British Association of Dermatologists Project Grant Report
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A study of the genetics behind primary orbital melanomas

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"I would like to offer my sincere thanks to the British Association of Dermatologists for this amazing award that allowed me to pursue this very interesting project and work with such a wonderful team"

Overview
Over summer 2016, I was greatly privileged to be able to conduct an 8-week long research project at Hammersmith Hospital under the supervision of Dr Anna Rose and Dr Channa Jayasena. During the duration of this project, I picked up many new skills and was fully involved as member of an amazing research team. I received plenty of personal guidance and had a thoroughly enjoyable time.

The project
The topic of research is on primary orbital melanomas (POM). POM is an extremely rare form of malignancy. To date, there have only been approximately 50 cases reported in various small cases series and case reports. Compared to the much more common cutaneous variant, very little is known about this rare condition.

Our aim is to investigate the underlying genetic mutations of POM. The genetic changes driving tumour behaviour often give important clues to its pathophysiology and also offer opportunities for new therapies. As the first step to unravelling the mysteries of this rare condition, we used Sanger sequencing to investigate common genetic mutations reported in cutaneous melanoma and uveal melanoma in our POM samples. Namely, these are the B-RAF, N-RAS, K-RAS, GNAQ, GNA11, EIF1AX and SF3B1 genes.

We are very fortunate to have a sample of 11 biopsy-confirmed POM DNA samples from Moorfields Eye Hospital. We believe this to be one of the largest study ever done on POM.

We obtained several positive results during the course of our research. It does appear that POM is a genetically distinct from its cutaneous and uveal counterparts. Most of our patients did not share in the mutations that had been so widely reported in cutaneous melanomas and uveal melanomas. We are currently in the midst of putting together a paper which we aim to publish in due course and share our findings in conferences.

Reflections/Learning points
It was an amazing 8 weeks that seem to have flown by. Having learnt about PCR and Sanger sequencing in classes, it was a delight to be able to see them in practical use. I have no doubt that having personally done these processes now, I have a better grasp of its principles and scientific basis. This is particularly true when things do not go as planned and critical thinking is needed to reassess, troubleshoot and reattempt.

Beyond the practical skills, I have also learnt a great deal about the research world; what exactly goes behind it, what considerations need to be taken and how a research project is planned and carried out from start to finish. This first-hand experience has certainly opened my eyes to a new world that books and classroom would never have been able to do. It has further reinforced my interest in the area of research and gave me a jump start into this field.

Finally, I think the most invaluable thing I have gained from this experience made possible by the generous grant is the opportunity to meet and get to know others, especially supervisors and mentors. I felt fully part of the team and was given excellent guidance. It was heartening to meet people a little ahead of me in the career paths who are such inspiration and role models. I would not have traded this opportunity for anything else.

In conclusion, I am very grateful towards all the supervisors who have mentored me in this process and all the people with whom I worked with during this project or crossed paths. I would once again like to thank the British Association of Dermatologists for their gracious funding which made it possible.