ORIGINAL RESEARCH PAPER
Volume-6 | Issue-9 | September-2017 | ISSN No 2277 - 8179 | IF : 4.176 | IC Value : 78.46
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

DEUTRANOMALIA: THE COMMONEST TYPE RED-GREEN C0LOR VISION DEFICIENCY IN KASHMIRIS

Physiology

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ABSTRACT

Background: Color vision deficiency is an abnormal condition characterized by the inability to clearly distinguish different colors of the spectrum. **Methods:** The study was done in the Postgraduate Department of Physiology, Government Medical College Srinagar. The study was conducted in OPDs of SMHS hospital and District hospitals of Kashmir. A sample 3110 was selected. Detailed informed consent was taken from subjects / patients and relevant information regarding name, age, gender, consanguity, marital status, education, medical history and drug history was collected on structured proforma. Results: A total of 3110 participants were included in the study.1462 (47%) were males. The mean age of the study participants was 34.72 +_14.862 years.871 (28%) and 892 (28.7%) belong to the age group of 21-30 & 31-40 years respectively. Among the types of color vision deficiency, Deuteranomalia is the most common type prevalent in the Kashmiri population i.e: 52.74% followed by Protanopia (21%). The least common defect was protanomalia (11.64%).Conclusion: Our study of color vision deficiency among adult Kashmiri population shows the significant gender based difference in the frequency of color vision deficiency, with higher in males (8.5%) than in females (1.3%). The assessment of color vision deficiency by Ishihara pseudo isochromatic plates is rapid, cost efficient and sensitive in detecting red-green color vision deficiency.

KEYWORDS

C.V.D in Kashmir Valley, Deuteranomalia, C.V.D, Most Common

Introduction:
Color vision is an illusion created by the interaction of billions of neurons in our brain. Without photoreceptors and nervous system there would be neither light nor color in the universe (1).Color blindness is the commonly used term for deficiency of color vision. Many people think anyone labeled as color blind only sees black and white but this is a big misconception as it is extremely rare to be totally color blind hence more correctly called color vision deficiencies. Color vision deficiency is an abnormal condition characterized by the inability to clearly distinguish different colors of the spectrum (2).

The retina is the light sensitive portion of the eye that contains (1) the cones, which are responsible for color vision (3), and the rods, which are mainly responsible for black and white vision and vision in the dark (4).Specialized photoreceptors differentially respond to narrow band of radiation which is experienced as “colors”. Resulting “vision” provides what are perhaps the richest and most varied phenomena of all perceptual experience. Photoreceptors trigger patterns of neuronal activity that begin visual processing during their trajectory through the retina (1).

The photosensitive compound in rods and cones of the eyes of humans and most other mammals are made up of a protein called an opsonin, and retinene, the aldehyde of vitamin A. Light activates retenine and this activates Gt2, a G protein which in turn activates phosphodiesterase, catalyzing the conversion of c-GMP to 5'-GMP, decreasing Na conductance and hence hyperpolarization.5 Humans have three different kinds of cones, and each expresses a photo pigment with a different absorbance spectrum. The three cones and their pigments were historically called blue, green, and red, respectively. They are now more commonly called S, M, and L (for short, medium, and long wavelengths).Color vision depends on the different spectral sensitivities of the three types of cones (5).The difference in peak sensitivity of the normal green and red cone photo pigments can largely be accounted for by just three amino-acid residues at positions 180, 277 and 285 of the opsin molecule. Variation in residue 180 (serine or alanine) are considered to represent polymorphism (6).

TYPES
Based on John Daltons (1798) three receptor theory, color vision deficiencies are classified into three types Tri, Di and monochromates.
1) Trichromates (one cone system weak) [Anomalous trichromacy] –It is the mildest form of CVD.

a) Protanomaly (which affects red cones)
b) Deuteranomaly (which affects green cones)
c) Tritanomaly (which affects blue cones)
1) Dichromates (two cone systems weak)
a) Protanopia (no functional red cones)
b) Deuteranopia (no functional green cones)
c) Tritanopia (no functional blue cones)
2) Monochromates (only one cone system present):- rare occurrence. The severest form of CVD in which color discrimination is absent.

(Suffix 'anomaly' means color weakness and suffix 'anopia' means color blindness).(Prefix Prot, dueter and trit refers to red, green and blue color defected cone system. Thus the term Protanopia refers to inability to appreciate red color and protanomaly means weakness (less sensitivity) to red color.

Commonest is red-green defectand Common defects of red-green color vision (in order of occurrence) Deuteranomaly, Deuteranopia, Protanopia, Protanomaly, Tritanomaly (7). Classes of Color Vision Defects (8)

Class Normal	Frequency	Retinal Cones S' M' L'	Inheritance
Protanopia (severe)	1%	S' M __	X-linked recessive
Protanomaly (mild)	1%	S' M' L'	X-linked recessive
Deuteranopia (severe)	1%	S' L'__	X-linked recessive
Deuteranomaly (mild)	5%	S' L' M'	X-linked recessive
Tritanopia (mild-severe)	1/1000	__ M' L	Autosomal dominant
Blue cone monochromacy (very severe)	<1/100,000	S'__ __	X-linked recessive
Achromatopsia (very severe)	1/30,000	__ __ __	Autosomal dominant

L' cones contain anomalous L pigments that have absorption maxima close to that of normal

M' cones contain anomalous M pigments that resemble normal
S' cones contain anomalous S pigments

Table I: Classes of Color Vision defects

Colors have three attributes; hue, intensity, and saturation (degree of freedom from dilution with white). There are three primary Colors – red, green, and blue.. For every color, there is a complementary color that when properly mixed with it, produces a sensation of white. Any spectral color and even extra-spectral color (e.g. purple) can be produced by mixing various proportions of primary colors. (The paints used in painting are not pure colors but mixtures of different pigments). Black is the sensation caused by absence of light. It is probably a positive sensation, because the blind eye does not "see black", it "sees nothing". Finally, the color perceived depends on the color of other objects in the visual field (9).

Because of the high frequency of X-linked color vision defects among males, it is estimated that about 16 per cent of women are carriers of red-green color vision defects (most of whom have normal color vision) and if they marry a color defective male, they may produce color defective female offspring. However, some heterozygotes may have color vision defects due to an extremely skewed X-inactivation that by chance has inactivated most of their normal X chromosome and thus express the mutant X chromosome (10).

As predicted by inheritance patterns of red-green (11), and blue-yellow (12) color vision deficiencies, the genes for human long-wavelength (L) and middle-wavelength (M) cone opnsweresi localized to the X-chromosome at Xq28, and the gene for the short-wavelength (S) cone opsin to an autosome, chromosome 7 at 7q32 (13).The location of the color-vision pigment genes on or near Xq28 raises the problem of the existence of larger deletions affecting genes that are tightly linked to the color-vision complex. Genes in this general location code for hemophilia A and B, glucose-6-phosphate dehydrogenase (G6PD), the fragile-X syndrome, conduction deafness with stapes fixation, Emery-Dreifuss myopathy, nephrogenic diabetes insipidus, dyskeratosis congenita, chondro dysplasiapunctata, an X-linked variety of bipolar affective illness, TKCR (torticollis, keloids, cryptorchidism, renal dysplasia) syndrome, myotubular myopathy, spastic paraplegia, and adrenoleukodystrophy (ALD) (14).

Color vision tests are used for a wide variety of purposes. Some of these include rapid screening of congenital red-green defects in industry, transportation and the military. The classification of discrimination ability within the population of congenital red-green defect is used for job assignment purposes. Another use of screening involves the recognition and diagnosis of congenital disorders for psychophysical or genetic study. In the clinic, screening is used for the recognition and differentiation of congenital and acquired disorders, for classification of acquired disorders in patients of eye diseases and in some cases, for the assessment of treatment or for tracking recovery from diseases or trauma. Finally, in education and industry, screening for both color vision defects and color aptitudes is used for vocational guidance in occupation or professions that require color judgment (15).

Pseudoisochromatic plates are the most famous type of color blindness test. Most people know them under the name Ishihara plates test, because Dr. Shinobu Ishihara was one of the first persons who designed a very reliable plate test, introduced in 1917 (16). Looking at the morbidity associated with color vision deficiency, we aimed at conducting this study in Kashmir division to estimate the burden of deuteranomalia, a type of color vision deficiency in adult Kashmiri population.

Material and Methods

Settings: The study was done in the Postgraduate Department of Physiology, Government Medical College Srinagar. The study was conducted in OPDs of SMHS hospital and District hospitals of Kashmir.It was an observational cross sectional study approved by institutional ethical clearance committee Government Medical College Srinagar (J&K) India. Based on population figures of Census 2011, out of a total population, a sample 3110 was selected. All apparently healthy subjects both males and females, visiting OPD's of the SMHS Hospital and district hospitals were included in the study. Detailed informed consent was taken from subjects / patients and relevant information regarding name, age, gender, consanguity, marital status, education, medical history and drug history was collected on structured proforma. Color vision testing was done using Ishihara Pseudoisochromatic plates 38 plates (latest edition).

Statistical Analysis: Data was entered in Microsoft Excel spread sheet 2007 and analyzed using SPSS v.20.0 software. Descriptive variable were expressed by mean and Standard Deviation. Categorical variables were analyzed using Chi-square test. p value < 0.05 was considered statistically significant.

Results

A total of 3110 participants were included in the study.1462 (47%) were males. The mean age of the study participants was 34.72 +_14.862 years.871 (28%) and 892 (28.7%) belong to the age group of 21-30 & 31-40 years respectively. Only 246 (7.9%) of the study participants were having history of Consanguineous marriage. Most of the participants 2544(81.8&) were educated and 2012 (64.7%) were married. [Table II]

Variable	Frequency (n=3110)	Percentage
Sex		
Male	1462	47.0
Female	1648	53.0
Age range in years		
<= 20	423	13.6
21 – 30	871	28.0
31 – 40	892	28.7
41 – 50	583	18.7
51 – 60	341	11.0
Marriage		
Consanguineous	246	7.9
Non-Consanguineous	2864	92.1
Educational Status		
Literate	2544	81.8
Illiterate	566	18.2
Marital Status		
Married	2012	64.7
Unmarried	1098	35.3

Table II: Demographic Characteristics of the Study Participants

Table III shows prevalence of color vision deficiency disorder in Kashmir division. The estimated prevalence is 4.7% [CI: 4.57-4.83].Among the types of color vision deficiency, Deuteranomalia is the most common type prevalent in the Kashmiri population i.e: 52.74% followed by Protanopia (21%). The least common defect was protanomalia (11.64%).

Variable	Frequency	Percent
Prevalence of C.V.D.		
Present	146	4.7
Absent	2964	95.3
Total	**3110**	**100.0**
Type of C.V.D.		
Deuteranomalia	77	52.74
Protanopia	31	21.23
Deutranopia	21	14.38
Protanomalia	17	11.64
Total	**146**	**100.0**

C.V.D: Color Vision Deficiency

Table III: Prevalence of C.V.D & Type of C.V.D in Study Participants
Figure 1 shows the frequency of types of color vision deficiency in the study population

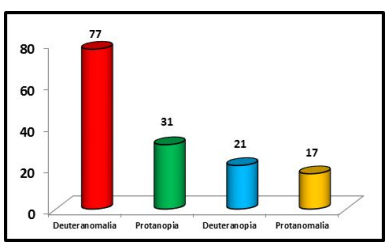

Figure 1: Frequency of Types of Color Vision Deficiency Disorders

The prevalence of color vision deficiency was highest among males (8.5%) than females (1.3%) and the relation was statistically significant with p< 0.001. Deuteranomalia was seen more among males (55%) than females (41%). One important finding to mention here is the higher prevalence of Protanopia & Protanomalia in females than men [Table IV].

Variable	Frequency	Percentage	p Value	
Sex			Chi-square=87.32 p-value<0.001	
Male	124	8.5		
Female	22	1.3		
Total	**146**	**9.8**		
Type of C.V.D	**Frequency In Males**	**Percentage**	**Frequency In Females**	**Percentage**
Deuteranomalia	68	54.8	9	40.9
Protanopia	26	21.0	7	31.8
Protanomalia	16	12.9	5	22.7
Deuteranopia	14	11.3	1	4.5
Total	**124**	**100**	**22**	**100**

C.V.D: Color Vision Deficiency

Table IV: Distribution & Type of C.V.D in Study Participants as per gender

DISCUSSION

The world of color presents no more surprising phenomenon than that of color blindness which is ever playing a mysterious back stage role in human experiences. Congenital color vision deficiency is one of the commonest inherited disorder of vision.

This study provides a description about the type of color vision deficiency in adult kashmiris.The frequency of color vision deficiency is significantly higher in males than females due to x-linked nature of deficiency. Since males have single x-chromosome while as females with two x-chromosomes can act as dosage compensation if mutation occurs. In our study Deutranomaly(mild green color defect) is the commonest type of color vision defect followed by Protanopia ,Deutranopia and Protanomaly.

]The type of color vision defect in males with decreasing order of frequency isDeutranomalia>Protanopia>Protanomalia>Deuteranopia while in females Deutranomalia>Deutranopia >Protanopia> Protano malia is the frequency.

SutenderNaresh (1995) (17) conducted a study entitled "Study of color blindness in Jat Sikhs".. The type of blindness in males in its descending order of occurrence was simple deutranomaly 0.92%, extreme deutranomaly 0.77%, protanopia 0.69%, simple protanomaly 0.61%, deutranopia 0.61%, extreme protanomaly 0.15% and tritanopia 0.08%. The sole blind female was simple deutranomaly type.

NavjotKaur, Avinash Kumar, GurinderKaur, JasjeetKaurDhillon, K. D. Singh (2011) (18) studied 1210 male and 800 female Tibetan population in Northern India between the age group of 11-60 years. The type of colour blindness in males in its descending order of occurrence was simple deuteranomaly 27.45% protanopia 17.64%, deuteranopia 17.64%, extreme deuteranomaly 15.68%, protanomaly 13.72%, extreme protanomaly 5.88% and tritanopia 1.96%. None of the female was colour blind.It is suggested that Caucasians have comparatively fewer green pigment genes than that of the Asians and the American Blacks (Cruze EM et al., 2010). It is also suggested by several researchers that green color receptors are commonly affected more than red or blue receptors.(Ahsana Shah et al, 2013).

Recommendations:

Color vision deficiency is currently under-diagnosed as the screening for color vision deficiency is not well appreciated and even Ophthalmologists neglect this aspect of vision. Increasing the awareness about color vision deficiency and its impact on various stages of life. Genetic counseling in the regions with high prevalence of color vision deficiency will decrease the birth of children with this deficiency. Population based screening for color vision deficiency is one of the most important steps to initiate appropriate measures and plan adaptive strategies. Test for color vision should be a part of ophthalmic examination. There are number of putative methods which may help in correcting color vision deficiency like (i) Tinted spectacles or contact lenses, (ii) Notch or band pass filters, (iii) Eyeborg, and (iv) Gene therapy.

Conclusion: Our study of color vision deficiency among adult Kashmiri population shows the significant gender based difference in the frequency of color vision deficiency, with higher in males (8.5%) than in females (1.3%). The assessment of color vision deficiency by Ishihara pseudo isochromatic plates is rapid, cost efficient and sensitive in detecting red-green color vision deficiency. Researches may attempt to formulate hypothesis regarding causal relationship, frequency and significance of color vision deficiency in human population. People with color vision deficiency face wide range of difficulties in day to day life but due to unawareness about their problem they either neglect or adapt to it. Early detection of the problem by active screening and by increasing awareness help the color vision deficient people to make adaptive and behavioral strategies to deal with potential difficulties they face during various stages of life.

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