

2016 International Summit in Human Genetics and Genomics

The International Summit in Human Genetics and Genomics, held September 1-30, 2016, was the first of a planned 5-year (2016-2020) trans-NIH initiative hosted by NHGRI that aims to advance genetics and genomics in research and medicine in low to middle income countries.

As mortalities from more common diseases and disorders decline, genetic disorders and congenital birth defects are taking an increasing proportion of resources allotted to health and medical care. Many countries are completely devoid of expertise in genetics and genomics, not only impacting the provision of care related to genetic disease, but also limiting training. The International Summit aims to help fill this gap.

With the generous support from Institutes/Centers at NIH (NIDCD, NIDCR, NEI, NHLBI, NIMHD, NINR, and FIC) and donations made to the Foundation for the NIH (FINH) by the Bill & Melinda Gates Foundation, Mayo Clinic, March of Dimes, American Dental Association Foundation, and Sequenom Corporation, nineteen (19) candidates from 13 countries (see attached world map) attended the first Summit. Participants included research scientists (3), physicians (12), dentist/scientist (1), Ph.D. nurses (2), and a medical school trainee (1).

The curriculum included three weeks of didactics in genomics research and clinical genetics, and one week of advanced training in the participants' areas of interest. There were over 50 speakers that delivered ~60 lectures on a variety of topics (ISHGG 2016_Agenda). In addition to didactics, the training included a bioinformatics workshop with hands-on activities in data analysis, exposure to genetics clinics and patients, and weekly field trips. The field trips included visits to academic and commercial institutions providing both clinical and laboratory services (Children's National Health System, GeneDx, Johns Hopkins University, NIH Intramural Sequencing Center, Kennedy Krieger Institute, and MedStar Washington Hospital Center). These visits helped familiarize the participants with the spectrum of experts involved in the provision of genetic/genomic services and testing. Based upon feedback, the patient panel was a favorite of the Summit. The panel included several patients talking to the participants, sharing insights, struggles, and continuing needs because of the diseases affecting them or their family members.

To measure outcomes, we assessed knowledge, interest, and learning on a daily basis through pre- and post-surveys. The survey results indicate that the Summit was a unique learning opportunity for participants and speakers, and they strongly encouraged its continuance.

The collective outcomes five months after the Summit have been remarkable (ISHGG 2016_Addendum). Participants have established collaborations with investigators at NIH, other US-based academic institutions, and amongst themselves. Many have published genomics-focused articles in their field of expertise (19) and submitted grant applications to NIH or other funding institutions (10). Our assessment of outcomes will continue for 5 years, as participants agreed to provide ongoing feedback as part of their acceptance into the program.

Based on the initial feedback, the Summit achieved its stated goals, and we look forward to learning more about long-term outcomes. Due to the resounding success of the inaugural Summit, we hope to expand and host up to 30 candidates in 2017. This, however, can only be achieved through partnership and support from our Institutes and Centers at NIH and the FNIH.

First International Summit in Human Genetics and Genomics

National Institutes of Health, Bethesda, Maryland, September 1 - 30, 2016

