

Multiple Cutaneous and Uterine Leiomyomatosis

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Clinical Case



36 y.o. healthy female

One year ago:

 sudden onset of at least 20 lesions over back, shoulders and upper arms

Mildly tender when rubbed

ROS: non-contributory

PMH: No skin cancers

PSH: Hysterectomy age 28 for multiple leiomyomas

Physical Exam

- 4-12mm pink-brown dermal papules
 - Mildly tender with palpation
 - No increase in erythema when rubbed

DDX:

- Leiomyomas
- Mastocytomas
- Neurofibromas
- Metastasis
- Adnexal tumors



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Cutaneous Leiomyomas

- Arise from smooth muscle in arrector pili, genitalia, breast or vasculature
- Equal sex distribution
- Brown-pink dermal papules or nodules
 - Multiple lesions may follow Blaschko's lines
 - Favor trunk
- Pseudo-Darier's:
 - Contraction of muscle when rubbed





•Pain: spontaneous or when exposed to cold •Unclear mechanism



Diagnosis: *Multiple Cutaneous and Uterine Leiomyomatosis*

Associations:

- Multiple Leiomyomatosis
- Uterine Fibroids
- Reed's Syndrome
- Family history of early aggressive Renal Cell Ca

Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC)

HLRCC

- 1954: Kloepfer et al. heritable predisposition to develop multiple cutaneous leiomyomas
 - 1973: Reed "multiple cutaneous and uterine leiomyomatosis"
- Autosomal Dominant
- 2002: Fumarate Hydratase
 - Kreb's cycle enzyme converts fumarate to malate
 - Thought to act as a tumor suppressor, but exact mechanism of tumorigenesis is unknown

CO.

FADH

HLRCC

Cutaneous Leiomyomas:

- 76% multiple or single
 40% 5 or fewer lesions
- Mean age: 25 (range: 10-47)

Uterine Fibroids:

- Almost 100% females
- Numerous/large
- Mean age: 30 (range: 18-52)
- Undergo symptomatic hysterectomy than general population

Uterine leiomyosarcoma:

•Unclear association, No families in North America reported

HLRCC

Renal Cell Carcinoma:

- ♦ 10-16%
- Median age of detection: 44
- Unilateral, solitary
 - Most: "type 2 papillary"
 - tubulo-papillary, collecting-duct carcinoma
 - <u>Aggressive</u>: often present with metastatic disease at time of diagnosis





roid





Surveillance and Treatment

Uterine Fibroids:

- Gynecology evaluation annually
 - Assess severity of fibroids
 - Evaluate for changes suggestive of leiomyosarcoma
- Most require surgery earlier than general population



How to know when your gynecologist is watching too much baseball.

Surveillance and Treatment

Renal Cell Carcinoma:

- Early detection and surgical intervention
 - Consider total nephrectomy for detectable renal masses
- Baseline CT with contrast or MRI
 - Repeat every 2 years if normal
 - PET-CT to ID metabolically active lesions
 - U/S alone may not be sufficient
- Urologic oncology surgery evaluation of tumors









Thank you!

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