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Prevalence and Gene Frequency of Colour Blind Disease among Three Tribal Demes of Kishanganj District, Bihar, India

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Abstract

Aim and Objective : The gene for colour blindness (Xq28) is commonly used by different geneticist as genetic marker for the study of variation in population. The present paper based on the gene frequency of CB and its analysis for the validity of relax a selection hypothesis in the three tribal population selected from district Kishanganj, Bihar, India. **Subject and Method:** The cluster stratified random sampling method was used for the study of prevalence of colour blindness gene. The three tribal demes Santhal, Munda and Oraon were selected and pooled sample sized was 322. The screening was based on Ishihara plates number 26,28,30,32,34 and 36 as the subjects were mostly illiterate. The calculation of allelic frequency was fundamentally based on Hardy and Weinberg Law with proper consideration of homozygous female and hemizygous male. **Finding and Disscussion:** The calculated allelic frequency forcolour blind gene (q) is 0.17 for female, 0.06 for male and 0.13 for combined population in Santhal whereas in Munda population the value is 0.19 for female, 0.04 for male and 0.14 for combined population. A high percentage values are observed for female carrier both in Santhal (28.18%) and Munda (31.54%). The χ^2 goodness of fit test value of phenotypic and genotypic variation in tribal demes are non-significant suggested that gene is fixed in population. The colour blind is absent from Oraonpopulation. These variations are associated with evolutionary factor like inbreeding in Santhal and Munda but in Oraon the probable cause is genetic drift. **Conclusion:** The findings provide a question mark on the universality of"relaxed selection hypothesis" as reported by some other workers alsothe high percentage of carrier female gives more chance for the appearance of disease in male of next generation due to criss-cross inheritance pattern. Hence, marriage counseling is recommended in these populations.

Introduction

The colour vision is evolutionarily an advanced character based on three classes of cones photoreceptors responsible for red, green and blue colour

vision ie trichromatic colour vision (Jacobs G.H.,1993). The human who represent dichromatic nature and do not able to differentiate the three colours are referred as colourblinds. It is a sex linked disease and associated with X chromosomes (Xq28), however the gene for blue pigment vision is located on chromosome 7. The disease is caused by recessive gene, present in homozygous conditions in female and hemizygous condition in male. It has been suggested that Opsine has array of two genesreferred as L and M. The close proximity of these two genes and their close sequence homology are responsible for miss-pairing during meiosis followed by intergenic and intragenic crossing over of genes. The products of these molecular events of L and M genes are responsible for high frequency of red green colour vision deficiency (Nathans et al. 1986;Feilet al.1990;Jorgensen et al.1990;Neitz and Neitz 2000).

The colour blindness gene is widely used as genetic marker for fundamental understanding of human variation. It has been known for a long time that males have much higher frequency of colour blindness than female (Wale and Oriowo 2008;Shah et al. 2013;Reddy et al.2017). On the basis of social stratification its frequency is lowest in hunting and gathering societies followed by farming societies with the highest frequency in the industrial society (Rowe and Stein ,1978;Naidu and Veeraju ,1978; Bhasin et al. 1992;Bhasinet al. 1994). The variation in prevalence in different strata has been explained on the basis of "selection relaxation hypothesis" (Post,1962; Piokford,1962;Post,1971;Kapoor et al.,1983;Ninhari 1993). The other hypothesis proposed is positive selection of mutant colour blindness gene in traditional culture (Adam, 1969). A critical review is also present on the basis of available data on the frequency of colour blindness in various ethnic groups and concluded that the Post – Pickard hypothesis of relaxed natural selection is not accepted universally (Adam,1969;Adam,1985).

The present paper is based on random sampling in tribal population of Kishanganj district of Bihar (India). The present study included three tribesSanthal, Munda and Oraon and pooled sapmle size was 322. The paper is presenting the prevalence rate of colour blindness in selected area and discussed on the probable evolutionary factors responsible for variation in allelic frequency of colour blind gene in selected population.

Subject and Methods

Study Area - In the present study three blocks Bahadurganj, Pothia and Kishanganj of this district were selected. Geographically it extends between $25^0$21' to $26^0$30' north latitude and between $87^0$7' to $88^0$19' east longitude. The boundary of district is shared with Nepal and Darjeeling of West Bengal in North, Araria in West, Purnea in South West and Uttar Dinajpurof West Bengal in East.

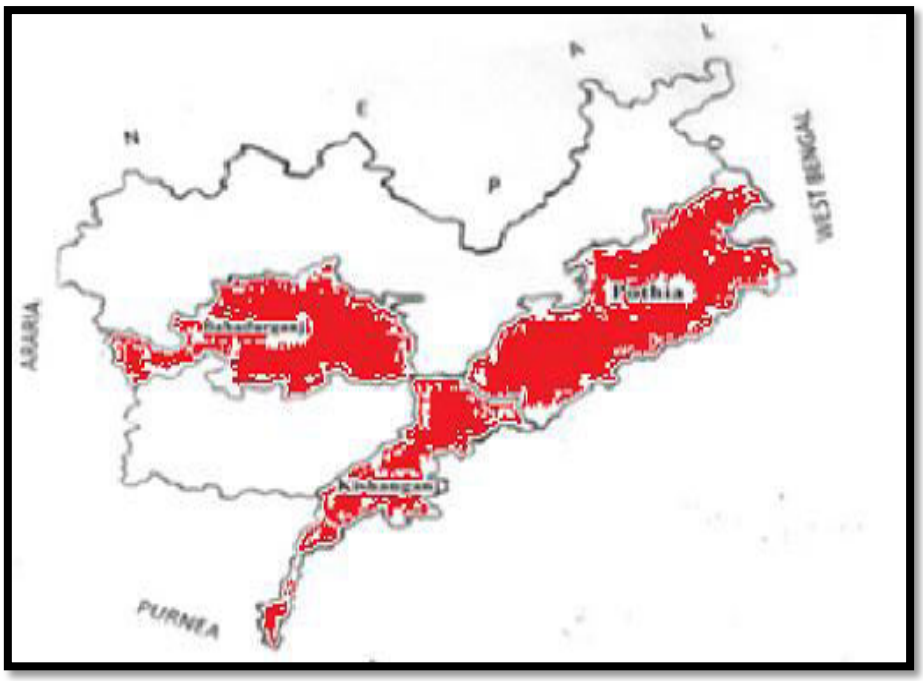


Fig – Represents Kishanganj district (Bihar, India) and coloured area included in present study.

Subject and Sampling – The tribal population were selected for the study. The district has 1690948 populations as per census 2011; among them 6.69% belongs to schedule caste and 3.8% belongs to schedule tribe. The three tribal demes Santhal, Oraon and Mundawere included in the present study. They were migrated to the area from Chhotanagpur of Jharkhand and West Bengal in200 – 250 years ago and were served as agriculture laborers by European land owners and local Zamindars. They are endogamous however Santhals are exogamous on the basis of their sub clan or *gotras*.
The cluster stratified random sampling was used carried from June 2020 to Dec 2020. The clusters were selected on the basis of secondary data of census. The sample size of Santhal, Oraon and Munda was 222, 50 and 50 respectively. The more number of Santhal in sample is due to its highest percentage (75%) in total tribal population residing in this area. The Oraon and Munda are present in small clusters in some villages of this districts.

Screening for Colour Blindness – Three details of random selected persons like name, age, sex etc were recorded in predetermined questionnaire. The consent of person was obtained before screening test of colour blindness. The screening was based on Ishihara plates (Ishihara J. H., 1959).The plates were placed at 75 cm from the subject and tilted at right angle to the line of vision in sufficient light. As the subjects were mostly illiterate, the Ishihara plate numbers 26,28,30,32,34 and 36 were used for the screening of colour blind persons.

Calculation and Statistical Methods – The basic formula for the calculation of allelic frequency is based on Hardy Weinberg Law (HWL). But due to its hemizygous nature in male, the formula for calculation of allelic frequency in total population and male is derived accordingly. The female follows $p^2+2pq+q^2 = 1$ of HWL and male follows $p+q = 1$. The allelic frequency in male, female and total population was calculated by derived formula, where Xc represented gene for colour blind and its homology X for normal gene.

A. For Male

$$Xc(q)=\frac{\%\ of\ colour\ blindness}{100} \qquad X(p)= 1 - Xc$$

B. For Female

$$Xc(q)= \sqrt{\frac{\%\ of\ colourblindness}{100}} \quad X(p)= 1- Xc$$

C. For-Combined $Xc(q) = \frac{1}{3}\ Xc\ (Male) + \frac{2}{3}\ Xc\ (Female)$ $X(p) = 1- Xc$

The chi square values were calculated for the determination of significance level (Srivastava N., 2010). All calculations were carried in MS Excel of Microsoft.

Result and Discussion

The prevalence of colour blindness in pooled data of tribes was observed 4.38% for male, 2.7% for female and 3.42% for combined population. If each deme was considered independently, Santhal represented 6.02, 2.88 and 4.05 percentage prevalence for male, female and combined populations respectively whereas in Munda population the values were 4.17%, 3.85% and 4% for male, female and combined populations respectively. The colour blindness gene was not observed in Oraon (Table–1). The calculated allelic frequency in pooled population as well as in both Santhsal and Munda demes represent variation. The frequency of colour blind gene (q) in Santhal is 6.03% for male, 16.97% for female and 13.32% for combined population. In Munda the frequencies are 4.17%, 19.62% and 14.47% for male, female and combined populations respectively (Table – 2). The χ^2 values of demes for male (0.00259, df =2), female (0.7009, df=2) and combined population (2.68168, df=2) are representing non-significant variations.

TABLE:1: REPRESENTS ACTUAL AND PERCENTAGE PREVALENCE OF COLOUR BLINDNESS IN ALL THE THREE DEMES SELECTED FOR THE STUDY FROM KISHANGANJ DISTRICT, BIHAR										
DEMES	N	MALE			FEMALE			COMBINED		
		NORMAL	CB	TOTAL	NORMAL	CB	TOTAL	NORMAL	CB	TOTAL
SANTHAL	222	78	5	83	135	4	139	213	9	222
		93.97	6.03	100	97.12	2.88	100	95.95	4.05	100
ORAON	50	30	0	30	20	0	20	50	0	50
		100	0	100	100	0	100	100	0	100
MUNDA	50	23	1	24	25	1	26	48	2	50
		95.83	4.17	100	96.15	3.85	100	96	4	100
POOLED	322	131	6	137	180	5	185	311	11	322
		95.62	4.38	100	97.29	2.71	100	96.58	3.48	100

Taple:2: Represents Allelic Frequency of Colourblind Genes among Three Demes of Tribal Population Residing in Kishanganj District of Bihar

Demes	Male		Female		Combined	
	p	Q	p	q	p	q
Santhal	0.9397	0.0603	0.8303	0.1697	0.8668	0.1332
Oraon	1	0	1	0	1	0
Munda	0.9583	0.0417	0.8038	0.1962	0.8553	0.1447
Pooled	0.9562	0.0438	0.8354	0.1646	0.8757	0.1243

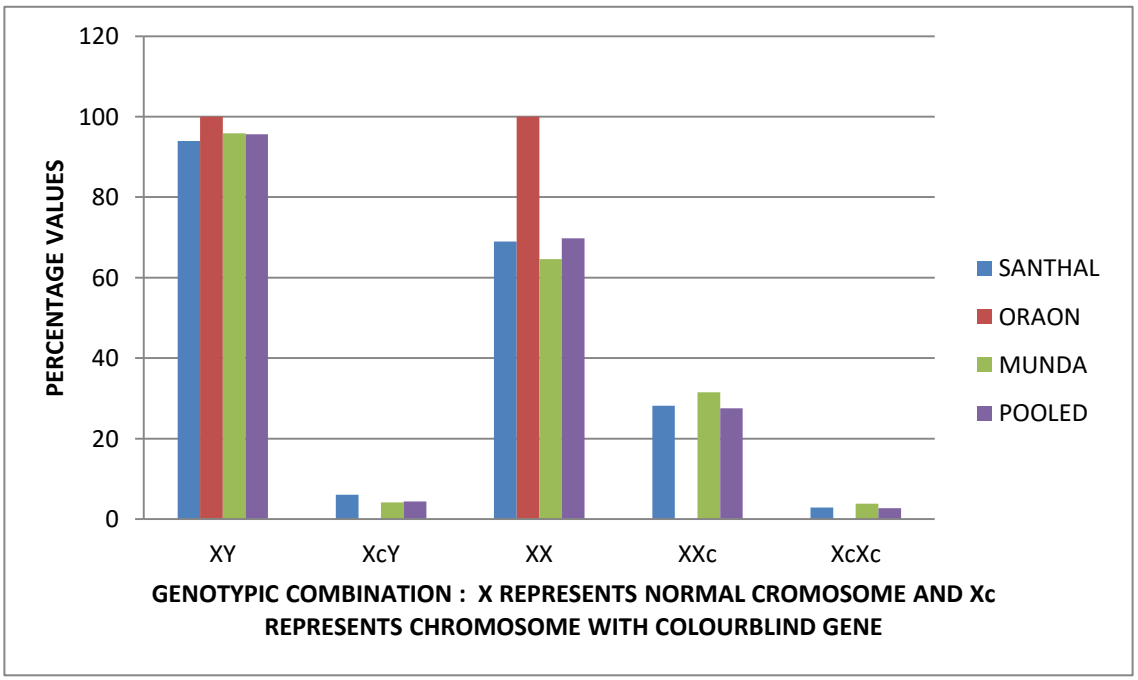
The genotypic prevalence of colour blindness is 4.38% for male and 2.71% for homozygous recessive female in pooled population.The heterozygous or carrier female is observed as 27.51% in pooled population. Santhal represents 6.03% male as diseased where as in female 2.88% is homozygous recessive and 28.18% is heterozygous or carrier. The similar nature is represented by Munda also. The 4.17% male has colour blind gene where as in female 3.85% are homozygous recessive and 31.54% are heterozygous or carrier for disease (Table – 3). The χ^2 goodness of fit test values represented non - significant variations (Santhalχ^2 = 1.0648 at df 1; Munda χ^2 = 0.8886 at df 1 and for pooled population χ^2 = 0.8728 at df 1).

Table:3: Represents Genoypic Frequency (Percentage Values) of Colourblind Gene in Both Sexs and Combind Polulation in All Three Demes of Tribal Population

Demes	Male		Female			Total
	XY	XcY	XX	XXc	XcXc	
Santhal	93.97	6.03	68.94	28.18	2.88	100
Oraon	100	0	100	0	0	100
Munda	95.83	4.17	64.61	31.54	3.85	100
Pooled	95.62	4.38	69.78	27.51	2.71	100

The present finding in Santhal and Munda represent similarity with the reporting of earlier works on both tribal and non tribal populations. The higher prevalence has been also reported from India (4.9%), Malaya (4.9%), China (5.4%) and Singapore (5.6%) (Shah,2013;Ninhari,1993; Basu et al., 1958;Basu,1964;). But generally low frequency of colour blindness is reported from tribal populations which followed the hypothesis of relaxed selection (Bhasin et al. 1992; Bhasinet al. 1994;Post,1962; Piokford, 1963;Post,1971;Kapoor et al.1983; Dutta1966). The hypothesis suggested that the accuracy of colour vision is related with socio-occupational factors. Initially the society was hunter – gatherer, they had more need of colour discrimination and condition was created for related selection. The works on nomadic population are also supported this hypothesis (Ninhari,1993;Bajpai,2017;Malhotra,2017; Malhotrs et al.1974). The high frequency of colour blindness gene in both Santhal and Munda of present area does not follow relaxed selection hypothesis. It is supposed that the high frequency is due to marriage among individuals of small population localised in present geographical area. Hence the evolutionary factor responsible for more frequency of colour blind gene is considered as inbreeding in present study as consanguineous marriage is considered an important factor for more prevalence of colour blindness (Shah et al.2013). The other remarkable finding of present study is absence of colour blind gene from Oraon in same area. In the present geographical area Oraon and Munda are very small in number and residing in 1-2 villages in Pothia block. It is supposed that the gene frequency become zero due to founder effect or genetic drift or the gene is eliminated from population to increase the survivorship benefits. However, more extensive study is necessary for the final conclusion.

Graph :1: Represents Genotypic Allelic Frequency of Colourblind Gene in All Thre Trbal Demes of Kishanganj



The colour blindness is a sex linked disease and carried in population by female through criss cross pattern of inheritance. The present study represents that the 28.18% female of Santhal and 31.54% female of Munda are carrier for the gene (Graph – 1). The statistically non – significant variation (in goodness of fit χ^2 test) suggested that the gene is naturally fixed in both populations. The present study suggests a mass screening drive and pedigree study in population reside in present area as well as marriage counselling in the population to decrease the colour blind gene frequency in the Santhal and Mundapopulation.

Conflict : No Conflict
Financial Assistance : No financial assistance

Contribution: NS was designed the study; A, SK and NK were collected literature, sample and tabulated the data; NS and RS were calculated gene frequencies and statistically analysed the data; NS, RK, A and AKB were prepared manuscript.

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References

1. Adam A.1969.Further quarry of colour blindness and natural selection. Soc. Biol. 16: 197 – 202.
2. Adam A.1985. Colour blindness in man: A call for re- examination of the selection – relaxation theory. In Y. R. Ahujha and J. M. Neel (Ed) "Genetic micro-differentiation in human and other animal populations." Proc. Int. Sym. Hydrabad (1983), Indian Anthropological Society, Delhi, pp. 181 – 194.
3. Bhasin M.K, Walter H and Danker – Hopfe H.1992. The distribution of genetical, morphological and behavioural traits among the people of Indian region. Kamla Raj Enterprises, Delhi.
4. Bhasin M.K, Walter H and Danker – Hopfe H.1994.People of India : An investigation of biological variability in ecological, geographical, ethno-economic and linguistic groups.Kamla Raj Enterprises, Delhi.
5. Basu A. C. 1964. The frequency of colour blindness in some population group of Maharashtra (India). Ann. Hum. Genet. London. 28: 129 – 131.
6. Bajpai V.B. 2017. Incidence of colour blindness among four verna population of Hindu religion of Lucknow district, Uttar Pradesh, India. Int. J. Humani. So. Sc., 7(1): 99 – 105.
7. Basu R. N, Kundu M. K and Boss J. C.1958. Colour blindness with sex linked heredity. J. Ind. Med. Assoc. 30 (1): 11 – 14.
8. Dutta P.C.1966. A review of the inherited defective colour vision variability and selection – relaxation among Indians. Acta. Genet. Stat. Med. 16: 327 – 339.
9. Feil R, Auboung P, Heilig R and Mandal J.L.1990. A 195 kb cosmid walk encompassing the human Xq28 colour vision pigment genes. Genomics. 6: 367 – 373.
10. Ishihara J. H.1993.Test for colour blindness. KanicharaShuppan Co. Ltd. Tokyo. 1959.
11. Jacobs G.H. The distribution and nature of colour vision among the mammals. Biol. Rev. Comb. Philos. Soc. 64: 413 – 471.
12. Jorgensen A.I, Deeb S.S and Motulsky A.G.1990. Molecular genetics of X chromosome linked colour vision among population of African and Japanese ancestry : high frequency of a shortened red pigment among Afro – Americans. Proc. Natl. Acad. Sci, USA. 87: 6512 – 6516.
13. Kapoor A. K, Satwanti and Kshatriye, G. K.1983.Incidenc of colour blindness in different Indian populations further examples of selection relaxation hypothesis. Acta. Anthropogenetica, 7: 1 – 14.

14. Malhotra K.C.2014. Natural selection and colour blindness; Fresh data an Indian caste. Genet. Res. 31: 203.
15. Malhotrs K.C, Muttalik G.S, Bhona B.W, Cate S.L and Fulmali P.M.1974. The incidence of colour blindness among four endogamous nomadic groups : An example of natural selection. Heredity. 32: 145 – 149.
16. Ninhari S.C.1993. Colour blindness and natural selection : Studies in four nomadic tribal groups from Andhra Pradesh, India. Anthrop. Anz, 51(2): 169 – 171.
17. Neitz M and Neitz J.2000. Molecular genetics of colour vision defects.Arch. Ophthalmo. 118: 691 – 700.
18. Naidu J.M and Veeraju.1978. The incidence of colour blindness among tribal population of Andhra Pradesh. Ann. Hum. Biol. 5: 2
19. Nathans J, Piantanida T.P, Eddy R. L, Shows T.B and Hogness D.S.1986. Molecular genetics of inherited variations in human colour vision.Science. 232: 203 – 210.
20. Neel J. B and Post R. H.1963. Transitory "positive" selection for colour blindness?.Eugen. Quart. 10; 33 – 35.
21. Post R H.1962. Population differences in red and green colour vision deficiency: A review and a query on selection – relaxation. Eugen. Quaqrt, 9: 131 – 146.
22. Piokford R. W. 1963.Natural selection and colour blindness. Eugen. Rev. 15 : 97 – 101.
23. Post R. H.1971. Possible case of relaxed selection in civilized population. Human Genetics, 13: 253 – 284.
24. Reddy A.V, Babu G.R and Prasad K.V.2017. Prevalence of colour blindness in school children in Guntur city, Andhra Pradesh.Int. J. Contemp. Med. Res. 17: 2266 – 2268.
25. Shah A. 2013.Hussain R, Fareed M, Afzal M. Prevalence of red green colour vision defect among Muslim males and females of Manipur, India. Iran. J. Public. Health. 42: 16 – 24.
26. Srivastava N. 2010.Introductory Biometry : Statistic and research Methodology. Pointer Publishers. Jaipur, Rajasthan, India, pp – 222 – 237..
27. Wale H and Oriowo O.M.2008. Colour vision screening among Saudi Arabian children. South. aft. Optom. Abdullah. Z. Alotaibi. 67: 56 – 61.