Genetic structure of human populations based on 5 gene loci: A preliminary report from Northern India

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ABSTRACT

The genetics of human populations reveals the evolutionary causes of population structure, population history, human diversity, racial divergence, adaptation, cultural and social behaviors and understanding human adaptation to local environmental constraints. The study aimed to determine the genetic relationship among six human population of Jammu and Kashmir, Northern India. This preliminary effort involved five gene loci of genetic traits, viz. ABO and Rh blood type, phenylthiocarbamide, color blindness and sickle cell trait. A total of 3679 healthy subjects were selected from six Muslim populations viz., Gujjar and Bakarwal (n = 762), Mughal (n = 560), Khan (n = 642), Malik (n = 564), Mir (579) and Syed (n = 572). The allele frequencies and heterozygosity levels show significant differences among six populations ($p < 0.05$). The phylogentic tree revealed that Mughal and Gujjar and Bakarwal (tribal population) are close to each other and differ from other four populations (Malik, Mir, Khan and Syed). Further studies in context genomic diversity, history and origin of these populations will be undertaken in future to get more conclusive proof.

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1. Introduction

Understanding genetic relationship among individuals from different populations is one of the best measures for retrieving ethnic and racial background. The study of genetic polymorphisms has been essential for the analysis of history, origin, divergence, migration and relationships among human populations with much fascinating and improved insights toward clinical and medical perspectives (Fareed and Afzal, 2013).

Ethnic history of India reveals that Indians belong to two different categories: The Dravidians or aborigines and the Aryans or Sanskrit-speaking group (Basu et al., 2003). Empirical studies indicate variable patterns of association among castes and tribes inhabiting wide geographic regions (Cordaux et al., 2003). The Dravidian and Austro-Asiatic speaking tribes inhabiting different geographic regions show wide genetic diversity which supports the hypothesis of their heterogeneous origin (Thangaraj et al., 2005). Whereas geographically proximate tribes and sub-tribes within a region reflect close genetic affinity irrespective of their cultural and linguistic differences (Gaikwad et al., 2006).

The populations of India and other South Asian countries offer great opportunities for studying socio-cultural and genetic variability. North Indian populations and those from Jammu and Kashmir (J&K) have historical, linguistic, cultural, and socio-religious significance for the Indian subcontinent (Fareed et al., 2012). The aims and objectives of the present study were to explore the relationship of six human populations using classical and serological genetic markers.

2. Materials and methods

2.1. Ethics statement

The study was approved by the Institutional Ethics Committee of Jawaharlal Nehru Medical College (JNMC), Aligarh Muslim University, India. We obtained informed written consent from participants, parents or guardians on behalf of minors/children participants involved in our study.

2.2. Geography and study population

Jammu and Kashmir (J&K) is the northernmost state of India, situated between 32.17° and 36.58° North latitude and 37.26° and 80.30° East longitude. To its North, is China and Russian Turkistan and on its East is Chinese Tibet while to the West are the North Western Frontier Provinces of Pakistan. The study was conducted in two twin districts (Rajouri and Poonch) of J&K (Fig. 1).

According to Indian census 2001, the human population of Jammu and Kashmir consists of 66.97% Muslims, 29.63% Hindus, 2.04% Sikhs, 1.12% Buddhists, 0.20% Christians, 0.024 Jains and 0.012% others. Muslims of Jammu and Kashmir belong to various castes such as Syed, Mughal, Malik, Mir, Bhatt, Khan, Lone, Pathan, Qureshi, Sheikh and many
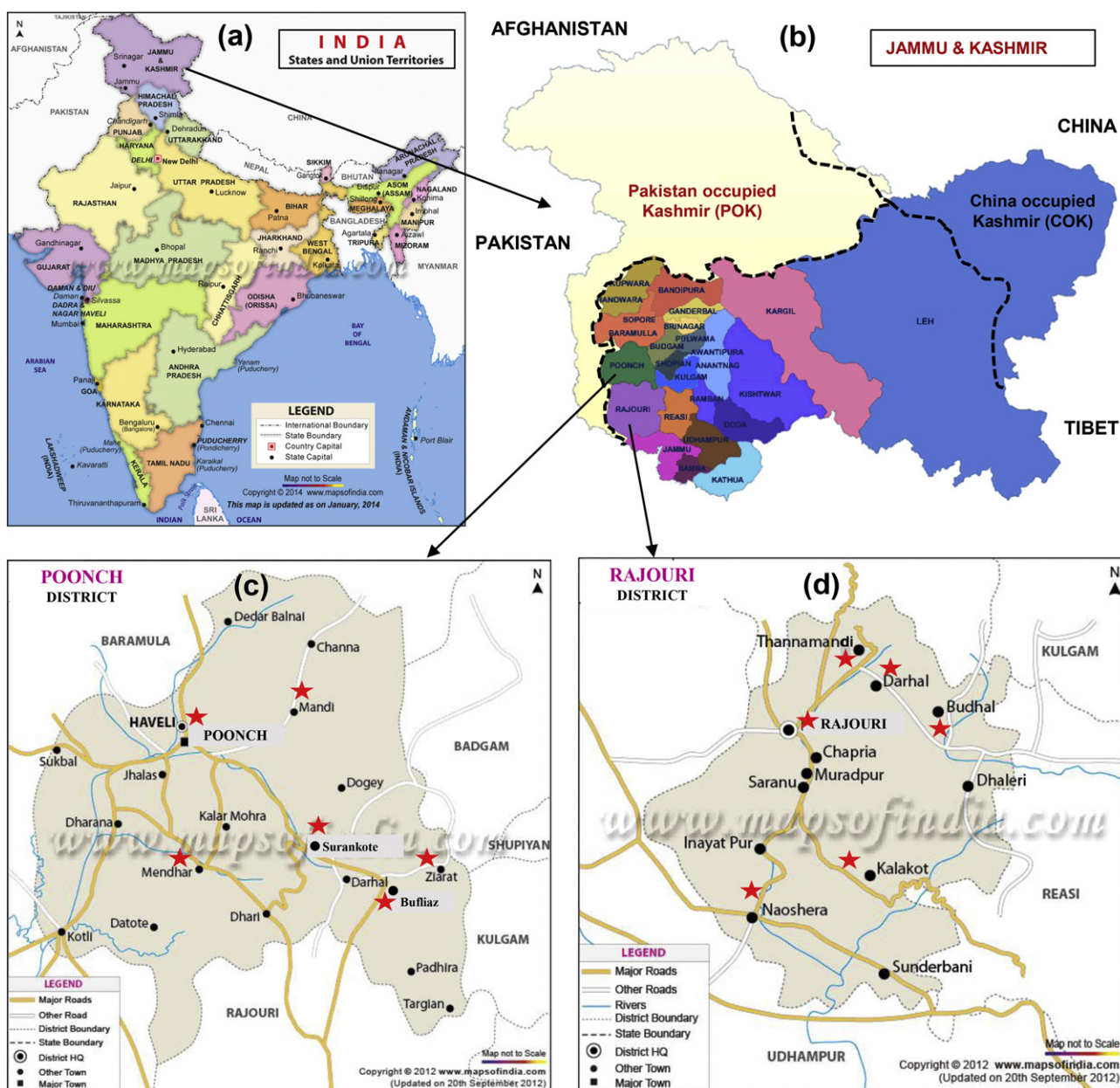
Fig. 1. Map of India (a) and map of Jammu and Kashmir (b). The populations studied from twin districts i.e., Poonch (c) and Rajouri (d). Star represents the regions surveyed.

others based on their occupations. Gujjar and Bakarwal belong to Muslim population and is the major tribe of the state, densely populated in Rajouri and Poonch districts.

The study was conducted during July 2011 through January 2014 and a total of 3679 healthy male and female subjects during household survey were selected from six Muslim populations viz., Gujjar and Bakarwal (n = 762), Mughal (n = 560), Khan (n = 642), Malik (n = 564), Mir (579) and Syed (n = 572). The age of the subjects ranged from 6 to 40 years. Care was taken to avoid the selection more than two subjects of the same sex from each family.

2.3. Procedure

ABO and Rh (anti-D) blood group testing done by using antisera (Tulip Diagnostics; Goa, India) (Fareed et al., 2014). Sickle cell trait was tested by wet sealed method using 2% freshly prepared sodium metabisulphite solution following Daland and Castle (1948) method (Fareed et al., 2016). The color vision impairment was determined using Ishihara color blindness test (38-plate edition) (Fareed et al., 2015). Serial dilution method of phenylthiocarbamide (PTC) was used to assess the taster and non-taster phenotypes (Fareed et al., 2012).

2.3.1. Analysis of gene frequency and genetic diversity

Gene frequency was calculated by using Hardy-Weinberg method ($p^2 + q^2 + 2pq = 1$). The genetic relationship among different population groups was determined by using the measure of genetic distance (D) (Nei, 1972). This 'D' measures the accumulated allele differences per locus or codon difference per unit length of DNA.