STRUCTURAL VARIATION: AN UPDATE

Since the first draft of the human genome published in 2001, there have been five new releases; the last being in 2013 (hg38). There is, however, still room for improvement and corrections. The main problems have been the variation that exists among different populations and the difficulty in assembling complex duplicated regions. Recently, two main papers have addressed these issues. One is dedicated to the assembly of regions rich in segmental duplications (Nature Methods). The second is the largest study on the structural variations (deletions, duplications, insertions, and inversions) present in the different human populations (Cell), including those thought to be introgressed from the Neanderthals. It represents a more complete worldwide catalogue of structural variants present in human populations, obtained by studying nearly 1000 individuals from 54 diverse worldwide populations. The implications of these variants in gene expression and gene selection are also considered.
A nice comment/summary can be found in Trends in Genetics.