VERRUCIFORM XANTHOMA BEHAVING BADLY

Marie Bertrand-Philippe, Ben Ryan

Sullivan Nicolaides Pathology, Bowen Hills, Qld, Australia

Verruciform xanthoma is an uncommon, asymptotic, mucocutaneous lesion which occurs predominantly in the oral mucosa of patients in their 5th to 7th decades. Other extraoral sites include skin of the anogenital area. It presents as a well circumscribed, rough, granular or pebbly plaque and can mimic a squamous cell carcinoma macroscopically. Recurrence is extremely rare. Histologically, verruciform xanthoma is characterised by a papillary epithelium with acanthosis and parakeratosis invaginating between the epidermal projection, as well as elongated rete ridges which sometimes coalesce at their base. Within the connective tissue papillae there is a collection of large xanthoma cells.

Forty-four cases of verruciform xanthoma have been reported in our laboratory over a 10 year period representing <0.001% of the dermatological cases. Although 38 of 44 (86%) of the lesions occurred in the common sites (oral and anogenital), six cases were present at atypical sites including ear, wrist, thumb and leg. We are presenting these atypical cases of verruciform xanthoma to highlight the importance of considering this diagnosis when reporting a lesion mimicking a SCC in locations where verruciform xanthomas are not usually present.

OESTROGEN RECEPTOR EXPRESSION IN A RARE CASE OF LEIOMYOMA OF DEEP SOFT TISSUE – A CASE REPORT

Ramzi Amin, Clarence Teo, Nicholas Koh

Department of Pathology, Tan Tock Seng Hospital, Singapore

Uterine leiomyoma is a common smooth muscle neoplasm. Gynaecologic type leiomyoma of deep soft tissue, on the other hand, is rare and commonly occurs in the retroperitoneum or abdominal cavity of young to middle aged women. We report a 42-year-old female who initially had hysterectomy performed for uterine leiomyoma at another institution but was also noted to have a 30 cm retroperitoneal mass. She was then referred to our institution where resection of the mass revealed a well-circumscribed tumour with histology features of a leiomyoma, supported by the expression of smooth muscle markers with immunohistochemical staining. Additionally, the lesional cells were both oestrogen-receptor and progesterone-receptor positive, a typical feature of this lesion which suggests derivation from hormonally sensitive smooth muscle cells in this extra-uterine location. Awareness of this benign entity is important to avoid a misdiagnosis of leiomyosarcoma so that unnecessary additional treatment can be avoided.

HISTOPATHOLOGY OF THE PLACENTA IN CONGENITAL SYPHILIS

Jamie Bishop, Jessica Matich

Histology Department, Middlemore Hospital, Auckland, New Zealand

Syphilis, a disease with a long and controversial past, has recently shown a resurgence in many countries. Cases of congenital syphilis are now being reported in New Zealand,

having previously been very rare prior to 2012. Congenital syphilis may lead to perinatal death or long term morbidity. The traditional histological triad seen in placentas consists of enlarged hypercellular villi, proliferative villous vasculature, and villitis. Necrotising funisitis, with a 'babypole' appearance at macroscopy, is another common finding. *Treponema pallidum* immunohistochemistry may assist in the diagnosis. The previous rarity of the disease means that many pathologists may not be very familiar with the histopathological features. We present two recent cases of congenital syphilis, with placental histology, as a reminder. Recognition of congenital syphilis is important in aiding diagnosis, leading to appropriate follow up and treatment for both mother and infant.

References

1. Ministry of Health. *National Syphilis Action Plan: An action plan to stop the syphilis epidemic in New Zealand*. Wellington: Ministry of Health, 2019.
2. Tsimis ME, Sheffield JS. Update on syphilis and pregnancy. *Birth Defects Res* 2017; 109: 347–52.
3. Genest DR, Choi-Hong SR, Tate JE, *et al*. Diagnosis of congenital syphilis from placental examination: comparison of histopathology, Steiner stain, and polymerase chain reaction for *Treponema pallidum* DNA. *Hum Pathol* 1996; 27: 366–72.

ROSETTE-FORMING GLIONEURONAL TUMOUR IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1 – A CASE REPORT

Jessica Bonavita, Peter Robbins

Department of Anatomical Pathology, PathWest Laboratory Medicine, QEII Medical Centre, Perth, WA, Australia

Rosette-forming glioneuronal tumour (RGNT) is a rare primary CNS neoplasm originally described arising in the fourth ventricle, and displaying a distinctive biphasic appearance of neurocytic and astrocytic components. Patients with neurofibromatosis type 1 (NF1) can have a variety of CNS manifestations, including neoplastic, and RGNT has been described in four cases to date. This case study reports a patient with NF1 who underwent resection of a left cerebellar lesion. Histology showed a lesion dominated by synaptophysin positive, neurocytic cells in a myxoid background, forming perivascular pseudorosettes and true rosettes surrounding a central eosinophilic fibrillary core. A second component of Olig2+, oligodendroglial-like cells was also noted in a nondescript glial background. Due to its readily recognisable appearance, a diagnosis of RGNT was made. This is now a fifth case report of RGNT, an already rare tumour, in a patient with NF1 and raises the interesting possibility of a potential association between the two.

SOLID PAPILLARY CARCINOMA OF THE BREAST WITH REVERSE POLARITY – A CASE REPORT OF A RARE ENTITY WITH A NOVEL IDH2 MUTATION

Jessica Bonavita, Celia Green, Ramela Rajakaruna,

Fabienne Griefu-Iacopetta, Benhur Amanuel, Felicity Frost

Department of Anatomical Pathology, PathWest Laboratory Medicine, QEII Medical Centre, Perth, WA, Australia

Solid papillary carcinoma with reverse polarity (SPCRP) is a rare breast carcinoma bearing resemblance to the tall cell variant of papillary thyroid carcinoma (PTC). These tumours have been

found to harbour mutations in the isocitrate dehydrogenase 2 (IDH2) gene. The patient presented with an impalpable 8 mm right breast lesion, detected on staging CT scan for known endometrial carcinoma. The lesion was well defined and hypo-echoic on ultrasound, and occult on mammogram. Core biopsy demonstrated a papillary neoplasm with focal atypical features. Excisional biopsy revealed a circumscribed papillary neoplasm composed of cuboidal to tall columnar cells with abundant granular eosinophilic cytoplasm. Nuclei were notably aligned away from the basal aspect of the cells and exhibited PTC-like nuclear features including grooves and intranuclear pseudo-inclusions. The cells diffusely expressed cytoplasmic CK5/6 and there was very focal nuclear positivity for ER. Calretinin was patchy and cytoplasmic. No intact myoepithelial layer was demonstrated. A diagnosis of SPCRP was made and ancillary molecular testing revealed a novel IDH2 variant c520G>A, p.(Ala174Thr). SPCRP is a rare tumour with a favourable prognosis, which exhibits distinctive histological and immunophenotypical features that are readily recognisable to pathologists.

UTERINE INFLAMMATORY MYOFIBROBLASTIC TUMOUR: A CASE REPORT

Jordan Butler¹, Lyndal Anderson¹, Diane Cominos², Robert Rawson¹

¹Department of Tissue Pathology and Diagnostic Oncology, NSW Health Pathology, Royal Prince Alfred Hospital, Sydney, Australia; and ²Histopathology and Cytopathology, Sullivan Nicolaides Pathology, Brisbane, Australia

Inflammatory myofibroblastic tumour (IMT) is a mesenchymal neoplasm with intermediate biological potential, which may be under-recognised in the uterus.^{1–3}

Morphological and immunophenotypic overlap exists between uterine IMT and some other uterine mesenchymal neoplasms, including smooth muscle tumours.² Morphological features suggestive of IMT may be subtle or focal and include myxoid change, tapered nuclei, and a lymphoplasmacytic infiltrate.¹

Chromosomal rearrangements involving the *anaplastic lymphoma kinase (ALK)* gene appear to be frequent in uterine IMT,^{1–3} and absent in other uterine mesenchymal neoplasms.² Therefore, ALK immunohistochemistry (IHC) and *ALK* fluorescent *in situ* hybridisation (FISH) appear to be useful tools to confirm the diagnosis of uterine IMT.³

Although most uterine IMTs follow a benign clinical course, local recurrence and metastases have been observed in a small subset.¹ Correct diagnosis is important to ensure accurate prognosis and optimal management, which may include a tyrosine kinase inhibitor.

We present a case of uterine IMT with morphology and IHC suggestive of a leiomyosarcoma with myxoid change. ALK IHC was positive and an *ALK* rearrangement was demonstrated with FISH, confirming the diagnosis of uterine IMT.

This case illustrates the importance of a low threshold for ALK testing in uterine mesenchymal tumours with morphological features suggestive of IMT.

References

1. Pickett JL, Chou A, Andrici JA, *et al.* Inflammatory myofibroblastic tumors of the female genital tract are under-recognized. *Am J Surg Pathol* 2017; 41: 1433–42.
2. Mohammad N, Haimes JD, Mishkin S, *et al.* ALK is a specific diagnostic marker for inflammatory myofibroblastic tumor of the uterus. *Am J Surg Pathol* 2018; 42: 1353–9.

3. Parra-Herran C, Quick CM, Howitt BE, *et al.* Inflammatory myofibroblastic tumor of the uterus: clinical and pathologic review of 10 cases including a subset with aggressive clinical course. *Am J Surg Pathol* 2015; 39: 157–68.

LOCALISED PULMONARY CRYSTAL-STORING HISTIOCYTOSIS WITH UNDERLYING EXTRANODAL MARGINAL ZONE LYMPHOMA: A CASE REPORT

Sunisha Chahal, J. J. Serfontein, Nicola Kingston

Department of Anatomical Pathology, LabPLUS, Auckland City Hospital, Auckland, New Zealand

Crystal-storing histiocytosis (CSH) is a rare entity, composed of a proliferation of non-neoplastic histiocytes with an accumulation of intracytoplasmic crystalline material. It is often associated with an underlying lymphoproliferative disorder, most often those that express monoclonal immunoglobulins, such as multiple myeloma and lymphoplasmacytic lymphoma. Here we report a case of localised pulmonary CSH in an asymptomatic 54-year-old man, found to have an incidental solid lesion in the right lung on computed tomography (CT). The lesion was composed of sheets of CD68 positive histiocytes that contained refractile eosinophilic crystalline inclusions, scattered lymphoid follicles of CD20 positive B lymphocytes, and occasional plasma cells. PCR for immunoglobulin heavy chain gene rearrangement confirmed the presence of a monoclonal population of B cells. These findings were supportive of a diagnosis of CSH with an underlying B cell/ plasma cell lymphoproliferative disorder, which was favoured to be an extranodal marginal zone lymphoma after clinical and radiological workup. Recognition of this rare entity is important as it may obscure an underlying lymphoproliferative disorder. A diagnosis of CSH should prompt careful exclusion of neoplasia, including clinical workup for systemic disease. In this case report we also discuss clinical features and diagnostic pitfalls.

WHOLE EXOME SEQUENCING (WES) ENHANCES THE DIAGNOSTIC RATE OF PERINATAL AUTOPSY: A PROSPECTIVE CLINICAL UTILITY TRIAL WITH IMPLICATIONS FOR PRENATAL DIAGNOSIS

Fiona Chan^{1,2,3}, Alison Yeung^{4,5}, Anand Vasudevan^{2,4}, Zornitza Stark^{4,6}, Stacey Prystupa⁵, Yuen Chan⁵, Trishe Leong⁷, Kerry Ireland-Jenkin^{7,8,9}, Susan Fawcett², Melissa Graetz⁸, Katherine Rose⁵, Samantha Ayres^{3,4}, Anna Jarmolowicz^{3,4}, Gemma Brett^{4,6}, Yael Praver^{3,5}, Heather Chaliner⁷, Candice Dao⁸, Tenielle Davis², Lisa Hui⁹, Mark Teoh⁵, Shelley Rowlands², Susan Walker⁸, Elly Lynch^{3,10}, Melissa Martyn^{3,10}, Belinda Chong^{4,6}, Clara Gaff^{3,10}, Sebastian Lunke^{4,6}, Jackie Collett^{1,2}, George McGillivray^{2,4}

¹Royal Children's Hospital, Melbourne, Australia; ²Royal Women's Hospital, Melbourne, Australia; ³Melbourne Genomic Health Alliance, Melbourne, Australia; ⁴Murdoch Children's Research Institute, Melbourne, Australia; ⁵Monash Medical Centre, Melbourne, Australia; ⁶Victorian Clinical Genetics Services, Melbourne, Australia; ⁷Austin Health, Melbourne, Australia; ⁸Mercy Hospital for Women, Melbourne, Australia; ⁹University of Melbourne, Melbourne, Australia; and ¹⁰Walter and Eliza Hall Institute, Melbourne, Australia