Paediatric and Perinatal Pathology: Congenital Tumours and Pseudotumours
Case 3

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Clinical History

• Male admitted at 24 hours of age
• Short hx of bilious vomiting and intolerance of feed
• Investigations (plain x-ray chest and abdomen):
  • Contrast studies normal passage of contrast into small bowel
    – Contrast studies → normal contrast passage into small bowel
    – Thoraco-abdominal right sided para-spinal mass
• “Feed and wrap” – MRI = 7.5 x 3.6 x 2.2 cm right sided paravertebral mass with extension into the intervertebral foramina
• Displacement and compression of the upper/mid thoracic spinal cord
• Dx: neuroblastoma (??lymphoma)
• No signs of spinal cord compression
Presentation MRI Scan

age: 2 days
Dx biopsy
Dx biopsy
Dx biopsy
Dx biopsy
Dx biopsy
Dx biopsy
Dx biopsy
Dx biopsy

Masson’s Trichrome
Dx biopsy

Vimentin
Dx biopsy

CD34
Dx biopsy

Ki67
Dx biopsy

CD45
Dx biopsy

INI1
Dx biopsy

NEGATIVE: myosin, SMM, αSMA, desmin, calponin, H caldesmon, S100, ERG
Dx biopsy

BCOR (Great Ormond Street Hospital, UK)
Dx biopsy 2 (split bx for review)
Dx biopsy (split bx for review)
Molecular Testing
FISH and RT-PCR

NO REARRANGEMENT/Negative:

- FUS
- ETV6
- NTRK3 or NTRK1
- PDGFRβ

- RT PCR for BCOR Exon 16 Internal Tandem Duplication (Dr R Allagio, Italia)
- (ERG neg → EWSR1 – SMAD3 rearranged fibroblastic tumour unlikely)
Dx

- “Primitive mesenchymal tumour with haemangiopericytomatous pattern”

- DD: infantile myofibroma / PMMTI...
Clinical History cont.

• Danger of cord compression

• Chemotherapy - Carboplatin / Etoposide x 2 (over 4 weeks/month)

• Vincristine / Cyclophosphamide / Doxorubicin x 4 (over 12 weeks)

• Surgery - right thoracotomy – (6 weeks after tx)
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection
Postchemotherapy resection

SMM
Postchemotherapy resection

SMA
Postchemotherapy resection

Ki67
Diagnosis:

Infantile myofibroma (IM)
Diagnosis: (Infantile) Myofibroma (IM)

A biphasic tumour composed of mature and immature myofibroblastic cells with a haemangiopericytoma-like pattern (solitary or multi-centric presentation).

History:

- Initially described as *congenital fibrosarcoma* by Williams and Schrum in 1951 (AMA Archives Pathology 1951; 51: 582-52).
- Stout (1951): *congenital generalised fibromatosis*.
Infantile myofibroma (IM)

- Prevalence: 1 : 150,000
- Usually present at birth or develop shortly thereafter; 90% of cases before the age of 2 years (age range 0-84 years)
- Various patterns of presentations:
  - SOLITARY (> 33% Head and Neck region (dermis, subcutis and striated muscle))
  - Solitary and multicentric IMs **not** involving the viscera > spontaneously regress.
Infantile myofibroma (IM)

Histology (1):

• Usually well circumscribed, unencapsulated lesion of spindle cells.
• Often biphasic/zonal architecture
• Periphery lighter (less cellular) bundles/lobules of ““mature” short fascicles or whorls of myofibroblasts with pale pink cytoplasm and long slender, tapered nuclei.
• Central zone darker, more cellular areas of immature/primitive cells with indistinct cytoplasm and basophilic nuclei; can show abrupt transition.
Infantile myofibroma (IM)

Histology (2):

- Pseudo chondroid areas (more basophilic matrix)
- Myoid balls contribute to classic lobular architecture
- Eosinophilic collagen present between cellular zones
- HPC-like pattern noted in 15 to 30% of lesions (non-specific finding)
- 30% of myofibromas may infiltrate around adjacent nerves, blood vessels, muscle, bone, salivary glands or adipose tissue
- Focal necrosis and occasional mitoses
Infantile myofibroma (IM)

Histology (3):

- Both components actin positive
  - smooth muscle actin/muscle specific actin
  - vimentin positive
  - CD34 up to 25% positive
  - desmin usually negative
  - S100 negative
Infantile myofibroma (IM)

*Site critical lesions → chemotherapy:*  
• Vincristine, Actinomycine-D, Vinblastine-Methotrexate, Interferon alpha and anti-oestrogen (Tamoxifen)  
• No clear treatment guidance (e.g. tumours with high mitotic rate)  
• Difficult to predict biological behaviour - with wide range of recurrence and regression published
Latest MRI Scan (08/2019)

age: 10 months days
Infantile myofibroma (IM) – Take home message:

• difficult dx if lesion is immature / monophasic and reveals no meaningful immunolabelling/immunophenotype
• site critical tumours may require chemotherapy
• difficult to predict biological behaviour, may recur several times despite chemotherapy
Merci beaucoup pour votre attention