ATS 2020 Highlights

Respiratory Structure and Function Early Career Professionals

Get to know members of the RSF Assembly

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Is your research clinical, basic science or translational?

Translational

Tell us about your research?

Under the expert supervision of Dr. Don Sin, my research explores the fundamental mechanisms of airways disease, and how this can potentially be translated into the clinical space. Most recently, my focus has been on the genomics of COPD risk and lung function decline, biomarker discovery, and deep phenotyping of COPD patients.

Where do you see yourself in 5 years?

I would like to lead a truly translational COPD service, where cutting-edge treatments and multi-disciplinary care are integrated with clinical and laboratory-based research programs.

What do you find is the major benefit of RSF Assembly Membership?

The sense of fellowship in the RSF Assembly is amazing. Getting to rub shoulders with some of the world's leading researchers, hearing about the history of respiratory science and the trailblazers of the profession, and connecting with so many talented early career professionals is inspiring.

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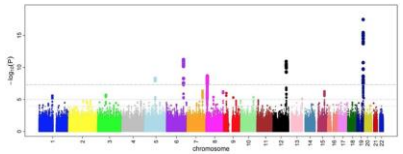
A: Developing a polygenic risk score for asthma (PRS_{Asthma})



Genome-wide association study (UK Biobank - asthma vs non-asthma)

SNP_1 ← GWAS → Phenotype

SNP_2 ← GWAS → Phenotype

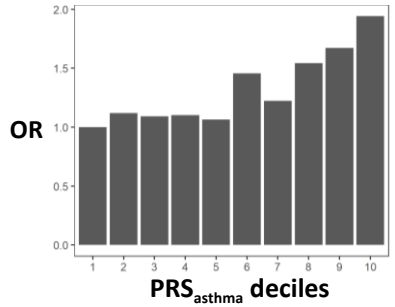
SNP_3 ← GWAS → Phenotype

...

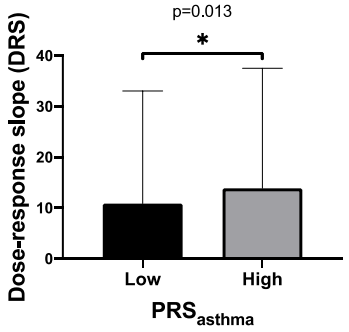
SNP_n ← GWAS → Phenotype

$\sum$ Weighted sum of risk alleles

B: PRS_{Asthma} predicts asthma Dx in COPD



C: PRS_{Asthma} assoc. with ↑ AHR in COPD



A polygenic risk score for asthma is associated with airway hyperresponsiveness in people with COPD

Objective: People with COPD may exhibit some clinical features usually associated with asthma, and it is likely that this subset of COPD patients have a genetic predisposition to the asthma phenotype. We therefore aimed to use a polygenic risk score for asthma (PRS_{Asthma}) to identify COPD patients with asthma-like features.

Methods: We first developed a PRS_{Asthma} using single-nucleotide polymorphisms (SNPs) previously associated with asthma in the UK Biobank cohort (n=33,593 cases, n=76,768 controls) (***Fig. A***). Next, we calculated the PRS_{Asthma} for genotyped COPD participants in the Lung Health Study (LHS; n=4,102) and examined its association with clinical features in this cohort.

Results: 27 SNPs were included in PRS_{Asthma}. Among COPD participants in the LHS, higher PRS_{Asthma} was associated with increased odds of having a past diagnosis of asthma (***Fig. B***) and greater AHR to methacholine (***Fig. C***).

Conclusion: A PRS for asthma, derived from patients with self-reported doctor-diagnosed asthma, is associated with increased a past diagnosis of asthma and AHR in a separate cohort of COPD patients. The PRS_{Asthma} may define a subpopulation of COPD patients with asthma-like features, which may be useful for diagnostic, prognostic or therapeutic purposes.

Supported by the MITACS Accelerate program. Supervisor: Dr. Don Sin

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