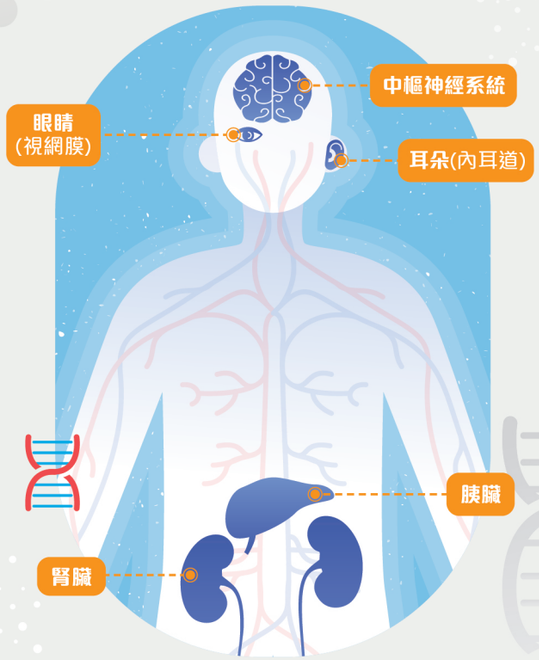
認識罕見病

希佩爾-林道病 (VHL)
(von Hippel-Lindau disease)

VHL會增加病人
在5大主要位置形成腫瘤和囊腫的風險[1]



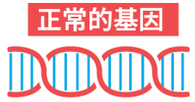
在香港，VHL的患病率估計為

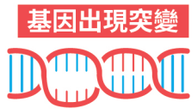
31,000 分之一至 91,000 分之一[1]

本資訊由美國默沙東藥廠有限公司及香港罕見疾病聯盟聯合提供以作公眾教育用途。以上資料只供參考用途，詳情必須向醫生查詢。

甚麼是希佩爾-林道病(VHL)？

VHL病是一種罕見的常染色體顯性遺傳病。由於3號染色體上的**VHL基因出現突變，引致部分器官形成腫瘤和囊腫的風險增加**[1]





- 5大主要位置腫瘤：眼睛、中樞神經系統、耳朵、胰臟、腎臟[1,*]
- 發病年齡：平均為26歲，97%患者會於65歲前發病[1]
- 遺傳模式：

50%機會遺傳給子女[1,†]

VHL的診斷方法

VHL可以透過基因檢測，檢驗3號染色體上的VHL基因有否出現突變。另外，如果符合以下條件，也有機會患上VHL[1,2]

同時驗出VHL致病基因突變或有直屬親屬確診

\+

出現至少一項VHL臨床表徵

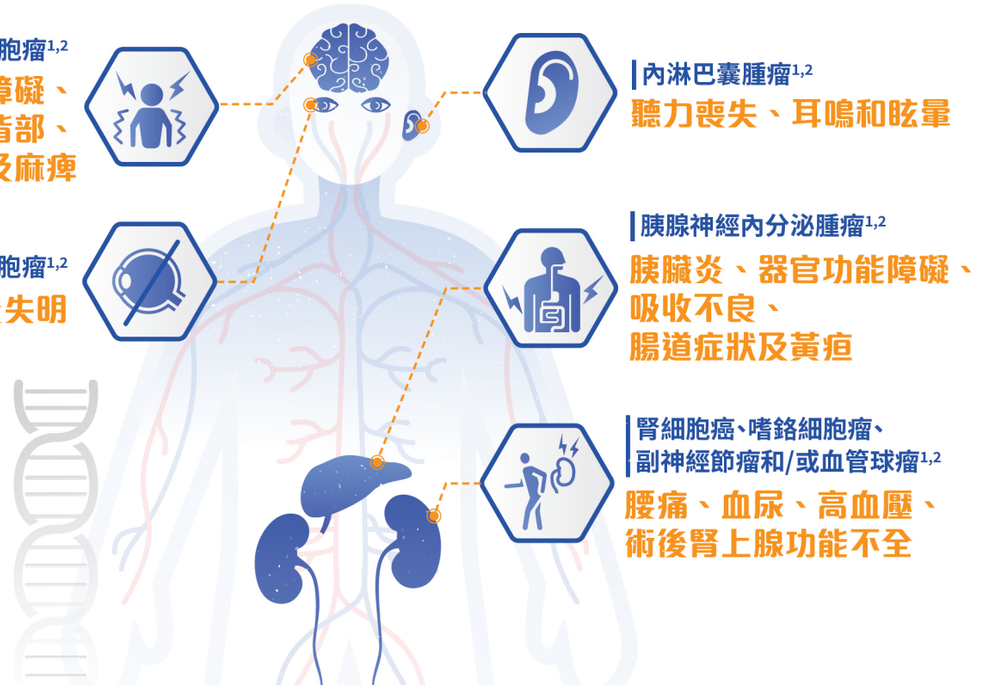
或

出現至少兩項VHL臨床表徵

\+

當中最少一項是血管母細胞瘤

VHL的臨床表徵



中樞神經系統血管母細胞瘤[1,2]
頭痛、協調障礙、眼球震顫、背部、手臂和腿部疼痛及麻痺

視網膜血管母細胞瘤[1,2]
視網膜剝離及失明

內淋巴囊腫瘤[1,2]
聽力喪失、耳鳴和眩暈

胰腺神經內分泌腫瘤[1,2]
胰臟炎、器官功能障礙、吸收不良、腸道症狀及黃疸

腎細胞癌、嗜鉻細胞瘤、副神經節瘤和/或血管球瘤[1,2]
腰痛、血尿、高血壓、術後腎上腺功能不全

*一般單一器官形成腫瘤並不等於患上VHL
†如果父母一方患有VHL

VHL的治療方法

手術切除是治療大多數VHL腫瘤的主要治療方法。不同的腫瘤也可以使用其他治療方法，例如小視網膜血管母細胞瘤可以使用激光治療，一些胰腺神經內分泌腫瘤可以使用化學治療[3]

“請諮詢您的醫生以了解合適的治療方案”

參考資料：

1. Rare Disease Hong Kong. von Hippel–Lindau Disease. Available from: https://rdhk.org/post/data?mid=15&id=13471&lang=en. [Accessed 11 Nov 2024].
2. van Leeuwaarde RS, Ahmad S, van Nesselrooij B, et al. Von Hippel-Lindau Syndrome. 2000 May 17 [Updated 2024 Feb 29]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1463/
3. Binderup MLM. et al. von Hippel-Lindau disease: Updated guideline for diagnosis and surveillance. European Journal of Medical Genetics. 2022:65(8):104538

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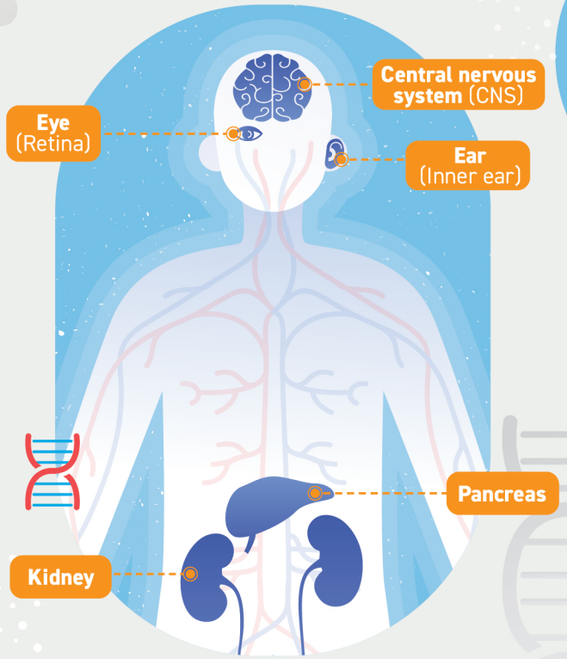
Deciphering the Myths of

VHL DISEASE
(von Hippel-Lindau disease)

VHL disease increases the risk of tumors and cysts in **5 MAJOR SITES** [1]



The prevalence is estimated to be between

1 in 31,000 to 1 in 91,000

individuals in Hong Kong[1]

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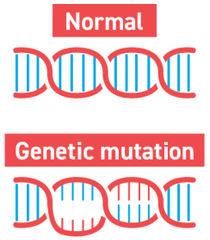
What is VHL disease?

VHL disease is an autosomal-dominant disease characterized by an **increased risk of tumors and cysts in certain organs caused by mutations in the VHL gene on chromosome 3**[1]



- Tumors in 5 main sites: Eye, central nervous system, ear, pancreas and kidney[1,*]
- Age of onset: 26 yo (mean), 97% of patients have symptoms by 65 yo[1]
- Pattern of inheritance:

50% chance of passing on VHL to child[1,†]

Diagnosing VHL disease

Identification of a heterozygous germline VHL pathogenic variant on molecular genetic testing establishes the diagnosis of VHL disease[1]

An individual may be considered to have VHL disease if they meet ⩾1 of the following criteria[1,2]:

Either a pathogenic VHL mutation or a first-degree relative with VHL disease

+

≥1 manifestation of VHL disease

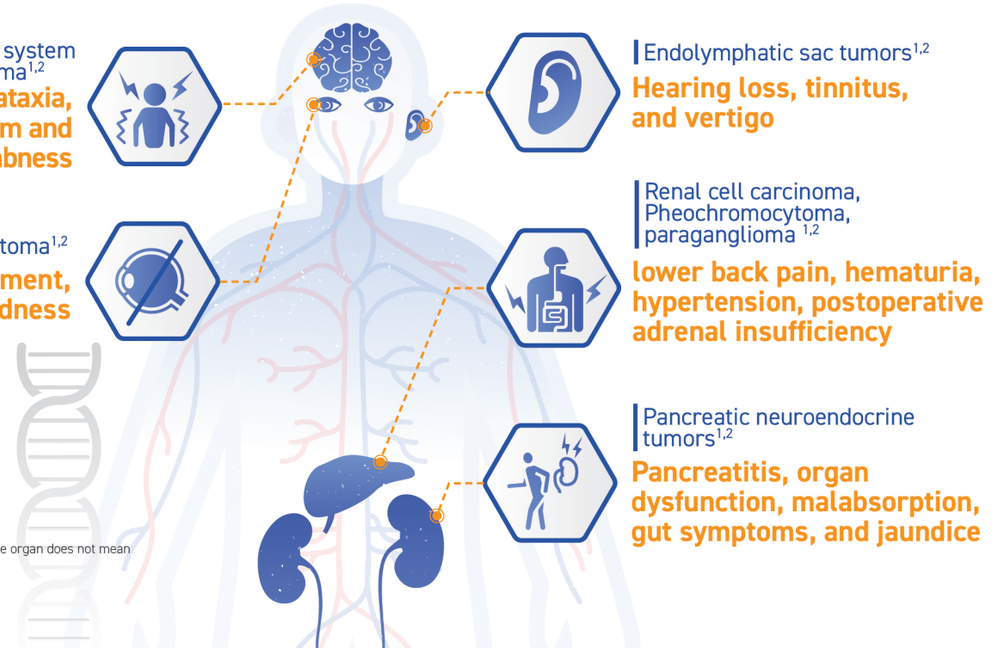
OR

≥2 manifestations of VHLdisease

+

≥1 of which is a hemangioblastoma

VHL disease clinical manifestations



Central nervous system hemangioblastoma[1,2]
Headaches, ataxia, nystagmus, back,arm and leg pain, andnumbness

Retinal hemangioblastoma[1,2]
Retinal detachment, and blindness

Endolymphatic sac tumors[1,2]
Hearing loss, tinnitus, and vertigo

Renal cell carcinoma, Pheochromocytoma, paraganglioma[1,2]
lower back pain, hematuria, hypertension, postoperative adrenal insufficiency

Pancreatic neuroendocrine tumors[1,2]
Pancreatitis, organ dysfunction, malabsorption, gut symptoms, and jaundice

*In general, the formation of tumors in a single organ does not mean that you have VHL disease
†If one parent has VHL disease

Treatment of VHL disease

Supportive care

Surgical removal is the cornerstone in the treatment of most vHL tumours. Choice of treatment may vary with different clinical manifestations: patients with small retinal hemangioblastoma can be treated with laser photocoagulation, some pancreatic neuroendocrine tumors patients can be treated with chemotherapy[3]

Consult your doctor for suitable treatment options

References:

1. Rare Disease Hong Kong. von Hippel–Lindau Disease. Available from: https://rdhk.org/post/data?mid=15&id=13471&lang=en. [Accessed 11 Nov 2024].
2. van Leeuwaarde RS, Ahmad S, van Nesselrooij B, et al. Von Hippel-Lindau Syndrome. 2000 May 17 [Updated 2024 Feb 29]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1463/
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