Correlation between Exons and Dispersed Repetitive DNA Distribution on the Human Genome

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1 Introduction

The human nuclear genome contains a large number of highly repeated DNA sequences. The \(Alu\) sequences are primate specific and are the most abundant family of repeated DNA sequences in the human genome. The human \(Alu\) sequence is approximate 300 bp long [2]. The \(L1\) sequence is a long interspersed nuclear element. \(L1\) is found in other mammals. Although their functions are not yet clear [4], some of them may affect gene functions or cause human diseases [3].

We have identified repeated DNA sequences from human genomic sequences in the region of 3p21.3-p22 and 9q32, both of which are more than 1M bp long. Our statistical analysis shows that the distributions of \(Alus\) and exons have a weak positive correlation and those of \(L1s\) and exons have a weak negative correlation.

2 Method

Genomic sequence data of the human chromosome 3p21.3-p22 and 9q32 as well as cDNA sequences on these regions were obtained by Y.Daigo et al. (unpublished data). The lengths of sequences on the chromosome 3p21.3-p22 and 9q32 are 1.2M bp and 1.0M bp respectively. While the region 3p21.3-p22 contains 14 genes, the region 9q32 has only 3 genes.

Repetitive sequences were identified by the computer program CENSOR [1] with Repbase(Release 5.0). We divided each sequence into non-overlapping 100k bp segment and counted the exon, \(Alu\) and \(L1\).

3 Result & Discussion

To characterize the exon, \(Alu\) and \(L1\) distributions, we compared their densities. The \(Alus\) and exons were more abundant in the 3p21.3-p22 region whereas the \(L1s\) were more abundant in the 9q32 region. To test the significance of these differences, we applied a statistical analysis technique known as two-sample paired \(t\)-test. The significant difference \((p<0.05)\) between the regions was observed in exon and \(Alu\) but not in \(L1\) (Table 1).

Next, densities of exon, \(Alu\) and \(L1\) in each 100k bp segment were plotted in Fig. 1. Due to extremely low gene content, no or little tendency was observed in the region 9q32. In 3p21.3-p22, positive correlation against exon density was observed for \(Alu(r = 0.42)\) and negative correlation against exon density was observed for \(L1 (r = -0.42)\). When the data in both regions were taken into
References

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Acknowledgements

Diamonds are in 1992.

dentify, a correlation coefficient is 0.79. Squares are 38-2-3. A comparison between exon density and exon
correlation is 0.65. B. Correlation between exon density and IV is greater than correlation between
A. Correlation between exon density and exon density. Correlation
P. Figure 1: Correlation between exon density and IV of exons.

Table: Comparison of exon IV and IV density

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<tr>
<th>Exon</th>
<th>IV</th>
<th>d/p (1/2)2</th>
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The base function of gene expression
between exons and introns might have an influence on
the base density of the observed corece
existence of direct relationships but
the base density in high. Although
these results suggest that IV
account, the correlation coefficient between exon and IV density became 0.62 while that between
across, the correlation coefficient dropped from 0.79 (data not shown).

To IIV, the lower correlation coefficient also dropped a little statistically signiﬁcant (r = 0.47). However, when the IVXN size was enlarged up
denovo, the correlation was weakened (r = 0.27).