



Dear Colleagues,

Welcome to the Feb 2014 (Issue 4) update of the POLARIS program. Supported by A*STAR's Strategic Positioning Fund, POLARIS is now just over one year old! Together with our partners at A*STAR, SingHealth and NUHS, the POLARIS team has been steadily working on several fronts to develop the field of precision medicine in Singapore.

Our first clinical test!

We are pleased to announce [our first clinical assay](#) - a *TGFBI* gene mutation test designed to aid in the diagnosis and management of patients with the ocular condition Stromal Corneal Dystrophy. The

POLARIS™ *TGFBI* test covers >99% of all known pathogenic mutations in the *TGFBI* gene, and was developed in partnership with the Corneal and External Eye Disease service at SNEC (A/Prof Jodhbir Mehta), SGH Dept of Pathology (A/Prof Tony Lim), and the NUHS Molecular Diagnosis Centre (A/Prof Evelyn Koay). Following POLARIS's core principles, the *TGFBI* test is a translation of local research efforts and leverages previous work at the Singapore Eye Research Institute (A/Prof Eranga Vithana). The test has been integrated with local clinical capabilities across the island, and to date five patients have already obtained a molecular *TGFBI* diagnosis using this test. More information regarding the [POLARIS™ *TGFBI* test](#) can be obtained through Dr Mehta at SNEC, or from the POLARIS web-site (<http://polaris.a-star.edu.sg/>).

POLARIS is Illumina CS Pro Certified!

We are pleased to announce that the POLARIS@GIS laboratory is now fully equipped with Illumina HiSeq2500 and MiSeq instruments, and also all required accessory equipment. Our goal is to produce top quality data, in a consistent and reproducible manner, within the required time. As part of our quality management process and conformance to regulatory standards, the POLARIS team has worked with Illumina to review laboratory and bioinformatics processes, and technical competence of the POLARIS@GIS staff through written and practical tests. We are proud that our team has passed the audit, and we have achieved Illumina CSPro certification for next-generation sequencing. Our collaborators and clients are thus assured that when their samples are sequenced by POLARIS@GIS, they will receive the same industry-leading data quality and service which they would receive if the service is provided directly by Illumina. Besides in-house POLARIS™ assays, this also includes commercially-available sequencing assays designed for use on Illumina instruments. More information about CSPro is available at <http://www.illumina.com/services/cspro.ilmn>



Community Outreach and Education

We have had the honour of organizing two major community outreach events in the past months. On 29th



November 2013, our first CME session entitled "Next-generation sequencing for the clinician" drew >60 participants. The audience listened to lectures by A/Prof Steven Rozen (Duke-NUS) on "The promise of next generation sequencing". A/Prof Rozen provided an introduction of emerging DNA sequencing technologies and useful case examples of how DNA sequencing can be used to improve patient care. Prof Wong Tien Yin (Group Director, Research, SingHealth; shown) then discussed "Genome wide association studies of human diseases: the view from the clinician", where he

challenged the research community to deliver findings of *true clinical utility*, where molecular information can be used to refine patient diagnosis, influence treatment decisions, and predict disease risk. Prof Wong also highlighted the importance of engaging key stakeholders early, particularly those involved in health financing and patient advocacy. POLARIS thanks A/Prof Rozen and Prof Wong for their thoughtful insights, and will take their lessons to heart as we develop our own programs.

Another community event was our 2nd POLARIS Community Meeting on 21st January 2014. Held in the SingHealth Academia building, we hosted 90 participants including clinicians, academic researchers, and representatives of industry (shown). The POLARISTM TGFBI Eye test was formally introduced at this event, and the audience was updated on progress of both the POLARIS@GIS and POLARIS@SingHealth facilities. Dr Chris Wong (POLARIS Chief Operating Officer) also presented a structure for engaging new partners in the POLARIS network, using the example of tuberculosis (TB) whole-genome sequencing as an example. The POLARIS TB project involves POLARIS, SGH (Dept of Infectious Diseases), TTSH, and the Saw Swee Hock School of Public Health at NUS. To partner with POLARIS, or to access the POLARIS technology platforms, please send an email to polaris@gis.a-star.edu.sg.



Genomic Medicine – An International Reality

Finally, like Singapore, many other countries are also developing clinical implementation strategies for genomic medicine. In our view, three notable events occurred in the past few months. First, in Dec 2013, the United States White House bioethics advisory panel issued a series of recommendations regarding the handling of patient “incidental findings” arising from genomic research. The panel advocated an “anticipate and communicate” strategy, and that clinicians and researchers involved in such research should anticipate the generation of such findings, and develop plans on how to transmit such findings back to patients. Such recommendations pave the way for Singapore to develop its own local recommendations on the handling of incidental findings, and we welcome interested parties to work with us on this topic. Second, in Nov 2013, the US FDA issued a series of approvals for the Illumina MiseqDX platform as a medical diagnostic device. This is a significant development as it represents the next step in the deployment of next-generation sequencing platforms in the clinic. Third, on a more cautionary side, in the same week the US FDA also issued a “cease and desist” order to the direct-to-consumer genomics company *23andMe*, citing concerns regarding the potential use of the companies’ Personal Genome ServiceTM for the diagnosis and treatment of diseases and other conditions. These developments reflect the fast-moving nature of the field, and POLARIS is devoting a significant portion of time and effort in monitoring and learning from these international experiences. Ongoing discussion is definitely needed to arrive at a consensus as to how such information can be utilized to maximize patient benefit.

In closing, 2014 will prove to an exciting year for both POLARIS and the field of genomic and precision medicine. We welcome enthusiastic partners to work with us to explore this “new frontier”!

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



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