



PEDIATRIC SURGERY Update © **Vol 10 No 02 FEBRUARY 1998**

'Official Publication of the Puerto Rico Association of Pediatric Surgeons'

Thyroid Nodules:FNAB

The need to differentiate malignant from benign thyroid nodules is the most challenging predicament in management. Present diagnostic work-up consists of ultrasonography (US), radionuclear scans (RNS) and fine-needle aspiration biopsy (FNAB). After reviewing our ten-year experience with twenty-four pediatric thyroid nodules we found nineteen benign and five malignant lesions. Benign nodules were soft, movable, solitary and non-tender. Malignant nodules were found during late adolescence, characterized by localized tenderness, a multiglandular appearance and fixation to adjacent tissues. US and RNS were of limited utility since malignancy was identified among cystic and hot nodules respectively. Suppressive thyroid hormone therapy was useless in the few cases tried. FNAB in eighteen cases did not limit the number of thyroid resections. It showed that the probability that a malignant nodule had suspicious or frankly malignant cytology was 60%. The specificity was 90%. This is the result of a higher number of patients with follicular cell cytology in the aspirate. No attempts should be made to differentiate follicular adenoma from carcinoma since capsular and vascular invasion cannot be adequately assessed by FNAB. The physical exam findings, persistence of the nodule, progressive growth and cosmetic appearance were the main indications for surgery. FNAB is a safe procedure that plays a minor role in the decision to withhold surgery. Its greatest strength is to anticipate in case of malignancy that a more radical procedure is probably needed. FNAB, US and RNS should not replace clinical judgement or suspicion as the most important determinants in management.

References

- 1- Lugo-Vicente HL: Pediatric Thyroid Nodules: Management in the era of FNA (submitted for publication).
- 2- Lugo-Vicente HL: Pediatric Thyroid Nodules: Insights in Management (in press).

Li-Fraumeni Syndrome

The Li-Fraumeni familial cancer syndrome is manifested by increased susceptibility of affected relatives to develop a diverse set of malignancies during early childhood. The major features of the syndrome include breast cancer, sarcomas of soft tissue and bone, brain tumor, leukemia and adrenal cortical carcinoma. More than one-half of the cancers overall and nearly one-third of the breast cancers were diagnosed before 30 years of age. Among females, breast cancer is the most common. Germline mutations within a defined region of the p53 gene have been found in families with the Li-Fraumeni syndrome. Persistence of the mutation in the germline suggests a defect in DNA repair in the family member first affected. Asymptomatic carriers of p53 germline mutation needs closed evaluation and follow-up for early detection and treatment in case neoplasia develops.

References

- 1- Garber JE, Goldstein AM, Kantor AF, Dreyfus MG, Fraumeni JF Jr, Li FP: Follow-up study of twenty-four families with Li-Fraumeni syndrome. *Cancer Res* 51(22):6094-7, 1991
- 2- Santibanez-Koref MF, Birch JM, Hartley AL, Jones PH, Craft AW, Eden T, Crowther D, Kelsey AM, Harris M: p53 germline mutations in Li-Fraumeni syndrome. *Lancet* 338(8781):1490-1, 1991
- 3- Tricker KJ, Prosser J, Condie A, Kelsey AM, Harris M, Jones PH, Binchy A, Crowther D, et al: Prevalence and diversity of constitutional mutations in the p53 gene among 21 Li-Fraumeni families. *Cancer Res* 54(5):1298-304, 1994
- 4- Frebourg T, Barbier N, Yan YX, Garber JE, Dreyfus M, Fraumeni J Jr, Li FP, Friend SH: Germ-line p53 mutations in 15 families with Li-Fraumeni syndrome. *Am J Hum Genet* 56(3):608-15, 1995
- 5- Strauss EA, Hosler MR, Herzog P, Salhany K, Louie R, Felix CA :Complex replication error causes p53 mutation in a Li-Fraumeni family. *Cancer Res* 55(15):3237-41, 1995

Omphalo-Mesenteric Remnants

Before placental circulation is established fetal nourishment occurs from the yolk sac through the omphalomesenteric duct. By the 5th to 7th intrauterine week the duct obliterates. Persistence of the duct might give rise to a wide spectrum of omphalomesenteric remnants (OMR). Most OMR are in the form of a Meckel's diverticulum toward the intestinal end. Other less OMR are in the form of a mucosa-lined sinus or blind pouch at the umbilicus, an umbilical polyp, an intra- or extraperitoneal cyst, a connective tissue cord, or a well-formed communication between the ileum and the umbilicus. Pluripotential ectopic tissues (gastric, duodenal, colonic or pancreatic) might be found within OMR causing further problems (bleeding, perforation, obstruction, intussusception). Clinically the infant manifests periumbilical reddening, ulceration, granuloma, fluid discharge (bowel content), or recurrent umbilical infections. Diagnosis should come to your mind with recurrent umbilical discharge, non-healing granuloma, cherry-red nodule, or a rosette-like opening. Differential diagnosis consists of local infection (omphalitis), urachal remnants, dermoid cysts or vascular malformations. Management is umbilical exploration and surgical excision after suspicion or radiographic diagnosis (US, sinogram) is established.

References

- 1- Moore TC: Omphalomesenteric duct malformations. *Semin Pediatr Surg* 5(2):116-23, 1996
- 2- Mothes W: [Complications caused by remnants of the omphalomesenteric duct] *Zentralbl Chir* 115(22):1431-4, 1990
- 3- Jauniaux E, De Munter C, Vanesse M, Wilkin P, Hustin J: Embryonic remnants of the umbilical cord: morphologic and clinical aspects. *Hum Pathol* 20(5):458-62, 1989
- 4- Gaisie G, Curnes JT, Scatliff JH, Croom RD, Vanderzalm T: Neonatal intestinal obstruction from omphalomesenteric duct remnants. *AJR Am J Roentgenol* 144(1):109-12, 1985
- 5- Schärli AF: Vitello-intestinal disorders, In Neill V. *Freeman's Surgery of the Newborn*, Churchill Livingstone Ed, UK, 1994, pag. 243-255
- 6- Chapter 48: Disorders of the Umbilicus, In Marc I. Rowe's *Essential in Pediatric Surgery*, Mosby Publishers, USA, 1995, pag. 441-445

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

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

Tel (787)-786-3495 Fax (787)-720-6103

E-mail: titolugo@coqui.net Internet: <http://home.coqui.net/titolugo>

PSU University Edition: <http://www.upr.clu.edu/psu>