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Institute for Quantitative & Computational Biosciences

**IQCB Seminar**
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10:30 AM

Lecture Hall (00.187) at BioZentrum I, Hanns-Dieter-Hüsch-Weg 15, 55128 Mainz


# Dr. Andy Berglund

The RNA Institute, University at Albany, USA


# Dysregulated RNA Splicing in Repeat Expansion Diseases and Development of Potential Therapeutics


Repeat expansions are the underlying cause for a large and ever-growing family of 50+ neurological, neuromuscular, and neurodegenerative diseases, which include the leading causes of amyotrophic lateral sclerosis (C9-ALS) and adult-onset muscular dystrophy (DM1). These diseases also share disease mechanisms including RNA gain-of-function and protein toxicity, and result in serious life-threatening symptoms that currently lack viable treatments. The aim of this talk is to explore the connection between dysregulated RNA splicing and repeat expansion diseases, using the most well studied "spliceopathy" myotonic dystrophy type 1 (DM1), and to explore current therapeutic developments for these diseases. In DM1, expression of toxic CUG expansion RNAs sequesters the MBNL family of splicing proteins into hallmark nuclear foci, resulting in dysregulation of thousands of alternative splicing events. Despite DM1 being one of the most highly heterogeneous diseases, making clinical care and trials difficult, key splicing events are one of the best biomarkers for the disease and are utilized in many of the current clinical trials. My group uses biochemical, cellular, genomic and computational approaches to better understand DM1 and other repeat expansion disease, then leverages that knowledge to develop novel small molecules and other novel therapeutics approaches. Finally, while research in DM is critical to advancing our understanding of the disease and developing treatments, it is also a valuable tool for training the next generation of RNA researchers and workers.


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***Keywords:*** *repeat expansion disease, myotonic dystrophy type 1, RNA dysregulation, alternative splicing, small molecule therapeutics, natural products, strength training, disease biomarkers.*



For further information please contact
IQCB coordinator Christine Driller
iqcb@uni-mainz.de | +49 (0) 6131 - 39 26517


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