Pr Joe Casey, Ph.D
Professeur titulaire
Department of Biochemistry, University of Alberta
Invité(e) par : Stéphanie Proulx

Date : Jeudi 8 novembre 2018
Heure : 14h00
Lieu : HSS, local L0-19

« Molecular defects and potential therapies in genetic corneal dystrophy »

Mutations of the gene, SLC4A11, cause childhood onset congenital hereditary endothelial dystrophy (CHED) and some cases of late-onset Fuchs endothelial corneal dystrophy (FECD). SLC4A11 encodes a membrane transport protein resident in the basolateral membrane of corneal endothelial cells. Our studies of the SLC4A11 gene product reveal that the protein normally acts as a water transporter, but possibly also acts as an ammonia and proton translocator. Studies of SLC4a11 disease alleles have shown that most mutations cause the protein to misfold and be retained inside the cell, which has led to the discovery of some compounds able to restore the diseased protein to the cell surface, with therapeutic potential. Our recent studies indicate that SLC4A11 has a role in cell adhesion, suggesting additional therapeutic directions.

Note :
Prière d’aviser vos étudiants gradués et stagiaires postdoctoraux afin d’avoir la participation de tous.

Visioconférence Sur demande