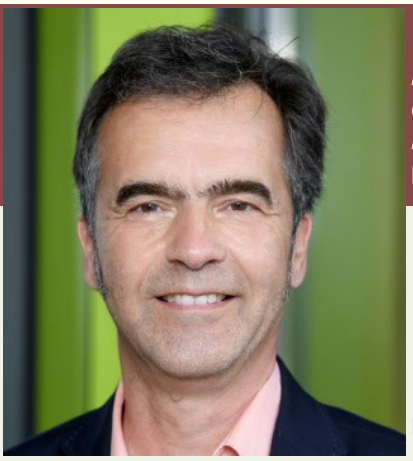
GENETICS, BIOINFORMATICS & SYSTEMS BIOLOGY COLLOQUIUM

THURSDAY, April 15th, 2021
12:00-1:00 PM
Held on Zoom

Click Here for Zoom Link!
Meeting Password: Genomics



Dr. Stefan Mundlos, MD

Full Professor of Human Genetics,
Chair of Institute for Medical Genetics, &
Head of Research Group Development & Disease
Max Planck Institute for Molecular Genetics, Berlin Germany

Decoding the Non-coding - Gene Regulatory Mechanisms Revealed by Rare Disease Genomics

Human Genetics has been extremely successful in identifying disease mutations in rare genetic conditions with Mendelian inheritance. However, even after whole exome sequencing, 50% to 70% of cases with suspected genetic cause remain undiagnosed. We have been focusing on disease mechanisms that are primarily driven by non-coding, regulatory mechanisms. The fine tuned regulation of gene expression is, to a large degree, the product of cis-regulatory elements, so called enhancers. We show that genomic rearrangements, also called structural variations (SVs), can cause re-wiring of enhancer-promoter contacts by disrupting topologically associated domains (TADs). While enhancers probably provide the majority of regulatory information, different types of non-coding RNAs also play important roles. I will present our recent results how mutations in a long non-coding RNA cause a develpomental malformation syndrome and how this lncRNA exerts it effects on the target gene.

Faculty Host: Joseph Gleeson, MD

Presented by:

IGM Institute for Genomic Medicine

UC San Diego Bioinformatics & Systems Biology

For ongoing updates on upcoming lectures:
Visit genomic.weebly.com

Organization Committee: J. Gleeson, F. Furnari, A. Majithia, T. Gaasterland
GBSBC Seminar Coordinators: R. White, S. Orosco

SPECIAL THANKS TO OUR SPONSORS:

illumina®

CCMI Cancer Cell Map Initiative