Medical mystery “branches” into a scientific success story

Collaborations are initiated when neighbours exchange ideas. This is a story of a boy with a puzzling disease causing symptoms ranging from inflammation of the gut, joints and skin, to blood cell abnormalities. Along the way, many SickKids physicians were involved in this case, combining expertise in rheumatology, gastroenterology, immunology, dermatology, haematology and pathology. Yet for a long time a diagnosis remained elusive. It was only after whole exome sequencing was performed, and another family with similar symptoms was identified, that clues to a potential cause began to emerge. This inspired cell biologists to look further – leading to basic mechanistic insights and identification of ARPC1B deficiency.

Panel Discussion with additional Panel Members

John Brumell, PhD, Cell Biology
Neil Warner, PhD, Cell Biology

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