MUHC Consortium for Rare Disease Research (CRDR)

Rare Disease Research Day:

Research and Reach-Out

Thursday, February 28, 2019

Drs. Sylvia & Richard Cruess Amphitheatre & Elspeth McConnell Atrium
1001 boul. Décarie, Montreal, QC
Block E, Room ES1.1129
RSVP and Event Info: goo.gl/NjTF63

Research Day Overview
To celebrate Rare Disease Day, the MUHC CRDR is planning a ½ day multidisciplinary event to showcase some of the rare disease clinical and research programs at the RI-MUHC. The event will be organized into short talks and posters, with a special focus on cross-sectional network opportunities for applied research. Invited participants from the McGill University Health Centre network include PhD researchers, clinician-scientists (from pediatric and adult specialties), trainees, research staff, industry, foundations, and patient advocacy groups.

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<tr>
<th>TIME</th>
<th>ACTIVITY</th>
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<tbody>
<tr>
<td>8:00 - 9:00</td>
<td>Arrival &amp; Registration for all members - <em>Light breakfast &amp; coffee served</em>&lt;br&gt;Poster and Exhibitor Table set-up</td>
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<td>9:00 - 9:15</td>
<td><strong>Introductions &amp; Opening remarks:</strong> Dr. Arnold Kristof</td>
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<td>9:15 – 10:30</td>
<td><strong>Part 1: Pediatric and Adult Rare Disease Research: From transition care to research collaborations</strong>&lt;br&gt;Dr. Eric Shoubridge: Mitochondrial disorders: Genetics and approaches to therapy&lt;br&gt;Dr. Catherine Argyriou: Retinal gene therapy for peroxisome disorders: preclinical studies&lt;br&gt;Dr. John Mitchell: Burden of disease in adult mucopolysaccharidoses and new therapeutic options&lt;br&gt;&lt;b&gt;Patient Perspectives&lt;/b&gt;&lt;br&gt;&lt;b&gt;Panel Discussion Moderators:** Dr. Donald Vinh, Dr. Jennifer Landry</td>
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<td>10:30 - 10:45</td>
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Consortium de recherche sur les maladies rares (CRDR)

Journée de la recherche sur les maladies rares :

S’unir pour la recherche

Jeudi le 28 février 2019

L’amphithéâtre Drs Sylvia & Richard Cruess et l’atrium Elspeth McConnell
1001 boul. Décarie, Montreal, QC
Bloc E, Salle ES1.1129
RSVP et info : goo.gl/NjTF63

Aperçu de la journée

Pour souligner la Journée des maladies rares, l’IR-CUSM et le Consortium de recherche sur les maladies rares organisent une demi-journée pour exposer les programmes cliniques et les recherches cliniques et fondamentales concernant les maladies rares disponibles à l'IR-CUSM. Afin de faciliter les échanges entre les différents intervenants et encourager la multidisciplinarité des projets, l’événement se tiendra sous forme de brèves discussions et présentations par affiches. Joignez les participants et représentants du Centre Universitaire de santé McGill pour cet événement: chercheurs académiques et cliniciens (spécialités pédiatriques et adultes), étudiants et personnel chercheurs, de l'industrie, associations de patients et fondations.

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Thank you to our sponsors / Merci à nos commanditaires
Background and Briefing Document - Prepared By Executive Committee

Rare diseases are often complex, and patients require multidisciplinary care teams that address their physical, psychological, and social needs. Patients with rare diseases also deserve access to timely diagnosis and treatment. To aid these individuals and their families, we desperately need a health delivery and evaluation system that will address all facets of their disorders, while uncovering novel diagnostic and therapeutic approaches. The MUHC has a diverse clinical and research expertise in pediatric and adult rare diseases, as well as expertise in pediatric to adult transition care. For this reason, we have developed a MUHC Consortium for Rare Disease Research.

Vision
The MUHC Consortium for Rare Disease Research will be a world-class clinical and translational research consortium aimed at improving the lives of people with rare diseases.

Mission
To facilitate patient-oriented, collaborative, trans-disciplinary health care and research in rare diseases across the lifespan.

Objectives

- Provide excellence in the clinical care and in the transition of care for patients with Rare Diseases
- Identify and consolidate clinicians and researchers with expertise in rare diseases
- Identify and integrate existing research infrastructure and resources, including the RI-MUHC platforms, the Centre for Innovative medicine (CIM), and genomics technology
- Facilitate recruitment of patients with Rare Diseases to the MUHC/RI-MUHC
- Conduct longitudinal studies of individuals with rare diseases and develop a database that captures detailed data and bio-specimens from all rare disease patients seen at the MUHC.
- Foster pre-clinical and pilot clinical studies that connect clinicians with scientists to accelerate research (including gene discovery, biomarker development, novel therapeutics and drug repurposing)
- Conduct clinical trials: Phase I, II, III, IV
- Develop innovative diagnostic and prognostic tests
- Partner with Rare Disease patient advocacy groups and other stakeholders to ensure that all pertinent needs are addressed