STRUCTURAL DIVERSITY AT 10q11.22

The highly variable 10q11.22 region contains copy-number variable genes linked to intellectual disability, bipolar disorder, and obesity. Fornezza et al. (1) analyzed 64 high-resolution haploid assemblies, revealing that the instability in this region is primarily caused by non-allelic homologous recombination. This process has generated 12 distinct haplotypes, with length differences of up to 2.4 Mb.

The link between this region and the above mentioned diseases raises the question of why natural selection did not act against this variation. Evolutionary analyses propose that the answer may lie in the AGAP gene, whose expansion might have contributed to enhanced synaptic plasticity and improved memory functions in primates and humans.

1. https://www.ncbi.nlm.nih.gov/pubmed/39322278