PEDIATRIC SURGERY *Update* 8
Vol. 33 No. 03 SEPTEMBER 2009

Waardenburg Syndrome

Waardenburg syndrome (WS) is an autosomical recessive or dominant trait condition derived from the neural crests and includes sensorineural hearing loss (bilateral or unilateral), and pigmentation disorder of the hair, eyes and skin (iris heterochromia, white forelock, lateral displacement of inner canthi of the eyes). Four types of the syndrome have been described. Though very rare, WS can be associated with Hircshsprung=s disease (Shaw-Waardenburg syndrome). Type IV WS is a term used to denote intestinal aganglionosis associated with the Waardenburg complex, consisting of hypopigmented spots on the skin, heterochromia irides, depigmented ocular fundus, telecanthus, and sensory deafness. The aganglionosis associated with WS is usually total colonic with extensive involvement of ileus. Clinically these affected babies present with intestinal obstruction findings such as bilious vomiting, abdominal distension, and inability to feed orally from the first few days of life. Mutations in the endothelin-3 endothelin B receptor, and SOX10 genes have been identified as causative genes of WS. The length of the aganglionic segment decides the prognosis of these children. The longer the affected bowel the worse the prognosis even in the face of restorative surgery (long bowel myectomy-myotomy).

References:

1-Tomiyama H, Shimotake T, Ono S, Kimura O, Tokiwa K, Iwai N: Relationship between the type of RET/GDNF/NTN or SOX10 gene mutations and long-term results after surgery for total colonic aganglionosis with small bowel involvement. J Pediatr Surg. 36(11):1685-8, 2001
2- Gnananayagam EJ, Solomon R, Chandran A, Anbarasi S, Sen S, Moses PD:Long segment Hirschsprung's disease in the Waardenburg-Shah syndrome.Pediatr Surg Int. 2003 Aug;19(6):501-3, 2003
3- Moore SW: The contribution of associated congenital anomalies in understanding Hirschsprung's disease. Pediatr Surg Int. 22(4):305-15, 2006
4- Jan IA, Stroedter L, Haq AU, Din ZU: Association of Shah-Waardenburgh syndrome: a review of 6 cases. J Pediatr Surg. 43(4):744-7, 2008
5- Espinosa R, Alonso Calderon JL: Neural crest disorders and Hirschsprung's disease. Cir Pediatr. 22(1):25-8, 2009
5- Karaca I, Turk E, Ortac R, Kandirici A: Waardenburg syndrome with extended aganglionosis: report of 3 new cases. J Pediatr Surg. 44(6):E9-13, 2009

Bishop-Koop Anastomosis

The survival of babies born with cystic fibrosis and meconium ileus was improved significantly with the development of intestinal surgical procedures in the second part of the past century. Such is the case of the temporary Bishop-Koop anastomosis (BKA), where the proximally dilated bowel is anastomosed end to side to the distal bowel which in turns is taken out as a stoma. Succus entericus can then pass from the proximal bowel to the

distal bowel all the way down to the anus or come out through the stoma if there is a distal functional obstruction. Beside meconium ileus, this anastomosis has been used in children suffering from various other intestinal anomalies such as jejuno-ileal atresia, necrotizing enterocolitis, volvulus, gastroschisis associated with bowel atresia, and apple-peel syndrome. In such situations the Bishop-Koop anastomosis is a safer procedure than primary end-to-end or end-to-side anastomosis, especially for the management of greatly different intestinal diameters. This occurs in cases of slowly distensible microcolons associated with the above conditions where the BKA is a temporary procedure suitable for the colon to grow properly.

References:

1- Kootstra G, Kamann HL, Okken A, Vander Vliet JA, Zwierstra RP, Slooff MJ, Krom RA, Kuijjer PJ: The Bishop-Koop anastomosis-a find in pediatric surgery. Neth J Surg. 32(3):92-6, 1980
2- Murshed R, Spitz L, Kiely E, Drake D: Meconium ileus: a ten-year review of thirty-six patients. Eur J Pediatr Surg. 7(5):275-7, 1997
3- Wit J, Sellin S, Degenhardt P, Scholz M, Mau H: Is the Bishop-Koop anastomosis in treatment of neonatal ileus still current?. Chirurg. 71(3):307-10, 2000
4- Fleet MS, de la Hunt MN: Intestinal atresia with gastroschisis: a selective approach to management. J Pediatr Surg. 35(9):1323-5, 2000
5- Kumaran N, Shankar KR, Lloyd DA, Losty PD: Trends in the management and outcome of jejuno-ileal atresia. Eur J Pediatr Surg. 12(3):163-7, 2002
6- Siman J, Trnka J: Stoma procedures in the congenital digestive tract malformations. Rozhl Chir. 86(7):347-52, 2007

Fetal Rectal Perforation

Perforation of the extraperitoneal rectal wall in newborns is very rare. Most described cases are term babies. Fetal distress is not noted at the time of delivery. All affected babies have a recognizable abnormality at birth mostly seen during the first five days of birth. The abnormality can be a perineal or buttock lesion, varying from a minor skin blemish, fistula, swelling to a rapidly enlarging aerocele with meconium staining of perineal tissue with subsequent skin rupture or isolated ascites. Diagnosis is suggested with lateral simple films. Meconium is expressed under pressure by fetal peristalsis through a defect in the lower rectal wall into normal tissue planes within the infralevator space to reach a subcutaneous level and up toward the abdominal cavity. No other congenital associated anomaly has been described. The position of the defect at the level of the pelvic floor is consistent with the watershed between the inferior and middle rectal arteries and might suggest a local ischemic insult, though the cause remains unknown. After resuscitation and antibiotherapy, surgery is mandatory. The procedure consists of fecal diversion achieved by sigmoid colostomy; in addition the perineal tissues are debrided and wide drainage is achieved. Prognosis is usually good with normal continence obtained later in most affected cases. Some cases has developed a rectal stenosis amenable to dilatation.

References:

1- Mitsudo SM, Boley SJ, Rosenzweig MJ, Campbell DE: Extraperitoneal pelvic meconium extravasation in a newborn infant. J Pediatr. 103(4):598-600, 1983
2- Davies MR, Cywes S, Rode H: Prenatal perforation of the extraperitoneal part of the rectum, associated with a developmental defect of the pelvic floor. Z Kinderchir. 39(4):271-3, 1984

3- Casaccia G, Giorlandino C, Catalano OA, Bagolan P: Prenatal rectal perforation: an unsuspected cause of isolated ascites. J Perinatol. 26(11):717-9, 2006
4- Sundararajan L, Patel D, Jawaheer G: Antenatal rectal perforation presenting in the neonate. Pediatr Surg Int. 24(5):601-3, 2008
5- Pitcher GJ, Davies MR, Bowley DM, Numanoglu A, Rode H: Fetal extraperitoneal rectal perforation: a rare neonatal emergency. J Pediatr Surg. 2009 Jul;44(7):1405-9

---

* Edited by: **Humberto Lugo-Vicente, MD, FACS, FAAP**
Professor /Academic Director of Pediatric Surgery, University of Puerto Rico - School of Medicine, Rio Piedras, Puerto Rico.
Address: P.O. Box 10426, Caparra Heights Station, San Juan, Puerto Rico USA 00922-0426.
Tel (787)-786-3495 Fax (787)-720-6103 E-mail: *titolugo@coqui.net*
Internet: http://home.coqui.net/titolugo

8 PSU 1993-2009
ISSN 1089-7739