

VHL UK/Ireland Fact Sheet



Von Hippel Lindau disease (VHL)

www.vhl-uk-ireland.org
www.vhl.org

- Sites at risk include: kidney, retina, spinal cord, brain, pancreas, adrenal gland and inner ear
- Studies of VHL gene have resulted in approval of multiple cancer drugs
- Particularly in the case of the kidney and pancreas, if not treated correctly, tumours can advance to malignant forms of cancer
- Currently, there are no approved drugs for treating of VHL.
- The VHL mutation is a dominant inherited trait: offspring have a 50% chance of inheriting the mutated VHL gene
- 20% of people with VHL are the first person in their families with this disease
- VHL affects people of all ethnicities in every country of the world
- The prevalence of VHL is approximately one-half of that of cystic fibrosis (1 in 32,000)
- Approximately 10,000 people in the United States are affected by VHL and 200,000 worldwide
- The VHL gene controls the major feeding pipeline of every tumour.
- Curing von-Hippel Lindau brings us one step closer to curing many forms of cancer



Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC)

www.hlrcinfo.org

- If you have HLRCC, it does not mean that you have cancer. It does mean that you are at risk for developing a type of kidney cancer that can be very dangerous
- You can be diagnosed with HLRCC if you have uterine fibroids and cutaneous leiomyoma's (skin bumps) confirmed by a dermatologist's biopsy. You can also be diagnosed with a genetic test
- Screening for HLRCC kidney tumours is essential. Right now it is recommended that individuals with HLRCC are screened once per year with an MRI with contrast
- MRI slices should be 1-3 mm to detect early tumours. (NOTE: HLRCC tumours can metastasize very quickly so catching them early is important.)
- The known prevalence of HLRCC is about 1500 patients worldwide. However it is thought that the condition is very under-diagnosed



Birt-Hogg-Dubé (BHD)

www.bhdsyndrome.org

- Possible symptoms of BHD include:
- Fibrofolliculomas are benign tumours/growths in the hair follicle and appear as skin papules. They are most commonly found on the neck, upper chest, upper back, and face, and can also be found on the ear and earlobe.
- Lung cysts and Spontaneous pneumothorax (collapsed lung).
- Kidney cysts, tumours and kidney cancer
- The prevalence of BHD is about 1 in 200,000

VHL UK/Ireland Support Team (supporting those affected by VHL, HLRCC and BHD)

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