



**Fig. S5**

Overview of the 187 human cell lines on which fiber FISH and/or Bionano optical mapping was performed. Fiber FISH mapping was carried out on five trios, five non-related probands, and two non related parents of individuals with the 22q11DS. Bionano optical mapping was performed on a total of 154 individuals from 5 super-populations. Both technologies were aligned in cell lines CHM1, GM12878, and three trios with a proband with the 22q11DS. Figures in which results are discussed are depicted at each cell line subset.