
The Genetics Commission Activity Update

Dan Lowenstein, Genetics Commission Chair

The Genetics Commission is pleased to report major progress on a number of initiatives developed over the past few years, along with two, entirely new concepts to be launched in the coming year.

First, in the domain of public education, Alica Goldman has overseen the development of an educational brochure entitled "Epilepsy and Genetics: Things You Want to Know," which addresses the essentials of human inheritance. The pilot educational material, designed for the English speaking population of the North America, was first beta tested in a focus group that included epilepsy patients, parents, and healthy individuals. The final document was then approved by the ILAE in 2013. Positive public reception of the material prompted Spanish translation aimed at the Spanish speaking Latin American population. This next version has been finalized and will be beta tested in a focus group and then submitted for review and approval by the ILAE by the end of 2014. At the same time, we plan to pursue the same strategy for creating similar brochures in Chinese, Japanese and other languages considered to be high priority by the ILAE.



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Second, the ILAE Consortium on Complex Epilepsies, sponsored by the Genetics Commission and led by Sam Berkovic, has achieved a major milestone in completing a genome-wide meta-analysis of almost 9,000 patients with epilepsy and over 26,000 controls. This effort, the first of its kind in epilepsy, brought together nine research groups from throughout the world under the aegis of the ILAE, and the initial results (currently submitted for publication) demonstrate the tremendous value that comes from open, large-scale, generous collaboration within the epilepsy research community.

Third, and in the new initiatives category, Ingo Helbig is developing an online communication strategy to share and discuss recent findings in the field of epilepsy genetics and related areas. To this end, the "Beyond the Ion Channel — the Channelopathist" blog of the European EuroEPI-NOMICS consortium (channelopathist.net) will be continued under the leadership of the Genetics Commission. This platform will be enhanced by the "Epilepsioeme" database, an online knowledge base for constant, concise and expert-reviewed information on epilepsy genetics that will be generated during the term of the current Genetics Commission. Our hope is that this novel approach will allow the ILAE Genetics Commission to reach out to clinicians and scientists and make epilepsy genetics more accessible for professionals and lay persons.¹⁴

Finally, recognizing that healthcare professionals have difficulty keeping up with advances in epilepsy genetics, the Genetics Commission, under the leadership of Nigel

Tan, has planned a new series on Genetic Literacy in Epilepsy. The goal of this series, to be published in *Epilepsia*, is to provide a concise, readable resource on genetic testing in epilepsy for the clinician on the ground so that he/she can apply that knowledge at point-of-care. The series will begin with a primer on genetics, and then progresses to cover frequently encountered epilepsies or epilepsy syndromes, e.g., genetic testing in epileptic encephalopathies, in progressive myoclonic epilepsies, or in IGEs / GGEs. We plan for each paper to start and end with a clinical scenario or case to ensure a strong and consistent clinical slant. The core of each paper will be a discussion centered around the case — who to test, why test, and what to do with the test result. Finally, the discussion returns to the case and a clinical conclusion is made. Each paper will also end with a short online multiple-choice test, with answers and feedback provided at the end of the test to reinforce learning.

Commission Members: Dan Lowenstein, Chair, Sam Berkovic, Peter de Jonghe, Alica Goldman, Ingo Helbig, Yuwu Jiang, Mitsuhiro Kato, Heather Mefford, Steve Petrou, Parthasarathy Satishchandra, Nigel Tan, and Helen Cross, MC Liaison.