Vol. 2 No. 3 (2025)
ONLINE ISSN :3007-309X
PRINT ISSN :3007-3081
Journal of Medical & Health Sciences Review

THE PREVALENCE OF COLOR VISION DISORDER (CVD) AND CONSANGUINITY IN POPULATION OF DISTRICT KARAK, KHYBER PAKHTUNKHWA (KP), PAKISTAN

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ARTICLE INFO:

Keywords: Color blindness, Ishihara test, City university test, Farnsworth D-15, consanguinity, X linked recessive, District Karak, KP, Pakistan.

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Article History:
Published on 03 August 2025

ABSTRACT

Objectives: To observe the prevalence of color vision disorder (CVD) in district Karak, Khyber Pakhtunkhwa (KP) Pakistan from September 2023 to September 2024.

Methodology: During this field work, samples of 555 (300 male and 255 female) individuals were collected from surrounding locations, i.e. Karak city, Metha khel, Landiwa, Thth-e-Nasrati, Gurguri, Latamber, Banda Dawood Shah and Shagi Banda. The data were collected via a predesigned questionnaire printed in Urdu and English, and the history of consanguinity of parents was also recorded.

Results: We used Ishihara test, City University test (CUT) and Farnsworth D-15 to identify color blind people, revealed that 28 people (5.04%) were color blind. Out of these, 19(67.85%) were males and 9(32.14%) were females. Among these, 28 color blind individuals, 16(57.14%) were red-green color blind, 5 (17.855) were red color blind, 5 (17.85%) were green color blind and 2 (7.14%) were blue color blind. Furthermore, consanguinity of parents was recorded and interestingly all affected individual parents had consanguineous marriages. Two different types of pedigree analysis were recorded too. In the collected data, people aged between 18 to 60 years were present.

Conclusion: We found a significant prevalence of CVD in males (6.3%) than in females (3.5%). The study is significant in that it reported the prevalence of color blindness for the first time in Southern KP, Pakistan. This study provides the foundational understanding of CVD and its prevalence in district Karak and a framework for further research and prevention strategies regarding CVD.

INTRODUCTION:

Worldwide, average color blindness prevalence is approximately 7 percent in males and 0.5 percent in females; however, its prevalence varies from one geographical region to another and from one race to another in different populations. Similarly, in India, prevalence of CVD is recorded as 3.89% in male and 0.18% in females[1]. According to a comparative study conducted in Pakistan between medical and nonmedical students, the total prevalence of color vision impairment was from 2.75 to 2.8% [2]. Human eyes have two types of photoreceptor cells located on retina: the rod cells and the cone cells. The Rod cells are responsible for differentiation between black and white colors, or between day and night, whereas the cone cells are responsible for discrimination among different color [3]. Human eyes respond to light through a series of events called photo- transduction[4].

Normally, blindness is caused by degenerative disorders of the retina called retinal dystrophies (RD) like disturbance in light sensation or response of photoreceptor cells to light, physical injury to eye or mutation in proteins responsible for normal vision. So, in this way, blindness is of two types. It may be complete blindness in which total vision loss occurs or it may be color blindness [5-7]. It may be caused due to disturbance in the cone cells, retina, photo perception or mutation in X chromosome [8]. Mostly, it is congenital and due to the missense mutation in genomic structure of L and M genes, located on long arm of the X chromosomes. Normally, the human eye can absorb three specific wavelength photons, i.e., red, green, and blue. Due to this absorbing capacity, normal human vision is trichromatic and is called trichromatic vision or trichromacy[9]. There are three types of color blindness: monochromatism, dichromatism, and achromatism. From molecular studies, it is concluded that color vision defects resulting from alteration, defects, absence, or malfunctioning of one photopigment result in dichromatism, two pigments absence or malfunctioning gives rise to monochromatic, and total absence or malfunctioning of all three photopigments results in achromatism or total color vision loss [10-12].

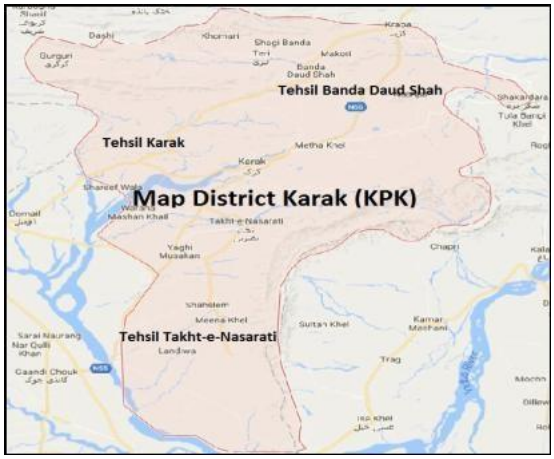
Published summaries of all the data available before 1960s showed that the prevalence of color blindness in male population is about 8 per cent in European Caucasians, 5 per cent in Asians, 4% in Africans and less than 2 per cent in indigenous Americans, Australians, and Polynesians [13]. Similarly, the prevalence of CVD in Muslim countries also varies. Its frequency is recorded from 5.41% to 7.86% in different cities of Turkey [14]. In different cities of Saudi Arabia, the prevalence of CVD is recorded 5.85% in male and 0.75% in females in Riyadh, 1.77% in Makkah [15]. Similarly, in India Prevalence of CVD is recorded as 3.89% in male and 0.18% in females [16]. Mughal et al studied the prevalence of CVD in different countries and reported its prevalence 2.1% in 7,542 males and 0.2 % in 3,519 females in India. In Potohara, Western Nepal, 10 to 19 years age group children were studied and the prevalence of CVD was recorded as 3.8% in 18 boys but no female was reported to be colour blind [17]. In Iran, the recorded prevalence for the first time was 8.18% in 93 studied male and 4.93% in 53 males, while 0.43% in 4 studied females and 0.32% in 3 studied females [18].

In Pakistan, the known percentages of CVD among males are 3.59%, 5.69%, 2.75%, 2.24%, 10.0%, 7.95 % and 3.1% and in females known percentages are 4.4%,1.64%, and 1.39 %. It means that the prevalence of CVD varies from province to province in Pakistan and many factors contribute to this variation like consanguineous marriages, environmental factors, etc. Due to these factors, the frequency varies from province to province and even from district to district. In spite of all these problems, the prevalence of CVD reported from different regions of

Pakistan in different published materials is 5.1% in Rawalpindi, 2.48% in Quetta, 3.1% in Southern Punjab [19]. In Karachi reported a total of 0.9% prevalence of CVD in 3437 individuals' sample. Among these CVD positive individuals, 1.45 were males and 0.4% were females. The prevalence of CVD in Peshawar was 2.91% in males and 0.83% in females among dental students at Sardar Begum Dental College Peshawar [20].

Materials and Methods Study Area

The study was conducted in different areas of District Karak, Khyber Pakhtunkhwa, Pakistan, from September 2023 to September 2024. The total area of Karak is 3372 sq km, and the total population is 815,878 (2023 Census). The majority of the people are very poor and have low economic status, and there is a lack of facilities regarding proper health and awareness about the disorder and its consanguineous transmission.



Study Design

The study was designed to know the prevalence of color vision disorder and its consanguinity factor in individuals of both sexes of all ages in different regions in Karak including city and its surrounding areas such as Karak city, Metha khel, Landiwa, Thath e Nasrati, Gurguri, Latamber, Banda Dawood Shah and Shagi Banda (Table 1).

Data Collection

The data was collected from different regions of district Karak after approval from the Ethical Review Committee of Government Postgraduate College, Karak (GPGC Karak). The sample size calculated was n=555. The study included all individuals' cases of vision disorder. A predesigned questionnaire, printed both in English and Urdu, was used to collect data. The history of consanguinity of parents was also recorded.

Table .1 Details of Data Collection

Serial No	Location	No of sample 2023-2024
1	Karak city	150
2	Metha khel	100
3	Tahth e Nasrati	70
4	Latamber	135
5	Landiwa	30
6	Gurguri	55
7	Banda Dawood Shah	12
8	Shagi Banda	3
Total		555

Screening

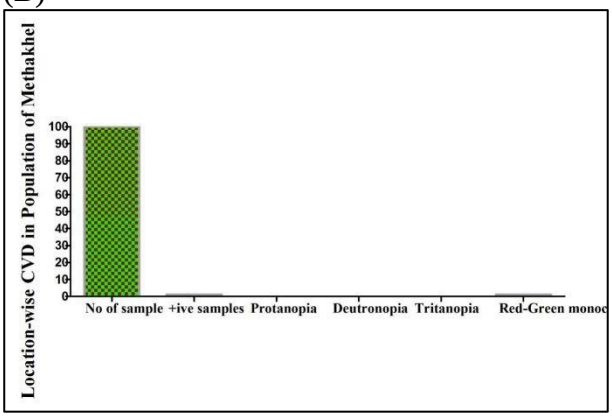
Total 38 different Ishihara plates were used for the detection of Red-Green monochromacy which contains various figures and lines [21]. while to find protanopia, deuteranopia and Tritanopia and making the diagnostic procedure more accurate and precise city university test (CUT) and Farnsworth D-15 test were performed. CUT contained 10 plates to assess colour vision. Each plate features a central coloured dot surrounded by four peripheral dots, designed to test for various colour vision disorders, including those related to protan, deutan, and tritan deficiencies [22]. The Farnsworth D-15 test is a method for screening CVD by having the patient arrange coloured discs in a sequence, with a reference disc provided.

It's a dichromatic test, meaning it's designed to distinguish between individuals with normal colour vision and those with mild to severe colour vision disorders. The test can help in identifying issues with red-green discrimination (deutan or protan) and blue yellow discrimination (tritan) [23].

Results

A total of 555 individuals were screened for color blindness both male and female. Before screening, a questionnaire was filled with all necessary information, including the personal profile (name, sex, age, profession, education, address, pedigree). In 555 individuals, 300 (54.05%) were males and 255 (45.94%) were females. Only 28 were found positive for CVD in both male and female individuals via diagnostic tests. Out of the 28 individuals, 19 (67. 85%) were male and 9 (32.14%) were female. Among the 28 positive individuals, 16 (57.14%) were positive for red-green color blindness,

(B)



(C)

5 (17.85%) were positive for red color blindness. Whereas 5 (17.85%) were positive for green color blindness and 2 (7.14%) were positive for blue color vision disorder (Table 2 and Fig 1).

Fig 1. Total prevalence of CVD in population of District Karak

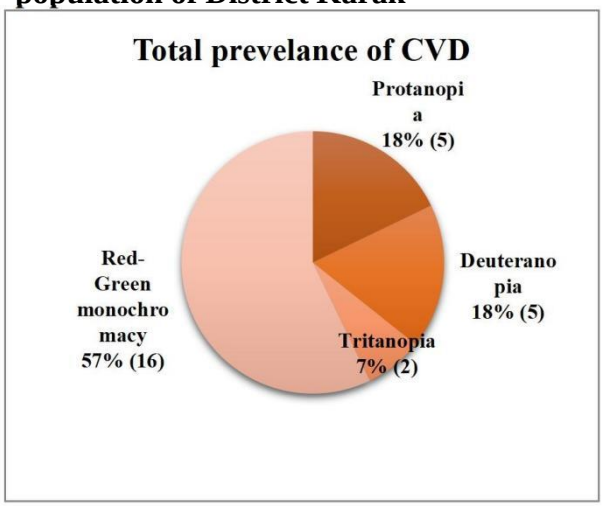
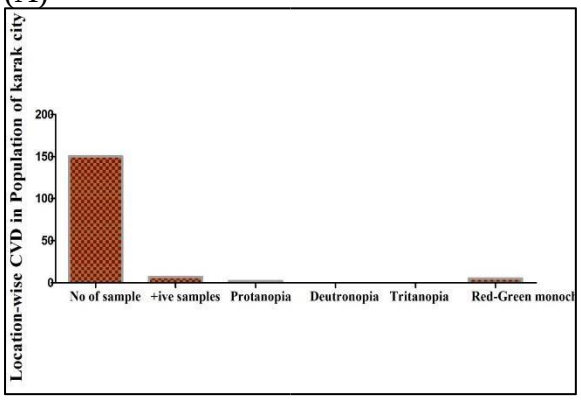


Figure 2. Location-wise prevalence of CVD in population of District Karak

(A)



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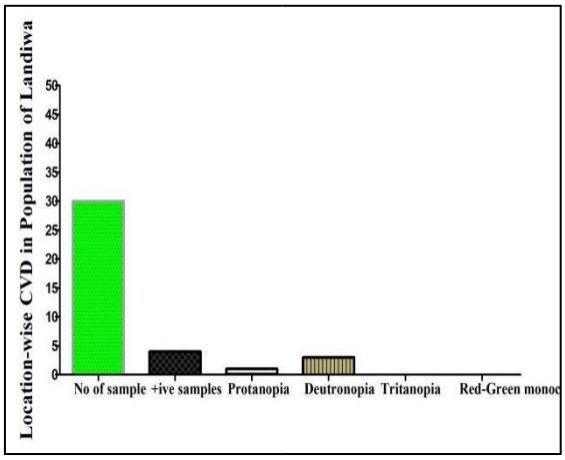

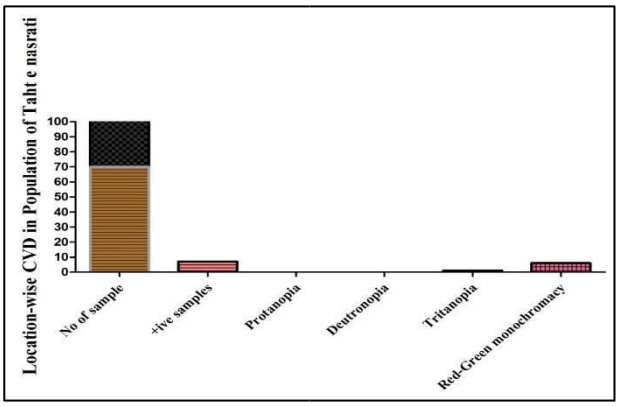

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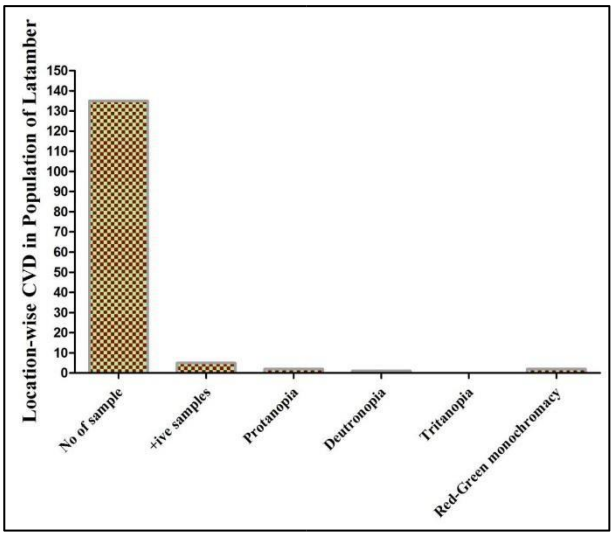

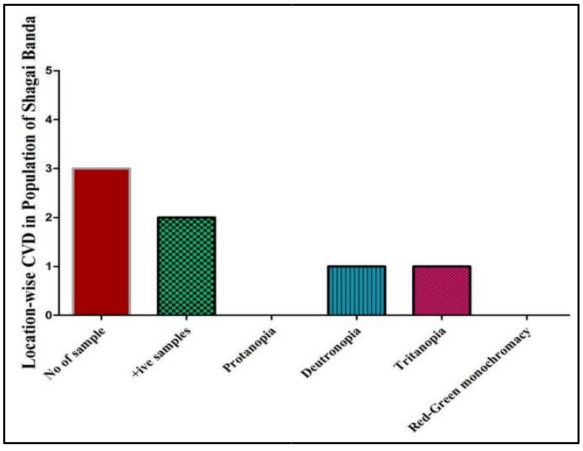

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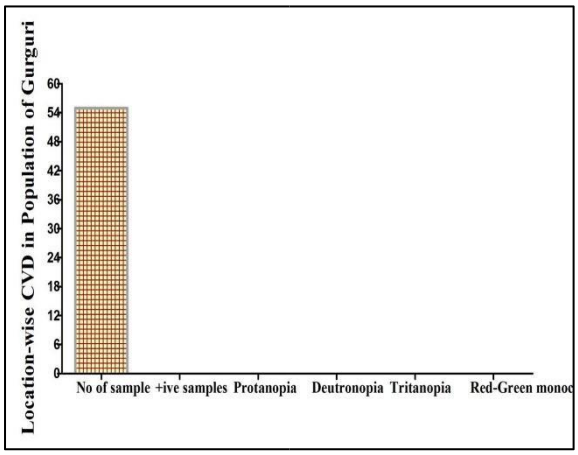

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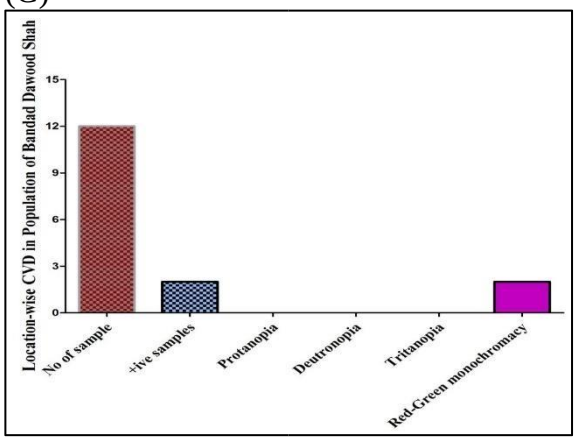
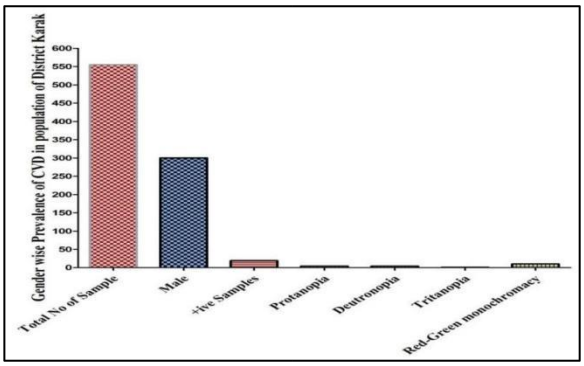


(H)

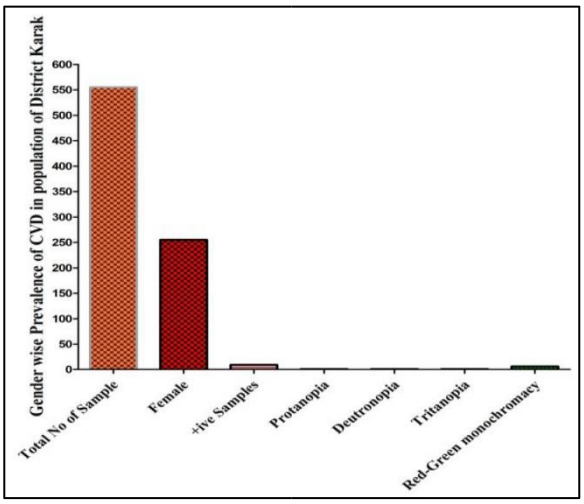
Gender wise Prevalence of CVD in

Next, we noticed that number of CVD female ratio were more significant than male population as shown in Fig.3.

Fig 3. Gender wise Prevalence of CVD in population of District Karak



(A)



(B)

population of District Karak

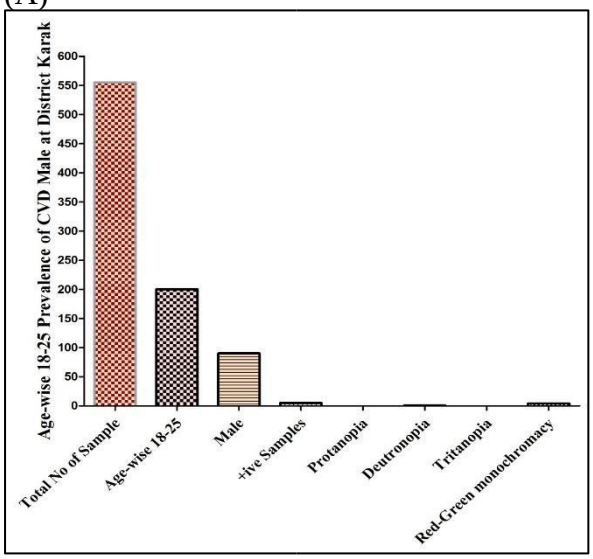
Age wise Prevalence of CVD in Male District Karak

Next, we examined the color blindness disorder on age basis. We found that 18-25 Protanopia 0%, Deuteranopia 1.11%, Tritanopia 0%, Red-Green monochromacy 4.44%. The age of were 26-35 Protanopia 2.86%, Deuteranopia 1.43%, Tritanopia 0%, Red-Green monochromacy 5.71%.

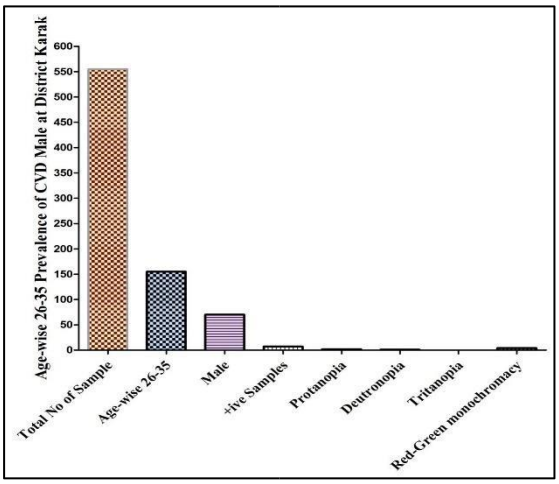
The age of were 36-45 Protanopia 1.12%, Deuteranopia 2.25%, Tritanopia 0%, Red-Green monochromacy 2.25%. The age of 46-60 were Protanopia 1.96%, Deuteranopia 0%, Tritanopia 1.96%, Red-Green monochromacy 0% as shown in Fig.4.

Fig 4. Age wise Prevalence of CVD in Male

(A)



(B)
District Karak



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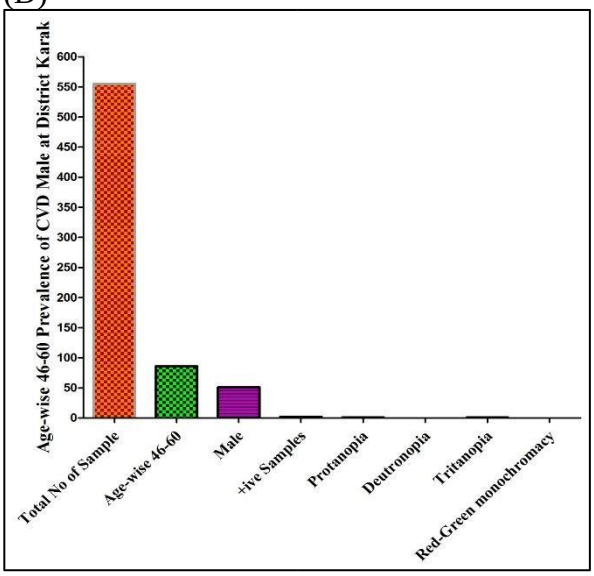
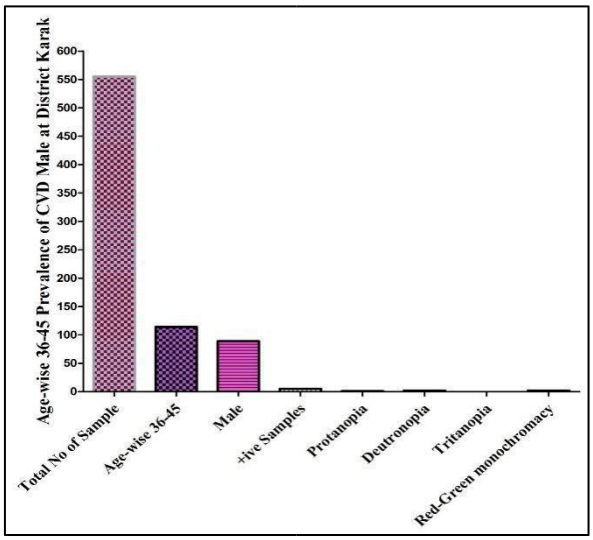




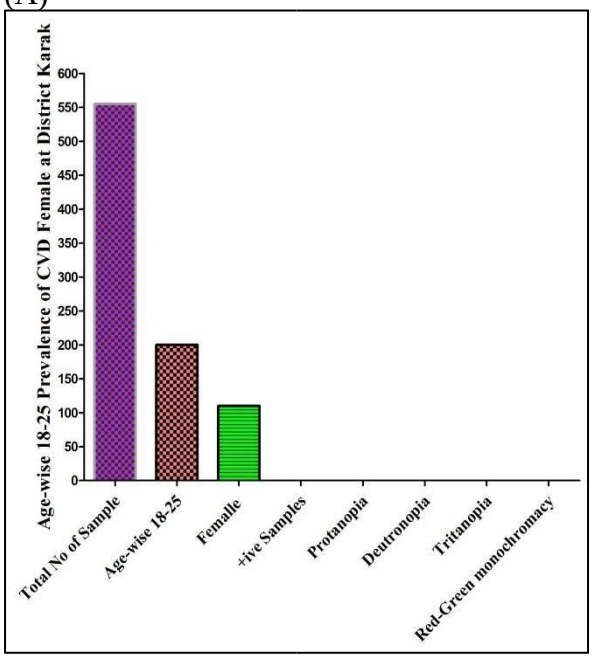
Age wise Prevalence of Color Blindness Disorder in Female District Karak

The age-wise female ratio was in of 18-25 Protanopia 0%, Deuteranopia 0%, Tritanopia 0%, Red-Green monochromacy 0%. The age-wise female ratio was in of 26-35 Protanopia 0%, Deuteranopia 0%, Tritanopia 1%, Red-Green monochromacy 0%. The age-wise female ratio was in of 36-45 Protanopia 4%, Deuteranopia 4%, Tritanopia 0%, Red-Green monochromacy 4%. The age-wise female ratio was in of 46-60 Protanopia 0%, Deuteranopia 0%, Tritanopia 0%, Red-Green monochromacy 5.71% as shown in Fig 5.

Fig 5. Age wise Prevalence of CVD in

Female of District Karak

(A)



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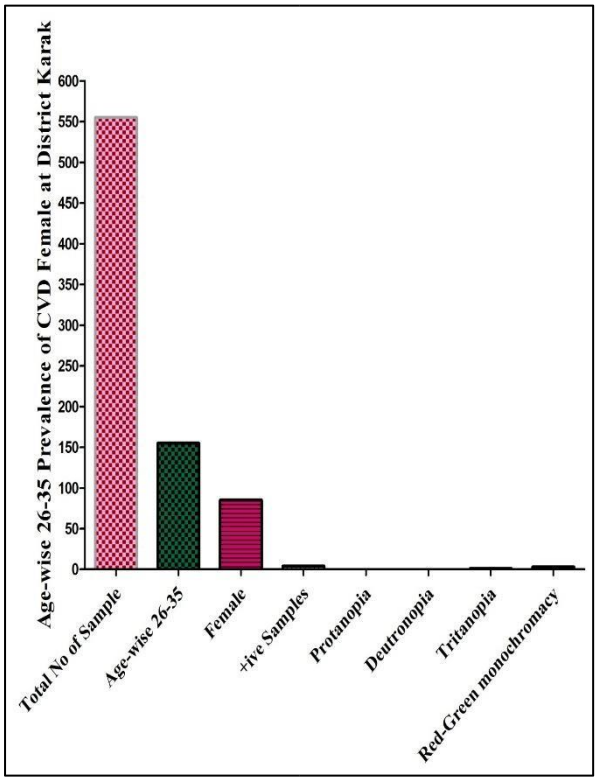

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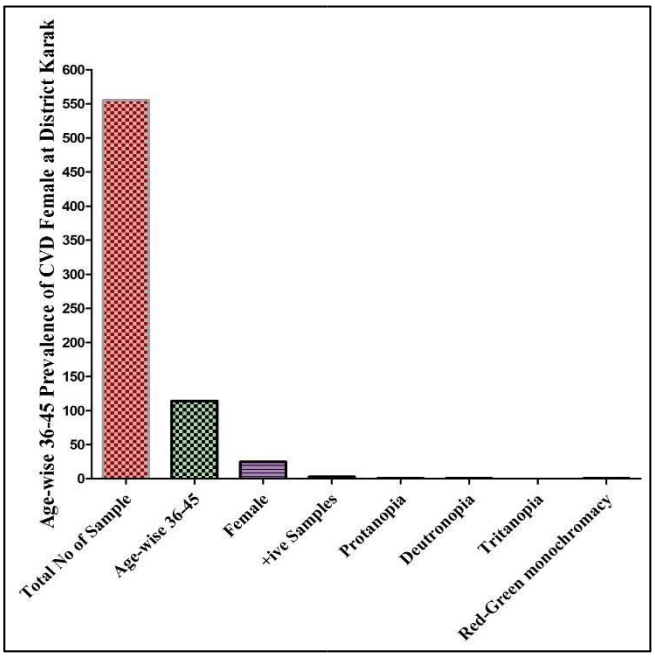

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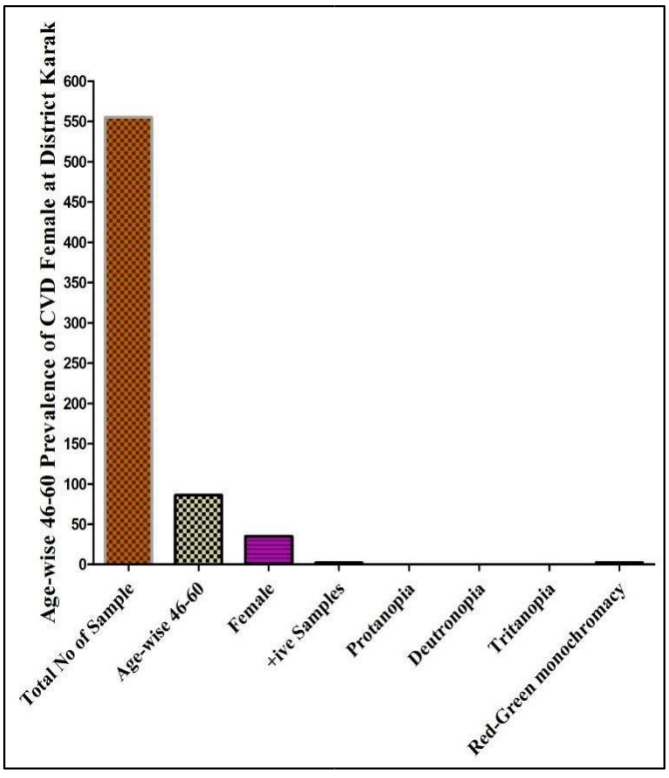

Consanguinity of the Disorder

In this study two different types of pedigrees were recorded. In one type pedigree, the female proband was identified with color blindness disorder who was married to a normal male, although they were not consanguineous related (Fig 6A). The result was two affected male individuals and four female carrier individuals were born. One of the carrier female individual was married to her close cousin who was normal. The resultant two male progenies were affected and one female was normal (Fig 6C).

Another type of pedigree was identified with male proband individual in our study Population consanguinity related analysis.

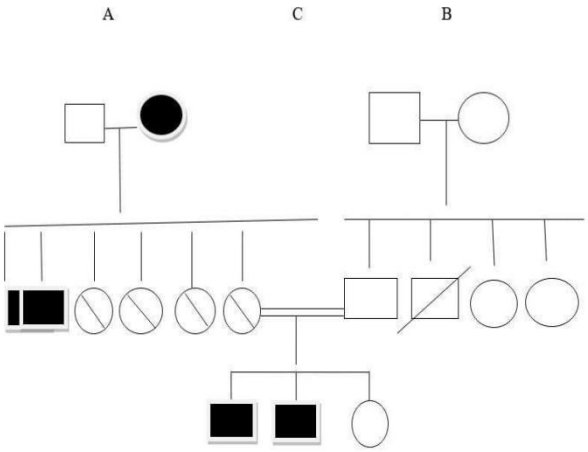

Fig 6. Pedigree of One female Proband Identified during Study

When an affected male married a normal woman, although they were not consanguineous related (Fig 7A), the result was a normal male and two carrier female individuals were born. One of the carrier's female is married to her close cousin who was affected then; two affected female individuals and one normal male individual were born (Fig 7C) and Table 4. Tritanopia is autosomal dominant disorder so consanguinity is not important for its inheritance from parents to their offspring's. As in our study two individual's male and female were recorded, their parents were normal but they were affected with tritanopia.

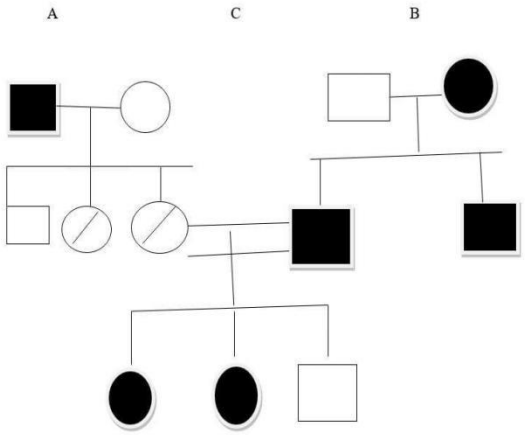

Fig 7. Pedigree of One Male Proband Identified during Study

Table 4. CVD in relation to consanguinity in

Consanguinity among parents of study individuals	Protanopia	Deuteranopia	Tritanopia	RedGreen monochromacy
Maternal Aunt Cousin (Khalazad)	0	2(M)	0	4(M)

Maternal Uncle Cousin (Mamonnzad)	1(M)	1(M)	1(M) (Mutated)	2(F)
Paternal Aunt Cousin (Phuphizad)	1(F)	1(M)	0	3(M)
Paternal Uncle Cousin (Chachazad)	2(M)	0	1(F) (Mutated)	2(M)
Second Cousin	0	0	0	3(F)
Distant Blood Relation	0	0	0	0
Same Subcaste (Bradri)	1(M)	0	0	1(F)
Same Caste	0	1(F)	0	1(M)
Different Caste	0	0	0	0
Total	5	5	2	16

DISCUSSION

Under normal lighting circumstances, a failure or diminished ability to detect color variations is known as color blindness or color vision disorder (CVD). CVD is a predominant X linked recessive disorder having genes located on the long arm of the X chromosome at loci 28 (Xq28), so its prevalence in males would be more as compare to females[24]. We examined 555 individuals among only 28 were positive. Among these positive, 19 were males and 9 were females. The previous studies revealed the same results of high prevalence rate of disorder in males 8% than females 0.4 % 25,26. Prevalence of CVD varies from area to area and caste to caste but its prevalence occurs mostly 3 to 8% in males and 0.3 to 0.5% in females and has been proved via many literatures [25]. In all forms of color blindness, red green color blindness is most common form, which is inherited as X linked recessive disorder[26]. Furthermore, we observed that the number of red color-blind individuals was similar to that of green color blind. Among 5 out of 28 (17 .85%) were red color blind and 5 (17.85%) were green color blind and all affected individuals' parents' marriages were consanguineous. Previous study has shown that protanopia was 1.94% and deuteranopia was 2.27%[27]. it can be concluded that CVD prevalence may vary from one region to another and from one race to another race[28]. Herein, the history of two families has been recorded in 8 location of district Karak in which both consanguineous and inter-caste marriages were recorded. In 2019 dichromacy and color vision impairment study was done in district Chiniot Punjab, Pakistan where the affected individual parents history was recorded in which 7 dichromatic individuals parents were paternal uncle cousins (First cousin), 4 dichromate parents were second cousins ,3 individual parents were maternal aunt cousins (second cousin), 1 individual parents were maternal uncle cousin (Mamuzad), 3 individual parents belonged to the same sub-caste (bradri), one patient parents were of the same caste while that of two individuals of different castes. Their study and our current study have similarity in family history of affected individuals because in this study, recorded consanguinity rate 6 red green monochromats ,2 protonopia individual parents were paternal aunt cousins (Chachazad),3 red green,1 protonopic and one deuteranopic individual parents were paternal aunt cousins (Phuphizad), 4 red green color blind ,and 2 deuteranopic individual parents were maternal aunt cousins (Khalazad),3 red green defective

individual parents were second cousins, 1 protonopic individual parents were of the same sub-caste 9 (bradri) while that of one deuteranopic individual parents were of the same caste [29].

According to consanguinity analysis protanopia, deuteranopia and red-green monochromacy are X-linked recessive disorders while tritanopia is autosomal chromosome number 7 linked dominant disorder [30]. Although it is very rare even 1 in 13000 effected and can pass from parents to children with 50% probability. In our study 2 individuals one was male and another was female recorded. Although tritanopia is not linked directly to consanguinity like other CVDs but consanguinity can increase the risk of certain autosomal dominant disorders[31]. The parents of tritanopic male and female individuals were maternal auntcousins and paternal uncle cousins they were normal but their children were tritanopic. This showed some type of autosomal mutation that affects male and female equally as recorded in this study and many other studies as well[32]. As it is very rare, so it is not considered in most of the studies or is neglected. CVD is being an X linked recessive disorder is deeply related with gender and consanguinity, but it has no relation with age. In this study almost, all age groups from 18 to 49 were included and it shows that color blind has no relation with age [33].

CONCLUSION

This, study is significant for reporting the prevalence of color blindness for the first time in southern KP, Pakistan. The study also shows relation of CVD with consanguinity. This study provides basics, awareness and a pathway for further studies. Furthermore, it is also observed that CVD being X linked recessive disorder is greatly related to consanguinity although, it may be onset disorder and may occur at any stage in life like other disease but mostly it is congenital. The findings reveal the prevalence of CVD as 6.3% in male and 3.5% in females in District Karak, KP, Pakistan. CVD although is not considered a serious disordered and even people are mostly unaware of its occurrence but sometime it become life threating and become the rejection cause of people in many occupations like medical, arm forces etc. Till now no special treatment is discovered to treat CVD like other congenital disorders, and the patients remain unaware of their condition even throughout life. This research provides a foundational understanding of CVD in district Karak KP Pakistan, providing a framework for further studies and prevention strategies.

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