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ORIGINAL ARTICLES

Lipomatous myofibroblastoma of the breast: case report with diagnostic and histogenetic considerations

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We report rare case of myofibroblastoma (MFB) of the breast comprised predominantly of a mature fatty component, representing approximately 70% of the entire tumour area. This tumour, designated “lipomatous MFB”, should be interpreted as the morphological result of an unbalanced bidirectional differentiation of the precursor mammary stromal cell, with the adipocytic component overwhelming the fibroblastic/myofibroblastic one. Lipomatous MFB is a rare variant of mammary MFB, which can mimic malignancy because of the close juxtaposition of fibroblasts/myofibroblasts with mature adipocytes, resulting in a finger-like infiltrative growth pattern of the former towards the latter. Histogenetic considerations and differential diagnostic problems with other bland-looking spindle cell tumours containing infiltrating fat are provided.

Cytological features of nipple adenoma in scraping smears

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Introduction. Nipple adenoma (NA) is a benign epithelial lesion of the breast that can clinically simulate Paget’s disease or invasive ductal carcinoma. Therefore, correct pre-operative diagnosis is important for appropriate management.

Methods. Cytological samples may be obtained by different methods such as fine needle aspiration, nipple discharge or nipple scraping. Herein, the cytological features of three cases of NA are described in which samples were derived from nipple scraping.

Results. In all three cases, patients were adult females presenting with a sub-areolar nodule, showing skin ulceration in 2 of 3 cases. The nipple scraping cytological smears were characterised by a bloody background with epithelial cells arranged in clusters or singularly, showing an irregular nuclei profile. These features could simulate a malignant process. However, at higher magnification, fine nuclear chromatin with inconspicuous nucleoli and presence of myoepithelial cells were helpful to exclude malignancy.

Discussion. NA may present “worrisome” cytological features on smears derived from nipple scraping. Therefore, knowledge of the cytological spectrum of this lesion is important to avoid misdiagnosis.

Cytologic features of solid pseudopapillary neoplasms of the pancreas: a single institutional experience based on evaluation of diagnostic utility of endoscopic ultrasound-guided fine needle aspiration (EUS-FNA)

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Background. Endoscopic ultrasound-guided fine needle aspiration (EUS-FNA) is an important modality for diagnosing solid and cystic pancreatic lesions. The objectives of this retrospective study are to review the cytologic criteria used to diagnose pancreatic solid pseudopapillary neoplasms (SPNs) and evaluate the utility of EUS-FNA by correlating cytologic and histologic samples.

Case reviews. Of the 924 pancreatic FNAs performed at our institution from January 2002 through February 2013, four histologically confirmed cases of SPN were identified; three had an initial cytologic diagnosis of SPN. All four cases lacked on-site evaluation. Cytologic smears were assessed by two reviewers for the presence of a cellular aspirate, fibrovascular stalks lined by neoplastic cells with pale to finely granular cytoplasm, and monotonous, oval nuclei containing delicate chromatin, inconspicuous nucleoli, and grooves and inclusions.

Three cases were diagnosed as SPN on cytologic examination and confirmed histologically. The remaining case was deemed a pancreatic endocrine neoplasm on cytology, but SPN on final histology. The most consistent cytologic feature we encountered was the presence of a cellular aspirate containing fibrovascular stalks lined by monotonous neoplastic cells with oval nuclei and nuclear grooves.

Conclusion. We conclude that EUS-FNA is an effective diagnostic tool in the diagnosis of pancreatic SPNs.

Neuroendocrine tumours of the pancreas: a clinicopathological study of nine cases including six insulinomas

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Background. Pancreatic neuroendocrine tumours (pNET) are relatively uncommon, accounting for 1-2% of all pancreatic neoplasms. They are characterised by varying clinical presentation, tumour biology and prognosis.

Aim. To provide an updated overview on clinicopathological features, treatment and outcome of pNET.

Patients and methods. In our retrospective study, we reviewed 9 cases of pNET that were diagnosed at the Pathology Department of Mongi Slim Hospital over an 11-year period (2003- 2013). Relevant clinical information and microscopic slides were available in all cases and were retrospectively reviewed. The latest WHO classification (2010) was adopted.

Results. Our study group included 3 men and 6 women (M/F ratio 0.5) with an age between 20 and 75 years (mean = 52 years). Pancreatic neuroendocrine tumours ranged in size from 0.5 to 10 cm (mean 4 cm). The sites of pNET were the head of the pancreas (n = 4), the body of the pancreas (n = 3) and the tail of the pancreas (n = 2). Enucleation of the tumour was performed in five cases. Three patients underwent distal pancreatectomy and splenectomy, whereas only one patient had central pancreatectomy. Histopathological examination of the surgical specimen coupled with immunohistochemical study established a diagnosis of pNET grade 1 (G1) in seven cases and grade 2 (G2) in two cases.

Conclusion. Pancreatic neuroendocrine tumours are a heterogeneous group of neoplasms with distinct tumour genetics, biology and clinicopathological features. Accurate clinical and pathologic diagnosis is an important first step in developing an appropriate management plan.

BRIEF ORIGINAL ARTICLE

Pre-miR146a expression in follicular carcinomas of the thyroid

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Introduction. Micro-RNA, a new class of small, non-coding RNAs, have been shown to be deregulated in several human carcinomas. In particular, SNP rs2910164 in pre-miR146a appears to be correlated with papillary thyroid carcinoma and may be involved in its genetic predisposition. Since data on follicular thyroid carcinomas (FTC) are lacking, we evaluated the involvement of SNP rs2910164 in FTC.

Methods. Thirty-nine cases of FTC and 20 follicular adenomas, defined according to WHO criteria, were selected. DNA and RNA were extracted from formalin-fixed paraffin-embedded blocks of both neoplastic and non-neoplastic areas. The DNA region of pre-miR146a, containing SNP rs2910164, was sequenced. Total RNA including miRNAs was used for stem-loop RT reactions, and applying a standard TaqMan PCR kit protocol for real-time PCR. Wilcoxon signed-rank test and Friedman test were used for statistical analyses.

Results. In 31% of FTC, the G allele was observed in neoplastic tissues, compared with the non-neoplastic areas ($p < 0.05$), whereas the CC phenotype was completely absent in tumours. Moreover, the expression of pre-miR146a was found to be significantly down-regulated in neoplastic tissues from FTC cases ($p = 0.043$), although no significant differences were seen in follicular thyroid adenomas.

Discussion. The expression profile of pre-miR146a can be correlated with FTC tumorigenesis. The G allele in SNP rs2910164 appears to be correlated with the transition from normal to neoplastic tissue. The GG and GC alleles appear to be associated with an increased risk for FTC, while the CC allele seems to play a protective role.

CASE REPORTS

Olfactory neuroblastoma with focal ganglioneuroblastic differentiation: a case report with literature review

S. Squillaci

Olfactory neuroblastoma (ONB) is a rare malignant neuroectodermal tumour, with clearly defined histologic and immunohistochemical

features, that typically arises in the superior nasal cavity. Although the classical clinicopathological features leave little room for misinterpretation, the wide variability in this tumour, including occasional divergent differentiation, may cause diagnostic difficulty. Herein, an unusual case of ONB with focal ganglioneuroblastic differentiation in an 81-year-old woman arising from the anterior ethmoid, filling the upper portion of the left nasal cavity and sparing the sinus cavities, is described. Histologically, the tumour was composed of atypical monotonous round cells that were positive for NSE, CD56, chromogranin, synaptophysin, neurofilament and calretinin and exhibited an irregular lobulated and nested growth pattern and sparse mitotic figures (3 to 4 mitoses per 10 HPF). Focally, the histology changed to ganglioneuroblastic differentiation consisting of large ganglion and spindle cells, positively staining for S-100, GFAP, CD99, neurofilament, calretinin, chromogranin and synaptophysin. Neuroblastomas, occurring in the nasal cavity, in analogy to other sites, tend to have an aggressive biologic behaviour and can histologically mimic other undifferentiated malignant neoplasms of the sinonasal tract. Differential diagnostic problems are discussed; a comprehensive review of the literature has also been performed with a focus on survival.

Papillary haemangioma: a case report of multiple facial location
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Papillary haemangiomas were recently defined as morphologically distinct and benign cutaneous haemangiomas showing a predominantly intravascular capillary proliferation within dilated thin-walled dermal blood vessels. We describe the case of a 45-year-old woman who presented with multiple eruptive red-bluish raised papules and nodules distributed over the skin of the chin that were related to a papillary haemangioma.

Juvenile hyaline fibromatosis: a case report

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Juvenile hyaline fibromatosis is a rare, hereditary disease with distinct clinical and histopathological features. Clinically, it presents with gingival hypertrophy, pappulonodular skin lesions and joint contractures. Bone involvement is usually an uncommon finding. We report a case of a 2-year-old patient, daughter of consanguineous parents, who presented since the age of 2 months with impairment of mental development, multiple joint contractures, motion limitation and nodules on the scalp. The calvarian lesions were surgically removed, and histopathological examination concluded to juvenile hyaline fibromatosis.

Uterine tumour resembling ovarian sex cord tumours presenting as multiple endometrial and cervical uterine polyps: a case report

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Background. Uterine tumours resembling ovarian sex-cord tumours (UTROSCT) are very rare, benign uterine tumours, composed solely of sex cord elements. These tumours have a polyphenotypic immunophenotype that favours a derivation from uterine mesenchymal stem cells.

Case report. A 43-year-old female presented with recurrent vaginal bleeding. On hysteroscopy, she had multiple endometrial and cervical polyps that were removed endoscopically. Histologically, the specimen contained epithelioid cells arranged in tubules, trabeculae and anastomosing cords, without significant cellular atypia or mitotic activity.

Immunohistochemical studies were performed. The tumour was found to be diffusely positive for vimentin, calretinin and desmin, focally positive for cytokeratin, CD99 and inhibin and negative for chromogranin and CD10. A subsequent total hysterectomy was performed and revealed neoplastic infiltration of the myometrium. **Conclusion.** A polyphenotypic immunophenotype is a characteristic feature of UTROSCT, and may be helpful in diagnosis and in exclusion of other lesions. Familiarity with this tumour by gynaecologists and pathologists is essential to avoid misdiagnosis: correct diagnosis of this neoplasm is important in patient management.

Giant pedunculated polypoid submucosal lipoma of the splenic flexure of colon: case report and review of the literature

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Lipomas of the colon are rare but clinically important conditions that require suitable evaluation for guiding appropriate therapy. The majority of lipomas arise from the submucosal layer in the ascending colon, especially near the ileocecal valve, which causes difficulties in diagnosis. Giant lipomas may be misinterpreted as a premalignant adenomatous polyp, particularly when arising in the left colon. A 38-year-old man presented with manifestations including hypogastric pain, constipation, loss of appetite and weight, accompanied by anaemia, nausea, vomiting and haematochezia. Colonoscopy revealed a large submucosal polyp about 5×4 cm, which was located at the splenic flexure of colonic. Surgery detected an oval polypoid tumour measuring 70×50×45 mm in size, having a pedunculated appearance and a stalk diameter of 20 mm. Histopathologic examination of the biopsy from the lesion confirmed diagnosis of a giant submucosal lipoma. In our experience, most giant colonic lipomas are found to be sessile and occur in the ascending colon in older patients. Herein, we report a pedunculated tumour in a 38-year-old male located in the splenic flexure of colon.

“Pure” primary large cell neuroendocrine carcinoma of the urinary bladder: case report, literature review and diagnostic criteria

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Introduction. Large cell neuroendocrine carcinoma (LCNC) is defined in the urinary bladder, as in other sites, as a high-grade neoplasm exhibiting neuroendocrine features at the H&E level, high mitotic activity and evidence of neuroendocrine differentiation by immunohistochemistry. We report a case of pure bladder LCNC with review of the literature.

Methods. A 68-year-old male presented with gross haematuria of two weeks' duration in October 2011. Transurethral resection and subsequently radical cystoprostatectomy (CP) with bilateral lymphadenectomy (L) were performed in December 2012.

Results. Urinary cytology identified malignant cells. Histologically, the tumour showed organoid nesting, trabecular growth, rosettes and perilobular palisading patterns, suggesting neuroendocrine differentiation. Immunohistochemical staining showed intense positivity for CD56.

Discussion. We examined all published pure bladder LCNC (12 cases) excluding mixed neoplasms. Small cell carcinoma of the urinary bladder pure LCNC of the bladder is a very aggressive malignancy, unresponsive to therapy, presents in an advanced stage and has a propensity for early metastasis. Prior to the advent of immunohistochemistry, such cases would most likely have been categorised as poorly differentiated, high-grade urothelial carcinomas.