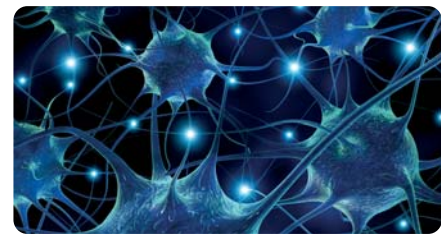


# Joint DANDRITE & MEMBRANES Lecture



**Thursday 15 Sept, 2016 at 10.15 - 11.00**

Venue: The AIAS Auditorium, building 1632, 2nd floor  
Høegh-Guldbergs Gade 6B, 8000 Aarhus C



**Dr. Kathleen J. Sweadner, Ph.D.**

Laboratory of Molecular Neurophysiology  
Massachusetts General Hospital & Harvard Medical School

## The molecular landscape of Na,K-ATPase mutations: genotype/phenotype relationships in human disease

To date, there are over 200 mutations in subunits of Na,K-ATPases that cause human diseases. Gene carriers present with a range of symptoms, however. In most cases, a clear genotype/phenotype relationship has not been found. Genomic data on random variants and human mutations in all four of the catalytic subunit isoforms show that each one has a different pattern of mutation. The structural distributions of known mutations also differ in significant ways. The evidence indicates that entire classes of mutations are missing from the known mutations. This suggests that entirely different human phenotypes and risk factors will eventually be discovered in this important gene family.

**Host:** Prof. Poul Nissen, DANDRITE - the Danish Research Institute of Translational Neuroscience, Dept. Molecular Biology and Genetics, Aarhus University