

To Study Prevalence and Genetic Pattern of Inheritance of Colour Vision Deficiency in School Going Children

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Abstract: Colour Vision is the ability to discriminate a light stimulus as a function of its wavelength. Colour vision deficiency is mainly of two types. The congenital anomaly is common chromosomal abnormality with an 'X - linked recessive pattern'. The acquired causes of colour blindness, such as damage to the eyes, nerves, brain, some metabolic disorders like diabetes, glaucoma, macular degeneration, chronic illness like sickle cell anemia, toxins, drug over dose such as digoxin, barbiturates, anti TB drugs, drug side effects like sildenafil and ethambutol. Human colour vision is classified according to Young Helmholtz Trichromatic Theory. The colour vision deficiency individuals have difficulty in comprehension due to increased reaction time. The Ishihara colour test is used to determine colour blindness using different colored and patterned plates. A total of 3172 students were chosen from various schools in Davanagere from class 5th to 10th and were examined for both visual acuity and colour blindness with the help of Snellen chart and Ishihara chart respectively with standard protocol and adequate lighting. If the students were found to be colour blind, then parents and siblings of those students were asked for consent and examined by same method. The results of the study out of 3172 students 66 students were found to be colour vision deficient using the Ishihara charts 38 plate edition. Among the 66 colour defectives, males were predominantly affected 63 of 66 (95.45%) as compared to females which was 3 of 66 (4.55%) with p value <0.000 which is highly significant. Protanopia (87.88%) was more prevalent than deuteranopia (10.61%). 1 girl was found to be achromatic (1.51%). After screening the parents and siblings of 31 colour defective students, males were found to be predominantly affected. Prevalence was more in consanguineous marriage when compared to non - consanguineous marriage.

Keywords: Colour Vision Deficiency, Genetic Pattern, Prevalence of CVD, School Children, Ishihara chart

1. Introduction

Colour vision is the ability to discriminate a light stimulus as a function of its wavelength. Light with wavelength between 360nm and 720nm causes a photoreaction on the human retina which leads to colour vision.¹ The term Colour Blindness was discovered by David Brewster and was formerly known as "Daltonism".² This was classified according to "Young Helmholtz Trichromatic theory".³

Colour vision deficiency is divided into Congenital and Acquired forms.⁴ The congenital anomaly is a common chromosomal abnormality in man with an "X linked recessive pattern", thus manifesting in the male offspring of a female carrier while female offspring are usually carriers.⁵

Human colour vision is normally trichromatic and requires at least 3 cone photo pigments, one from each of 3 well separated spectral classes and are referred to as RED, GREEN, BLUE pigments.⁹ Hence these are the Primary colours. Colour Blindness is classified into: Anomalous Trichromatism and Dichromatism.⁶ Trichromatism is further divided into Protanomaly (mild red blind), Deuteranomaly (mild green blind), Tritanomaly (mild blue blind).¹ Dichromatism is when ability to perceive one of the colours is completely absent. If red is absent Protanopia, Green is Deuteranopia, Blue is tritanopia.⁶

In general Red Green defects shows the highest prevalence.⁷ The Ishihara colour test is a test to determine colour blindness¹⁰ using different coloured and patterned plates. It is the most widely used screening test for Red Green deficiency.⁶

This study will not only give a clear idea about the prevalence rate of congenital colour blindness in children of Davanagere but will help detect many unknown cases and will help spread awareness regarding the disorder and its importance. This will help teachers and parents understand better regarding the child's learning disabilities, reassure the child, adopt and apply new methods of teaching, provide supportive measures, guidance in choosing and adjusting with the suitable profession etc. Hence, this study was done to determine the prevalence rate of Congenital Colour Blindness in school going children of Davanagere and their genetic pattern of inheritance.

2. Review of Literature

Colour vision deficiency (CVD) or Colour Blindness is the inability or decreased ability to distinguish different colours under normal lighting conditions.¹³ The ability to have trichromatic vision distinguishes humans from nonprimates.⁹

Colour blind individuals have difficulty in comprehension due to increased reaction time.¹⁰ Colour vision deficiency does not cause complete blindness and there is no available therapeutics that treat Colour vision deficiency.¹⁴ Congenital colour vision deficiency is an X chromosome linked recessive, autosomal dominant and very rarely autosomal recessive inherited trait.¹⁵ A recent study has revealed the missense mutation in a hybrid L/M cone opsin gene leading to X linked cone dystrophy and colour vision deficiency.¹⁶ There are acquired causes of colour blindness, such as damage to the eyes, nerves, brain, some metabolic disorders like diabetes, glaucoma, macular degeneration, chronic illness like sickle cell anaemia, toxins, drug over dose such as digoxin, barbiturates, anti TB drugs, drug side effects like sildenafil, ethambutol etc.¹⁷

A high proportion of school children are unaware of their colour vision status and undiagnosed CVD could pose a handicap to the scholarly performance of an affected student.¹⁸ Colour blind persons unaware of their disability, may choose professions, which may not be suitable for them like traffic policemen, train driver or technician in colour industries, which require proper colour perception leading to lesser efficiency in work as well as may cause accidents.⁷ The Ishihara colour test is a test to determine if a person has colour blindness.¹⁰ It is the most widely used screening test for red - green colour deficiency.¹⁹

Objectives:

- 1) To determine the prevalence rate of colour blindness.
- 2) To know the different types of colour blindness and the most prevalent type.
- 3) To study the prevalence rate between male and female separately and compare.
- 4) To know the pattern of genetic inheritance.

Sample size: 3172 students.

Inclusion Criteria:

Students of class 5th to 10th of various schools of Davangere (even students with myopia of less than - 6D)

Exclusion Criteria:

Recent trauma to eye, any eye injuries, congenital or acquired obvious ocular pathologies, high progressive myopia of - 6D and above.

3. Methodology

A total of 3172 students from various schools in Davangere of class 5th to 10th will be chosen. Consent will be taken and they will be called roll number wise to a room in the respective school where clinical examination of both eyes will be done to check for VISUAL ACUITY and COLOUR BLINDNESS. Visual acuity will be checked using the Snellen Chart and Colour deficiency will be tested using the Ishihara's type tests for colour blindness, 38 plates edition.

Colour vision testing plates will be held at 75cm from the student and tilted at right angle to the line of vision. Test will be conducted for all students in similar settings, it being adequately lighted room resembling natural day light. Student will be asked to read numbers seen on test plates and answers will be noted down. Time given for telling the number on the plate will be 5 seconds.

Assessment of the reading of the plates will be made and results will be given as per the instructions given are identified to be colour blind will undergo a repeat examination. If positive, then parents and siblings of those students will be asked for consent and then examined by the same method described above and results will be obtained. A pedigree chart will be then drawn to study the genetic inheritance pattern.

Prevalence rates in males and females will be expressed separately and will then be compared. Prevalence rates will be interpreted in the form of percentage and calculated using the formula:

(Prevalence= Number of all current cases of a specific disease existing at a given point x 100/Estimated population at the same point in time)

4. Results

A total of 3172 students from 6 different schools of Davangere were screened out of which 1453 were females and 1719 were males. Using the Ishihara colour vision deficiency 38 plates edition 66 of 3172 students (2.08%) were found to be colour blind.

Among the 66 colour defectives, males were predominantly affected that is 63 of 66 (95.45%) as compared to females which was 3 of 66 (4.55%) with p value <0.001 which is highly significant. The two types of Red Green deficiency seen was Protanopia and deuteranopia. Protanopia (87.88%) was more prevalent than deuteranopia (10.61%). 1 girl was found to be achromatic (1.51%).

Amongst the 58 protanopes 56 were males and 2 females with p<0.01 which is significant, 7 deuteranopes in which all 7 males and no females. Hence, protanopia is more common than deuteranopia.

After screening the parents and the siblings of the colour defective students for tracing the genetic pattern of inheritance, males were found to be predominantly affected as compared to females which confirm the possibility of X linked recessive pattern implying males as affected and females mostly are carriers. In many of the cases of consanguineous marriages both the children were found to be colour defective showing its relation to genetic inheritance.

Table 1: Distribution of Colour Blindness gender wise

Colour Blindness	Gender		Total	Chi Square Test	
	Male	Female		P Value	Significant
Affected	63 (3.66%)	3 (0.21%)	66 (2.04%)	P<0.001	Highly Significant
Not Affected	1656 (96.3%)	1450 (99.79%)	3106 (96.91%)		
Total	1719	1453	3172		

Table 2: Distribution of Protanopia and Deuteranopia

Colour Blindness	Male (n=63)	Female (n=3)	Total	Chi Square Test
Protanopia	56	2	58	P<0.01 Significant
Deuteranopia	07	0	07	P>0.05, Not Significant
Achromacy	0	1	01	P<0.001, Highly Significant

5. Discussion

Colour can be described in terms of hue (which is determined by the wavelength) and saturation (determined by the amount of white light mixed). Colour blindness is a nonfatal disorder; therefore colour blind people usually remain unaware of the defect since their vision is otherwise normal.

Congenital defect are usually bilateral and other visual functions usually not affected. But they are unfit for jobs which require precise colour perception which leads to decreased efficiency and accidents. They may even get rejected in interviews for such jobs which put them through an emotional turmoil. Defective colour vision even hinders the scholastic performance of children. This study even focused on the visual acuity and was found that a large number of children have refractive error and even children with visual acuity 20/200 did not have correction of any form.

They were made aware of the possible corrections in the form of spectacles and surgical treatment like LASIX. Anterior segment abnormalities were also examined for and strabismus, dry eye and conjunctivitis were mostly seen.

This study mainly focused on prevalence of colour vision deficiency which was tested using the ISHIIHARA COLOUR PLATES screening method. This method is not diagnostic but a very effective mass screening method with minimal possibility of false positive and false negative.

This study even made possible to create awareness amongst students, parents and teachers about Colour vision deficiency and its effects which was a totally new concept to the majority of population. This can help provide the colour defective children the support they need and adapt to newer better methods of teaching and guide them regarding the career options and choices.

In this study 66 among the 3172 students (2.04%) screened were found to be colour defective, males were predominantly affected that is 63 of 66 (95.45%) as compared to females which was 3 of 66 (4.55%) with p value <0.01. Protanopia (87.88%) was more prevalent than deuteranopia (10.61%). 1 girl was found to be achromatic (1.51%). Amongst the 58 protanopes 56 were males (96.55%) and 2 females (3.44%), 7 deuteranopes in which all 7 males (100%) and no females. Hence, protanopia is more common than deuteranopia.

This was similar to the study conducted by Alla Venkata Pitchi Reddy et al where the prevalence was found to be 1.9%, males predominantly affected 90.3% as compared to females 9.7%. Protanopia (90.3%) was more prevalent than deuteranopia (9.7%). Moudgil et al showed the prevalence of colour blindness to be 1.89% which included 1.69% males and 0.184% females. It shows that 90.2% were protanopes and 9.8% were deuteranopes. Among the protanopes, 92.7% were males and 7.3% were females and in deuteranopes percentage of males affected was 66.7% and females were 33.33%. Males were affected more than females.

A study by Balasundaram R, Reddy SC found colour deficiency in total 45 persons (3.2%) which includes 42 males (6.7%) and 3 females (0.4%). Mohd Fareed et al observed the prevalence of CVD which was observed to be 7.52% in males and 0.83% in female children in Jammu province. B Dakshayani and MR Gangadhar reported percentage frequency of red green colour blindness was 1.12% in Hakkipikkis of Mysore district of Karnataka state.

According to our present study and in comparison with other similar studies the prevalence of colour blindness is higher in males when compared to females. The prevalence is more in case of consanguineous marriages when compared to nonconsanguineous marriages thereby reflecting the connection of the disease to genetic inheritance mostly X linked recessive inheritance pattern.

The government must take initiative for colour blind awareness among the population. Education, screening and prenatal counselling for the disease could help in minimizing the occurrence of the disorder. Awareness among the family and society regarding the disorder and necessary support and attention is required for the healthy development of mental status of the individual suffering from this disorder. Programmes regarding career choices should be conducted.

6. Conclusion

Comprehensive assessment of colour vision impairments provides genderbased differences and prevalence of CVD was found higher among males (3.66%) as compared to females (0.21%) children. The screening was done using the Ishihara 38 plate edition. The test provided efficient sensitivity and specificity. Protanopia and Deuteranopia were identified where protanopia was more prevalent (87.88%) than deuteranopia (10.61%). It mainly affects the male children and more in consanguineous marriages affirming that the disorder is mainly genetic and it has X linked recessive pattern of inheritance.

The disorder does not cause complete non perception of colours but it does hinder the day to day activities. It affects the scholastic performance of the children and even affects the efficiency of the individual when involved in occupations like traffic policeman, histopathologists, pilots etc may even cause accidents so it is important to spread awareness regarding CVD about which majority of the population is unaware or less aware. It is important for the individual to know about his/her colour vision status and be given proper moral support and career counselling.

7. Summary

Colour vision deficiency is a common visual disorder but many individuals are totally unaware of their colour vision status. Such individuals face difficulties due to increased reaction time for perception of colours and can only experience a few hundred hues of colour instead of millions of shades of colours. The problem mainly lies in the L and M cones and its pigments. It is a genetic disorder and mainly affects the males and the daughters remain unaffected suggesting an X linked recessive pattern of inheritance.

In the present study 3172 students out of which 1453 were girls and 1719 were boys of 6 different schools were screened using the Ishihara 38 plate edition. 66 of 3172 students were colour defective.

Amongst the 66 students 63 were boys and 3 were girls showing the predominance of the disorder among males. The prevalence of protanopia was found to be more than deuteranopia and 1 girl was found to be achromatic. A majority of colour defective individuals complained of having photophobia.

Colour blindness handicaps the individuals for many day to day activities like visualizing the traffic signals and train signals etc and even for certain occupations like lab technicians, military etc who require precise colour perception. CVD causes decreased efficiency or even accidents in case of such occupations. Hence, awareness regarding CVD and its effects is important as this gives an individual a better chance at choosing an appropriate career; seek special help for scholastic requirements and even moral supports from teachers, family and society. This helps in the better mental development of the children and can grow in a healthy supportive environment being aware of their colour vision status.

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