International Journal of Community Medicine and Public Health
Suliman O et al. Int J Community Med Public Health. 2026 Apr;13(4):2073-2076
http://www.ijcmph.com
pISSN 2394-6032 | eISSN 2394-6040

DOI: https://dx.doi.org/10.18203/2394-6040.ijcmph20261052

Case Report

Shah-Waardenbrug syndrome type IV presenting with long segment Hirschsprung's disease in a neonate: a case report

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Received: 21 February 2026
Accepted: 20 March 2026

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ABSTRACT

Shah–Waardenburg syndrome, also known as Waardenburg syndrome type 4, is a rare genetic condition characterized by the combination of pigmentary abnormalities and Hirschsprung disease resulting from neural crest developmental defects. We present a case of a 5-day-old male neonate with dysmorphic features, bilious vomiting, abdominal distension, and failure to pass meconium, later confirmed to have total colonic aganglionosis. Early surgical intervention and histopathological evaluation were essential in confirming the diagnosis and guiding management. A 5-day-old male neonate born at term by normal vaginal delivery at home to a 20-year-old mother with limited antenatal care was referred with refusal to breastfeed since birth and progressive bilious vomiting beginning on the second day of life. The infant had not passed meconium spontaneously. There was no history of similar illness in the family. Shah–Warrensburg syndrome with total colonic aganglionosis is a rare but life-threatening condition that should be suspected in neonates presenting with intestinal obstruction and characteristic dysmorphic features. Early recognition and prompt surgical intervention are crucial to prevent morbidity and mortality. Histopathological confirmation remains the gold standard for diagnosis and guides definitive management. A multidisciplinary approach involving neonatology, pediatric surgery, and genetics is essential for optimal outcomes. Long-term follow-up is required to address surgical, nutritional, and developmental challenges.

Keywords: Shah–Warrensburg syndrome, Total colonic aganglionosis, Hirschsprung's disease

INTRODUCTION

Warrensburg syndrome is a rare neurocristopathy that may follow autosomal dominant or autosomal recessive inheritance patterns, depending on the gene involved. It is characterized by variable combinations of pigmentary abnormalities of the skin, hair, and eyes, congenital sensorineural hearing loss, and distinctive craniofacial features such as dystopia canthorum.[1] These manifestations arise from defective migration, proliferation, or differentiation of neural crest cells, which play a critical role in the development of melanocytes, craniofacial structures, and components of the peripheral nervous system.[2-7]

Shah-Waardenbrug syndrome, also known as Waardenbrug syndrome type IV, represents a more complex variant in which the classic phenotype is associated with Hirschsprung's disease.[2] The intestinal involvement results from aganglionosis of the distal bowel due to failed development of the enteric nervous system, leading to functional intestinal obstruction in the neonatal

period.[5] Clinically, affected neonates often present with delayed passage of meconium, progressive abdominal distension, bilious vomiting, and feeding intolerance.[3]

The extent of aganglionosis in Shah–Waardenbrug syndrome is variable, ranging from short-segment disease to total colonic aganglionosis, the latter representing one of the most severe forms.[8] Long-segment aganglionosis accounts for approximately 15–20% of cases of Hirschsprung's disease and is associated with significant surgical and nutritional challenges. These patients commonly require staged surgical procedures and prolonged postoperative care. Long-term morbidity may include persistent bowel dysmotility, frequent stools, electrolyte disturbances, and impaired growth.[4] Therefore, early diagnosis of Shah–Waardenbrug syndrome is crucial to prevent life-threatening complications such as Hirschsprung's-associated enterocolitis and bowel perforation, and to facilitate timely surgical intervention, genetic evaluation, and coordinated multidisciplinary management.[5]

CASE REPORT

Physical examination

The neonate appeared mildly dehydrated and febrile, with a respiratory rate of 54 breaths per minute and a pulse rate of 109 beats per minute. Dysmorphic features were noted, including very light, depigmented hair, blue irises, a broad nasal bridge, and laterally displaced ears. The abdomen was distended with visible bowel loops, yet remained soft and non-tender on palpation. On digital rectal examination, the anus was normally positioned, and a gush of meconium was expelled following withdrawal of the examining finger. Bowel sounds were hypoactive, and no palpable abdominal masses were detected. The remainder of the systemic examination, including cardiovascular and neurological assessments, was unremarkable (Figure 1).

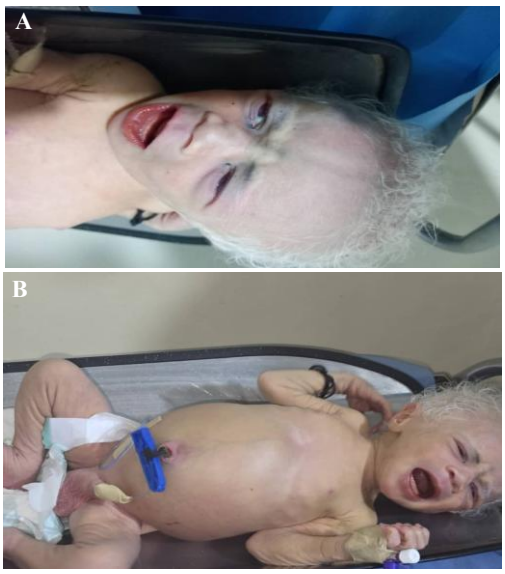

Figure 1 (A and B): Dysmorphic and clinical features.

Laboratory investigations

The total count was within normal limits, with the platelet count slightly above the usual upper limit. Serum sodium levels indicated mild hypernatremia, while potassium values were at the upper limit of the normal range. Urea and creatinine levels were elevated, suggesting possible dehydration or impaired renal perfusion (Table 1).

Table 1: Laboratory tests.

Parameter	Result	Notes
White blood cell count (WBC)	8,000/mm³	Total count is normal
Hemoglobin	422,000/mm³	Slightly above usual upper limit
Sodium	148 mmol/l	Mild hypernatremia
Potassium	5.2 mmol/l	Upper normal range
Urea	50 mmol/l	Elevated
Creatinine	1.1 mg/dF	

Imaging

Erect plain abdominal radiograph demonstrated multiple dilated bowel loops with several air-fluid levels and a paucity of gas in the distal colon, findings consistent with distal bowel obstruction. No free intraperitoneal air was observed on the supine view, the loops appeared centrally located with mild diffuse distension. These radiographic features supported the clinical suspicion of Hirschsprung's-associated obstruction (Figure 2).

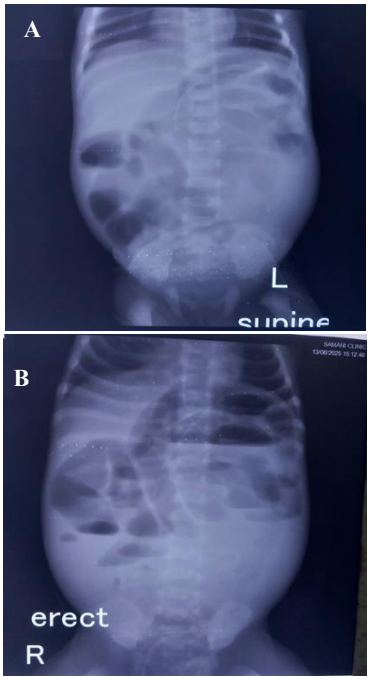

Figure 2 (A and B): Erect and supine radiologic features.

Intra-operative findings

Exploratory laparotomy revealed dilated proximal bowel and narrowed distal ileum and colon. Multiple intraoperative seromuscular biopsies were taken, and a diverting ileostomy was performed (Figure 3A and B).

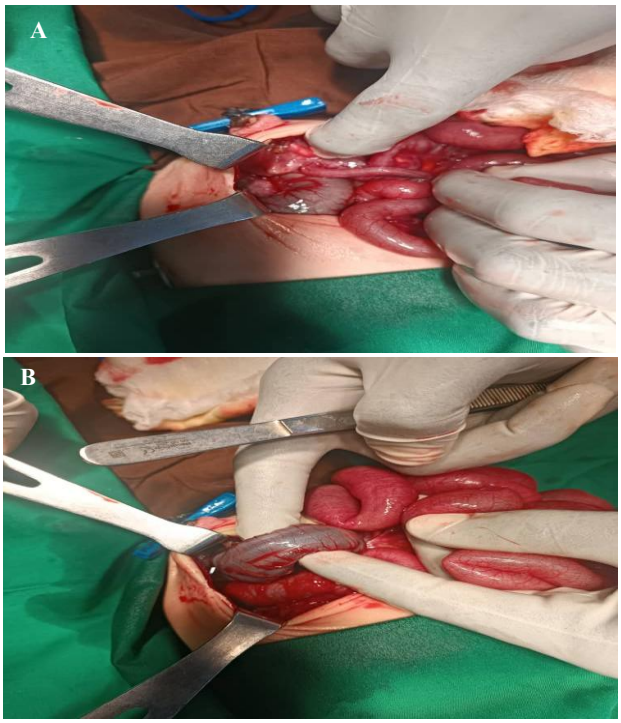

Figure 3 (A and B): Intraoperative findings.

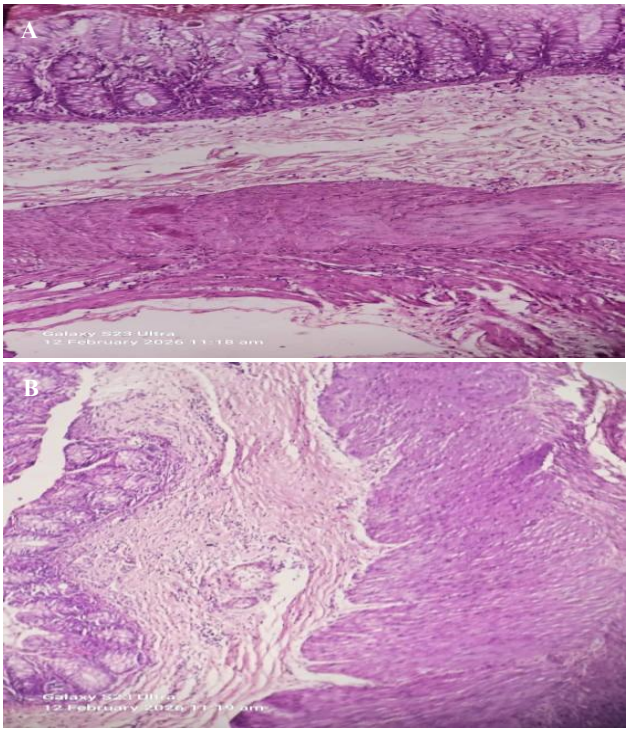

Figure 4 (A and B): Histopathology findings.

Histopathology

Rectum, sigmoid colon, transverse colon, and cecum: Complete absence of ganglion cells in both the submucosal (Meissner's) plexus and the myenteric (Auerbach's) plexus, accompanied by marked hypertrophy of submucosal nerve bundles, consistent with aganglionosis.

Ileum: normal histological architecture with preserved ganglion cells identified in both the submucosal (Meissner's) plexus and the myenteric (Auerbach's) plexus, with no evidence of nerve fiber hypertrophy (Figure 4A and B).

Management

The patient was stabilized with intravenous fluids, nasogastric decompression, and broad-spectrum antibiotics. A diverting ileostomy was performed to relieve intestinal obstruction and decompress the dilated bowel. Definitive surgical management is planned as a delayed pull-through procedure once the infant is clinically stable and nutritionally optimized.

Outcome

The postoperative course was uneventful, with good stoma function and gradual resolution of abdominal distension. The infant tolerated enteral feeding with appropriate weight gain during follow-up. He remains clinically stable and is scheduled for definitive pull-through reconstruction at a later stage.

DISCUSSION

Shah–Waardenbrug syndrome is a rare variant of Waardenbrug syndrome characterized by the association of pigmentary abnormalities, craniofacial dysmorphism, and Hirschsprung's disease.[6] The underlying pathophysiology involves mutations affecting genes such as EDNRB, EDN3, and SOX10, which impair the migration and differentiation of neural crest cells during embryogenesis.[9] As a result, both melanocytes and enteric ganglion cells are affected, explaining the simultaneous presence of depigmentation and intestinal aganglionosis.[1] Clinical expression is heterogeneous, and the severity of gastrointestinal involvement ranges from short-segment disease to total colonic aganglionosis, as observed in severe reported cases.[8]

Long-segment aganglionosis represents a particularly severe form of Hirschsprung's disease and often presents in the neonatal period with progressive abdominal distension, bilious vomiting, and failure to pass meconium.[3] Early diagnosis is critical, as delayed recognition increases the risk of Hirschsprung's-associated enterocolitis, which may rapidly progress to sepsis and significant morbidity.[5] In our case, the presence of characteristic pigmentary features raised early suspicion

for a syndromic diagnosis, prompting timely evaluation and confirmation.

Management is challenging and typically requires a staged surgical approach. An initial diverting enterostomy is commonly performed to decompress the bowel, improve feeding tolerance, and allow weight gain. Once growth and clinical stability are achieved, a definitive pull-through procedure can be planned. Postoperative care is equally important, as these patients frequently experience persistent dysmotility, electrolyte imbalances, and nutritional difficulties.[4] A multidisciplinary team including pediatric surgery, gastroenterology, nutrition, audiology, and genetics is essential for optimizing outcomes.[7]

Long-term prognosis varies and depends on the extent of aganglionosis, timing of diagnosis, and prevention of enterocolitis.[3,8] Children with Shah–Waardenbrug syndrome may also require ongoing management of associated features such as sensorineural hearing loss or developmental delay.[6,7,10] Early recognition of the syndrome, vigilant monitoring for gastrointestinal complications, and comprehensive postoperative nutritional support are key factors in improving survival and quality of life.

CONCLUSION

This case underscores the importance of recognizing dysmorphic features associated with neonatal intestinal obstruction. Early diagnosis of Shah–Waardenbrug syndrome with long segment aganglionosis allows timely surgical intervention and improves outcomes. Long-term follow-up is essential for nutritional rehabilitation and bowel function monitoring.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Suliman O, Bireama R, Abdalla E, Elnour A, Nugud F. Shah-Waardenbrug syndrome type IV presenting with long segment Hirschsprung's disease in a neonate: a case report. Int J Community Med Public Health 2026;13:2073-6.