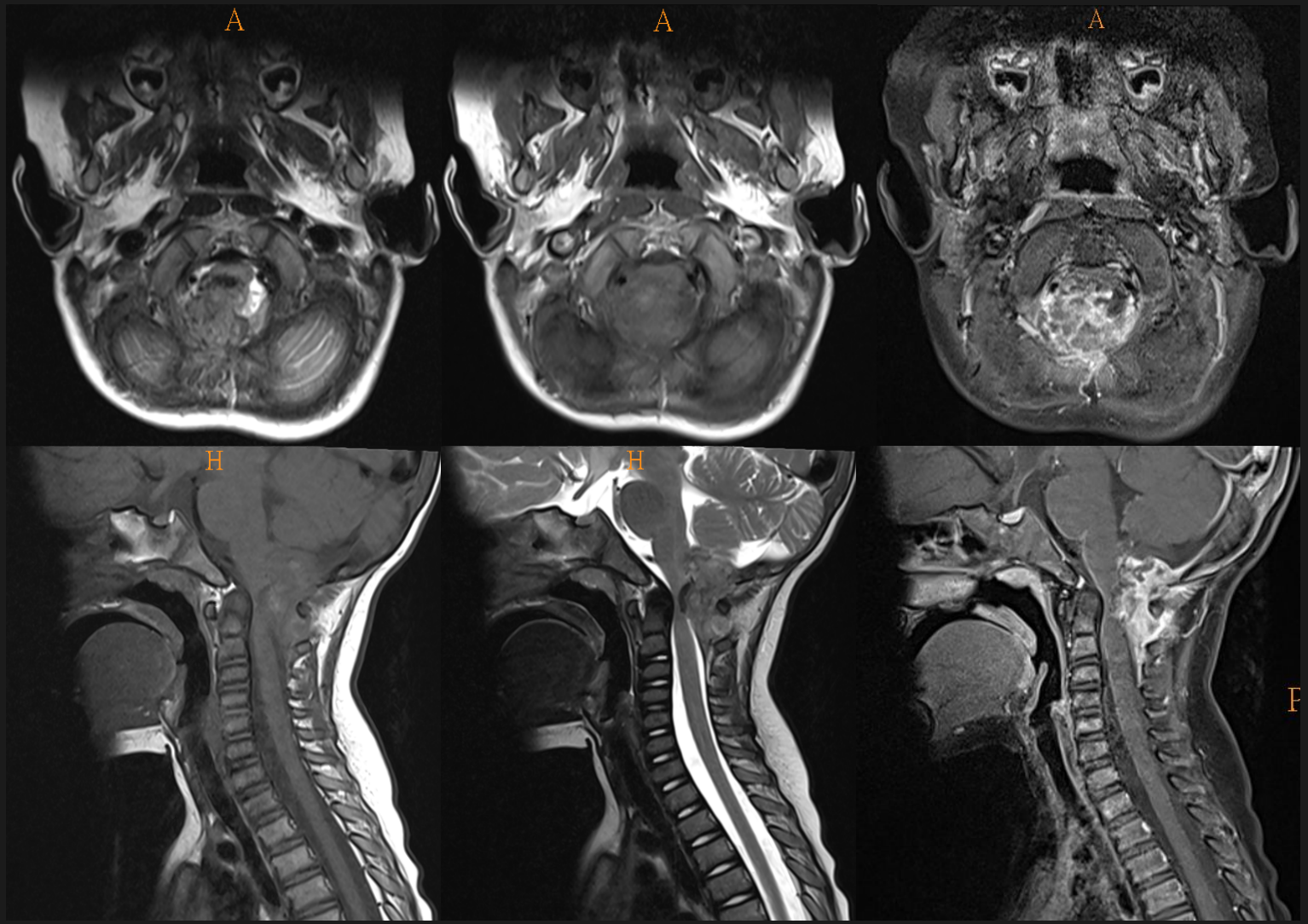
CASE 2

16453560

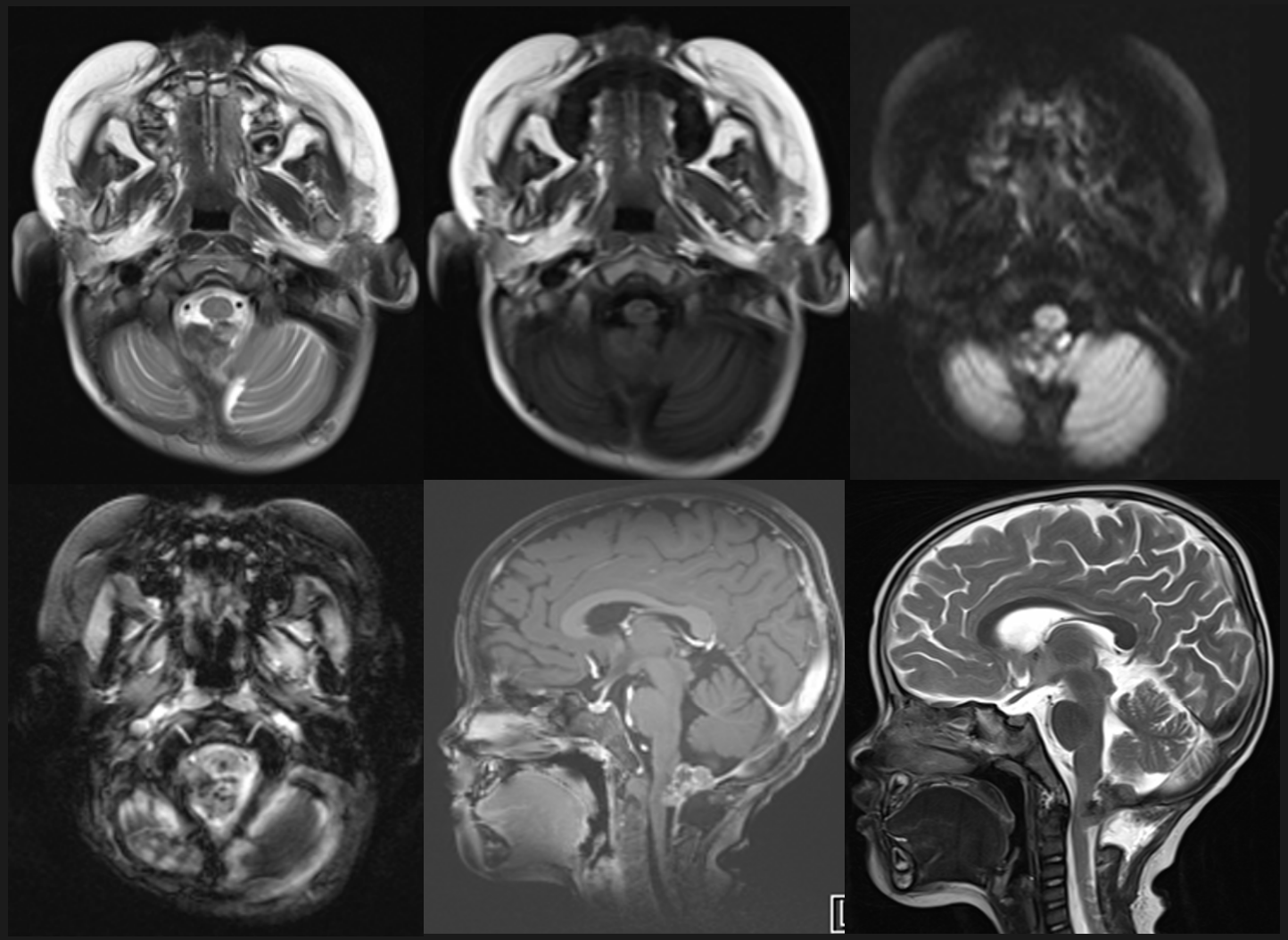
- 2-year-1-month-old, girl
- No specific past histories
- Limited range of motion of neck for 3 months
- Sudden onset of right limb weakness (especially in the right arm) at night for a few weeks
- The patient was taken to 高雄長庚H for help.
- Emergent brain MRI was performed on 2018-04-25.



- MRI (2018-04-25):
  - An irregular enhancing mass involving the posterior compartment of craniovertebral junction, result in remarkable compression to the C1 cord at the foramen magnum level.

Clinical Course

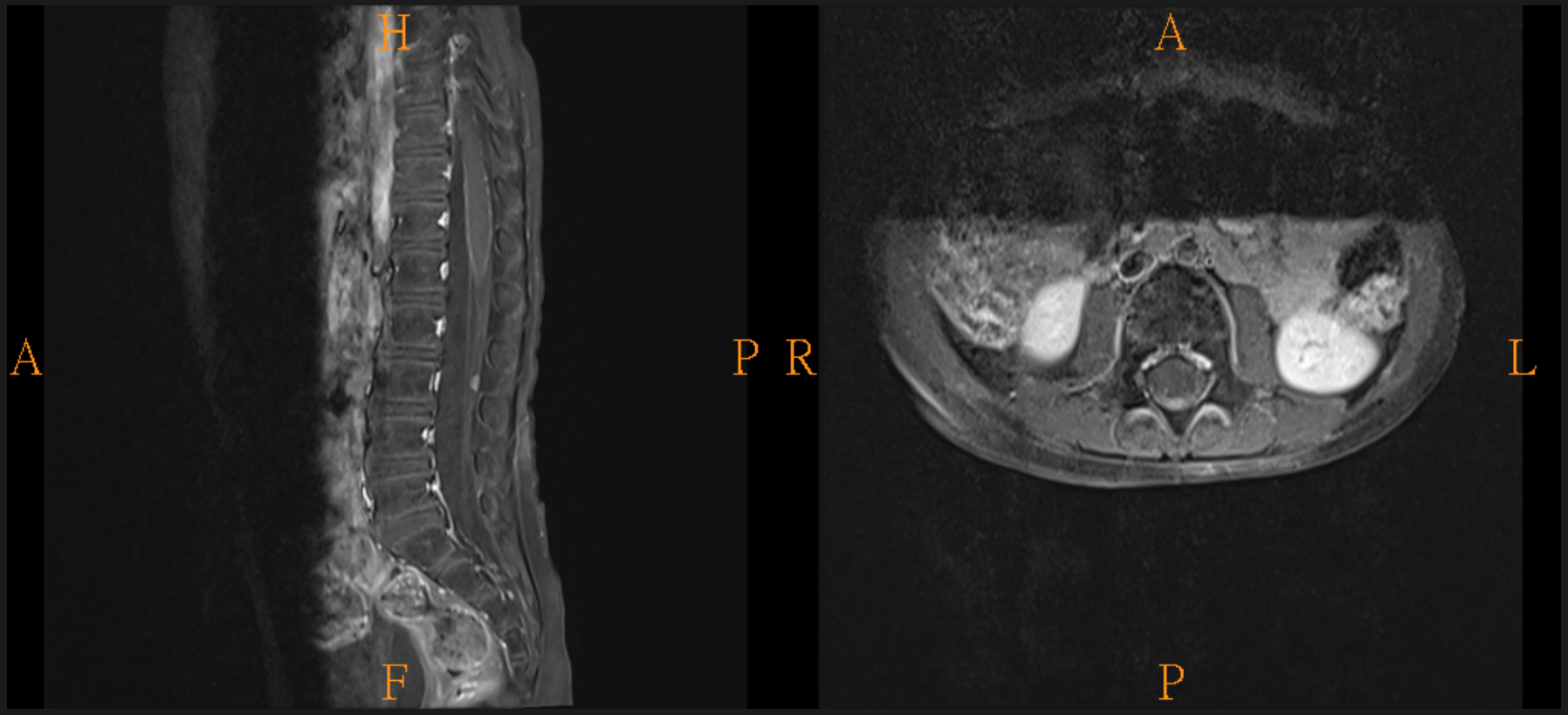
- Emergent C1 laminectomy and decompressive removal of the extradural part of the tumor was performed on the same day.
- Postoperatively, the motor weakness of right side limbs recovered.
- Follow-up MRI of brain and spine were performed on 2018-05-05.



- MRI (2018/05/05):
  - residual posterior fossa tumor extending intradurally from the foramen magnum to dorsal medulla region with marked cord compression at C1 level.

Clinical Course

- The patient was referred to our hospital for further surgical removal of tumor (2018-05-08)
- Pathology (2018-05-08): Malignant round and epithelioid cell neoplasm with bone involvement and CSF seeding
- Subsequent chemotherapy, intrathecal chemotherapy and CCRT
- Disease remains stable until 2019/04/12 (12 months)



- MRI (2019-04-12):
  - Intradural enhancing nodules are noted at cauda equina, the bony level of L3, and at the dependent portion of the thecal sac, probable CSF seeding.

Clinical Course

- Laminotomy at L2-4 level with tumoral removal and implantation of Ommaya catheter at L3/4 level (2019-04-16).
- Pathology (2019-04-16):
  - Spine/cauda equina, L3 level, laminotomy and biopsy, NUTM1-rearranged malignant round and epithelioid cell neoplasm
- Chest, abdomen to pelvis CT: no other abnormal findings
- Intrathecal chemotherapy (2019-04-22)

Differential Diagnosis

- The location of craniovertebral junction tumors is predominantly ventral, and a very small number had a lateral or a dorsal location.
  - Ventral tumors (anterior foramen magnum): meningioma, chordoma, fibrous dysplasia, aneurysmal bone cyst, osteoblastoma, and schwannoma; plexiform neurofibroma (ventral to the clivus and upper cervical spine)
  - C1/2 tumors: eosinophilic granuloma (atlas and axis vertebra), Ewing's sarcoma (lateral mass of C1)
  - Dorsal tumors (posterior foramen magnum): meningioma, ependymoma, pilocytic astrocytoma, arachnoid cyst, and medulloblastoma

Differential Diagnosis

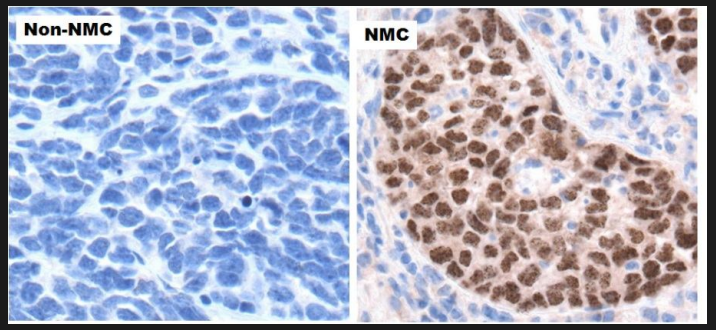
- Meningioma
  - Usually middle-aged; craniocervical region usually associated with neurofibromatosis
  - Extra-axial, dural "tails“, often along clivus
- Ewing’s sarcoma
  - Typically between 10 to 20 y/o, slight male predilection
  - Involves vertebral body before neural arch: Permeative intramedullary mass ± soft tissue mass (50%)
  - Sacrum most common spinal site
  - Heterogeneous contrast enhancement, Areas of central necrosis common

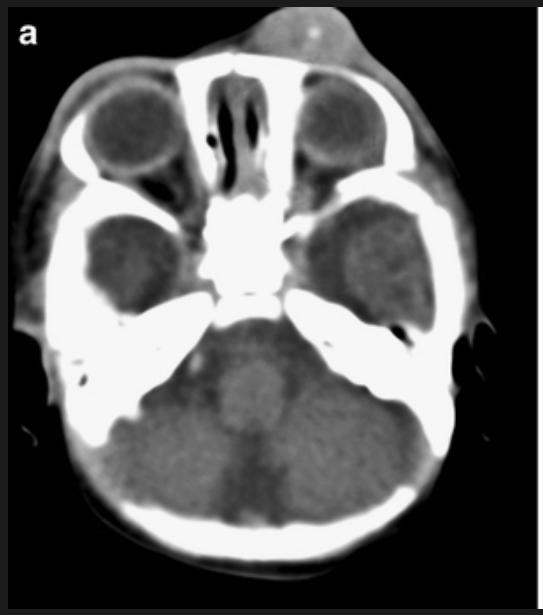
Diagnosis: NUT midline carcinoma

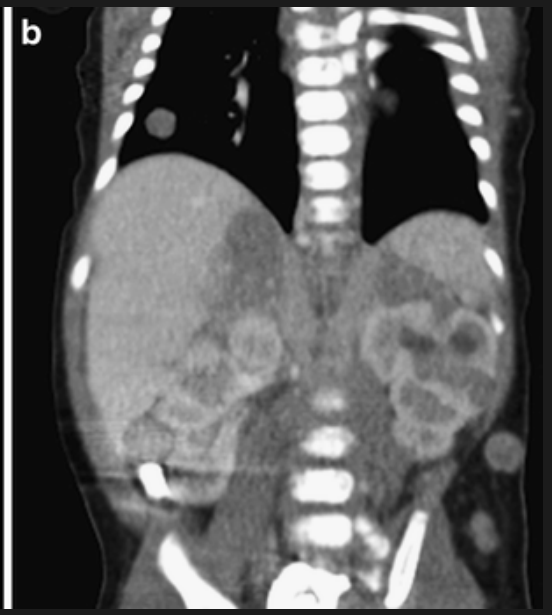
- Firstly reported in 1990 in Japan: a 22-year-old woman with presumed thymic carcinoma that was refractory to chemotherapy and radiation
- In the United States, the first case was reported in a 12-year-old girl with an epiglottic mass
- A rare and aggressive tumor (average survival 9.5 months) that has primarily been reported in children and young adult (0-78 y/o) without gender predilection

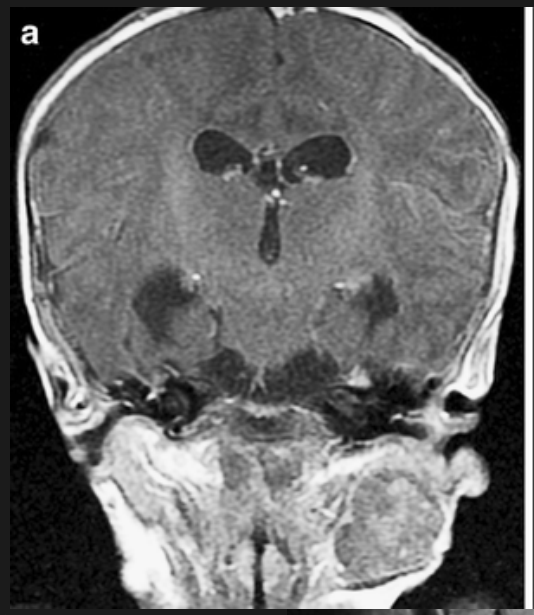
NUT midline carcinoma

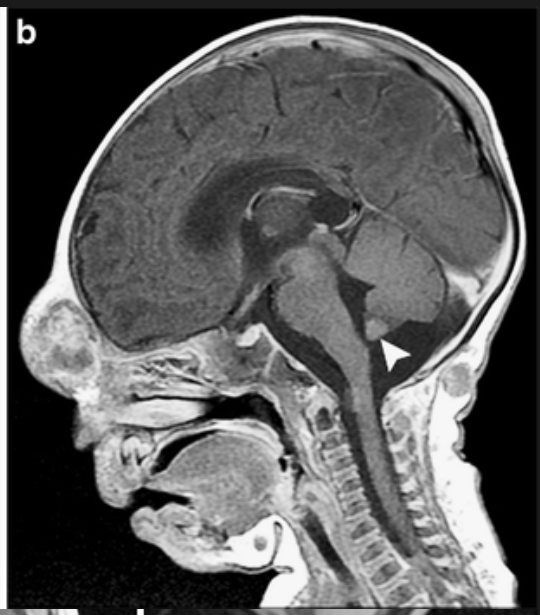
- NMC does not arise from any specific tissue type or organ. It presents as a poorly differentiated carcinoma originating from midline locations (head, neck and mediastinum)
- Rearrangement on the nuclear protein in testis (NUT) gene, located on t(15;19), resulting in BRD4-NUT oncogene expression
- Diagnosed by positive karyotyping for t(15;19) and confirmed with FISH
- Metastatic disease is common and can be extensive.



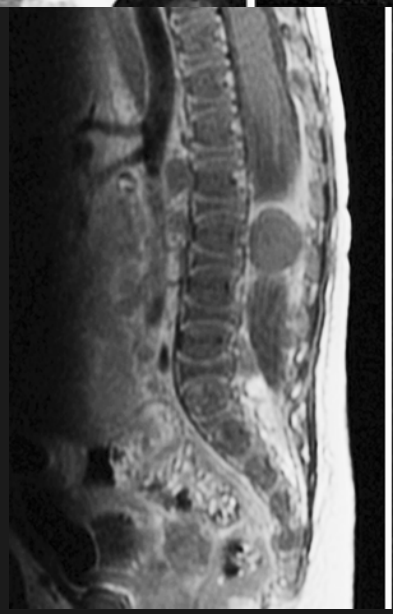


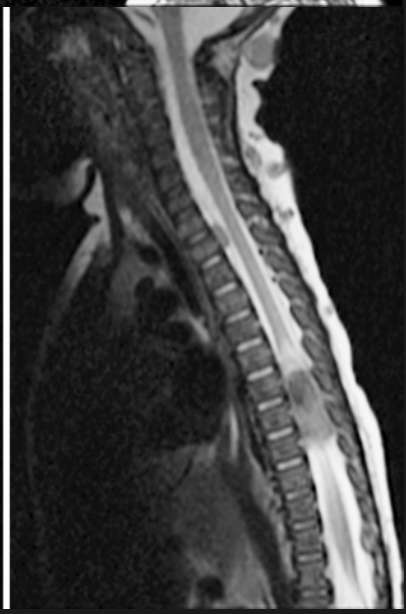




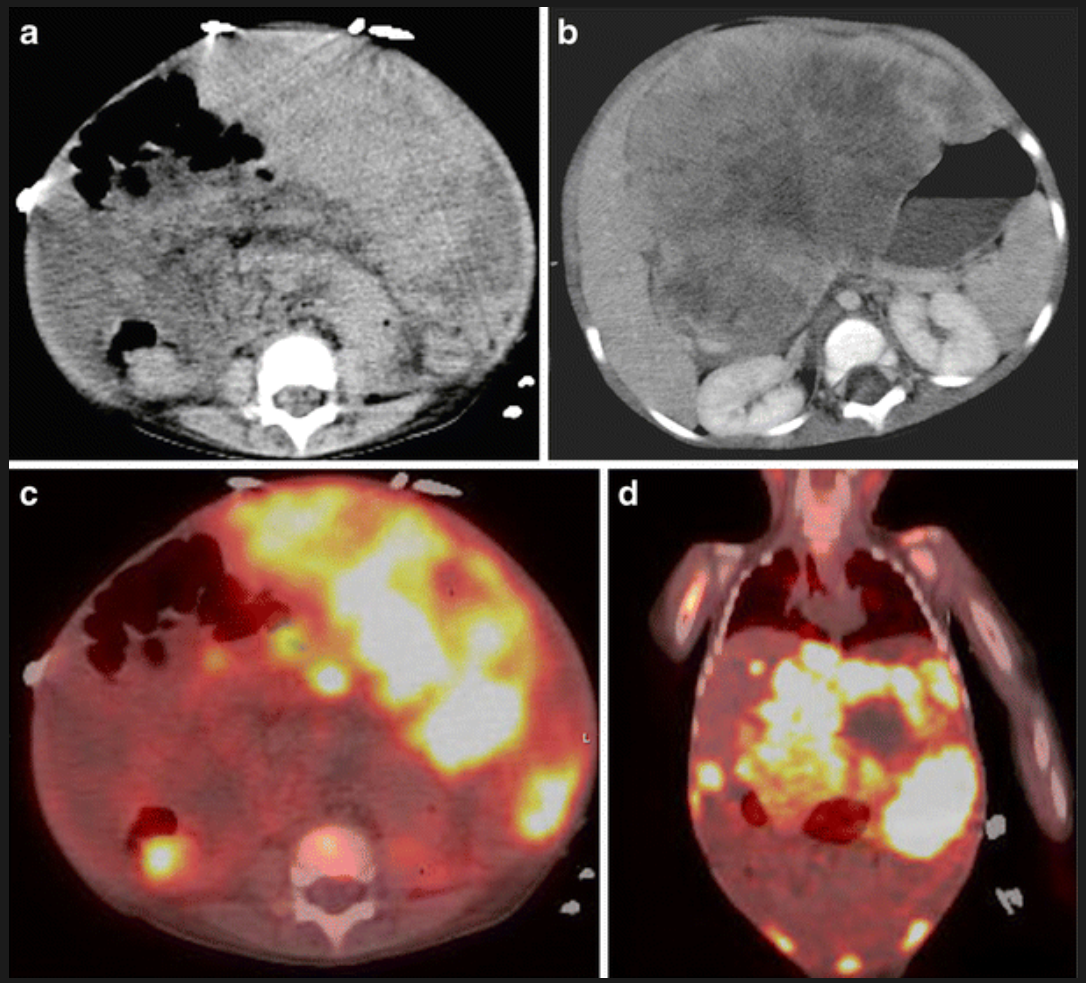


- A newborn boy presented with a rapidly progressive left supraorbital mass
- Ill-defined right suprarenal mass extending across the midline to the left suprarenal space
- A large mass in the central pelvis between the bladder and rectum
- Multiple bilateral renal cortical-based hypodensities
- Left parotid mass as well as multiple intracranial and intraspinal metastatic lesions
- DDx: metastatic stage IVs neuroblastoma? rhabdomyosarcoma or other sarcomatous lesions?
- Extensive leptomeningeal spread of disease and death soon after





Aruna Palsani, et al. NUT midline carcinoma: an imaging case series and review of literature. Pediatric Radiology (2012)



- A 2-year-old Chinese boy with a 3-week history of fevers, sweating, vomiting and abdominal distention.
- A poorly defined, large hypodense heterogeneous mass centered in the right upper quadrant, involving the left lobe of liver and the pancreatic head.
- A separate nodule in the right hepatic lobe and multiple lower lobe pulmonary nodules.
- DDx: metastatic hepatoblastoma?
- Significant progression of the pulmonary metastatic disease with large pleural effusions and subsequent death from respiratory failure

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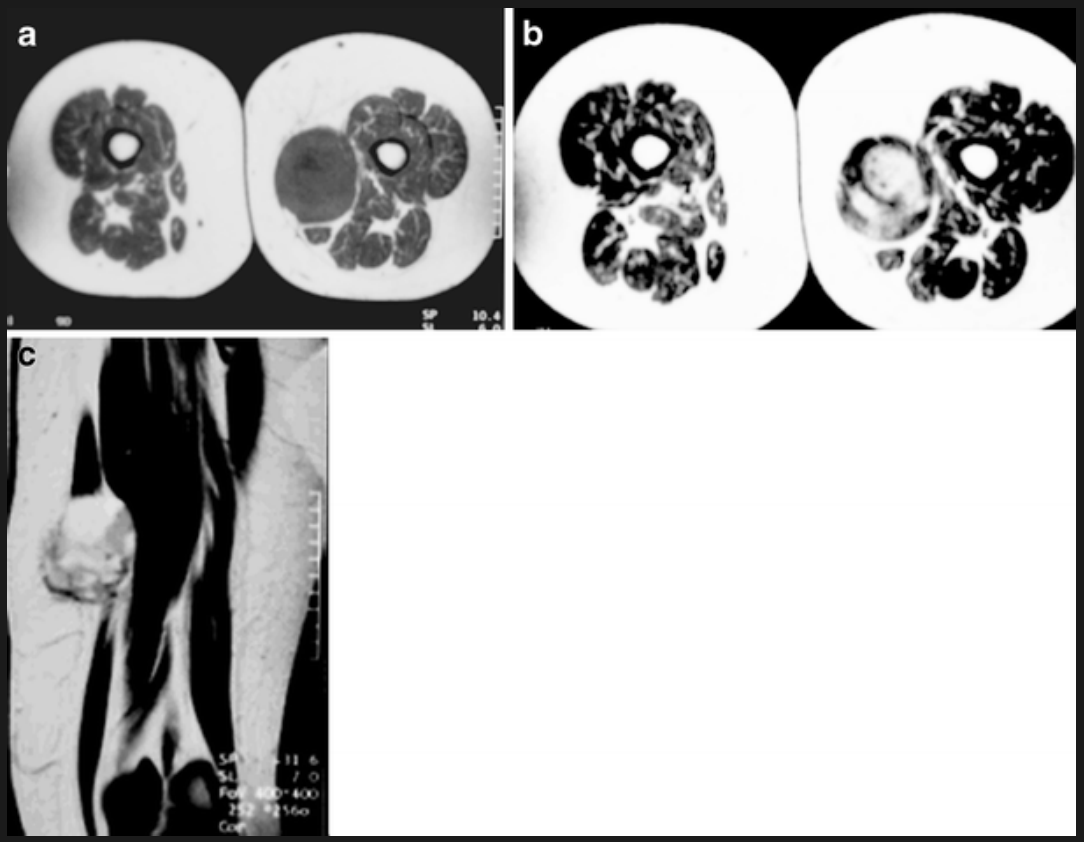
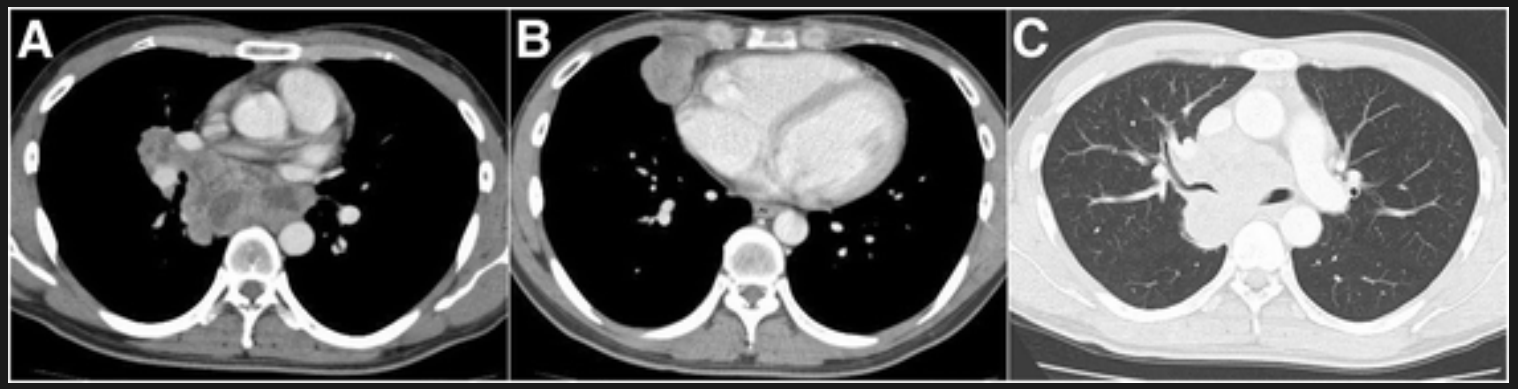
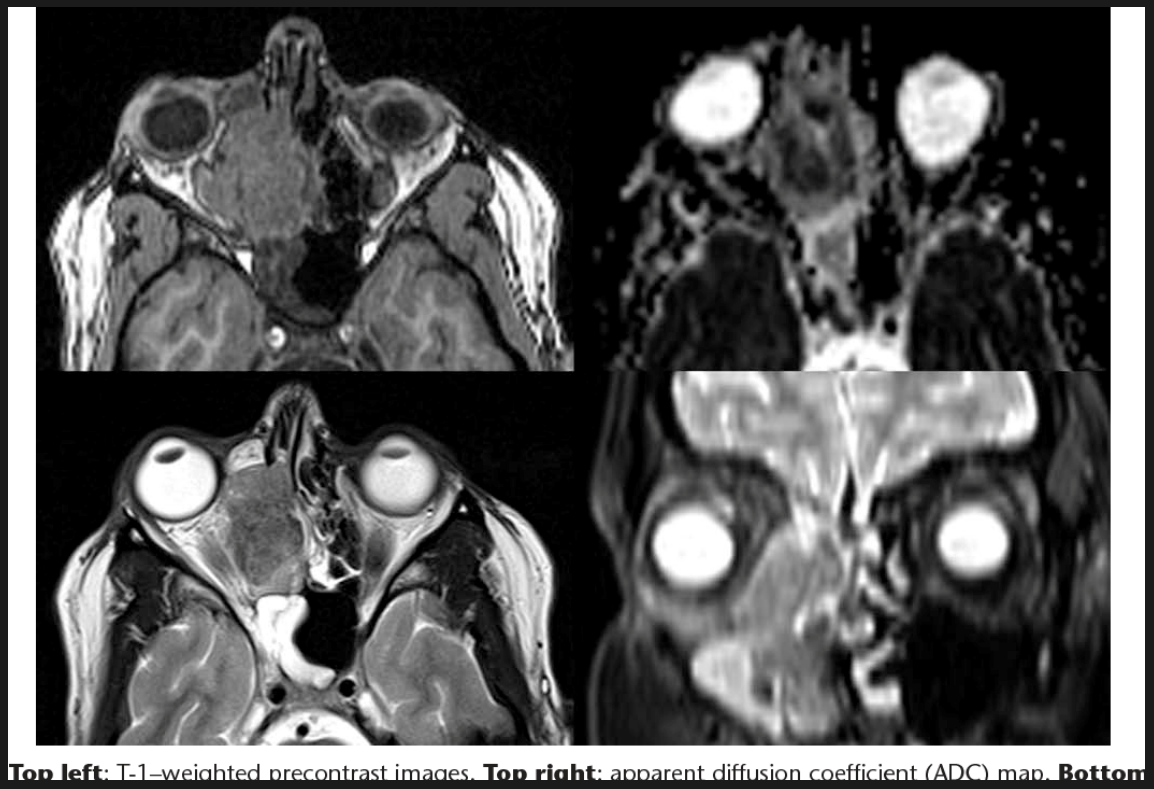


- A 4-year-old Caucasian girl with a solitary
- A T1 hypointense heterogeneously enhancing mass that was T2 hyperintense in the medial compartment of the left thigh
- Relapsed after tumoral removal and chemotherapy.
- Extensive pulmonary metastatic disease with bilateral pleural effusions and subsequent death from respiratory failure.

Aruna Palsani, et al. NUT midline carcinoma: an imaging case series and review of literature. Pediatric Radiology (2012)





Top left: T-1–weighted precontrast images, **Top right**: apparent diffusion coefficient (ADC) map, **Bottom**

Table 1 Clinical features of Nut midline carcinomas

Age	Range = 0–78 yr
Sex	14 Female; 14 Male
Location	Head and neck = 14
	Sinonasal = 6
	Nasopharynx = 2
	Larynx = 1
	Epiglottis = 1
	Orbit = 2
	Major salivary gland = 2
	Thorax = 11
	Mediastinum/Thymus = 7
	Lung = 1
	Trachea = 1
	Other = 3

Edward B. Stelow. A Review of NUT Midline Carcinoma. Head and Neck Pathol (2011)

Imaging Findings

- Intralesional necrosis is present
- The tumor can encase and infiltrate adjacent structures
- CT: heterogeneous low density
- MRI:
  - T1: heterogeneous hypointense
  - T2: heterogeneous and low-level hyperintense
  - +C: heterogeneous enhancement
  - DWI: may demonstrate restricted diffusion
- PET scan: FDG-avid

Imaging Findings

- Most striking feature: aggressive and multifocal nature with exponential interval growth of tumor
  - Diagnosis of this tumor by imaging is not likely.
  - Differential considerations usually depend on the site of the largest presenting tumor, the presumed site of origin.
- Treatment including surgery, radiation therapy, and chemotherapy
  - Combination standard cytotoxic regimens applied in other carcinomas, sarcomas, germ cell tumors, and other solid tumors
  - No clearly superior regimen