A Rare Tumor

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Case 1

• 43F c LUQ pain, stabbing, worse with mov’t
• No nausea/vomiting/constipation.
• PMHx: asthma, uterine fibroids
• PSHx: caesarian section x 2
• FHx: younger sister with breast cancer
Physical Examination

• 97.4  104/56  62  BMI 38

• S1/S2 RR

• Equal breath sounds b/l

• Abd: obese, soft, mild LUQ tenderness

• CBC  5.6>11.2/37.6<363

• BMP  137/4/105/23/5/0.66<72
Hospital Course

- Hematology & Surgery consults
- Discharged home. F/u in clinic – mild abd pain
- IR biopsy – inconclusive
- Admitted for surgical resection vs open biopsy
Preoperative Planning

- Vascular surgery
- GU consultation
- Bowel prep
Intraoperative Findings

- Small amount murky peritoneal fluid.
- Mesenteric mass involving branch of SMA
- Small bowel resection
Postoperative Course

- Transferred to floor and kept NPO
- Clear diet on POD#3
- Discharged home on POD#7
- F/up in clinic

- PATHOLOGY: *lymphangioleiomyoma*
  - 20 X 17 X 7.5 cm
Case 2

- 19M s/p scrotal trauma c pain/bleeding
- Incidental finding of right groin swelling
- Mass present since birth
- PMHx: none
- PSHx: none
- Tobacco + marijuana smoker
- Review of Systems: 20 lb weight loss
Operative Course

- Patient underwent incisional biopsy
- CBC 6.6>10.2/33.2<151
- 138/4/106/26/20/0.76<91
Intraoperative Findings
Postoperative Course

• Discharged home on POD#1 with dressing
• POD#10 returned to UHB for drainage
• Re-educated about wound care mgmt

• PATH: *lymphangioma vs Kaposi’s Sarcoma*
  – 6 x 3 x 2 cm
Lymphangioleiomyomatosis (LAM): very rare
1 in 1,000,000 women
Systemic disease, though considered benign
Affects women in reproductive age
Pulmonary system most affected
LAM Subtypes

• Spontaneous LAM
  – Worldwide prevalence 1 in 1 million
  – More significant pulmonary disease

• Tuberous sclerosis complex-LAM
  – Associated with mutation in TSC-2 gene
  – Cases of children and males described
Histology

• Smooth muscle
  – Trabecular
  – Fascicular
  – Papillary

• Immunohisto stains
  – α smooth muscle actin
  – Vimentin
  – Desmin
  – HMB-45
Extrapulmonary LAM

• Exceedingly rare

• Clinical features:
  – Abdominal mass
  – Pain
  – Chylous ascites

• Mainly located in retroperitoneum, pelvis

• Only four cases reported in mesentery
Clinical Presentation

• Dyspnea on exertion
• Pneumothorax, often recurrent
• Chylous pleural effusion
• Gradual obstructive disease
Diagnostic Studies

- CXR offers little
- High-resolution chest CT
- CT abdomen/pelvis
- Rule out TSC, “Epiloila”
Disease Progression

• Life expectancy variable: 91-92% alive @ 10yrs

• Poor prognostic factors
  – Dyspnea on presentation (47% 10 yr survival)
  – ↓FEV1/FVC
  – Increase in percentage of predicted TLC
  – Predominantly cystic LAM
Treatment

- Anti-estrogen/Progesterone supplementation
- Gonadotropin-releasing hormone agonists
- Oophorectomy
- Lung transplantation
Novel Treatments

• Doxycycline (antiangiogenic/Abx properties)
• Aromatase inhibitors
• mTOR inhibitors (sirolimus)
  – mTOR pathway activation → uncontrolled prolif.
MILES Trial

• “Multicenter Int’l LAM Efficacy of Sirolimus”
• Double-blind, randomized, multi-center
• Two stages: 12 months on tx, 12 mos off
• N= 89
• In tx group, improved FVC, FRC, quality of life
• Lung function deteriorated after stopping tx
I’m a Surgeon...
What Do I Need to Know?

• Higher risk of pneumothorax in OR
• Lymphangioleiomyomas may rupture, bleed
• Sirolimus can impair wound healing
• Future CT fellows... may need lung transplant!
Summary

• Rare disease affecting young women
• Dyspnea, pneumothorax: MC presentation
• Extrapulmonary dz may pre-date pulmonary
• Progressive deterioration of respiratory fxn
• Hormone therapy & mTOR inhibitors
• May require lung transplantation
3rd Worldwide LAM Awareness Day

Saturday June 1st 2013

Lymphangioleiomyomatosis (LAM) is a rare disease which is only found in women. The condition mainly affects the lungs which are progressively destroyed over a number of years. This leads to increasing breathlessness and lung collapse and can be fatal. Although clinical trials of promising treatments are in progress, there is no cure for LAM and for many patients lung transplantation is the only means of survival.

Saturday June 1st 2013 has been designated as the 3rd Worldwide LAM Awareness Day by the WLPC (Worldwide LAM Patient Coalition). Please see the links on the right for details of your national organization.
References

- Hancock E et al. Lymphangioleiomyomatosis: a review of the literature. Respiratory Medicine 2002 (96) 1-6
- Jun YJ et al. Extrapulmonary lymphangioleiomyoma presenting as a mesenteric mass. Basic and Applied Pathology 2011 (4) 63-65
TSC and LAM

- Tuberous sclerosis: chromosome 9, **hamartin**
- LAM: chromosome 16, **tuberin**
- Proteins for cell growth via mTOR pathway