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Inheritance of Eye Colour in Population of Mysuru City, Karnataka, India

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Abstract: The study was performed to investigate the inheritance of eye colour in Mysuru city population using 260 individuals which comprised 124 brown, 87 black, 27 cat eyes, 7 dark brown, 4 green, 3 brownish green, 2 blue eye. It is due to the presence of melanin pigment within the iris for the visual expression of human eye colour. Although segregation of blue-brown eye colour has been described using a simple Mendelian dominant-recessive gene model this is too simplistic, and a new molecular genetic perspective is needed to fully understand the biological complexities of this process as a polygenic trait.

Keywords: Perspective, Polygenic, Segregation Mapping, Chromatin, Eye colour, Inheritance, Melanin

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Introduction

Eye Colour is a polygenic phenotypic character determined by two distinct factors: the pigmentation of the eye's iris (Prota *et al.*, 1998; Wielgus and Sarna, 2005) and the frequency dependence of the scattering of light by the turbid medium in the stroma of the iris (Denis, 1979). In humans, the pigmentation of the iris varies from light brown to black depending on the concentration of melanin in the iris pigment epithelium (located on the back of the iris), the melanin content within the iris stroma (located at the front of the iris), and the cellular density of the stroma (Wang *et al.*, 2005). The appearance of blue and green, as well as hazel eyes results from the Tyndall Scattering of light in the stroma, a phenomenon similar to that which accounts for the blueness of the sky called Rayleigh Scattering (Sturm. and Larsson, 2009). Neither blue nor green pigments are ever present in the human iris or ocular fluid (Mason, 1924; Denis, 1979). Eye colour is thus an instance of structural colour and varies depending on the lighting conditions, especially for lighter-coloured eyes.

The presence of melanin pigment within the iris is responsible for the visual impression of human eye colouration with complex patterns also

evident in the tissue, including Fuchs' crypts, nevi, Wolffian nodules and contraction furrows. The genetic basis underlying the determination and inheritance of the traits has been the subject to debate and research from the very beginning of quantitative trait studies in humans. Although segregation of blue-brown eye colour has been described using a simple Mendelian dominant-recessive gene model, this is too simplistic, and a new molecular genetic perspective is needed to fully understand the biological complexities of this process as a polygenic trait. Nevertheless, it has been estimated that 74% of the variance in human eye colour can be explained by one interval on chromosome 15 that contains the OCA2 gene. Fine mapping of this region has identified a single base change rs12913832 T/C within intron 86 of upstream HERC2 locus that explains almost all of this association with blue-brown eye colour. A model is presented by whereby this SNP (single nucleotide polymorphisms), serving as a target site for the SWI/ SNF family member HLTF, acts as part of a highly evolutionary conserved regulatory element required for OCA2 gene activation through chromatin remodelling. Major candidate genes possibly affecting iris patterns are also discussed, including MITF and PAX6.

The brightly coloured eyes of many bird species result from the presence of other pigments, such as pteridines, purines, and carotenoids (Oliphant, 1987). Humans and other animals have many phenotypic variations in eye colour (Morris, 2006).

The genetics and inheritance of eye colour in humans is complicated. So far, as many as 15 genes have been associated with eye colour inheritance. Some of the eye-colour genes include OCA2 and HERC2 (White and Montserrat, 2010). The earlier belief that blue eye colour is a simple recessive trait has been shown to be incorrect. The genetics of eye colour are so complex that almost any parent-child combination of eye colours can occur. However, OCA2 gene Polymorphism, close to proximal 5'regulatory region, explains most human eye colour variation (Duffy *et al*., 2007).

Eye Colour is an inherited trait influenced by more than one genes (Grant and Lauderdale, 2002; Sturm and Frudakis, 2004). These genes are sought using associations to small changes in the genes themselves and in neighbouring genes. These changes are known as single nucleotide polymorphisms or SNPs. The actual number of genes that contribute to eye colour is currently unknown, but there are a few likely candidates. Rotterdam (2009) found that it was possible to predict eye colour with more than 90% accuracy for brown and blue using just six SNPs (Liu *et al*., 2009). There is evidence that as many as 16 different genes could be responsible for eye colour in humans; however, the main two genes associated with eye colour variation are OCA2 and HERC2, and both are localized at Chromosome 15 (White and Montserrat, 2010).

The gene OCA2 when in a variant form, causes the pink eye colour and hypopigmentation common in human albinism (The name of the gene is derived from the disorder it causes, oculocutaneous albinism type II). Different SNPs within OCA2 are strongly associated with blue and green eyes as well as variations in freckling, mole counts, hair and skin tone.

The polymorphisms may be in an OCA2 regulatory sequence, where they may influence the expression of the great product, which in turn affects pigmentation (Duffy *et al*., 2007). A specific mutation within the HERC2 gene, a gene that regulates OCA2 expression, is partly responsible for blue eyes (Kayser *et al*., 2008). Other genes implicated in eye colour variation are SLC24A4 (Sulem *et al*., 2007) and TYR. Study on eye colour variation into hue and saturation values using high-resolution digital full-eye photographs found three new loci for a total of ten genes, and now about 50% of eye colour variation can be explained (Liu *et al*., 2010).

Iris colour can provide a large amount of information about a person, and a classification of colours may be useful in documenting pathological changes or determining how a person may respond to ocular pharmaceuticals (German *et al*.,

1998). Classification systems have ranged from a basic light or dark description to detailed gradings employing photographic standards for comparison (German *et al*., 1998). Others have attempted to set objective standards of colour comparison (Fan *et al*., 2003).

Normal eye colours range from the darkest shades of brown to the lightest tints of blue (Sturm and Frudakis, 2004). For standardized classification, at once simple yet detailed enough for research purposes, Seddon *et al*. (1990) developed a graded system based on the predominant iris colour and the amount of brown or yellow pigment present. There are three pigment colours that determine, depending on their proportion, the outward appearance of the iris, along with structural colour. Green irises, for example, have some yellow and the blue structural colour. Brown irises contain more or less melanin. Some eyes have a dark ring around the iris, called a limbal ring.

Eye colour in non-human animals is regulated differently. For example, instead of blue as in humans, autosomal recessive eye colour in the skink species *Corucia zebrata* is black, and the autosomal dominant colour is yellow-green. (Jones and Schnirel, 2006)

As the perception of colour depends on viewing conditions (e.g. the amount and kind of illumination, as well as the hue of the surrounding environment), so the perception of eye colour. Generally, eye colour depends on how much of the pigment melanin is in the iris, the coloured part of the eyes. A small number of people may notice their eye colour changing with age. The more pigment, the darker the eyes. The only pigment is brown; the colour of the eyes in determined by the level of pigmentation in the eyes. Research shows that up to 16 genes may influence eye colour.

Brown eyes are by far the most common. In fact, approximately 80 per cent of the world's population has brown eyes. Because they are deeply pigmented, Brown eyes are more in the world's population. Because they are deeply pigmented, brown eyes are more protected from the sun than other coloured eyes.

There's a saying out there that all people with blue eyes are related. Legend has it that at one time, everyone had brown eyes, but a genetic mutation in a single individual 10,000 years ago led to blue eyes. Whether that's true or not, we do not know for sure. What we do know is that about 10 per cent of the world's population has blue eyes today.

The word "hazel" does not really describe a colour as much as a combination of colours. Hazel eyes have hints of green, blue, gold, and brown. About 5 per cent of the population has hazel eyes. The number of people who have true amber eyes is unclear, but it is definitely one of the rarest colours. Unlike hazel eyes, amber eyes do not contain hints of other colours. Only 3 per cent of the world's population has gravy eyes, which understandably may be confused with blue eyes. Although it may not appear to be so, green eyes are the least common. Green eyes have more melanin than blue eyes, but only 2 per cent of the people in the world have green eyes.

Materials and Methods

A total number of 260 individuals were studied including Brown- 124, Black- 89, Dark brown- 7, Green- 4, Greyish brown-4 Brownish green-3, and Blue eyes -2. These eye colours are analysed based on a melanin pigment.

Results and Discussion

In this study, only frequency of distribution of eye colours among the sampled individuals is presented. The study revealed that Black eye colour is the most frequent in more than 35% of the population, followed by the Brown eye colour appeared in 32% of the population, cat eye in 11%, Dark Brown in 7%, Green and Brownish Green 4% each, Blue in 2% of the population (Fig. 1). Thus, Eye pigment distribution can be taken as a standard genetic trait with multiple variations, in population genetic studies.

The inheritance seems to be unpredictable in

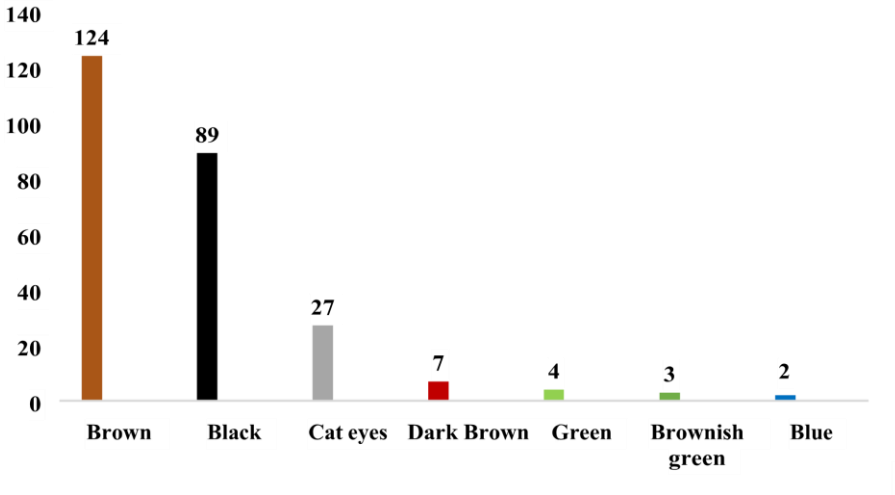

Fig. 1: Number of individuals of different eye colour.

majority of cases, because brown eye parents have either black or Grey or brown children. That is to say that brown is dominant and has many alleles. But it is well known that the eye colour is polygenic inheritance, determined by three different genes for brown, blue and gravy eye. So, the formation of colour may also be due to interaction of these genes.

Eye colour inheritance and distribution can be taken as ideal example of polygenic interaction and inheritance in human. Eye colour results from varying degrees of melanin produced in the melanocytes of the iris. The process that produces melanin, known as melanogenesis, requires numerous proteins. TYR, the enzyme responsible for pigment throughout the body, uses tyrosine to begin the chemical pathway. With the help of dopachrome tautomerase and TYR-related protein 1, eumelanin, darker pigments, is synthesized; with cysteine, pheomelanin, yellow-red pigments, is produced. Once the pigment is produced, MC1R, membrane-associated transporter protein, and a proteins (OCA2) mature the melanosomes to be used in the cells.

Two major genes on chromosome 15 affect the quantity and quality of the melanin produced by melanogenesis. HERC2, a larger ubiquitin ligase, contains the promoter region for OCA2, the protein. When a T is replaced with a C in rs12913832 of intron 86, OCA2 transcription is depressed, resulting in a blue-eyed individual.

This epistatic relationship demonstrates the significance of introns and how a single base change greatly affects an aspect of the individual. OCA2 contains regions for the numerous eye colours, but one SNP is a strong predictor for brown/blue eyes. A change in rs1800407 causes a change in the protein, Arg419gln, and a change from brown to blue eyes. Therefore, the residue change causes a problem with the P protein, and melanin maturation decreases.

Aside from these two genes, the genes involved in melanogenesis and other minor genes also contain regions that account for eye colour. Having little effect on eye colour, many of them deal primarily with hair and skin pigmentation. Lastly, disorders involved in eye colour include ocular albinism and heterochromia. Depending on how little pigment the melanocytes produce, albinism causes red or violet eyes. Producing multi-coloured irises, heterochromia stems from mutation in certain cells of the iris.

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