



Dear Colleagues,

Welcome to the July 2014 (Issue 5) update of POLARIS! Time flies and POLARIS is now just over one and a half years old. Over the past six months, the team has been extremely busy and focused on building the required infrastructure and systems needed to translate promising local genomic research findings back to Singapore patients.

POLARIS engages with the Ministry of Health (MOH)

After commissioning the POLARIS@GIS lab, we have directed our efforts towards establishing administrative, wet-bench and bioinformatics SOPs

for the generation, analysis, and clinical reporting of next-generation-sequencing (NGS) data. Guided by A/Prof Tony Lim, our POLARIS clinical director and SingHealth experts, an official application to certify POLARIS@GIS has now been submitted to MOH. Prompted by this application, MOH officials are now working with the Singapore Academy of Medicine on official recommendations and guidelines for using NGS clinical diagnostics in our local healthcare system. We eagerly await the MOH response and continuing our engagement with policy-makers. On a related note, the



The POLARIS@SingHealth Lab at work

POLARIS@SingHealth facility has officially opened! Located on Level 7 of the Academia Building, the POLARIS@SingHealth group will take the lead on developing assays related to RNA-based diagnostics. By resourcing each POLARIS sister lab to interrogate different categories of diagnostic platforms, we hope to establish diverse communities comprising distinct “peaks of excellence”, each pursuing disruptive technology platforms in genomic and other –omic type applications.

POLARIS Collaboration with NUS Centre for Biomedical Ethics (CBME)

To ensure that POLARIS projects are developed in a responsible manner, we have partnered with CBME to explore several topics related to genomic medicine, such as patient consent frameworks for research data aggregation, re-contact of individuals based on genomic data, and mechanisms for managing incidental findings emerging from genomic sequencing. By engaging with bioethics experts, we hope to contribute towards the establishment of local community standards for the ethical use and governance of patient genomic data. The issue of incidental findings, in particular, represents a major challenge for the implementation of genomic medicine. Unlike incidental findings emerging from other diagnostic tests such as radiology (Solomon et al., 2014 *NEJM*), incidental findings arising from genomic data may impact not only the patient but also family members, since genetic variants of clinical significance can be inherited. Also, with the increasing take-up for genetic and genomic testing, we foresee greater demand for local personnel qualified and trained in genetic counselling. To contribute to the overall pool of genetic counsellors in Singapore, POLARIS is pleased to introduce the first of our genetic counsellors, Yasmin Bylstra, recently hired from her previous position as genetic counsellor at the Royal Melbourne Hospital, Australia, who will work with our clinical, research, and bioethics colleagues. Also, by working with SingHealth, we have identified local individuals who are being sent for overseas genetic counsellor training. We believe that upon their return these valuable individuals will represent a positive addition to the overall Singapore healthcare scene.

POLARIS in the International Arena

While we are first-and-foremost focused on the local implementation of genomic medicine, the work of POLARIS has not gone unnoticed by the international scientific community. For example, earlier this year, the POLARIS *TGFBI* test, developed in collaboration with the Singapore National Eye Centre, was highlighted

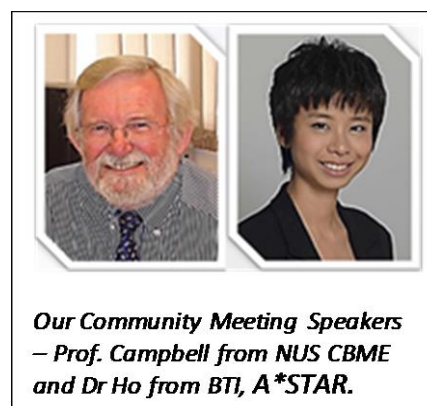
by Dr Teri Manolio, Director of Genomic Medicine at the National Human Genome Research Institute (NIH) in a presentation to the US Advisory Council on genomic medicine, as an example of how countries outside the USA are developing genomic medicine efforts. Dr Manolio's presentation can be viewed [online](#). Members of the POLARIS Core team have also been invited to join the Global Alliance for Genomic Health (G4AGH) as part of its Regulatory and Ethics Workgroup (REWG). Briefly, [G4AGH](#) is an alliance of over 190 international institutions in healthcare, research, disease advocacy and information technology, working collectively to help accelerate the potential of genomic medicine to advance human health. Led by luminaries in genomics (David Altshuer, David Haussler, Tom Hudson), GA4GH aims to establish a common framework of harmonized approaches for the effective and responsible sharing of genomic and clinical data. Through its membership in G4AGH, POLARIS will be able to obtain a sense of the "pulse" of genomic medicine efforts globally, and will use this information to align its efforts to achieve maximal impact.

Global Momentum in the Progress of Genomic Medicine

Beyond Singapore, several notable developments have also recently occurred in the arena of genomic medicine. On the industry front, the field has been revolutionized by the recent introduction of new sequencing machines such as the *Illumina HiSeq X Ten*, which promises to make good on the long-awaited \$1000 genome. At an initial cost of US\$10 million (system requirements state a minimum of 10 machines per order), to date over 100 HiSeq X Tens have been ordered, from research institutions, CROs, and biomedical startup companies in the USA, Europe and Asia. In the literature, a groundbreaking paper published by Gilissen et al. in the June issue of *Nature* reported the use of whole-genome sequencing (WGS, based on Complete Genomics technology) to increase the diagnostic accuracy of patients with severe intellectual disability. Importantly, the authors reported that WGS delivered an improved diagnostic accuracy rate of 62%, compared to 12% for array-based and 27% for exome-based methods respectively. Finally, in March 2014, the Chinese government in a sudden move issued a statement banning all medical applications of 'gene sequencing technology products', covering applications for "disease prevention, diagnosis, care, treatment, monitoring, health status evaluation and prediction of genetic diseases". Evaluations by experts suggest that this move may be related to concerns related to preventing potential abuses of genetic testing especially in prenatal testing (specifically referred to in the announcement). Although this ban was reserved on July 1st with the China Food and Drug Administration approving NGS-based products developed by BGI (Beijing Genomics Institute), such events strongly argue that the progress of genomic medicine must be conducted within a robust framework that ensures such technologies are used in a responsible, ethical and high-quality manner.

Save the Date -> Next POLARIS Community Meeting on August 20th

In closing, we wish to invite interested parties to our next POLARIS community meeting, to be held on **August 20th at 7:30 am** under the auspices of the SingHealth Grand Rounds program. We will have two speakers - Prof. Alastair Campbell, Director of NUS CBME, will discuss the bioethics of incidental findings emerging from genomic research, and Dr Ho Ying Swan, our POLARIS metabolomics champion will describe how her team is working with clinicians to use metabolomics for identifying novel signatures associated with disease progression. It promises to be an exciting affair, and CME points will be awarded. More details about the meeting can be found [here](#).



Best regards,
Patrick Tan
(On behalf of the POLARIS Team)



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