

FOR IMMEDIATE RELEASE

Canadian researchers discover gene related to the appearance of aging

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HALIFAX - Scientists in Atlantic Canada have found a gene that may play a role in skin aging. Researchers were investigating the genetic cause of a rare disorder known as cutis laxa type 2 (CL2), which causes skin on the hands, feet and face to be loose and older looking, as well as growth and developmental delays including effects on the brain. In the process, researchers found some interesting correlations with the synthesis of proline, a chemical associated with skin and joint health.

The findings are published in the current online issue of American Journal of Human Genetics (www.cell.com/AJHG), and are part of the *Atlantic Medical Genetic and Genomics Initiative* (AMGGI), an ambitious, multi-partner gene-discovery project, managed by Genome Atlantic, and funded by Genome Canada/Genome Atlantic, Capital Health, IWK Health Centre, Dalhousie University, Nova Scotia Research Innovation Trust, Dalhousie Medical Research Fund, Nova Scotia Health Research Foundation and others as listed at www.amggi.ca.

Several Maritime Canadian families with CL2 were identified by clinicians at the IWK Health Centre's Maritime Medical Genetics service. Through genomic research, scientists were able to identify the gene responsible for CL2 in these patients, citing an interruption of the metabolism of the amino acid, proline. The gene, pyrroline-5-carboxylate reductase 1, carries out the final step of proline synthesis.

Proline is a major component of connective tissue and skin proteins, collagen and elastin. It can be created by the body, and is also found in our diets. Some skin creams, cosmetics and vitamin supplements already include proline, touting its health benefits.

However, proline is not completely understood. Researchers know that it helps make 'kinks' in protein chains, critical for correct folding of these proteins. But it seems to play an independent role in protection from cellular stress. Proline made internally in our bodies may have a biological role distinct from that of dietary proline.

"This reinforces our understanding of proline as an integral part of skin health," says Dr. Mark Samuels, AMGGI co-lead. "It provides the impetus for further work that could help us understand the development of skin, the largest, and one of the most complex organs of the body. It shows that you can't mess with proline without causing a dramatic effect on many systems, including brain development."

The gene is one of six novel discoveries made to date through the AMGGI project. Among them, a gene related to sudden cardiac death in Newfoundland and Labrador and one linked to haemoglobin production.

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