Case Report

Bilateral Symmetric Retinal Pigmentation Versus Heterochromia: A Case of Waardenburg Syndrome

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This report describes a 22 year intellectual female who presented with poor vision while working on a microscope. Slit lamp biomicroscopic examination showed heterochromia iridis and retinal pigmentary changes that disclosed Waardenburg Syndrome (WS) type 2. Except of white forelock, we could not find any other association or anomaly elsewhere. Ophthalmological examination revealed WS type 2 with heterochromia iridis. She also had the specific pattern of bilateral posterior heavy retinal pigmentation versus peripherally albinotic pattern. This retinal finding in WS type 2 is the first report in literature to the best of our knowledge.

Waardenburg syndrome (WS) is an uncommon autosomally inherited and genetically heterogeneous disorder of neural crest cell development. It consist of six distinctive features: lateral displacement of the medial canthus and lacrimal punctae, broad and high nasal root, hypertrichosis of medial part of the eyebrows, partial or total heterochromia iridis, white forelock, and congenital deaf mutism[1].

The diagnosis of WS is made clinically. Genetic testing is available for confirmation of diagnosis and prenatal diagnosis if the mutation in the family has been identified[2].

Failure of neural crest-derived melanocyte differentiation results in a spectrum of phenotypic presentations that are subdivided into 4 clinical types. Type 1 is the classic form of WS with dystopia canthorum (lateral displacement of the inner canthi), type 2 characterized by the presence of white forelock, unilateral or bilateral deafness, but without the dystopia canthorum. WS3 have hypoplasia of limb musculature and/or contractures of elbows and fingers, and WS4 have Hirschprung disease in addition to the other common feature of WS[3].

The others report describe variable retinal pigmentary disturbances, the extensive albinoid areas nasally and in posterior pole, versus the temporal region which showed a homogeneous area of dense hyperpigmentation[4, 5].

We report here a case of WS, for its rarity, variability of systemic and clinical eye involvement in literature. The heterchoromia with symmetrically bilateral posterior retinal hyperpigmentation versus peripheral hypopigmentation was the unique finding that it is not reported till now.

CASE REPORT

A 22 year old girl with white forelock complained of poor vision while working on a microscope. She had centrally placed white forelock in the frontal area without any associated depigmentation of scalp or elsewhere on the body. Her nasal root was not broad without wide intercanthal distance. She had not any complain of hearing or any others anomaly elsewhere. On ophthalmological examination, Pupils were mid dilated with sluggish reaction and full eye movements. The best corrected vision was 20/25 with 1 diopter of myopic correction in both eyes. Anterior segment examination was within normal limit except

the presence of unilateral right blue iris. Fundoscopic examination showed bilateral symmetrical posterior hyperpigmentation and peripheral hypopigmentation of the retina Fig. 1 – 2. The abstinence of high illumination and spectacle prescription reduced her complaint. The white forelock could not be showed due to patient's non consent.

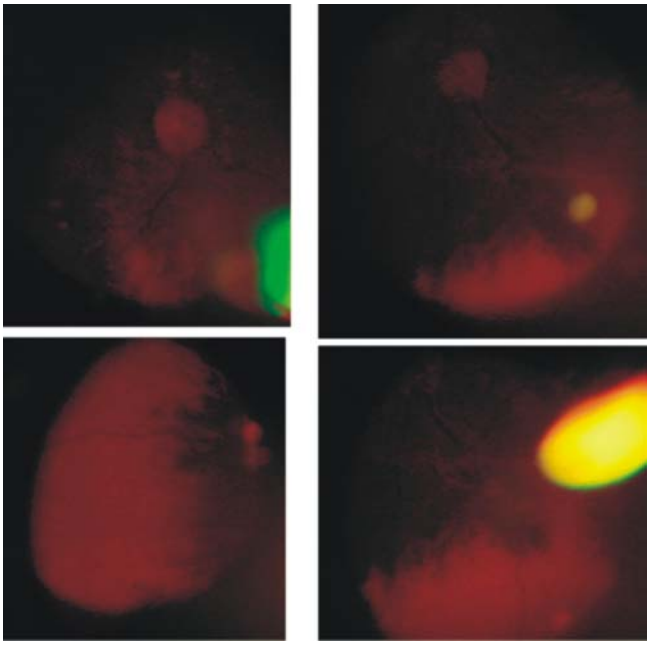

Fig. 1: Right and left posterior pole retinal hyper-pigmentation versus peripheral hypopigmentation.

DISCUSSION

We present a 22 year old female with heterochromia and unusual retinal pigmentary change with waardenburg syndrome type 2. She did not have any obvious classical systemic or family history but her ocular examination helped us discover another variable feature of WS_2.

The white forelock and right blue iris versus left brown iris, was accompanied by bilateral central retinal hyperpigmentation and peripheral hypopigmentation in our case that not observed till now. The peripheral retinal hypopigmentation had similarity to funduscopic change that may be albinotic pattern or may show pigment mottling in the periphery as described by Tagra Sunita et al[6]. Tagra Sunita et al also describe that except the deafness all features of Waardenburg type 1 and 2 are essentially benign and cosmetic in nature and do not necessitate active intervention but the visual complaint in this case without causative finding may warrant more knowledge about funduscopic change and heterochromia.

Study of Abah et al[7] among deaf student demonstrated four (0.6%) students that had iris heterochromia, three bilateral and one unilateral involvement, they also had white forelock hair and together with the deafness were presumed to have Waardenburg syndrome but none of the students had the funduscopic finding of typical salt and pepper retina of congenital rubella[7].

Another case report by Manish Mahta et al[8] also showed choroidal depigmentation with classical salt and pepper retina appearance in a two year deaf female with brilliant blue iris[8].

A case report of Waardenburg by Naeimeh Tayebi described a 2 year old girl with waardenburg syndrome whose father was also affected. The ophthalmic examination revealed dystopia canthorum and telecanthus, accommodative isotropia in either eyes without heterochromia or any other eye anomaly2. This case did not have any misalignment.

According to our knowledge the fundoscopic finding of WS2 in our case introduced another variable feature of Waardenburg which was associated with bilateral similar retinal pigmentary change in spite of dissimilar iridis in a female.

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