

## **Report Nigel Clarke Memorial Bursary**

### **Dr Lilian Downie**

In 2016-2017 I was the clinical lead of a project recruiting infants who were diagnosed with congenital hearing impairment to have exome sequencing and microarray. The aim of the project was to characterise the genetic causes of congenital deafness in our population. An additional component to the project was to offer additional findings information from the exome data.

Last year at HGSA I presented the data for this cohort, the rate of diagnosis was 56% which was a significant increase from standard of care (20%). We also found that having an early molecular diagnosis did have an impact on care. 36% of infants received a non-syndromic diagnosis and were discharged from further screening or surveillance 36% (38/106) received a non-syndromic HI diagnosis and were discharged from further screening or surveillance 9% (10/106) were moved to a screening protocol tailored to their genetic diagnosis 2% (2/106) had a specific treatment offered 4% (4/106) had a complex neurodevelopmental syndrome diagnosed that informed medical care.

A significant hurdle remains in obtaining funding for genomic testing of infants diagnosed with congenital deafness beyond this project. Government requires evidence of cost effectiveness for the healthcare system to provide ongoing funding. In order to address this issue I used the money awarded for this bursary to attend a three day course run by the University of Melbourne, Practical methods for health economic evaluation.

This course provided me with background knowledge on cost analysis, analysis of outcomes and preparing data as well as practical exercises on using modelling software. Importantly it also gave me the opportunity to network with health economists and other medical professionals interested in the area of health economics. I now have the necessary skills to perform a health economic analyses on the data set from this project and provide the necessary information to funding bodies.

I wish to thank the HGSA for this award.