When color matters: Waardenburg syndrome

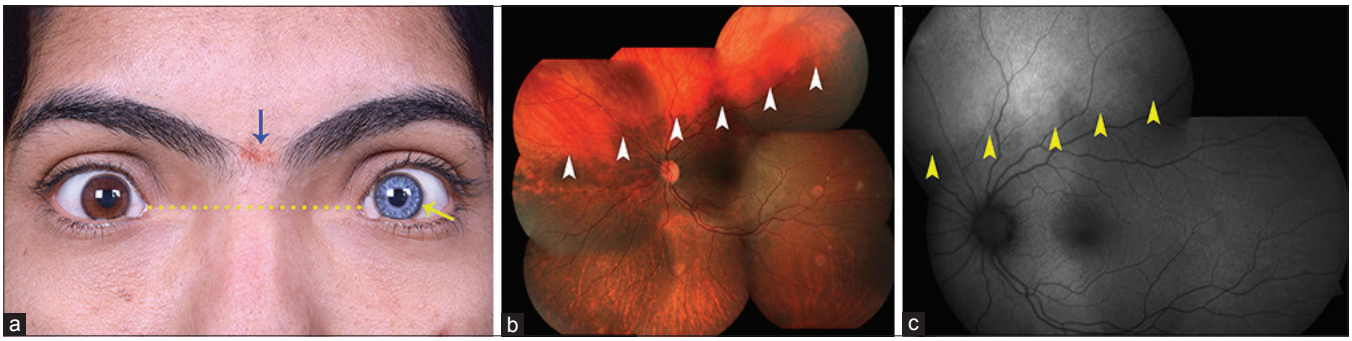

Figure 1: Waardenburg Syndrome (a) External photograph showing telecanthus. Yellow arrow indicates the left heterochromia iridum. **(b):** Montage Fundus photograph of the left eye. White arrowheads indicate the junction of the hypopigmented choroid and the normal choroid. (c) Montage autofluorescence. Yellow arrowheads indicate the area of hyperautofluorescence

A 27-year-old Asian-Indian lady, systemically normal, with diffuse iris hypopigmentation, telecanthus, mild synophrys with a congenital white forelock treated with hair color and 20/20 vision in both eyes presented for treatment of the heterogeneous iris color [Fig. 1a]. Fundus examination revealed a sectoral superonasal hypopigmented choroid in the left eye [Fig. 1b]. Fundus autofluorescence revealed hyperautofluorescence through the hypopigmented choroidal region in the left eye [Fig. 1c], indicating unmasking of scleral autofluorescence. The patient was diagnosed as a case of Waardenburg syndrome Type 1 and was offered a cosmetic contact lens. Audiometry on both sides and gastrointestinal examination was normal.

Waardenburg syndrome has four subtypes distinguished by their physical characteristics and sometimes by their genetic cause.[1-5] Iris and choroidal hypopigmentation are central to all the subtypes [Fig. 1]. Telecanthus is more frequent in type 1, and hearing loss is more frequent in Type II. Type III includes limb anomalies predominantly of upper limb. Type IV is associated with Hirschsprung disease, an intestinal disorder that causes severe constipation or blockage of the intestine. According to the phenotypic presentation, the systemic work-up to classify the patient into one of the subtypes is crucial.

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Conflicts of interest

There are no conflicts of interest.

Meeting presentation

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