A Case Series of Waardenberg Syndrome

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Waardenburg syndrome (WS) is a rare autosomally inherited and genetically heterogeneous disorder of neural crest cell development with distinct cutaneous manifestations. We report here a case series of patients of a single family, presented with assemblage of complete heterochromia, dystopia canthorum, synophrys and broad nasal root. Other family members with presence of heterochromia and telecanthus have been delineated in pedigree. In our case series second generation of family also found to be affected, which is rarely reported till now. To our knowledge no local cases have been reported till date.

Key words: Waardenberg syndrome, Heterochromia, Telecanthus.

CASE REPORT

Our first patient is a 22 year old male presented to the outpatient department of our tertiary eye care hospital with chief complaints of difference in color of his eyes with decreased vision in right eye (Fig. 1). His Best corrected visual acuity at presentation was 6/24 p OD with -3.50 x -1.00 at 90 degree and 6/6 OS. Patient also has complain of decreased hearing in right ear. He was the last child of a non-consanguineous marriage among 7 siblings with one affected brother as well. (3 brothers and 3 sister). His birth and developmental history did not reveal anything significant. His detailed family history revealed that his mother has telecanthus along with 1 brother and 2 nephews and 2 nieces affected as well.

On systemic examination, he was moderately built with an average height, weight and normal IQ. He had premature graying of hair with absence of any associated depigmentation elsewhere on the body. His ENT and abdominal examinations were normal.
On ocular examination, gross inspection shows broad nasal root and synophrys. The palpebral fissure height of right eye was 1.5 cm and 1.4 cm in left eye along with lateral displacement of canthi. He also had dystopia canthus with interpupillary distance of 8 cm and inner canthal distance of 5.7 cm. Hirschberg corneal reflex was central but medially sclera was visible o a lesser extent.

Complete heterochromia due to hypoplastic iris, blue in color was noted in right eye (Fig. 2). Sectoral atrophy of iris seen in left eye. Besides this anterior segment examination was otherwise normal in both the eyes. IOP was in normal limits. Pupillary reactions were normal. Right fundus was albinotic showing hypopigmentation and temporal disc pallor and left fundus was normal (Fig. 3). Gonioscopy revealed...
grade IV angle in both eyes, normal iris vessels seen in angle of right eye.

Our second patient is 46 years old male, elder brother of patient. His history revealed presence of white forelock of hairs at birth (due to hair dying at early age patient has no old picture) and premature graying of hairs, dystopia canthorum, synophrys and heterochromia irides. His visual acuity was 6/6 in both eyes. Right eye shows sectoral iris atrophy and complete heterochromia found out in left eye. Both fundii were normal.

![Fig. 7: Pedigree chart. Dark blue box: first patient. Light blue box: other affected males. Red circles: affected females.](image)

**DISCUSSION**

Waardenburg consortium proposed diagnostic criteria for diagnosing WS types. According to it, for placing patient in category of WS I, patient should have 2 major or 1 major + 2 minor criteria present. WS II is characterized by sensor neural hearing loss and heterochromia iridis but absence of dystopia canthorum. WS III (Klein–Waardenburg syndrome) resembles type I with supplementary musculoskeletal abnormalities. These patients have hypoplastic muscles and contractures of the upper limbs. WS IV is associated with Hirschsprung disease. Liu et al. suggested method for diagnosis of WSII, which requires presence of at least two major criteria and the same study propounded the use of premature graying as one of the mature criteria instead of dystopia canthorum5.

We present a case series of a single family with several members affected of first and second generation. According to the above mentioned criteria our first and second patient falls in category of WS I. since all of these are only cosmetic problems therefore they do not require compulsory treatment.

It is plausible that in certain cases WS may remain undiagnosed until other family members with similar features seek medical attention. One of the purposes to present this case series is to bring in light the significance of detailed examination of all the family members to recognize undiagnosed cases. Clinical features of WS are mainly cosmetic problems for which no definitive treatment exist, except for few oculoplastic procedures which can be done for broad medial canthus. In some cases muted diseases, such as, Sensorineural deafness, bony abnormalities or Hirschsprung disease are found to be associated with WS which results in deterioration in quality of life. An ophthalmologist can help these patients by making an early diagnosis which may aid in the initiation of early treatment, social and vocational training, and rehabilitation of these patients.

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**REFERENCES**