BACKGROUND SELECTION AND BIASED GENE CONVERSION AFFECT MORE THAN 95% OF THE HUMAN GENOME AND BIAS DEMOGRAPHIC INFERENCES

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Genomic diversity evolves under evolutionary factors such as mutations, selection, gene flow and demography. Across a genome, it varies with recombination in complex ways. For instance, purifying selection is expected to remove diversity with decreasing recombination rate when neutral diversity linked with deleterious variants is removed (background selection, BGS).

In this talk we offer to disentangle the effect on genomic diversity of natural selection from that of demography to properly reconstruct the history of species. By using the high-quality human genomic data, we show that BGS and a second mechanism determined by base composition, termed GC-biased gene conversion (gBGC) together affect as much as 95% of the variants of our genome. Interestingly, synonymous sites and non-transcribed regions are also affected, albeit to different degrees such that their use for demographic inference can lead to strong biases. However, this talk illustrates the possibility to identify a set of SNPs that is mostly unaffected by BGS or gBGC, by conditioning on genomic regions with recombination rates above 1.5 cM/Mb and mutation types (C↔G, A↔T). We will show that this neutral set of variants avoids these biases in the reconstruction of human history. If time permits, we will discuss other intricate links between recombination and evolutionary processes across the genome, especially in regions of very low recombination.