

Research Grants 2024

Brain tumour | A ddPCR-based ctDNA Liquid Biopsy for Glioma

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Gliomas, the most common brain cancer remain a devastating diagnosis, disproportionately affecting young people and despite best treatment with surgery, chemotherapy and radiotherapy, they have a universally dismal prognosis.

A circulating molecular biomarker (known as a “liquid biopsy”) does not currently exist for brain tumours but is desperately needed. Liquid biopsies for other cancers are now available in the clinic and provide the huge advantage of being able to diagnose and guide treatment for patients via a simple blood test rather than invasive surgery or tissue biopsies. This problem is particularly important for brain tumours, because they require a high-risk neurosurgical procedure to access biopsy tissue. Furthermore, tissue biopsies can be subject to sampling error, whereas a blood test is more able to give a complete picture of the tumour gene mutation profile, which can then be used to predict outcome.

Our research group has been working on glioma circulating biomarkers for more than 10 years and has a large world-leading biobank of matched tissue and blood samples. This test could also distinguish between true tumour progression and ‘pseudoprogression’, which is enlargement of the tumour on the MRI due to inflammation rather than tumour growth.

Since 2018 we have been focusing on developing a liquid biopsy for glioma based on circulating tumour DNA (ctDNA) in plasma (from blood tests). ctDNA has the advantage of extremely high specificity for gliomas, because the known mutations in the tumour can also be detected in the blood and therefore used for diagnosis or monitoring.

This grant funding in 2024 is to focus on one specific part of the research program which is validating a ddPCR test (digital droplet PCR) for a set of specific glioma gene mutations, including IDH1, IDH2, TERTp and EGFRvIII as well as novel mismatch repair mutations in glioma (MSH2/6) that we have identified with deep sequencing (results submitted to Neuro-oncology Advances pending review).

In 2024, we are pleased to welcome back Dr Jordan Jones who has successfully completed his PhD in 2021 in glioma liquid biopsy, and who has just finished neurosurgery training to be the RMH brain tumour research fellow. Dr Jones will be conducting the majority part of this research project which will run in combination with the liquid biopsy gene profiling project run by the PMCC group, which will test our custom ctDNA panel in plasma. This grant covers the sequencing and consumables part of the budget for this project.

Grant \$92,000