Prevalence and Allele Frequency of Red-Green Color Vision Defects among School Children in Repi Primary School in Addis Ababa, Ethiopia

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Research Article

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Abstract

Background

X-linked red-green color blindness is the most widespread form of vision impairment. The study aimed to determine the prevalence and allele frequencies of red-green color vision impairments among school children in Repi primary school in Addis Ababa, Ethiopia.

Methods

A cross-sectional survey was employed involving 2400 healthy subjects (females = 850 and males = 1550) in Repi primary school. The Ishihara pseudo-isochromatic 24 plate edition was used to test the colour vision of students under natural day light condition.

Results

The prevalence of congenital colour vision deficiency (CCVD) was 2.29%; 50 (3.23%) males and 5 (0.56%) females were affected with CCVD. The frequencies of achromacy, deutan and protan in male subjects were 0.13%, 1.75%, and 0.50%, respectively. Both Deutan and protan were highest among students of Amhara ethnic origin 2.85 % and 1.08%, respectively.

Conclusions

The overall prevalence of CCVD found in the present study was lower compared to the previous studies done in Ethiopia. There was clear variation in the prevalence of color vision deficiency among students of various ethnic groups. Proper screening, education and counseling are needed to minimize impacts of CCVD in the country, and can also be beneficial for the affected subject in tackling difficulties in everyday work and for proper choice of future profession.

Background

Three classes of cone photoreceptors lining the retina of the eye, namely, the blue or short wave –sensitive (S), the green or middle –wave sensitive (M), and the red or long (L) wave sensitive determine colour vision in humans [1, 2]. The three major classes of congenital colour vision deficiency are protan, deutan and tritan types. Recessive mutations and rare genomic rearrangements on the long arm of the X chromosome encode the gene products that causes protan and deutan defects [3, 4]. However, genomic mutations encoding blue retinal cone pigment located on the autosomal chromosome 7 are responsible for tritan defects [1, 2]. None congenital colour vision defects could be due to neurological disease, metabolic disorders, drug toxicity, drug overdose etc [2, 5].

The benefits of analyzing the phenotypic and allelic frequencies of congenial colour visions population wise are manifold, for example, occupations/jobs and in routine life that involve precise colour matching to mention few [6]. Managing and organizing data on the prevalence and causes colour vision defects population wise are crucial for vision defect prevention and control; career choice, and prioritization of research activities depending on the severity of problems in the various groups. It is recommended that school children should undergo early diagnosis of colour vision defects to adjust better to tasks at school and to make career choice [2, 5]. To date the number of studies done in Ethiopia, to assess the prevalence of congenital colour vision deficiency are hardly few. Therefore, the objective of the present study was to assess the prevalence congenital colour vision deficiency and allele frequencies among school children at Repi primary school in Addis Ababa, Ethiopia.

Methods

Study area and study subjects

Descriptive cross-sectional survey was conducted to determine the prevalence of congenial colour vision deficiency. The study was done at Repi primary school is located in Addis Ababa; focusing on second cycle in grade 7 and 8 students who have normal eye vision condition and who gave their consent were included in the study. A total of 2400 students participated in this study out of which 850(35.42%) were females and 1550 (64.58%) were males with an age range from 12 to 16 years with mean age of 15 years. As the sample sizes of the some ethnic groups were small, we had combined them together to have a balanced sample size.

Colour Blindness Test

The Ishihara pseudoisochromatic plates were used to evaluate the colour vision of students under natural day light condition [7]. The number plates of the Ishihara chart were used and the abnormality is differentiated accordingly. The test was done in adequate lighted room resembling natural day light. Testing was carried out by holding the plates 75 cm away from the student and tilted at right angle to the line of vision. Each student was then

asked to read the numbers seen in the test plates 1 to 17 in less than 5 seconds and answers were noted down. The normality or defectiveness of color vision and also the type of color blindness were determined by the assessment of the reading of the plates. Thus, an assessment of the reading of plates 1 to 15 determines the normality or defectiveness of color vision. If 13 or more plates are read correctly, the color vision is regarded as normal. If only 9 or fewer plates are read correctly, the color vision was regarded as red-green deficient. Achromacy subjects read correctly plate one only. The plates 16 and 17 are used to differentiate deutan and protan types of colour vision deficiency.

Allele Frequency Analysis

Assuming that the populations are non-consanguineous, the frequencies of the normal allele (p), deutan allele (q) and protan allele (r) for color blind subjects were calculated based on the Hardy–Weinberg law using the gene counting method as was done by [2]. The expected allele frequencies of females were calculated based on the affected males allele frequencies but the actual allele frequencies of females were calculated based on the affected females as performed by Mitiku *et al.*[2].

Results

Phenotypic frequency of congenital colour vision deficiency

In the present study the prevalence of CCVD in the overall population, male and female subjects were 2.29% (95% CI: 1.8 to 2.78), 3.23% (95% CI: 2.8 to 4.00) and 0.56% (95% CI: 0.34 to 0.84), respectively (Table 1). Among the male subjects the highest frequency was recorded for Guraghe followed by subjects from Silte ethnic origin (Table 1). Colour blindness was recorded among subjects from 8 ethnic origins, namely; Amhara, Dorze, Gurage, Kembata, Oromo, Silte, Tegar and Wolaita (Table 1). The frequencies of achromacy, deutan and protan were: 0.13%, 1.75% and 0.50%, respectively (Table 2). The distribution of achromacy, deutan and protan among the male subjects were: 0.19%, 2.45% and 0.77%, respectively (Table 2). The distribution of achromacy, deutan and protan among the male subjects were: 0.19%, 2.45% and 0.77%, respectively (Table 2).

Table 1
The prevalence of congenital color vision deficiency by gender

Ethnic groups	Male			Female			Combined		
	Normal (%)	Affected (%)	Total	Normal (%)	Affected (%)	Total	Normal (%)	Affected (%)	Total
Amhara	352(95.91)	15(4.09)	367	94(97.91)	2(2.08)	96	446(96.33)	17(3.67)	463
Oromo	415(97.19)	12(2.81)	427	276(99.28)	2(0.72)	278	691(98.01)	14(1.99)	705
Dorze	387(1.77)	8(2.03)	395	95(100.00)	0 (0.00)	95	482(98.37)	8(1.63)	490
Guraghe	74(93.67)	5(6.33)	79	86(100.00)	0 (0.00)	86	160(96.97)	5(3.03)	165
Silte	161(95.83)	7(4.17)	168	157(100.00)	0 (0.00)	157	318(97.84)	7(2.15)	325
Other*	108(97.30)	3(2.70)	111	138(0.99)	1(0.01)	139	246(98.4)	4(1.60)	250
Total	1497(96.77)	50(3.23)	1547	845(99.41)	5(0.56)	850	2342(97.71)	55(2.29)	2397
*Awi (4/0), Hadiya (25/0), Kembata (75/1), Tegar(65/2)and Wolaita (80/1). The numbers in the parenthesis are sample size and CCVD affected subjects of the respective ethnic group.									

Table 2
Phenotypic frequency of achromacy and the different types of CCVD among male and female students of various ethnic groups

Ethnic Group	Male				Female				Combined			
	Achromacy (%)	Deutan (%)	Protan (%)	Total	Achromacy (%)	Deutan (%)	Protan (%)	Total	Achromacy (%)	Deutan (%)	Protan (%)	Total
Amhara	2(0.54)	10(2.71)	5(1.36)	369	0(0.00)	2(2.08)	0(0.00)	96	2(0.43)	12(2.85)	5(1.08)	465
Oromo	1(0.23)	9(2.10)	3(0.70)	428	0(0.00)	2(0.2)	0(0.00)	278	1(0.142)	11(1.56)	3(0.42)	706
Dorze	0 (0.00)	7(1.77)	1(0.25)	395	0 (0.00)	0(0.00)	0 (0.00)	95	0(0.00)	7(1.43)	1(0.20)	490
Gurage	0 (0.00)	4(5.06)	1(1.2)	79	0 (0.00)	0(0.00)	0 (0.00)	86	0(0.00)	4(2.42)	1(0.61)	165
Silte	0 (0.00)	6(3.5)	1(0.60)	168	0 (0.00)	0(0.00)	0 (0.00)	157	0(0.00)	6(1.85)	1(0.31)	325
Others	0(0.00)	2(0.00)	1(0.00)	111	0(0.00)	1(0.00)	0(0.00)	138	0(0.00)	2(0.80)	1(0.40)	249
Total	3(0.00)	38(0.00)	12(0.00)	1550	0(0.00)	5(0.00)	0(0.00)	850	3(0.13)	43(1.75)	12(0.50)	2400

Allele Frequency And Heterozygosity

The frequency of CCVD allele (Table 3) in the present study is lower compared to males in some Indian populations [10] but the frequency is comparable to recent study done in Hawassa, Ethiopia [2]. Similarly, the level of heterozygosity found in the present study among female students of Guraghe, Amhara and Oromo ethnic origin was low compared to that of the population in India [10]. The average heterozygosity for females in the present study was lower compared the prevalence in a general population [17] and a recent study in Hawassa [2]. Furthermore, the frequency of double heterozygote among females in this study is lower relative to that Mitiku *et al.* [2]. Females who had a recessive allele for deutan on one X chromosome, but a recessive allele for protan on the second X chromosome are called compound heterozygote [17]. The expected frequency of the compound heterozygote female in the present study is lower compared to the frequency in the general population [17]; and among university students [2]. Similarly, the expected frequency CCVD among the females lower relative to that of the general public (0.39%)[17].

Table 3 Genotype frequency distribution among male and female

ethnic groups	Gender								
	Male				Female				
	Normal	Deutan	Protan	Total	Deutan	Protan	Heterozygote	Double heterozygote	Color blind
Amhara	352 p =0.96	10 q =0.03	5 r=0.01	367	q ² =0.0009	r ² =0.0001	2(pq+pr) =0.0768	2qr=0.001	q ² + r ² =0.001
Oromo	415 p =0.97	9 q =0.02	3 r =0.01	427	q ² =0.0004	r ² =0.0001	2(pq +pr) =0.0582	2qr=0.0004	q ² + r ² =0.001
Dorze	387 p =0.98	7 q =0.02	1 r=0.00	395	q ² =0.0004	r ² =0.000	2(pq +pr) =0.039	2qr=0.000	q ² +r ² =0.0004
Guraghe	74 p =0.94	4 q=0.05	1 r =0.01	79	q ² =0.003	r ² =0.0001	2(pq +pr)=0.112	2qr=0.001	q ² + r ² =0.003
Silte	161 p =0.96	6 q =0.03	1 r =0.01	168	q ² =0.0009	r ² =0.0001	2(pq +pr)=0.0768	2qr=0.001	q ² + r ² =0.001
Others	108	2	1	111	q ² =0.0004	r ² =0.0001	2(pq +pr)=0.0582	2qr=0.0004	q ² + r ² =0.001
Total	P=0.97 1497 p =0.97	Q=0.02 38 q =0.02	R=0.01 12 r =0.01	1547	q ² =0.0004	r ² =0.0001	2(pq +pr)=0.0582	2qr=0.0004	q ² + r ² =0.001

Discussion

Phenotypic frequency of congenital colour vision deficiency

The overall incidence of CCVD in this study is comparable to the prevalence rates of 2.2 % in Durban, South Africa [8], and slightly higher than 1.5% the report among school children in Zaria, Northern Nigeria [9]. However, it is lower compared to incidence rates 5.28 % in Manipur, India [10], 3.28% in Shekhan City/Kurdistan region, Iraq[11] and 2.85% among university students [2]. The difference in the incidence of CCVD from the current study could be due to difference in study populations, cultural/religious, geographical, ethnicity and techniques of color vision test [2, 5, 6, 10, 12].

The prevalence CCVD in male was lower relative to 3.6 % in Wolkite town [13], male school children Pokhara, Western Nepal [14] and Harari (Ethiopia) [12]. Male students of Guraghe ethnic showed the highest prevalence of color blindness followed by Silte, while the students from Dorze ethnic showed least incidence (Table1). The higher incidence in Guraghe and Silte could be due to the practice of some degree of consanguineous marriage in these ethnic groups albeit the degree of such marriage is rare Ethiopia [2, 12]. The prevalence of color blindness for females is generally low and the finding of this study is no exception. The incidence of CCVD observed in females is lower compared to females in some Indian populations [10] and in Qazvin (Iran)[15] but is larger compared to females in Saudi Arabia [16]. The higher prevalence among males as compared to females shows the genetic causation of the disorder. It is a sex linked recessive trait with defective gene on X-chromosome. Therefore, females may be carriers but are rarely affected because they possess two X-chromosome. On the other hand, males are more prone to develop these disorders as they possess one X-chromosome. Genetically, it reinforces the fact of X-linked recessive nature of the defect [2, 17, 18].

The frequencies of achromacy, deutan and protan in male subjects were (0.13%), (1.75%), and (0.50%), respectively (Table2). These frequencies are relatively low compared to recent study done by Mitiku *et al.* [2] among university students at Hawassa, Ethiopia. A comparable frequency of deutan and protan are reported in done in Gish–Abay town by Wale *et al.* [19]. The slight difference in the prevalence could be due to genetic, geographic, population movements and cultural/religious factors [2, 12, 18]. Deutan cases occur in higher percentage than protan for both male and female subjects. Male subjects from Guraghe ethnic origin had the highest dutan prevalence of followed by Silte, while the subjects from other ethnic combined population showed zero prevalence. The prevalence of deutan in males the present study is lower relative to earlier work done among school children in Abeshge district, Ethiopia [5]. The differences in the frequencies could be attributed to massive population movements and intermixing with other ethnic in metropolitan areas such as Addis Ababa.

The prevalence of CCVD in the present study was lower compared to population in the Middle East, North Africa and Southeast Asia. The lower frequencies of various types of CCVD in the present study compared to populations in the Middle East [20, 21], in North Africa [22,23] and in Southeast Asia [10, 20], could be due to the common practices of consanguineous marriage these countries/regions. In Ethiopia consanguineous marriage is rare as it is not promoted because of cultural, religious and legal factors [2, 5, and 12]. Factors like population movements, the molecular structure the gene on the X chromosome and natural selection could also contribute to the difference in the prevalence of CCVD between population and geographic regions [2,22,23]. Population movements between regions and within countries could lead in the rise of the magnitude of exogamous union that that might contribute the increase the incidence of CCVD.

Allele frequency and heterozygosity

The frequency of CCVD allele (Table3) in the present study is lower compared to males in some Indian populations [10] but the frequency is comparable to recent study done in Hawassa, Ethiopia [2]. Similarly, the level of heterozygosity found in the present study among female students of Guraghe, Amhara and Oromo ethnic origin was low compared to that of the population in India [10]. The average heterozygosity for females in the present study was lower compared the prevalence in a general population [17] and a recent study in Hawassa [2]. Furthermore, the frequency of double heterozygote among females in this study is lower relative to that Mitiku *et al.* [2]. Females who had a recessive allele for deutan on one X chromosome, but a recessive allele for protan on the second X chromosome are called compound heterozygote [17]. The expected frequency of the compound heterozygote female in the present study is lower compared to the frequency in the general population [17]; and among university students [2]. Similarly, the expected frequency CCVD among the females lower relative to that of the general public (0.39%)[17].

Colour Blindness and career choice

Data on prevalence of CCVD are useful for identifying needs for treatment and rehabilitation services, planning, career choices, and focusing research priorities [2, 5, 12, 24,25]. Undiagnosed color vision defect could pose a handicap to the performance of affected students. Color blindness is a nonfatal disorder; therefore colour blind people usually remain unaware of the defect since their vision is otherwise normal [2, 24]. In this study the awareness of school children about colour vision status was low. This intern could affect students in future career choice. Normal color vision is important for our daily life work such as to recognize the traffic signals during crossing roads or to build career in several professions like engineering, soil sciences, chemistry, electricity and electronics, etc [10,24, 25].

Conclusions

The overall prevalence of congenital colour vision deficiency in the present study was low compared to earlier studies in Ethiopia. School children's awareness about CCVD was very low. Colour vision defects may remain undetected and school children remains unaware about the disability. Such

undetected defects could cause problems during learning which then resulted into a decline in interest and lesser efficiency in work, preventing the students to reach up to their maximal potential. Therefore, establishing continuous visual screening programs for school children is of paramount importance to minimize the potential impacts of colour vision defects on school children learning capacities. Anomaloscope, the gold standard in color vision test, was not used as it was not available in our set up.

List Of Abbreviations

CCVD

congenial colour vision deficiency

IRB

Institutional Review Board

Declarations

Ethics approval and consent to participate

The study was conducted after ethical approval of the Institutional Review Board (IRB) of Hawassa University (Ethiopia), College of Natural and Computational Sciences; and after informed consent letters were obtained from legal guardians and principal of the school after explanation about the procedures of the test. Currently, the reference number ethical clearance letter is not available but if need be we could request the committee and present the reference number in the future.

Consent for publication

Consent for publication is not applicable as this study did not include names, images, or videos relating to individual participants.

Availability of data and materials

The data used in this study are available from the corresponding author on reasonable request.

Competing interests

We declare that there are no conflicts of interest amongst the co-authors. All the co-authors have contributed from the very inception of the manuscript and until the final version of the manuscript.

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Authors' contributions

WAM was involved in the study design and manuscript writing and revision; AGM was involved in data collection and analyses and ZGT was involved in the study design, data analyses, manuscript writing and revision. All authors read and approved the manuscript.

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