

Contribution of Whole Genome Sequencing to Brain Tumour Biology – Year 1 Report

Progress Report of Study “Contribution of Whole Genome Sequencing to Brain Tumour Biology”

Reference Number 19-006-AF3 on HRCDC Website <https://hrcdc.ie/>

Data Controllers: Beaumont Hospital and Genuity Science (formerly Genomics Medicine Ireland or GMI)

Report prepared by Principal Investigator:

Dr. Michael Farrell michaelfarrell@beaumont.ie

Genomics Summary

At present, technological advances have made it possible to sequence an entire human genome (i.e., whole genome sequencing) in a matter of days and at a cost of around 800€. It is uncommon for whole genome sequencing results to immediately result in treatable targets for any disease, especially common diseases where there are substantial environmental or behavioural contributions to the disease. But when married to really detailed information about a particular disorder such as brain tumours, genomic studies can generate important new information that allow us to better predict how an individual brain tumour will behave. Such a rapidly developing science as genomics raises social, ethical, and legal issues which must be addressed in a thoughtful manner as experts and the public try to balance risks to an individual’s rights, especially to rights to privacy, with potential benefits of this new science to society at large. Improved genomics literacy for the public and for health care workers must be the goal of everyone involved in genomics.

Progress of the Beaumont Brain Tumour Research Study

Background

The Beaumont Brain Tumour Study in collaboration with Genuity Science Ireland (formerly Genomics Medicine Ireland or GMI) was established to marry the genomic information derived from archived brain tumours with subsequent clinical behaviour in order to form a Brain Tumour Information System [BTIS] that could be interrogated and used to better predict behaviour of new tumours in new patients. This study, of up to 10,000 samples, is a Beaumont led research study for the benefit of patients. Genuity Science Ireland are supporting the study by generating genomic data from the tumour samples and providing the data to Beaumont Hospital, at no cost to the Hospital, which they will use to establish the BTIS. Genuity Science will also keep a copy of the data generated, and will collaborate with academic and commercial research partners, in a strictly controlled manner, in the hopes that insights gained through its studies will lead to the development of new treatments and diagnostics.

The study originally received a waiver of consent from the Beaumont Research Ethics Committee in July 2016. All sample and data transfers were compliant with applicable regulations at that time. Beaumont Hospital and Genuity Science paused the study following the introduction of the Health Research Regulations in August 2018. No samples have been sent to Genuity Science since August 2018. The study [19-006-AF3] received a conditional consent declaration under the Health Research Regulations on November 6th, 2019, which remains valid until December 31st, 2023. The study remains on hold until the conclusion of the Public Notice Period on April 1st 2021

Changes to Study

To date there have not been any material changes made to the study and the consent declaration remains valid December 31st, 2023 and currently, changes are not proposed to the exit strategy

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outlined in the original application. No changes have been required to be made to the Data Impact Assessment statement.

Have the specific conditions attached to the consent declaration have been met?

In relation to the consent declaration granted by the Appeal Panel pursuant to the Health Research Regulations 2018 (“Consent Declaration”), there were 3 conditions attached:

Condition 1: Sharing of Research Data

Condition 1 of the Consent Declaration requested that the Beaumont and Genuity Science undertake to make the genomic data collected during the health research project, publicly available on a recognised genomic data repository in a de-identified and appropriately controlled way, once public policy has been set in that regard. As the study has been paused pending conclusion of the now extended Public Notice Period, no action has been taken to date in relation to this condition. Furthermore, public policy has not yet been formulated in Ireland with regard to the sharing of genomic research data in a public forum.

In the meantime, and in alignment with the purpose of this Condition, in response to a request from HRB-CRCI (Clinical Research Coordination Ireland), summary details of the Beaumont Brain Tumour Study have been submitted to the European Commission’s EU Survey platform [1+MillionGenomes] Initiative group:

https://ec.europa.eu/eusurvey/runner/1plusMG_Survey2020

Condition 2: Publicity Campaign

The second condition of the Consent Declaration requested a publicity campaign to be carried out by the Beaumont and Genuity Science, drawing attention to the health research project, and affording patients (or their Next of Kin) the opportunity to withdraw their brain tumour samples and personal data from the health research project. In conjunction with Brain Tumour Ireland [BTI] and following a meeting with the Board of BTI [January 12th 2020], a Public Notice which outlined details of the study was drawn up and published in the Irish national press (Irish Times, Irish Independent, Irish Daily Mail, Irish Times Weekend Edition, Sunday Independent) during the weekends of March 13-14-15 and April 24-25-26. The timing of the commencement of the Public Notice Period was undertaken in consultation with the HRCDC. As the publication of the first notice unfortunately coincided with the beginning of the national Covid-19 lockdown the notice period was extended for a further 90 days, with publication of the public notice again during the weekend of July 17-18-19. In response to feedback from the public, and in response to COVID-19 a further extension to the opt-out period has been arranged, and is set to conclude on April 1st 2021. The current declaration remains valid pending successful annual reviews until December 31st, 2023.

Discussions in relation to the format and nature of a possible further extension as described above are currently ongoing between Beaumont Hospital, Genuity Science and The Department of Health and Children.

A dedicated phone line was set up [01-7977726] and a postal address [BTIS, Beaumont Hospital, Dublin 9] together with an email address, btis@beaumont.ie, were provided. A website which outlined the project in detail was created www.btis.ie.

Additionally, the project received considerable publicity in the national press, Dáil Eireann and on social media and Public Radio during the Public Notice Period.

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Responses to Public Notices

To date, 527 contacts have been received in response to the Public Notice including 5 postal enquiries, 290 telephone calls and 232 emails. There were 10,548 web-site visits. There have been 245 Opt-Outs. Where a patient chose to opt out, an OPT-OUT confirmation email was sent to the patient together with a copy of an amendment which detailed the patient's OPT-OUT status and which was placed in the patient's neuropathology record – the primary source of all tumour samples and patient data, thereby preventing inadvertent use of the brain tumour in question in the Brain Tumour Study. For those samples that had been previously sent to Genuity Science in accordance with the then applicable regulations (prior to Aug 2018), including Research Ethics Committee Approval, a withdrawal request was sent from Beaumont to Genuity Science and internal processes were followed to ensure data deletion and destruction.

The lead Investigator provided detailed information in relation to the project to patients and relatives who contacted Beaumont to directly address any concerns or queries raised. Queries raised by and feedback received from individuals during this period have been used to improve and enhance the information available to the public on our website, www.btis.ie, in particular the FAQ section.

Condition 3: Annual Report

The third condition of the Consent Declaration requested that an annual report be published, detailing the progress of the health research project. This report is in satisfaction of this Condition for the year ending 31 December 2020.

Conclusion

The study investigators wish to thank the many patients and families of patients who are allowing us to enter their brain tumour tissue into this unique and important Irish study. We are acutely conscious of the need to better inform the public about the value of Genomics in medicine and are acutely aware of the many privacy issues associated with the rapid developments in genomics. The developments have been so rapid that at times it is extraordinarily difficult to allay fears about re-identification of individuals. Improving genomics literacy is a primary goal of the study investigators who welcome open, honest, and calm debate about a technology that that will have an influence on all of our lives. Genomics must be available to all.

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Dr. Michael Farrell



Date:

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