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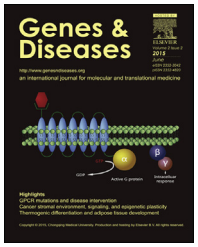

FULL LENGTH ARTICLE

Prevalence and gene frequency of color vision impairments among children of six populations from North Indian region

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Abstract X-linked red–green color blindness is the most widespread form of vision impairment. The study aimed to determine the prevalence and gene frequencies of red–green color vision impairments among children of six different human populations of Jammu province. A total of 1028 healthy subjects (6–15 years of age) were selected from five Muslim populations and the color vision impairments were determined using the Ishihara's test of color deficiency. The gene frequency was calculated using Hardy–Weinberg equilibrium method. The prevalence of color vision deficiency (CVD) ranged from 5.26% to 11.36% among males and 0.00% –3.03% among females of six different populations. The gender based differences in the frequency of CVD was found to be statistically significant ($p < 0.0001$), with a higher prevalence among male (7.52%) as compared to female (0.83%) children. We observed high frequency of deutan as compared to protan defects. The incidences of deuteranomaly (5.68%) and deuteranopia (2.27%) were higher among male children of Syed population while the frequencies of protanomaly (1.94%), protanopia (1.28%) and achromacy (2.27%) were the highest among male subjects of Khan, Malik and Syed populations, respectively. The allele and genotype frequencies showed cogent differences among six populations. The population based assessment of CVDs help patients to follow adaptive strategies that could minimize the risks of the disease.

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Introduction

Color blindness, or color vision deficiency (CVD), is the inability or decreased ability to perceive color differences under normal lighting conditions. CVD can be classified as congenital or acquired. The prevalence of congenital color blindness is about 8% in males and 0.4% in females, results either from alterations or absence in the absorption spectrum of photopigment.[1] The frequency of color blindness vary among different ethnic populations across the world. A recent study from Eastern India has reported 8.73% of males and 1.69% of females as color blind.[2]

The discrimination of color in humans depends on unequal stimulation of three (red, green and blue) cone types to lights of different wavelengths. Normal human color vision is trichromatic, based on the presence of three spectrally-distinct types of cone photoreceptors in the retina that are maximally sensitive to light at 420, 530 and 560 nm (short, middle and long wavelength sensitive cones; S, M and L, respectively).[3] Mutations and rearrangements in the genes encoding the long, middle, and short wavelength sensitive cone pigments are responsible for color vision deficiencies.[4] A recent study has revealed the missense mutation in a hybrid L/M cone opsin gene leading to X-linked cone dystrophy and color vision deficiency.[5]

The blue pigment gene is located on chromosome 7, while the red and green pigment genes are located on long arm of the X-chromosome (Xq28). The mothers who are carriers of the abnormal gene have a chance of 50% abnormal color vision for sons. The CVD fathers transmit their X-chromosomes to daughters only, which leads to all daughters as carriers and sons with normal color vision. A simplified inheritance pattern of sex-linked red–green color blindness is shown in Fig. 1.

The two broad categories of 'red–green' defects are protan and deutan. The protan defects are characterized by an absence or anomaly of L-cone function, while deutan defects are characterized by an absence or anomaly of M-cone function. Deuteranopia or protanopia arises due to the absence of photopigment of the green or red cone, whereas the photopigment response of the green cones is shifted towards that of the red cones and leads to deuteranomaly or vice versa (protanomaly).[6]

Estimating the CVD phenotypes and gene frequencies among different populations has several benefits in occupations or jobs and in routine life that involve precise color matching. These include telecommunication and electrical mechanics, seamen, train drivers, air traffic controllers, painters and several other jobs as well as daily routine work deemed of color recognition. The North Indian human populations and populations from Jammu and Kashmir (J&K) have historical, linguistic, cultural, and socio-religious significance for the Indian subcontinent.[7] The aim of this preliminary study was to estimate the phenotype and gene frequencies of color vision impairments among the children of six different Muslim populations of Jammu region.

Materials and methods

Ethics statement

The study was approved by Institutional Ethics Committee of Jawaharlal Nehru Medical College (JNMC), Aligarh Muslim University, India. We obtained the written informed consent from the parents, caretakers, or guardians on behalf of the minors/children participants involved in our study.

Population and study design

The J&K is the Northern most state of India, situated between 32.17 and 36.58 North latitude and 37.26 and 80.30 East longitude. To its North is China, Russia and Turkistan and on its East is Chinese Tibet and on the West are the north western frontier provinces of Pakistan.[8] The study was conducted in Rajouri and Poonch districts of J&K, during April 2013 through December 2013. The incidence of consanguineous marriages ranges from 35% to 50% among these populations.[9,10] Family pedigrees were made up to five generations back (volunteered by the parents) helped in ascertaining the consanguinity status of their marriage. The information provided by the parents was also cross checked by seeking help from the elder members of the family. Only non-consanguineous families were taken in our study to follow Hardy–Weinberg equilibrium for gene frequency analysis. A total of 1028 healthy children (6–15 years of age) were selected from 815 families of six Muslim populations viz., Gujjar and Bakarwal (n = 184), Mughal (n = 160), Khan (n = 190), Malik (n = 167), Mir (n = 173), and Syed (n = 154). Care was taken to avoid the selection of two or more children of the same sex for each family. The family based children sample include: (a) single child (n = 602) from 602 families and (b) two children (n = 426) of different sex from 213 families. Fig. 2 depicts the steps involved in the recruitment process. The details of sample size under study are presented in Table 1.

Measures and procedure

The color vision impairment or CVD was determined using the Ishihara's Test of Color deficiency (38-plate edition). The plates make up several different test designs as follows:

(a) Transformation plates: The CVD patients recognize a different figure from individuals with normal color vision.
(b) Vanishing plates: Only normal color vision individuals could recognize the figure.
(c) Hidden digit plates: Only CVD patients were able to spot the sign.
(d) Diagnostic plates: These were used to differentiate red–green color blindness (protan and deutan defects).

The subjects were approached through their parents (at residence) and their name, age, sex, class and school