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Colour Vision Deficiency and Consanguinity in Pakistani Pukhtoon Population

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Abstract

Background: Color vision deficiency is an X-linked recessive condition. This is more common among males as compared to females. This inherit from maternal grandfather to grandson usually. This study was aimed to find prevalence of color vision deficiency and consanguinity in Pukhtoon population of Pakistan.

Methods: Three hundred and fourteen (314) high school individuals of both genders were screened for color vision deficiency using pseudoisochromatic Ishihara Plates. Consanguinity of parents were also recorded. Frequency of color vision deficiency over entire sample was used to calculate prevalence. Chi-square is used to see association between color vision deficiency and consanguinity.

Result: Prevalence of CVD among males and females were 5.32% and 2.06%. Prevalence of consanguinity among parents of participants were 73.9%.

Conclusion: Study showed prevalence of CVD among males as comparable to already reported but among females the rate is relatively higher. Rate of consanguinity among study population is also higher than previously reported ones.

Introduction

Colour vision is an evolutionary trait among primates. This trait shows many polymorphism like monochromacy, dichromacy, trichromacy and tetrachromacy in different organisms based on their evolution. In humans, normal colour vision is trichromatic although dichromacy is also common. Anomalous trichromacy is also present and known as deuteranomaly and protanomaly. Dichromacy also has two forms, i.e., deuteranopia and protanopia. Hybrid *L/M opsin* gene is responsible for colour vision in humans. *L/M opsin* gene is present on X chromosome X and is a recessive trait. All types of variation in *L/M opsins* express as anomalous trichromacy or dichromacy and commonly known as red-green colour vision deficiencies. [1] In human males, prevalence of colour vision deficiencies is 3 to 8% in different populations. On the other hand, prevalence of colour vision deficiencies in females among different populations is 0.2 to 1.7% [2].

Epidemiological studies are of key importance in understanding of genetic trait in any population [3]. In an earlier work, we described colour vision deficiencies in the Punjabi population of Pakistan [2]. Being an evolutionary trait, colour vision has evolved differently in different species based on their subsistence. Similarly, it may had evolved differently in different human populations living in different subsistence. Current paper aims at presence of colour vision deficiency trait among Pukhtoon population of Pakistan. Pukhtoon is an ethnic group largely found in Pukhtunis of Central Asia, Afghanistan and Khyber Pakhtunkhwa (KPK) Province of Pakistan [4]. Terms of Pukhtoon, Pushtoons, and Afghans are synonymous in their identity. Exact origin of Pukhtoon is unknown, however, many claims to be descendants of Lost Tribes of Israel. A telltale makes Pukhtoons descendant of Afghana, grandson of King Saul of Israel. Similarly, sub-ethnic group of Pukhtoons, Yousufzai claims to be descendants of Joseph [5,6]. Another ethnic group living together with Pukhtoons is Syed. Syed are descendants of old Arab tribe, Quresh. This study includes Yousufzai Pukhtoons and Syed living in Buner area of KPK province of Pakistan and aimed to identify colour vision deficient families and prevalence of consanguinity among them.

Methods

This was a cross sectional study conducted in high school children of Buner, Khyber Pakhtunkhwa province of Pakistan. Any individual with reported eye disease was excluded from the study. Simple random sampling was used and 314 individuals (169 males and 145 females) were screened. Pseudoisochromatic Ishihara Plates (36 plates edition) were used to identify colour vision deficient individuals in well day-lighted classrooms. A pre-designed questionnaire was used to collect data. History of consanguinity of parents was also recorded. Frequency of colour vision deficiency and consanguinity was used in order to calculate prevalence. SPSS (version 22.0) was used for statistical analysis. Study was approved by Ethical Review Committee of University of Health Sciences Lahore, Pakistan. An informed consent was obtained from each participant prior to examination.

Results

Out of 314 individuals screened, 309 were Yousufzai Pukhtoons, 4 were Syed and 1 was of non-Pukhtoon origin. Gender based division can be seen in Table 1. Out of these individuals, 12 were found to be colour vision deficient on Pseudoisochromatic Ishihara Plates. Among males, 9 were colour vision deficient thus reporting prevalence as 5.32%. While among females, 3 were found to be colour vision deficient thus a high prevalence as 2.06%.

Colour Vision Deficiency	Gender of individuals	
	Male	Female
Normal	160	142
Colour Vision Deficient	9	3
Total	169	145

Table 1: Colour Vision Deficiency and Gender Distribution

Consanguinity of parents of study individuals was also recorded and is reported in Table 2. 30.3% individuals showed their parents were married to first cousins while 19.8% to their maternal first cousins so increasing chances of inheriting X-linked traits. Only 26.1% parents married outside of their tribe, showing a very high rate of consanguinity, i.e., 73.9%. However, among 12 individuals who were CVD, parents of 7 individuals were cousins, i.e., 58.3% consanguinity among parents of CVDs.

Father married to		Frequency	Percent
Maternal First Cousin	Khalazad (Aunt's daughter)	30	9.6
	Mamoonzad (Uncle's daughter)	32	10.2
Paternal First Cousin	Phoophizad (Aunt's daughter)	9	2.9
	Chachazad (Uncle's daughter)	24	7.6
	Second Cousin	10	3.2
	Distant Blood Relation	51	16.2
Same sub-caste	Bradri	24	7.6
	Same Caste	52	16.6
	Different Caste	82	26.1
	Total	314	100.0

Table 2: Consanguinity among parents of Study individuals

Discussion

Current study reported prevalence of red-green color vision deficiency as 5.32% among males while our previous study reported 5.26%. Thus both studies revealed almost the similar prevalence so closest among two different Pakistani populations [2]. Other studies in Pakistan reported 3 to 8% in Punjab and Sindh areas [2,7-9].

However, prevalence among female participants is found too high than expected, i.e., 2.06%. Being an X-linked recessive trait, expected prevalence is 0.5% as reported in many studies including our own previous study [2]. However, availability of small sample size of females may be one of the reasons of bias in study population. Also in study area of KPK, all the maternal and paternal families live in the same area usually thus maybe population bottleneck like phenomenon. This may be another reason of higher rate of CVD trait among

participants. Another reason is higher rate of consanguinity. Moreover, color vision is an evolutionary trait and useful in foraging thus a further study involving L/M opsins sequencing followed by analysis by multiple population genetics aspects is necessary to make an interpretation.

Considering consanguinity reported by this study, this is the highest reported rate of consanguinity among Pukhtoon population, i.e., 73.9%. This rate is based on the population of Buner district in Khyber Pukhtoonkhwa. Already reported consanguinity rates were based on studies done in Bajur and Malakand districts. etc., and were 58.3% and 66.4% respectively [10,11]. However, these rates of consanguinity among Pukhtoon populations are relatively lower than other Pakistani populations. Like in Pakistani Punjabi population, the rate of consanguinity is higher than pakhtoon populations, i.e., 80% and 84.82% [2,12]. Either it is Punjabi or Pukhtoon population, consanguinity is almost always cultural in Pakistani population [13]. This cultural consanguinity is not only present in different Pakistani populations but also in neighbor countries including Afghanistan and India etc. Currently, there are many campaigns by Pakistani public health institutions to avoid consanguinity especially among families reported with clinically significant recessive diseases.

Colour vision deficiency among males of Pukhtoon population is representative as other Pakistani populations. But colour vision deficiency among females is relatively high probably due to consanguinity and higher consanguinity rate.

Authors' Contribution

MSZ: Research idea, data collection and manuscript writing
MSA: Research idea, study design and statistical analysis
ARH: Data entry
MA: Research idea, manuscript writing

Competing Interest

The authors declare that they have no competing interests.

References

1. Kawamura S, Melin AD (2017) Evolution of genes for color vision and the chemical senses in primates. In: Saitou N, editor. Evolution of the Human Genome I: The Genome and Genes. Tokyo, Japan: Springer Japan. pp. 181-216.
2. Akhtar MS, Aslamkhan M, Zar MS, Hanif A, Haris AR. Dichromacy: Colour Vision Impairment and Consanguinity in Heterogenous Population of Pakistan. The International Journal of Frontier Sciences, (2019); 3(1): 41-56.
3. Akthar MS. Role of Epidemiological Studies in Disease Prevention. The International Journal of Frontier Sciences, (2019); 3(1): 1-2.
4. Omrani B. the durand line: history and problems of the Afghan-Pakistan border. Asian Affairs, (2009); 40(2): 177-195.
5. Britannica E. Encyclopaedia Britannica. Millennium 4th Edition Copyright, (1994); 20031994-2003.
6. Dani AH. Pakistan through ages: History of Pakistan. 2007; 1. Sang-e-Meel Publications.
7. Faiz S, Zehra S, Qureshi F, Mumtaz S. Colour Blindness Among Male Professional Drivers: A Cross-sectional Study In Karachi, (2019); 9(2): 124-127.
8. Khairoalsindi OA, Almasoudi BM, Bamahfouz AY, Alghamdi AA, Siddiqui MI. Prevalence and determinants of color vision defects among preparatory university students at Makkah, Saudi Arabia. Middle East African Journal of Ophthalmology, (2019); 26(3): 133.
9. Nazeer M, Bashir S, Rafiq N. Color Vision Deficiency in Medical Students in Jammu & Kashmir, India. Galician Medical Journal, (2019); 26(1): 23-26.
10. Sthanadar AA, Bittles AH, Zahid M. Civil unrest and the current profile of consanguineous marriage in Khyber Pakhtunkhwa Province, Pakistan. Journal of Biosocial Science, (2014); 46(5): 698-701.
11. Pervaiz R, Faisal F, Serakinci N. Practice of consanguinity and attitudes towards risk in the Pashtun population of Khyber Pakhtunkhwa, Pakistan. Journal of biosocial science, (2018); 50(3): 414-420.
12. Aslamkhan M, Ali A, Barnett H. Consanguineous marriages in rural west Pakistan. Annual Reptorts of University Medical School ICMRT, (1969); 69: 181-192.
13. Aslamkhan M. Clinical genetics and genetic counselling in Pakistan. Journal of Genes and Cells, (2015); 1(2): 31-33.