

Australasian Association of Clinical Geneticists Education Bursary
Recipient report

Amount: AU\$1200, used as travel fund for the 12th International Skeletal Dysplasia (ISDS) Meeting in Istanbul, Turkey (29th July to 1st August 2015), representing the trainees from Australasia.

Objectives achieved:

1. Poster presentation

Poster entitled "*Pseudodiastrophic dysplasia: two cases delineating and expanding the pre- and postnatal phenotype*" was presented during the conference. The familial case of pseudodiastrophic dysplasia in two siblings from non-consanguineous Caucasian parents has generated tremendous amount of interests. The discussion has further improved my knowledge about the condition and helped me formulate the outline of the manuscript for peer-reviewed journal. The paper was accepted for publication in the American Journal of Medical Genetics (Part A) on the 21st December 2015. I am the first author of the journal. The molecular cause for pseudodiastrophic dysplasia is not yet delineated. Further testing using next generation sequencing is underway for gene discovery.

2. Oral presentation

I had the opportunity to present an oral session entitled "*Best Practices in the Evaluation and Treatment of Foramen Magnum Stenosis in Achondroplasia During Infancy*" on behalf of Professor Ravi Savarirayan due to scheduling clash. Through preparation for the oral session, I had the opportunity to research the topic in-depth, fortifying my knowledge on the clinical management of this important complication of achondroplasia. The oral presentation also increased my confidence in presenting in international conferences.

3. Knowledge expansion and net-working

I have a special interest in skeletal dysplasia. During the conference, I have benefited most from the lectures by invited international experts, and poster and oral sessions, which have increased my knowledge on skeletal dysplasias. The conference has deepened my understanding on the pathophysiology of different types of skeletal dysplasia, and also kept me up-to-date with the current research in other institutions around the world. I also met and established contacts with many fellow clinicians and researchers for future collaboration.

Last but not least, I had the opportunity to see the beautiful city of Istanbul and experienced its rich culture. It was indeed an invaluable and unforgettable experience. I would like to extend my gratitude to the AACG for the bursary, which has made all these possible.

Patrick Yap

Date: 19th April 2016