Texas Tech University. Joint BioMathematics, Statistics and Applied
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Modeling neutral evolution of Alu elements using a branching process

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ABSTRACT. Alu elements occupy about eleven percent of the human genome
and are still growing in copy numbers. Since Alu elements substantially impact the
shape of our genome, there is a need for modeling the amplification, mutation and
selection forces of these elements. Our proposed theoretical neutral model follows a
discrete-time branching process described by Griffiths and Pakes. From this model,
we derive a limit frequency spectrum of the Alu element distribution, which serves as
the theoretical, neutral frequency to which real Alu insertion data can be compared
through statistical goodness of fit tests. Departures from the neutral frequency
spectrum may indicate selection. A comparison of the Alu sequence data, obtained
by courtesy of Dr. Jerzy Jurka, with our model shows that the distributions of Alu
sequences in the AluY family systematically deviate from the expected distribution
derived from the branching process. This observation suggests that Alu sequences
do not evolve neutrally and might be under selection.