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WAARDENBURG SYNDROME: RARE CASE REPORT

Paediatrics

Dr. Pankaj Narwade — Department Of Surgery Nkp Salve Institute Of Medical Sciences, Digdoh Hills, Hingna Road, Nagpur 440019, Maharashtra, India

Dr. Roopal Khobragade* — Department Of Paediatrics, Nkp Salve Institute Of Medical Sciences, Digdoh Hills, Hingna Road, Nagpur 440019, Maharashtra, India *Corresponding Author

Dr. Balwant Khobragade — Department Of Paediatrics, Nkp Salve Institute Of Medical Sciences, Digdoh Hills, Hingna Road, Nagpur 440019, Maharashtra, India

Dr. Manisha Albal — Department Of Paediatric Surgery Nkp Salve Institute Of Medical Sciences, Digdoh Hills, Hingna Road, Nagpur 440019, Maharashtra, India

ABSTRACT

The Waardenburg-Shah syndrome (WSW) is an autosomal recessive disease with varied presentation where Hirschsprung's disease and the Waardenburg syndrome are seen together. Majority of patients have total colonic aganglionosis with or without small bowel involvement. This is a rare syndrome so we present in this study a neonate full term male 2.7 kg delivered by caesarian section presented with white forelock and multiple colonic biopsy confirmed to have Hirschprungs disease.

CONCLUSION: This is a rare disorder in neonatal period with varied presentation. Surgical management may be used in these patients to gain some time for the child to grow and to decrease complications associated with total parenteral nutrition. However prognosis is very poor. An early diagnosis and improvement of hearing impairment with timely intervention are the most important for children with Waardenburg syndrome.

KEYWORDS

Waardenburg syndrome, Hair hypopigmentation, Sensorineural hearing loss

INTRODUCTION

Waardenburg syndrome is a group of genetic conditions that can cause hearing loss and changes in coloring (pigmentation) of the hair, skin, and eyes. Although most people with Waardenburg syndrome have normal hearing, moderate to profound hearing loss can occur in one or both ears. The hearing loss is present from birth (congenital). People with this condition often have very pale blue eyes or different colored eyes, such as one blue eye and one brown eye. Sometimes one eye has segments of two different colors. Distinctive hair coloring (such as a patch of white hair or hair that prematurely turns gray) is another common sign of the condition. Overall, the syndrome affects around 1 in 42,000 people Nayak CS, Isaacson G. (2003), and genetically heterogeneous disease accounts for >2 % of the congenitally deaf population Morell R, Spritz A. (1997). The features of Waardenburg syndrome vary among affected individuals, even among people in the same family.

There are four recognized types of Waardenburg syndrome, which are distinguished by their physical characteristics and sometimes by their genetic cause. Types I and II have very similar features, although people with type I almost always have eyes that appear widely spaced and people with type II do not. In addition, hearing loss occurs more often in people with type II than in those with type I. Type III (sometimes called Klein-Waardenburg syndrome) includes abnormalities of the arms and hands in addition to hearing loss and changes in pigmentation. Type IV (also known as Waardenburg-Shah syndrome) has signs and symptoms of both Waardenburg syndromeand Hirschsprung disease, an intestinal disorder that causes severe constipation or blockage of the intestine.

CASE SUMMERY

Neonate 2.7 kg male baby delivered by caesarian section presented with Bilious vomiting , Failure to pass meconium in the first 72 hours , Jaundice and Progressive abdominal distention. Parent had 2 siblings with similar presentation: 1st sibling died on day of life-1 and 2nd died on Day of life -6 with unknown cause. Baby on examination had white forelock of hair in frontal area and massively distended abdomen, pale irises, and absence of reaction to any sound. It was admitted to NICU for further management.

Fihure 1- Dermatological Lesions In Waardenburg Syndrome (showing White Forelock)

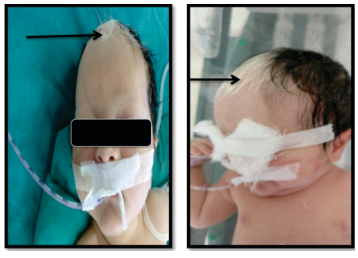


Figure 2- Barium Study Was Suggestive Of Dilated Bowel Loops With Suspicion Of Hirschprung Disease.

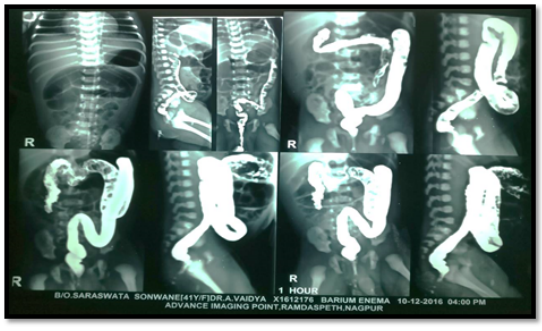
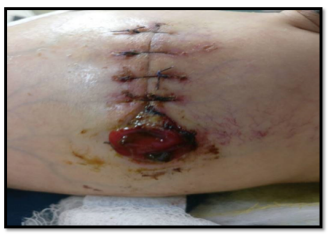


Figure 3- Ileostomy Performed With Biopsy From Distal Collapsed Segment.



HPE Report s/o aganglionosis in colon and terminal ileum, compatible with Hirschsprung disease.

A diagnosis of Shah- Waardenburg syndrome(SWS) was made

The iloestomy started to function 4th postoperative day, and the baby initially showed the signs of improvement.

The iloestomy started to function 4th postoperative day, and the baby initially showed the signs of improvement.

However, the baby died on 6th day because of sepsis.

- SWS is the association of Waardenburg syndrome (WS) with Hirschsprung disease.
- Only 48 cases are reported in English literature till 2002 .
- The incidence of WS is 2/100,000 and of HD 2/10,000.
- WS Characterized by
- Congenital sensory neural hearing loss
- Partial albinism
- Lateral displacement of inner cannthi.
- Diagnostic criteria for Waardenburg syndrome :
- five major criteria
- five minor criteria.

DIAGNOSTIC CRITERIA FOR WAARDENBURG SYNDROME

According to the diagnostic criteria proposed by the Waardenburg consortium, a person must have two major or one major plus two minor criteria to be diagnosed as Waardenburg syndrome from Farrer A, Kenneth M, and Amos GJ. (1992).

MAJOR CRITERIA:-
MINOR CRITERIA:-

a) Skin hypopigmentation
b) Synophrys
c) Broad/high nasal root
d) Hypoplastic alae nasi
e) Premature graying of the hair.
f) The clinical diagnosis of WS requires at least 2 major criteria or 1 major and 1 minor criteria .

There are four types of Waardenburg syndrome out of which type, IV is very rare and is known as SHAH WAARDENBURG SYNDROME and Waardenburg syndrome in association with Hirschprung Disease

SHAH WAARDENBURG SYNDROME (AR) (Endothelin EDN, EDNRB and SOX 10)

a) Waardenburg syndrome + Hirschprung disease
b) Bilious vomiting
c) Enterocolitis-associated diarrhea
d) Failure to pass meconium in the first 24 hours of life
e) Infrequent, explosive bowel movements; difficult bowel movements
f) Jaundice
g) Poor feeding
h) Progressive abdominal distention
i) Tight anal sphincter with an empty rectum

CLINICAL TYPE

a) Waardenburg syndrome is a group of rare genetic conditions. Features vary among affected individuals, even among people in the same family.
b) Shah-Waardenburg syndrome is very rare. Defective migration of the neural crest derived cell lines, melanocytes and the neuroblasts (contributing the enteric ganglion cells) during the embryonic phase, has been postulated as a cause of this disorder .
c) Bowel involvement in Shah-Waardenburg syndrome is characteristic in the form of aganglionosis in the myenteric (Auerbach) plexus and the submucous (Meissner) plexus, with long-segment Hirschsprung disease.

The unusual finding in this case was total colonic aganglionosis and familial incidence of Shah- Waardenburg syndrome.

a) Patients with Shah-Waardenburg syndrome and total colonic aganglionosis cases usually present within first month of life with delayed passage of meconium (beyond 48 hours) or constipation since birth or with features of neonatal small bowel obstruction (as seen this case), although cases presenting beyond the neonatal period have been reported.
b) The differential diagnoses of such presentation are ileal atresia, neonatal small left colon syndrome, meconium ileus, meconium plug syndrome, and intestinal neuronal dysplasias.

CONCLUSION

All the primary care Initial surgical approach in Shah-Waardenburg syndrome is histopathological confirmation of diagnosis by routine seromuscular colonic biopsy and stoma formation depending upon the involvement of the colon. Frozen section examination if available remains a useful diagnostic modality for this purpose with its inherent advantages of prompt intraoperative diagnosis. The definitive treatment of Hirschsprung's disease is performed at a later date. Genetic counselling must be provided for families with this disorder and Mutational analysis was not possible due to resource constraints.

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Conflict of Interest

The authors declare that they have no conflict of interest.

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