

CDS SeminarSeries

24.04. **22.05.** 19.06. 17.07.

Bernhard Haubold

MPI for Evolutionary Biology

Do Mouse Genomes Contain Homozygous Lethal Loci?

Recent whole genome sequencing of 16 inbred mouse strains revealed a surprisingly large number of heterozygous positions (Lilue et al., 2018). The authors regard the vast majority of these as artifacts. However, a small fraction of the positions identified might be genuinely heterozygous if homozygosity of either of the two haplotypes is lethal. To identify candidate loci with genuine heterozygosity, we looked for loci in the genome of mouse strain C57BL/6NJ with two criteria: no close paralogs, and least 10 heterozygous positions. We identified regions without close paralogs using our program macle, which rapidly performs a sliding window analysis of local similarity to the rest of the genome (Pirogov et al., 2018). Approximately 500 genes in regions of low self-similarity contained more than 10 heterozygous SNPs. Among the three most heterozygous genes thus identified was Thap1 on chromosome 8. According to prior work, heterozygous knockouts of Thap1 affect motor control and homo-zygous knockouts are embryonic lethal. It would thus be interesting to further investigate, whether selection might after all play a role in keeping this and similar loci heterozygous in C57BL/6NJ.

> 22 May 2019, 17:00 Lecture hall 0.40, Zülpicher Straße 47a

Hosted by Thomas Wiehe (twiehe@uni-koeln.de)

