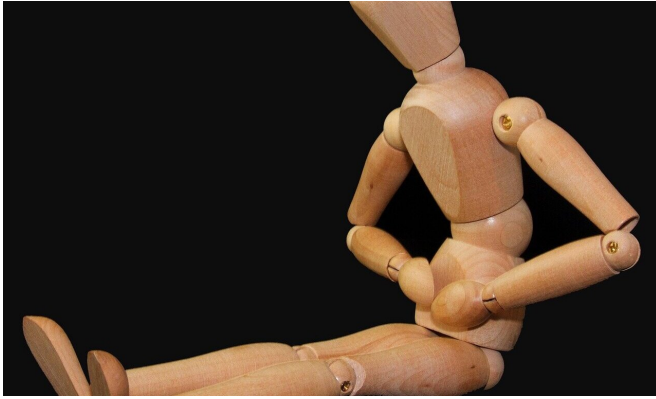


Cure found for rare form of inflammatory bowel disease

13 October 2020, by Mike Addelman



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A rare genetic condition which causes inflammatory bowel disease can be successfully treated by bone marrow transplant, according to University of Manchester and Manchester University NHS Foundation Trust researchers.

The disease, called G6PC3 deficiency, affects around one in a million people and causes inflammation of the bowel, as well as lung infections.

The team also showed that in affected patients, [white blood cells](#) called neutrophils trigger inflammation when exposed to gut bacteria, despite treatment with commonly available biological therapies.

The patients were treated at the Royal Manchester Children's Hospital—part of Manchester University NHS Foundation Trust (MFT)—one of the world's leading centers for pediatric bone marrow transplantation, under the direction of Professor Rob Wynn.

The study is published in the *Journal of Leukocyte Biology*.

Dr. Anu Goenka carried out the laboratory work as part of his Ph.D. He was funded by both a Medical Research Council Clinical Research Training Fellowship, as well as a Fellowship from the European Society for Pediatric Infectious Diseases (ESPID).

G6PC3 is important in sugar metabolism, critical for providing energy for neutrophils, which struggle to divide and function when it is deficient.

Neutrophils—which form pus—are deployed by the body to remove bacteria, particularly in the gut and lungs. The team examined neutrophil function and response to [bone marrow transplant](#) in four children with G6PC3 deficiency-associated IBD. The children's IBD had failed to respond to other immune therapies including steroids and biologics.

After the treatment, the symptoms of IBD went into remission in all of the patients. Three of the patients are now three to four years post-transplant and still in remission. Dr. Peter Arkwright, senior lecturer at the University of Manchester and consultant in pediatric immunology, Royal Manchester Children's Hospital, led the study. Professor Tracy Hussell, director of the Lydia Becker Institute of Immunology and Inflammation and NIHR Manchester Biomedical Research Center researcher, was co-lead.

Dr. Arkwright said: "Although IBD caused by G6PC3 deficiency is extremely rare and difficult to diagnose, it's thrilling that we have found a way to successfully treat it in these four children. It's rare to say a cure has been found for any disease, but I think in this case, it's perfectly accurate to say this. Our paper has also shown beyond any doubt that IBD G6PC3 deficiency can cause IBD."

Children with the deficiency are often diagnosed in the first few years of their life, though some aren't diagnosed until they are 11 and 12. And because of difficulties in detecting the disease or receiving

transplant, adults may also be affected. Bone Marrow transplant donors are either relatives or members of a bone marrow registry. The donated marrow is infused into a vein once the child has had chemotherapy. Around 10 days after transfusion, new neutrophils appear—although the immune system takes between three and four months to recover. Most patients stay in isolation in a ward for three to four weeks before going home to isolate for a further few months.

More information: Anu Goenka et al. Neutrophil dysfunction triggers inflammatory bowel disease in G6PC3 deficiency, *Journal of Leukocyte Biology* (2020). DOI: [10.1002/JLB.5AB1219-699RR](https://doi.org/10.1002/JLB.5AB1219-699RR)

Provided by University of Manchester

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