

Waardenburg–Shah Syndrome: Diagnostic and Surgical Challenges in a Resource-Limited Setting — A Rare Case Report

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Introduction and Importance: Waardenburg–Shah syndrome (WSS) is a rare congenital disorder characterized by the coexistence of Waardenburg syndrome features and Hirschsprung disease. It results from abnormal neural crest cell migration, leading to variable inheritance patterns and diverse phenotypic presentations.

Case Presentation: We report a 13-day-old Somali neonate admitted with abdominal distension, bilious vomiting, poor feeding, and fever since birth. Physical examination revealed sharply demarcated hypopigmented patches on the chest and face, bilateral blue irides, and craniofacial dysmorphism. Automated auditory brainstem response (AABR) screening indicated bilateral “refer” outcomes, suggesting sensorineural hearing impairment. Abdominal imaging suggested Hirschsprung disease, and laparotomy revealed long-segment aganglionosis with ganglion cells preserved only in the ileum. Sequential biopsies confirmed absence of ganglion cells in the appendix and sigmoid colon. The patient underwent prophylactic appendectomy, creation of a loop ileostomy, and later definitive repair using the modified extended Duhamel procedure—a pull-through technique that combines a retrorectal pathway with side-to-side anastomosis to bypass the aganglionic segment while preserving continence. She recovered well and was discharged on postoperative day twelve.

Clinical Discussion: Diagnosis of WSS relies on recognizing the coexistence of pigmentary abnormalities, auditory impairment, and intestinal aganglionosis. Clinical presentation varies depending on the length of the aganglionic segment, ranging from late-onset constipation in short-segment cases to neonatal intestinal obstruction in long-segment disease. Management requires a multidisciplinary approach, including early hearing evaluation, dermatologic assessment, and surgical correction tailored to disease extent. In low-resource settings without access to genetic testing, clinical evaluation and histopathology remain crucial for accurate diagnosis and treatment planning.

Conclusion: This case highlights the diagnostic challenges and multidisciplinary care required for Waardenburg–Shah syndrome in a resource-limited environment. Early recognition of characteristic features, supported by timely surgical intervention and coordinated care, is essential for optimizing outcomes in affected neonates.

Keywords: Waardenburg–Shah syndrome, Waardenburg syndrome type IV, Hirschsprung disease, long-segment aganglionosis, modified extended Duhamel procedure, case report

Introduction

Waardenburg syndrome (WS) is a rare, genetically inherited auditory-pigmentary disorder that arises from abnormal neural crest cell migration. It is characterized by a wide spectrum of clinical features and is subdivided into four recognized types (WS1–4), with types 3 and 4 being particularly uncommon.^{1,2}

Waardenburg–Shah syndrome (WS type IV) represents an especially rare congenital variant, marked by significant phenotypic variability. It combines the pigmentary manifestations of WS—such as a white forelock, premature graying of hair, and iris pigmentary abnormalities (heterochromia or homochromia)—with congenital sensorineural deafness and Hirschsprung disease.³ The estimated prevalence of Waardenburg–Shah syndrome is approximately 1 in 50,000 live births.¹

The inheritance pattern is variable, with *SOX10* mutations most often displaying autosomal dominant transmission, whereas *EDNRB* and *EDN3* mutations are typically inherited in an autosomal recessive manner.^{3–5}

We report a Somali neonate presenting with intestinal obstruction and syndromic features consistent with Waardenburg–Shah syndrome, emphasizing the diagnostic and management challenges of this rare condition in a resource-limited setting. This case contributes to the limited literature by documenting a rare long-segment form of the disease successfully managed in Somalia, where access to genetic testing and comprehensive audiologic evaluation remains limited. To our knowledge, only a few cases have been reported from Somalia or the wider Horn of Africa. By presenting this case, we highlight the diagnostic and surgical complexities encountered in low-resource environments and demonstrate that, with coordinated multidisciplinary care, favorable outcomes are achievable even in constrained settings.

Case Presentation

A 13-day-old term female neonate was admitted to the neonatal intensive care unit (NICU) with abdominal distension present since birth, associated with poor feeding, bilious (green) vomiting, and fever.

She was delivered in Garowe, a rural area of Somalia, to a 30-year-old multiparous mother (G7P7) with no history of chronic illness. The pregnancy was uneventful, and there were no reports of maternal fever, rash, or complications. Delivery occurred via normal spontaneous vaginal delivery at a local hospital. The neonate cried immediately after birth, although Apgar scores were not documented. She passed both urine and meconium shortly after delivery. There was no family history of Waardenburg syndrome or Hirschsprung disease.

On admission, the neonate was hemodynamically stable and appeared generally well. Dermatological examination revealed sharply demarcated hypopigmented patches on the scalp, face, and upper chest, extending beyond the nipple line (Figure 1A–D). The abdomen was distended with visible superficial veins but no palpable organomegaly. The anal canal was patent with minimal stool in the rectum. Respiratory assessment showed normal effort with SpO₂ >92% on room air, and auscultation revealed vesicular breath sounds without added sounds. Cardiovascular examination revealed normal S1 and S2 without murmurs.

Ophthalmologic evaluation (OCT) revealed bilateral iris hypopigmentation (blue irides) (Figure 2). Automated Auditory Brainstem Response (AABR) screening performed at 35 dB nHL yielded bilateral “refer” results. Because AABR provides only pass/refer outcomes and does not generate waveforms such as wave V, these findings indicate screening failure and raise suspicion of bilateral sensorineural hearing loss. Diagnostic ABR—required for waveform analysis and confirmation—was not available at our facility; thus, the assessment was performed using the available AABR system while the infant was in natural sleep (Figure 3A and B).

Initial investigations included routine blood work, chest and abdominal radiography, and ultrasonography. Abdominal X-ray demonstrated markedly dilated bowel loops, primarily involving the distal colon and rectum, with a clear transition zone between the dilated proximal bowel and narrowed distal rectum, consistent with Hirschsprung disease (Figure 4A and B).

At exploratory laparotomy, the colon was diffusely narrow with an “unused colon” appearance, and a transition zone was identified approximately 15 cm proximal to the ileocecal valve. Sequential biopsies were obtained from the appendix (prophylactic appendectomy), sigmoid colon, and ileum at the planned stoma site. A loop ileostomy was fashioned in the upper left abdomen through an oblique incision, and the patient was transferred back to the NICU.

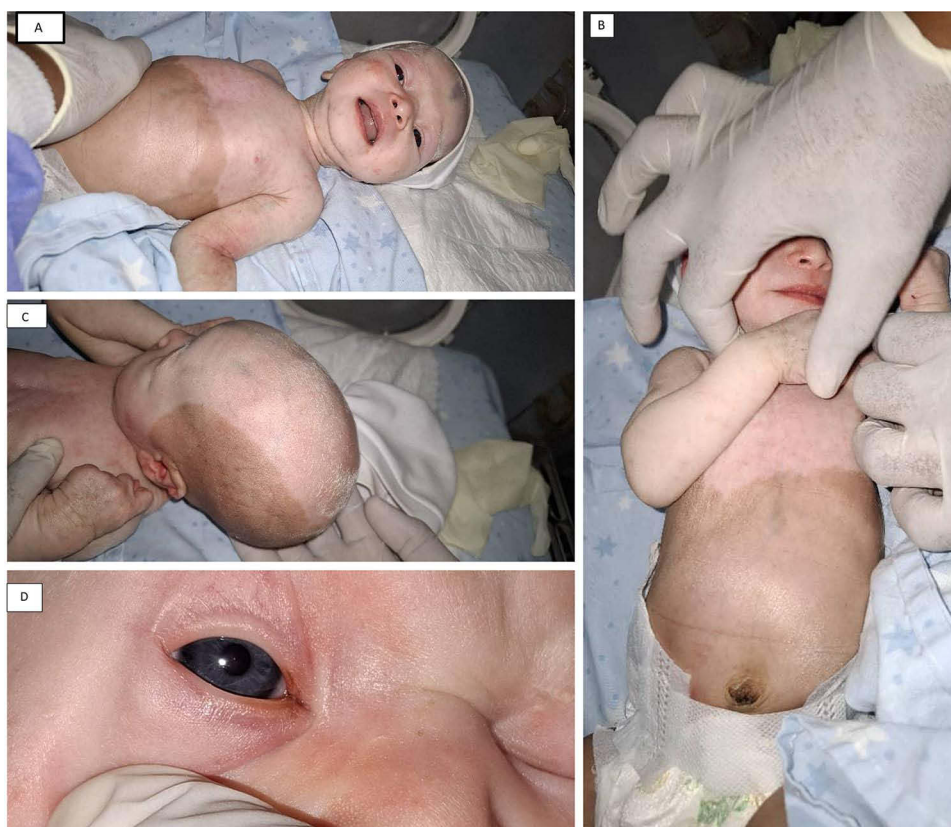


Figure 1 Clinical features of Waardenburg syndrome in a neonate. (A) Hypopigmented patch extending from the scalp to the upper chest with sharply demarcated borders. (B) Hypopigmentation involving the upper chest and extending to both arms. (C) Large hypopigmented scalp area with clear and well-defined margins. (D) Close-up image of the right eye demonstrating marked iris hypopigmentation (blue iris).

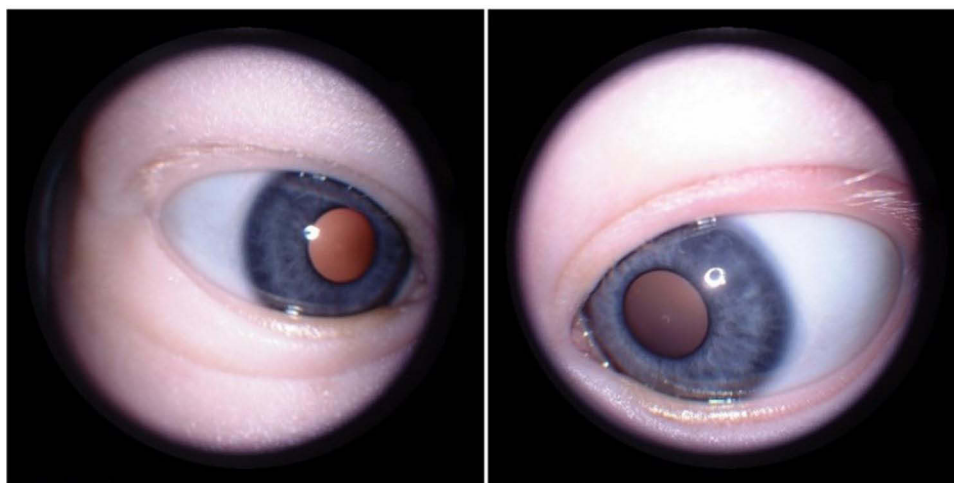


Figure 2 Ophthalmologic examination of the patient. Anterior segment photographs demonstrating bilateral iris hypopigmentation (blue irides), consistent with the ocular features of Waardenburg syndrome.

Histopathology revealed the presence of ganglion cells in the ileal biopsy, while both the sigmoid colon and appendix showed absence of ganglion cells and hypertrophied nerve bundles, confirming the diagnosis of Hirschsprung disease (Figure 5A–C).

A summary of the key clinical, radiological, and histopathological findings is presented in Table 1.

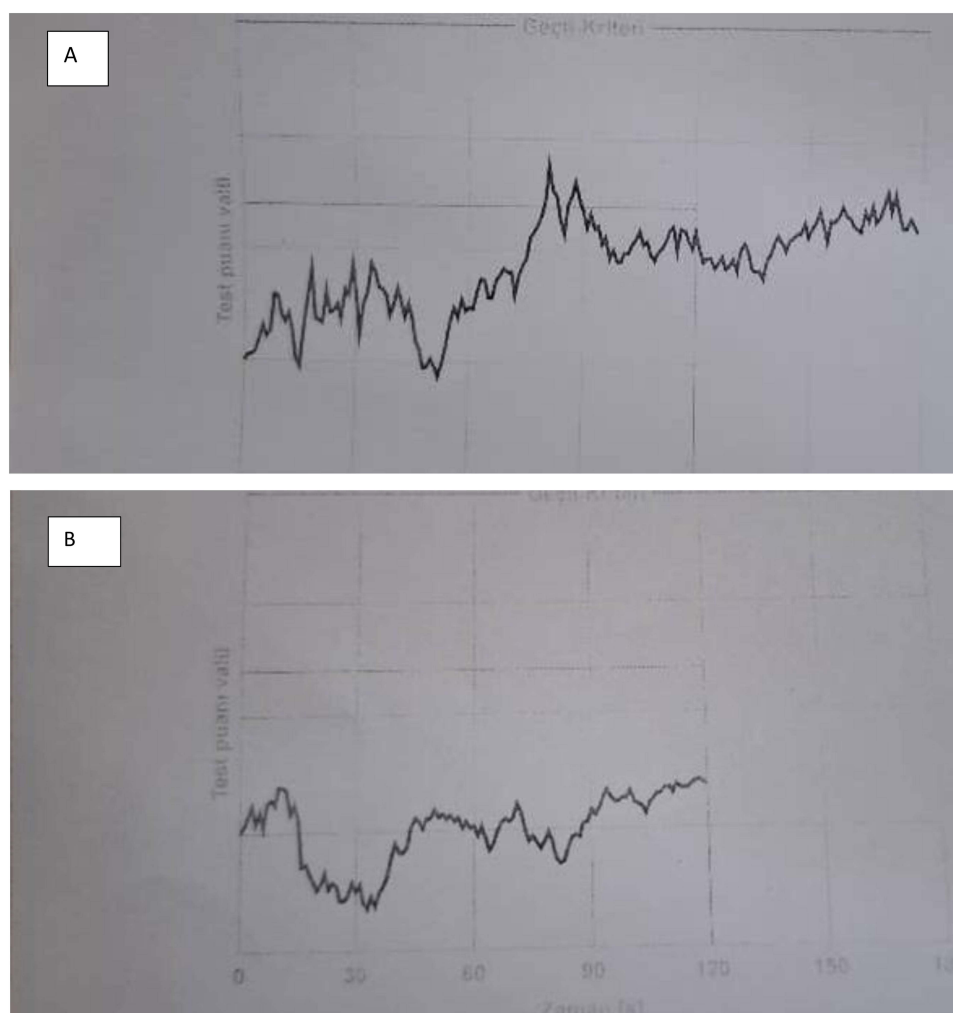


Figure 3 Automated Auditory Brainstem Response (AABR) screening at 35 dB nHL. **(A)** Left-ear AABR showing a “refer” result with no detectable screening response. **(B)** Right-ear AABR also demonstrating a “refer” outcome. Because AABR provides only pass/refer outputs and does not generate diagnostic waveforms (such as wave V), the bilateral “refer” results indicate screening failure suggestive of sensorineural hearing loss. A diagnostic ABR, which would allow waveform interpretation and degree-of-loss determination, was not available at our facility at the time of evaluation.

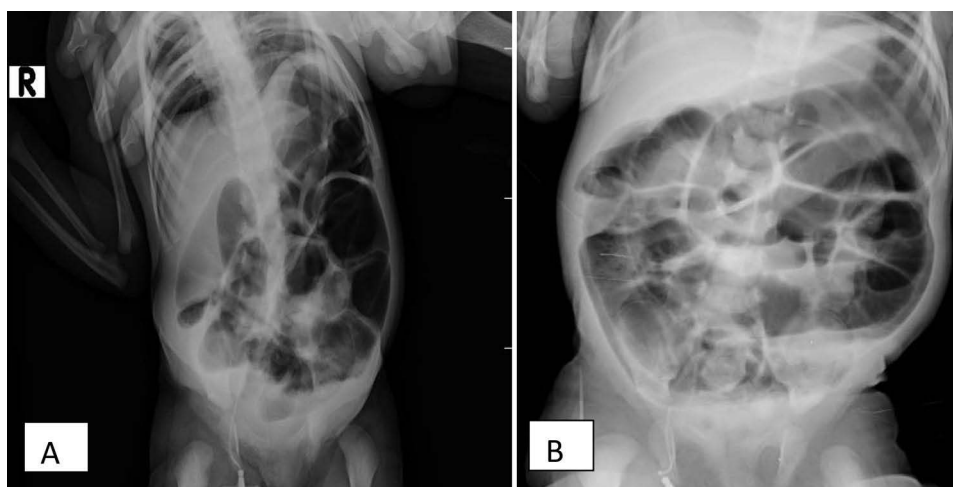


Figure 4 Abdominal radiological findings. **(A and B)** Abdominal imaging showing marked dilation of bowel loops involving the distal colon and rectum, with a distinct transition zone between dilated proximal bowel and a narrowed distal segment at the proximal rectum—findings characteristic of Hirschsprung disease.

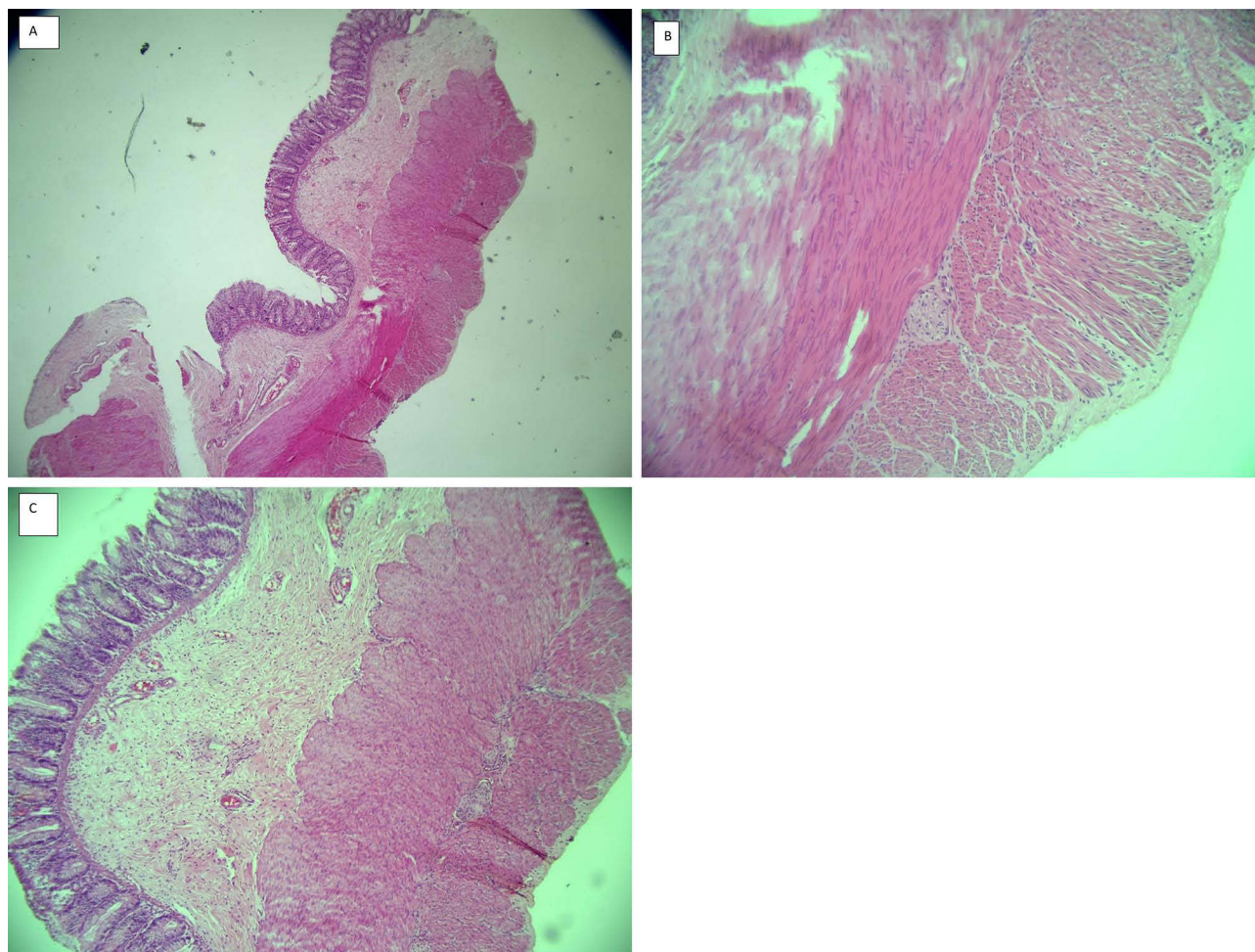


Figure 5 Histopathological findings of sigmoid colon biopsy in Hirschsprung disease. **(A)** Hematoxylin and eosin (H&E) section at ×4 magnification showing mucosal and submucosal architecture. **(B)** H&E section at ×10 magnification demonstrating absence of ganglion cells in the myenteric region and the presence of hypertrophic nerve bundles (arrow). **(C)** H&E section at ×10 magnification confirming complete absence of ganglion cells in both the submucosal and myenteric plexuses.

Subsequently, the patient underwent definitive correction with a modified extended Duhamel procedure, which was successfully performed. Postoperative recovery was uneventful, and the patient was discharged on the 12th postoperative day in good condition.

Table 1 Summary of Diagnostic Findings in the Neonate

Diagnostic Domain	Findings
Clinical Presentation	Abdominal distension since birth; poor feeding; bilious vomiting; fever
Dermatologic Findings	Sharply demarcated hypopigmented patches on scalp, face, and upper chest
Ophthalmologic Findings	Bilateral iris hypopigmentation (blue irides) on OCT
Auditory Testing (ABR)	Absent wave V bilaterally at 35 dB nHL → bilateral sensorineural hearing loss
Respiratory Examination	Normal effort; SpO ₂ >92% on room air; vesicular breath sounds

(Continued)

Table 1 (Continued).

Diagnostic Domain	Findings
Cardiovascular Examination	Normal S1/S2, no murmurs
Abdominal X-ray	Dilated bowel loops; transition zone at distal colon/rectum consistent with Hirschsprung disease
Intraoperative Findings	Unused colon appearance; transition zone ~15 cm proximal to ileocecal valve
Histopathology	Ileum: ganglion cells present; Sigmoid colon and appendix: absent ganglion cells with hypertrophied nerve bundles → confirmed Hirschsprung disease
Final Diagnosis	Waardenburg–Shah syndrome (Type IV) with Hirschsprung disease

Notes: Concise overview of the clinical, dermatological, ophthalmological, auditory, radiological, intraoperative, and histopathological findings that contributed to the diagnosis of Waardenburg–Shah syndrome (Type IV) with Hirschsprung disease.

Discussion

Waardenburg–Shah syndrome (WSS) is a rare clinical entity whose diagnosis relies on recognizing the constellation of features from both Waardenburg syndrome (WS) and Hirschsprung disease (HSCR). Our patient exhibited multiple phenotypic features consistent with WS, including hypopigmented skin patches, bilateral blue irides, craniofacial dysmorphism, and bilateral sensorineural hearing loss confirmed by ABR screening. The coexistence of intestinal obstruction with radiological findings of a transition zone and histopathological confirmation of aganglionosis established the diagnosis of Hirschsprung disease, thereby fulfilling the criteria for WSS.

Waardenburg syndrome was first described by Petrus Johannes Waardenburg in 1951, with characteristic features including lateral displacement of the medial canthi, heterochromia of the iris, white forelock, and congenital deaf-mutism.⁵ Over the following two decades, several studies further classified WS into subtypes: type I with dystopia canthorum, type II without dystopia canthorum, type III (Klein–Waardenburg syndrome) with upper limb anomalies, and type IV (Waardenburg–Shah syndrome) associated with Hirschsprung disease.^{6–9}

Diagnostic criteria for WS are based on five major and five minor features. The major criteria include sensorineural hearing loss, iris pigmentary abnormalities such as heterochromia, segmental or brilliant blue eyes, hair hypopigmentation (white forelock or localized white patches), dystopia canthorum (lateral displacement of the inner canthi), and having a first-degree relative with a confirmed diagnosis. The minor criteria consist of congenital skin depigmentation (leukoderma or white patches), medial eyebrow flare (synophrys), broad nasal root, hypoplasia of the alae nasi, and premature graying of the hair.^{1,2,5,6,10}

The clinical presentation of WSS varies according to the length of the aganglionic segment. Patients with short-segment involvement often present later with chronic constipation, malabsorption, or enterocolitis, whereas those with long-segment involvement typically present in the neonatal period with intestinal obstruction, bilious vomiting, abdominal distension, and feeding difficulties.¹⁰

Diagnosing WSS requires a high index of suspicion, as its rarity and phenotypic variability complicate recognition, particularly in the absence of family history.^{5,6,9} A standard evaluation includes a detailed family history, clinical and audiological examination of relatives, imaging to detect intestinal obstruction, rectal biopsy with histopathological confirmation of aganglionosis, and auditory brainstem response (ABR) testing to assess sensorineural hearing loss.^{10,11} Genetic counseling is also essential, as it guides families on inheritance, prognosis, and the importance of timely interventions.^{10,11}

In our case, the neonate presented with abdominal distension and bilious vomiting, alongside hypopigmented patches, bilateral blue irides, and bilateral sensorineural hearing loss confirmed by ABR. Imaging and histopathology supported the diagnosis of Hirschsprung disease, while the constellation of pigmentary, ocular, and auditory features confirmed WSS. Although genetic testing was not available, careful clinical recognition enabled timely diagnosis and early surgical as well as supportive management, emphasizing the value of multidisciplinary care in resource-limited settings.

Management of WSS is inherently multidisciplinary, addressing hearing loss, skin care, and intestinal involvement. Surgical correction of Hirschsprung disease must be tailored to the length of the aganglionic segment: the Soave endorectal pull-through or Swenson pull-through is commonly performed in short-segment disease, while the modified extended Duhamel procedure is preferred for long-segment involvement; in cases of total colonic aganglionosis, the Kimura–Stringel operation may be indicated.^{10,12,13} Thus, the choice of surgical technique and overall management strategy varies according to whether short- or long-segment disease is present.

In our patient, long-segment aganglionosis with preservation of ganglion cells in the ileum was initially managed by ileostomy, followed by definitive correction using the modified extended Duhamel procedure, which was successfully performed.

In contrast to the Mongolian WS4 cases, which benefited from genetic confirmation and comprehensive multidisciplinary management, and the previously reported Somali case that culminated in early septic mortality, our patient experienced a smooth postoperative course, reflecting key differences in disease severity, clinical presentation, and perioperative resources, although such comparisons are inherently limited by single-case observations.^{1,5}

The rarity of WSS continues to pose diagnostic and management challenges, especially in low-income regions where neonatal care often focuses only on acute surgical emergencies. In our context, the diagnosis was achieved through careful clinical evaluation, imaging, and histopathology despite the lack of genetic testing facilities. This case emphasizes the importance of multidisciplinary collaboration—incorporating neonatology, pediatric surgery, pathology, and audiology—even in resource-limited hospitals.

Conclusion

Waardenburg–Shah syndrome is a rare but important consideration in neonates presenting with intestinal obstruction accompanied by pigmentary abnormalities and hearing deficits. This case demonstrates that, even in the absence of genetic testing, meticulous clinical evaluation supported by histopathological confirmation can reliably establish the diagnosis. Definitive surgical management, such as the modified extended Duhamel procedure, coupled with coordinated multidisciplinary follow-up, offers an effective treatment pathway in resource-limited settings. Early recognition and timely intervention remain essential to reducing morbidity and improving survival. Strengthening regional collaboration and leveraging telemedicine for earlier diagnosis and specialist input may further enhance outcomes for similar rare congenital syndromes in low-resource environments.

Research Involving Human Participants and/or Animal

This study involved human participants. Ethical approval was waived by the Institutional Review Board of Mogadishu Somali–Türkiye Recep Tayyip Erdoğan Training and Research Hospital, as per institutional policy for single case reports. All procedures were conducted in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

Data Sharing Statement

The data supporting the findings of this study are available from the corresponding author upon reasonable request.

Informed Consent

Written informed consent was obtained from the patient’s legal guardians for participation in the study and for the publication of anonymized data and clinical images.

Acknowledgment

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Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

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Disclosure

The authors declare that they have no conflicts of interest in this work.

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