# *Letters to the Editor*

## Shah-Waardenburg Syndrome

A 3-day-old full term male neonate was admitted with history of bilious vomiting since birth. On examination, he had a prominent white forelock of hair and massively distended abdomen. Audiologic evaluation (BERA) revealed complete bilateral sensorineural hearing loss. A clinical diagnosis of Shah-Waardenburg syndrome or Waardenburg syndrome type 4 was made. The abdominal roentgenogram revealed dilated bowel loops but no air-fluid levels. Barium enema showed a featureless normal caliber colon with no obvious transitional zone; the small bowel loops were distended.

On failure of conservative management, an exploratory laparotomy was undertaken on $D_{18}$ of life that revealed distended proximal jejunal and ileal loops; the 15 cm of terminal ileum and the colon were contracted (*Fig.1*).

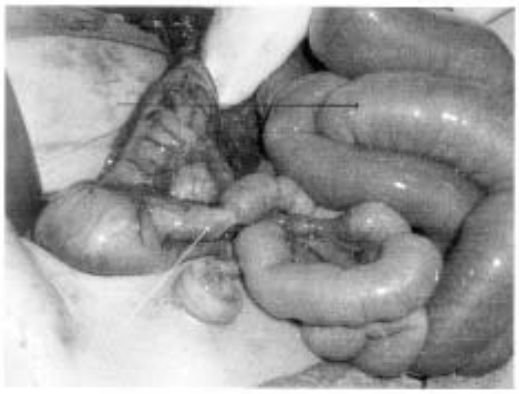

*Fig. 1. Intra-operatively, the colon and distal ileum were found contracted (white arrow); the ileum proximal to the transition zone was distended (black arrow).*

Multiple sero-muscular biopsies were taken from colon and terminal ileum; appendectomy was also performed. A divided ileostomy was performed at the transition zone. The histopathology of gut biopsies confirmed aganglionosis in colon and terminal ileum. Child underwent modified Kimura's procedure at 10 weeks of age but later succumbed to sepsis at 3 months of age.

Waardenburg syndrome (WS) was first described in 1951 (now called WS type 1) with 6 components lateral displacement of the medical canthi combined with dystopia of the lacrimal puncta and blepharophimosis, prominent broad nasal root, hypertrichosis of the medial part of the eyebrows, white forelock, heterochromia iridis and deafmutism(1). WS4 is the association of Waardenburg syndrome with Hirschsprung disease. Only 48 cases are reported in English literature till 2002(2).

**Y.K. Sarin ,**
**V. Manchanda,**
*Department of Pediatric Surgery,*
*Maulana Azad Medical College,*
*New Delhi 110 002, India.*
*E-mail: yksarin@hotmail.com*

**REFERENCES**

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2. Toki F, Suzuki N, Inoue K, Suzuki M, Hirakata K, Nagai K, *et al*. Intestinal aganglionosis associated with the Waardenburg syndrome: report of two cases and review of the literature. Pediatr Surg Int. 2003; 19: 725-728.