are very different from other sequencing technologies for which existing analysis tools were designed.

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<sup>4</sup>.

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."

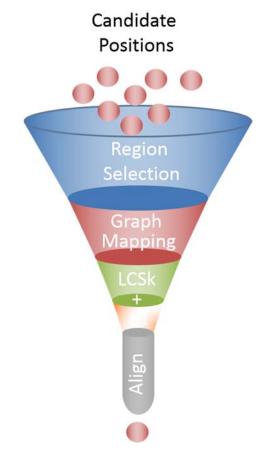
"GraphMap is a great contribution to the nanopore sequencing community. As MinION read lengths get longer and sequencing throughput increases, optimised tools like GraphMap will facilitate fast and accurate data analysis," added Prof Mark Akeson from the UC Santa Cruz Genomics Institute & Biomolecular 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."

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<sup>4</sup> http://www.nature.com/nature/journal/v530/n7589/full/nature16996.html

#### **IMAGE**



(Image source: A\*STAR's Genome Institute of Singapore)

A visual depiction of the 'read-funneling' approach used in GraphMap to align nanopore reads.

#### **Notes to Editor:**

The research findings described in this media release can be found in the scientific journal *Nature Communications*, under the title, "Fast and sensitive mapping of nanopore sequencing reads with GraphMap" by Ivan Sovic<sup>1,2\*,</sup> Mile Sikic<sup>3,4\*</sup>, Andreas Wilm<sup>1</sup>, Shannon Nicole Fenlon<sup>1,5</sup>, Swaine Chen<sup>1,6</sup>, Niranjan Nagarajan<sup>1#</sup>

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Full text of the *Nature Communications* paper can be accessed online from: <a href="http://www.nature.com/ncomms/2016/160415/ncomms11307/full/ncomms11307">http://www.nature.com/ncomms/2016/160415/ncomms11307/full/ncomms11307</a>. <a href="http://www.nature.com/ncomms/2016/160415/ncomms11307/full/ncomms11307">http://www.nature.com/ncomms/2016/160415/ncomms11307/full/ncomms11307</a>.

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## **About A\*STAR's Genome Institute of Singapore (GIS)**

The Genome Institute of Singapore (GIS) is an institute of the Agency for Science, Technology and Research (A\*STAR). It has a global vision that seeks to use genomic sciences to achieve extraordinary improvements in human health and public prosperity. Established in 2000 as a centre for genomic discovery, the GIS will pursue the integration of technology, genetics and biology towards academic, economic and societal impact.

The key research areas at the GIS include Human Genetics, Infectious Diseases, Cancer Therapeutics and Stratified Oncology, Stem Cell and Regenerative Biology, Cancer Stem Cell Biology, Computational and Systems Biology, and Translational Research.

The genomics infrastructure at the GIS is utilised to train new scientific talent, to function as a bridge for academic and industrial research, and to explore scientific questions of high impact.

For more information about GIS, please visit <a href="www.gis.a-star.edu.sg">www.gis.a-star.edu.sg</a>

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