Open Peer Review on Qeios

Runs of Homozygosity

Francisco Ceballos¹

1 University of Witwatersrand

(ROH). Contiguous regions of the genome where an individual is homozygous across all sites. This arises if the haplotypes transmitted from the mother and father are identical, having in turn been inherited from a common ancestor at some point in the past. It is important to note that this notion does not rely on a known pedigree and does not require an (arbitrary) baseline population (the first generation of ancestors or founders in a pedigree). However, ROH in practice are required to have an (arbitrary) inimum size, depending on the density of genotypes available, to distinguish identity-by-descent from chance.