Waardenburg Syndrome Type I

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Received: 13 September 2019 / Accepted: 23 December 2019 / Published online: 27 January 2020
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Waardenburg Syndrome Type I (WS) is a rare genetic disorder, with pigmentary anomalies of eyes, skin and deafness [1]. A 5-y-old girl born at term, of non-consanguineous marriage presented with inability to hear and speak since birth. Both parents were deaf. Other developmental milestones were normal. Examination revealed white forelock, heterochromia iridium, dystopia canthorum (Fig. 1A), and depigmented macules over dorsum of hand and legs (Fig. 1B). No waveforms were noted in brain stem auditory evoked potentials, suggesting bilateral auditory pathway dysfunction.

The four types of WS are distinguished by their physical characteristics [2]. Dystopia canthorum is common in type I and hearing loss occurs more often in type II. Type III includes limb abnormality and Type IV is associated with Hirschsprung disease. We diagnosed WS type I based on clinical criteria. Differentials include piebaldism which has islands of increased pigmentation in the hypomelanotic areas and sometimes with hyperpigmented borders. Early diagnosis and management of deafness in Waardenburg syndrome improves the quality of life.

Fig. 1 Photograph of the child showing (A) white forelock on the frontal region of scalp along with heterochromia irides and (B) a well defined depigmented macule present bilaterally, symmetrical on the hands extending to mid forearm and bilateral feet extending on to both lower legs

Compliance with Ethical Standards

Conflict of Interest None.

References

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