

GeNeEvent - Fourth International Conference on Birth Defects (ICBD) & Fifth Annual National Conference of the Society for Indian Academy of Medical Genetics (SIAMGCON 2018)

The fourth International Conference on Birth Defects and Fifth Annual National Conference of the Society for Indian Academy of Medical Genetics was held in Christian Medical College, Vellore from 13th to 15th December, 2018. It was organized by CMC, Vellore and the Indo-UK Genetic Education Forum, under the aegis of SIAMG. The conference was followed by post-conference workshops on cancer genetics, cardiovascular genetics and genetic counselling and a CME on rare diseases. A number of distinguished international and Indian faculty attended the conference and shared their experience and expertise. The academic sessions included deliberations on various aspects related to the diagnosis and management of birth defects. The highlights of the meeting included the Dr SS Agarwal Oration by Dr Dhavendra Kumar from Cardiff University, UK; the Dr IC Verma Outstanding Researcher Award lecture by Dr Radha Rama Devi, Clinical Geneticist and Metabolic Disorders specialist from Hyderabad; and presentation of the Dr SS Agarwal Young Scientist Award to Dr Anju Shukla from Kasturba Medical College, Manipal, for identification of a novel causative gene *AIMP2* for neurodevelopmental disorder. There were a number of interesting free papers as well as posters presented by enthusiastic, budding medical geneticists. The conference and workshops were appreciated by all the attendees and SIAMG was lauded for its ongoing contributions towards the field of Medical Genetics in India.



GeNeEvent - Felicitation function organized for Prof. IC Verma at New Delhi

23rd December 2018

We salute your vision, up to date knowledge, infectious enthusiasm, communication, writing and editing skills and the spirit to go on! Your sense of humour, your simplicity, kind and child like heart are adorable. Your contributions in the field of Medical Genetics are outstanding which sowed the seeds of this specialty in India which is now blooming and flourishing. We all love you Sir!



Dr S S Agarwal Young Scientist Award 2018



Dr S S Agarwal Young Scientist Award of SIAMG was won by Dr Anju Shukla, currently working as Associate Professor at Department of Medical Genetics, Kasturba Medical College, Manipal. Her paper describes two consanguineous families with two affected children each with microcephaly, refractory seizures, intellectual disability and spastic quadriparesis. This work is published in the Journal of Human Genetics and reports the first human disease associated with deleterious mutations in *AIMP2*. The condition is recently catalogued in Online Mendelian Inheritance in Man as Leukodystrophy, hypomyelinating, 17 (# 618006). Her focus of research is discovery of novel phenotypes and the underlying mechanisms of rare Mendelian neurodevelopmental disorders through application of genomic techniques. (Shukla A, Das Bhowmik A, Hebbar M, Rajagopal KV, Girisha KM, Gupta N, Dalal A. Homozygosity for a nonsense variant in *AIMP2* is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriparesis. *J Hum Genet.* 2018; 63:19-25. doi: 10.1038/s10038-017-0363-1)