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PREVALENCE AND ALLELE FREQUENCY OF CONGENITAL COLOUR VISION DEFICIENCY IN ILORIN, NIGERIA

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ABSTRACT

Colour vision deficiency (CVD) is the inability or decreased ability to see colour, or perceive colour differences under normal lighting conditions and this affects a significant percentage of the population. This study investigated prevalence and allelic frequency of congenital colour vision deficiency among high school children in Ilorin, Kwara State, Nigeria. 3025 high school children (1849 males and 1176 females) were randomly selected and screened for congenital CVD using the standard Ishihara test. Hardy-Weinberg equilibrium method was used to calculate allele frequency of CVD among the studied populations. Prevalence of CVD ranged from 5.26% to 11.36% among males and 0.00% to 3.03% among females of six different populations. The results showed that 2.6% of the 1849 males showed congenital CVD and 0.4% of the 1176 females showed congenital CVD. Proper and regular screening of infants and individuals of pre-high school ages for CVD is therefore recommended. This will hopefully ensure early detection and influence the choice of career in occupations such as armed forces, aviation, electrical, rail road, maritime, teaching, geologists, and health care professions which require standard and normal colour vision for optimum performance at work.

Keywords: Allelic Frequency; Colour test; Colour vision; Congenital; Ishihara; Prevalence

INTRODUCTION

Vision is the primary means of integration between individuals and the external environment. Colours have three attributes; hue, intensity and saturation (Barret et al., 2010). The human eyes have two types of photoreceptor cells, rods and cones, which serve different functions. The cones mediate coloured vision (Maureen and Jay 2000). Human retinas are trichromatic where the normal- green, blue and red sensitive cones are necessary for normal vision (Lee, 2004). A deficiency or absence of a pigment leads to

colour vision deficiency. Depending on the pigment absent, colour deficiency can be classified into the protan (red), deutan (green) and tritan (blue-yellow) types. The tritan CVDs are rare autosomal dominant traits and have a prevalence of 1 in 15,000 to 1 in 50,000 (that is, 0.02-0.007%) of the population (Pokorny et al., 1979). The colour vision deficiency could be congenital or acquired. Congenital colour vision deficiency is inherited and occurs due to genetic mutations that affect the expression of the full complement of normal cone

photoreceptors. Acquired CVD occurs due to factors that impair normal vision e.g. certain diseases, malnutrition etc. (Simunovic, 2010). *The genes that produce photo-pigments are located on the X-chromosomes and are expressed with higher probability in males than females because male sex carries one X-chromosome and females carry two X-chromosomes (Ahsana et al., 2013).*

Colour vision screening amongst high school children in Saudi Arabia showed that out of the 836 males and 800 females randomly selected and screened for red-green colour vision defects using Ishihara test, 5.85% males and 0.75% females had colour vision deficiency. This result shows that the prevalence among the males was higher than females (Oriowo *et al.*, 2008). A similar study carried out among children of six populations in North Indian region showed that amongst the 1028 child cohort, the colour vision deficiency frequency was found to be higher 41 (7.52%) among males than in females 4 (0.83%) (Mohd *et al.*, 2015). A study performed in Lagos with 5580 males and 5405 females in primary and secondary schools also showed that the frequency of red-green colour blindness was higher in males (3.60%) than in females (0.81%) (Williams *et al.*, 1998). The problems arise among CVD individuals working in industries (paint, textile, plastic, decorates, furniture), transport (rail, road, aviation, maritime), defense (police, armed force, fire and rescue services) and other occupations (electricians, technician, telecommunication, mechanics) (Mohd *et al.*, 2015). Thus, estimating the CVD phenotypes and gene frequencies among different population has several benefits in jobs and routine life that involve precise colour matching. The aim of this study is to investigate the prevalence and gene frequency of congenital colour vision deficiency in Ilorin, Kwara State, Nigeria. This study is relevant because there is no documented information on gene frequency of colour vision deficiency in Kwara State, Nigeria.

MATERIALS AND METHODS

This study investigated allele frequency and prevalence of CVD among 3025 individuals was drawn of which 1849 (61.1%) were males and 1176 (38.9%) were females. The screening of colour vision deficiency in this study was carried out using Ishihara pseudo isochromatic plates which are the most used plates for colour vision screening. The study was carried out in four local governments in Kwara State, Nigeria. Kwara state is located in the North Central geopolitical zone. It is the ninth largest state in Nigeria. Samples were collected from males and females in secondary schools in Ifelodun, Ilorin East, Ilorin South and Ilorin West Local Government areas.

Ishihara (2014) pseudo isochromatic plates were used to test the students for CVD. One of the peculiarities of red-green deficiencies is that blue and yellow colours appear to be clear compared with red and green colours. The students were shown the complete range of Ishihara's plates under day light conditions at normal reading distance and asked to read off the plates 1-38 of the Ishihara's colour album. Time consumed for reading each plate was not more than 3 seconds. The plates were shown to each student once only. Of the Plates 1 – 15, if 13 or more plates were read correctly, the colour vision was regarded as normal. If only 9 or less plates were read correctly, the colour vision was regarded as colour vision deficient and thus classified as colour blind. Those who read not more than two plates (Plate 1 inclusive) as either normal or red-green defective are classified as totally colour blind (monochromats) while the red-green defectives (dichromats) were further classified as deutans or protans based on their ability to read Plates 16 and 17 correctly.

The data obtained was presented in simple percentages. The chi square (χ^2) test was used to determine the level of significant differences as shown below:

$$\chi^2 = \frac{(\text{Observed} - \text{Expected})^2}{\text{Expected}}$$

Hardy-Weinberg equation was used to calculate the allelic frequency. Taking C allele to be p, and c allele to be q, then gene frequencies for colour blindness were calculated by using $(p+q=1)$. Where,

$$c = \sqrt{\frac{\text{Population of cv deficient}}{\text{Total population}}}$$

RESULTS

Figure 1 shows the percentage of males and females with normal vision and CVD in the overall population. The normal vision males and females recorded were about 58.5% and 38.5% of the population, respectively. The CVD males and females were 2.6% and

0.4% of the population, respectively. Table 1 shows the prevalence of congenital colour vision deficiency and allelic frequency in males and females in the population. The Table illustrates that the colour vision deficiency is more prevalent in males in the population than the females. 4.33% of the 1849 males in the population are colour vision deficient showing an allelic frequency of 0.21 and in comparison with the 95.67% of the male population with normal vision and allelic frequency of 0.79. Of the 1176 females in the population, 1.02% exhibit deficiency in colour vision with an allelic frequency of 0.10 as opposed to the other 98.98% with normal vision and allelic frequency of 0.90.

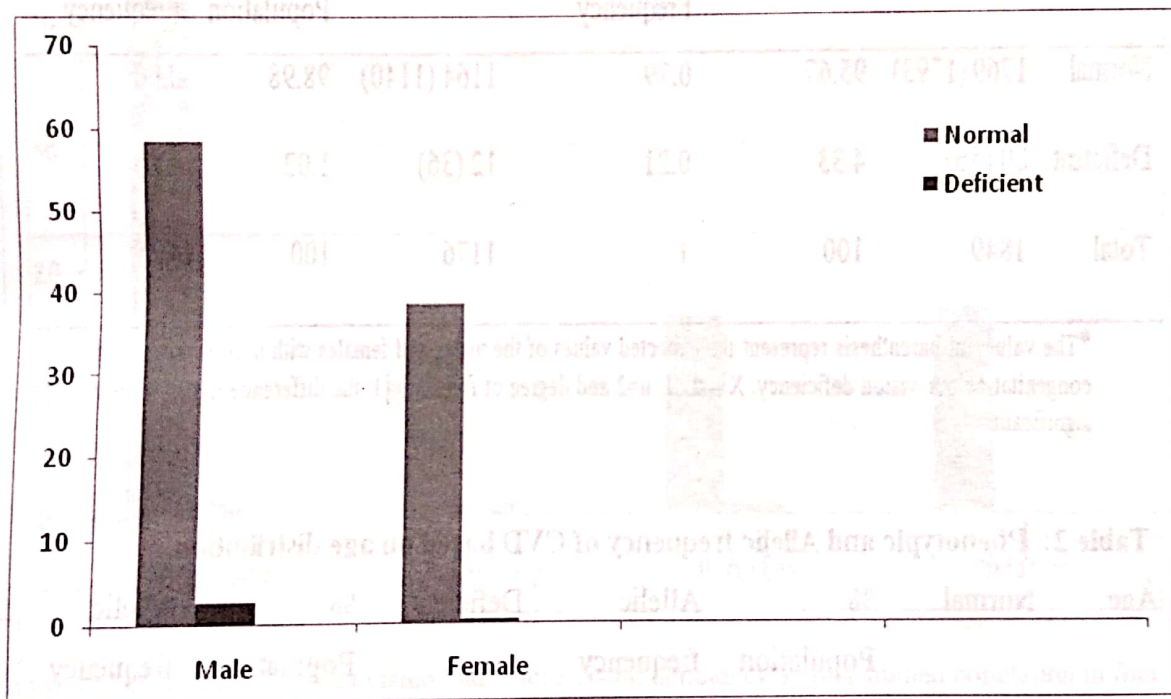


Figure 1:Prevalence of male and females that have normal vision and CVD in the overall population.

Table 2 shows the phenotypic and allelic frequency of colour vision deficiency based on age distribution. Out of 170 students in the 0-10 demographic, 1.18% showed deficiency in colour vision and allelic frequency of 0.11. 3.13% of the 2842 students in the 11-20 demographic were

colour vision deficient with allelic frequency 0.18. The 13 students with ages ranging from 21-30 in the sample population showed 7.69% colour vision and allelic deficiency of 0.28.

Figure 2 shows the percentage of normal vision and colour vision deficiency in the

sampled Local governments in the population. Ilorin South Local Government showed the highest percentage of colour deficiency of the 3025 students sampled. It showed that 1.42% of the total sample population is colour vision deficient. The other local governments (i.e. Ilorin West, Ilorin East and Ifelodun) showed 0.86%, 0.69% and 0.23% colour vision deficiency, respectively.

Table 1: Prevalence of CVD and Allelic Frequency Based On Gender

Gender	Males			Females		
	Population	% Population	Allelic Frequency	Population	% Population	Allelic Frequency
Normal	1769 (1793)	95.67	0.79	1164 (1140)	98.98	0.90
Deficient	80 (56)	4.33	0.21	12 (36)	1.02	0.10
Total	1849	100	1	1176	100	1

*The values in parenthesis represent the expected values of the males and females with normal vision and congenital colour vision deficiency. $X^2 = 27.1$, $n=2$ and degree of freedom=1, the difference is statistically significant.

Table 2: Phenotypic and Allelic frequency of CVD based on age distribution.

Age	Normal	% Population	Allelic frequency (C)	Deficient	% Population	Allelic frequency (c)
0-10	168 (165)	98.82	0.89	2 (5)	1.18	0.11
11-20	2753 (2756)	96.87	0.82	89 (86)	3.13	0.18
21-30	12 (12.6)	92.3	0.72	1 (0.4)	7.69	0.28

*The values in parenthesis represent the expected values. The $X^2 = 2.9$ and $df = 2$. The values are significantly different at $p = 0.20$

Table 3: Prevalence and allelic frequencies of males and females with normal vision and congenital colour vision deficiency in the population.

Population	Males				Females			
	Normal	C	CVD	c	Normal	C	CVD	c
Ifelodun	261 (262)	0.86	5 (4)	0.14	230 (229)	0.91	2 (3)	0.09
Ilorin South	986 (998)	0.79	45 (33)	0.21	518 (506)	0.90	5 (17)	0.10
Ilorin West	238 (245)	0.71	20 (13)	0.29	207 (200)	0.88	3 (10)	0.12
Ilorin East	284 (287)	0.81	10 (7)	0.19	209 (206)	0.90	2 (5)	0.10

*the values in parenthesis are the expected values of the population. $X^2 = 26.1$, $df=9$ at value $p=0.002$, the difference is significant.

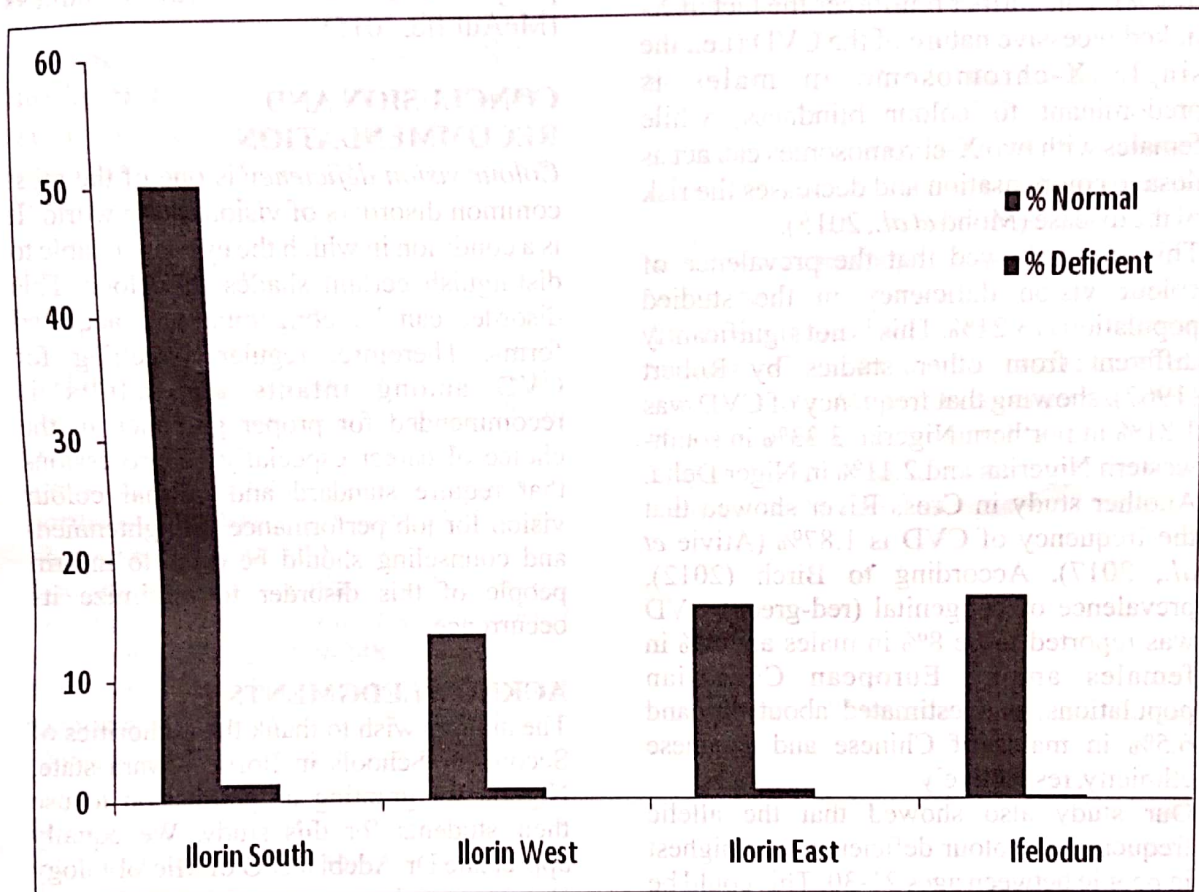


Figure 2: Percentage of normal vision and colour vision deficiency among studied population in four different local governments in Kwara State, Nigeria.

Table 3 shows the allelic frequency of the congenital colour vision deficiency in the Ilorin West, Ilorin South, Ilorin East and Ifelodun local governments in Ilorin. The allelic frequency of congenital colour vision deficiency was highest in males in Ilorin West (0.29) and lowest (0.09) in females in Ifelodun compared to other local governments.

DISCUSSION

This study gives a comprehensive account of the prevalence and gene frequency of congenital colour vision deficiency in Ilorin East, Ilorin West, Ilorin South and Ifelodun local governments in Kwara State. Our results showed that the congenital colour vision deficiency was more prevalent in males than females. Results from this study do not deviate from a study carried out in

Lagos which showed that the prevalence of CVD is higher in males (3.60%) than in females (0.81%) (Williams *et al.*, 1998), which suggests that the presence of an extra X chromosome in females decreases the prevalence of the deficiency unlike males that possess just one X chromosome. Another study in Ibadan by Ugalahi *et al.* (2016) showed that CVD prevalence is higher in males (3.8%) than in females (0.9%). This further reinforces the fact of X-linked recessive nature of the CVD (i.e., the single X-chromosome in males is predominant to colour blindness, while females with two X-chromosomes can act as dosage compensation and decreases the risk of the disease (Mohd *et al.*, 2015).

This study showed that the prevalence of colour vision deficiency in the studied population is 3.21%. This is not significantly different from other studies by Robert (1967), showing that frequency of CVD was 1.81% in northern Nigeria, 3.33% in south-western Nigerian and 2.11% in Niger Delta. Another study in Cross River showed that the frequency of CVD is 1.87% (Ativie *et al.*, 2017). According to Birch (2012), prevalence of congenital (red-green) CVD was reported to be 8% in males and 4% in females among European Caucasian populations, and estimated about 4% and 6.5% in males of Chinese and Japanese ethnicity, respectively.

Our study also showed that the allelic frequency of colour deficiency was highest in people between ages 21-30. This could be due to the risk of eye defects increasing with age. A study carried out in northeast Iran showed that the prevalence of CVD has increases in individuals in older ages because they have a higher risk of developing ocular defects like glaucoma, diabetes and are exposed to environmental factors that can increase the risk of CVD (Hashemi *et al.*, 2017).

Since colour blindness is a non-fatal disorder, people are unaware of this defect. A study in Central Ethiopia reported that only 0.19% of the students screened were aware that they were colour deficient and overall only one student reported that he had

undergone eye examination at least once in his life time (Mulusew and Yilikal, 2013). Although CVD cannot be treated, several therapies have been proposed (e.g. electrical eye stimulation, Iodine injections, large doses of vitamins) (Natu, 1987). They could also use lenses to help them perceive colors more accurately (Badawy *et al.*, 2018). Visual aids could also be employed to help people cope with colour blindness (McAuliffe, 2015).

CONCLUSION AND RECOMMENDATION

Colour vision deficiency is one of the most common disorders of vision in the world. It is a condition in which the eyes are unable to distinguish certain shades of colour. This disorder can be congenital and acquired forms. Therefore, regular screening for CVD among infants and adults is recommended for proper guidance in the choice of career especially on professions that require standard and normal colour vision for job performance. Enlightenment and counseling should be done to inform people of this disorder to minimize its occurrence.

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