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Genetic mutation found in familial chronic diarrhea syndrome

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When the intestines are not able to properly process our diet, a variety of disorders can develop, with chronic diarrhea as a common symptom. Chronic diarrhea can also be inherited, most commonly through conditions with genetic components such as irritable bowel syndrome. Researchers in Norway, India, and at the HudsonAlpha Institute for Biotechnology have identified one heritable DNA mutation that leads to chronic diarrhea and bowel inflammation.

Shawn Levy, Ph.D., faculty investigator at HudsonAlpha said, "Based on the effects seen from this one mutation, we are hopeful that the work will aid in understanding of much more common diseases like Crohn's and irritable bowel syndrome, which also have inflammation and diarrhea as symptoms."

The Norwegian family studied for the paper published today in *The* New England Journal of Medicine has 32 living members with a number of related inflammatory bowel conditions. Such a large family allowed scientists in Norway to use traditional genetic linkage methods to narrow down the potential DNA mutation to one portion of chromosome 12, and then to a specific gene called GUCY2C.

The Norway group asked Levy and his group at HudsonAlpha to confirm initial findings on this mutation as well as determine if there were other mutations that could contribute to the disorder. "Our exome sequencing was able to rule out other mutations and demonstrate that the one change in the GUCY2C gene was common to the disease," commented Levy.

The protein made from the GUCY2C gene is involved in transmitting specific chemical signals from food consumed to the cells inside our bowels. But the family members with chronic diarrhea have a mutation that makes the protein constantly "on," or transmitting much more signal than it should. Based on this new understanding, the scientists are now evaluating possible drug treatments based on the function of the affected protein. They can also recommend that GUCY2C be reexamined in more common bowel inflammation syndromes, as it may contribute to pathology for thousands of people worldwide.

More information: The article "Familial Diarrhea Syndrome Caused by an Activating GUCY2C Mutation," by Fiskerstrand et al. can be found at the website www.nejm.org .

Provided by HudsonAlpha Institute for Biotechnology

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