

COMMISSION ON GENETICS OF EPILEPSY



Sam Berkovic

List of Members

Chair	Sam Berkovic (Australia)
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Finance	Shinichi Hirose (Japan) Thomas Sander (Germany) Peter DeJonghe (Belgium)
Information Officer	Nigel Tan (Singapore) Sanjay Sisodyia (UK) Dan Lowenstein (USA)
Education Secretary	Marcello Kauffman (Argentina) Alica Goldman (USA) Carla Marini (Italy)
Committee Liaison	Michel Baulac

Aims

- Initiative 1: Improve the genetic literacy of ILAE members
- Initiative 2: Develop information sources regarding genetics of epilepsy for the public, taking into account regional sensitivities, cultural factors and possible stigmatization related to epilepsy and genetics.
- Initiative 3: Assist in coordination of international efforts to understand the basis of complex epilepsies that will require large multinational cohorts.

Mission

Improving knowledge and application of genetics in epilepsy across the world.

Commission Activities from July 2012 through June 2013

- Initiative 1:
- EpiGAD has been regularly maintained, updated and audited.
 - Guidelines on SCN1A testing completed and published in *Epilepsia*.
 - Liaison with GeneTests website continues; draft document underway
- Initiative 2: Pamphlet for general information on epilepsy has been developed
- Initiative 3: The ILAE Consortium on Complex Epilepsies has been established and is functioning well. It has become a useful body for communication between genetics groups, in addition to its direct activities

Accomplishments (2012-2013)

- The major accomplishments have been
- Publication of guidelines for SCN1A testing (Hirose S, Scheffer IE, Marini C, De Jonghe P, Andermann E, Goldman AM, Kauffman M,

Tan NCK, Lowenstein DH, Sisodiya SM, Ottman R, Berkovic SF for the Genetics Commission of the International League Against Epilepsy. SCN1A Testing for Epilepsy: application in clinical practice. *Epilepsia* 2013; 54: 946-52.

- Successful functioning of the ILAE Consortium on Complex Epilepsies. We have accumulated a data set of GWAS data on 10,000 patients with epilepsy from Europe, UK, North America, Hong Kong and Australia. A phenotyping committee has standardized the clinical data, the analysis committee has standardized analytic methods across sites and a meta-analysis is in progress. A meeting to plan the first publication occurred in Montreal (June 2013). Publication anticipated in 2014.

As previously highlighted, the ILAE Consortium previously attracted the attention of NINDS and we were actively involved in planning and executing a successful meeting in San Diego (September 2010) which led to a White Paper on the subject and a Request for Applications from NIH which funded a \$25 million "Center without Walls" on Epilepsy Genetics that is now highly successful.

Recommendations for Future Work

Increased energy needs to be put into educational aspects of the Commission. The internationalization and large-scale collaboration of genetic research is a priority and the Commission has the reach and authority to be the body to effect this.

New Commission Members (2013-2017)

Dan Lowenstein, Chair (USA)
Sam Berkovic (Australia)
Peter de Jonghe (Belgium)
Alica Goldman (USA)
Ingo Helbig (Germany)
Yuwu Jiang (China)
Mitsuhiro Kato (Japan)
Heather Mefford (USA)
Steve Petrou (Australia)
Parthasarathy Satishchandra (India)
Nigel Tan (Singapore)
Helen Cross, EC Liaison (UK)