



## Incidence of Protanopia and Deuteranopia, Defects of Colour Vision in Quetta, Pakistan

Hamida,<sup>1</sup> Tehmina Sajid,<sup>1</sup> Amna Bibi,<sup>1</sup> Nabeela Tariq,<sup>1</sup> Naheed Sajjad,<sup>2</sup> Kashif Umer,<sup>3</sup> Muhammad Ali<sup>4</sup> and Rehana Iqbal<sup>3\*</sup>

<sup>1</sup>Zoology Department, Sardar Bahadur Khan Women's University, Quetta, Pakistan

<sup>2</sup>Biotechnology Department, Sardar Bahadur Khan Women's University, Quetta, Pakistan

<sup>3</sup>Institute of Pure and Applied Biology, Bahauddin Zakariya University, Multan.60800

<sup>4</sup>Government College University, Faisalabad.

### ABSTRACT

Colour vision deficiency (CVD), an X-linked recessive disorder, is predominantly present in males. Congenital disorders usually occur due to abnormality in any one or all three cone photoreceptors. Protanopia and deuteranopia result when long wavelength (L) photopigments (red) and middle wavelength (M) photopigments (green) are missing, respectively. The study aimed to screen the inherited colour vision defects among random populations of different ethnicity in Quetta. The study subjects (n=1450; males=452; females=998; mean age=23.17±9.38) were randomly selected and examined for CVD using the Standard Ishihara Chart. Results revealed that 5.75% (26) males and 1% (10) females were colour vision deficient. The distribution of protanopia and deuteranopia among males were 2% (9) and 3.8% (17), respectively. Females showed 0.6% (6) and 0.4% (4) protanopia and deuteranopia, respectively. No significant difference (P>0.05) was observed in the prevalence of CVD among different age groups. Familial cases were far more prevalent than the sporadic cases. Among ethnic groups the highest proportions of CVD in males and females were observed in Pathan (7.39% and 1.16%, respectively). No significant difference (P>0.05) in the proportion of disorder was found among different ethnic groups. Overall 2.48% of the populations of Quetta had this abnormality with deuteranopia being more prevalent. The proper screening of CVD at early age can help individuals avoid certain occupational hazards and such studies can also be helpful in decreasing the proportion of disorder by discouraging consanguinity.

### Article Information

Received 18 June 2015

Revised 26 November 2015

Accepted 10 January 2016

Available online 1 June 2016

### Authors' Contribution

MA designed and supervised the study. H, TS and AB collected the data. NT and NS performed experimental work. KU statistically analyzed the data. RI wrote the article.

### Key words

X-linked recessive, photoreceptor, Ishihara chart, consanguinity.

### INTRODUCTION

Colour vision deficiency (CVD) is the inability to perceive differences among certain colours of spectrum. Normal colour vision is actually trichromatic that is the combination of red, green, and blue lights (Curcio *et al.*, 1990). It requires three types of cone photopigments that differ in their relative spectral sensitivities, and are generally termed as red (long wavelength), green (middle wavelength), and blue (short wavelength) cone pigments (Cruz *et al.*, 2010). There are three types of inherited CVD: monochromacy, dichromacy and anomalous trichromacy. Monochromacy, being so rare, is the total absence of colour vision and occurs when two or all three of the cone pigments are missing. Dichromacy occurs when only one of the cone pigments is missing and is categorized into protanopia (characterized by the complete absence of red cone), deuteranopia (characterized by the absence of green cone) and tritanopia (complete absence of blue cone). Anomalous trichromacy involves reduced sensitivity to one of the

three cone pigments and includes protanomaly, deuteranomaly and tritanomaly in which the spectral sensitivity of the red, green and blue cone receptors is altered. Achromatopsia is the most severe kind of colour vision defect (Adams *et al.*, 2009).

Red and green CVD (protanopia and deuteranopia, respectively) have the highest frequency in the general populations (Oriowo and Alotaibi, 2008). The genes causing red-green defects are localized on the long arm of the X-chromosome at Xq28 (Filosa *et al.*, 1993; Norn, 1997; Deeb and Kohl, 2003), whereas the blue pigment gene is located on an autosome, chromosome 7 at 7q32 (Nathans *et al.*, 1986; Motulsky, 1988; Deeb and Kohl, 2003). The tritanopia type, a rare autosomal dominant disorder, occurs in 0.002-0.007% in the population (McIntyre, 2002). Achromatopsia, a rare autosomal recessive disorder, occurs when the functions of all three cone receptors are lost and is the complete inability to distinguish between different colours. It has a prevalence of about 0.003% of the population (Deeb, 2004; Alexander *et al.*, 2007). Colour vision defect may be congenital or acquired. Acquired CVD may be caused due to factors such as damage to the optic nerves, metabolic disorders (*e.g.* diabetes), eye diseases (*e.g.* glaucoma, macular degeneration), chronic illness (*e.g.*

\* Corresponding author: [rehanaiqbal82@gmail.com](mailto:rehanaiqbal82@gmail.com)

0030-9923/2016/0004-1045 \$ 8.00/0

Copyright 2016 Zoological Society of Pakistan

sickle-cell anaemia), drug over-dose (*e.g.*, barbiturates, digoxin, anti-tubercular drugs) (Ganong, 2005).

Colour vision defect usually remains undiagnosed that results in serious handicaps. It is considered as occupational hazard all over the world and poses problems for the affected individuals such as difficulty in recognition of traffic signals (13%), job disabilities (25%), judgment difficulties in daily routines (75%) and the choice of a career (33%) (Holroyd and Hall, 1997). People with abnormal colour vision must be advised not to be in the professions such as pilots, electrical jobs, police, navigators, aircraft maintenance workers and certain jobs in armed forces (Guest *et al.*, 2011). Similarly, a relatively higher rate of road accidents has been reported by the colour deficient people (Verriest *et al.*, 1980). Colour is often used as a sign in the practice of medicine. There are many commonly used diagnostic and descriptive terms indicating the importance of colour recognition in medical such as jaundice, erythema, cyanosis, rubella and melaena (Spalding, 1999a).

CVD has been reported in many populations from many countries. In Pakistan, there is very little data available reporting the distribution of protanopia and deuteranopia in the general population. Therefore, the present study was designed to evaluate the prevalence of protanopia and deuteranopia in the random populations of Quetta, Pakistan.

## MATERIALS AND METHODS

This cross sectional study was designed and approved from institutional ethical review committee, Sardar Bahadur Khan Women University, Quetta. After brief introduction to CVD, the consent of all individuals was taken. Information obtained from the individuals included name, gender, age, ethnic group, type of colour vision defect, eye sight problem, and family history of the abnormality. A total of 1450 individuals comprising 452 (31.2%) males and 998 (68.8%) females with mean age  $23.17 \pm 9.38$  (ranged from 8 to 83 years) participated in the study. The study population consisted of 521 Pathan, 510 Baloch and 419 participants from other ethnic groups (*i.e.* Punjabi, Hazara, Sindhi and Urdu speaking).

Colour vision defect was tested using the standard Ishihara's colour plate chart (Pseudo-isochromatic plates). The chart consisted of 38 test plates, each containing printed figure/pattern made up of coloured spots on a background of similarly shaped coloured spots. The spots of background and spots of the figure were of different colours. The figures were made up of coloured spots intentionally that were likely to appear the same as the background to the colour-deficient individuals. The participants were asked to read the figures/impression at a

distance of 75cm and tilted at right angle to the line of vision (Karim and Saleem, 2013). The individuals who read all the plates correctly was considered normal while the one who either could not read the coloured figures or read incorrectly was regarded to be colour vision deficient. The types of defective colour vision were investigated with the help of the key provided with the chart.

## Statistical analysis

Data were statistically analyzed on SPSS-16 to find out frequencies of protanopia and deuteranopia among the studied population. Chi-square test was applied to determine the difference in the proportion of CVD among different age groups and ethnic groups at 5% significance level.

## RESULTS

The results of the current study showed that 5.75% (26/452) males and 1% (10/998) females were colour vision deficient. The overall percentage of CVD in the random populations of Quetta city was 2.48% (36/1450). Out of 26 affected males, 9 (2%) were protanopes and 17(3.8%) were deuteranopes while among females 6 (0.6%) were protanopes and 4 (0.4%) were deuteranopes (Table I, Fig. 1).

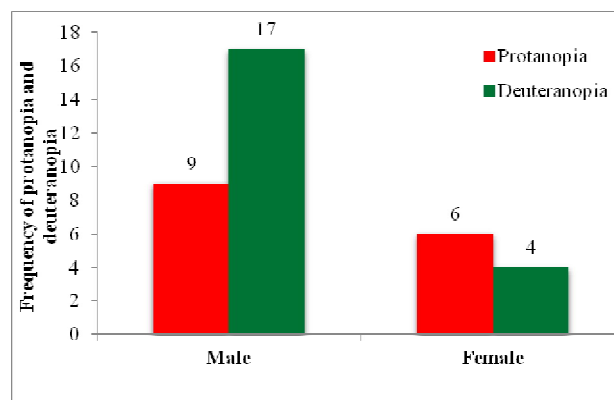


Fig. 1. Frequency of protanopia and deuteranopia among male and female populations residing in Quetta.

Table II shows age wise comparison of colour vision deficiency among individuals. The colour vision disorder was observed in almost all age groups. However, difference (Chi-square= 4.681, df= 6,  $P= 0.585$ ) in the prevalence of CVD among different age groups was not statistically significant. The study also revealed that 93.3% (14/15) protanopia cases were familial while 6.7% (1/15) were sporadic. Out of 21 deuteranopes 18 (85.7%)

were familial and 3 (14.3%) cases were sporadic (Table III).

**Table I.- Gender wise frequency distribution of protanopia and deuteranopia in Quetta.**

	Male	Female	Total
Total	452	998	1450
Normal colour vision	426	988	1414
N (%)	(94.25)	(99.00)	(97.52)
Deficient colour vision	26	10	36
N (%)	(5.75)	(1.00)	(2.48)
Protanopia	9	6	15
N (%)	(2.0)	(0.6)	(1.03)
Deuteranopia	17	4	21
N (%)	(3.8)	(0.4)	(1.45)

**Table II.- Age wise frequency distribution of colour vision deficiency.**

Age (Years)	Total N	Colour vision deficient N (%)	Normal N (%)
5-14	264	4 (1.5)	260 (98.5)
15-24	333	12 (3.6)	321 (96.4)
25-34	256	6 (2.3)	250 (97.7)
35-44	231	5 (2.2)	226 (97.8)
45-54	140	5 (3.6)	135 (96.4)
55-64	116	3 (2.6)	113 (97.4)
>64	110	1 (0.9)	109 (99.1)
Total	1450	36	1414

**Table III.- Frequency distribution of familial and sporadic cases among protanopia and deuteranopia**

Color vision defect	Total Cases	Familial N (%)	Sporadic N (%)
Protanopia	15	14 (93.3)	1 (6.7)
Deuteranopia	21	18 (85.7)	3 (14.3)

The distribution of CVD among males in the study population demonstrated that 7.39% (13/176) of Pathan, 5.36% (9/168) of Baloch and 3.70% (4/108) cases from other ethnic groups (*i.e.* Punjabi, Hazara, Sindhi and Urdu speaking) were found affected with the abnormality. In females, 1.16% (4/345) of Pathan, 0.88% (3/342) of Baloch, and 0.96% (3/311) female cases from other ethnic groups (*i.e.* Punjabi, Hazara, Sindhi and Urdu speaking) were colour vision deficient. Data analyzed by chi-square test at 0.05 significance level revealed no significant differences (Chi-square = 2.487,

df= 2,  $P= 0.288$ ) in the prevalence of CVD among different ethnic groups (Table IV).

## DISCUSSION

Since CVD is a genetic condition, its occurrence differs from one geographical area to the other and from population to population. Prevalence rate of the deficiency in Caucasians is 8% in males and 0.4% in females (Spalding, 1999b). The prevalence of CVD in the current study among males (5.75%) was similar to the findings observed in Saudi Arabia (5.85%) by Oriowo and Alotaibi (2008), and in Philippine (5.17%) by Cruz *et al.* (2010). Mian *et al.* (1991) also reported almost similar distribution of CVD among males (4.89%) in another study conducted in Quetta, Pakistan. However, the results of this study are relatively lower than earlier findings that reported 8.73% in Manipur, India (Shah *et al.*, 2013), 8.72 % reported in Jordan (Al-Aqtum and Al-Qawasmeh, 2001), 8.47% in Iraq (Karim and Saleem, 2013) and 8.18% in Tehran (Modarres *et al.*, 1996) but higher than 1.12% reported in India (Dakshayani and Gangadhar, 2006), 2.56% in Iran (Khalaj *et al.*, 2014), and 3.8% in Nepal (Niroula and Saha, 2010).

The CVD being inherited as an X-linked recessive disorder, males are affected more as compared to females. The females usually act as carriers (Guyton and Hall, 2005). Therefore, the incidence of CVD observed in the present study in females (1%) was partially in line with the results observed in Saudi Arabia 0.75% (Oriowo and Alotaibi, 2008), Iran 0.93% (Khalaj *et al.*, 2014) and India 1.69% (Shah *et al.*, 2013).

The occurrence of CVD in the random population was found among all age groups with statistically no significant difference. Most of the protanopia and deuteranopia cases were familial while only a few cases were sporadic. The higher prevalence was observed in Pathan in both males and females that showed 7.39% and 1.16% affected individuals, respectively. The present study also indicated the deuteranopia type of CVD to be more prevalent than protanopia.

Although few therapies have been proposed (*e.g.* iodine injection, electrical eye stimulation, large doses of vitamins), yet there are currently no surgical procedures or treatments exist to improve the chromatic vision of the individual (Richer and Adams, 1984). Experimental study on adult red-green colour deficient primates (gene therapy) cures colour blind monkeys (Mancuso *et al.*, 2009).

Colour recognition in some jobs like doctors, educational trainers and drivers is essential and hence detection of colour vision deficiency at an early age can be useful to avoid certain occupational hazards. Several

**Table IV.- Phenotypic frequency of CVD among different ethnic groups of Quetta.**

Ethnic Group	Male			Female		
	Normal N (%)	CVD N (%)	Total	Normal N (%)	CVD N (%)	Total
Pathan	163 (92.61)	13 (7.39)	176	341 (98.84)	4 (1.16)	345
Baloch	159 (94.64)	9 (5.36)	168	339 (99.12)	3 (0.88)	342
Others	104 (96.30)	4 (3.70)	108	308 (99.04)	3 (0.96)	311

studies indicated that doctors and nurses affected with colour vision deficiency performed worse than those with normal colour vision in certain medical practices (Spalding, 1999b).

The present study indicated overall 2.48% incidence in the population of Quetta. Further investigation on CVD with sufficiently larger sample size can determine the exact rate of this genetic condition.

### CONCLUSION

The present study revealed high proportion of color vision deficiency in Quetta population, predominant in males. Proper screening, education and counseling can help minimize this disorder in our country and can also be beneficial for the affected subjects in tackling difficulties in everyday work, proper choice of future profession, avoidance of consanguineous marriages and its genetic inheritance to the next generation.

### ACKNOWLEDGMENT

The authors acknowledge the cooperation given by the study populations to accomplish this study.

#### Statement of conflict of interest

The authors declare that they have no competing interests.

### REFERENCES

- Adams, A.J., Verdon, W.A. and Spivey, B.E., 2009. Color vision. In: *Duane's foundations of clinical ophthalmology* (eds. W. Tasman and E.A. Jaeger), 15th ed. Lippincott Williams & Wilkins; Philadelphia, Pa: chap 19.
- Al-Aqtum, M.T. and Al-Qawasmeh, M.H., 2001. Prevalence of colour blindness in young Jordanians. *Ophthalmologica*, **215**:39-42.
- Alexander, J.J., Umino, Y., Everhart, D., Chang, B., Min, S.H., Li, Q., Timmers, A.M., Hawes, N.L., Pang, J.J., Barlow, R.B. and Hauswirth, W.W., 2007. Restoration of cone vision in a mouse model of achromatopsia. *Nat. Med.*, **13**:685-687.
- Cruz, E.M., Cerdana, H.G., Cabrera, A.M., Garcia, C.B., Morabe, E.T. and Nanagas, M.L.R., 2010. Prevalence of color vision deficiency among male high-school students. *Philipp. J. Ophthalmol.*, **35**:20-24.
- Curcio, C.A., Sloan, K.R., Kalina, R.E. and Hendrickson, A.E., 1990. Human photoreceptor topography. *J. Comp. Neurol.*, **292**:497-523.
- Dakshayani, B. and Gangadhar, M.R., 2006. Red green colour blindness among the hakkipikkis: a tribal population of Mysore District, Karnataka. *Anthropologist*, **8**:141-142.
- Deeb, S.S., 2004. Molecular genetics of colour vision deficiencies. *Clin. exp. Optom.*, **87**: 224-229.
- Deeb, S.S. and Kohl, S., 2003. Genetics of color vision deficiencies. *Dev. Ophthalmol.*, **37**:170-187.
- Filosa, S., Calabro, V., Lania, G., Vulliamy, T.J., Brancati, C., Tagarelli, A., Luzzatto, L. and Martini, G., 1993. G6PD haplotypes spanning Xq28 from F8 C red/green colour vision. *Genomics*, **17**:6-14.
- Ganong, W.F., 2005. *Review of medical physiology*. 22nd ed. McGraw-Hill Companies.
- Guest, M., D'Este, C., Attia, J., Boggess, M., Brown, A., Tavener, M., Gibson, R., Gardner, I., Harrex, W. and Ross, J., 2011. Impairment of colour vision in aircraft maintenance workers. *Int. Arch. Occup. Environ. Hlth.*, **84**:723-733.
- Guyton, A.C. and Hall, J.E., 2005. *Textbook of medical physiology*. 11th ed. Elsevier Publication, p. 633.
- Holroyd, E. and Hall, D.M., 1997. A re-appraisal of screening for colour vision impairments. *Child: Care Hlth. Dev.*, **23**:391-398.
- Karim, J.K. and Saleem, M.A., 2013. Prevalence of congenital red-green color vision defects among various ethnic groups of students in Erbil City. *Jordan J. Biol. Sci.*, **6**:235-238.
- Khalaj, M., Barikani, A. and Mohammadi, M., 2014. Prevalence of color vision deficiency in Qazvin. *Zahedan J. Res. med. Sci.*, **16**:91-93.
- Mancuso, K., Hauswirth, W.W., Li, Q., Conner, T.B., Kuchenbecker, A.J., Mauck, M.C., Neitz, J. and Neitz, M., 2009. Gene therapy for red-green color blindness in adult primates. *Nature*, **461**:784-787.
- McLntyre, D., 2002. *Colour blindness – Causes and effects*. Dalton Publishing, Chester, UK, pp. 4-141.
- Mian, A., Ali, M. and Rafique, S., 1991. Frequencies of colour blindness in different ethnic groups of Quetta, Pakistan.

- Pakistan J. Zool.*, **23**:153-155.
- Modarres, M., Mirsamadi, M. and Peyman, G.A., 1996. Prevalence of congenital colour deficiencies in secondary-school students in Tehran. *Int. Ophthalmol.*, **20**:221-222.
- Motulsky, A.G., 1988. Normal and abnormal color-vision genes. *Am. J. Hum. Genet.*, **42**:405-407.
- Nathans, J., Piantanida, T.P., Eddy, R.L., Shows, T.B. and Hogness, D.S., 1986. Molecular genetics of inherited variation in human color vision. *Science*, **232**:203-210.
- Niroula, D.R. and Saha, C.G., 2010. The Incidence of color blindness among some school children of Pokhara, Westren Nepal. *Nepal Med. Coll. J.*, **12**:48-50.
- Norn, M., 1997. Prevalence of congenital colour blindness among inuit in East Greenland. *Acta Ophthalmol. Scand.*, **75**:206-209.
- Oriowo, O.M. and Alotaibi, A.Z., 2008. Colour vision screening among Saudi Arabian children. *S. Afr. Optomol.*, **67**:56-61.
- Richer, S. and Adams, A.J., 1984. Development of quantitative tools for filter aided dichromats. *Am. J. Optom. Physiol. Optics*, **61**:246-25.
- Shah, A., Hussain, R., Fareed, M. and Afzal, M., 2013. Prevalence of red-green color vision defects among Muslim males and females of Manipur, India. *Iran. J. Publ. Hlth.*, **42**:16-24.
- Spalding, J.A.B., 1999a. Medical students and congenital color vision deficiency; unnoticed problems and the case for screening. *Occup. Med.*, **49**:247-252.
- Spalding, J.A.B., 1999b. Color vision deficiency in medical profession. *Br. J. Gen. Pract.*, **49**:469-475.
- Verriest, G., Neubauer, O., Marre, M. and Uvijls, A., 1980. New investigations concerning the relationships between congenital colour vision defects and road traffic security. *Int. Ophthalmol.*, **2**:87-99.