



Research We Fund



Project:

Why does colonoscopy fail to prevent cancer of the large bowel in people with Lynch syndrome?

Research team:

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Institution: University of Melbourne

Cancer type: Colorectal

Years funded: 2020-2022

What is the project?

Cancer of the colon and rectum (CRC) is one of the most common in Australia and a leading cause of cancer-related mortality. Around 5% is caused by Lynch Syndrome, which is known as interval CRC, but this is often unpreventable despite regular screening. We aim to analyse the DNA of a large sample of interval CRCs to determine their pathway to development, examining whether most arise from the traditional carcinoma pathway or from a novel DNA mismatch repair (MMR) pathway. This could have important implications for CRC prevention in the future.

What is the need?

Lynch Syndrome is the most common hereditary cancer syndrome and carries a high risk of interval CRC. Colonoscopy

and polypectomy screenings are very effective at preventing CRC development in people without Lynch Syndrome but are often unsuccessful in preventing interval CRC in people who do have the genetic condition. This is a big clinical problem. Determining the relative incidence of interval CRC through traditional tumour development pathways and the novel one we will be studying could be a game changer for clinical management and alternative modes of treatment and could help reduce diagnoses in Australia.

What are you trying to achieve?

The aim of our research is to understand the mechanism CRC uses to develop in people with Lynch Syndrome. This project we will develop a deeper understanding regarding how CRC develops so we can determine more effective ways to prevent it.

Project timeline

Timeline	2020	2021	2022
Dissect and analyse tissue from Colon Cancer Family Registry, prepare DNA samples, and prepare library for whole exome sequencing (WES); collect medical records of study participants			
Continue tissue dissection, analysis, DNA extraction and library preparation; continue to collect and abstract medical records			
Complete WES for 100 interval CRC samples, finalise medical record abstraction for 120 participants; finalise analysis of data and submit findings for publication			

“Cancer of the colon and rectum is one of the most common in Australia. This research could be a game-changer for clinical management and alternative modes of screening.”

