

Gene desert area in the 1q21.1 makes the frequent contacts with nucleoli in different human cell lines

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There are large human genome regions where no genes were detected. These regions are called gene deserts (Ovcharenko et al., *Genome Res.* 2005. 15:137-145). There are some indications suggesting that nevertheless these regions could play an important role in regulation of expression of remote genes (Harimendy et al., *Nature.* 2011. 470:264-268). There is about 800-kb long gene desert area in chr1 with coordinates from 142,800 to 143,600 kb (hg19). We analyzed the whole-genome contacts of nucleoli in HEK293T, K562, and hESM01 human cell lines using 4C (circular chromosome conformation capture) approach. The same patterns of very frequent contacts of nucleoli were detected inside this gene desert in all three lines analyzed. No genes in the area are indicated in IGB Browser (hg19). Genome-wide annotations from ENCODE genome segmentations show the presence of repressed chromatin, CTCF sites, small isles of transcribed regions, and weak enhancers in the desert. Surprisingly, we detected strong PARP1 binding sites in the region. No promoters or TSS are present in the desert. Our previous data strongly demonstrated that the contacts of rDNA clusters with different chromosomes are involved in development (Tchurikov et al., *Cells.* 2019). Nucleoli are the largest membrane-less organelles in nuclei. Potentially they could directly spread active or repressed chromatin states at the contacts sites via phase separation mechanisms. The conservation of nucleoli contacts in this gene desert in different cell types strongly suggests the importance of the region in 3D organization of chromosome 1. The study was supported by the grant from Russian Science Foundation No. 21-14-00035.