

June 1, 2018

Hi Everyone

Quebec's 'Founder Effect' has made it a rich repository for rare recessively inherited disorders. Over time genetically oriented members of our Faculty have mined this repository with great effect leveraging clinical access with scientific discovery. This week's Departmental Acknowledgment goes to one of our more successful such miners **Paul Goodyer**.

Paul, a pediatric nephrologist, has focused on cystinosis, a lysosomal storage disorder in which a defective transporter leads to the accumulation of cysteine, an amino acid, and eventually the formation of cysteine crystals. A multi-systemic disorder, cystinosis has prominent renal effects that lead to progressive renal failure by adolescence or early adulthood. Paul's work at the bench has led to steady gains in our knowledge of cystinosis and led to his being granted earlier this Spring, as PI, a large \$2M plus three year grant from a combination of public and private funders (Genome Quebec, Genome Canada, Cystinosis Foundation and Eloxx Pharmaceuticals [an Israeli biotech firm]) that will explore a novel molecular therapy that offers the prospect of substantive disease amelioration.

Please join me in congratulating Paul on his granting success.

Have a great weekend everyone!

Michael

Michael Shevell, MDCM, FRCPC, FCAHS
Chairman, Department of Pediatrics
Professor (with Tenure)
Departments of Pediatrics and Neurology/Neurosurgery
McGill University

Guyda Chair in Pediatrics

Pediatrician-in-Chief
Montreal Children's Hospital/
McGill University Health Centre (MUHC)

Follow Me on Twitter: @McGillPeds