

It All Lies in Her Eyes

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Pak J Ophthalmol 2016, Vol. 32 No. 4

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Eye colour is one of the most notable traits in determining facial features. In this case report, we present a 19 year old girl with isolated congenital heterochromia iridis. She presented to outdoor patient department for a routine check up. On detailed examination, we did not find any ocular or systemic abnormality. Isolated heterochromia iridis is an exceedingly rare condition world wide and in our population as well. Ophthalmologist should be aware of this condition and should be able to rule out different syndromes and ocular/systemic conditions that may be associated with it.

Key words: Heterochromia iridis, melanin, Fuch's heterochromic uveitis.

Heterochromia iridis is a term coined for a clinical situation when there is a difference in the colour of both iris. The normal iris can be of darker (hyperchromia) or lighter (hypochromia) hue. And there can be many genetic or acquired factors that can lead to heterochromia iridis¹. Heterochromia iridis happens due to relative loss or gain of melanin pigment granules in the effected eye. These pigment granules reside in melanocytes whose number remains constant. The amount of melanin in each melanocyte is genetically regulated. This loss or gain of pigment can be congenital or acquired and isolated or composite with other systemic conditions². There are many systemic conditions that can lead to sectoral or complete heterochromia iridis e.g, Fuch's heterochromic uveitis, congenital Horner's syndrome, Waardenburg's syndrome, hypomelanosis of Ito and linear scleroderma³⁻⁵.

Those conditions leading to hypochromia iridis include Waardenburg's syndrome, Horner's syndrome, incontinentia pigmenti, Fuch's heterochromic uveitis etc⁶. The effected iris can be darker in colour in certain acquired or congenital conditions like siderosis, topical use of prostaglandin analogue for glaucoma, Nevus of Ota, Sturge Weber syndrome, pigment dispersion syndrome etc.

Isolated congenital heterochromia iridis is an exceedingly rare condition and there is no literature

evidence to report its incidence. Henceforth, we report this rare ocular condition to document its presence in Pakistani population.

CASE REPORT

This 19 year old female presented to the outdoor patient department (OPD) of Sharif Medical City Hospital, Raiwind, Lahore in June 2016 for a routine checkup. She came to the OPD with her mother and one sibling and the family had been aware to two different iris colours since birth. According to the

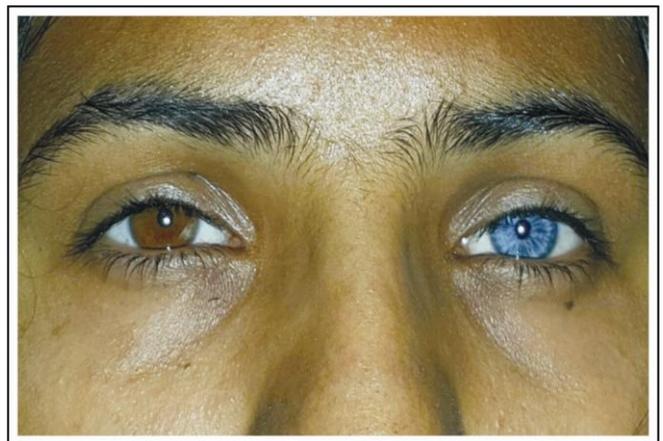


Fig. 1: Isolated Congenital Heterochromia Iridis in a 19 year old girl.

mother, the child was born at her home (village in Raiwind) with the assistance of a midwife through spontaneous vaginal delivery. The child was full term and had uneventful antenatal course. The only significant postnatal event was neonatal jaundice. Her vaccination record was sketchy and mother could not recall about completing her daughter's vaccination. The patient had no significant ocular complaint at the time of her presentation. The patient had 2 siblings (both females) and their eyes colour was dark brown. The parents also had brown coloured eyes and they were second order cousins.

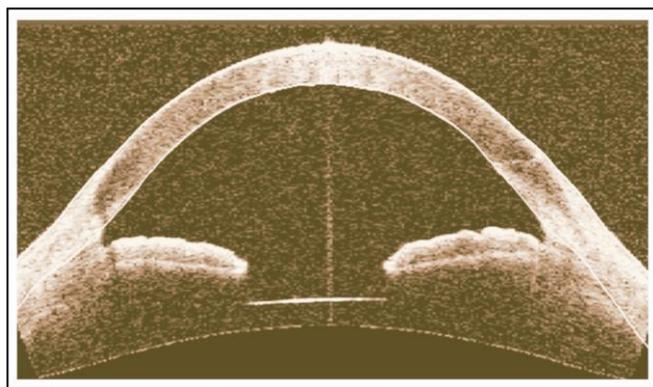


Fig. 2: Anterior Segment Optical Coherence Tomograph of Left Hypochromic eye of 19 year old girl.

On examination, the patient had 20/20 visual acuity in both eyes with minimal or no refractive error after cycloplegic refraction. Her anterior segment examination was unremarkable apart from stark difference in colour of her iridis (Figure 1). Posterior segment examination showed mild hypo pigmentation of left fundus with normal optic disc, macula and retinal periphery. Her intraocular pressure was 14 mm Hg and 16 mm Hg in right and left eye respectively. The patient was also checked by a female doctor for any hypopigmentation on rest of the body but results were negative. The patient was also thoroughly examined for the presence of any signs that may direct us towards any of the syndromes (Fuch's heterochromic uveitis, Horner's syndrome, Waardenburg's syndrome) mentioned above but her examination was completely unremarkable apart from heterochromia iridis. An anterior segment optical coherence tomography was also performed delineate the anatomy of iris and anterior segment; and also to rule out iris tumours and cysts which may render iris

hypo pigmented (Figure 2). The patient was not concerned about the heterochromia but was still offered the choice of using cosmetic contact lens if she desired so.

An informed consent was taken from the patient for publishing her rare ocular condition along with pictorial reference to her heterochromia iridis.

DISCUSSION

The different iris colours are based on the presence of amount of brown and yellow pigment with distinguishes iris based on predominant colour which may be blue, grey, green or various hues of brown.⁷ Patients having dark brown iris are rich in melanin whereas other shades of iris colour have relatively lower melanin content; blue colour completely lacking melanin. It may be inherited, or caused by genetic mosaicism or chimerism. The scientific consensus is that a lack of genetic diversity is the primary reason behind heterochromia. This is due to a mutation of the genes that determine melanin distribution at the 8-HTP pathway, which usually only become corrupted due to chromosomal homogeneity².

Very early reports from the start of century supported the idea that eye colour was inherited as a Mendelian trait. It approved the notion that blue eyes were autosomal recessive where brown eyes were autosomal dominant traits. This doctrine suggested that two blue eyed parents could not give birth to a brown eyed child but later reports nullified this idea. It was then perceived that eye colour is a polygenic trait. Chromosomal studies implicated multiple genes playing a role in determining eye colour but the most notable of them came out to be OCA-2 gene located on the long arm of chromosome 15. Apart from causing heterochromia iridis, its mutation has also been linked with oculocutaneous albinism, Angleman and Prader Willi syndromes⁸.

Although, heterochromia iridis has been reported multiple times in association with other syndromes, its literature evidence is minimal when reported as the only clinical finding. Similar to our case, one case of 15 year old Nigerian girl was reported by Omolase where the only clinical finding was heterochromia iridis with normal ocular and systemic examination⁹. He also reported a case of bilateral hypochromia iridis with normal ocular and systemic examination in a 6 months old Nigerian girl¹⁰. Kocak reported a very interesting case of heterochromia iridis in a 5 year old girl who also had wooly hair nevus and ipsilateral pigmentary

demarcation lines. Woolly hair nevus refers to a clinical entity where a patch of scalp hair is hypopigmented and curlier than rest of scalp hair. This constellation of symptoms is also a solitary occurrence with no other such reference in literature. The rest of ocular and systemic examination in this patient was unremarkable.³ Other reported cases of heterochromia iridis have been associated with rare systemic syndromes; the most notable of them being Waardenburg's syndrome⁶.

CONCLUSION

We reported this rare case to entice interest of ophthalmologists in this rare clinical presentation. Since heterochromia iridis is also associated with other ocular and systemic clinical conditions, so such a patient always warrants a complete examination to rule out any sinister condition associated with heterochromia iridis.

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