A four-day-old neonate presented with severe abdominal distention, intermittent bilious vomiting, typical black lock, partial albinism and heterochromatic iris. Ileostomy was done proximal to the transition zone at mid-ileal region (50 cms proximal to ileo-cecal region) All biopsy specimens till the transitional zone were negative for ganglion cells. Distinct features of total colonic aganglionosis and rectosigmoid Hirschsprung's disease are delineated here.

**Key words:** Hirschsprung's disease, Shah-Waardenberg syndrome, total colonic aganglionosis

### Introduction

Total colonic aganglionosis (TCA) is rare in neonates. Patients presenting with bowel obstruction, albinism, white forelock of hair and deafness should alert clinician of possibility of Shah-Waardenberg syndrome or its variation. ABCD syndrome, an expression of Shah-Waardenberg syndrome is presented here. ABCD syndrome includes albinism, black lock, cell migration disorder of the neurocytes of the gut and deafness. It is due to homozygous mutation in exon 3 (R201X) of the EDNRB gene.

### Case Report

A four-day-old neonate first born of non-consanguineous parents, normally delivered was transferred to our institution for severe abdominal distention and intermittent bilious vomiting. He had passed meconium after 48 hours of life.

On examination, he was dehydrated and had tachycardia. He had typical black lock and partial albinism [Figure 1]. The baby had heterochromatic iris and did not appear to respond to sound. Abdomen was distended but soft with visible bowel loops. Suspicion of Waardenberg syndrome (WS) associated with HD was raised. Erect X-ray abdomen revealed multiple air-fluid levels.

Post resuscitation laparotomy was done. Terminal 50 cm of ileum and colon were collapsed. Proximal dilatation was significant. An ileostomy was done proximal to the transition zone at mid-ileal region.

All biopsy specimens from colon, ileum and appendix till the transitional zone were evaluated. Whole-mount preparations showed lack of Interstitial Cells of Cajal- MY and ganglion cells [Figure 2] and a markedly reduced number of NADPH-positive nerve trunks as in TCA. The baby succumbed after 1½ months of fight for survival.

Genetic study in this patient could not be done.

### Discussion

Shah-Waardenberg syndrome includes sensorineural deafness, hypopigmentation of skin, hair and irides and HD due to homozygous mutation in EDNRB gene or
EDNB gene or hetero-zygous mutation in gene SOX10.[1]

ABCD syndrome has the following:
A- Albinism
B- Black lock
C- Cell migration disorder of the neurocytes of gut
D- Deafness

It occurs due to homozygous nonsense mutation in exon 3 (R201X) of the EDNRB gene. It is an autosomal recessive neural crest syndrome.[2,3]

HD is known do be associated with genetic disorder like Waardenberg syndrome. Further, Waardenberg syndrome has got various subtypes where Shah-Waardenberg syndrome is the fourth subtype.[4,5] Thus ABCD syndrome is variant expression of Shah-Waardenberg syndrome. We have come across only two-three reported cases of ABCD syndrome.

Whole-mount preparation shows lack of Interstitial Cells of Cajals and a markedly reduced number of NADPH-positive nerve trunks. We found the same findings in our case.

Immunohistochemistry using peripherin, neuronal nitric oxide synthase (nNOS) and c-kit antibody is often done in TCA and recto-sigmoid HD. In TCA it shows lack of or weak nNOS in short nerve trunks of myenteric, submucous plexus and muscle layer whereas weakly positive nNOS trunks are seen in the recto-sigmoid HD bowel. Peripherin immunoreactivity (IR) is also reduced in TCA as compared to recto-sigmoid HD bowel.[6]

References


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