WAARDENBURG SYNDROME – A CASE REPORT

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We report an eight-year-old girl who presents bilateral congenital sensorineural hearing loss and prominent left heterochromia iridis. Besides, there are some distinct facial characteristics such as lateral displacement of the medial canthi combined with distopia of the lacrimal puncta, broad nasal root and confluent eyebrows. Her left blue eye also shows prominent choroidal vasculature as an albinotic fundus. After detailed examination and inquiry about family history, Waardenburg syndrome is diagnosed. The clinical manifestations, mode of inheritance, diagnostic criteria and clinical classification of Waardenburg syndrome are also addressed.

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