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REVIEW

PDX-1 (Pancreatic/Duodenal Homeobox-1 Protein 1)

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The homeodomain-containing transcription factor pancreatic duodenal homeobox 1 (PDX-1) plays a key role in pancreatic development and β -cell function. It is a major regulator of transcription in pancreatic cells, and transactivates the insulin gene by binding to a specific DNA motif in its promoter region. Glucose also regulates insulin gene transcription through PDX-1. It has been shown that PDX-1 is required for maintaining pancreatic islet functions by activating gene expression and has a dual role in pancreatic development. It initially contributes to pancreatic formation during embryogenesis and subsequently regulates the pancreatic islet cell physiology in mature islet cells. Because of this key role in the embryologic development of the pancreas, PDX-1 expression has been investigated in pancreatic cancer cell lines and human tumors. Moreover, a few reports have described expression of PDX-1 in other human neoplasms and have investigated its potential role in differential diagnosis, but data on normal human tissues are lacking. Understanding the molecular mechanisms of pancreas formation, and especially the function of PDX-1, may contribute to the improved treatment and prevention of debilitating diseases such as diabetes, insulinomas and pancreatic carcinomas. Nevertheless, further studies are needed concerning its possible application in routine practice.

ORIGINAL ARTICLES

Lipomatous angiofibroblastoma of the vulva: diagnostic and histogenetic considerations

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We report a rare case of angiofibroblastoma (AMFB) of the vulva, composed predominantly of a mature fatty component, representing approximately 60% of the entire tumour. The tumour, designated as "lipomatous AMFB", should be interpreted as the morphological result of an unbalanced bidirectional differentiation of the presumptive precursor stromal cell resident in the hormonally-responsive stroma of the lower genital tract, with the adipocytic component overwhelming the fibroblastic/myofibroblastic one. The close admixture of adipocytes with spindled/epithelioid cells of the conventional AMFB resulted, focally, in a pseudo-infiltrative growth pattern and pseudo-lipoblast-like appearance, raising problems in differential diagnosis, especially with well-differentiated lipoma-like liposarcoma and spindle cell liposarcoma. Awareness of the possibility that vulvo-vaginal AMFB may contain large amount of lipomatous component is crucial to avoid confusion with other bland-looking spindle cell tumours containing infiltrating fat.

A peculiar fibroma-like lesion of superficial soft tissue: morphologic and immunophenotypic evaluation

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A peculiar lesion of superficial soft tissue characterised by fibroma-like morphology and an immunohistochemical profile consisting of CK+, VIM+, CD34+, CD31+/-, FLI1+ and INI-1 retained is described. The lesion entered into differential diagnosis with the so-called fibroma-like variant of epithelioid sarcoma, with the entities defined as ES-like/pseudomyogenic haemangiopericytoma and the recently identified entity defined as superficial CD34+ fibroblastic tumour. All of these entities share a common morphological structure, but differ in their immunophenotypic profile.

CASE REPORTS

Mixed stromal and smooth muscle tumours of the uterus: a report of two cases

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Mixed stromal and smooth muscle uterine tumours, defined as those containing at least 30% of each component as seen by routine light microscopy, are rare. This report describes the morphological features of two such tumours diagnosed in 44-year-old and 50-year-old females complaining from recurrent uterine bleeding that was unresponsive to medical treatment. Morphological and immunohistochemical evaluations were performed, and a final diagnosis of mixed endometrial stromal nodule and smooth muscle tumour of the uterus was rendered in both cases.

Primary mediastinal angiosarcoma: a rare observation in a patient with 8-year-survival

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Background. Vascular tumours of the mediastinum are rare, accounting for 1-2% of all mediastinal tumours in this location. Angiosarco-

mas are most often encountered as sporadic lesions, typically in the scalp or face of elderly patients. However, they can occur in any anatomic site. Mediastinal angiosarcomas (MA) are very rare with less than 50 cases reported.

Case report. The authors describe the case of a 38-year-old woman whose past medical history was consistent for a MA that was diagnosed in 2003. This tumour was treated by complete surgical resection followed by radiation therapy and chemotherapy. Diagnosis was based on histologic examination. In 2011, the patient presented a pleural localisation of the angiosarcoma and died one month after admission, 8 years after diagnosis of the MA.

Conclusion. MA is a very rare tumour causing a diagnostic dilemma. Clinical and radiologic findings are non-specific, and final diagnosis is based on histologic examination. The case described is unusual considering the long period of survival, which may be explained by the treatment modalities associating complete surgical resection, chemotherapy and radiation therapy.

Dermatofibrosarcoma protuberans of the vulva: a mesenchymal tumour with a broad differential diagnosis and review of literature

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Dermatofibrosarcoma protuberans (DFSP) is a malignant cutaneous soft tissue tumour, which rarely presents in the vulva. We report an unusual case of this tumour involving the vulva. A 61-year-old female presented with a mass in the left mons pubis. Subsequent excisional biopsy of the mass was performed. Histologic evaluation of the specimen showed a spindle cell lesion consisting of fibroblast-like cells arranged in a storiform pattern. On average, there were 2 to 3 mitotic figures per 10 high power field (hpf). The neoplastic cells showed extension into the surrounding fibroadipose tissue. A panel of immunohistochemical stains including CD34, S-100, melan-A, HMB-45, vimentin and smooth muscle actin (SMA) were tested. The neoplastic cells showed diffuse staining with CD34 and vimentin, while the rest were negative. Based on the morphologic and immunohistochemical staining pattern, a diagnosis of DFSP was rendered. The patient underwent two subsequent resections before she had clear resection margins. The postoperative course was unremarkable. The patient is disease free without recurrence after a follow-up of 12 months. DFSP infrequently involves the vulva and should be considered in the differential diagnosis of other spindle cell lesions presenting in this unusual site. The role of immunohistochemical staining with CD34 is imperative in establishing the diagnosis. The rate of local recurrence is high, but it rarely shows metastasis. Treatment of choice is wide local surgical excision with close follow-up to detect recurrence.

Unusual presentation of metastatic adenoid cystic carcinoma: a challenge in aspiration cytology of the thyroid

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Introduction. Adenoid cystic carcinoma is a malignant neoplasm most commonly originating in the salivary glands. Its occurrence elsewhere is rare and its metastasis to the thyroid gland has been described only once.

Case report. We describe the case of a 66-year-old man who presented for a swelling in the midline neck of six months duration. A solitary palpable nodule was identified in the isthmus region of the thyroid. Fine needle aspiration of the nodule revealed high cellularity, a partial microfollicle-like pattern and the presence of small hyaline globules. The neoplastic population was composed of monomorphic cells with basaloid appearance. Thyroid primitivity was excluded on the basis of the negativity for TTF1 and thyroglobulin. As the patient referred an ulcerative lesion of the inferior lip, fine needle aspiration cytology of the lesion was performed, yielding a diagnosis of adenoid cystic carcinoma.

Conclusion. The present case highlights the need to be aware of possible metastatic thyroid localisation of adenoid cystic carcinoma also originating in minor salivary glands of the oral cavity. This is a very rare event, but it should be taken into consideration and clinical and cytological findings must be carefully examined.

Gastrointestinal stromal tumour of the stomach with osseous differentiation: a case report

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Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal neoplasm of the gastrointestinal tract, while osseous metaplasia of this tumour is an unexpected event. To date, no cases have been reported in the literature. Herein, we report a case of a 60-year-old man affected by a GIST with benign osseous metaplasia and mature bone formation. We also discuss the pathogenesis of intratumoural ossification and review the relevant literature. The prognostic significance of ossification in GIST remains unclear because of the limited cases reported.