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Research group discovers genetic mutations that cause intestinal obstruction

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A research group from Ben-Gurion University of the Negev (BGU) and Soroka University Medical Center led by Prof. Ohad Birk has discovered genetic mutations that lead to intestinal blockages in newborns from two Bedouin tribes in Israel.

The new paper published in the *American Journal of Human Genetics* identifies mutations in gene GUCY2C that abrogates its function. The mutations were identified in two different Negev Bedouin tribes where there were instances of intestinal obstructions in newborns without any of the other effects of Cystic Fibrosis (CF). The GUCY2C gene is known to activate the CF gene and expresses solely in the intestine.

According to the researchers, "Mutations in the GUCY2C gene might serve to protect against diarrheal infections such as *E. coli*. Unlike normal laboratory mice that die of severe diarrhea when infected with *E. coli* bacteria, mice with a GUCY2C mutation do not. Apparently, the mutation might have evolved in the Bedouin to make them more immune to diarrheal diseases and the loss of fluids in their harsh desert climate."

Prof. Birk's group is continuing the research to determine whether more subtle changes in this gene control the tendency for diarrhea or constipation in the population at large. So far, his research group has discovered the genetic mutations that lead to more than 20 diseases in human beings. Birk is the head of BGU's Morris Kahn Laboratory of Human Genetics in the National Institute for Biotechnology in the Negev

and of the Genetics Institute at Soroka University Medical Center.

Provided by American Associates, Ben-Gurion University of the Negev

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