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Red Flags to identify patients with renal tumors most likely to benefit from referral to genetics

Reproduced and adapted with permission from the Canadian Urological Association. Reaume, *et al.,* 2013. Canadian guideline on genetic screening for hereditary renal cell cancers. *Can Urol Assoc J.* 7(9-10):319-23

Patients with any renal tumour (benign or malignant) **AND** any one of the following: a. Bilaterality or multifocality b. Early age of onset (≤45 years of age) c. 1st or 2nd degree relative with any renal tumour d. A history of pneumothorax* e. One of the following dermatologic findings: i. Skin leiomyomas* ii. Skin fibrofolliculomas/ trichodiscomas* f. One of the following associated tumours: i. Pheochromocytoma/ paraganglioma* ii. Hemangioblastoma of the retina, brainstem, cerebellum or spinal cord* iii. Early onset of multiple uterine fibroids (<30 years of age)* g. Lymphangiomyomatosis* h. Childhood seizure disorder* **or 1st degree relative with same*	Patients with non-clear cell carcinoma with unusual associated features (e.g., chromophobe, oncocytic or hybrid tumours)	Patients, with or without RCC, who report a family member (any) with a known **clinical or laboratory diagnosis** of any one of the following genetic conditions: a. Von Hippel-Lindau syndrome b. Birt-Hogg-Dubé syndrome c. Hereditary leiomyomatosis and renal cell cancer d. Hereditary papillary renal cell cancer e. Hereditary paraganglioma/ pheochromocytoma f. Tuberous sclerosis

RCC – Renal Cell Cancer

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Brief summary of some RCC hereditary syndromes described in Reaume *et al.*, 2013

Genetic testing is available for all of the conditions listed. All conditions have variable expressivity [*variation in clinical features (type and severity) of a genetic disorder between affected individuals, even within the same family*.]

Condition	**Von Hippel-Lindau syndrome**	**Birt-Hogg-Dubé syndrome**	**Hereditary leiomyomatosis and renal cell cancer**	**Hereditary papillary renal cell cancer**	**Hereditary paraganglioma/ pheochromocytoma**	**Tuberous sclerosis Complex**
Inheritance	Autosomal dominant	Autosomal dominant	Autosomal dominant	Autosomal dominant	Autosomal dominant	Autosomal dominant
# of individuals with new (de novo) mutations / no family history	20%	Unknown because a sufficient number of parents has not been evaluated for subtle manifestation	Unknown because a sufficient number of parents has not been evaluated for subtle manifestation	-	unknown	66%
Histology	Clear cell RCC	Chromophobe RCC/oncocytic RCC	Papillary type 2 RCC	Papillary type 1 RCC	Clear cell RCC	Epithelial (various) or mesenchymal (angiomyo-lipoma)
Features	— Renal tumours — CNS hemangioblastomas — Retinal hemangiomas — Adrenal pheochromocytoma/ paraganglioma — Pancreatic neuroendocrine tumours — Endolymphatic sac tumours — Epididymal cystadenomas — Broad-ligament tumours	— Skin fibrofolliculomas — Pulmonary cysts — Renal tumours	— Skin leiomyomas — Renal tumours — Uterine leiomyomas	Renal only	Adrenal pheochromocytoma / paraganglioma	— Skin (adenoma sebaceum, shagreen spots) — Retinal hamartomas — CNS lesions (including tubers) — Cardiac lesions — Renal tumours — Teeth/gum lesions — Bone cysts