



## ***BMC-GPMLS: Distinguished lecture series***

# **Professor Rudolf Jaenisch**

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**Title: „Epigenetics, stem cells and disease research“**



The development of the iPS cell technology has revolutionized our ability to study development and diseases in defined *in vitro* cell culture systems. The talk will focus on the use of gene editing and stem cells for the study 1) of epigenetic regulation in development and 2) our efforts to use iPS cell technology to get mechanistic insights into sporadic Parkinson disease.

*1. Monitoring the dynamics of DNA methylation at single cell resolution during development and disease:* DNA methylation is a broadly studied epigenetic modification that is essential for normal mammalian development. Current methods to quantify methylation provide only a static "snap shot" of DNA methylation, thus precluding the study of real-time methylation dynamics during cell fate changes. We have established a new approach that enables monitoring loci-specific DNA methylation dynamics at single-cell resolution.

*2. Parkinson's disease (PD):* A major effort of the lab is devoted to study Parkinson's disease (PD). We have generated isogenic pairs of iPS and ES cells that differ exclusively at the A53T or E46K mutation of the synuclein gene, both dominant point mutations that cause early onset of PD. However, the great majority of PD is polygenic with many loci that contribute to disease risk as identified in GWA studies. Most of these risk loci identified in GWA studies are localized to regulatory regions (as opposed to coding sequences), but these studies remain largely descriptive providing little or no mechanistic insight. Using CRISPR/Cas gene editing we have generated isogenic neurons that differ at specific GWAS SNPs in an effort to molecularly define the effect of risk alleles on down stream gene expression and cellular phenotype.

**Time: Monday, June 27<sup>th</sup> , 14:30-15:30**

**Location: DeCODE Genetics, Fróði auditorium**