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Publisher
Pacini Editore S.r.l.
Via Gherardesca, 1
56121 Pisa, Italy
Tel. +39 050 313011
Fax +39 050 3130300
info@pacinieditore.it
www.pacinimedicina.it

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REVIEWS

Everything you always wanted to know about GIST (but were afraid to ask)

An update on GIST pathology

R. Ricci, L. Saragoni

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors of the gastrointestinal tract. The discovery of the occurrence of activating *KIT* mutations and KIT expression in GISTs opened the way to the unequivocal diagnosis of these tumors and to their successful treatment with imatinib, a tyrosin kinase inhibitor. Since then, research progress revealed molecular GIST triggers alternative to *KIT*, implying heterogeneous analytic approaches and prognostic expectations. Several targeted therapies, variably specific for each GIST trigger, have been developed or are being investigated. Thus, GISTs eventually revealed a family of diseases rather than a single tumor type. All these events had an unprecedented impact on pathology practice, constituting at the same time a heavy burden and an exciting challenge, ultimately putting pathologists in the spotlight as never before.
This review will discuss the most recent advances concerning GISTs, highlighting the tasks of pathologists facing these tumors, with an emphasis on traps potentially compromising a correct diagnosis.

Immunohistochemical evaluation of mismatch repair proteins in colorectal carcinoma: the AIFEG/GIPAD proposal

A. Remo, M. Fassan, G. Lanza on behalf of AIFEG and GIPAD

Microsatellite instability (MSI) is a hypermutable phenotype that usually arises from either a germline mutation in components of the mismatch repair (MMR) machinery (i.e. hMLH1, MSH2, MSH6 and PMS2) in patients with Lynch syndrome (LS) or somatic hypermethylation of the hMLH1 promoter in sporadic carcinomas. In all colorectal cancers (CRC) is possible to identify the MMR deficiency through protein expression by immunoistochemistry (IHC). Recently, the predictive role of MMR deficiency in reduced chemotherapy benefit and the introduction of universal screening for Lynch syndrome suggest to include MMR testing into routine clinical practice. In this scenario is mandatory to update the minimal requirements for MMR IHC standardization and evaluation. According to international guidelines, these are the GIPAD and AIFEG suggestions for MMR IHC testing.

Computed tomography - histology correlations of unusual lung tumors

N. Landini, G. Milanese, E. Zambrini, I. Ariozzi, L. Gnetti, A. Carloni, F. Nosenzo, M. Silva, N. Sverzellati

A large variety of rare benign and malignant tumors may sporadically affect the lung. Computed tomography (CT) findings of unusual primary lung tumors are often nonspecific. However, there are some rare pulmonary tumors with imaging features overlapping those of other conditions, thus making radiologic diagnosis challenging. The aim of this review was to correlate CT and histopathological features of a variety of unusual lung tumors to better clarify when and to what extent radiological diagnosis is reliable.

Kikuchi-Fujimoto disease: a clinicopathologic update

F. Pepe, S. Disma, C. Teodoro, P. Pepe, G. Magro

Kikuchi-Fujmoto disease (KFD), also known as "histiocytic necrotizing lymphadenitis", is a rare lymphadenitis of unknown origin, but with an excellent prognosis. It is more common in Asia, but isolated cases are also reported in America, Africa and Europe. The disease can have an acute or subacute course, usually develops in 1 to 3 weeks, with spontaneous resolution in 1-4 months. The main clinical sign is cervical lymphadenopathy, especially in the posterior cervical triangle with bulky and painful lymph nodes, usually affecting only one side; rare cases of generalized lymphadenopathy can be seen. This common clinical presentation can also be accompanied by nausea, vomiting, weight loss, weakness, headache and arthralgia. An extranodal extension of the disease, including involvement of skin, eye, and bone marrow localizations, has been rarely described. Most patients have leukopenia or neutropenia with a relative leukocytosis. At an ultrasound exploration of the affected lymph nodes, a hypoechoic aspect can be seen, with an external, thick and irregular hyperechoic ring. As there are no specific tests for KFD, the final diagnosis is histologically-based from lymph node excisional biopsy. Histological examination shows paracortical foci of coagulative necrosis containing karyorrhectic debris, which are surrounded by numerous CD68+/myeloperoxidase (MPO)+ histiocytes, CD68+/CD123+ plasmacytoid dendritic cells, and a minority of small- to large-sized CD8+lymphocytes and immunoblasts. Differential diagnosis mainly includes systemic lupus erithematous (SLE)-related lymphadenopathy and large cell lymphoma. The histological absence of neutrophils, plasmacells, as well as hematoxylin bodies, is a feature which argues against the diagnosis of SLE. In addition, the absence of auto-antibodies and anti-nuclear antibodies is useful in ruling out an autoimmune disorder. Early diagnosis of KFD is crucial to prevent the patients undergo extensive investigations related to suspected malignant lymphomas or other diseases.

ORIGINAL ARTICLES

The diagnostic value of cytohistological urine analysis and cytokeratin 20 in malignant and atypical urothelial cells

S. Negri, P. Biavati, A. Bondi

Introduction. To determine the ability of cytohistology and cytokeratin 20 (CK 20) expression in malignant and atypical cells (AUC) from urine to serve as a diagnostic tool for assessing urothelial carcinoma (UC).
Methods. Diagnoses from 55 urine cytological samples from 55 patients were analyzed and correlated with subsequent biopsy findings. A total of 50 archived urine slides from patients that received a cytological diagnosis and histological follow-up were selected for immunostaining with monoclonal CK 20 antibodies and elaborated by Z-test for proportions.
Results. The majority of all positive or atypical smears (24; 89%) were confirmed through histological analysis. The majority of urinary cytological diagnoses reported as negative (15; 54%) were also confirmed through biopsies. The overall sensitivity, specificity, PPV, and NPV were 65%, 83%, 89%, and 54%, respectively. All 13 smears cytologically determined to contain malignant cells, with subsequent biopsies confirming UC, exhibited strong positive staining with the CK 20 antibody. All cases evaluated as benign both cytologically and histologically had negative CK 20 staining. Of the 15 AUC cases with lesions confirmed through biopsies, 11 (73%) had atypical cells that stained positive for CK 20.
Discussion. Our results demonstrate the diagnostic value of urinary cytology and confirm CK 20 as an adjunct marker for the diagnosis of UC and for the triage of AUC.

Solid papillary carcinoma of the nipple: an in situ carcinoma or an expansive growth tumor?

D. Tacchini, L. Vassallo, M.A.G.M. Butorano, V. Mancini, T. Megha

Papillary breast lesions are a heterogeneous group of tumors which mainly arise in the central mammary region, ranging from benign to malignant. Among them, solid papillary carcinoma (SPC) represents a very uncommon variant with indolent clinical behavior and excellent prognosis. The categorization of papillary lesions as benign, atypical or malignant is often difficult even for experienced pathologists. Furthermore, for prognostic purposes, to decide whether to consider a lesions as in situ when it is not associated with frank invasive foci of carcinoma may be problematic.

We present a case of solid papillary carcinoma arising in the nipple with an expansive and circumscribed growth, mimicking an in situ lesion of the breast on the hematoxylin and eosin stained sections, but in which a myoepithelial layer around neoplastic nodules could not be detected by using immunohistochemistry. To the best of our knowledge, primary origin in the nipple is very rare for SPCs and it has been described only once in the literature. The case we herein illustrate is of interest not only because of its origin in the nipple, but also because of its not in situ, but invasive, although expansive and not infiltrative growth. In the differential diagnosis, nipple disorders as adenoma and syringomatous adenoma, usual ductal hyperplasia (UDH), papilloma, intracystic papillary carcinoma, lobular carcinoma in situ, ductal carcinoma in situ and skin adnexal tumors are considered.

Case reports

Splenic histiocyte-rich pseudotumor following chemotherapy for non Hodgkin diffuse large B cell lymphoma

A.G. Abdou, M. Kandil, M.S. Eldien, R. Abdallah

Chemotherapy may induce mass lesion in rare conditions, which can be easily mistaken as a residual tumor mass. In this report, we describe a mass affecting spleen in a patient received chemotherapy for non Hodgkin diffuse large B cell lymphoma. This mass proved histologically to be non neoplastic formed of sheets of histiocytes and xanthoma cells, which is called histiocyte-rich pseudotumor. This report describes this rare lesion and the possible differential diagnosis.

A puzzling ovarian tumour: pregnancy luteoma with diffuse endometriosis

F. Limaiem, S. Bouraoui, S. Bouslama, A. Lahmar, S. Mzabi

Background. Pregnancy luteoma is a distinctive non-neoplastic hormone dependent lesion arising in pregnancy and mimicking an ovarian tumour. Fewer than 200 cases have been described in the English-language literature. Its clinical and morphological features are characteristic and must be considered in order to prevent diagnostic misinterpretation.

To the best of our knowledge the association of pregnancy luteoma with endometriosis has not been reported in literature to date.

Case report. A 30-year-old pregnant woman with no particular past medical history, consulted her gynaecologist at 17 weeks gestation for routine check-up. The patient was asymptomatic and did not show any signs of virilization. Ultrasonography disclosed a left adnexal heterogeneous mass measuring 7 cm in diameter with intramural vegetations. The right ovary was unremarkable. The patient underwent salpingo-oophorectomy considering the imaging findings were suspicious for malignancy. Histologically, the lesion was constituted of large sheets of luteinized polygonal cells with abundant eosinophilic cytoplasm and small round nuclei devoid of atypia and mitotic figures. In addition, there were several ectopic endometrial glands surrounded by abundant decidualized or edematous stroma. Immunohistochemically, these glands were immunoreactive for cytokeratin 7. The final pathological diagnosis was pregnancy luteoma associated with diffuse endometriosis.

Conclusions. Because of its relative rarity, pregnancy luteoma is likely to be clinically misinterpreted and overtreated, as in the present case.

Unusual pleural effusion from vulvar squamous cell carcinoma: report of a case and review of the literature

S. Erra, G. Patrucco, G. Speranza, S. Barbero, F. Brucculeri, G. Taverna

Vulvar tumors are not very common and account for about 4% of all cancers affecting the female genital organs. Frequently, malignant neoplasia of this site have squamous phenotype and the rare cases of metastasization are reported in the locoregional lymph nodes and in the surrounding organs.

We report a case of metastasization of a vulvar squamous cell carcinoma in an unusual place such as the parietal pleura, in a relapsing patient that was submitted to a surgical vulvectomy the previous year.

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