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Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

BRIEF SUMMARY OF SOME RENAL CELL CARCINOMA HEREDITARY SYNDROMES (1/2)

Genetic testing is available for all six of the conditions listed.

All conditions have variable expressivity [variation in clinical presentation (onset, features and severity) even within the same family].

Renal Cell Carcinoma abbreviated to RCC

Reviewed Jan 2023

VON HIPPEL-LINDAU SYNDROME

- Autosomal dominant
- # of individuals with no family history / new (de novo) pathogenic/likely pathogenic gene variants = 20%
- Histology: Clear cell RCC
- Features:
  - Renal tumours
  - Central nervous system hemangioblastomas
  - Retinal hemangiomas
  - Adrenal pheochromocytoma/ paraganglioma
  - Pancreatic neuroendocrine tumours
  - Endolymphatic sac tumours
  - Epididymal cystadenomas
  - Broad-ligament tumours

BIRT-HOGG-DUBÉ SYNDROME

- Autosomal dominant
- # of individuals with no family history / new (de novo) pathogenic/likely pathogenic gene variants = Unknown
- Histology: Chromophobe RCC/oncocytic RCC
- Features:
  - Skin fibrofolliculomas
  - Pulmonary cysts
  - Renal tumours

HEREDITARY LEIOMYOMATOSIS AND RENAL CELL CANCER

- Autosomal dominant
- # of individuals with no family history / new (de novo) pathogenic/likely pathogenic gene variants = Unknown
- Histology: Papillary type 2 RCC
- Features:
  - Skin leiomyomas
  - Renal tumours
  - Uterine leiomyomas

Reaume, et al., 2013. Canadian guideline on genetic screening for hereditary renal cell cancers. Can Urol Assoc J. 7(9-10):319-23

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POINT OF CARE TOOL

BRIEF SUMMARY OF SOME RENAL CELL CARCINOMA HEREDITARY SYNDROMES (2/2)

Genetic testing is available for all six of the conditions listed.

All conditions have variable expressivity [variation in clinical presentation (onset, features and severity) even within the same family].

Renal Cell Carcinoma abbreviated to RCC

Reviewed Jan 2023

HEREDITARY PAPILLARY RENAL CELL CANCER

- Autosomal dominant
- # of individuals with no family history/new (de novo) pathogenic/likely pathogenic gene variants = Unknown
- Histology: Papillary type 1 RCC
- Features: Renal only

HEREDITARY PARAGANGLIOMA/PHEOCHROMOCYTOMA

- Autosomal dominant
- # of individuals with no family history/new (de novo) pathogenic/likely pathogenic gene variants = Unknown because a sufficient number of patients have not been evaluated for subtle manifestation
- Histology: Clear cell RCC
- Features: Adrenal pheochromocytoma/paraganglioma

TUBEROUS SCLEROSIS COMPLEX

- Autosomal dominant
- # of individuals with no family history/new (de novo) pathogenic/likely pathogenic gene variants = 66%
- Histology: Epithelial (various) or mesenchymal (angiomyo-lipoma)
- Features:
  - Skin (adenoma sebaceum, shagreen spots)
  - Retinal hamartomas
  - Central nervous system lesions (including tubers)
  - Cardiac lesions
  - Renal tumours
  - Teeth/gum lesions
  - Bone cysts

Reaume, et al., 2013. Canadian guideline on genetic screening for hereditary renal cell cancers. Can Urol Assoc J. 7(9-10):319-23