May 13, 2023

Cure RTD Awards $25,000 Grant to Dr. Alex MacKenzie at Childrens Hospital of Easter Ontario (CHEO), Canada

The Cure RTD Foundation has awarded a $25,000 grant to Dr. Alex MacKenzie and colleagues for the project "Targeting Protein Mistargeting in Riboflavin Transporter Deficiency."


Dr. Alex MacKenzie

One of the key impacts of pathogenic mutations underlying recessive disorders is they can cause protein mistargeting. The mutated but often still functional protein hangs up in endoplasmic reticulum rather than proceeding to it normal site of action within the cell. Recent evidence suggests that protein mistargeting is occurring for certain SLC52A mutations causing Riboflavin Transporter Deficiency.

In this study, HepG2 cells modified with SLC52A2 mutations known to cause RTD will be used investigated for mistargeting of the riboflavin transporter protein (RFVT). Once identified, drug and genetic screens will be used to enhanced cell survival and growth. The hope is drugs which bring this misdirected but still partially functional protein to the membrane will be revealed that lead to new treatment approaches for RTD.

Meet Dr. Alex MacKenzie

Dr. Alex MacKenzie received his MD in 1983 and his PhD in medical biophysics in 1986, both from the University of Toronto. He received his pediatric certification in 1989 from the University of Ottawa. From 2000 to 2010, he served as the CEO and director of the Children's Hospital of Eastern Ontario (CHEO) Research Institute as well as vice president of research for CHEO. He also served as vice president of research at Genome Canada from 2003 to 2004. Dr. MacKenzie works on the molecular genetics of pediatric disease with a current research focus on the molecular genetics of spinal muscular atrophy and riboflavin transporter deficiency.

Cure RTD Research Funding

This grant to Dr. Alex MacKenzie is part of Cure RTD's Basic Research and Drug Discovery programs that we're currently announcing for our 2023-2024 grant cycle.