British Association of Dermatologist
Medical student project prize report

Genetics research into Peeling Skin Syndrome at the Centre for Life

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I am extremely delighted to have been awarded the British Association of Dermatologist winter project prize. During my previous summer holidays, I decided to take some time out to sit in dermatology clinics with Dr Rob Ellis. Having piqued my interest, I was given the opportunity to undertake a lab project with his team. I had felt that there was little time spent undertaking lab work and combined with my interest in dermatology, this made for the perfect student selected component for me to undertake during my time at medical school. This award has been fundamental in allowing me to undertake genetics research at the institute of genetic medicine at the centre for life in Newcastle. I was given the opportunity to work with Dr Neil Rajan, a Wellcome Intermediate Clinical Fellow in which I undertook a project looking into genetic mutations in a patient with a suspected case of Peeling Skin Syndrome.

Peeling skin syndrome is an inherited disorder leading to spontaneous peeling of the skin. There are currently a number of known mutations in specific subtypes with further mutations being researched. Nevertheless, the condition is often misdiagnosed and patients may be treated for other conditions such as psoriasis. Our patient was a 50 year old man who was initially treated for parapsoriasis, however, his clinical symptoms included erythematous peeling of the skin with a lack of scaling. We elected to investigate identifiable or novel mutations within the Corneodesmin (CDSN) gene on chromosome 6 and Cystatin A (CSTA) gene on chromosome 3 due to their relationship with generalized peeling skin subtypes.

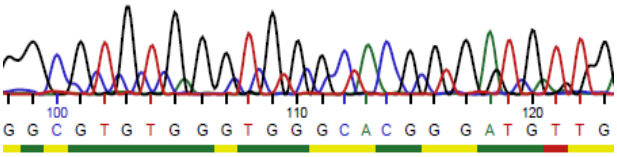
During my time in the lab, I undertook a variety of tasks including the running of polymerase chain reactions, electrophoresis, Sanger sequencing and then interpreting the readings. It all seemed like a blur when I started, however, going through the process from start to finish including creating and ordering primers and making agarose gels has allowed me to gain a comprehensive awareness of the role of genetics in medicine. This has helped put into perspective what I had actually learnt during my time at medical school. This experience has been invaluable in allowing me to understand how genetics works, and the work required in order to attain an understanding

of mutations. This direct correlation of a mutation with disease has been interesting to perceive and it has been fascinating to study how various mutations effect a patients symptoms.

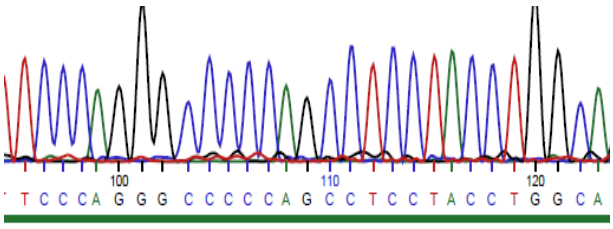
The work undertaken in a lab differed to what I had expected. A great deal of patience is required and one is not able to attain results right away and often the results gained were not clear or correct. For example, running a clean sequence is not something which occurred right away which meant, I was having to alter my methods, run PCR's and electrophoresis again and then send these off for sequencing again (See figure 1). I was able to run sequences for both CDSN and CSTA during my time and I am currently in the process of analysing and writing up our results. This experience has given me a great deal of respect for those whom work in the lab and the important role they play in medicine. I was also given the opportunity to work with some very kind people including a PhD student Majid Arefi who was extremely helpful and a great mentor during my time at the centre for life.

Figure 1: Showing difference in sequencing results with A being the first run and B being the second run

A



B



Attaining this award has allowed me to gain valuable experience and a thorough understanding into Peeling Skin Syndome. Additionally, I feel this has enhanced my exposure into the world of genetics and the vital role it plays and will continue to play in the role of dermatology. I have gained valuable knowledge and a greater appreciation of dermatology through this whole process as through my time, I was given the opportunity to undertake clinical work with Dr Rob Ellis who

heads the dermatology research at James Cook hospital and has been an inspiration. This whole process has been a wonderful experience and I have further been given the opportunity to participate in other projects. I thank the BAD from the bottom of my heart for this award.