MedicalXpress

First diagnostic test for hereditary children's disease

June 16 2011

A breakthrough in genetic research has uncovered the defect behind a rare hereditary children's disease that inhibits the body's ability to break down vitamin D. This discovery has led researchers to develop the first genetic and biochemical tests that positively identify the disease.

Idiopathic Infantile Hypercalcemia (IIH) is among the top ten most common inherited diseases. The researchers estimate that one in every 47,000 people – around 600 Canadians and 6,000 Americans – may suffer from IIH, but there was no way until now of confirming the diagnosis.

"Developing a positive diagnostic test for IIH is a major step in understanding this disease," says co-lead researcher Glenville Jones, a professor in the Department of Biomedical and Molecular Sciences. "We hope the test will be made available for the approximately 600 Canadians who may be afflicted with IHH."

The body's inability to break down vitamin D results in an excess of calcium in the blood. Children with IIH suffer from calcifications and tissue hardening throughout the body, as well as calcification of the kidneys and renal failure.

"This is classic case of research going from the bench to the bedside," explains Dr. Jones. "Our research started in the laboratory but the findings will have a definite impact on the health of Canadians."

This research was conducted in collaboration with pediatricians Martin Konrad and Karl-Peter Schlingmann from the University Children's Hospital in Munster, Germany, and funded in part by the Canadian Institutes of Health Research. The findings were published yesterday in the *New England Journal of Medicine*.

Provided by Queen's University