



THE UNIVERSITY OF  
MELBOURNE

Faculty of Medicine,  
Dentistry and Health  
Sciences

## Rare Disease Oncogenomics Lab

Translational research of rare and less-common cancers.

The Rare Disease Oncogenomics (RADIO) laboratory is dedicated to translational research of rare and less common cancers. We use advanced methods to survey the genomic landscapes of cancer cells and their tumour “microenvironment” to better understand the biology of rare diseases, develop better diagnostic and prognostic tests, identify biomarkers of therapeutic response and find new therapeutic targets for more precise cancer treatment.

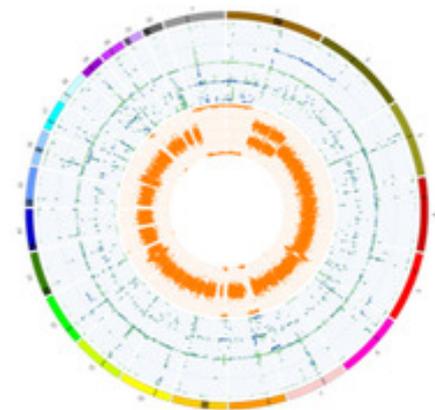
### Opportunity for Honours or Masters:

#### Circulating nucleic acid biomarkers for surveillance in Merkel cell carcinoma

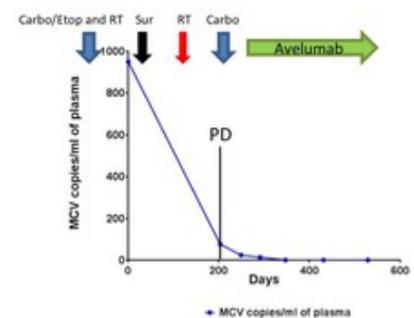
Merkel cell carcinoma (MCC) is a rare but highly aggressive and often fatal neuroendocrine skin cancer linked to viral infection and excessive sun exposure. The majority of MCC (~65%) present as early stage disease, which nevertheless has an estimated 5-year survival of only 50 percent, meaning that progression is common. Surveillance of disease recurrence is therefore an important part of patient management. Our laboratory has developed a biomarker study involving the collection of clinically well annotated biospecimens from >100 MCC patients, including blood and tissue samples taken before and during treatment.

This mature biobank represents an excellent resource to investigate the potential utility of nucleic acid biomarkers to monitor MCC response to treatment. The project will involve the detection of somatic mutations in cell free (cf) DNA extracted from blood plasma using targeted massively parallel DNA sequencing and digital droplet PCR. The detection of the tumour DNA in blood will be compared to carefully curated clinical data and FDG-PET imaging in these patients. A potential extension of this work for a masters project would involve the exploration of more novel methods for the detection of RNA in blood plasma or serum samples.

For more information, visit [medicine.unimelb.edu.au/clinical-pathology](http://medicine.unimelb.edu.au/clinical-pathology)



Massively parallel DNA sequencing



Detection of blood cfDNA in MCC

### Contact us

Rare Disease Oncogenomics  
Dr Richard Tohill

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](mailto:clinical-pathology@unimelb.edu.au)