When to think about hereditary RCC

Patients with non-clear cell RCC with unusual associated features, such as:

- Papillary type I
- Papillary type II
- Chromophobe, oncocytoma, oncocytic hybrid

Patients with or without RCC, who report a family member with a known clinical or genetic diagnosis of any one of the syndromes in the table below;

Patients with RCC and history of a second cancer suggestive of any one of the syndromes below:

ASSOCIATED CLINICAL FINDINGS

**Von Hippel-Lindau**
FIH (ccRCC)
- Annual imaging alternating between US and MRI
- Annual ophthalmologic examination
- Annual 24-hour urine catecholamines/metanephrines, plasma metanephrines
- 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
  - Clinical breast exam every 6–12mo (age ≥35y)
  - Annual mammography and MRI of breast (age ≥30y)
  - Annual random endometrial biopsies and/or transvaginal US (age ≥30y)

**Hereditary papillary RCC (HPRC)**
MET (Papillary type I)
- Annual MRI of abdomen

**Hereditary leiomyomatosis and RCC (HLRCC)**
FIH (Papillary type II)
- Annual MRI of abdomen
- Annual MRI of head and neck
- Annual 24-hour urine catecholamines/metanephrines, and plasma metanephrines

**Paraganglioma, pheochromocytoma, GI stromal tumors**

**SDH-associated kidney cancer**
SDH (ccRCC, chromophobe, oncocytoma)
- Annual CT or MRI of abdomen
- Annual gynecologic assessment and transvaginal US

**Birt-Hogg-Dubé (BHD)**
FLCN (Oncocytoma, mixed oncocytic, chromophobe)
- Not established: consider annual abdominal imaging
- Annual dermatologic assessment
- Annual ophthalmologic assessment

**Uveal melanoma, melanoma, mesothelioma**

**BAP1 Hereditary Cancer Syndrome**
BAP1 (ccRCC)
- US of abdomen every 6mo
- Annual rapid full body MRI
- Annual MRI of brain
- CBC, LDH, ESR every 6mo

**Li-Fraumeni Syndrome**
TP53 (various)
- Annual mammography and MRI of breast
- Colonoscopy every 2–5y
- Annual dermatologic examinations

**Reference:** Dr. Raymond Kim MD/PhD Medical Genetics
Princess Margaret Cancer Centre
Referral: Dr. Raymond Kim MD/PhD
Dr. Michael Jewett
Dr. Raymond Kim
UHN

**Authors:**
Nicole S. Kim
Dr. Michael Jewett
Dr. Raymond Kim
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