International Summit in Human Genetics and Genomics, 2017

The Second International Summit in Human Genetics and Genomics was hosted by NHGRI on the NIH campus, from August 31st –September 29th, 2017. It was the second 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; defined by the World Bank). Based on last year's success, four additional ICs (NCATS, NCI, NIAID, NIDA) participated this year.

As mortalities from more common diseases and disorders decline, genetic disorders and congenital birth defects consume a disproportionate amount of resources allotted to health and medical care. Many LMICs lack expertise in genetics and genomics which impacts the provision of care related to genetic diseases and limits the training in this field. This takes a toll on the welfare of these nations. The International Summit is helping to alleviate these issues by filling the knowledge gap in human genetics and genomics.

With the generous support from Institutes at NIH (FIC, NCATS, NCI, NEI, NHLBI, NHGRI, NIAID, NIDA, NIDCD, NIDCR, NIMHD, NINR) and donations made to the Foundation for the NIH (FINH) by Mayo Clinic and the March of Dimes, 26 candidates from 24 countries (refer to world map on reverse side) attended the 2017 Summit. Participants included research scientists (3), physicians (12) physician/scientists (5), dentist/scientist (3), a nurse and genetic counselor, and two PhD candidates, one of whom was a physician.

The curriculum included 12 days of didactics in genomic research and clinical genetics medicine, and 4 days of tailored training in the participants' areas of interest. There were 58 speakers delivering over 60 lectures on a variety of topics (ISHGG 2017_Agenda). In addition to didactics, the training included a hands-on bioinformatics workshop, a grant writing session, exposure to genetics clinics and patients, and 7 fieldtrips. The field trips included visits and lectures at academic and commercial institutions providing both clinical and laboratory services (Children's National Health System, GeneDx, Johns Hopkins University, NIH Clinical Center, NIH Intramural Sequencing Center, Kennedy Krieger Institute and MedStar Washington Hospital Center). These visits helped inform participants of the spectrum of experts involved in the provision of genetic services and testing. Based upon feedback, the patient panel was a favorite of the Summit. It included several patients that shared their insights, their struggles and continuing needs, because of the genetic diseases affecting them or their family members. One participant described the Summit as "life changing and career enhancing."

To measure outcomes, we assessed knowledge, interest and learning, daily, through pre- and post-surveys. The results indicated that the Summit was 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.

Our one year outcomes from the 2016 Summit have been remarkable (ISHGG 2016_Annual Addendum). Participants have established collaborations (27) with investigators at NIH, other US-based academic institutions, and amongst themselves. Many have published articles in genetics/genomics related to their field of expertise (51) and written/received grants to/from NIH or other funding institutions (24). Our assessment of outcomes will continue for 5 years.

The Summit has trained 45 professionals from 37 countries, and based on the survey feedbacks (2017) and the annual outcomes (2016), the Summit is making great strides in achieving its goals. Due to its demand and success, we hope to expand and host up to 30 candidates in 2018. This however, will only be possible through partnership and support from our Institutes and Centers at NIH and the FNIH.

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National Institutes of Health, Bethesda, Maryland, September 1 - 29, 2017