

## ALU insertion / deletion polymorphisms in Jatt Sikh population of Jammu, J&K

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### ABSTRACT

Four Alu insertion/deletion polymorphisms (Alu PLAT, Alu ACE, Alu PV92 and Alu APO) were studied in jatt sikh population from Jammu region, J&K. Blood samples were collected from fifty unrelated healthy donors. DNA was isolated and amplified by PCR and subjected to agarose gel electrophoresis. Gene frequencies were calculated and were used to calculate heterozygosity and average heterozygosity. All the four loci are polymorphic in nature and showed high levels of heterozygosity. The average heterozygosity is recorded as 0.4608 to 0.4928.

**Key words:** ALU insertion/deletion polymorphisms; jatt sikh; allele frequency; average heterozygosity.

### INTRODUCTION

The Alu insertions are short interspersed nuclear elements (SINEs) present in the human genome. The insertion of an Alu-element at a particular locus can be regarded as a unique event as once inserted, most Alu elements, being stable genetic markers. Alu deletions are rare and even then the deletion leaves behind a foot print. Alu insertion/deletion polymorphisms have gained importance in the study of genetic structure of human populations as they lack selection pressure. A number of populations in India and other parts of the world are found to be highly polymorphic for these markers. They have been used extensively in recent years to trace human history (Batzer et al. 1996; Stoneking *et al.* 1997; Majumder *et al.* 1999; Watkins et al. 2001). In the present investigation, we present the allele frequencies of four Alu markers in Jatt Sikh population from Jammu, J&K.

### MATERIAL AND METHODS

Jatt Sikh (also Jat Sikh) refers to a sub group of the Sikh ethnoreligious group from the

Indian subcontinent. They form the majority of the Sikh community. About 5 ml of intravenous blood samples were collected in vials containing EDTA. DNA was extracted by using inorganic (salting out) method (Miller et al. 1988). PCR reactions were carried out in a 25 $\mu$ l volume containing 100 ng DNA, 200  $\mu$ M dNTPs, 1.5 mM MgCl<sub>2</sub>, 25 ng each primer, 1.25 U Taq polymerase, 50 mM KCl 10 mM Tris – HCl (pH 8.4). 30 cycles of 94 $^{\circ}$  C for 4 min, 58 $^{\circ}$  C for 1 min, 72 $^{\circ}$  C for 1 min were used for ACE in a thermocycler, 30 cycles of 94 $^{\circ}$  C for 4 min, 54 $^{\circ}$  C for 1 min, 72 $^{\circ}$  C for 1 min were used for PV92, 30 cycles of 94 $^{\circ}$  C for 4 min, 50 $^{\circ}$  C for 1 min, 72 $^{\circ}$  C for 1 min were used for APO and 30 cycles of 94 $^{\circ}$  C for 4 min, 60 $^{\circ}$  C for 1 min, 72 $^{\circ}$  C for 1 min were used for PLAT. PCR products of each marker was visualized in UV– light after separation in a 2% Agarose gel (1.5% for Alu ACE) and ethidium bromide staining.

### RESULTS AND DISCUSSION

All the four loci are found to be polymorphic in nature and showed high levels of heterozygosity. The allele frequencies and heterozygosities for the

insertion/deletion alleles for the four loci studied in the Jatt Sikh population are given in table 1. The heterozygosity is recorded as 0.4608 to 0.4928 which is close to the values that of study conducted by Kaur *et al.* (2002) on jatt Sikhs of Punjab. It may be pertinent to point out here that Majumder *et al.* (1999) reported consistently high levels of average heterozygosity in 14 populations from India ranging from 0.351 to 0.449. The present study population also exhibits high levels of heterozygosity. The Alu APO was found to be most heterozygous in the study conducted.

Finally it may be concluded here with the observation made by Majumder *et al.* (1999) that consistent with the findings of classical markers, the Alu insertion/deletion markers show high levels of genomic diversity in Indian populations. Therefore, further studies are to be taken up on Alu markers in more Indian populations in near future.

**Table 1: Showing heterozygosity and average heterozygosity of four Alu (Alu ACE, Alu APO, Alu PV92 and Alu PLAT) polymorphic loci in 50 individuals of Gujjar population of Jammu region of J&K**

Alu marker	Heterozygosity
Alu ACE	0.4712
Alu APO	0.4928
Alu PV92	0.4608
Alu PLAT	0.4920
Average heterozygosity	0.4805

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