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CASE REPORT

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A Rare Case of Waardenburg Syndrome Type II

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Abstract

Introduction: Waardenburg syndrome is an autosomal dominant inherited genetic condition where abnormal melanocyte distribution during embryogenesis results in areas of patchy depigmentation. The loss of pigmentary cells in skin, eyes, hair and stria vascularis of cochlea are the cause of various clinical manifestations of this syndrome. The incidence of this syndrome globally is 1/42000 live births. **Case Report:** One day old male baby presented with symmetrical depigmented patches over both lower legs, white forelock, hearing loss, broad nasal bridges since birth. Baby was delivered at 37 weeks period of gestation after an uneventful pregnancy through normal vaginal delivery; history of similar complaints in the father present. No h/o consanguinity among parents. **On Examination:** Symmetrical solitary well defined depigmented patches of size 4x5cm present over both lower legs around knee joints. White forelock present over the frontal region of head. Broad nasal bridge with heterochromia of eyes present. Sensorineural hearing loss detected after audiometry test. Hence clinically diagnosed as Waardenburg syndrome type II. **Conclusion**: This is a rare case of Waardenburg syndrome satisfying criteria for type II. Knowledge of this syndrome helps in its early identification and diagnosis which in turn helps in its early management.

Keywords: Waardenburg Syndrome, White Forelock, Depigmented Patches

1 Introduction

Waardenburg syndrome (WS) is a group of genetic conditions inherited in an autosomal dominant fashion. It is named after Dutch ophthalmologist and geneticist Petrus Johannes Waardenburg, who described it in 1951[1]. During embryogenesis, there is an abnormal distribution of melanocytes, which results in patchy areas of depigmentation. It is a rare disease, caused by loss of pigmentary cells in eyes, skin, stria vascularis of the cochlea, and hair[1].

Melanocyte development is coordinated by a network of genes that function in a temporal, spatial, and dose-dependent manner. Germline mutations in genes that regulate melanocyte development occur in patients with Waardenburg syndrome (WS). Over the last three decades, our understanding of Piebaldism and WS has improved and continues to evolve with advancing molecular techniques.

Waardenburg syndrome (WS) are neurocristopathies characterized by incomplete penetrance and high levels of variable expressivity[2].

Loss-of-function mutations in MITF or its regulatory genes can cause a striking pattern of depigmentation characteristic of Waardenburg syndrome[2].

Waardenburg syndrome (WS) is an auditory-pigmentary disorder that accounts for 2%-3% of congenital deafness. The estimated worldwide incidence is 2 to 3 cases per 100 000 population and equally affecting both genders and all races.28,29 There are four subtypes of WS that are defined phenotypically[2].

The diagnosis of WS1 can be established clinically using the Waardenburg Consortium criteria. WS2, WS3, and WS4 are defined by the absence of dystopia canthorum (ie, lateral displacement of the inner canthi), presence of musculoskeletal abnormalities, and aganglionic megacolon, respectively.

2 Case Report

Clinical History: One day old male baby presented with symmetrical depigmented patches over both lower legs, white forelock, hearing loss, broad nasal bridges, difficulty in passing Meconium since birth. Baby was delivered at 37 weeks period of gestation after an uneventful pregnancy through normal vaginal delivery; history of similar complaints in the father present. No h/o consanguinity among parents.

On Examination- Symmetrical solitary well defined depigmented patches of size 4x5cm present over both lower legs around knee joints. White forelock present over the frontal region of head. Broad nasal bridge with heterochromia of eyes, broad nasal root with hypoplastic nasal alae present. Brainstem Evoked Response Audiometry (BERA) test detected sensorineural hearing loss. All these clinical features without dystopia canthorum points to the clinical diagnosis of Waardenburg syndrome type II.

Fig. 1

Fig. 2

Fig. 3

Fig. 4

- Fig. 1 & Fig. 2 shows hypertelorism, broad nasal bridge, white forelock, hypoplastic ala nasi.
- Fig. 3 & Fig. 4 shows symmetrical depigmented patches on both knees.

3 Discussion

A suspected diagnosis of Waardenburg syndrome should be accompanied by a thorough history and physical examination. A family history is important in identifying familial cases and should focus on pigment abnormalities, premature graying of the hair, hearing loss, and gastrointestinal complications. A detailed physical examination should be performed with particular attention to the craniofacial structures, genitalia, and potential neurological symptoms, including any delay in the developmental milestones.

The pattern of depigmentation and/or presence of a white forelock helps to differentiate them from vitiligo and other hypopigmented conditions Nevus anemicus, Nevus depigmentosus, Tuberous sclerosis complex (TSC) and Hypomelanosis of Ito in the newborn. A skin biopsy is not necessary or adequate in confirming the diagnosis of WS[4].

The management of Waardenburg syndrome involves a multidisciplinary team approach, patient education, and early intervention in selected patients. All patients with WS, and their families, should be offered genetic counselling and testing.

Skin depigmentation associated with Piebaldism and WS is stable but lacks melanocytes and inflammation; as a result,

light therapy and corticosteroids have no role in therapy. Optimal skin photo-protection should be recommended. Management with cosmetic camouflage and hair dye can improve quality of life and provide significant emotional benefit.

Waardenburg syndrome is a high-risk indicator for hearing loss; as such, even if the newborn screening is passed, the American Academy of Pediatrics recommends a referral for at least one diagnostic auditory assessment. The comprehensive audiological evaluation should be performed no later than 3 months of age. Confirmed hearing loss requires appropriate referral (otolaryngology, speech-language pathology, audiology, genetics) and intervention by no later than 6 months of age. Congenital sensorineural hearing loss treated with cochlear implants improves language, communication, and cognitive skills.

During follow-up visits, the patient should be asked about constipation because Hirschsprung disease may not be apparent early in life.

5 Conclusion

The increasing use of whole-genome sequencing will hopefully identify novel genes that are responsible for the unexplained cases of Waardenburg syndrome WS. We reported a rare case of Waardenburg syndrome satisfying criteria for Type II. Knowledge of this syndrome helps in its early identification and diagnosis which in turn helps in its early management.

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