

Erdheim-Chester Disease: a case report and role of Neurosurgery in ECD

Rare form of non-Langerhan cell histiocytosis with multisystem involvement

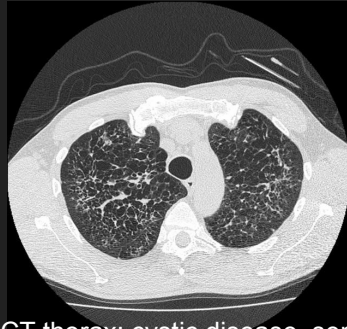
In Hong Kong, 1 case reported in 2007 and another reported in 2013

Involvement to long bone, brain, lung, skin, pituitary, posterior orbit etc

Imaging: plain XR, CT, MRI, some may require histological diagnosis



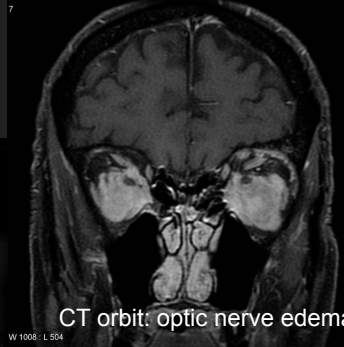
Knee XR: bilateral, symmetric metaphyseal and diaphyseal sclerosis, cortical thickening



CT thorax: cystic disease, septal thickening



CT brain: dural accumulations mimic meningiomas, with enhancing soft tissue masses



CT orbit: optic nerve edema, retrobulbar masses



CT abd: hairy kidney sign: irregular symmetric infiltration of the bilateral perirenal and posterior pararenal spaces coated aorta sign: periaortic soft tissue

52 year-old lady

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Presented with bilateral lower limbs weakness, numbness, unsteady gait, visual disturbance, headache Aug 2019

MRI showed leptomeningeal lesion at posterior fossa and lumbosacral nerve root

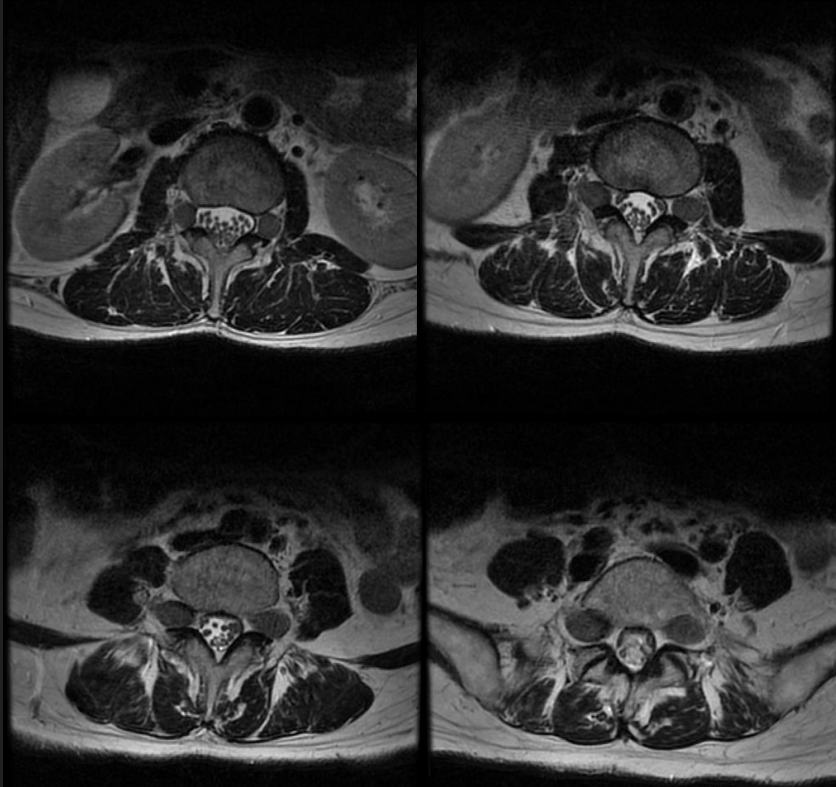
LP performed: Cytology showed large number of histiocytes, tumour markers negative

Leptomeningeal biopsy and VP shunt done Oct 2019. Pathology negative

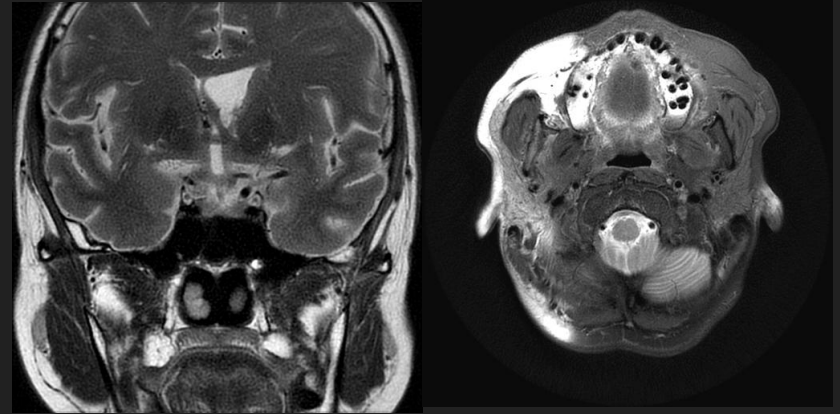
Given a course of prednisolone, azathioprine, IVIG, hydrocortisone

Responded to dexamethasone

MRI



MRI spine June 2020 showed bilateral thickening of spinal nerve root. Possibility of infiltrative or neoplastic disease. Swelling of dorsal root ganglion of LS spine and C spine, cauda equina



MRI brain July 2020 showed focal left para-sellar lesion, mild mass effect with narrowing of adjacent left internal carotid artery, yet still patent. Differential diagnosis includes inflammatory, neoplastic or granulomatous lesion.

Biopsy

Multidisciplinary meeting discussion

- Intracranial lesion in close proximity to left ICA
- Decided for L5/S1 laminectomy and biopsy of lesional nerve root.

L5/S1 laminectomy and nerve root biopsy was performed

- Yellowish thick caseating chalky tissue yielded after laminectomy
- Yellowish fluid after dura punctured
- Incisional biopsy of right nerve root

Pathology

Specimen: atypical histiocytosis in fibrous tissue with necrosis

Positive for CD68, CD163

Negative for S100, CD1a, LCA, CK, BRAF and HMB45

Pathology concluded histiocytosis consistent with Erdheim Chester Disease, BRAF V600E negative, target sequencing negative for CNS tumor panel.

In contrast to Langerhans cell histiocytosis (LCH), no S-100 nor CD1 are detected, but CD68 is positive.

CD68 - mononuclear lineage, malignancy histiocytosis

CD163 - in CSF, upregulated in inflammatory disease

S100 - proteins are normally present in cells derived from the neural crest, histiocytes

CD1a - CD1a, in particular, is a specific marker for Langerhans cells

LCA - Leukocyte common antigen is an marker for hematopoietic neoplasms of lymphoid type from neural, epithelial and mesenchymal derivatives like osteosarcoma

CK - cytokeratin expression profile allows the identification of epithelial cells

HMB45 - monoclonal antibody that reacts against an antigen present in melanocytic tumor

Discussion

Now patient has FU in Neurology

- On Peginterferon alfa-2a, hydrocortisone
- Plan for MARK mutation testing for potential MARK inhibitors

Diagnosis of ECD is made based on a mix of clinical findings, imagings and histological diagnosis

Typically present with bone lesion, which is not evident in this case

Involvement of neurosurgical structures reported in literature: pituitary, hypothalamus, dura

In the absence of more accessible lesions, neurosurgical input may be required for biopsy