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# Epilepsies linked to mTOR1 pathway


**演者** **Dr. Stephanie Baulac**

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**日時** 2017 年 7 月 14 日（金）18:00 － 19:30

**会場** M&D タワー21 階大学院講義室 1

**講演要旨**


Focal epilepsies, the most frequent epilepsy type, have long been thought to be acquired disorders; several focal epilepsy syndromes are now proven to be monogenic disorders. While earlier genetic studies have demonstrated a strong contribution of ion channel and neurotransmitter receptor genes, or synaptic secreted protein genes, later work has revealed a new class of genes encoding components of the mechanistic target of rapamycin (mTOR) signal transduction pathway. The mTOR pathway controls a myriad of essential biological processes like cell growth and protein synthesis in response to a variety of signals. Recently, germline mutations have been found in genes encoding the components of the GATOR1 complex (DEPDC5, NPRL2, NPRL3), a repressor of mTORC1 in familial focal epilepsies. These mutations are increasingly recognized as causing a wide and yet evolving spectrum of focal epilepsy syndromes, with and without cortical structural abnormalities (usually focal cortical dysplasia). Brain somatic mutations in the gene encoding mTOR itself (MTOR) have recently been linked to malformation of cortical development such as focal cortical dysplasia.
This seminar will review the evolving clinical and molecular spectrum of GATOR1-related epilepsies, and present recent data on the neurobiology of mTORC1-related epilepsies using knockout rodent models.


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