Research Grants 2025

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 could circumvent the high-risk neurosurgical procedures traditionally needed to access biopsy tissue. Furthermore, surgical tissue biopsies which are only tiny pieces of the whole tumour, can be subject to sampling error, whereas a blood test is more able to give a complete picture of the wholistic gene mutation profile.

Our research group focuses 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.

A very exciting update is that our work over the last 2 years, with the assistance of the RMH Neuroscience Foundation grant funding, has culminated in 2 published papers in 2024 in Neuro-oncology Advances.

Firstly, we found that using ddPCR we can detect key DNA mutations in the blood of glioma patients including IDH mutations in low grade glioma. Second, using Next Generation Sequencing (NGS) techniques on blood (AVENIO) from high grade gliomas, we reported the early detection of DNA mismatch repair mutations caused by chemotherapy leading to resistance. This is a first in the world discovery that is garnering a lot of international attention and offers of collaboration. For instance, we have recently been invited to collaborate to provide our expertise on glioma ctDNA to a prospective trial in the UK and also to research with the University of Toronto.

We now plan to extend our work significantly and validate our ctDNA tests clinically. Over the last 6 months we have been prioritising ddPCR for IDH1 mutation for the ANHEART Phase 0 trial running at RMH (PI – Jim Whittle) and this has allowed us to optimise our ddPCR methods and techniques which are working well. This further funding allows us to focus on expanding the ddPCR work including IDH1, IDH2, TERTp and EGFRvIII as well as novel mismatch repair mutations in glioma (MSH2/6) mentioned above to the whole of our stored biobank samples.

Grant $72,000

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