

## **International Summit in Human Genetics and Genomics, 2018**

The International Summit in Human Genetics and Genomics was hosted by NHGRI on the NIH campus, from August 29<sup>th</sup> –September 29<sup>th</sup>, 2018. It was the third of the 5- year (2016-2020) trans-NIH initiative to advance genetics and genomics in research and medicine, in low to middle income countries (LMIC; World Bank). Based on last year's success, two additional ICs (NICHD, NINDS) participated this year, for a total of 13 ICs, including FIC, NCI, NEI, NHGRI, NHLBI, NIAID, NIDA, NIDCD, NIDCR, NIMHD and NINR.

As mortalities from more common diseases and disorders decline, genetic disorders and congenital birth defects consume a disproportionate level of resources allotted to health care. Many LMICs lack expertise in genetics and genomics. This impacts the required training for the provision of care and contributes negatively to the welfare of these nations. The International Summit aims to alleviate these issues by filling the knowledge gap in human genetics and genomics.

With generous support from ICs at NIH and donations made to the Foundation for the NIH (FINH) by Mayo Clinic, March of Dimes, Medtronic and ASHG, 26 candidates from 17 countries (refer to world map on reverse side) attended the Summit. Participants included research scientists (4), physicians (15) physician/scientists (3), dentist (1), dentist/scientist (1) and nurses (2).

The curriculum included 11.5 days of didactics in genomic research and medical genetics, and 4.5 days of tailored training in the participants' areas of interest. Speakers (n=65) delivered over 70 lectures on various topics (ISHGG 2018\_Agenda). In addition to didactics, the training included hands-on workshops in bioinformatics, grant writing, genetics clinics and patients, and fieldtrips. The field trips included visits and lectures at academic and commercial institutions, that provided clinical and laboratory services (Children's National Health System, GeneDx, Johns Hopkins University, Kennedy Krieger Institute, Maryland Public Health Laboratories, MedStar Washington Hospital Center, National Center for Advancing Translational Sciences, NIH Clinical Center, NIH Intramural Sequencing Center). These visits informed the participants of the spectrum of experts, infrastructure and systems involved in the provision of genetic services and testing. As in previous years, the patient panel was a favorite of the Summit. Several patients shared their insights, struggles and continuing needs, because of the genetic diseases affecting them or their family members. Participants described the Summit as "invaluable", "magnificent" and "[making] an impact on my life and career."

To measure outcomes, we assessed prior knowledge, learning and interest through daily pre- and post-curriculum surveys. The results were consistent with those of previous years which describe the Summit as a unique learning opportunity for participants and speakers, and they strongly encouraged its continuance. All activities were rated as imparting knowledge and important to include in next year's summit.

The 2017 annual outcome (2016, 2017 batches) has been remarkable (ISHGG 2017\_Annual Outcome). Participants established collaborations (51) with investigators at NIH, other US-based academic institutions and amongst themselves; Published articles (147, in various stages), in their field of expertise and related to genetics; Wrote/received grants to/from NIH or other funding agencies (49); Initiated new research and/or clinical projects (43) and influenced the course of ongoing research projects (35). Our assessment of outcomes will continue for 5-years with every batch.

Based on the survey feedback (2018) and 2017 outcomes, the Summit has made good progress in achieving its goals and is in popular demand. Thus, in 2019, we hope to host up to 30 candidates. This however will only be possible with support from our Institutes and Centers at NIH and the FNIH.

**Peru**



Mario Reynaldo Cornejo-Olivas



Milagros Mariasela Dueñas Roque

**Chile**



Sandra N. Nakousi

**Argentina**



Malena Daich Varela

**Tunisia**



Olfa Messaoud EP Ezzeddine

**Sierra Leone**



Joseph Edem-Hotah

**Ghana**



Dorothy Ama Kemevor-Asima

**Nigeria**



Olanisun O. Adewole

**Ukraine**



Olena Savina

**Uganda**



Winters Primus Muttamba

**Rwanda**



Jean Nepo Utumatwishima

**Tanzania**



Sofia A. Ottaru

**Kenya**



Jesse N Gitaka

**Ethiopia**



Mekonen Eshete Abebe

**Pakistan**



Muhammad Asif Naem

**Thailand**



Pongtawat Lertwilaiwittaya

**Indonesia**



Karolus Y. Woitila Wangi

**India**



Sweta Das



Mehul A. Mistri



Aradhana Dwivedi



Atanu Kumar Dutta

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Olaniyi O. Taiwo

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Fatimah Biade Abdulkareem