


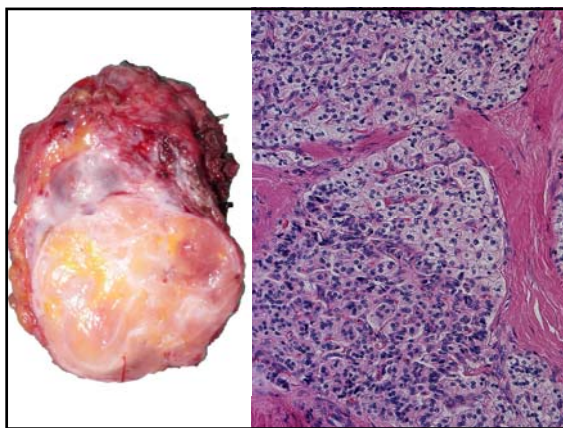
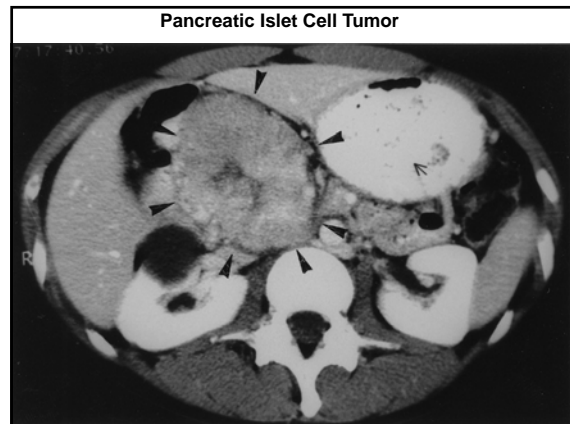
**VHLFA Annual Meeting and Conference**  
Houston, Tx, January 26-28, 2012

**Pancreatic Islet Cell Tumors**  
(Pancreatic Neuroendocrine Tumors - PNETs)

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**20yr, male, Bilateral Pheo, ICT**

Bilateral Adrenalectomy  
Steroid Dependent

**PNETs and VHL**

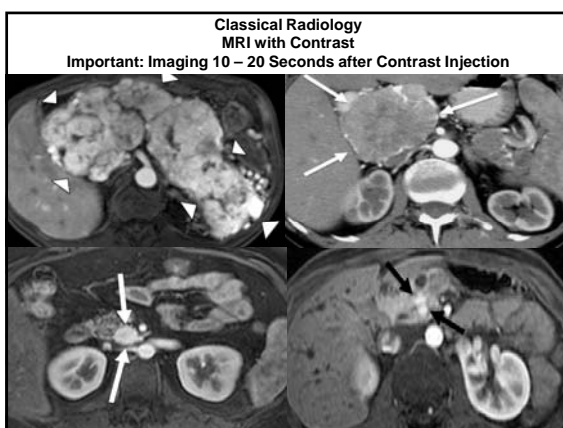
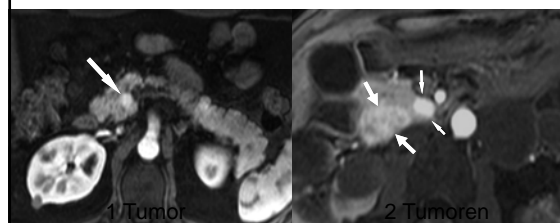
- Prevalence
- Serial Tomography
- Nuclear Medicine Imaging
- Growth
- Malignancy
- Genetics
- Treatment

### Prevalence

- The Freiburg VHL Registry 590
- Patients with ICT and Abdominal MRT 48
- Additional Patients with ICT but only with CT 11
- Patients with Autopsy 1/4
- Prevalence 60/590 10%
- Gender: Females:Males 39:21
- Age at Diagnosis 13-75
- Symptomatic 1/60

### Number of VHL ICTs

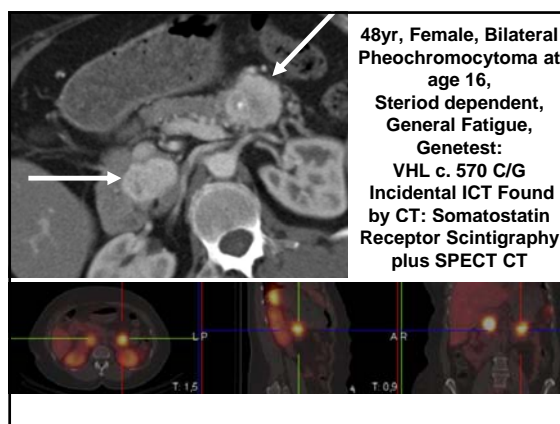
Single 74,5%      Multiple 25,5%

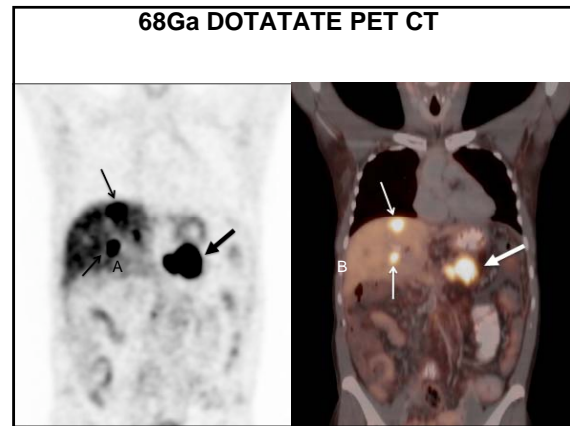
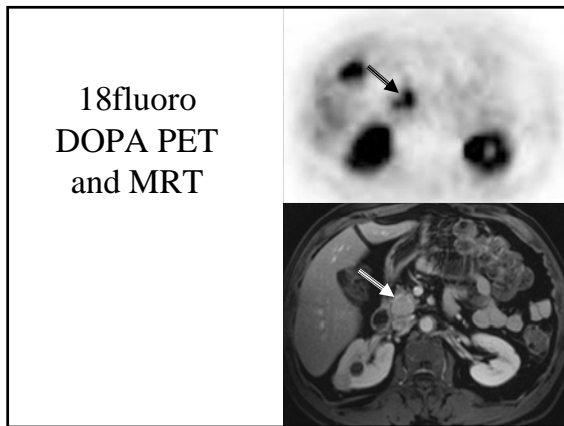


### MRI and CT Imaging

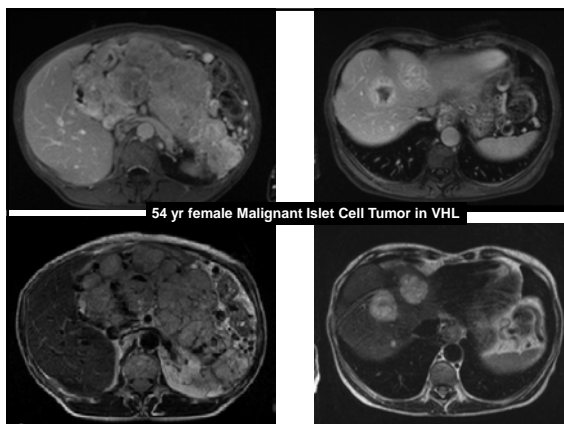
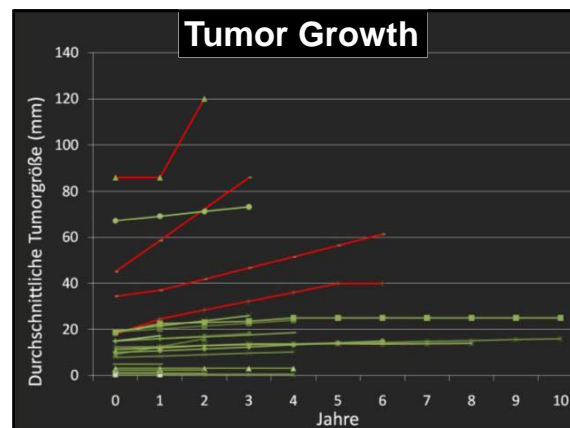
- MRI (n=23)
- Sensitivity 100%
- Specificity 96%
- CT (n=19)
- Sensitivity 89%
- Specificity 79%

MRT  
vs.  
CT





Nuclear Medicine Imaging			
• Octreoscan (99Tc/111In)	(n=14)		
• Sensitivity	12+	86%	
• 18fluoro DOPA PET	(n=7)		
• Sensitivity	2+	29%	
• 68Ga DOTATATE PET	(n=4)		
• Sensitivity	4+	100%	
• 123iodo MIBG	(n=6)		
• Sensitivity	1+	16%	
• 18fluoro FDG PET CT	(n=2)		
• Sensitivity	2+	100%	



Malignant PNETs in VHL	
• N=	16 / 60
• Age at diagnosis of malignancy:	16 – 62, mean 38 yrs
• Site of metastases:	
	Liver 52%,
	LN 39%,
	Bone 9%
• Size of Malignant ICT 3-x cm	

VHL Germline Mutations Associated with ICT					
34 Different Mutations and 4 Large Deletions					
208 G>T	E70X	1			
221 T>G	V74G	1	464-2 A>G	Splice	2
224-6 del			467 A>G	Y156C	1
TCT	T76delF	1	472 C>G	L158V	1
240 T>G	S80R	1	478>79 del AG	E160AfsX12	2
266 T>C	L89P	2	482 G>A	R161Q	5
277 G>C	G93R	1	488 T>A	L163H	1
292 T>C	Y98H	3	499 C>T	R167W	7
340 G>A	G114S	1	500 G>A	R167Q	4
340+1			533 T>C	L178P	1
G>T	Splice	1	548 C>A	S183X	1
349 dup	W117Lfs	1	583 C>T	Q195X	1
T	X15	1	593 T>A	L198Q	2
357 C>G	F119L	2	Del Exon 1	Gross Del	2
362 A>G	D121G	1	Del Exon 1-2	Gross Del	1
364-365			Del Exon 1-3	Gross Del	1
GC>AT	A122I	1	Del Exon 3	Gross Del	4
393 C>A	N131K	1	not found	-	2
394 C>T	Q132X	1			
445-458	N150Sfs	1			
del 14 bp	X19	1			
453 C>G	H151M	1			
461 C>T	P154L	1			

### Location of PNETs in VHL

