CASE REPORT

Waardenburg-Shah Syndrome: a rare case in an Indian child

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SUMMARY

A 7-year-old male child presented with a history of discolouration of right eye since birth. On examination visual acuity was 6/6 on Snellen's chart in both eyes; anterior segment was within normal limits except for the brilliant blue discolouration of the inferior quadrant and superior quadrant of right iris and left eye iris, respectively. Both eyes had a clear lens and fundus findings were within normal limits. A detailed history from parents revealed that the child had difficulty in hearing and slurring of speech. In addition, the child had repeated episodes of constipation with bilious vomiting during infancy for which a diagnosis of fungal sepsis with Hirschsprung's disease was made and the child had to undergo a mid-sigmoid loop colostomy for that. A diagnosis of Waardenburg-Shah Syndrome was made and the child was referred for hearing and speech rehabilitation.

BACKGROUND

Waardenburg-Shah Syndrome (WSS) is a rare disease with an incidence of around 1 in 50 000 cases.¹⁻³ This has been rarely reported by Indian patients.¹ Early diagnosis of these cases plays a key role by allowing timely rehabilitation of these children. The first presenting symptom can often be the bluish discolouration of the eyes and a meticulous eye evaluation can help in early diagnosis of these cases.

CASE PRESENTATION

A 7-year-old male child presented with a history of discolouration of right eye since birth. The child's parents also gave a history of hearing difficulty and slurring of speech during childhood. The child's prenatal, natal and postnatal course of development was uneventful. Medical records revealed that the child had repeated episodes of constipation and bilious vomiting since birth. The child was diagnosed to have fungal sepsis with Hirschsprung's disease and he underwent mid-sigmoid loop colostomy at the age of 7 months. No other member of the family had similar symptoms. Ocular evaluation revealed a visual acuity of 6/6 on Snellen's chart in both the eyes. There was brilliant blue discolouration of the inferior quadrant of iris in right eye and superior quadrant of iris in the left eye (figures 1 and 2). The anterior segment examination was within normal limits in both the eyes. There were no lenticular changes in either of the eyes. Fundus examination was also normal. The right side of forehead had few hypopigmented patches (figure 3). Audiometry of the child revealed sensorineural deafness.

So our patient had sensorineural deafness, pigmentary abnormalities of the iris, hypopigmented patches on forehead and Hirschsprung's disease. Fulfilment of two major and one minor criteria led us to make a clinical diagnosis of WSS (table 1).⁴

TREATMENT

The ophthalmology department carried out no active intervention. The parents were counselled

Figure 1 Clinical photograph showing brilliant blue discolouration of inferior quadrant of iris in the right eye and superior quadrant of iris in the left eye.

Figure 2 Clinical photograph showing brilliant blue discolouration of the superior quadrant of iris in the left eye.

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Figure 3 Clinical photograph showing hypopigmented patches on the right side of forehead.

Table 1 Diagnostic criteria* for Waardenburg-Shah syndrome^{1–3}

Major criteria	Minor criteria
1. Sensorineural hearing loss	1. Skin hypopigmentation (congenital leucoderma/white patches on body)
2. Iris pigmentary abnormality (complete heterochromia iridium, segmental heterochromia, or complete hypoplastic blue iridis (brilliant blue iridis))	2. Synophrys: medial eyebrow flare
3. Hair hypopigmentation (white forelock or white hairs at other parts of body)	3. Broad nasal root
4. Dytrophia canthorum (lateral displacement of inner canthi)	4. Hypoplasia of nasal alea
5. First-degree relative previously diagnosed with Waardenburg syndrome	5. Premature hair greying (age <30 years)

*Criteria proposed by the Waardenburg Consortium. An affected individual must have 2 major criteria or 1 major plus 2 minor criteria.

about the nature of the disease. The child was referred to the concerned specialists for further management and rehabilitation services.

DISCUSSION

Dutch ophthalmologist Petrus Johannes Waardenburg first described Waardenburg syndrome in 1951.⁴ It is a hereditary condition with four different phenotypes labelled WS1, WS2, WS3 and WS4.³ WS1 has features of telecanthus, pigmentary abnormalities of hair, skin and eyes and congenital deafness. WS2 presents with features similar to WS1 but without telecanthus. WS3, also known as the Klein-Waardenburg syndrome, presents with severe limb abnormalities and contracture. WS4; also known as WSS is an extremely rare condition. In addition to the features of WS1 it has an additional feature of Hirschsprung's disease.³ The syndrome has been postulated to be due to an abnormality of neural crest derived melanocytes.

A patient of WSS with short segments of intestinal involvement presents at a later age with features of chronic

constipation, malabsorption and enterocolitis, compared to those with long segments of intestinal involvement who present with symptoms of intestinal obstruction; such as bilious vomiting, abdominal distension and inability to feed orally starting from the first few days of life.^{2 5} The diagnosis of Hirschsprung's disease is usually made based on history, physical examination, plain abdominal X-ray, barium enema, anorectal manometry and rectal biopsy. Past medical records of our case did reveal a diagnosis of Hirschsprung's disease, but we could not find any mention of WSS. Until now only few cases of WSS has been reported from India and it is not unusual for a physician to miss the diagnosis.^{2 6 7} Wadhvani *et al* reported a girl child from India being diagnosed as WSS at the age of 16 years. The presentation was similar to our case except for an additional feature of hypopigmented fundus that was not present in our case.²

Treatment of WSS depends on the presenting feature and associated abnormalities. Visual acuity is usually not affected. Early diagnosis is important in order to address the hearing and speech abnormalities so as to allow overall normal development of the child. In addition, genetic counselling can help to assess the risk of having the disease in subsequent siblings. An alert ophthalmologist can contribute by diagnosing the disease at an early stage.

Learning points

- Waardenburg-Shah syndrome is an extremely rare condition.
- Early diagnosis is vital to ensure proper growth and development of the child.
- Discolouration of iris is often the presenting symptom.
- Ophthalmologists must be aware of this rare syndrome.

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Contributors RP had a role in clinical examination of the case, collection of the data/consent and preparation of the first draft of the manuscript. SS had a role in preparing the manuscript and subsequent revisions of the manuscript. PKM had a role in planning, final editing of the manuscript and the figures and is responsible for the overall content as guarantor.

Competing interests None declared.

Patient consent Obtained.

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