

# DBMR Research Conference

Seminar Room F502  
Murtenstrasse 50, 3008 Bern

Date **June 30, 2022, 12:00 – 13:00**

Title **Epigenomic Strategies for Discovery and Characterization of Enhancers in Human Disease**

Speaker **Dr. Axel Visel**  
**Lawrence Berkeley National Laboratory and Joint Genome Institute**  
**School of Natural Sciences, University of California, Merced (US)**

**Bio:** Dr. Visel received his Ph.D. in 2004 from the Max Planck Institute in Hanover, Germany, where he developed novel tools for large-scale in situ gene expression analysis in mouse embryos. His current research interests cover a wide range of functional genomics approaches aimed at understanding the biological functions encoded in the genomes of animals, plants, and microbes.

An area of particular interest that he will be talking about are the functions embedded in the non-coding DNA of the human genome. In particular, his group has been developing methods for the identification and characterization of distant-acting enhancer sequences and has extensively used mouse models to study the role of these enhancers in development, disease, and evolution of vertebrates.

Dr. Visel is a Senior Staff Scientist at Lawrence Berkeley National Laboratory and the Deputy Director of Science at the Joint Genome Institute (JGI), a Genome Science User Facility funded by the U.S. Department of Energy. In addition, Dr. Visel also holds an appointment as an Adjunct Professor at the School of Natural Sciences at the University of California, Merced.

**Abstract:** The human genome harbors tens of thousands of distant-acting gene regulatory sequences that play important roles in the development and function of the human body. Multiple converging lines of evidence from experimental and human genetic studies indicate that both common and rare sequence variants involving enhancers play major roles in Mendelian and complex human disease phenotypes. However, the underlying molecular mechanisms are difficult to study due to our limited understanding of the in vivo functions of enhancers, as well as their molecular and functional interactions with their respective target genes. We use a combination of sequence-based molecular approaches (ChIP-seq), large-scale transgenic mouse studies (<http://enhancer.lbl.gov>), and CRISPR genome editing in the mouse model to study the in vivo function of enhancers in developmental, evolutionary, and disease-related processes. Using examples from our ongoing work, I will illustrate how these studies provide insight into the function and evolution of distant-acting regulatory sequences, and how improved tools for understanding the impact of sequence variation on enhancer function offer a starting point for understanding their role in human disease and potentially open new avenues for precision medicine.

**Dr. Axel Visel has been invited by Ass. Prof. Dr. Marco Osterwalder, Osterwalder Group Leader, Cardiovascular Diseases, Department of BioMedical Research, University of Bern**

An apéro will follow the Research Conference. This is a hybrid RC.

- To attend the virtual RC on zoom, please scan the code.
- To attend the RC in person, kindly email [tranvu@dbr.unibe.ch](mailto:tranvu@dbr.unibe.ch) to make a reservation



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**Next DBMR Research Conference**

**September 5, 2022**  
**Host: Platform for Stem Cell Research and Regenerative Medicine (SCRM)**  
*Speaker and Title tba*



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