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Analysis of ALU repeats for the identification of some polymorphic variants in blood samples

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The human genome contains over one million copies of primate-specific retrotransposed elements called Alu. These elements are a short nucleotide sequence, approximately 300 nucleotides long, with a recognition site for the restriction enzyme AluI. They are non-coding regions of the chromosome and have been found in several chromosomes. The centromeric regions in chromosomes 1, 5, 7, 9, 10, 16 and 19, and distal heterochromatic regions in the long arm of chromosome Y, which generally present some polymorphism, have ALU sequences. From recent data, it has been estimated that Alu elements are currently amplified in the human lineage at a rate of one insertion in every 100–200 births. Therefore, it is possible that mutations in these recent Alu retrotransposition events could cause genetic disorders and contribute to some human diseases like neurofibromatosis, haemophilia and breast cancer. In the present work, the authors apply the Alu technique in ten blood samples for which the indication for cytogenetic study was mental retardation or infertility and which presented cytogenetic polymorphisms, different from the usual cytogenetic ones. Cell culture banding was performed according to routine protocols at the laboratory. Ten GTLmetaphases were analysed from each sample and the polymorphisms were confirmed with CBL banding. Alu sequences in those polymorphisms were investigated. The authors present the results obtained and compare them with data in the literature.