AMSER Case of the Month:

16-year-old female with history of ataxia and hyperreflexia, found to have incidental suprarenal mass

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Patient Presentation

- HPI: 16-year-old female with history of ataxia and hyperreflexia
  - No history of trauma. Negative ROS.
- PMHx: right hemiplegia, hyperreflexia, foot deformity
- PSHx: hemangioma resection (age 1), dental surgery (age 5)
- FMHx: unknown-patient was adopted from another country
- SHx: lives with adoptive parents, doing well academically in high school
- No medications
- No allergies

Physical Exam

- Vital Signs: T: 36.8C BP: 103/66 HR: 79 RR: 18 Sp02: 98% room air
- General: Alert and oriented, No acute distress.
- HEENT: PERRL, EOM intact, normocephalic
- Neck: Supple, Non-tender, No lymphadenopathy, No thyromegaly.
- Respiratory: Lungs are clear to auscultation, Respirations are non-labored.
- Cardiovascular: Regular rate, Normal rhythm, No murmur.
- Gastrointestinal: Soft, Non-tender, No organomegaly.
- Lymphatics: No lymphadenopathy.
- Musculoskeletal: no scoliosis, no varus/valgus deformity
- Integument: hypopigmented lesions possibly café-au-lait spots
- Feet: foot deformity with her toes curling down while walking.
- Neurologic: Alert, Oriented, Cranial Nerves II-XII are grossly intact, hyperreflexia
- Psychiatric: Cooperative.

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

Variant 4: **Ataxia of any acuity. No history of trauma. Suspected spinal or spinal vascular process. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
MRI spine area of interest without and with IV contrast	Usually Appropriate	O
MRI spine area of interest without IV contrast	Usually Appropriate	O
MRA spine area of interest with IV contrast	May Be Appropriate	O
CTA spine area of interest with IV contrast	May Be Appropriate	Varies
MRA spine area of interest without IV contrast	May Be Appropriate	O
Arteriography spine area of interest	May Be Appropriate	Varies
CT myelography spine area of interest	May Be Appropriate	Varies
CT spine area of interest with IV contrast	May Be Appropriate	Varies
CT spine area of interest without IV contrast	May Be Appropriate	Varies
CT spine area of interest without and with IV contrast	Usually Not Appropriate	Varies
Radiography spine area of interest	Usually Not Appropriate	Varies

MRI spine with and without contrast was ordered by the pediatric neurologist

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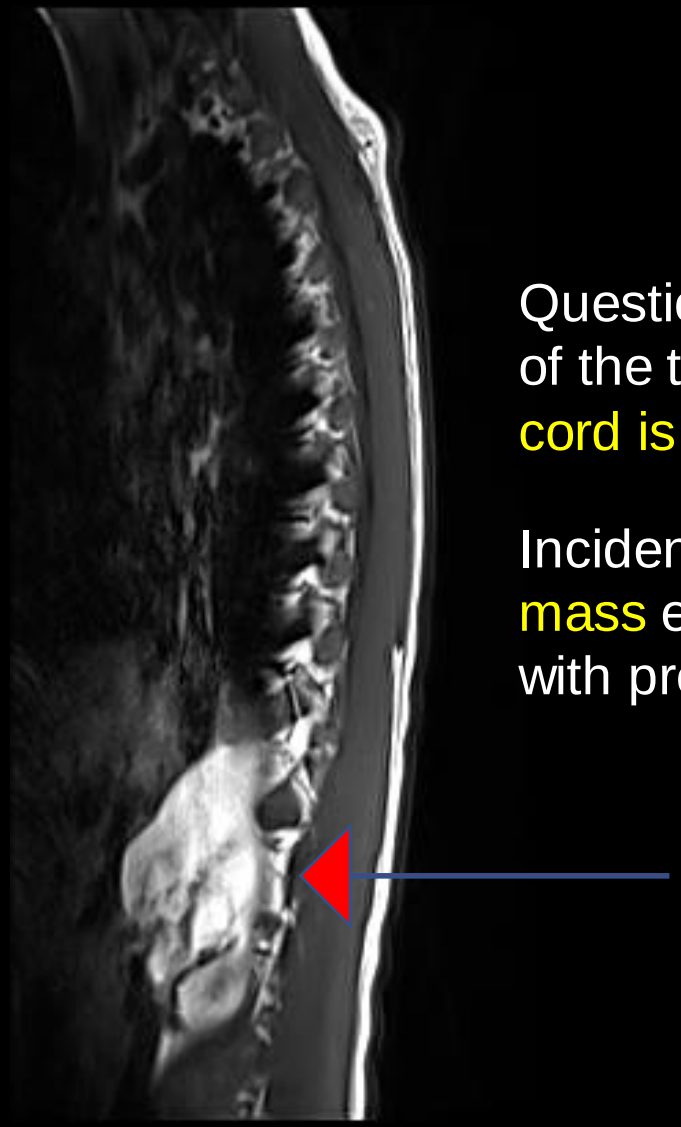
MRI Spine (unlabeled)

MRI Spine (labeled)

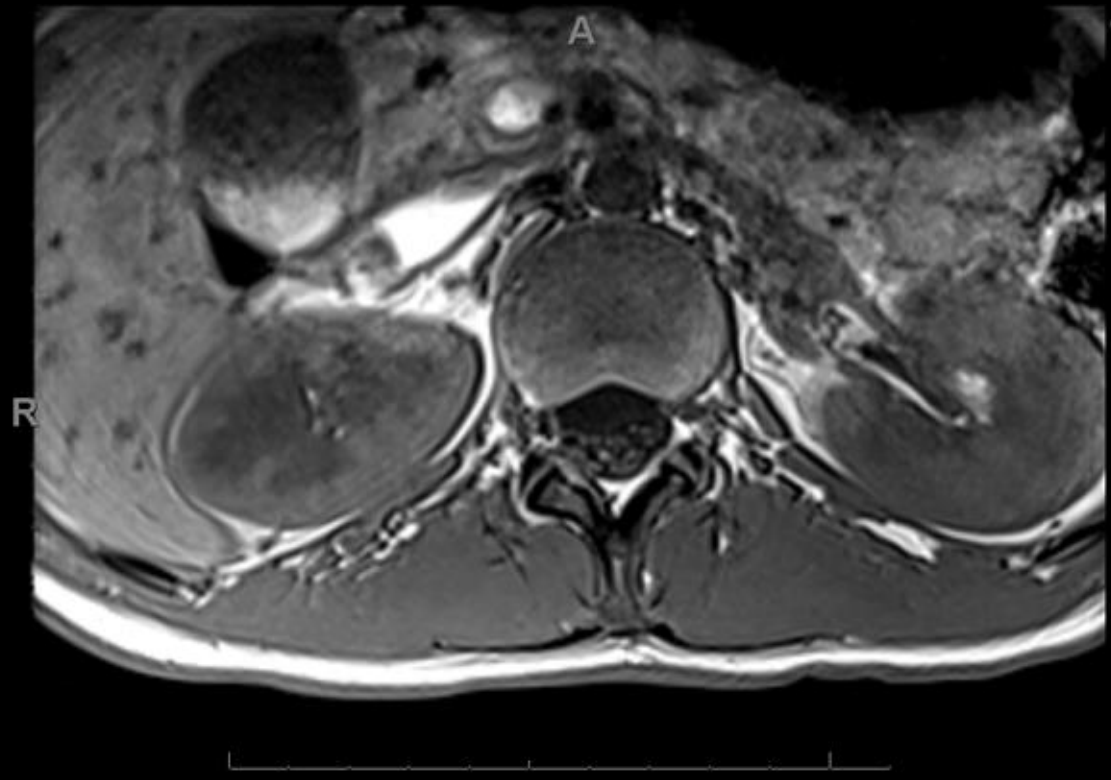


Questionable mild subjective thinning of the thoracic cord, otherwise spinal cord is unremarkable.

Incidental finding of right suprarenal mass estimated 9.3 x 7.0 x 3.2 cm with preponderance of fat

MRI Spine (unlabeled)

MRI Axial T2



MRI Axial T1

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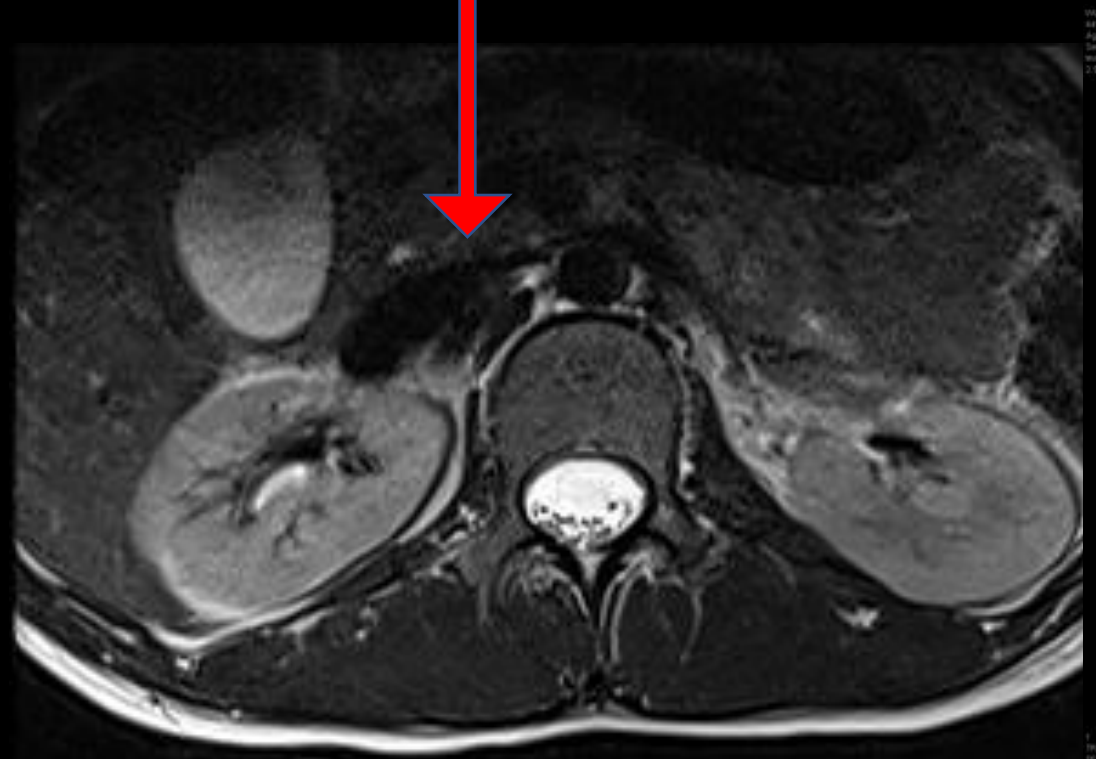
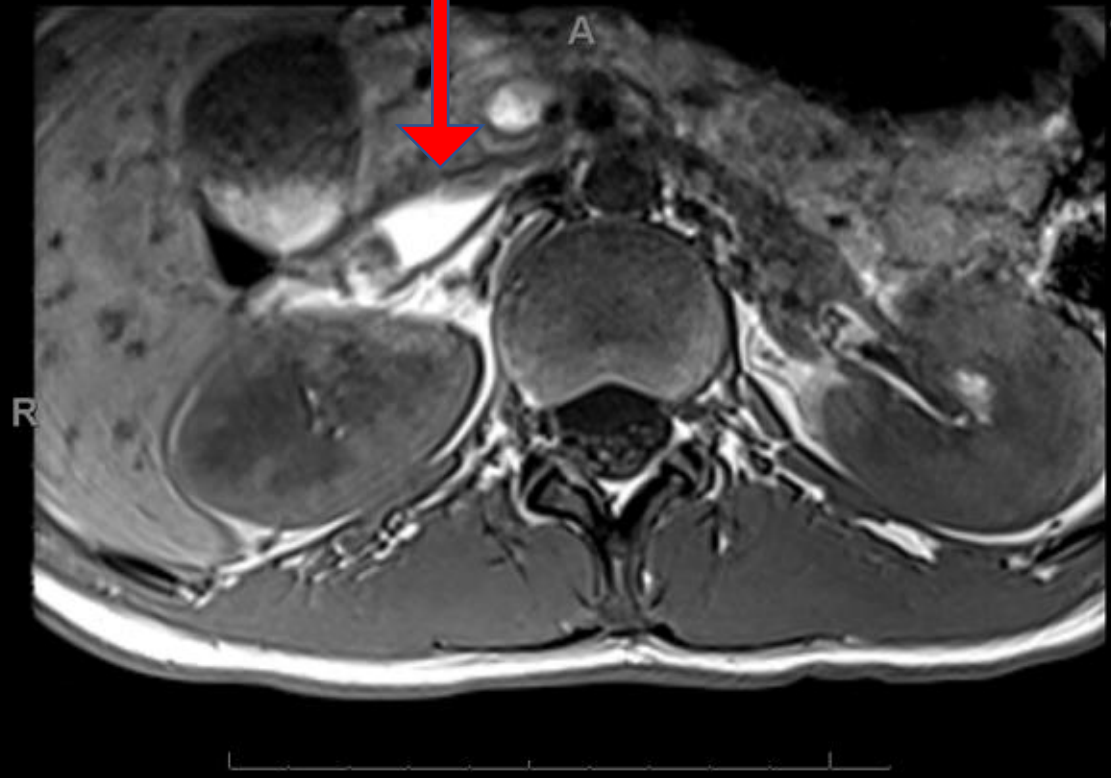
MRI Spine (labeled)

Right suprarenal mass estimated 9.3 x 7 x 3.2 cm in maximal representative dimensions. Majority of mass follows fat signal characteristics.



MRI Axial T2



MRI Axial T1

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Now what?

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

Variant 3: **Soft-tissue mass. Nondiagnostic initial evaluation (ultrasound and/or radiograph). Next imaging study.**

Procedure	Appropriateness Category	Relative Radiation Level
MRI area of interest without and with IV contrast	Usually Appropriate	O
MRI area of interest without IV contrast	Usually Appropriate	O
CT area of interest with IV contrast	May Be Appropriate (Disagreement)	Varies
CT area of interest without IV contrast	May Be Appropriate	Varies
CT area of interest without and with IV contrast	Usually Not Appropriate	Varies
FDG-PET/CT area of interest	Usually Not Appropriate	☢☢☢☢

MRI already done

CT abdomen and pelvis with and without contrast was ordered by the pediatric neurologist

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CT abdomen and pelvis (unlabeled)

CT abdomen and pelvis (labeled)

Right suprarenal retroperitoneal mass, heterogenous with significant internal fat density, linear and nodular areas of soft tissue density, and coarse internal calcifications.

This lesion displaces the right adrenal gland laterally and the right kidney inferiorly.

CT abdomen and pelvis (unlabeled)

CT abdomen and pelvis (labeled)

Differential Diagnoses

- Teratoma
  - Most likely given presence of elements from all 3 embryological layers
- Adrenal myelolipoma
- Ganglioneuroma
- Neuroblastoma
- Pheochromocytoma
- Adrenal cortical tumor
- Adrenal myelolipoma

Workup

- Pertinent labs
  - HCG <5 (nml <5)
  - CA-125 14 (nml <35)
  - VMA 4.1 (nml <8.2)
  - AFP 1.4 (nml <8.8)
  - CA 19-9 14 (nml <35)
  - Metanephrines 47 (nml <88)
  - LD 155 (nml <360)
- Seen by multiple consultants
  - The patient's ataxia and hyperreflexia were ultimately considered unrelated to the tumor

Specialist	Pertinent Findings	Assessment/Plan
Orthopedics	Normal hip and leg radiographs Toe hyperflexion, hip circumduction	Friedreich's ataxia or Charcot-Marie-Tooth. Consult PM&R, if no improvement consider EMG.
Endocrinology	Normal plasma metanephrines etc.	No further endo workup needed
Pediatric Surgery	Normal HCG/AFP (teratoma tumor markers) and catecholamines	Open surgical resection due to size of mass, send for pathology

Final Diagnosis:

Mature teratoma.

Pathology report:

"Mature teratoma with prominent glioneuronal proliferation. Various components including a glioneuronal infiltrate with associated fibrosis, ependyma, meningothelium, cutaneous adnexal structures, bone, and cartilage. Overall the findings are those of a teratoma with mature elements."

Case Discussion: Germ Cell Tumors

- Teratoma is the most common type of germ cell tumor (GCT)
  - Neoplasm derived from germ cells
  - Can be benign (mature) or malignant (immature)
  - Commonly grow in the gonads (i.e. ovaries and testes)
  - Tumor markers include HCG, AFP, LDH

- Extra-gonadal GCTs (EGGCTs) can occur anywhere
  - Most commonly brain, mediastinum, and retroperitoneum
  - Incredibly rare
  - Epidemiology ~2 in 1 million (1.8-3.4 per 1 million)
  - Bimodal age distribution: <4 years to age 20-40s

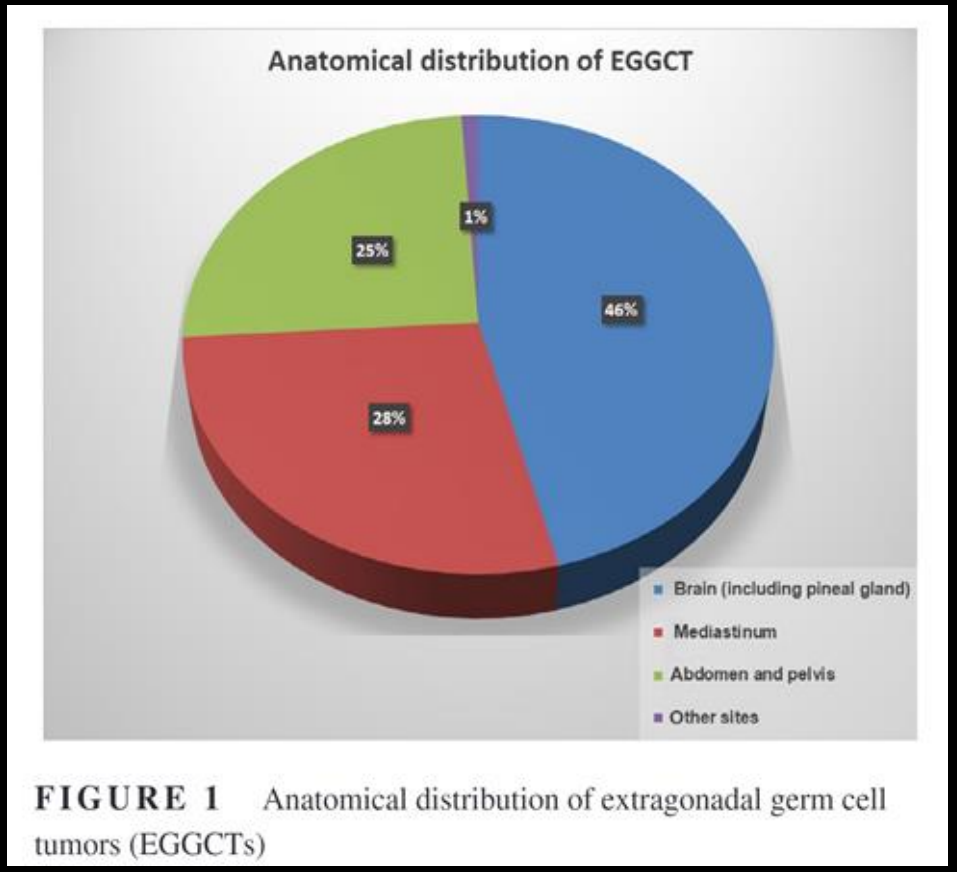

FIGURE 1 Anatomical distribution of extragonadal germ cell tumors (EGGCTs)

Ronchi et al., 2019

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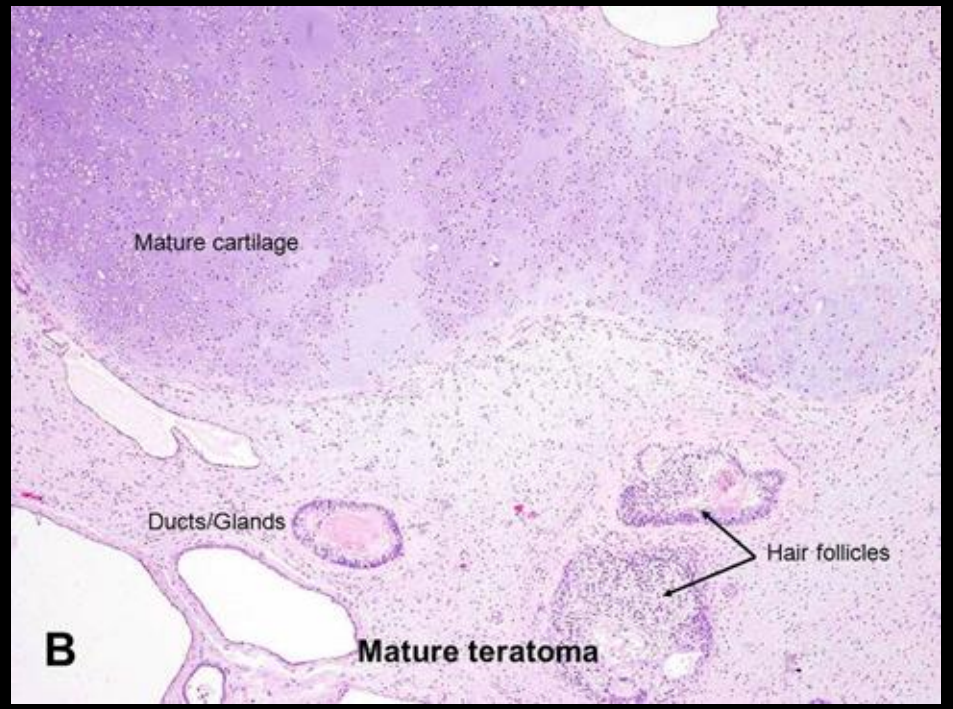
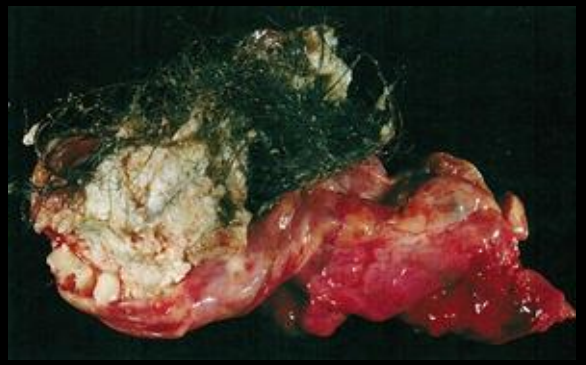
Case Discussion: Matue Teratoma

- Very little research exists on extra-gonadal teratomas
- Pathophysiology of extragonadal GCTs poorly understood
  - Hypothesis: migration defect during embryological development

- Histology: contains 3 embryologic layers
  - Ectoderm – skin, hair, teeth, neuronal tissue
  - Endoderm – GI, solid organs
  - Mesoderm – muscle, cartilage, bones

- Pathology: solid or cystic, greasy with keratin and teeth
  - Rokitansky's protuberance often seen - area of projection





Case Discussion: Teratoma Treatment

- May be clinically asymptomatic or symptomatic
  - Symptoms may include abdominal or pelvic pain, fertility issues, etc.
- Diagnosis involves imaging and labs
- Prognosis is typically good for *mature* teratomas
  - Depends on histology, location, age, and other factors
  - Malignant transformation occurs in 0.2-2% of cases
- Treatment involves surgery and/or chemotherapy
  - Surgery – ovary sparing vs. non-sparing
  - Chemotherapy – PEB combo of cisplatin, etoposide, and bleomyc

Summary

- A 16 y/o female with ataxia and hyperreflexia was found to have an incidental suprarenal mass on spinal MRI
- The soft tissue, fat, and calcified components on imaging (CT and MRI) suggest teratoma
- Teratomas are the most common type of germ cell tumor (GCT)
- Extragonadal GCTs are extremely rare
  - In this patient's case, the teratoma was located in the right suprarenal space in the retroperitoneum
- Treatment involves surgery

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