

Another piece found in disease puzzle

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(PhysOrg.com) -- An international research team led by a Simon Fraser University scientist is closer to piecing together a picture of what causes a potentially fatal, rare disease whose symptoms are prevalent in the general population.

SFU cell biologist Michel Leroux has helped discover how mutations in a specific protein known as ARL6 or BBS3 may contribute to the development of Bardet-Biedl syndrome (BBS).

Obesity, blindness and [kidney dysfunction](#) are some of the common illnesses in the general population that collectively characterize BBS, a complex [hereditary disease](#).

The May issue of the [Journal of Biological Chemistry](#), an international science publication, has published the latest findings of Leroux and his colleagues from Toronto, Calgary and the United States.

Using a technique called X-ray crystallography, they have uncovered the shape of BBS3 and discovered how mutations in specific [amino acids](#), which constitute the protein, cause it to malfunction.

The scientists have also determined that BBS3 is not only located at the base of cilia, which are small hair-like [appendages](#) that operate like cellular antennas, but is also necessary for their function.

The activities of cilia are now understood to be critical for sensory physiology—including vision, smell and hearing—as well as proper

development of organs.

“So any dysfunction of a protein that’s crucial to the proper operation of cilia by virtue of its location, like BBS3, would have serious consequences,” explains Leroux.

These latest findings further solidify Leroux’s and his colleagues’ earlier discovery that defects in cilia function cause BBS and reaffirm the role of cilia in multiple facets of human health and disease.

Over the last 10 years, SFU scientists, including Leroux and geneticist Willie Davidson, have contributed to international efforts to identify a total of 14 human [genes](#) associated with BBS.

Scientists believe that piecing together BBS’s genetic puzzle will open up research doors to understanding a number of common diseases—including obesity—affecting populations worldwide.

More information: www.jbc.org/content/early/2010/05/14/jbc.M109.070953.long

Provided by Simon Fraser University

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