



# AGENDA

RSVP by email to  
[inga.murawski@mail.mcgill.ca](mailto:inga.murawski@mail.mcgill.ca)  
This event is CME accredited

- Meeting** Rare Respiratory Diseases Workshop
- Date** October 22<sup>nd</sup> and 23<sup>rd</sup>, 2015
- Location** Research Institute of the McGill University Health Centre  
1001 boul Decarie, Block E, Montreal, QC H4A 3J1
- Room** Block E Auditorium ES1.1129
- RSVP** This workshop is open to the public  
Kindly RSVP by email to [inga.murawski@mail.mcgill.ca](mailto:inga.murawski@mail.mcgill.ca)

### Workshop Overview:

Rare respiratory diseases (RRD) are a group of disorders that have common features with regard to organization of clinical and translational research. The purpose of this workshop is to review the local and international experience in rare respiratory disease research in order to define optimal paradigms and platform as well as to identify current gaps in this area. This workshop will feature local and international experts in rare respiratory diseases in an effort to formalize current and future approaches in this area.

### Objectives:

- To provide an overview of rare respiratory diseases as a clinical entity
- To described clinical and translational research infrastructures and methodologies for the study of RRD and discuss common concepts and hurdles
- To describe genetic and molecular approaches to studying RRD in patient cohorts and in pre-clinical models
- To put forth recommendations on the optimal organization of clinical and translational research in RRD

### Workshop Planning Committee

Chair: Dr. Arnold Kristof, McGill University  
Co-Chair: Dr. Qutayba Hamid, McGill University  
Co-Chair: Dr. Basil Petrof, McGill University

**Meakins-Christie Laboratories**  
**Seymour Heisler Memorial Lecture Fund**  
**The Dr. Dorothy Wiselberg Memorial Fund**

This workshop is generously supported by:





## Thursday October 22, 2015 - morning

Time	Description
07:45	<i>Coffee and light breakfast served</i>
<b>WELCOME REMARKS: Goals and Objectives of the Workshop</b>	
08:15	Arnold Kristof, McGill University: <i>Workshop overview and objectives</i>
<b>SESSION 1: Global and Local Approaches to Studying Rare Respiratory Diseases</b> <b>Session Chair:</b> Dr. Phil Gold, Professor of Physiology and Oncology, McGill University	
<b>DOCTOR DOROTHY WISELBERG MEMORIAL LECTURE</b>	
08:30	Bruce Trapnell, University of Cincinnati: <i>Rare respiratory diseases: an overview</i>
<b>SESSION 2: Infrastructure for Clinical and Translational Research in Rare Respiratory Diseases</b> <b>Session Chair:</b> Dr. Phil Gold, Professor of Physiology and Oncology, McGill University	
09:30	Joel Moss, National Heart, Lung, and Blood Institute, NIH: <i>Lymphangi leiomyomatosis: learning from natural history studies</i>
10:00	<i>Health Break</i>
10:15	Jennifer Landry, McGill University: <i>Bronchopulmonary dysplasia: using pediatric to adult transition cohorts in longitudinal research</i>
10:45	Adam Shapiro, McGill University: <i>Primary ciliary dyskinesia: pediatric perspectives, longitudinal cohorts, and transition clinics</i>
11:15	Frank McCormack, University of Cincinnati: <i>Rare lung disease clinical networks</i>
11:45	<b>DISCUSSION: General discussion on current infrastructure knowledge, examples, and gaps</b>
12:15	<i>Lunch</i>



## Thursday October 22, 2015 - afternoon

Time	Description
<b>SESSION 3: Preclinical Models and Mechanisms of Disease</b> <b>Session Chair:</b> Dr. Salman Qureshi, McGill University	
13:00	<b>SEYMOUR HEISLER MEMORIAL LECTURE</b> Dave Thomas, McGill University: <i>Correctors of protein trafficking diseases</i>
14:00	Bruce Trapnell, University of Cincinnati: <i>Pulmonary alveolar proteinosis: mouse models and human disease</i>
14:30	Martin Kolb, McMaster University: <i>Pre-clinical models of pulmonary fibrosis</i>
15:00	<i>Health Break</i>
<b>SESSION 4: Clinical Studies, Genotyping and Phenotyping, and Biomarker Development</b> <b>Session Chair:</b> Dr. James Martin, Professor of Medicine, McGill University	
15:15	Lisa Young, Vanderbilt University: <i>Lessons learnt from Hermansky-Pudlak syndrome</i>
15:45	Ivan Rosas, Brigham and Woman's Hospital, Boston: <i>Clinical trials in Idiopathic pulmonary fibrosis: modifiable phenotypes and novel therapies</i>
16:15	Benjamin Smith, McGill University: <i>Phenotyping by imaging to discover rare modifiers in COPD</i>
16:45	<b>DISCUSSION: General discussion on current methodologies, common concepts, and gaps</b>
17:15	<i>End of Day 1</i>
18:00	<b>Speaker Dinner (by invitation) Restaurant Graziella - 116 Rue McGill, Montréal, H2Y 2E5</b>



## Friday October 23, 2015 - morning

Time	Description
8:00	<i>Coffee and light breakfast served</i>
8:15	Arnold Kristof, McGill University: <i>Goals and Objectives for Day 2</i>
8:30	Wayne Critchley, Canadian Organization for Rare Disorders: <i>Canada's Rare Disease Strategy</i>
8:45	Gail Ouellette, Regroupement Québécois des Maladies Orphelines
<b>SESSION 4 (ctd): Clinical Studies, Genotyping and Phenotyping, and Biomarker Development</b> <b>Session Chair:</b> Dr. Sabah Hussain, McGill University	
9:00	Maimoona Zariwala, University of North Carolina: <i>Genetic Approaches in Rare Lung Diseases: using genetic techniques to uncover novel mechanisms of disease in Primary Ciliary Dyskinesia</i>
<b>SESSION 5: Moving Beyond the Lung: RRD of Neuro-musculoskeletal Origin Spanning Pediatric and Adult Care</b> <b>Session Chair:</b> Dr. Larry Lands, McGill University	
09:30	Alex MacKenzie, University of Ottawa: <i>Spinal muscular atrophy, genetic approaches to studying rare diseases, and the state of rare disease research in Canada</i>
10:00	Basil Petrof, McGill University: <i>Pompe disease and other metabolic disorders affecting respiratory muscles</i>
10:30	Frank Rauch, Shriners Hospital, McGill University: <i>Osteogenesis imperfecta: clinical and genetic aspects</i>
11:00	<i>Break</i>
11:10	<b>DISCUSSION: Current translational strategies, strengths and weaknesses as related to rare respiratory diseases, recommendations, and next steps.</b>
12:00	<i>Workshop adjourned</i>



RI-MUHC Translational Research  
in Respiratory Diseases Program



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### DOCTOR DOROTHY WISELBERG MEMORIAL LECTURE FUND



The Dr. Dorothy Wiselberg Memorial Symposium on Thoracic Diseases was established in 1981 by Dr. Wiselberg’s sisters, Rose and Fanny, to honor her memory. Dr. Wiselberg graduated from McGill University in 1930 and pursued post-graduate training in New York. Upon completing her training, she was invited to join the staff of the Royal Edward Chest Hospital (now Montreal Chest Institute) by Dr. Hugh Burke. Dr. Wiselberg remained an active member of the attending staff until her death in 1970. One of the first female physicians to work in the field of pulmonary diseases in Montreal, Dr. Wiselberg administered to patients suffering from tuberculosis for much of her career. In later years she was a member of the Medical Board of the hospital and held a teaching position at McGill University. Throughout her career, Dr. Wiselberg was noted for her great kindness and for the love and respect she engendered from her patients and those who worked with her.

### SEYMOUR HEISLER MEMORIAL LECTURE FUND

Seymour Heisler (1943-1992) was a native of Montreal who received his undergraduate and doctoral training at McGill. He did his post-doctoral research at the Karolinska Nobel institute in Stockholm Sweden. He also did a sabbatical at the National Institute of Health in Bethesda Maryland with the late Nobel Laureate Julius Axelrod. He dedicated himself to research and the pursuit of knowledge as Professor at the Department of Pharmacology at Université Laval in Quebec City and as Director of the Cellular Bioregulation Unit at its university hospital (CHUL). Dr. Heisler returned to McGill in 1990, joining the Department of Medicine and the Divisions of Respiratory and Gastroenterology. He served as a research director at the McGill Meakins-Christie Laboratories with a major commitment to the biochemistry and physiology of the secretory abnormalities underlying the devastating illness, cystic fibrosis. Dr. Heisler was a truly respected mentor who inspired and fostered the careers of many young physicians and scientists currently holding positions of leadership in universities and laboratories around the world. His serious dedication to research was leavened by a keen and dry sense of humor that characterized both his scientific collaborations and personal friendships. He is remembered as a committed, innovative researcher, and loyal friend.

