

# A Clinical Study of Colour Vision Status of Female Relatives of Indian Males with Defective Colour Vision

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## ABSTRACT

**Introduction:** Around 15% of females are carriers of congenital colour blindness. Variable colour vision status exists in these heterozygous carrier females due to various factors such as retinal mosaicism and the likelihood of the presence of an additional cone with different photosensitivity. These heterozygous carriers are considered to be tetrachromats.

**Aims:** To determine the colour vision status of female relatives of Indian colour blind males.

**Materials and Methods:** In this cross-sectional observational study, screening of males was done by Ishihara plates, and those with Colour Vision Defects (CVD) underwent a digital Farnsworth Munsell hue test (FM 100 test). First degree female relatives and a control group of normal females underwent the same tests. Statistical analysis was done using one-way ANOVA test, Tukey's test and unpaired t-test.

**Results:** Thirty one colour blind males accounted for 3.04% of the 1017 males screened. All participating female relatives, i.e., 31 obligatory carriers (23 mothers and 8 daughters) and

12 sisters, had normal test results on Ishihara plates as well as FM 100 test, barring one mother. FM error score above 70 is considered to be abnormal. The FM error scores in the obligatory carrier females ranged from 39 to 62, while that in sisters' group ranged from 36-67. This error score was lower in control group with range from 14 to 49. Although, all female relatives made errors within the normal range, they had statistically significant higher FM error scores than the controls.

**Conclusion:** Obligate carriers have normal colour vision on Ishihara plates. Higher error scores, albeit within normal limits, were seen on FM 100 test in obligate carriers than controls. Poor performances of genetic tetrachromats on the FM 100 test can be attributed to the fact that these tests are designed specifically for trichromatic normal individuals, and thus, may not be sensitive enough to detect the superior colour discrimination abilities of tetrachromats. Detection of tetrachromacy will require customisation of colour vision tests as well as their availability to clinicians.

**Keywords:** Congenital colour blindness, Digital farnsworth munsell 100 hue test, Heterozygous obligatory carriers, Human tetrachromacy

## INTRODUCTION

Congenital X-linked recessive defects are the most common type of Colour Vision Defects (CVDs) [1,2], with 15% of females being carriers of the trait [3]. Mothers and daughters of colour blind males are obligatory carriers [4] and hence, should have normal colour vision [5]. However, variable colour vision status has been reported in these heterozygous carrier females [3,6-8], due to a random inactivation of one of the two X chromosomes, leaving only one functioning X chromosome which may be normal or abnormal [9]. The retinas of heterozygous carriers have a mosaic of cone population having mutant alleles as well as normal alleles expressing four different types of cone pigments, i.e., one anomalous red or green pigment in addition to the normal red, green and blue cone pigments [3]. Hence, such heterozygous carriers possess four types of cones and may be considered to be tetrachromats with an additional cone pigment having a different photospectral sensitivity [3,10]. However, all genetic tetrachromats may not manifest tetrachromacy phenotypically, because of the random inactivation phenomenon.

There are previous reports of daughters and mothers of trichromatic anomalous males, who are able to discern many more hues than a normal person [11]. So far, only one tetrachromatic female with supernormal colour vision has actually been identified to date [11]. This extremely rare detection may be attributed to the fact that the current, routinely used, colour vision tests are not designed to detect the possible superior colour discrimination in putative tetrachromatic carriers.

There are no previous studies on this aspect of colour vision in the Indian population; although, there are reports on the prevalence of colour blindness [12-14]. Against the above background, it was decided to study the colour vision status of female relatives of Indian colour blind males in a clinical setting and compare it with controls.

## MATERIALS AND METHODS

A cross-sectional, observational study was conducted from April to July 2015, at a teaching hospital, after due approval by the institutional ethics committee. Healthy adult males, below 40 years of age, were enrolled from the college campus, hospital and community outreach centres. They were screened for colour blindness. Female relatives of colour blind males and a control group of normal female relatives of non-colour blind males were also enrolled in the study. All participants were enrolled after obtaining informed consent.

Individuals taking drugs known to affect colour vision, such as tamoxifen, sildenafil, ethambutol, metronidazole, digoxin, and chloroquine were excluded from the study, as also those with a history of ocular or optic nerve disorders, or trauma.

### Study Materials

Ishihara pseudo-isochromatic charts (38 plates), and a FM 100 test [15] were used to evaluate colour vision.

CVDs were detected with transformation plates, vanishing plates, and hidden digit plates of the Ishihara charts, Red (Protan) and

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