

## CANP 2024 DIAGNOSTIC CASES

### Case 1

Assem S. Alrumeh<sup>1,2</sup>, Phedias Diamandis<sup>1,2</sup>, Andrew F. Gao<sup>1,2</sup>

<sup>1</sup>Dept. of Laboratory Medicine and Pathobiology, University of Toronto, Toronto, ON, Canada

<sup>2</sup>Laboratory Medicine Program, University Health Network, Toronto, ON, Canada

### Clinical summary:

A 72-year-old woman presented with back pain and was found to have a lytic lesion of the T7 vertebral body.

Recently, she was diagnosed with a hepatic leiomyosarcoma at an outside hospital after having had progressive abdominal pain, unintentional weight loss, and decreased appetite. Her other past medical history included hypertension, hypercholesterolemia, type 2 diabetes mellitus, and remote renal transplant.

CT of the spine showed a lytic lesion in the posterior T7 vertebral body and right pedicle with encroachment of the spinal canal. Prior MRI of the abdomen revealed a well-defined 4.5 x 4.0 cm mass in the liver along with an ovoid lesion adjacent to the left obturator externus muscle.

A T7 laminectomy was performed for tumour excision.

### Material submitted

1 H&E of spinal tumour

### Discussion points

1. Differential diagnosis?
2. Additional workup?
3. Pathogenesis?

## Case 2

Christopher Newell<sup>1</sup> & Christopher Dunham<sup>2,3</sup>

<sup>1</sup> Department of Pathology and Laboratory Medicine, University of Calgary, Calgary, AB, Canada

<sup>2</sup> Division of Anatomic Pathology, British Columbia Children's Hospital, 4500 Oak Street, Vancouver, BC, V6H 3N1, Canada.

<sup>3</sup> Department of Pathology and Laboratory Medicine, University of British Columbia, Vancouver, V6T1Z3, BC, Canada.

### Clinical Summary:

A 31-year-old woman (G8, T3, P1, A3, L4) was referred for fetal diagnostics at BC Women's Hospital after routine ultrasound studies at mid-gestation (21 weeks, 6 days) revealed severe hydrocephalus. The brain anomalies were suspected to be primary (query holoprosencephaly) and a poor prognosis rendered by Medical Genetics. The family elected for termination of pregnancy (24 weeks, 1 day gestational age) via intrafetal digoxin injection followed by dilatation and uterine extraction (D&E).

For the current pregnancy, clinical investigations revealed unremarkable protective serology and blood work. No gestational diabetes screen was performed and the mother denied exposures to alcohol, smoking or illicit drugs. Notably, the parents were first cousins. Pregnancy history was remarkable for three prior spontaneous abortions and a twin gestation in 2022 involving an unaffected female fetus and abnormal male fetus that was diagnosed with ventriculomegaly at 4-months gestation. The parents were told that this male fetus "would not survive". The etiology of the ventriculomegaly was presumed to be due to either infection or anemia as the mother has been sick with several episodes of fever during the pregnancy. Clinical investigations at that time revealed no evidence of infection, although no genetic investigations were pursued. Macrocephaly of the male fetus necessitated Caesarean section and at the time of birth this fetus was stillborn. The female twin was liveborn and is currently healthy.

A complete autopsy was performed. Due to the D&E procedure, the head was not completely intact. Recognizable portions of brain were placed in formalin for fixation.

### Materials submitted:

1. One gross image at time of brain dissection.
2. One hematoxylin and eosin (H&E) stained digitally scanned slide.

### Questions:

1. What are the gross findings?
2. What are the major histological findings?
3. What is the differential diagnosis?
4. What further investigations are warranted to confirm the diagnosis?

### **Case 3**

Karina C. Martin<sup>1</sup>, Stephen Yip<sup>1</sup>, Ian Mackenzie<sup>1</sup>

- 1- Department of Pathology & Laboratory Medicine, University of British Columbia, Vancouver, British Columbia, Canada

#### Clinical Summary:

A 49-year-old female, with a history of iron deficiency anemia and GERD, was investigated for longstanding low back pain. MRI of the lumbar spine and sacrum revealed hyperintense lesions with surrounding bone marrow edema in the bilateral sacral ala, iliac wings, acetabuli, and right femoral neck. Extensive imaging studies were undertaken to further characterize these lesions. A 3.2 x 2.8 x 3.3 cm heterogeneously enhancing right frontal dural-based mass that was <sup>68</sup>Ga-DOTATOC avid was identified. Neurosurgery was consulted and a right frontal craniotomy and resection of tumor was performed. Intra-operative findings showed a significantly vascular right frontal dural-based tumor.

#### Materials Submitted:

1. A: H&E

#### Questions:

1. What is the differential diagnosis and which immunohistochemical stains would aid in narrowing the differential?
2. Which additional molecular tests would help confirm the diagnosis?

#### **Case 4**

Arnulf H. Koeppen, MD, VA Medical Center, Albany, New York, USA and Albany Medical College

**Case report:** A 56-year-old New York State police officer was found comatose in his patrol car in a remote area. He was admitted to a regional hospital but died in 3 days from acute renal failure. A forensic pathologist sent the brain for diagnosis

#### **Submitted material:**

1. Slide of cerebellum, stained by hematoxylin and eosin

#### **Questions:**

1. What is the diagnosis?

## Case 5

Shervin Pejhan<sup>1</sup>, Jeff Terry<sup>2</sup>, Jon Bush<sup>2</sup>, Tony Ng<sup>3</sup>, Christopher Dunham<sup>2</sup>

<sup>1</sup>University of Western Ontario

<sup>2</sup>University of British Columbia, Division of Anatomic Pathology, BC Children's Hospital (BCCH)

<sup>3</sup>University of British Columbia, Division of Anatomic Pathology, Vancouver General Hospital (VGH)

A previously well, 11-year-old boy, began developing back pain and tripping while playing basketball 3 weeks prior to surgery. Back pain and left leg weakness led to visits with a chiropractor and physiotherapy. This weakness subsequently progressed to involve both legs after 2 weeks and became so severe he required crutches and could not stand without assistance. Urinary incontinence prompted presentation to the emergency department at an outside hospital. Abdominal ultrasound, x-rays of the pelvis and abdomen, and urine studies were performed, and the patient was diagnosed with "slipped capital femoral epiphysis". An MRI was scheduled, but worsening symptoms including progressive weakness and sensory loss in both legs, as well as urinary incontinence, prompted early re-presentation to the same emergency department. An MRI at that time revealed a well circumscribed 9 x 6 x 6 mm intradural extramedullary mass anterior to the thoracic spinal cord. There was no involvement of the neural exit foramina. The mass was T1 and T2 isointense and demonstrated intense homogenous enhancement. Despite significant compression, only minimal T2 hyperintensity was detected in the adjacent spinal cord.

The patient was transferred to BCCH and underwent surgery involving a T4 laminectomy and decompression. The left T4 pedicle was resected in order to access the tumor. T3-5 posterior instrumentation and fusion was performed to stabilize the spine and a gross total resection was thought to be achieved.

Materials submitted:

1. 1 H&E-stained representative digitally scanned slide.

Questions:

1. Diagnosis?
2. Molecular alteration?
3. Any additional therapy required?

## **Case 6**

Jacob A. Houpt 1, Hao Li 1, Lee-Cyn Ang 1, David Ramsay 1, Cynthia Hawkins 2, Qi Zhang 1

1 Department of Pathology and Laboratory Medicine, London Health Sciences Centre, London, ON, Canada.

2 Hospital for Sick Children, Toronto, ON, Canada.

### Clinical Summary

A 19-month-old left-hand dominant female was found to have a mass in the left frontoparietal lobe with concern for diffuse leptomeningeal involvement for which surgical resection was undertaken. Despite also undergoing an extensive regimen of chemotherapy, a recurrence was detected along the anterior aspect of the resection cavity 3 years later. Repeat excision was undertaken followed by radiation to the tumour bed. Following the second surgery, she was left with right-sided hemiplegia and hemiparesis.

At 19 years of age (approximately 15 years later), she began experiencing multiple 3-minute episodes of sudden repetitive horizontal eye movement, breathlessness, and facial droop, occasionally preceded by light-headedness and double vision. She was diagnosed with focal epilepsy which proved to be drug-resistant. Electroencephalography identified epileptogenic foci arising from the left frontoparietal resection cavity. Prompted by this, the clinician requests a retrospective review of the pathological diagnoses.

### List of Submitted Materials:

1: HE of original left frontoparietal tumour.

### Questions for Discussion

1: What is the differential diagnosis?

2: What special stains, IHCs, or ancillary testing would be most informative?

## Case 7

Leslie E Hamilton, MD FRCPC D-ABP; Department of Pathology and Laboratory Medicine; University of Ottawa; CHEO and The Ottawa Hospital

This 4 year old girl was brought to hospital in status epilepticus (lasting ~ 45 minutes), with the history of a prodromal febrile illness. During the prodromal illness, bloodwork and blood cultures performed elsewhere had been unremarkable, and she was prescribed amoxicillin. Her medical history was also notable for B cell non-Hodgkin lymphoma, diagnosed 6 months prior. She had completed chemotherapy approximately 3 months earlier, with no evidence of residual disease at that time.

She was emergently managed and admitted to the PICU. CT imaging of the brain was normal. She was able to be extubated the following day, but within 48 hours showed significant neurologic deterioration with further seizure activity and a progressively worsening encephalopathy, with confusion, decreased level of consciousness, minimal words, and reduced right-sided movements. Initial pan-cultures and serology were negative. Initial CSF analysis was clear/colourless with 6 total nucleated cells (normal 0-5  $10^6/L$ ); and a normal glucose and total protein. CSF cytology was negative for malignancy.

Initial MR imaging performed the day after admission showed focal areas of T2/T2 FLAIR hyperintensity and edema, with restricted diffusion in the cortex of the bilateral parieto-occipital and posterior temporal lobes. Repeat MRI showed evolving findings (see Images).

A stereotactic biopsy of the left parietal region was performed.

Submitted materials:

1. MRI images
2. Scanned slides of biopsy, H&E slides x 2

Questions:

1. What are the major histopathological findings?
2. What is the differential diagnosis?
3. What ancillary studies are recommended?

## **Case 8**

Marc A. Khoury<sup>1</sup>, Julian Spears<sup>2</sup>, Robert Moreland<sup>3</sup>, David G. Munoz<sup>4</sup>

1. Institute of Medical Science, Division of Neurosurgery, University of Toronto,
2. Division of Neurosurgery, St. Michael's Hospital, Unity Health Toronto
3. Department of Medical & Diagnostic Imaging, St. Michael's Hospital, Unity Health Toronto
4. Department of Laboratory Medicine, St. Michael's Hospital, Unity Health Toronto & Department of Laboratory Medicine & Pathobiology, University of Toronto

### **Clinical Summary**

A 76-year-old male with a history of cognitive decline and coordination difficulties lasting 3-4 months presented to the hospital. Complaints of walking and bumping into objects were reported along a similar duration, likely due to left sided homonymous hemianopsia. Past medical history includes multiple system issues, including heart failure with reduced ejection fraction secondary to ischemic cardiomyopathy with CRT-D (Cardiac resynchronization therapy defibrillator) in place, hypertension, complicated left ventricular thrombus requiring lifelong apixab, pulmonary fibrosis, emphysema, T2DM with nephropathy and neuropathy. No known family history was reported. Patient also has a history of smoking, 100 packs-year as well as a past bladder carcinoma resected in 2022, with recurrence in 2015. MRI was ruled out by non-compatible device. CT scan revealed a new, right parieto-occipital solid-cystic lesion with a small (5 mm) contrast-enhancing nodule. There was associated intracranial mass effects with sulcal effacement, compression of the right lateral ventricle and millimetric leftward midline shift. Slight growth of a parafalcine extraaxial previously detected mass was identified. The right parietal lesion was resected.

### **List of Submitted Materials.**

1. CT scan image
2. H&E slide (scanned)

### **Questions for discussion**

1. Imaging differential diagnosis
2. Histopathological diagnosis
3. Pathogenesis

## Case 9

Shervin Pejhan<sup>1</sup>, Ryan Wang<sup>2</sup>, Sachin Pandey<sup>3</sup>, Joseph Megyesi<sup>2</sup>, Claire Coire<sup>4</sup>, Lee Cyn Ang<sup>1,2</sup>

<sup>1</sup> Department of Pathology & Lab Medicine, Western University, ON, Canada

<sup>2</sup> Department of Clinical Neurological Sciences, Western University, ON, Canada

<sup>3</sup> Department of Medical Imaging, Western University, ON, Canada

<sup>4</sup> Department of Pathology, Trillium Health Partners, Mississauga Hospital, ON, Canada

### Clinical Summary:

A 76-year-old gentleman presented to the hospital with nausea, and vomiting, after his diabetes medication was switched from Empagliflozin to metformin. He had also a worsening gait instability for more than a year, and a coincidental headache during that admission that ended up in a CT-head which showed a mass in the 4<sup>th</sup> ventricle without obstructive hydrocephalus. His past medical history was curious for a low-grade prostatic adenocarcinoma (stage pT1a), diagnosed by TURP more than 4 years ago, left lower lung lobectomy of unknown reason in 2011, and a spinal tumor resected in 2002. The MRI showed a T2 weighted hyperintense mass centered in the 4th ventricle (2.9 x 2.9 x 2.5 cm). The lesion demonstrated heterogeneous enhancement, with apparent solid and cystic components. He underwent suboccipital craniectomy and resection of the 4th ventricular mass.

### List of submitted materials:

1. MR images of the lesion
2. H&E-stained smear of intra-operative consult
3. H&E-stained slide of the resected tumor

### Questions for Discussion:

1. What are the differential diagnoses for this lesion?
2. What special stains, IHCs, or ancillary testing would be most informative?