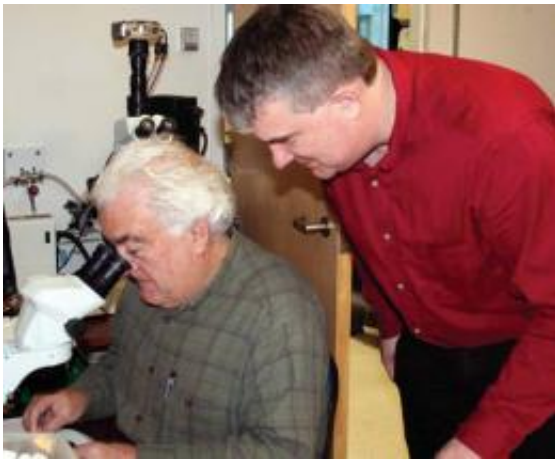


# Fruit fly researcher discovers genetic mutation that causes muscular problems, mobility issues

November 8 2010, By Raquel Maurier

---



Andrew Simmonds, an associate professor of cell biology in the Faculty of Medicine & Dentistry, and John Bell, a professor in biological sciences, have discovered the root cause of how a genetic mutation prevents muscles from attaching to each other, leading to mobility issues.

(PhysOrg.com) -- A researcher and his team in the Faculty of Medicine & Dentistry at the University of Alberta have discovered the root cause of how a genetic mutation prevents muscles from attaching to each other, leading to mobility issues.

For the last 15 years, Andrew Simmonds, an associate professor of cell biology, has been working in his lab to determine the impact of

mutations in certain genes that perform similar functions in both people and fruit flies. John Bell, a professor in biological sciences started researching the topic 15 years ago with a team, which included Simmonds as a graduate student. When Bell recently retired, Simmonds and other team members decided to continue the research.

“We said the work was too valuable to not keep going,” says Simmonds, who, along with his graduate student Hua Deng, were originally looking at how mutations in a specific gene could lead to congenital heart defects. However, when he made certain mutations in the gene, not only did it cause problems in the heart, but also caused musculoskeletal and mobility issues.

Simmonds says that by uncovering mutations that cause these developmental defects in the [fruit flies](#), physicians can look for similar [mutations](#) when diagnosing genetic diseases in people that affect mobility and muscles. He hopes other researchers will take these results forward and, ultimately, that treatments might be developed to target the effects of this mutated gene. Half the battle of discovering a treatment for any condition, he says, is to first identify the cell, protein or gene that is causing the health problem in the first place.

Bell notes that basic science research is crucial because it builds the foundation for additional research.

“It’s curiosity driven research. It is building the foundation for applied research. In this case the research has spin-offs for human medical research.”

The results of the team’s work were recently published in *Molecular Biology of the Cell*, a journal of the American Society of Cell Biology.

Provided by University of Alberta

Citation: Fruit fly researcher discovers genetic mutation that causes muscular problems, mobility issues (2010, November 8) retrieved 24 April 2024 from

<https://medicalxpress.com/news/2010-11-fruit-genetic-mutation-muscular-problems.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.