## When to think about hereditary RCC

Patients with any renal tumour (benign or malignant) AND any one of the following:

- Bilaterality or multifocality
- Onset of ≤50 years of age
- 1st or 2nd degree relative with any renal tumour
- Pneumothorax (BHD/FLCN)

### Dermatologic findings:
- Skin leiomyomas (HLRCC/FH)
- Skin fibrofolliculomas/trichodiscomas (BHD/FLCN)

### Associated tumours:
- Pheochromocytoma/paraganglioma (VHL, SDH)
- Hemangioblastoma of the retina, brainstem, cerebellum or spinal cord (VHL)
- Early onset of multiple uterine fibroids (<30 years of age) (HLRCC/FH)
- Lymphangiomyomatosis (TSC)
- Childhood seizure disorder (TSC)

### ASSOCIATED CLINICAL FINDINGS

### SYNDROME / GENE / (RCC HISTOLOGY)

#### Von Hippel-Lindau

- **VHL (ccRCC)**
- Annual imaging alternating between US and MRI
- Annual ophthalmologic examination
- Annual 24-hour urine catecholamines/melanoph thriller
- Annual audiometry
- MRI of brain and spine every 2y

#### Tuberous sclerosis complex

- **TSC1/TSC2 (Angiomyolipoma, RCC)**
- MRI of abdomen every 1–3y
- MRI of brain every 1–3y (age ≥25 y) ± EEG
- CT of chest for women every 5–10y or symptomatic males
- Annual dermatologic examination
- Dental exam every 6mo
- Annual ophthalmologic examination
- ECG every 1–3y

#### PTEN hamartoma syndrome/ Cowden syndrome

- **PTEN (various)**
- All patients:
  - US of abdomen every 1–2y (age ≥40y)
  - Annual US of thyroid
  - Colonoscopy every 5y (age ≥35y)
  - Dermatologic examination
  - Women:
    - Clinical breast exam every 6–12mo (age ≥25y)
    - Annual mammography and MRI of breast (age ≥30y)
    - Annual random endometrial biopsies and/or transvaginal US (age ≥30y)

#### Hereditary papillary RCC (HPRCC)

- **MET (Papillary type I)**
- Annual MRI of abdomen

#### Hereditary leiomyomatosis and RCC (HLRCC)

- **FH (Papillary type II)**
- Annual MRI of abdomen
- Dermatologic examinations every 1–2y
- Annual gynecologic assessment and transvaginal US

#### SDH-associated kidney cancer

- **SDH (ccRCC, chromophobe, oncocytoma)**
- Annual MRI of abdomen and MRI of head and neck
- Annual 24-hour urine catecholamines/melanoph thrillers, and plasma melanoph thrillers

#### Birt-Hogg-Dubé (BHD)

- **FLCN (Oncocytoma, mixed oncocytic, chromophobe)**
- Annual CT or MRI of abdomen or every 3y in persons without renal lesions
- Baseline CT of chest
- Annual dermatologic examination

#### Uveal melanoma, melanoma, mesothelioma

- **BAP1 Hereditary Cancer Syndrome**

#### Li-Fraumeni Syndrome

- **TP53 (various)**
- US of abdomen every 6mo
- Annual rapid full body MRI
- Annual MRI of brain
- CBC, LDH, ESR every 6mo

### SUGGESTED SURVEILLANCE IN ADULTS

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- **MET (Papillary type I)**
- Annual MRI of abdomen

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- **FH (Papillary type II)**
- Annual MRI of abdomen
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### Reference:

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### 2015

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**Monthly Renal Genetics Rounds**

3rd Thursday Monthly GI Clinic North,
Submit cases to Rehab Chahin
rehab.chahin@uhn.ca

**Referral**

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