



# 28 Congreso Sociedad Canaria de Urología

21 al 23  
SEPTIEMBRE 2023

Palacio de Congresos  
**ExpoMeloneras**





# 28 Congreso Sociedad Canaria de Urología

ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE  
CÉLULAS RENALES BILATERAL

Palacio de Congresos  
**ExpoMeloneras**

# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL



95%  
Unilaterales



1-5 %  
Bilaterales

# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL

Yes, **But...**

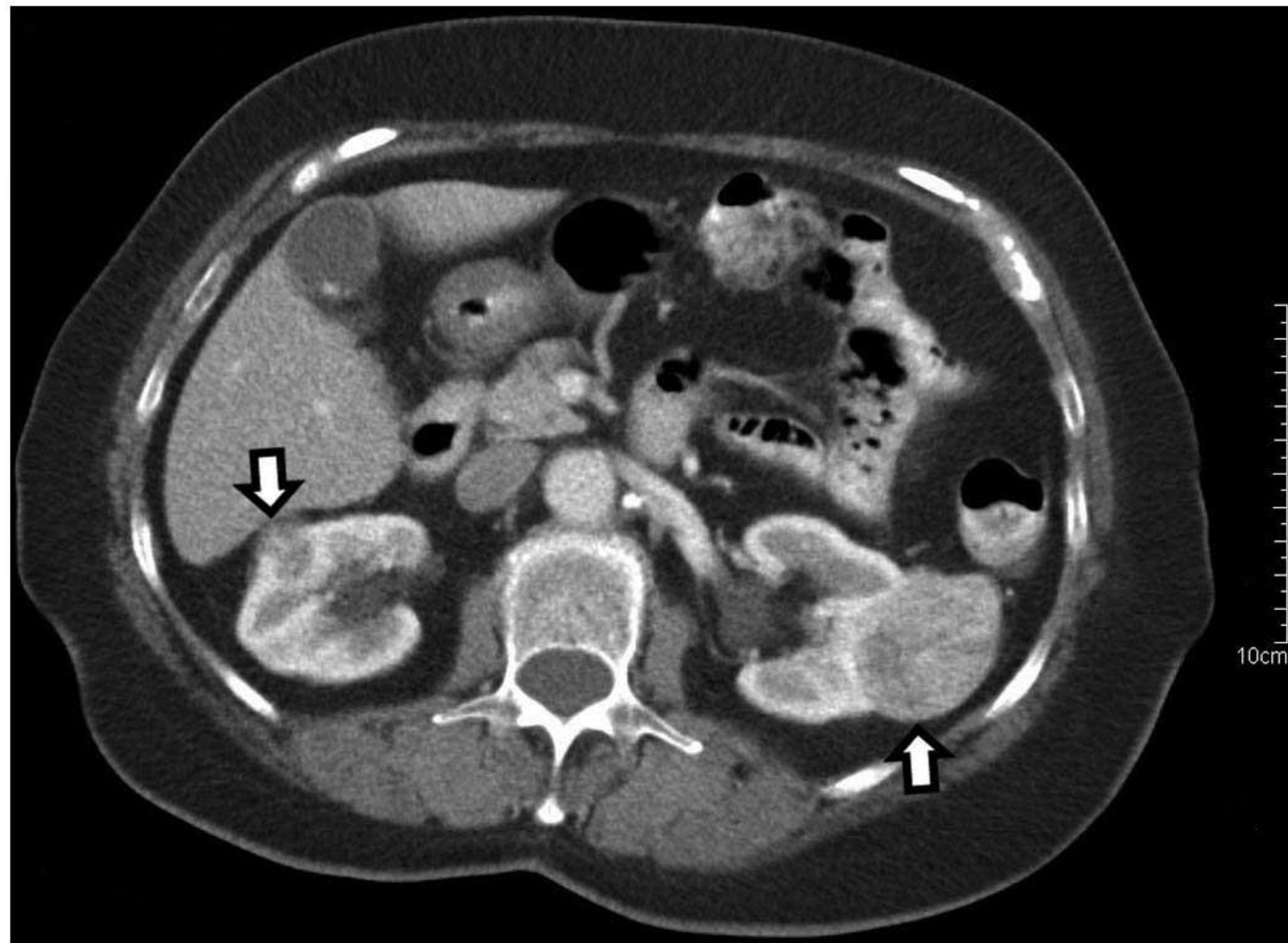
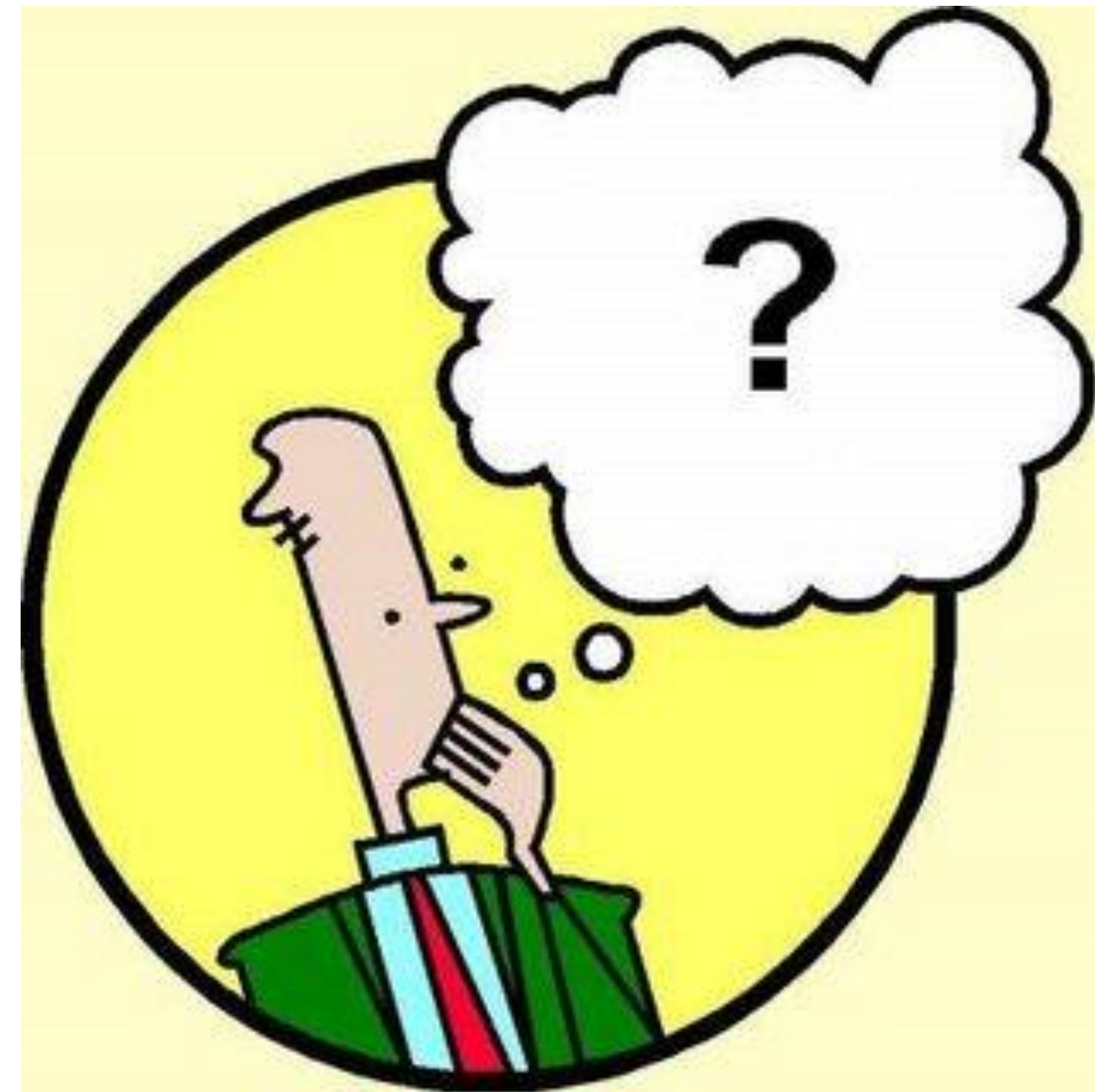
1-5 %  
Bilaterales







# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL





# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL



- **Historia Familiar. Estudio Genético**
- Riesgo Oncológico
- Preservación Nefronal
- Precisión Diagnóstica
- Diseño terapéutico
- Estrategias Diagnósticas intraoperatorias



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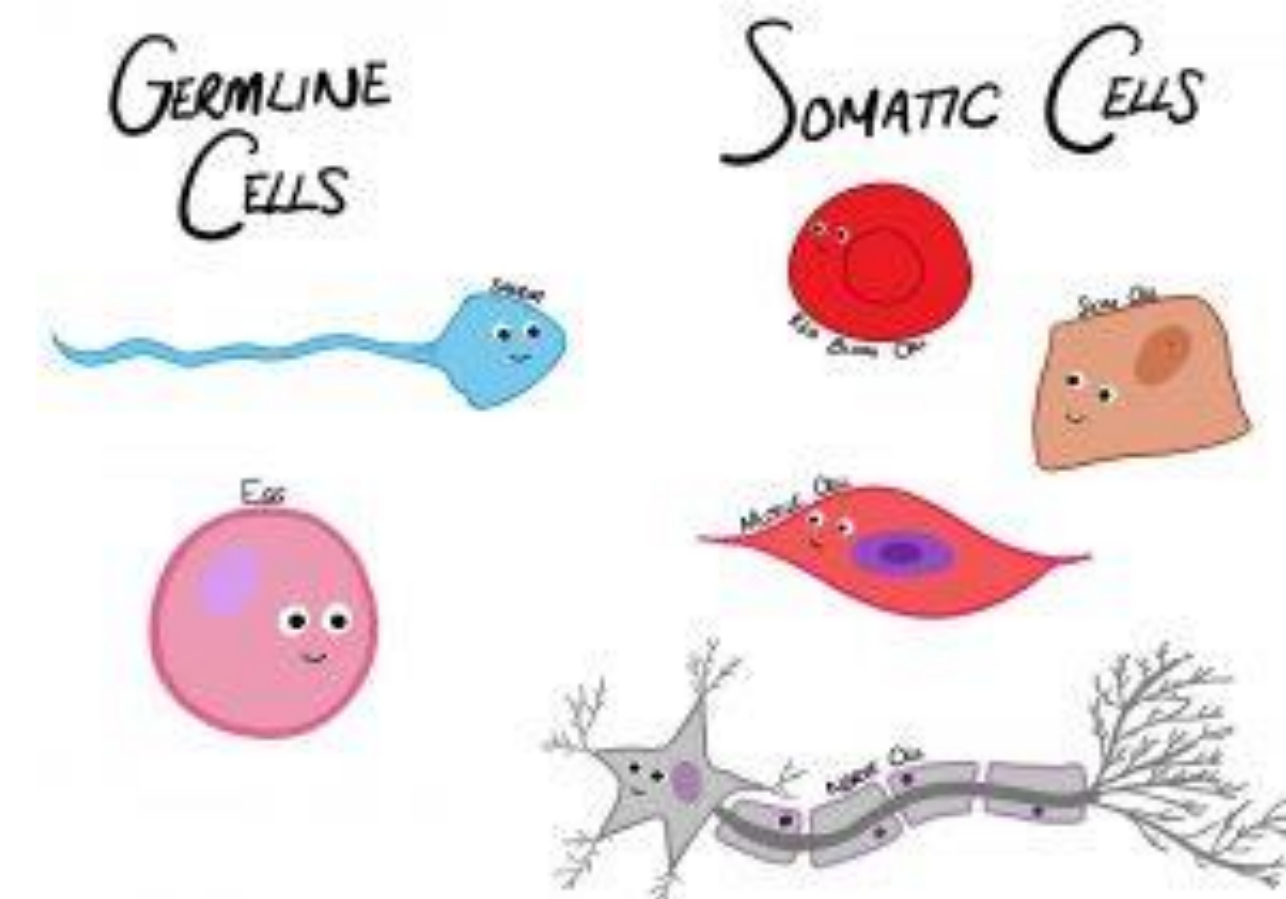
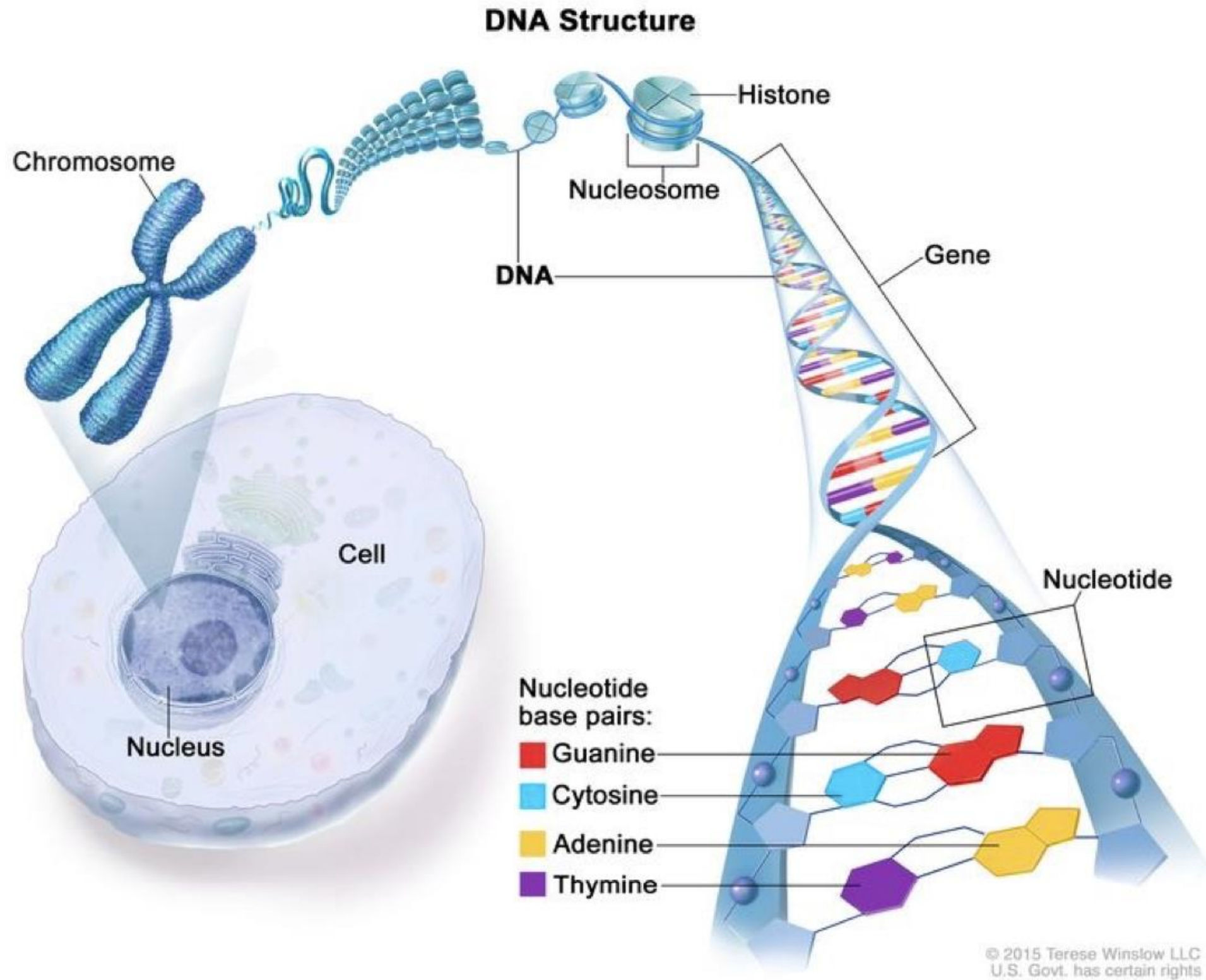
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# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL

## CONSIDERACIONES GENÉTICAS

- **1-5 % RCC Bilateral** (Predisposición Genética)
- **5-8% RCC Hereditarios** (INFRAESTIMADO)
- **38 % M1+ Mutación Linea Germinal**



Mucci, L.A., et al. Familial Risk and Heritability of Cancer Among Twins in Nordic Countries. JAMA, 2016. 315: 68.

Bratslavsky, G., et al. Genetic risk assessment for hereditary renal cell carcinoma: Clinical consensus statement. Cancer, 2021. 127: 3957



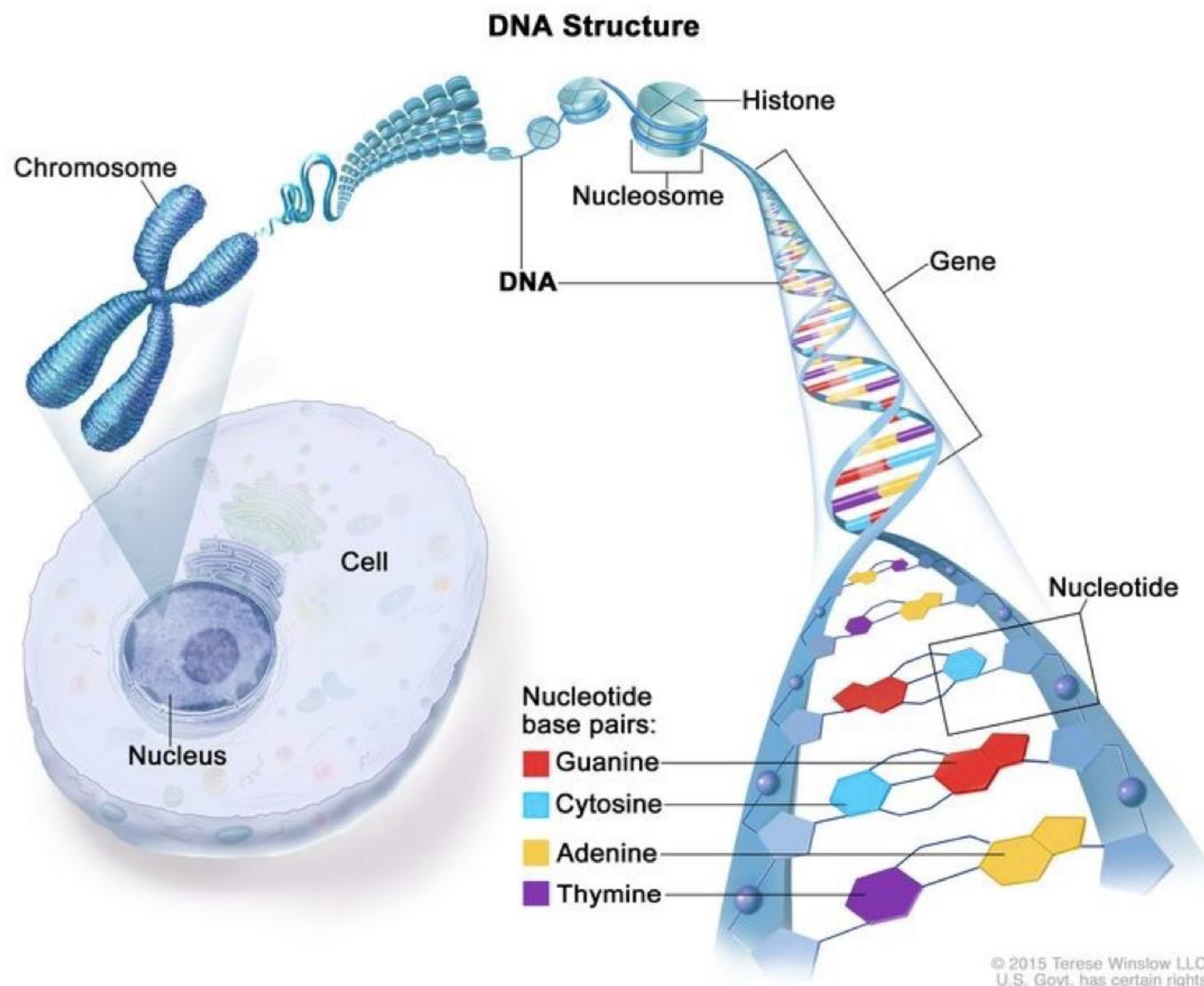








# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL



## CONSIDERACIONES GENÉTICAS

- **Abordaje Multidisciplinar.**
- **Comité Genético.**
- **Ausencia de Manifestaciones clínicas o Historia familiar NO excluye una causa genética**



# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL





# ABORDAJE DIAGNÓSTICO DEL CARCINOMA DE CÉLULAS RENALES BILATERAL



**EDAD !!!**

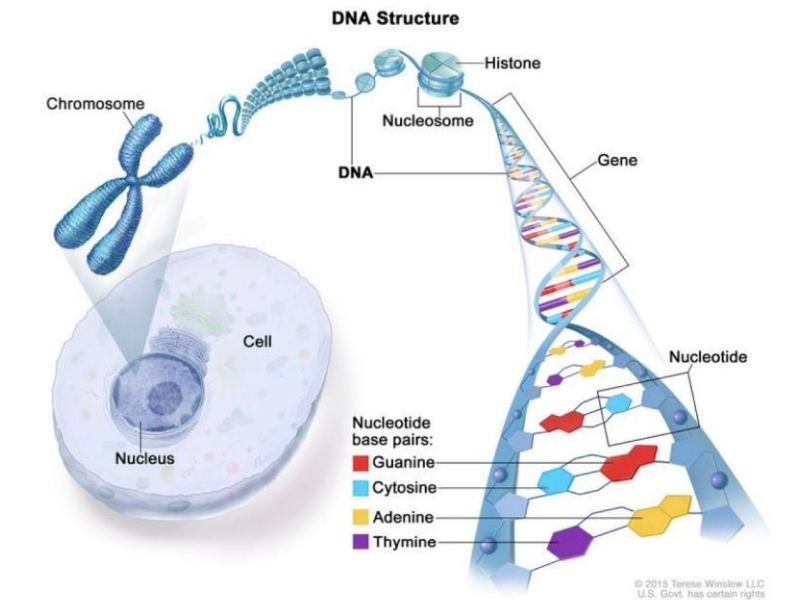
- **Edad Media: 37 años**
- **<46 años (70% RCC-Hereditarios)**
- **Histologías Características !!!**



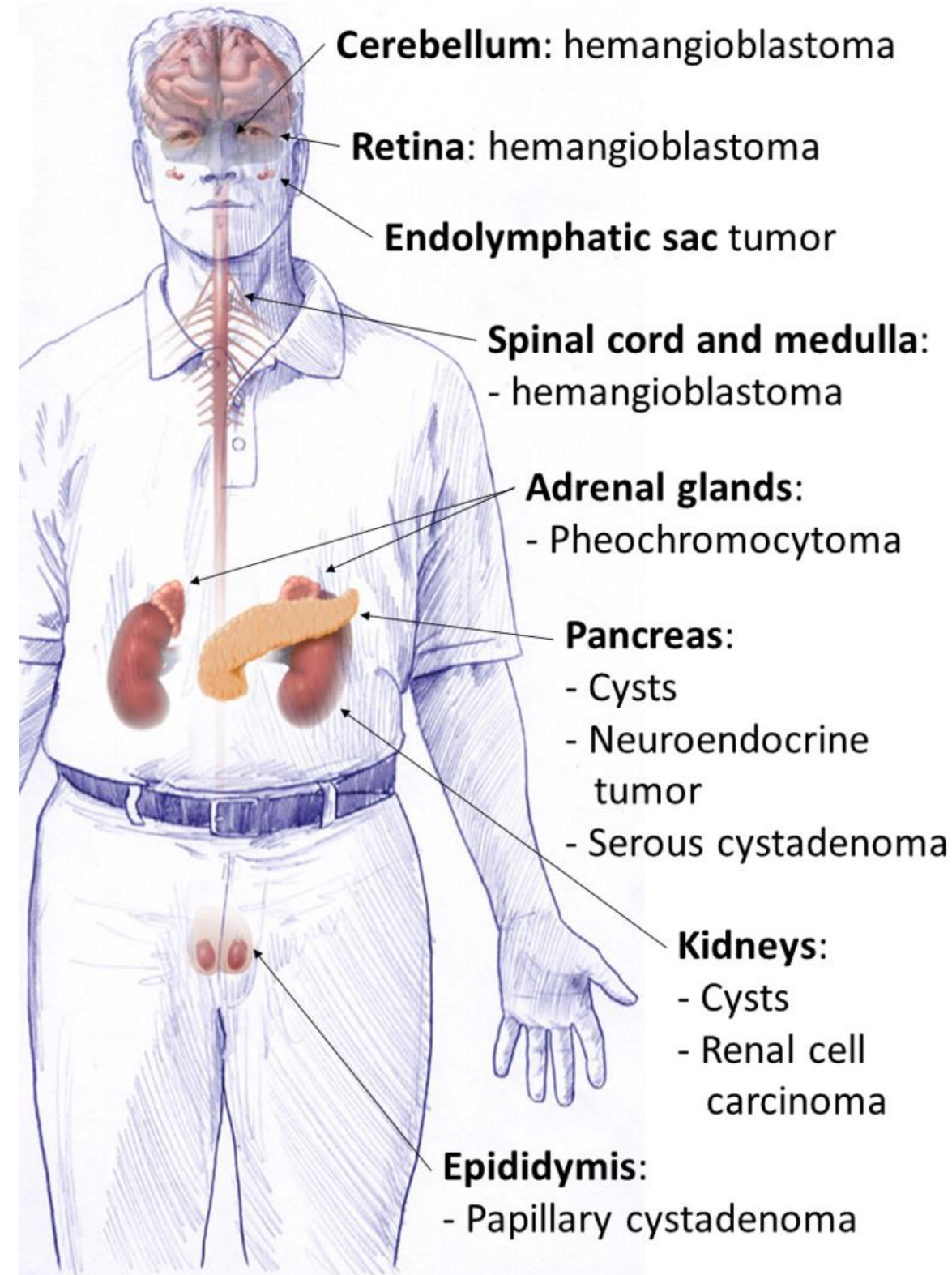


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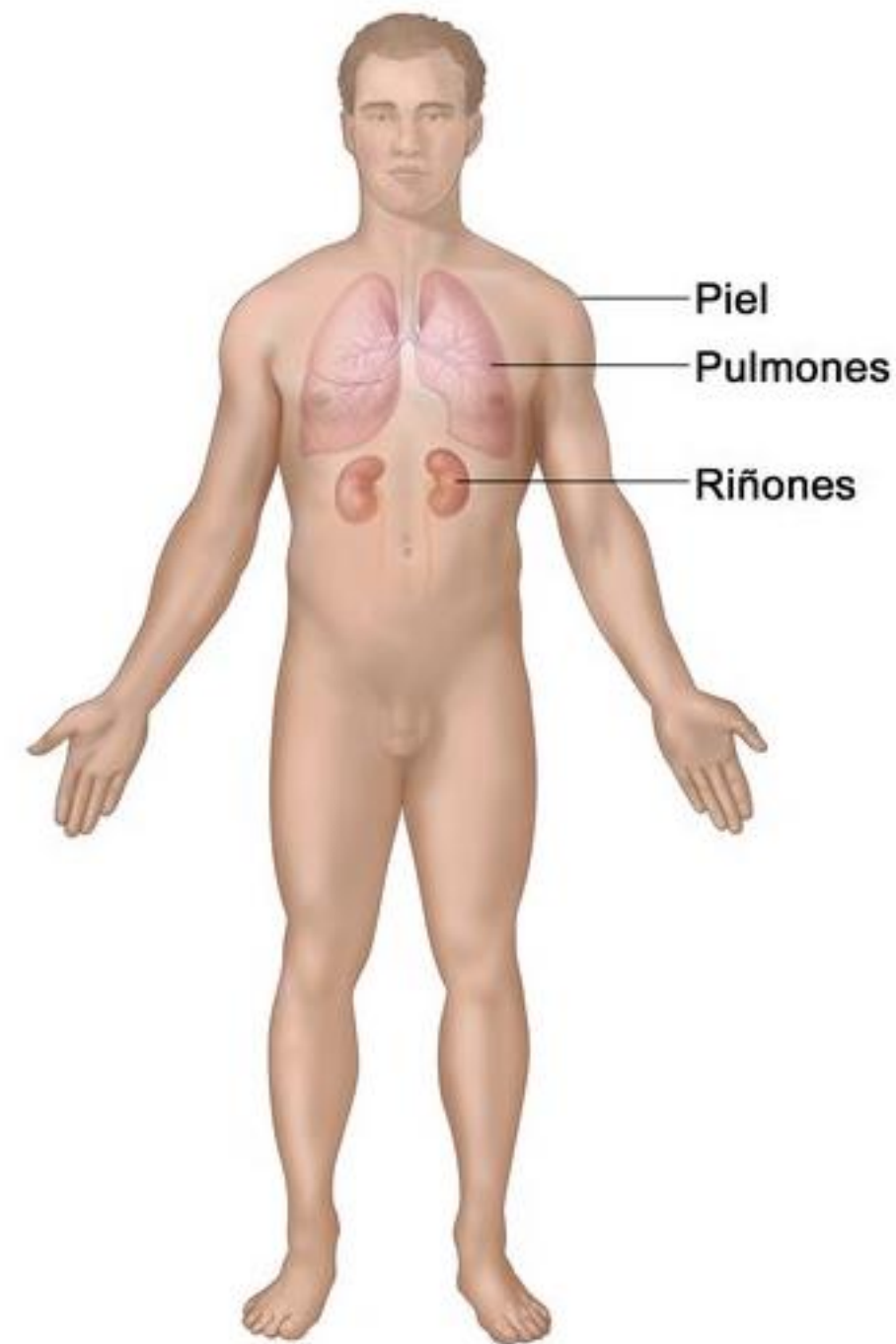
## CONSIDERACIONES GENÉTICAS



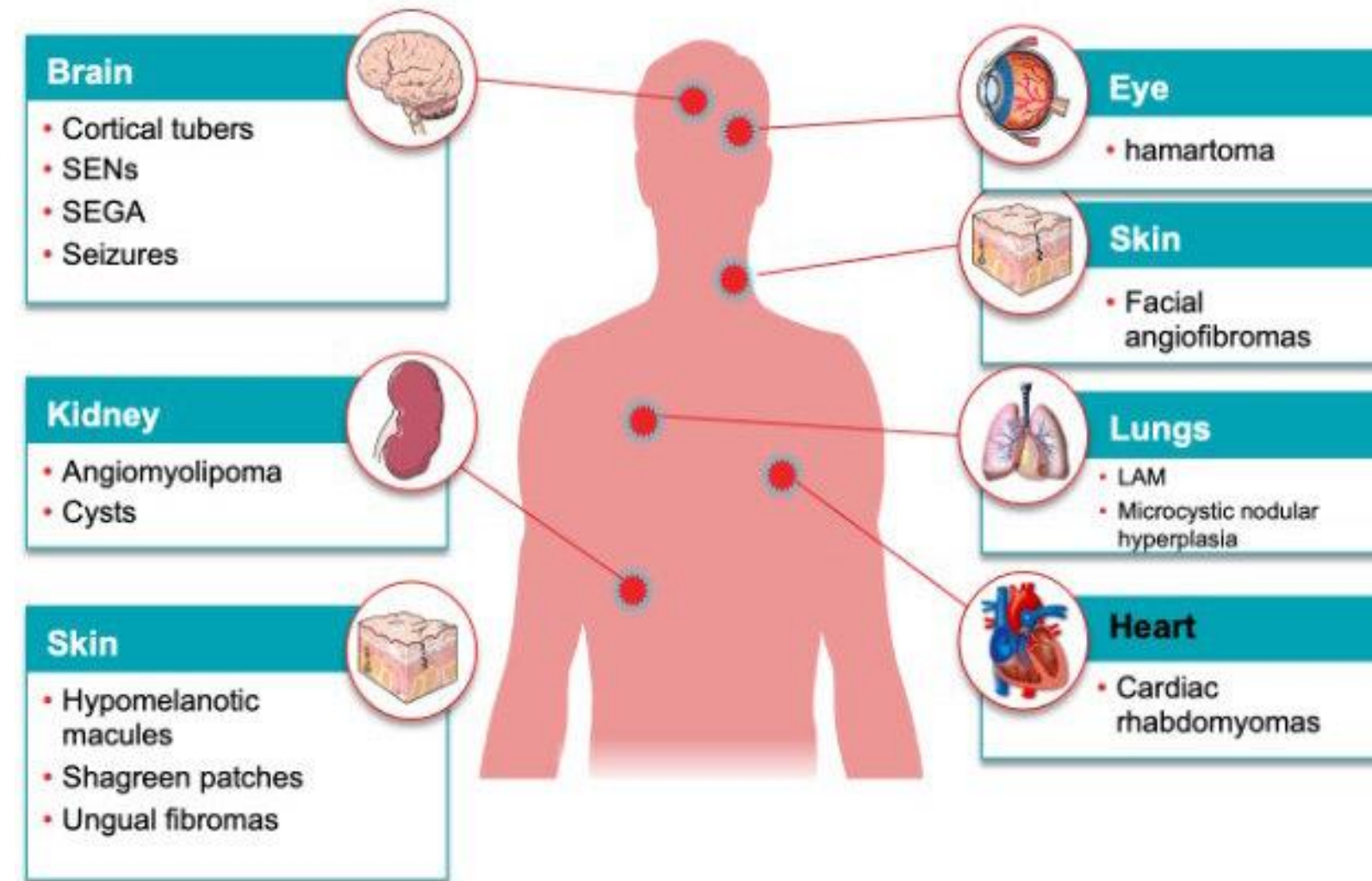
### Von Hippel Lindau



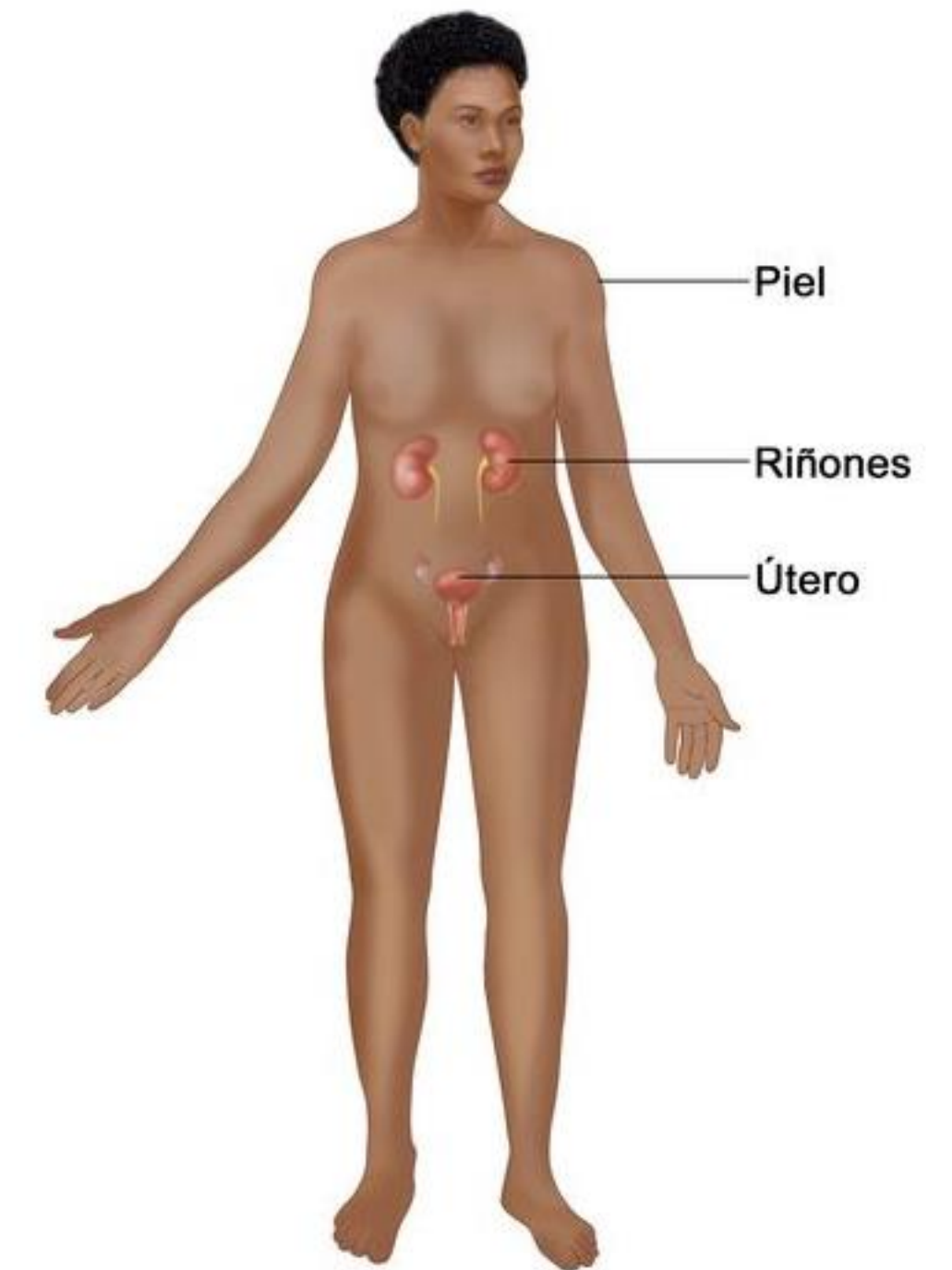
### Birt Hogg Dubé



### Esclerosis Tuberosa



### Deficit Fumarato Hidratasa (Leiomiomatosis Hereditaria)



Mucci, L.A., et al. Familial Risk and Heritability of Cancer Among Twins in Nordic Countries. JAMA, 2016. 315: 68.

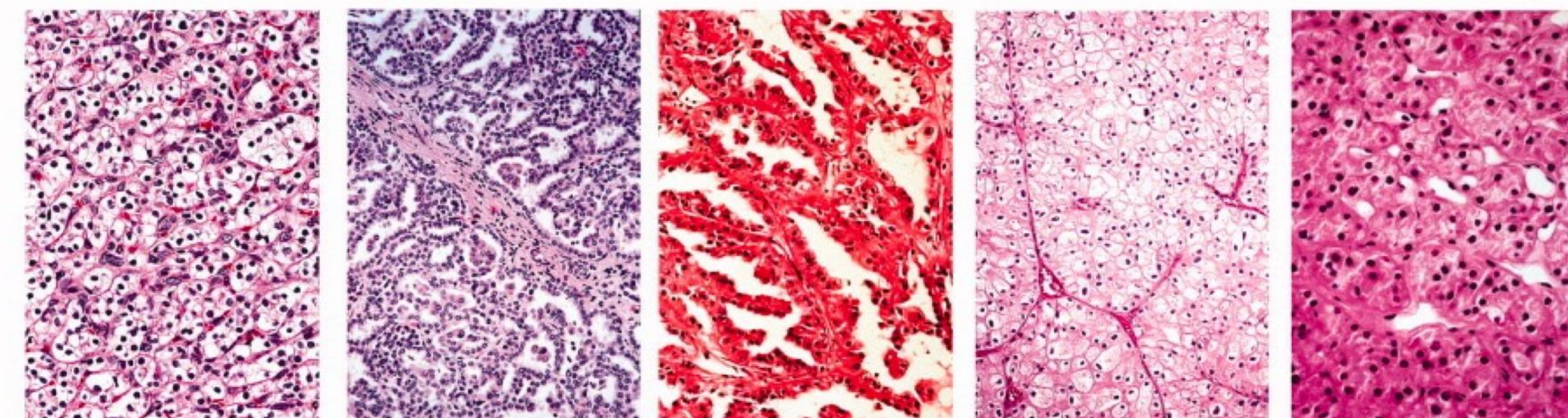
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## HEREDITARY RCC SYNDROMES OVERVIEW

Syndrome/Gene	Common Histologies	Inheritance Pattern Major Clinical Manifestations	Other Specialists Involved in Screening
von Hippel-Lindau (VHL)/ <i>VHL</i> gene	Clear cell	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li><a href="#">Table 2</a></li> </ul>	<ul style="list-style-type: none"> <li>Neurosurgery</li> <li>Ophthalmology</li> <li>Audiology</li> <li>Endocrinology</li> <li>Endocrine surgery</li> </ul>
Hereditary papillary renal carcinoma (HPRC)/ <i>MET</i> gene	Papillary	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li>Multifocal, bilateral renal cell tumors</li> </ul>	<ul style="list-style-type: none"> <li>Nephrology</li> </ul>
Birt-Hogg-Dubé syndrome (BHDS)/ <i>FLCN</i> gene <sup>1,2</sup>	Chromophobe, hybrid oncocytic tumors, clear cell, oncocytomas, angiomyolipomas, papillary RCC	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li>Cutaneous fibrofolliculoma or trichodiscoma, pulmonary cysts, and spontaneous pneumothorax</li> </ul>	<ul style="list-style-type: none"> <li>Pulmonology</li> <li>Dermatology</li> </ul>
Tuberous sclerosis complex (TSC)/ <i>TSC1</i> , <i>TSC2</i> genes	Angiomyolipoma (and other PEComas), renal cysts, eosinophilic solid and cystic RCC, RCC with fibromyomatous stroma, eosinophilic vacuolated tumor, low-grade oncocytic tumor, clear cell	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li><a href="#">Table 1</a></li> </ul>	<ul style="list-style-type: none"> <li>Neurology</li> <li>Dermatology</li> </ul>
Hereditary leiomyomatosis and renal cell cancer (HLRCC)/ <i>FH</i> gene	HLRCC associated RCC or FH-deficient RCC	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li>Leiomyomas of skin and uterus, unilateral, solitary, and aggressive renal cell tumors. PET-positive adrenal adenomas</li> </ul>	<ul style="list-style-type: none"> <li>Gynecology</li> <li>Dermatology</li> </ul>

## Human Renal Epithelial Neoplasms



Type:	Clear Cell 75%	Papillary Type 1 5%	Papillary Type 2 10%	Chromophobe 5%	Oncocytoma 5%
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Hereditary Gene:	VHL	Met	FH	BHD	
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Sporadic Gene:	VHL (92%)	Met (13%)	Unknown	Unknown	
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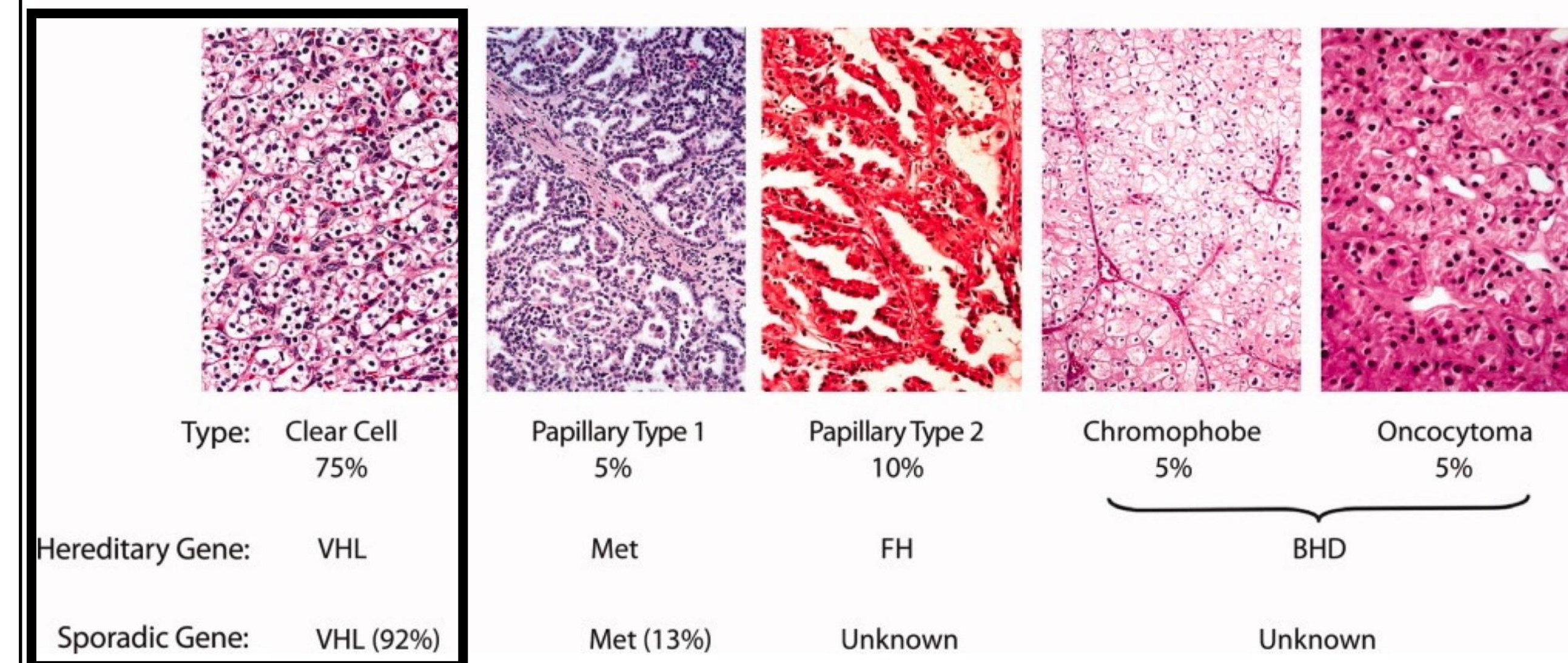


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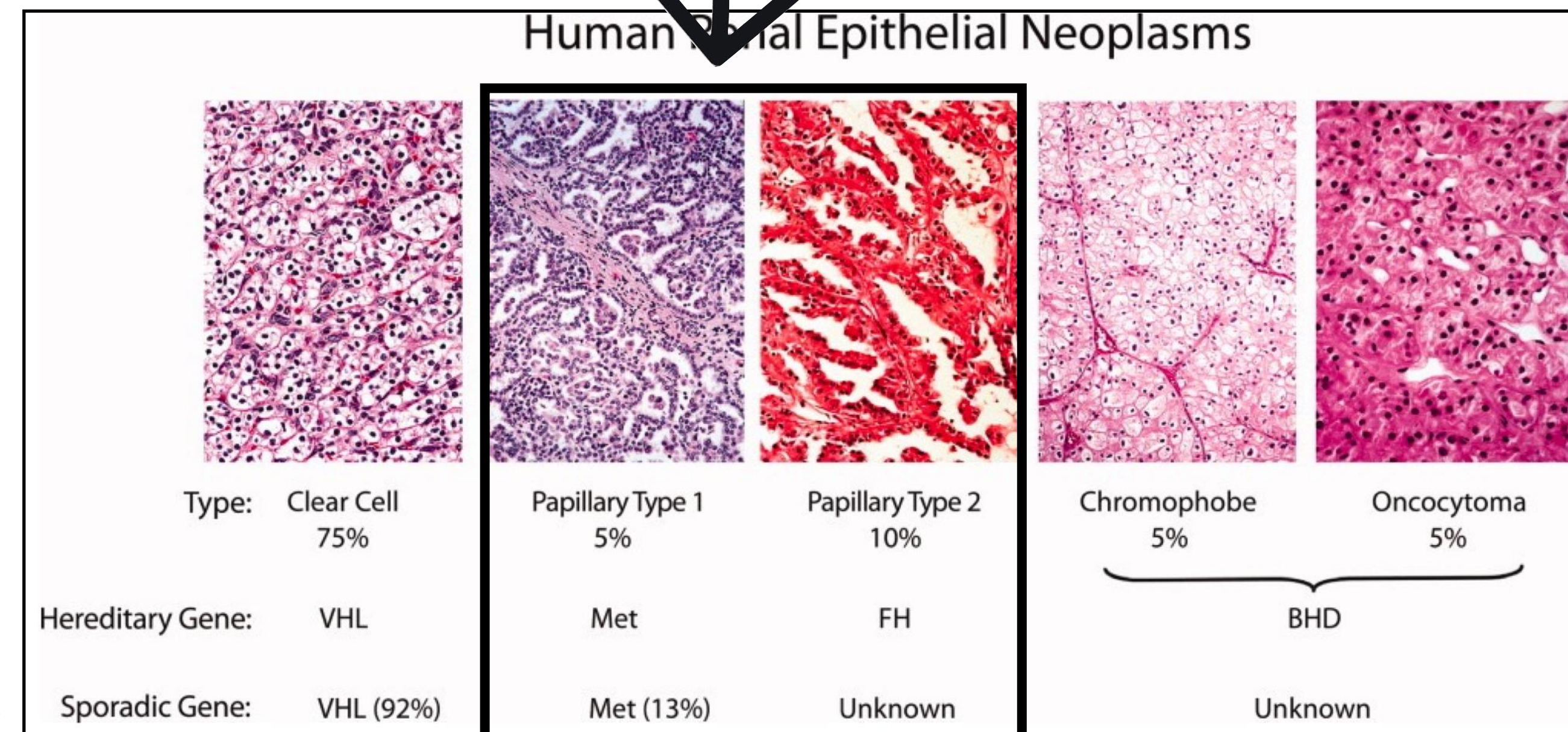
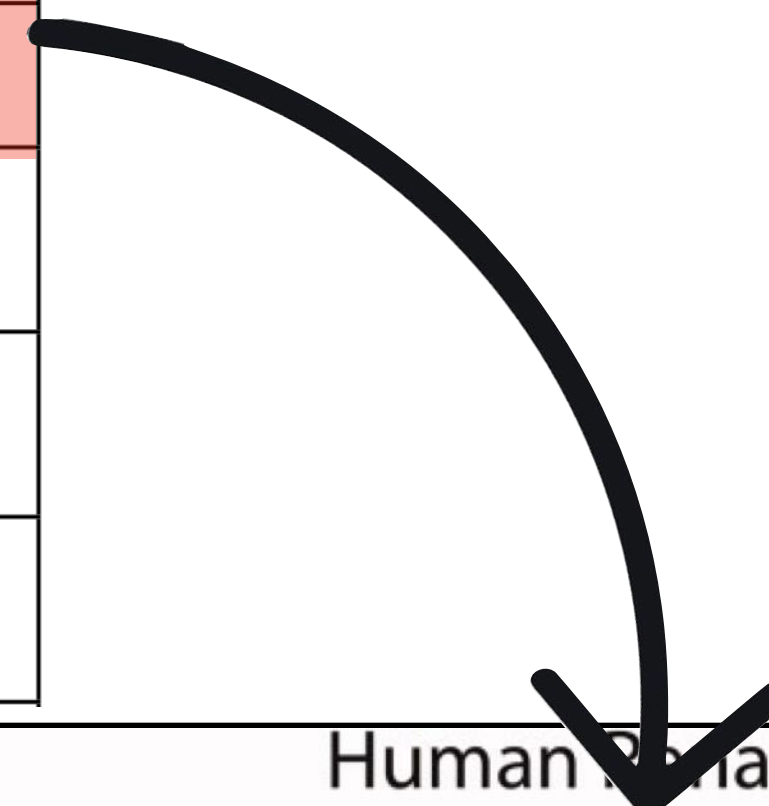


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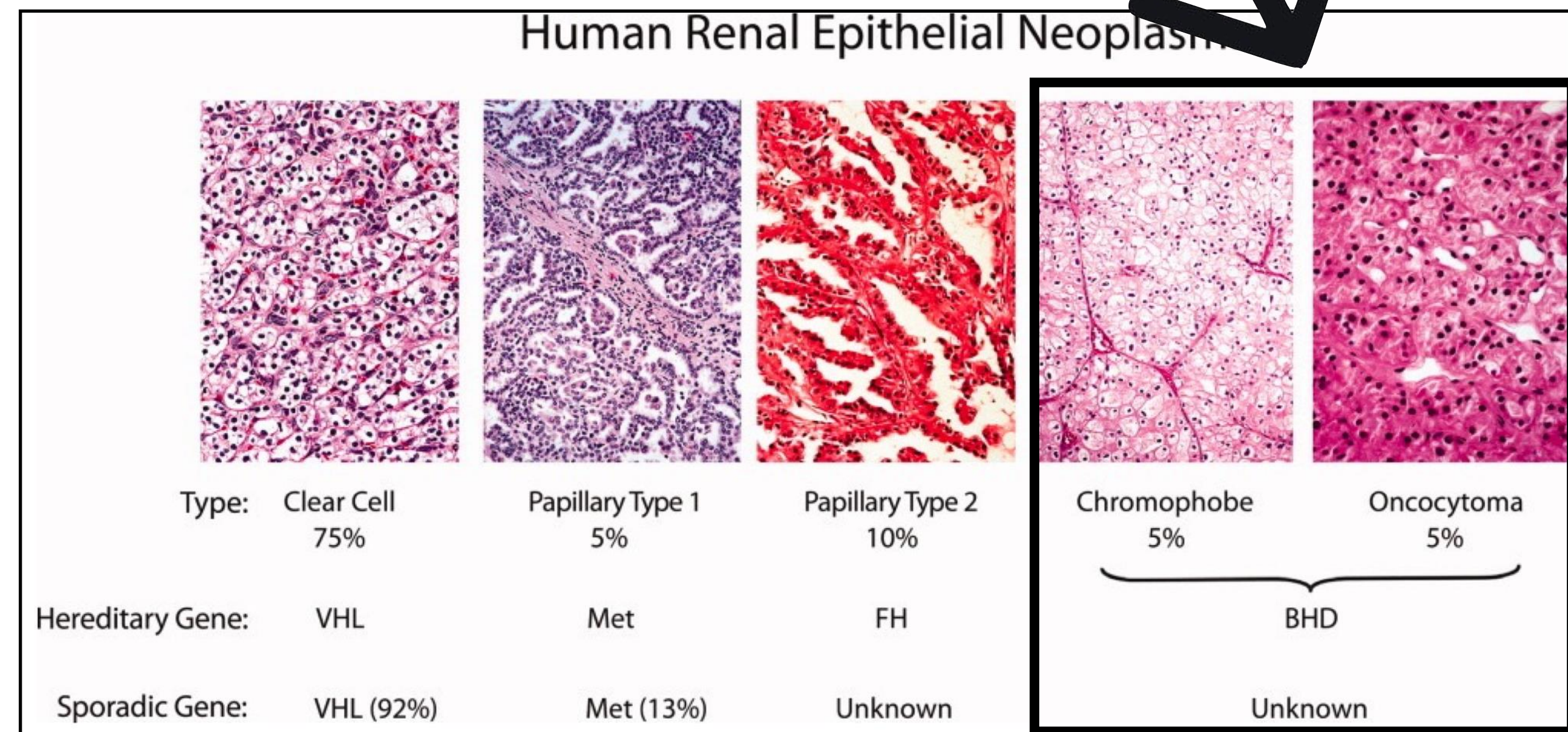


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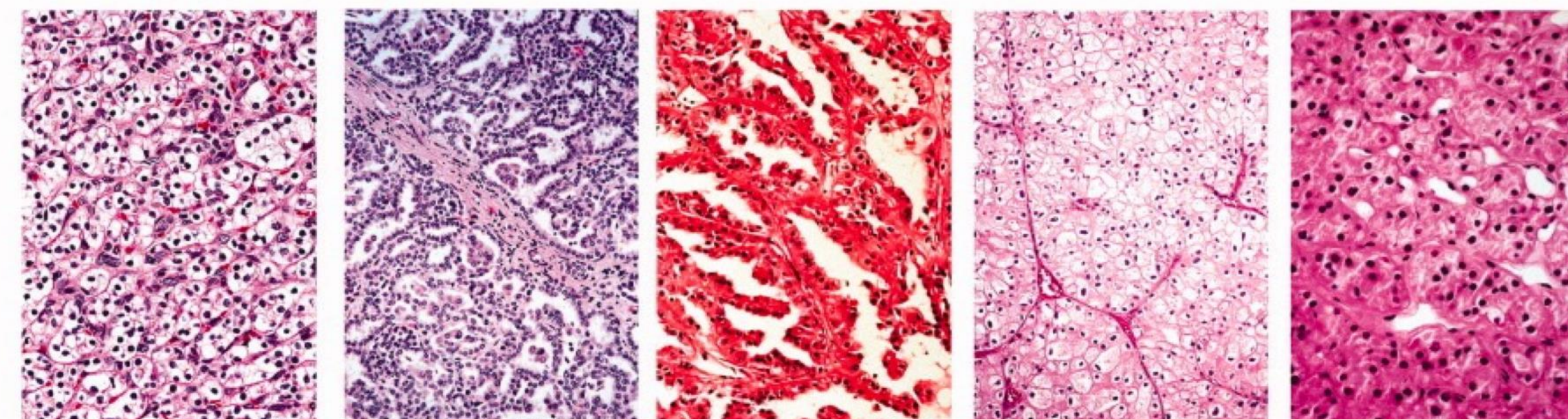
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## Human Renal Epithelial Neoplasms



Type:	Clear Cell	Papillary Type 1	Papillary Type 2	Chromophobe	Oncocytoma
	75%	5%	10%	5%	5%

Hereditary Gene:	VHL	Met	FH	BHD	
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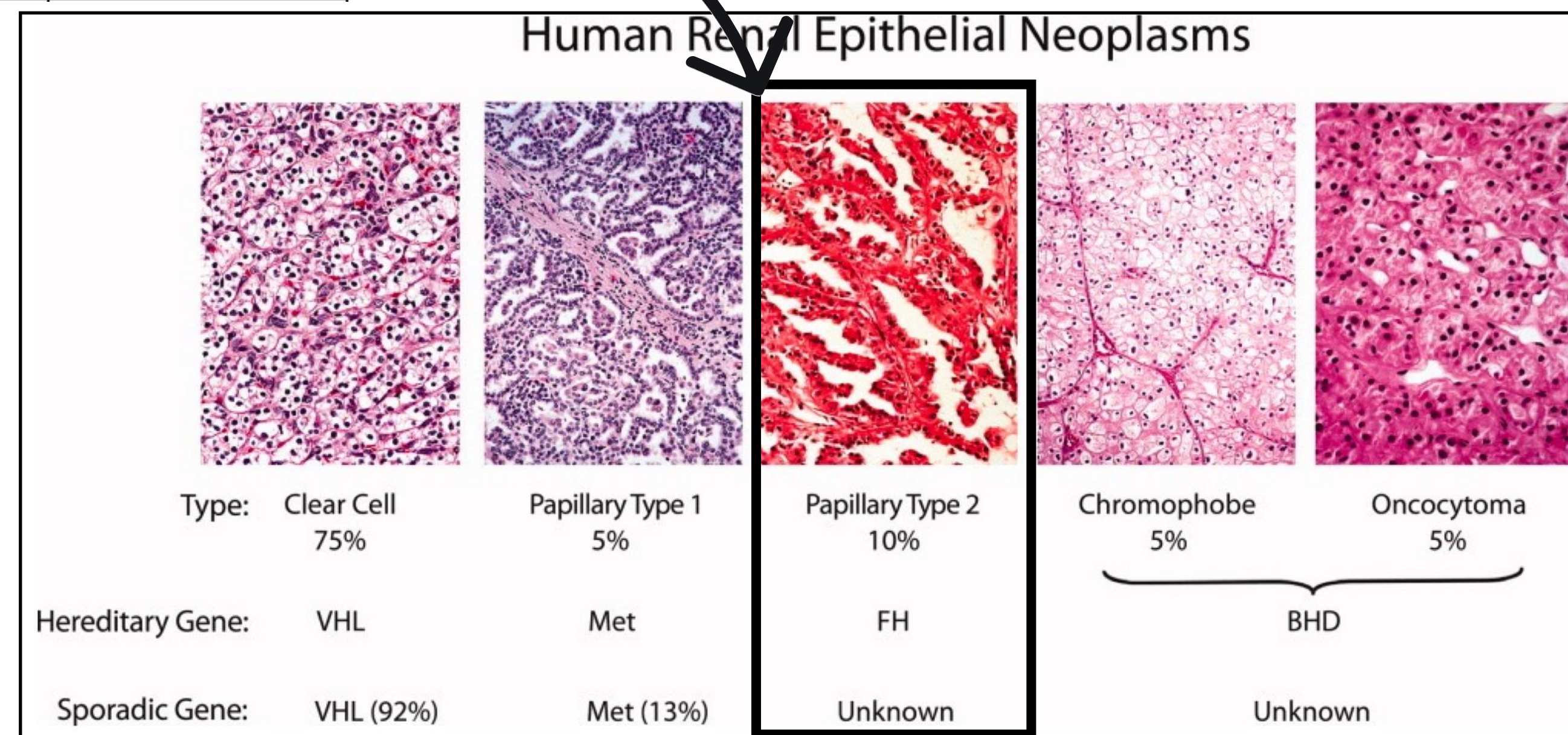
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## HEREDITARY RCC SYNDROMES OVERVIEW

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Hereditary papillary renal carcinoma (HPRC)/ <i>MET</i> gene	Papillary	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li>Multifocal, bilateral renal cell tumors</li> </ul>	<ul style="list-style-type: none"> <li>Nephrology</li> </ul>
Birt-Hogg-Dubé syndrome (BHDS)/ <i>FLCN</i> gene <sup>1,2</sup>	Chromophobe, hybrid oncocytic tumors, clear cell, oncocytomas, angiomyolipomas, papillary RCC	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li>Cutaneous fibrofolliculoma or trichodiscoma, pulmonary cysts, and spontaneous pneumothorax</li> </ul>	<ul style="list-style-type: none"> <li>Pulmonology</li> <li>Dermatology</li> </ul>
Tuberous sclerosis complex (TSC)/ <i>TSC1</i> , <i>TSC2</i> genes	Angiomyolipoma (and other PEComas), renal cysts, eosinophilic solid and cystic RCC, RCC with fibromyomatous stroma, eosinophilic vacuolated tumor, low-grade oncocytic tumor, clear cell	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li><a href="#">Table 1</a></li> </ul>	<ul style="list-style-type: none"> <li>Neurology</li> <li>Dermatology</li> </ul>
Hereditary leiomyomatosis and renal cell cancer (HLRCC)/ <i>FH</i> gene	HLRCC associated RCC or FH-deficient RCC	<ul style="list-style-type: none"> <li>Autosomal dominant</li> <li>Leiomyomas of skin and uterus, unilateral, solitary, and aggressive renal cell tumors. <u>PET-positive adrenal adenomas</u></li> </ul>	<ul style="list-style-type: none"> <li>Gynecology</li> <li>Dermatology</li> </ul>



Schmidt LS, Nickerson ML, Warren MB, et al. Germline BHD-mutation spectrum and phenotype analysis of a large cohort of families with Birt-Hogg-Dubé syndrome. *Am J Hum Genet* 2005;76:1023-1033.  
 Sattler EC, Steinlein OK. Birt-Hogg-Dubé Syndrome. 2006 Feb 27 [Updated 2020 Jan 30]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. *GeneReviews* [Internet]. Seattle (WA): University of Washington, Seattle;1993-2020.  
 Peña-Llopis S, Vega-Ruweather bin-de-Celis S, Liao A. BAP1 loss defines a new class of renal cell carcinoma. *Nat Genet* 2012;44:751-759.  
 Hakimi AA, Ostrovskaya I, Reva B. Adverse outcomes in clear cell renal cell carcinoma with mutations of 3p21 epigenetic regulators BAP1 and SETD2: a report by MSKCC and the KIRC TCGA Research Network. *Clin Cancer Res* 2013;19:3259-3267



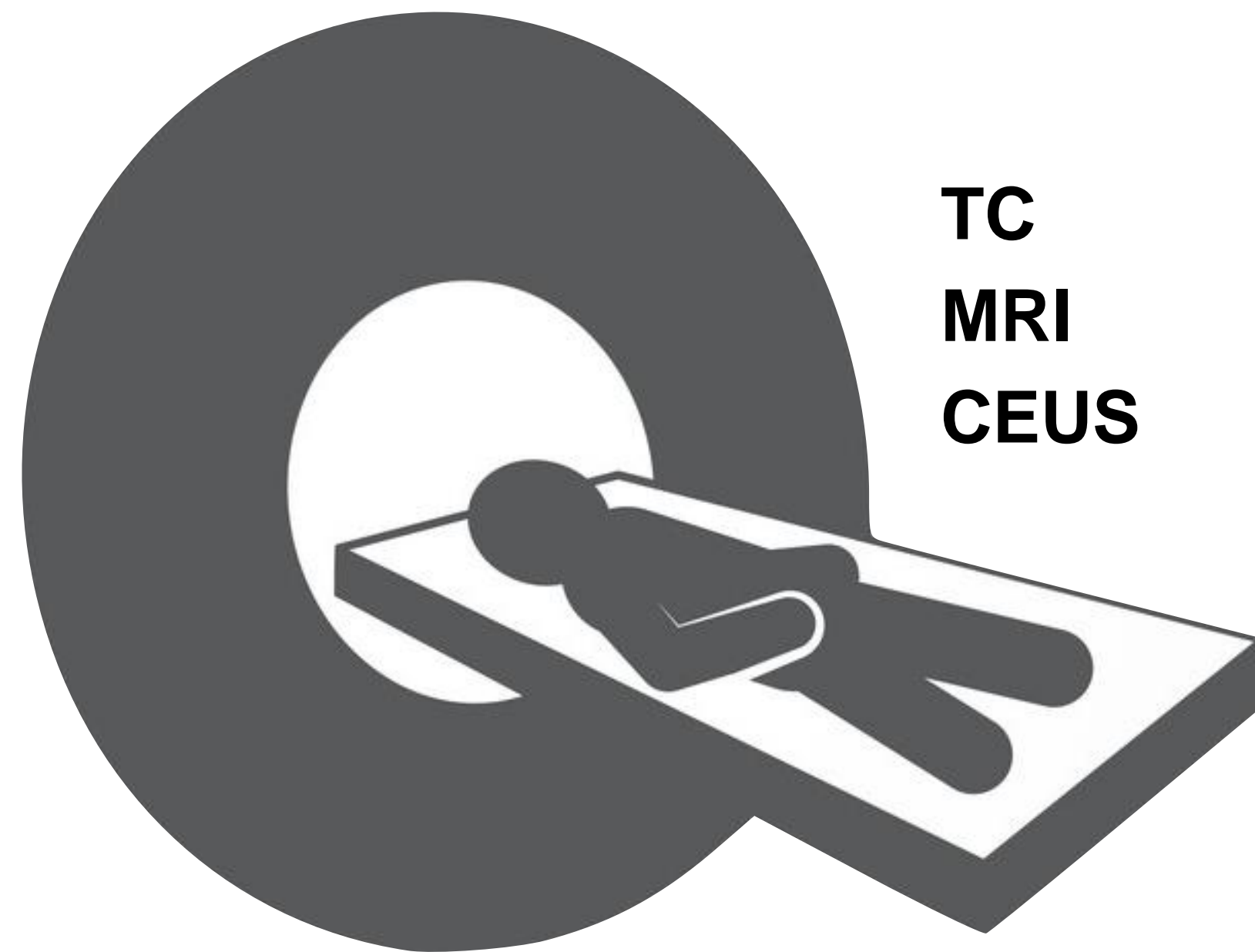
## CRITERIA FOR FURTHER GENETIC RISK EVALUATION FOR HEREDITARY RCC SYNDROMES<sup>a</sup>

1. An individual with a <u>close blood relative<sup>b</sup></u> with a <u>known pathogenic/likely pathogenic variant in a cancer susceptibility gene</u>
2. An individual with RCC with any of the following criteria: <ul style="list-style-type: none"> <li>▶ <u>Diagnosed at age <math>\leq 46</math> y<sup>c</sup></u></li> <li>▶ <u>Bilateral or multifocal tumors</u></li> <li>▶ <u><math>\geq 1</math> first- or second-degree relative<sup>b</sup> with RCC</u></li> </ul>
3. An individual whose tumors have the following <u>histologic characteristics:</u> <ul style="list-style-type: none"> <li>▶ Multifocal papillary histology</li> <li>▶ HLRCC-associated RCC, RCC with fumarate hydratase (FH) deficiency or other histologic features associated with HLRCC</li> <li>▶ Birt-Hogg-Dubé syndrome (BHDS)-related histology (multiple chromophobe, oncocytoma, or oncocytic hybrid)</li> <li>▶ Angiomyolipomas of the kidney and one additional tuberous sclerosis complex (TSC) criterion in the same person (<a href="#">Table 1</a>)</li> <li>▶ Succinate dehydrogenase (SDH)-deficient RCC histology<sup>d</sup></li> </ul>
4. An unaffected individual <sup>e,f</sup> with any of the following criteria: <ul style="list-style-type: none"> <li>▶ <u><math>\geq 2</math> first- or second-degree relatives<sup>b</sup> with RCC (on the same side of the family)</u></li> <li>▶ <u>Any first-degree relative who meets the criteria in boxes 2 or 3 who is unable or unwilling to genetically test</u></li> </ul>



# Evaluación Diagnóstica

Técnicas de imagen en el diagnóstico de la mas renal



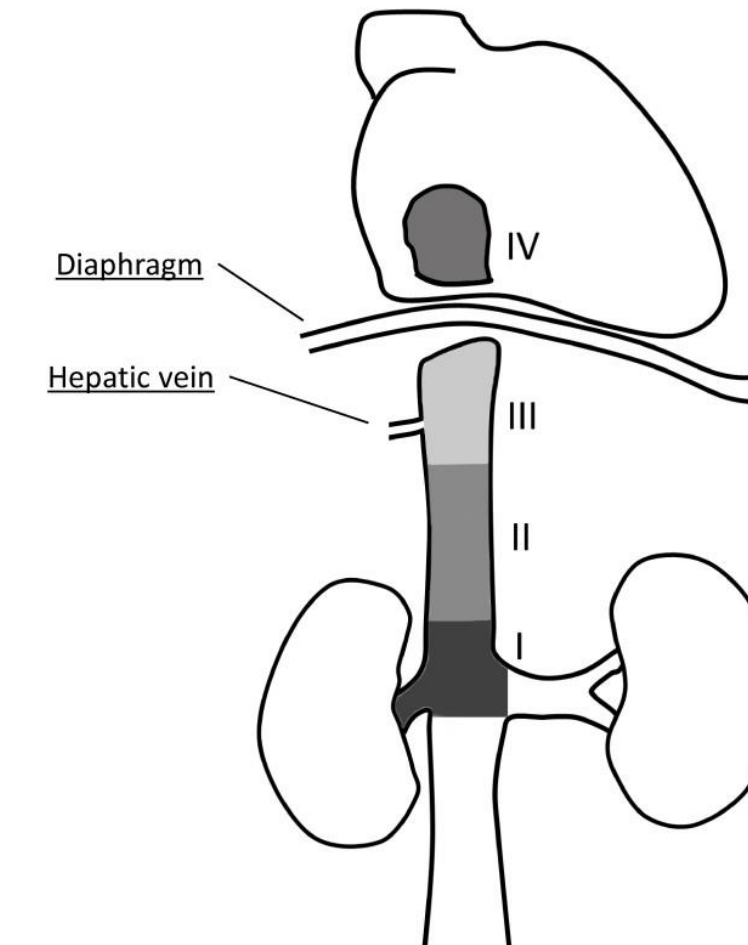
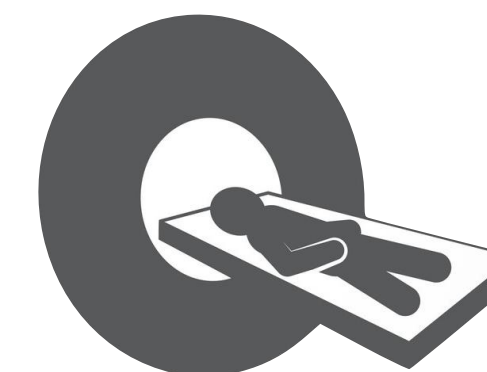


# TC multifásico



S: >90% (81-94%)  
E: >85% (51-90%)

## MRI



Útil para el diagnóstico y el estadiaje completo. GOLD STANDARD — Rápido

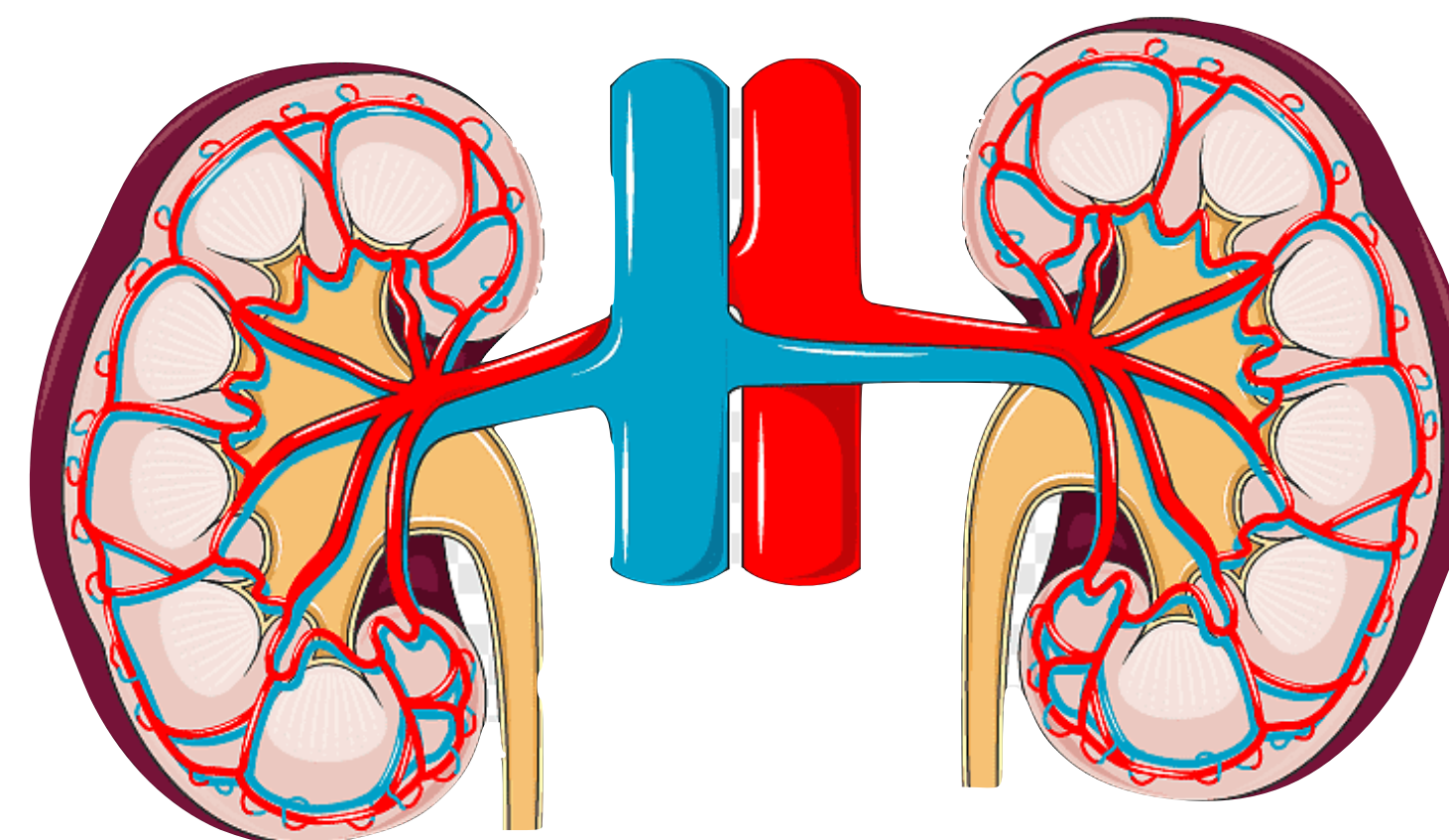
caracterizar el subtipo histológico, así como las características intrínsecas del tumor y el compromiso vascular, lo que tiene implicaciones pronósticas quirúrgicas..



### Rápido



### Barato



**Estadiaje Local: T y N**  
**Estadiaje Distancia**  
**Compromiso Venoso**  
**Fx Riñón Contralateral**

Vogel C, Ziegelmüller B, Ljungberg B, Bensalah K, Bex A, Canfield S, Giles RH, Hora M, Kuczyk MA, Merseburger AS, Powles T, Albiges L, Stewart F, Volpe A, Graser A, Schlemmer M, Yuan C, Lam T, Staehler M. Imaging in Suspected Renal-Cell Carcinoma: System



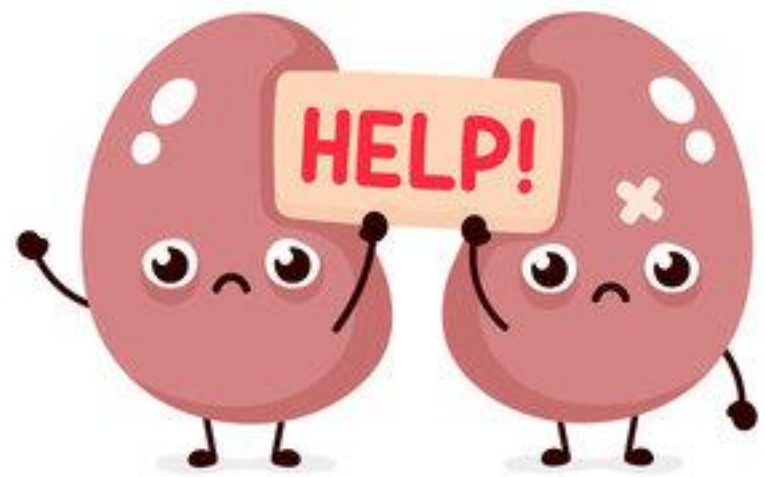


# TC multifásico



Útil para el diagnóstico y el estadiaje completo. GOLD STANDARD — Rápido

caracterizar el subtipo histológico, así como las características intrínsecas del tumor y el compromiso vascular, lo que tiene implicaciones pronósticas quirúrgicas..



**Insuficiencia renal**



**Alergia Contrastes**



**Pediatrica / Embarazo**



**MRI**

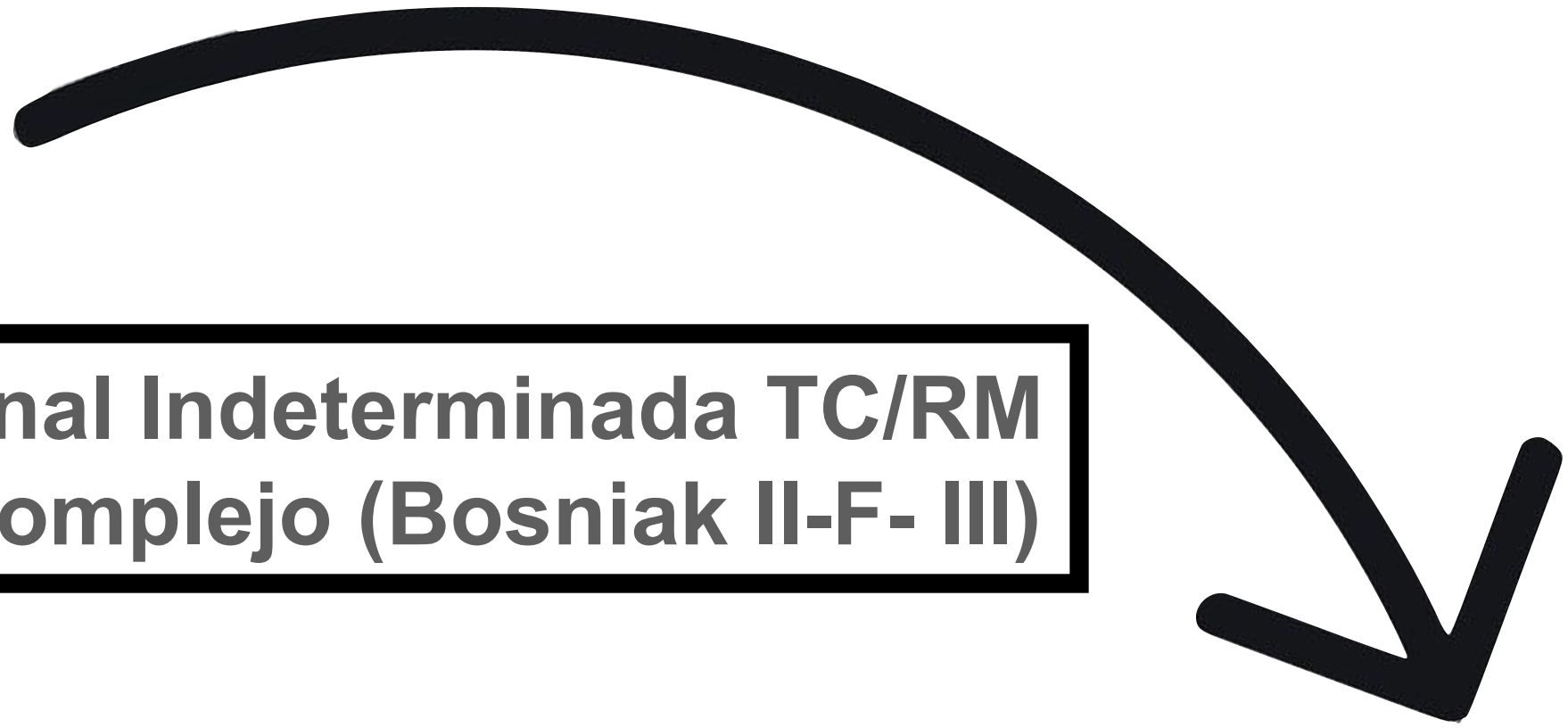
Vogel C, Ziegelmüller B, Ljungberg B, Bensalah K, Bex A, Canfield S, Giles RH, Hora M, Kuczyk MA, Merseburger AS, Powles T, Albiges L, Stewart F, Volpe A, Graser A, Schlemmer M, Yuan C, Lam T, Staehler M. Imaging in Suspected Renal-Cell Carcinoma: System



# TC multifásico



Masa Renal Indeterminada TC/RM  
Quiste Complejo (Bosniak II-F- III)



## CEUS



**SENSIBILIDAD Y ESPECIFICIDAD**  
Comparable Con TC y RM



available at [www.sciencedirect.com](http://www.sciencedirect.com)  
journal homepage: [euoncology.europeanurology.com](http://euoncology.europeanurology.com)



## Comparison of the Diagnostic Performance of Contrast-enhanced Ultrasound with That of Contrast-enhanced Computed Tomography and Contrast-enhanced Magnetic Resonance Imaging in the Evaluation of Renal Masses: A Systematic Review and Meta-analysis

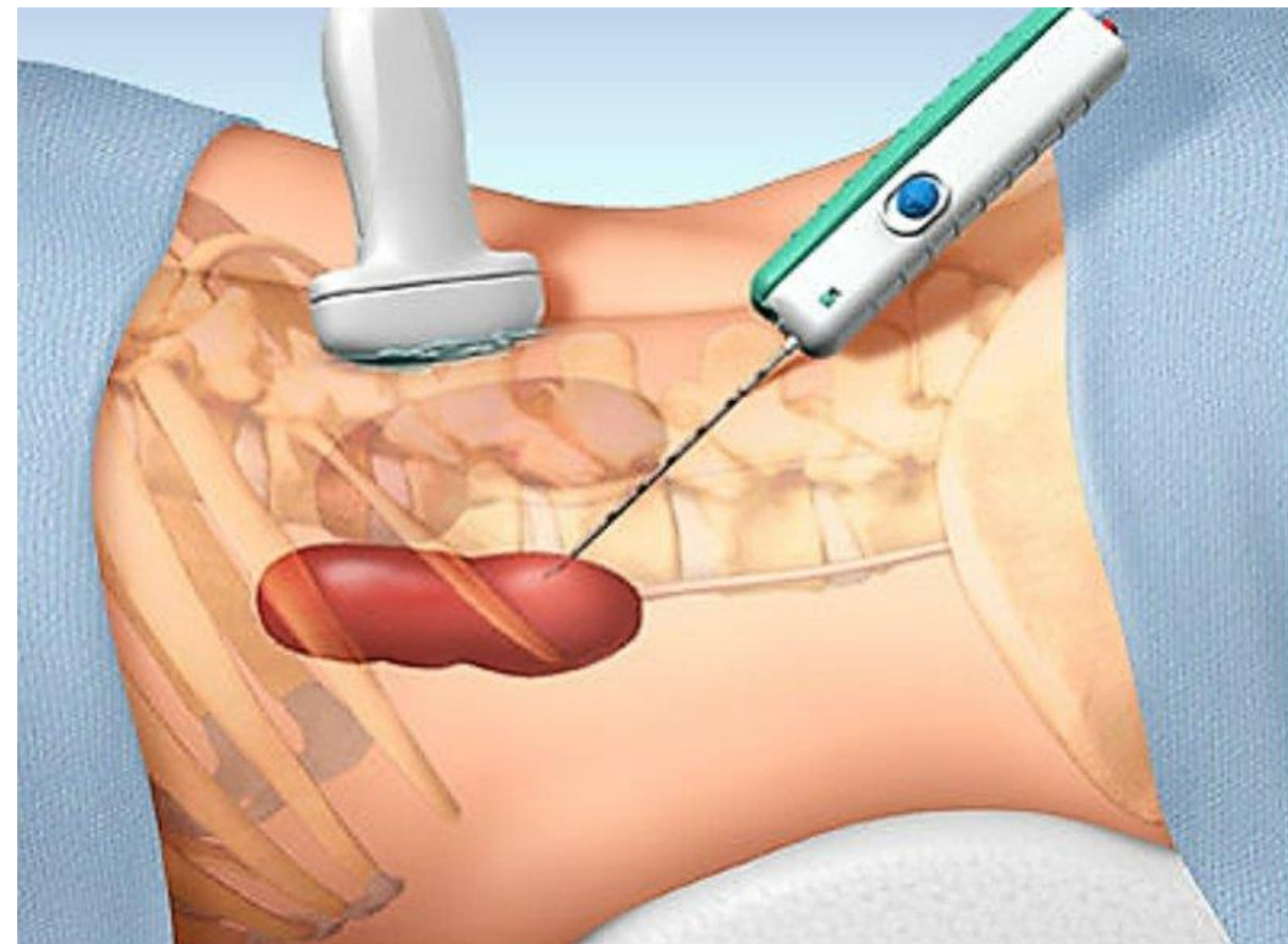
Marc A. Furrer<sup>1,\*</sup>, Samuel C.J. Spycher<sup>1</sup>, Sophia M. Büttiker<sup>1</sup>, Tobias Gross, Piet Bosshard,

Furrer MA, Spycher SCJ, Büttiker SM, Gross T, Bosshard P, Thalmann GN, Schneider MP, Roth B. Comparison of the Diagnostic Performance of Contrast-enhanced Ultrasound with That of Contrast-enhanced Computed Tomography and Contrast-enhanced Magnetic Resonance Imaging in the Evaluation of Renal Masses: A Systematic Review and Meta-analysis. *European Urology Oncology*. 2023;16(1):1-10.





## Biopsia



**TRUCUT x2-3**

**S y E Malignidad > 99%**

**Precisión Histológica > 90%**

**Precisión Grado 62% (High - Low 85 %)**



## Biopsia



TC RM CEUS Indeterminada

Vigilancia Activa  
Previo Tratamiento Focal  
M+

TRUCUT x2-3

S y E **Malignidad** > 99%

Precisión **Histologica** > 90%

Precisión **Grado** 62% (High - Low 85 %)



## Biopsia



TC RM CEUS Indeterminada

Vigilancia Activa  
Previo Tratamiento Focal  
M+

**Vigilancia Expectante** (comórbidos y frágiles)

**Diagnóstico Radiológico** → Tratamiento radical  
(alta precisión pruebas imagen)

**Masas quísticas** (menor rendimiento y precisión diagnóstica. excepto si áreas con patrón sólido: **quistes Bosniak IV**)

TRUCUT x2-3

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Precisión **Histológica** > 90%

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## Biopsia



TC RM CEUS Indeterminada

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TRUCUT x2-3

S y E **Malignidad** > 99%

Precisión **Histológica** > 90%

Precisión **Grado** 62% (High - Low 85 %)



**SEGURIDAD**

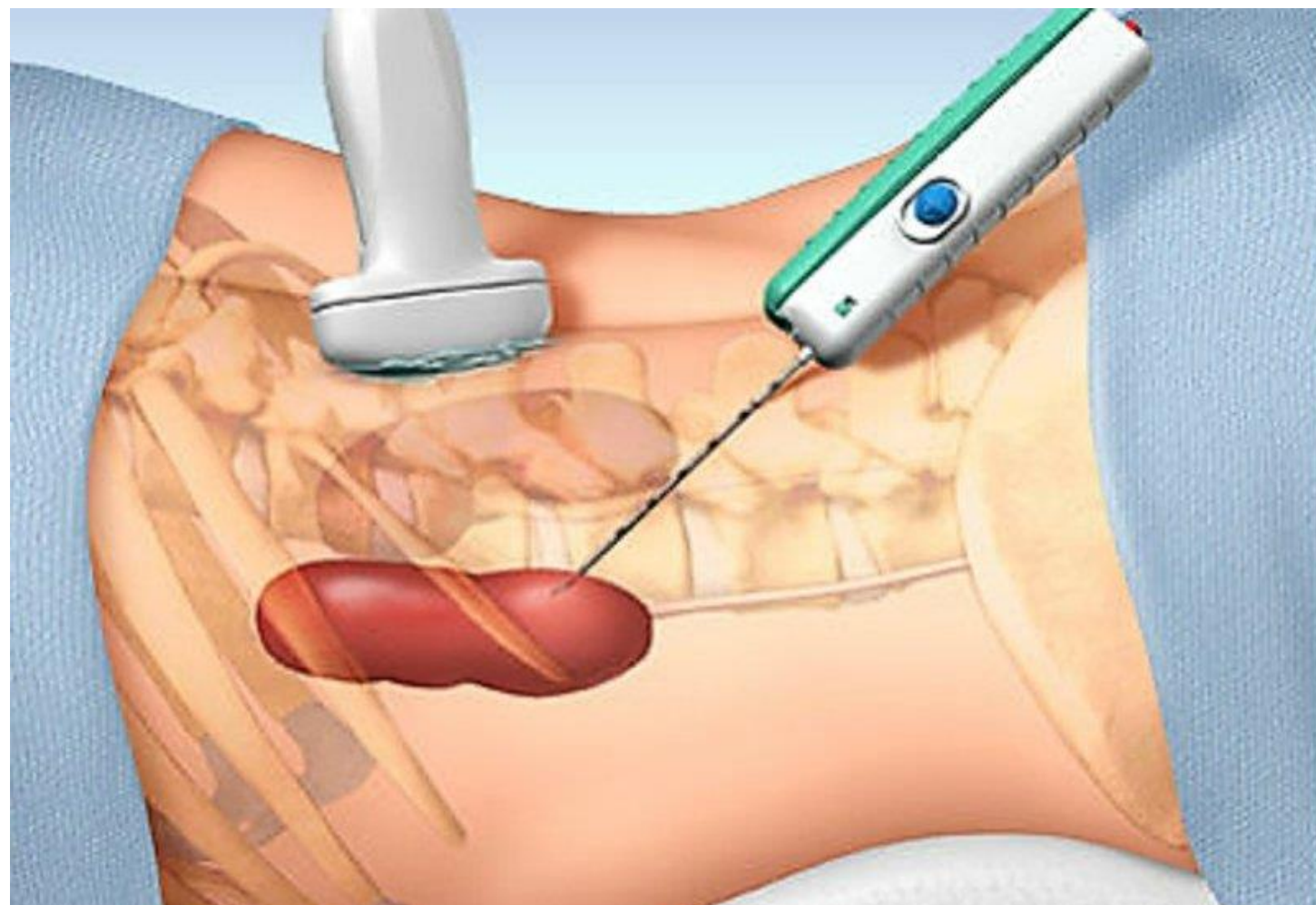
Baja tasa de complicaciones





## Renal Tumor Biopsy: Rationale to Avoid Surgery in Small Renal Masses

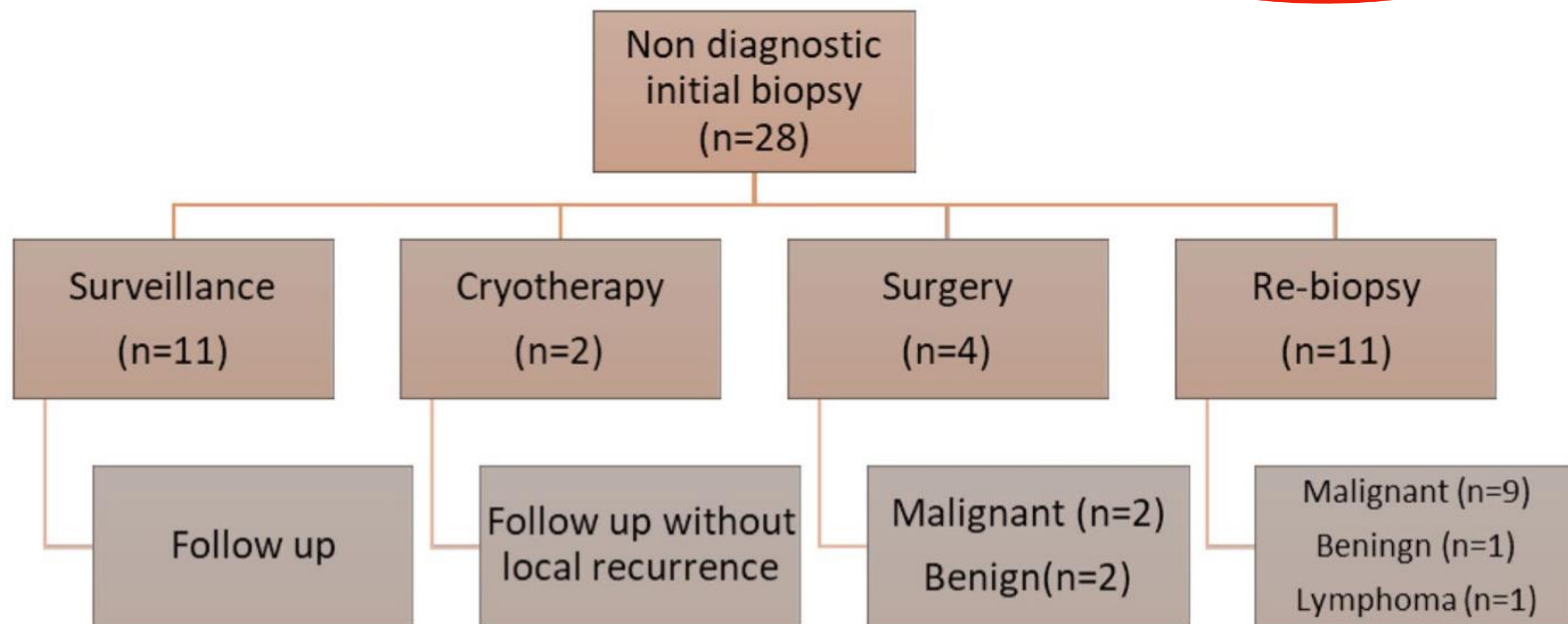
B.S. Amaral<sup>1,2</sup> · P. Macek<sup>1</sup> · A. Arora<sup>1,3</sup> · C.L. Pazeto<sup>1</sup> · A.S. Zugail<sup>1,4</sup> · A. Mombet<sup>1</sup> · A. Fregeville<sup>5</sup> · M. Lefevre<sup>6</sup> ·



28% BENIGNO !!

**Table 2** Histopathology of 165 diagnostic biopsies of small renal masses

Benign masses (n=50)		Malignant masses (n=115)	
Type	n (%)	Type	n (%)
Oncocytoma	37 (74)	Clear cell RCC	67 (58.2)
Angiomyolipoma	11 (22)	ISUP 1	3 (4.4)
Other	2 (4)	ISUP 2	55 (82)
		ISUP 3	4 (5.9)
		ISUP 4	-
		Not specified	5 (7.4)
		Papillary RCC	29 (25.2)
		Type 1	19 (65.5)
		Type 2	9 (31)
		Not specified	1
		Chromophobe RCC	14 (12.1)
		RCC (unspecified subtype)	1
		Metastasis from another primary	2
		Urothelial carcinoma	1
		Lymphoma	1





• Bilaterales o Jóvenes —> **RCC - Hereditario ??**



• 5 Síndromes:

- Von Hippel Lindau // Birt Hogg Dubé // Esclerosis Tuberosa
- Deficit Fumarato Hidratasa (Leiomiomatosis Hereditaria)

• **Histologías:** Papilar múltiple, Cromofobo múltiple-oncocitomas, AML, etc

• TAC —> **CEUS**

• **Biopsia**



TC RM CEUS Indeterminada

Vigilancia Activa  
Previo Tratamiento Focal  
M+



**Vigilancia Expectante** (comórbidos y frágiles)  
**Diagnóstico Radiológico** —> Tratamiento radical (alta precisión pruebas imagen)  
**Masas quísticas** (menor rendimiento y precisión diagnóstica. excepto si áreas con patrón sólido: quistes Bosniak IV)





# 28 Congreso Sociedad Canaria de Urología

**MUCHAS GRACIAS !!**

Palacio de Congresos  
**ExpoMeloneras**