

# セミナーのお知らせ

演者： Dr. Pleasantine Mill

所属： MRC Human Genetics Unit, Institute for Genetics and Molecular Medicine, Edinburgh, UK.

演題： Genetic And Biochemical Characterization Of Mammalian Cilia Compartmentalization

日時： 平成26年 9月26日(金) 15:00~16:00

場所： 医学部講義棟2F、第2講義室

**講演要旨**：Patterning of the embryo is a tightly controlled process which requires the correct integration and interpretation of key developmental signals. Small microtubule-based projections, or cilia, found on almost every mammalian cell play a critical role in signal transduction during development and disease. This is because cilia have a unique composition that is enriched in many receptors and signalling components, analogous to an antenna for focusing signalling responses. Dysfunction of the cilium underlies a group of human genetic disorders, termed ciliopathies. Despite a growing list of genes implicated in cilia biology, we have very little idea what the encoded proteins actually do in cells, or how mutations cause the disease. Consequently most of these cilia-related diseases are currently untreatable. Using mouse as a genetic model of human disease, we aim to understand how cilia are maintained as a specialized signalling compartments. We are also searching for cell-type specific differences how traffic to the cilia, necessary for ciliary function, is regulated. Work from our studies on pan-ciliary retrograde IFT mutants as well as factors restricted to subset of cilia which are motile will be presented. Identifying novel ciliary components and characterizing their functions will lead to a better understanding for the diagnosis and clinical management within ciliopathy spectrum.

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