Case Report:
Blue Eyed Boy.

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Abstract: We report a case of blue eyes of Waardenburg Syndrome, with findings of hypopigmentation on the posterior pole.

Key Words: Blue eyes, Waardenburg Syndrome

Case Report
A 22 years old male came to our department for regular check up. History was given by his mother as he was congenitally deaf and mute. As per the mother, he had no visual complaints. She gave history of premature greying of hair in the boy and white areas of hypopigmentation over the forearms. He was the first of two children, born out of a non-consanguinous marriage with the sibling being healthy. His birth history and development history were insignificant. On being asked, she revealed history of dissimilar coloration of eyes in maternal grandmother as well as premature greying of eyelashes in herself.

On examination, visual acuity was 6/6, N6 in both eyes. Pupillary reactions were normal. Extraocular movements were normal and full in both eyes. Systemic examination revealed a white forelock of hair in the left frontal area with generalised premature greying of hair. There was a bunch of white hair seen in left eyebrow along with rows of hair joining the two eyebrows. There was a broad nasal root with hypoplastic alae nasi. There were multiple hypopigmented patches along with white hair on both the forearms. On ocular examination, the horizontal palpebral fissure was 25 mm in both eyes along with lateral displacement of canthi. There was dystopia canthus with interpupillary distance of 55 mm and inner canthal distance of 40mm. Medially sclera was visible to a lesser extent giving an impression of convergent squint, however Hirschberg test revealed a central corneal reflex. Brilliant blue irides were noted bilaterally.

Rest of the anterior segment examination was normal in both

Figure 1: Characteristic brilliant blue irides, synophrys, dystopia canthus, hypoplastic alae nasi, premature greying of hair
We found hypopigmentation at the posterior pole while past authors have reported hyperpigmentation. There is a need to further explore this aspect of the disease and describe the various possible retinal pictures of the disease. Fundus picture has not been given due importance in literature while describing this condition and has not been considered as a major or minor criteria.

**Conclusion:**
A timely diagnosis of this striking syndrome could facilitate improvement in speech discrimination in these patients, by cochlear implantation, hence leading to a better quality of life.[6] It would also prove useful in genetic counselling of expectant mothers in such families.

**References**