PEDIATRIC SURGERY *Update* ©
Vol. 51 No. 06 DECEMBER 2018

Cecal Diverticulum

Diverticulum in the cecum are very rare in children and adults. It is estimated that for every 300 appendectomies the surgeon will encounter one cecal diverticulum. Most cecal diverticulum are congenital in origin containing all four layers of the bowel (true diverticulum), instead of the more common false diverticulum that does not contain the muscular layer in the left colon seen mostly in adults patients. Cecal diverticulum results from pulsion of the fetal cecum in the 6th week of pregnancy. Cecal diverticulum can cause inflammation, hemorrhage or perforation. The incidence of right colon diverticulum among all patients with diverticulosis is between 60-80% in the Asian population. Since the child develops right lower quadrant pain the initial diagnosis is appendicitis. Fecal material can accumulate in the most distal part of the diverticulum leading to ruptured. The diverticulum is solitary and tends to produce problems in younger patients. Most are situated posteriorly and near to the ileocecal valve. In the less common anterior diverticulum there might be more rapid progression to perforation and peritonitis. Finding an unexpected inflammatory mass in the cecum during an emergency laparoscopy or laparotomy for appendicitis might end in a right hemicolectomy if the diagnosis of a benign solitary diverticulum is not entertained. Symptoms mimic those of acute appendicitis (right iliac fossa pain, fever and leukocytosis) depending on the grade of inflammation and walling off from the omentum. Diagnosis of a cecal diverticulum can be made or suggested with CT-Scan. Colonoscopy is a valuable diagnostic tool in the diagnosis of diverticular disease. Preoperative diagnosis is difficult in emergency cases. Management of cecal diverticulitis is segmental resection of the diverticulum base (diverticulectomy) along with concomitant appendectomy. In few occasions right hemicolectomy might be required in cases with perforation risk or severe surrounding tissue inflammation.

References:

1- Kalcan S, Basak F, Hasbahceci M, et al: Intraoperative diagnosis of cecal diverticulitis during surgery for acute appendcitis: Case series. Ulus Cerrahi Derg 32: 54-57, 2016
2- Chen CJ, Chuang JP: Conservative Surgery for Right Colon Perforation Leads to Better Long-Term Outcomes in Children: A 21-year Experience. Pediatr Neonatol. 56(3):159-64, 2015
3- Martens T, Fierens K: Giant cecal diverticulum in a child. J Pediatr Surg. 46(6):e23-5, 2011
4- Kamocki Z, Jaroszuk J, Zaraba K, Kadra B: Acute inflammation of the true cecal diverticulum--case report. Pol Przegl Chir. 83(8):461-4, 2011
5- Rubio PA: Laparoscopic resection of a solitary cecal diverticulum. J Laparoendosc Surg. 4(4):281-5, 1994

Mayer-Rokitansky-Kuster-Hauser Syndrome

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome refers to a normal female karyotype 46XX with congenital aplasia or severe hypoplasia of the structures that derived from the müllerian ducts including the upper two-third of the vagina, uterus and fallopian tubes. Occurs in one of 4500 female births. The syndrome becomes type II when other associated malformations are found such as renal unilateral agenesis, renal ectopia, horseshoe kidney, skeletal-vertebral anomalies, scoliosis, auditory system anomalies, heart defects and syndactylism or polydactylism. Associated malformations are present in almost half of patients with unilateral renal agenesis the most common. The defect occurs during organogenesis 4th to 8th week gestation transmitted as a dominant autosomal with incomplete penetration and variable expression. The patient presents with primary amenorrhea as initial symptom with normal secondary sexual characteristics since the ovaries are normal. Pelvic ultrasound is the first diagnostic study to perform. This is followed by MRI which is usually diagnostic of the anomaly. This is followed with X chromatin, karyotype and female hormones plasma levels. In case of doubt diagnostic laparoscopy can precise anatomy. Management should start in late adolescent. The objective of treatment is the creation of a functional neovagina satisfactory for intercourse. Progressive perineal dilatation is the preferred initial approach due to the minimally invasive nature, high success rate and low cost. If this method fails then surgical vaginoplasty using inverted skin flap (McIndoe method) or U-flap (William method) is indicated. Some authors use a piece of sigmoid colon as neovagina. This is a major procedure with potential complications such as excess mucous production, vaginal stenosis, prolapse, diversion colitis, bowel obstruction and even neovaginal carcinoma. To have children these patients will need in vitro fertilization with a gestational carrier.

References:

1- Pizzo A, Lagana AS, Sturlese E, Retto G, Retto A, De Dominici R, Puzzolo D: Mayer-rokitansky-kuster-hauser syndrome: embryology, genetics and clinical and surgical treatment. ISRN Obstet Gynecol. 2013;2013:628717. doi: 10.1155/2013/628717. Epub 2013

2- Londra L, Chuong FS, Kolp L: Mayer-Rokitansky-Kuster-Hauser syndrome: a review. Int J Womens Health. 7:865-70, 2015

3- Saleem M, Iqbal MZ, Jam MR, Ahmad M, Mirza B: Colovaginoplasty in a case of mayer-rokitansky-kuster-hauser syndrome. APSP J Case Rep. 5(1):7, 2014

4- Thomas E, Shetty S, Kapoor N, Paul TV: Mayer-Rokitansky-Kuster-Hauser syndrome. BMJ Case Rep. 2015 May 15;2015. pii: bcr2015210187. doi: 10.1136/bcr-2015-210187.

5- Bombard DS 2nd, Mousa SA: Mayer-Rokitansky-Kuster-Hauser syndrome: complications, diagnosis and possible treatment options: a review. Gynecol Endocrinol. 30(9):618-23, 2014

6- Hall-Craggs MA, Williams CE, Pattison SH, Kirkham AP, Creighton SM: Mayer-Rokitansky-Kuster-Hauser syndrome: diagnosis with MR imaging. Radiology. 269(3):787-92, 2013

DICER1 Syndrome

DICER1 syndrome also known as pleuropulmonary blastoma familial tumor susceptibility syndrome is a rare genetic disorder that predisposes patents to development of benign and malignant tumors. They include cystic nephroma, embryonal rhabdomyosarcoma, multinodular goiter, thyroid cancer, ovarian Sertoli-Leydig cell tumor, pituitary blastoma and others. The DICER1 gene is located in

chromosome 14, position q32.13 and encodes an endoribonuclease Dicer protein playing a role in protein translational control. The mutation causes loss of function of cancer suppressors genes or gain of function in genes that contribute to the onset of cancer in an active manner (oncogenes). Pleuropulmonary blastoma, a rare cancer of the lung or pleura in children, is a manifestation of DICER1 syndrome. The simultaneous occurrence of Sertoli-Leydig ovarian cell tumor and thyroid carcinoma is a reliable indicator of DICER1 syndrome. A rare form of Hodgkin lymphoma arising from T cells, instead of mature B cells, has been associated with DICER1 syndrome. Pineoblastoma may be associated with a DICER1 mutation. Other rare associations with DICER1 syndrome include developmental delay, lung cyst, overgrowth, macrocephaly and Wilms tumor. DICER1 syndrome has been recognized as an autosomal dominant disease, inherited and expressed in a haploinsufficient manner. The DICER1 gene encodes an enzyme that is involved in the biogenesis of microRNAs. DICER1 Germline mutations are identified as nonsense mutations leading to stop codons within the coding sequence leading to cancerous and noncancerous tumors. The spectrum of DICER1-related tumors and the young age at presentation suggest early surveillance of at-risk patients is critical.

References:

1- Robertson JC, Jorcyk CL, Oxford JT: DICER1 Syndrome: DICER1 Mutations in Rare Cancers. Cancers (Basel). 2018 May 15;10(5). pii: E143. doi: 10.3390/cancers10050143.

2- Bueno MT, Martinez-Rios C, la Puente Gregorio A, et al: Pediatric imaging in DICER1 syndrome. Pediatr Radiol. 47(10):1292-1301, 2017

3- Rutter MM, Jha P, Schultz KA, et al: DICER1 Mutations and Differentiated Thyroid Carcinoma: Evidence of a Direct Association. J Clin Endocrinol Metab. 101(1):1-5, 2016

4- Schultz KA, Yang J, Doros L, et al: DICER1-pleuropulmonary blastoma familial tumor predisposition syndrome: a unique constellation of neoplastic conditions. Pathol Case Rev. 19(2):90-100, 2014

5- Doros L, Yang J, Dehner L, et al: DICER1 mutations in embryonal rhabdomyosarcomas from children with and without familial PPB-tumor predisposition syndrome. Pediatr Blood Cancer. 59(3):558-60, 2012

6- Slade I, Bacchelli C, Davies H, et al: DICER1 syndrome: clarifying the diagnosis, clinical features and management implications of a pleiotropic tumour predisposition syndrome. J Med Genet. 48(4):273-8, 2011

***Edited by: Humberto Lugo-Vicente, MD, FACS, FAAP**

Professor of Pediatric Surgery, University of Puerto Rico - School of Medicine, Rio Piedras, Puerto Rico. Director - Pediatric Surgery, San Jorge Childrens Hospital.

Address: P.O. Box 10426, Caparra Heights Station, San Juan, Puerto Rico USA 00922-0426.

Tel (787)-999-9450 E-mail: *titolugo@coqui.net*

Internet: http://home.coqui.net/titolugo

ISSN 1089-7739