

Waardenburg Syndrome: A Rare Disorder with an Uncommon Manifestation in a Neonate

Sir,

Waardenburg syndrome (WS) is an autosomal recessive disorder that occurs in one out of every 40,000 to 50,000 live births.¹ As a clinical entity, it has four recognisable subtypes with multisystem involvement of hair, skin, iris, and gastrointestinal system. Hearing loss is a crucial aspect of the disorder. Type IV WS, associated with Hirschsprung's disease (HD), is also known as Shah-Waardenburg syndrome (SWS), with only a few reported cases in the literature.² The patient, in this case, exhibited symptoms of WS along with intestinal obstruction, however, aganglionosis was not detected on intestinal biopsy as anticipated. Additionally, the baby had extensive skin pigmentation, which has not been documented in previous cases.

A male infant was delivered *via* lower segment caesarean section at 35 ± 2 weeks of gestation, with good Apgar at birth. The mother's antenatal history revealed a diagnosis of gestational diabetes and polyhydramnios. Antenatal scans indicated dilated gut loops and elevated Doppler ultrasound indices. The parents were consanguineous, with a family history of WS in the paternal uncle and aunt.

Postnatally, the baby boy weighed 2.75 kg and displayed a respiratory rate of 56 breaths per minute and a heart rate of 156 beats per minute. He had syndromic facies, such as a broad nasal bridge, a white forelock, and large and multiple hypopigmented lesions on the abdomen, limbs, and forehead (Figure 1).



Figure 1: Hypopigmented lesions on limbs, abdomen and forehead.

The positive family history and typical phenotypic clinical features were suggestive of a diagnosis of WS. He had a distended abdomen with no other visible systemic abnormalities. Following consultation with a paediatric surgical team, the workup revealed jejunal atresia. A laparotomy was performed which showed Type IV jejunal atresia with a constricted portion of the distal ileum. A jejunioileal anastomosis was performed, and a functional ileostomy was established in the right iliac fossa. Biopsies were taken and sent for histopathology, with anticipated results to show aganglionosis, as WS has a high association with HD, especially when intestinal obstruction is

present. However, the biopsy results showed normal ganglionic segments, ruling out HD. Following a 10-day postoperative period of nil per oral (NPO), the patient was gradually able to tolerate full feeds and was discharged from the clinic for routine follow-up.

We present a unique case in which a child had clinical features suggestive of SWS; however, the biopsy did not reveal HD which is reported to be the cause of intestinal obstruction in literature. Earlier, we reported a case of SWS wherein the child exhibited clinical features of WS, and the biopsy showed aganglionosis.³ Other cases have also been documented in the literature.^{1,4,5} Nonetheless, there are no reported cases of WS with intestinal obstruction not due to HD. Moreover, hypomelanotic skin lesions are typically present in such children, but widespread lesions as observed in this case, are rare.

In summary, it is important to note that not all instances of intestinal obstruction in WS are caused by HD. Intestinal obstruction can be an isolated finding, along with other classical manifestations of the syndrome. Hence, it is important to confirm all such cases through intestinal biopsy.

PATIENT'S CONSENT:

Consent was taken from the parents of the patient.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

AK: Conception and design of the work, acquisition of data, drafting the work, and critical revision for important intellectual content.

UA: Design of the work, analysis of the data, and critical revision for important intellectual content.

MH: Design of the work, acquisition and interpretation of the data, and drafting the work for important intellectual content.

All authors approved the final version of the manuscript to be published.

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Arshad Khushdil, Umair Anees and Moez Hussain
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Department of Paediatrics, Pak Emirates Military Hospital,
Rawalpindi, Pakistan
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Correspondence to: Dr. Arshad Khushdil, Department of Paediatrics, Pak Emirates Military Hospital, Rawalpindi, Pakistan

E-mail: drarshad104589@yahoo.com

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