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Researchers identify genetic mutation responsible for serious disorder common in Inuit

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Researchers have identified the cause for a disorder common in Inuit people that prevents the absorption of sucrose, causing gastrointestinal distress and failure to thrive in infants. The study, published in *CMAJ* (*Canadian Medical Association Journal*), identified a genetic mutation responsible for the disorder, called congenital sucrose-isomaltase deficiency (CSID).

CSID is a rare disorder in people of European descent, but is more common in Inuit people living in northern Canada, Greenland and Alaska, with rates estimated between 5% and 10%. The disorder prevents the body from absorbing particular sugars, especially sucrose, and generally presents after weaning with the introduction of sucrose-containing foods to an infant's diet. Breastfed infants are usually without symptoms.

Researchers isolated DNA from the blood of a child from Baffin Island in Nunavut, Canada's largest territory, in whom severe chronic diarrhea developed while taking sucrose-containing infant formula. They then sequenced the sucrose-isomaltase (SI) gene in this child to identify the specific genetic mutation responsible for the disorder. The same mutation occurred in about 3% of healthy Inuit control individuals living in Canada's Arctic, which led the team to conclude that a common 'founder' mutation is responsible for the high prevalence of CSID in people of Inuit descent. Until this study, a common mutation for the

disorder had not been identified.

"People with CSID may remain asymptomatic unless they consume sucrose, which is why persons consuming a Western diet are more likely to become ill," said Dr. Matthew Lines, Department of Pediatrics, University of Ottawa and Children's Hospital of Eastern Ontario (CHEO), Ottawa, Ontario, Canada. "Timely recognition of this condition and initiation of appropriate therapy is paramount. Our study should prompt physicians to consider CSID, and to review the sucrose content of a patient's diet, and specifically that of infant formula if applicable."

Dr. Lines added that as result of the research team's findings CSID, which formerly required an intestinal biopsy for diagnosis, can now be diagnosed with a simple blood test.

More information: *Canadian Medical Association Journal*, www.cmaj.ca/lookup/doi/10.1503/cmaj.140657

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