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Y- Chromosome Polymorphism in the Mongolian Origin Population of Sikkim-Darjeeling Himalaya

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Abstract:
The gender identity of a person is the end result of genetic, hormonal, morphologic and environmental factor. The study includes an analysis of the variability of length in the Y-chromosome in the normal population of Mongolian origin extending from foothills/plain to altitudes of 8500 ft and above. A wide variability in the length of Y-chromosome has been indicated between the races. Long Y-chromosome is most frequent in the Kirati Complex and the Hill Tribes. Observation was compared with other available reports; also possible mechanism of the Y-chromosome length heteromorphisms and their role in ethnic variation are discussed. Possible involvement of constitutive heterochromatin variability, a class of chromosomal polymorphism, in evolutionary diversification and adaptation has been assessed in the ethnic populations of Mongolian races inhabiting different altitudes and geographical conditions.

Keywords: *Mongolian, constitutive heterochromatin, Y-chromosome, polymorphism, tolerance*

1. Introduction

Genetic polymorphism has been demonstrated in man for many characteristic features including blood groups, serum protein, tissue enzyme and hemoglobin. Before the discovery of banding techniques, the length of the Y-chromosome was known to vary from person to person and from one ethnic group to another (Cohen et al. 1966, Ghosh & Singh 1975, Verma et al. 1984, Potluri et al. 1987 John, 1988, Davila-Rodriguez et al. 2011, Thomas et al. 2015). Some autosomal regions were also found to differ in various racial groups (Lubs and Ruddle 1971, Nand et al.1981, Decsey et al. 2006, Movafaq et al. 2007, Ibraimov et al. 2013). The banding procedure especially C-banding have added a new dimension in this area of cytogenetic research. These have led to the identification of a new class of genetic polymorphism involving constitutive heterochromatin which can be demonstrated especially CNG and CBG methods established by Arrighi and Hsu (1971), Sumner (1972) and Sumner (1982) respectively.

Sexual dimorphism is the end result of genetic and biochemical factors. In mammals the sex chromosome X and Y evolved from a homologous pair of chromosome that differs only at the locus regulating sex determination (Ohno 1979, Jaboling et al. 2003, Jaboling 2008). Intensive study of the problem of the relationship of the Karyotype to the phenotype began after establishing the chromosomal nature of some human diseases in the early 1960s.Even before the advent of banding techniques, heteromorhism of the Y-chromosome of man was known to vary from person to person and from one ethnic group to another (Denver Conference 1960, Oakey and Taylor-Smith 1990). By quantitative evaluation, Beltron et al. (1979), Potluri et al. (1987) and Kostrzewa et al. (2013) observed significant heterogeneity between individuals in the length of the Y-chromosome. The morphology of the Y-chromosome is heritable, is relatively constant in male relatives and exhibits racial variation. Most of this variation is limited to the length of the long arm and its distal heterochromatic region. The long arm of the Y contains repetitive sequence of DNA that are both specific and non-specific to the Y- chromosome. However, the biological significance of the variation of Y-chromosome length is still poorly understood. Previous reports have shown significant intra- as well intergroup variation in the length of the Y-chromosome (Sumi and Tursi 1982, Verma et al. 1984, Agarwala and Datta 1985, Potluri et al. 1987, Chettri 1989, Jobling 2008, Ibraimov et al. 2013, Kostrzewa et al. 2013).

Though the biological and clinical implications of human chromosome polymorphism are poorly understood, yet, it has been suggested that these heteromorphism are limited to certain groups (Craig-Holmes et al.1971, Davila-Rodriguez et al. 2011, Ibraimov et al 2013, 2014). A possible correlation has been demonstrated between variability of the heterochromatic regions to the chromosome 1,9,16 and Y and anthropometric characters. There are repeated emphasis on the selective value of certain chromosomal polymorphism in adaptation of human population to certain extreme environmental conditions and has been contemplated for a comparative analysis of Y- chromosome polymorphism in different racial groups of Mongolian origin for ascertaining selective value, if any, of chromosomal polymorphism in adaptation to varying environmental and geographical conditions. Cytological observations have been carried out on Mongolian origin ethnic groups living under different climate-geographical conditions of Sikkim-Darjeeling Himalaya, which provides a unique opportunity in undertaking a systematic study on the heterochromatin polymorphism and variability in the length of Y-chromosome. The present investigation covers exclusively the Mongolian races including generally hill

inhabitating Kirati Complex comprising of Rai, Mangar, Tamang and Gurung, and the high altitude dwellers consisting of Bhutia and Sherpa and the Rajbansis of plain.

2. Material and Methods

Blood sample of 174 Mongolian origin subjects were collected from different parts of Darjeeling-Sikkim. All were phenotypically normal male, apparently healthy randomly selected and between the ages of 14 to 50. The samples were cultured using the standard micro method (Hungerford 1965). Six to seven day old slides were treated to obtain the differentially stained c-bands using the methods of Arrighi and Hsu (1971) and/or Sumner et al. (1971) with some modifications. Slides were stained with 5% Giemsa and Chromosomes were examined microscopically and photographed. The measurements were done on five good quality metaphase spreads selected according to chromosome size similarity and lack of overlapping of the chromosomes.

3. Results

The ethnic populations studied in the present investigation are the hill inhabiting Hill Tribes and the Kirati Complex and the Rajbansis of plain, all of Mongolian origin. The size of the human Y-chromosome varies considerably, the morphology of the Y is heritable, long arm contains repetitive sequence of DNA and is relatively constant in male relatives and exhibits racial variation. Y-chromosome is one of the smallest among human chromosomes and is mainly concerned with the formation of testis (Lyon 1983, Grumbach 1979, Oakey et al. 1990, Jabling 2008). However, the biological significance of the variations of Y-chromosome length is still poorly understood. Most of this variation is limited to the length of the long arm and its distal heterochromatic segment. This polymorphism in size of the heterochromatic portion, as well as loss of the part of distal euchromatic portion of the long arm, is consistent with normal male sex differentiation and not associated with recognized phenotypic effects. The length of Y-chromosome in males varies with varying frequencies in different population. The incidence of the long Y-chromosome has been noted and observation is presented in Table 1 and Fig. 1. The Hill Tribes, comprising of Sherpas, Lepchas and Bhutia, mostly living in the high hills depicted a moderately high percentage 5.26%) of long Y-chromosome. The Kirati Complex comprising of Rai, Limbu, Tamang, Mangar and Gurung and living in Hills and foothills revealed a relatively high percentage (7.14%) of ling Y-chromosome. The Rajbansis individuals living in the Plains showed a comparatively low percentage (2.50%). The total percentage of long Y-chromosome is 4.26% in the Mongolian origin population of Sikkim-Darjeeling hill.

The frequency of increased (+) and decreased (-) heterochromatin material in the investigated Mongolian racial groups have been depicted Table 2 and Fig. 2.The percentage of increased (+) and decreased (-) heterochromatin in the Hill Tribes is 5.76% and 4.95% respectively. In the Kirati Complex as well the percentage of increased heterochromatin (5.12%) is higher than the decreased heterochromatin (4.86%). However, in the Rajbansis the percentage of decreased heterochromatin is slightly higher (4.61%) than the increased heterochromatin (4.334%). Further analysis reveals that the frequency of the decreased heterochromatin (-) material is 4.95% in the Hill populations and is closely followed by the Plain population (4.61%). However, it appears that the Hill populations have more heterochromatin variant (+) type (5.44%) than the plain population (4.34%).

4. Discussion

The ethnic populations studied in the present investigation are the Hill Tribes, the Kirati Complex and the Rajbansis, all of Mongolian origin. Since the first report of the existence of variability in the length of Y-chromosome among different races (Cohan et al. 1966) a number of investigators have observed significant heterogeneity of this chromosomal parameter between individuals and among different populations (Borvik et al. 1977, Simi and Tursi 1982, Verma et al. 1984 Agarwala and Datta 1985, Berntein et al.1986, Potluri et al. 1987, Chettri 1989, Davila-Rodriguez et al. 2011,Ibraimov et al. 2013, 2014).The present study indicates wide variability in the length of Y-chromosome even between the populations of Mongolian origin. Long Y-chromosome is most frequent in the Kirati Complex followed by the Hill Tribes and the Rajbansis. Previous reports have shown significant variation in the length of Y-chromosome within and between the races (Ghosh and Singh 1975, Cavelli et al. 1984, Monsalve et al. 1980, Jobling and Taylor-Smith 2003, Thomas et al. 2013). The present study not only indicts the existence of interracial heterogeneity in the length of Y-chromosome but also reveals that the high altitude dweller, in general have a relatively high frequency of long Y-chromosome.

A detail analysis of the frequency of constitutive heterochromatin variation reveals variation even between populations. The frequency of heterochromatin variation is comparatively high in the Hill Tribes closely followed by the Kirati Complex and low in the Rajbansis. Earlier studies in the same geographical area involving different ethnic populations have demonstrated significant variation between sub-groups within a population (Agarwala and Datta 1985, Chettri 1989). Further, the present observation not only established the existence of interracial heterogeneity of chromosomal heterochromatin but also reveals that the hill dwellers, namely, the Hill tribes and the Kirati Complex exhibit, in general, a relatively high frequency of constitutive heterochromatin polymorphism. The possible existence of interracial heterogeneity of chromosomal polymorphism has been examined by some investigators in few populations (Oakey and Taylor-Smith 1990, Lillian et al. 2005, Decsey et al. 2006, Davila-Rodriguez et al. 2011, Ibraimov et al. 2013). A Comparative study on heterochromatin polymorphism made of Central Asia by Ibraimov et al. (1982, 1986, and 2014) in some highland and steppe Mongolian. The significant reduction noted in the amount of heterochromatin in all the three representatives of highland populations as compared to the steppe populations of Central Asia has been demonstrated by them as an adaptation of the highland populations of extreme environmental conditions, in particular to the high altitude. The results of the previous and present investigations demonstrate that a considerable amount of variation and changed in the heterochromatin can be tolerated with no apparent harmful effect on the phenotype. The present investigation further reveals that the degree of tolerance of the genome for

changes involving heterochromatin varies between the populations. The hill populations, by and large, livings at high altitude belts with low temperature are more tolerant to heterochromatin variability.

5. Conclusion

The present investigation covers exclusively the Mongolian origin races generally hill inhabitating the Hill Tribes, the Kirati Complex and the Rajbansis of plains. The variability of length in the Y-chromosome in the normal population of diverse ecological conditions has been suggested amongst different populations. Present observation also depicts a wide variability on the length of Y-chromosome between the populations of Mongolian origin population of this geographic region. Long Y-chromosome is most frequent in the Kirati Complex and the Hill Tribes. A comparative analysis of the frequency of increased and decreased heterochromatin material in the populations shows a tendency towards increased in the Hill Tribes and the Kirati Complex, while, a reverse situation has been noted in the Rajbansis. The significant increase in the amount of heterochromatin in the hill population living at high altitude with low temperature suggest the possible selective value of chromosomal heterochromatin material in the adaptation of human population to extreme environmental conditions.

Population	No. of chromosome Examined	Variants No. %	Total %age of variants
Hill Tribes	38	+ + 5.26	4.26
Kirati Complex	56	+++ 7.14	
Rajbansis	80	++ 2.50	

Table 1: Frequency of long Y-chromosome recorded in Mongolian origin racial groups
+ = Long Y-chromosome

Population	Heterochromatin * Increased Decreased
Hill Tribes	5.76% 4.95 %
Kirati Complex	5.12 %4.86 %
Rajbansis	4.34 %4.61 %

Table 2: Increased and decreased heterochromatin in Mongolian origin racial groups
**Sum total of 1, 9, 16 and Y- chromosome*

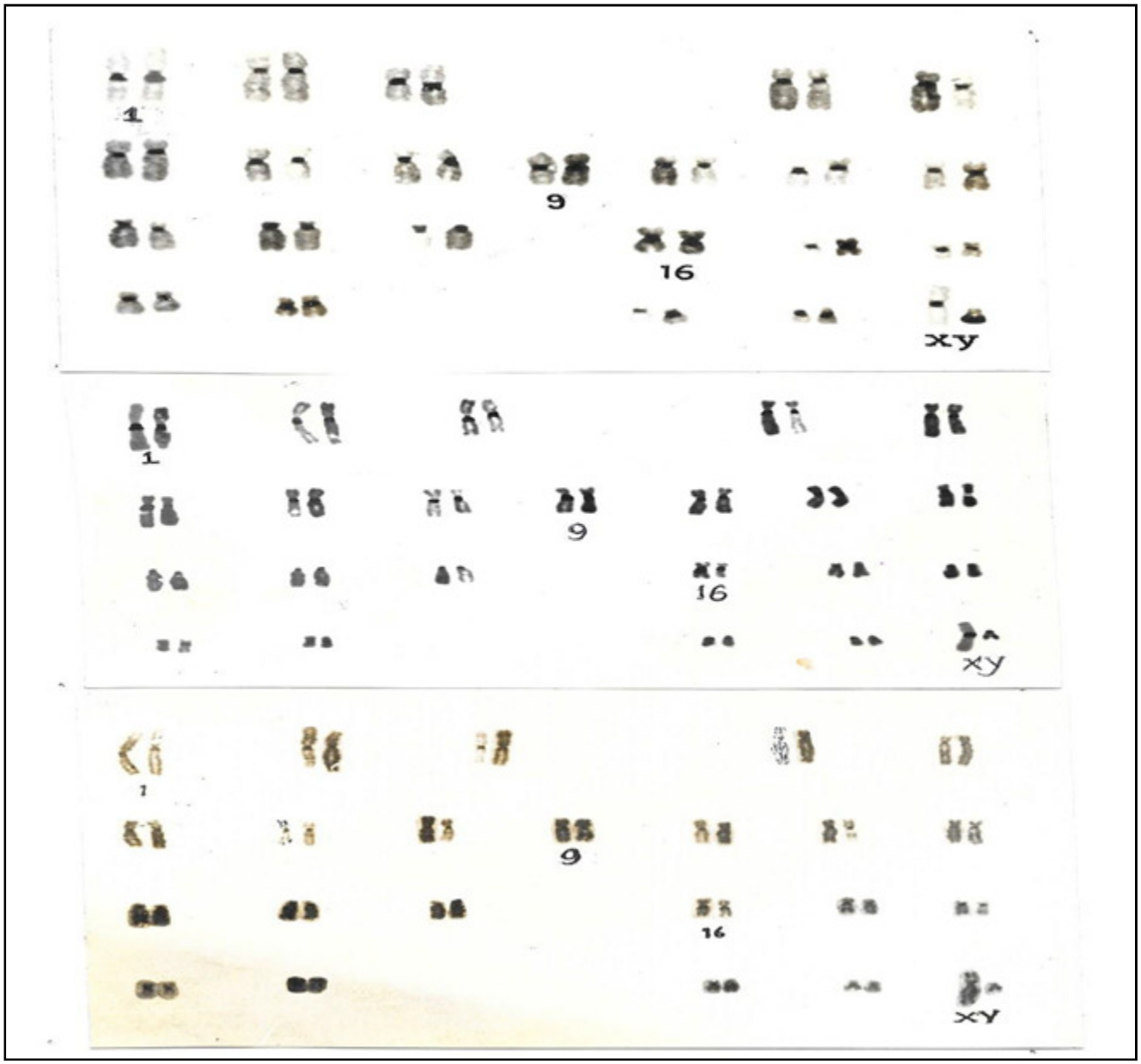

Figure 1: Karyotype showing heteormorphic constitutive heterochromatin in chromosomes 1,9,16 and y recorded in three ethnic Mongolian origin groups.

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