WAARDENBURG SYNDROME, A CASE REPORT

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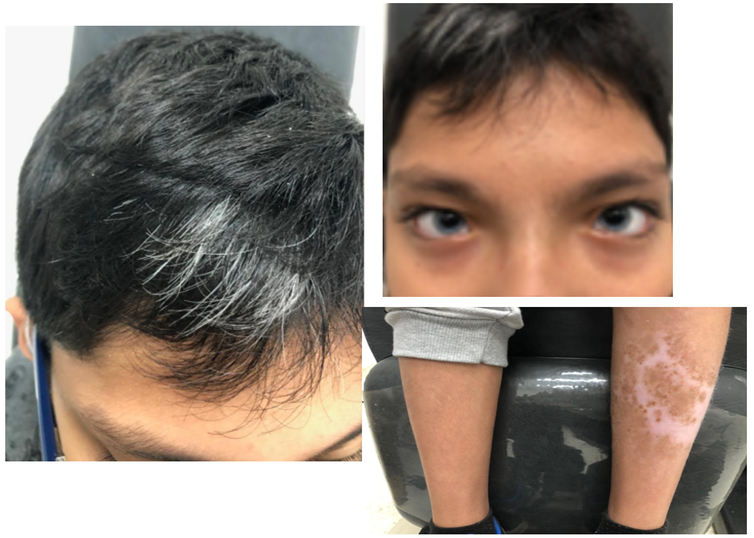


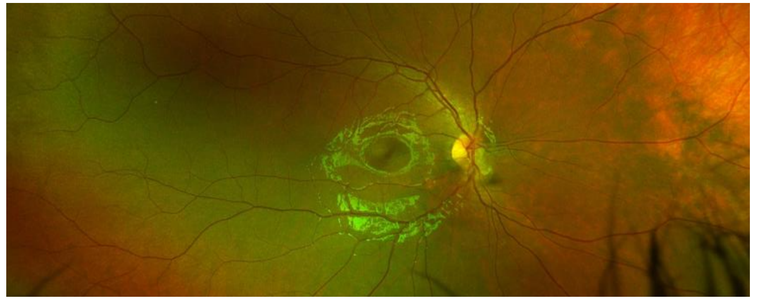
PURPOSE: To report a case of Waardenburg Syndrome

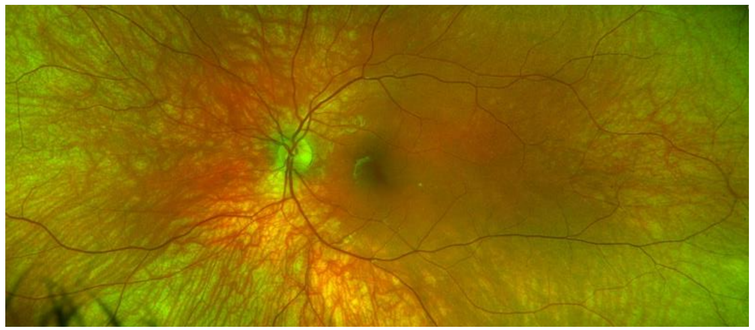
METHODS: Case report; literature review

RESULTS: Nine-years old, white, female, complaining of decreases visual acuity (VA) on her left eye (OS) for 1 year. She has neurosensorial deafness in her right ear. VA was 20/20 OU and the intraocular pressure was within normal range. Eye examination showed depigmentation of the iris and at fundus on OS. Physical examination showed clusters of white skin spots and localized gray hair. Genetic testing was positive for PAX3 mutation.

DISCUSSION: Waardenburg Syndrome (WS) type 1 is autosomal dominant and is caused by mutations of several genes that affect the division and migration of neural crest cells during embryonic development. It has major (congenital neurosensorial deafness, white forelock, complete hypoplastic blue iris, dystopia canthorum) and minor diagnostic criteria (skin hypopigmentation and broad high nasal root). Two major or 1 major and 2 minor are sufficient for the diagnosis. The reported patient has 4 major criteria, and 2 minor criteria.

Patients with WS have fundus hypopigmentation in 67%. Despite of the broad range of abnormalities in this condition, VA generally remains good, so no treatment is needed.





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