Title:
The risk of colorectal cancer in individuals with mutations of the cystic fibrosis transmembrane conductance regulator (CFTR) gene: an English population-based study

Lay Title:
The risk of developing bowel cancer in people with cystic fibrosis, or cystic fibrosis causing mutations.

Authors:
Rebecca J. Birch\textsuperscript{1,2}, Daniel Peckham\textsuperscript{1,3}, Henry M. Wood\textsuperscript{1}, Philip Quirke\textsuperscript{1}, Rob Konstant-Hambling\textsuperscript{4}, Keith Brownlee\textsuperscript{5}, Rebecca Cosgriff\textsuperscript{5}, Genomics England Research Consortium*, Nicholas Burr\textsuperscript{6}, Amy Downing\textsuperscript{1,2}

Affiliations:
\textsuperscript{1}Leeds Institute of Medical Research at St James’s, University of Leeds
\textsuperscript{2}Leeds Institute for Data Analytics, University of Leeds
\textsuperscript{3}Leeds Teaching Hospitals NHS Trust
\textsuperscript{4}NHS England
\textsuperscript{5}Cystic Fibrosis Trust, London, UK
\textsuperscript{6}Mid Yorkshire NHS Trust


1 Genomics England, London, UK
2 William Harvey Research Institute, Queen Mary University of London, London, EC1M 6BQ, UK.
What was your research question?
Is having cystic fibrosis (CF) associated with a higher risk of getting bowel cancer? Is this also the case for people who carry a single CF causing mutation?

Why is this important?
Previous studies have suggested that people with CF may have a higher risk of developing bowel cancer than people who do not have CF. These studies have also shown that people who carry CF causing mutations but do not have CF, also potentially have a higher risk of developing bowel cancer. Following the introduction of highly effective modulator therapy for most people with CF in the UK, life expectancy has increased and is expected to continue to increase. Bowel cancer is more common with increasing age.

What did you do?
The number of people with bowel cancer and Cystic Fibrosis was calculated using routinely collected healthcare data. Using data from the 100,000 Genomes Project, the presence of CF causing mutations in patients with bowel cancer was measured.

What did you find?
Patients who had Cystic Fibrosis and bowel cancer were of an average age of 52 years, this is significantly younger than in patients without Cystic Fibrosis, which was an average of 73 years. This study found that people with CF are five times more likely to develop bowel cancer than people without and it found that the overall rate of CF causing mutations in the bowel cancer population was higher than was expected. The absolute risk of any individual getting bowel cancer remains relatively low.

What does this mean and reasons for caution?
CF is linked to an increased risk of bowel cancer and the rate of CF causing mutations in a population with bowel cancer was higher than would be expected. This study shows that further research is needed and a screening programme should be developed appropriate for both people with CF and those carrying a CF causing mutation. Most bowel cancers in people with CF occurred before they would be eligible for the national screening programme.

What’s next?
Work will be undertaken to look at more cancer sites in addition to bowel cancer, both in people with CF and those who carry CF causing mutations.
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