

DKF Research Conference

Langhans Hörsaal Pathologie
Murtenstrasse 31, 3010 Bern

Date October 3, 2016, 5 pm – 6 pm

Title **Advances in genetics of epilepsy**

Speaker Prof. Dr. Eric Leguern, MD, PhD, Neurogenetics Laboratory,
Department of Medical Genetics and ICM (Brain and Spine Institute),
Hôpital Pitié-Salpêtrière, F-Paris

Prof. Dr. Eric Leguern, MD, PhD, is head of the Neurogenetics Laboratory at La Pitié-Salpêtrière hospital in Paris. He is also head of the research team “Genetics of Epilepsy” at the ICM (Institut du Cerveau et de la Moelle épinière: Brain and Spine Institute) in the same hospital. He is Professor of Medical Genetics at the Pierre and Marie Curie Faculty of Medicine (Pierre and Marie Curie University – Paris / Sorbonne University).

Abstract

Advances in genetics of epilepsy

Epilepsy is a frequent neurological disorder affecting about 1% of the population. It is characterized by recurrent and spontaneous epileptic seizures, which are transient episodes of abnormal synchronous neuronal firing in one or more brain regions.

Epilepsy can be broadly divided into two groups: partial epilepsy (also known as focal epilepsy) and generalized epilepsy (GE). Epilepsies can be further classified into: 1) Epilepsies of unknown origins; 2) Structural or metabolic epilepsies, caused either by a structural lesion (post trauma or tumors) or by an infection or a metabolic disturbance or 3) Genetic (idiopathic) epilepsies.

The genetic factors predisposing to genetic epilepsies remain elusive, despite the high heritability of the disease. Advances in genetics of epilepsy have been made predominantly on monogenic forms, where the standard strategies of gene mapping and cloning could easily be applied. Since 1998, our group has been interested in these familial forms of epilepsy. We identified mutations in genes encoding ion channels like SCN1A (sodium channel subunit $\alpha 1$ gene), GABARG2 ($\gamma 2$ GABAA receptor subunit gene) or HCN1 (Hyperpolarization-Activated Cyclic Nucleotide-Gated Potassium Channel 1 gene) but also proteins with various functions like PDCH19 (Protocadherin 19) or DEPDC5 (DEP Domain-Containing Protein 5). Studying these genes and proteins has brought a lot of questions about inheritance and physiopathology, and answering these questions is the key to a better care for patients and their families.

Prof. Dr. Eric Leguern has been invited by Prof. Dr. Hugues Abriel, Department of Clinical Research, University of Bern.

December 5, 2016

Next DKF Research Conference

Dr. Michaela Kneissel, Global Head, Musculoskeletal Disease Area,
Novartis Institutes for Biomedical Research, Novartis Pharma AG, Basel

The DKF Research Conference takes place from 5 pm – 6 pm and will be followed by an apéro.

Everybody is welcome!



Departement Klinische
Forschung DKF
Murtenstrasse 35
3008 Bern
Tel. +41 31 632 09 98
Fax +41 31 632 09 46
verena.frazao@dkf.unibe.ch
www.dkf.unibe.ch



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