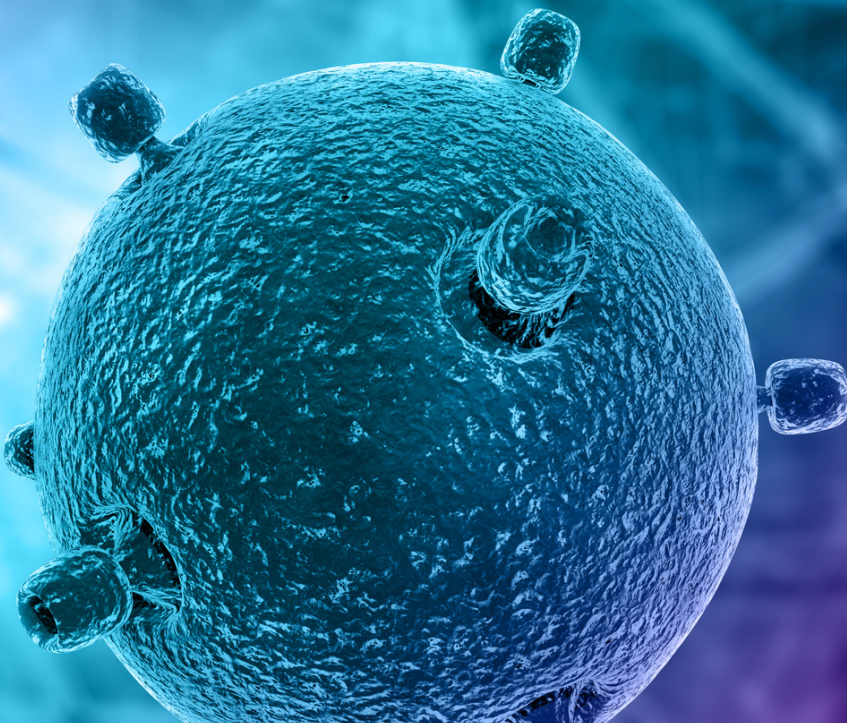




THE UNIVERSITY OF
MELBOURNE

Faculty of Medicine,
Dentistry and Health
Sciences



Colorectal Oncogenomics Group

Identifying and investigating subtypes of colorectal cancer.

The Colorectal Oncogenomics Group's research program is focused on the identification of clinically and biologically relevant subtypes of colorectal cancer including hereditary colorectal cancer and polyposis syndromes. The Colorectal Oncogenomics Group uses genomic, epigenomic and transcriptomic profiling integrated with immune cell profiling, histopathological characterisation, environmental/lifestyle risk factors and clinical data to determine the underlying aetiology of colorectal tumourigenesis so that greater steps can be made towards personalised risk stratification for early detection and prevention of this disease.

Opportunity for PhD, Honours or Masters:

Developing a multifactorial classification approach for germline variants in the colorectal cancer susceptibility genes POLE/POLD1

In the current era of genetic testing for inherited Colorectal Cancer (CRC), multiple susceptibility genes are tested simultaneously, enabling cost-effective, personalised CRC risk determination. The increasing clinical problem resulting from multi-gene panel testing is the burden of rare genetic variants identified in these high risk genes and trying to assign pathogenicity to a variant. Recently identified CRC susceptibility genes, POLE and POLD1, have been added to these multi-gene panels without well developed and validated models for variant classification. As such, the majority of rare variants identified in POLE and POLD1 are classified as variants of uncertain clinical significance (VUS). This classification has significant implications for the patient as clinicians cannot assign the most

appropriate clinical management, leaving them potentially at risk of developing CRC or being over-treated. This project will develop a multifactorial approach to variant classification for the POLE and POLD1 genes inclusive of in silico prediction tools, tumour molecular and histological features, protein structure, RNA expression, clinical and family cancer history data. It will develop expertise in genomics, molecular biology, bioinformatics, and statistical analysis. Outcomes will have significant international impact through improved risk categorization for patients identified to carry a rare variant in POLE and POLD1 genes. A stipend is available for this project.



Contact us

Colorectal Oncogenomics Group
Associate Professor Daniel Buchanan

University of Melbourne
Department of Clinical Pathology
Victorian Comprehensive Cancer
Centre
Level 10, 305 Grattan Street
Melbourne, Victoria 3000

+61 3 8559 6142
clinical-pathology@unimelb.edu.au

 For more information, visit medicine.unimelb.edu.au/clinical-pathology