PAEDIATRIC DERMATOLOGY

WAARDENBURG SYNDROME TYPE – IV VARIANT WITH AN UNUSUAL PRESENTATION OF COMPLETE AGANGLIONOSIS OF LARGE INTESTINE

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INTRODUCTION : Waardenburg syndrome is an inherited disorder often characterized by varying degree of hearing loss, changes in skin and hair pigmentation. There are four clinical variants depending on its associated conditions. Type IV variant also called Waardenburg Shah syndrome with Hirschsprung disease is a rare variant with predominant autosomal recessive inheritance. Our patient had an exceptional rare association of extended long segment of aganglionosis of entire large bowel with associated cutaneous manifestation.

CASE REPORT : A full term male baby weighing 3.5 kg was admitted in NICU for bowel obstruction. On the 10th day of life, upon detailed examination, there was presence of white forelock over the frontal area with fine vellus hair, white in color over the cheeks and two well defined hypopigmented patchs of 3 x 4 cm were seen on the right elbow and right knee. Bilateral low set ears was present. Barium enema showed gross dilatation of stomach with small bowel filled with gas (microcolon). Other hematological parameters were within normal limits. On failure of conservative management, an exploratory laparotomy was undertaken which revealed contracted jejunum and large bowel upto rectum. Multiple sero muscular biopsies were taken from colon and terminal ileum. The histopathology of gut biopsies confirmed aganglionosis in colon and jejunum with compensatory hypertrophic myentric nerve plexus. But within 2 days post laprotomy baby succumbed to death before further gene evaluation for specific mutation was done.

CONCLUSION : Our case has been reported for its rarity of complete absence of gangliosides from jejunum to entire large intestine upto rectum along with classical cutaneous features of Waardenburg syndrome. Prognosis of such babies of this variant is associated with high morbidity and mortality.