AN ALU BASED TEST FOR HUMAN GENDER DETERMINATION

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While several PCR-based methods are currently available for the determination of gender, there are pitfalls associated with each of them. Amelogenin, the most widely utilized locus for human sex determination, has been reported to yield erroneous results in some cases. This is due to a Y chromosome deletion that is present in 0.6% of the general population and may range as high as 8% among South Asians. We have developed a novel method of gender determination based on the PCR amplification of *Alu* insertions within homologous regions of the X and Y-chromosomes. When *Alu* insertion events become fixed within non-recombining homologous regions of the sex chromosomes, they provide a novel way of differentiation between the sex chromosomes by PCR. We have identified multiple loci that meet this criterion and have designed PCR based assays for two of these loci. The accuracy of the test was verified on a sample of over 500 individuals. These *Alu* insertions provide a simple PCR-base gender assay that is quickly and easily resolved on an agarose gel. In addition, the use of multiple loci will significantly reduce the possibility of misidentification due to local deletions.