SingHealth Duke-NUS
Institute of
Precision Medicine

Director's Message

We are delighted to present the final quarterly update of 2018 from the SingHealth Duke NUS Institute of Precision Medicine (PRISM). This year has seen PRISM celebrate numerous firsts. Among others, our clinical and research teams published their work in well-regarded journals; *Genetics in Medicine* and *PLOS Biology*. These achievements are an affirmation of PRISM's ability to perform cutting-edge research on clinical genetics as well as biomedical informatics. Apart from that, we also conducted genomics education symposiums geared at increasing the awareness of genomics for diagnosing genetic disorders among clinical practitioners in Singapore. We are looking forward to 2019, which will see new milestones in PRISM's mission to facilitate the delivery of genomic medicine in

We also invite researchers looking into disease genetics to make use of the SingHealth Exome Consortium (SEC) database, which now contains variants from **more than 2300 samples**. The SEC database contains variants from control/healthy exomes contributed by SNEC/SERI, NCCS, KKH, NUS and PRISM/NHCS that have been re-analysed using a uniform industry-standard pipeline. Our collaborators have recently published studies that made use of SEC data, demonstrating its utility in significantly cutting down the number of candidate disease-causing variants that require in-depth analysis. The database can also serve as a population-matched control cohort for various research projects seeking to identify population genetic risk factors in Singaporeans.

If you just need to know if your variant of interest has ever been seen in a local cohort, you can query our Beacon server at http://beacon.prism-genomics.org. However, if you need to know the local allele frequency for your variant of interest, please contact us at prism@singhealth.com.sg.

Best,
Patrick Tan
Director, SingHealth Duke-NUS Institute of Precision Medicine

Publications

PRISM contributes to NCCS study on pediatric cancer predisposition syndromes
SH Chan et al, DOI: 10.1038/s41525-018-0070-7

The Cancer Genetics Service (CGS) team at the National Cancer Centre Singapore (NCCS) recently published a paper reporting on germline mutations associated with childhood cancers in Singapore. Among 108 pediatric cancer patients that underwent whole-exome sequencing (WES), 10% were found by the team to harbor mutations in cancer predisposition genes (e.g. TP53, VHL, NF1). They also evaluated the utility of clinical checklists in identifying patients likely to be affected by cancer predisposition syndromes. The PRISM informatics team contributed their bioinformatics capabilities in the area of exome sequencing data analysis, as well as interrogating the SEC database for local variation data.

Announcements

Upcoming course - Genetics Education for Physicians

PRISM and KKH are jointly organizing a course for physicians interested to know more about genetics. Taking place on the 12th of January 2019 at the SGH campus (Academia Building), the course will cover basic genetics, genetic testing methodologies, variant interpretation, genetic counseling, bioethics as well as prevailing standards. Registration fees are S$250 for SingHealth staff and S$300 for non-SingHealth staff (excl. GST). For more information or to register, please visit the SingHealth Academy page or contact ask@singhealthacademy.edu.sg. The **deadline** for registration is the **21st of December 2018**.

SingHealth Exome Consortium Release Notes
Version: Release **01122018**
- Current sample count: **2305**
- Removed closely-related individuals (up to 2nd-degree relatives)

News Around The World

New Study Published in Journal of Clinical Oncology Supports Universal Genetic Testing for all Breast Cancer Patients

Revealing the brain's molecular architecture

Israel to Sequence 100K People, Create Genomic Database to Support 'Digital Health'

What Happens When You're Convinced You Have Bad Genes

Australian National Phenome Centre: new facility could find autism prevention

How Indian biotech is driving innovation

Owkin launches global patient data sharing network for training AIs

Hong Kong could become a hub for genetics-based drugs discovery

Scientists develop 10-minute universal cancer test

We Need More Diversity in Our Genomic Databases

Alzheimer's Reclassified into Six Subgroups

How does the precision medicine initiative affect me?

A tiny startup wants to pay you for your DNA, and it could lead to the next wave of medical innovation

The UK has sequenced 100,000 whole genomes in the NHS

100,000 Reasons Genomics is the Future of Healthcare

FDA recognizes ClinGen as a resource for development of clinical tests