McGill UNIVERSITY

Department of Anatomy and Cell Biology

Hosted by Dr. Chantal Autexier

Human RTEL1 and genome maintenance: Telomeres or centromeres? Most certainly both.

Dr. Arturo Londono

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Regulator of Telomere ELongation helicase 1 is absolutely required for the maintenance of long telomeres and for genome replication in the mouse. In humans, mutations in the gene are associated with a severe form of Dyskeratosis Congenita, the Hoyeraal-Hreidarsson Syndrome (HHS). HHS patients with mutations in RTEL1 have short telomeres and genome instability, display developmental abnormalities affecting the central nervous system and usually die from bone marrow failure and immunodeficiency. We have recently shown that RTEL1 plays unanticipated roles in RiboNucleoprotein trafficking, thus potentially broadening the underlying bases of the disease. I will present unpublished data regarding the role of human RTEL1 at telomeres as well as its role in the correct functioning of the mitotic apparatus. The implications for HHS will be discussed.

Wednesday, December 13, 2017
11:30 am

Strathcona Anatomy Building
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Room 2/36

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