An unusual intrathoracic mass in an adolescent with CKD

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Case Overview

16 y/o young male presented with a three-month history of intermittent chest pain, heartburn, dysphagia, and post prandial non bloody, non bilious vomiting.

Referred to Cardiology

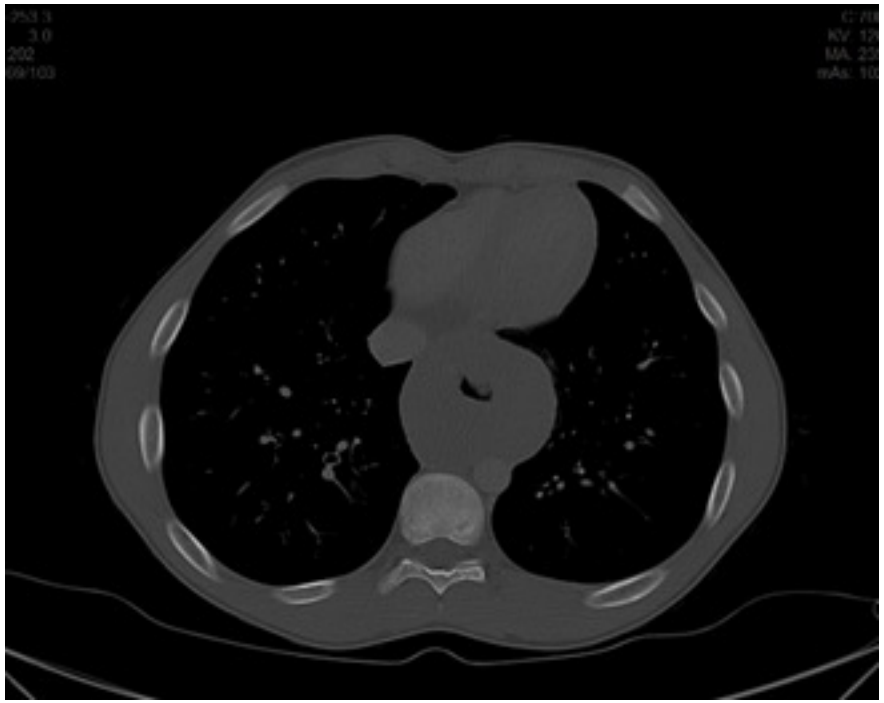
BP: 134/80 mmHg

EKG: Prolonged QT interval and left ventricular hypertrophy.

Echo: A mass posterior to the left ventricle with mild compression on the posterior wall of the ventricle.

Started on Nadolol
Chest CT w contrast

Case Overview – Outpatient CT Scan



The CT scan demonstrated a circumferential distal esophageal mass causing severe narrowing of the esophagus.

Referred to the Hospital

Case Overview

Review of Systems

The patient denied any headaches, tinnitus, epistaxis, edema, gross hematuria or orthopnea.

Family History

MGF with throat cancer in his 30s - died. Two maternal great aunts died of cancer - ovarian and breast. PGF died of colon cancer. Mom with a benign eye tumor that required surgery.

No known family history of kidney disease.

Physical Examination

BP: **144/97 mmHg** HR: 68 bpm
Temp:37. 1C RR: 16 rpm SpO2: 100%

CV: RRR, no murmurs

Resp: Clear to auscultation

Abd: Soft, No hepatomegaly, No ascites

Extremities: No swelling

Plan for Admission to Heme/Onc Service

Case Overview – Initial Labs

Chemistry	
Sodium	141
Potassium	4.5
Chloride	111
CO2	**18**
Glucose	85
BUN	**64**
Creatinine	**8.69**
Calcium	**6.6**
Phosphorus	**7.2**
Albumin	3.3

Urinalysis	
UA Blood	2+
UA Protein	200
Casts	None
UA RBC	**22**
UPCR	3.03

Nephrology Consult

What is your differential diagnosis?

Differential diagnosis

- Rhabdomyosarcoma
- Adenocarcinoma
- Squamous Cell Carcinoma
- Chloroma
- Leiomyoma
- Lymphoma
- Glomerular disorder
- Nephronophthisis
- Renal dysplasia
- ATN

What initial studies would you obtain?

Workup

CBC	
WBC	8.9
Hgb	9.2
Hct	27.6
Plt	391

Chemistry	
Sodium	140
Potassium	4.2
Chloride	112
CO2	**16**
Glucose	91
BUN	**62**
Creatinine	**8.29**
Calcium	**6.9**
Ionized ca	**3.59**
Phosphorus	**7.3**
Albumin	**2.8**

UA	
Sg	**1.010**
PH	**6.5**
Protein	**200**
Blood	**+1**
RBC	**16**
WBC	**3**

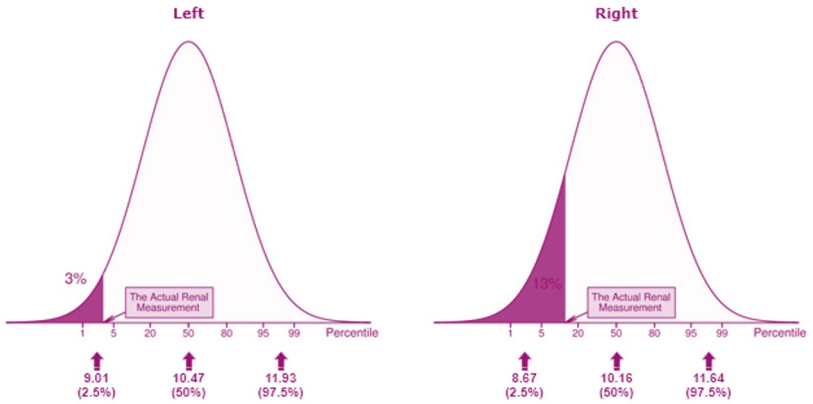
Endocrinology	
PTH	**1122.5**

Immunology	
ESR	**33**

Renal US

Renal ultrasound

- Right kidney length is 9.1 cm.
- Left kidney is 9.3 cm.
- Left kidney appears echogenic.
- Bladder was normal.



Esophageal Mass Biopsy

Esophageal Mass Biopsy

- Smooth muscle tumor, consistent with **leiomyoma**.
- Distal and Middle Esophagus:
  Mild esophagitis
- Proximal Esophagus:
  Acute esophagitis with ulceration.
  HSV, CMV, adenovirus negative.

Differential diagnosis

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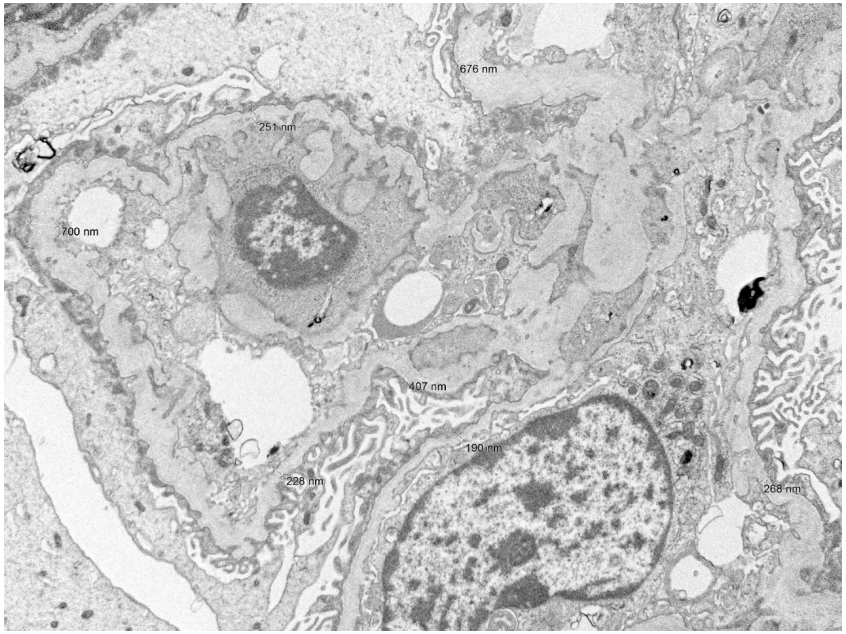
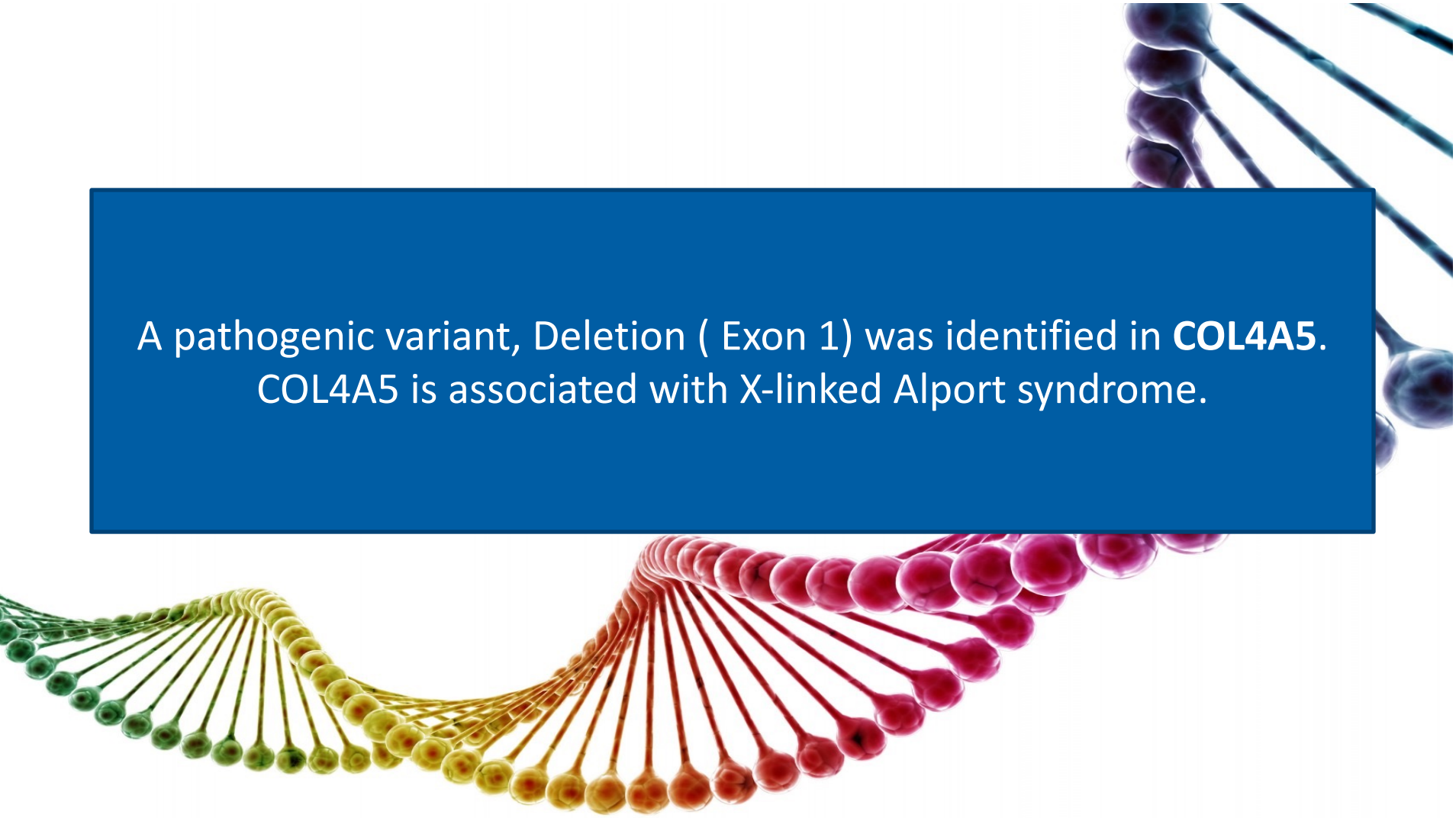
Kidney Biopsy

- ESKD with **78% globally sclerosed glomeruli** (7/9)
- **Severe** interstitial fibrosis and tubular atrophy, 70% each.
- Acute tubular injury
- Nephrocalcinosis
- Mild arteriolar sclerosis
- **Abnormal EM findings suggestive of a type IV collagen abnormality.**



Genetic Test



A pathogenic variant, Deletion (Exon 1) was identified in **COL4A5**. COL4A5 is associated with X-linked Alport syndrome.

X-linked Alport Syndrome with diffuse leiomyomatosis

X-linked Alport Syndrome with diffuse leiomyomatosis

- **Alport syndrome** is characterized by kidney disease with microscopic hematuria, hearing loss, and ocular abnormalities.
- It is mainly caused by a mutation of the gene-encoding **type IV collagen** in the glomerular basement membrane.
- More than 1000 different mutations in the **COL4A3, COL4A4, and COL4A5 genes** have been found in patients with Alport Syndrome.

Diffuse leiomyomatosis

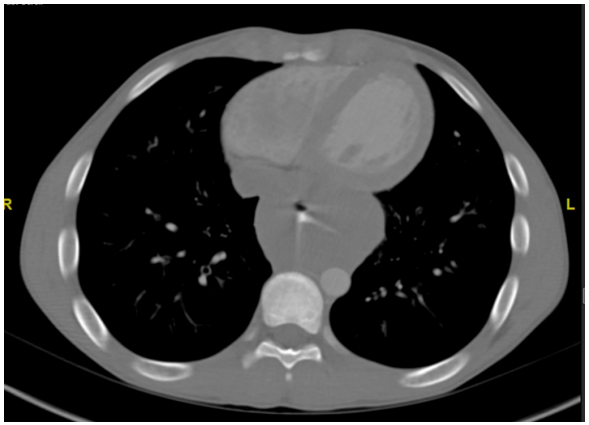
- **Benign smooth muscle** tumor characterized by abnormal proliferation of well differentiated smooth cells.
  - Gastrointestinal tract
  - Respiratory tract
  - Female reproductive tract
- The genetic defect associated to diffuse leiomyomatosis and Alport syndrome consists of **deletion mutations** involving the 5’ ends of the **COL4A5 and COL4A6** genes on the X chromosome.

Back to our patient …

Case Overview

- The patient underwent peritoneal dialysis catheter placement and **peritoneal dialysis** was initiated given the advanced CKD state.

- **Pediatric Surgery** and **Pediatric Oncology Team** managed his esophageal lesion.



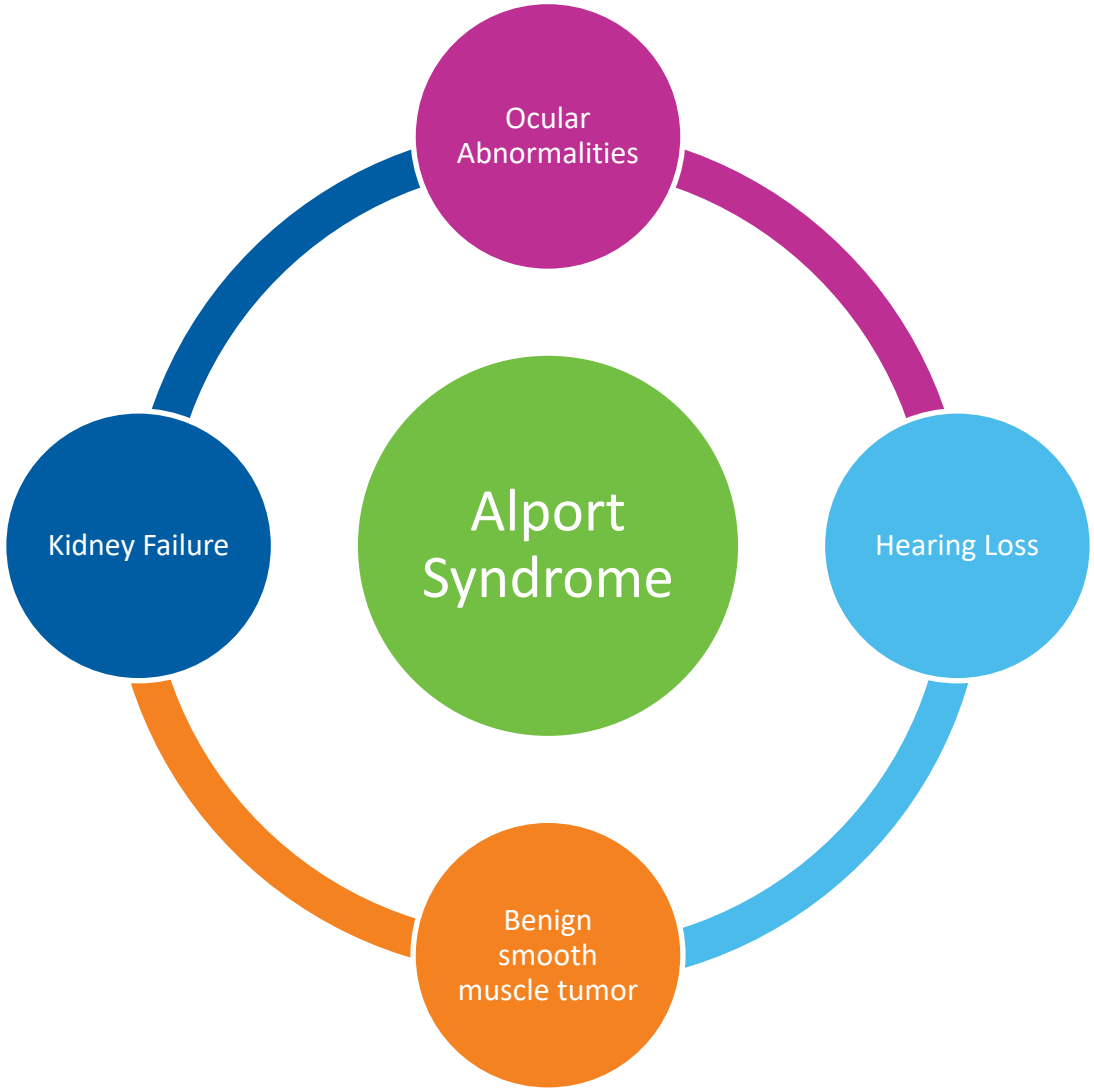
Esophagram

Significant (greater than 75%) luminal narrowing and irregularity distal esophagus secondary to known esophageal mass.

Midthoracic esophagus markedly dilated with altered esophageal motility.

- He underwent exploratory laparotomy with **esophagectomy** and gastric pull through.

Pearls


Ocular Abnormalities
Kidney Failure
Alport Syndrome
Hearing Loss
Benign smooth muscle tumor

Pearls

- This case highlights the importance of recognizing **rare presentations** of Alport syndrome.
- Diffuse leiomyomatosis results from deletions associated with **5' ends** of **the COLA4A5 and COL4A6** genes.
- It is a **benign** condition characterized by the overgrowth of **visceral smooth muscles** in the esophagus, gastrointestinal tract, and female reproductive tract.

Pearls

- Diffuse esophageal leiomyomatosis (DEL) usually presents with **dysphagia**, **emesis**, and **gastroesophageal reflux**.

- Patients may experience **aspiration** of esophageal and gastric content resulting in **respiratory manifestations**.

Acknowledgments

- Dr. Chinnadurai
- Dr. Reyes
- ADC Planning Committee

References

- Nozu, K., Minamikawa, S., Yamada, S., Oka, M., Yanagita, M., Morisada, N., Fujinaga, S., Nagano, C., Gotoh, Y., Takahashi, E., Morishita, T., Yamamura, T., Ninchoji, T., Kaito, H., Morioka, I., Nakanishi, K., Vorechovsky, I., & Iijima, K. (2017). Characterization of contiguous gene deletions in COL4A6 and COL4A5 in Alport syndrome-diffuse leiomyomatosis. Journal of human genetics, 62(7), 733–735. https://doi.org/10.1038/jhg.2017.28
- Garcia-Torres, R., Cruz, D., Orozco, L., Heidet, L., & Gubler, M. C. (2000). Alport syndrome and diffuse leiomyomatosis. Clinical aspects, pathology, molecular biology and extracellular matrix studies. A synthesis. Nephrologie, 21(1), 9–12.
- Komatsu H, Goda T, Nozu KDe novo X-linked Alport syndrome in a 3-year-old girlBMJ Case Reports CP 2019;12:e230183

Questions ?