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A CASE OF WAARDENBURG SYNDROME AND AGANGLIONOSIS*

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Waardenburg's syndrome is characterized by a broad nasal root, pigmentation disturbance and congenital deafness while aganglionosis is described as the partial or complete lack of ganglion cells in the alimentary tract. This report describes a five-day-old male infant with Waardenburg's syndrome associated with total aganglionosis of the colon, ileum and distal jejunum and draws attention to the causal relationship between these two entities. *Key words: Waardenburg's syndrome, Hirschsprung's disease, neural crest.*

The principal characteristics of Waardenburg's syndrome are facial deformities exhibited by a broad nasal root, pigmentary disturbance, and congenital deafness. It is inherited as an autosomal dominant condition with variable penetrance. Aganglionosis is a congenital anomaly characterized by partial to complete colonic obstruction associated with the absence of intramural ganglion cells in the alimentary tract[1, 2]. This report presents both anomalies in one patient and stresses the etiological relationship.

Case Report

A five-day-old male infant was referred to the Pediatric Surgery Department of Dicle University Hospital for evaluation of intestinal obstruction. The baby was the first - born child of a first cousin marriage with the family history being otherwise unremarkable. The mother was 21 years old and the father 23. The patient's weight was 2800 g and length 49 cm. Physical examination revealed an extensive white forelock, broad nasal root, megalocornea, hypopigmentation of the irides and retina, and parietal deviation of the lacrimal puncta. He was totally unresponsive to noise. A cleft palate, systolic murmur along the mesocardiac region and bilateral hydrocele were striking features. Plain X - ray showed gas - fluid levels and an unused colon was found with contrast enema (Figs. 1 and 2). After correction of fluid and electrolyte imbalances and other preoperative

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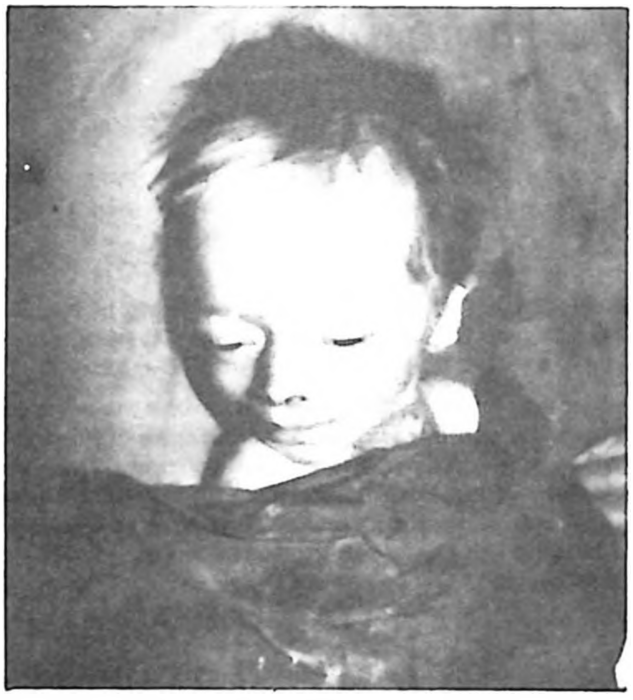

Fig. 1: Extensive white forelock and broad nasal root indicative of Waardenburg's syndrome.

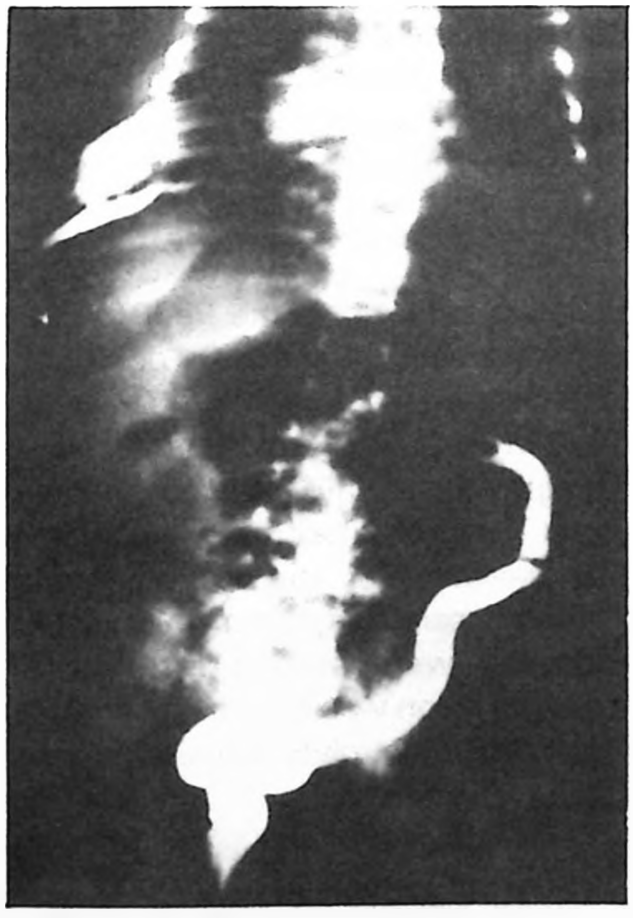

Fig. 2: Unused colon of patient with aganglionosis.

assessments, an exploratory laparotomy was performed. The whole colon and 25 cm of the terminal ileum were very narrow and believed to be aganglionic. An ileostomy was performed. Three biopsies were taken from above, below and at the site of the ileostomy. The ileostomy was not effective during the postoperative period. Parenteral nutrition was initiated. The infant died of sepsis on the 14th postoperative day. A postmortem examination revealed total aganglionosis extending to the upper jejunum. Hepatomegaly, edema, hyperemia of the other intraabdominal viscus and reactionary fluid were striking features of the intraabdominal examination. The pancreas and thyroid tissues were found to be normal. The retroperitoneal area was scrutinized for the sympathetic ganglion chain and surrenal glands. The surrenal glands were present at their normal location; both had normal findings. Intrathoracic examination revealed patent ductus arteriosus.

Discussion

As the neural folds fuse to form the neural tube, some neuroectodermal cells lying along the crest of each neural fold lose their epithelial affinities and attachments to neighboring cells. These are termed neural crest cells and are differentiated as melanocytes, Schwann cells, dorsal root ganglion cells (cochlea), sympathetic ganglion cells, chromaffin cells of the surrenal medulla, celiac ganglion cells and plexuses of the intestinal tractus[3]. If there are differentiation or migration problems of the neural crest cells, some of the pathological features of Waardenburg's syndrome and Hirschprung's disease can be seen together. Although these two conditions seldom occur together (2: 100,000 and 2: 10,000, respectively), numerous cases have been reported of Waardenburg's syndrome associated with Hirschprung's disease, indicating a possible etiological relationship. Moreover, after a review of the literature, we found that the length of the aganglionic intestinal segment is directly correlated with the variety of symptoms relevant to anomalies of neural crest origin[4-6]. This was also the case in our patient who had multiple anomalies and an aganglionic segment consisting of the entire colon, ileum and distal jejunum.

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