**PRESS RELEASE**

# CUHK study reveals heavy medical burden of von Hippel–Lindau disease and calls for enhanced multidisciplinary care and support

(Hong Kong, 28 August 2025) – **von Hippel–Lindau (VHL) disease is a rare autosomal dominant hereditary syndrome that can lead to the development of tumours in multiple organs. A study conducted by the Faculty of Medicine at The Chinese University of Hong Kong (CUHK) provides the first analysis of the clinical profile of local VHL patients. The findings reveal that patients experience repeated hospital admissions, surgeries, and dialysis procedures throughout their lives, placing a significant burden on the healthcare system. The researchers also introduced new medications that have evidence to reduce the number of surgeries, and multidisciplinary care to improve overall patient outcomes. The study has been published in the *Hong Kong Medical Journal*.**

The retrospective study reviewed patient records from 1993 to 2024 across five public hospitals, identifying 32 VHL patients. The mean age at first presentation was 28 years. All patients developed tumours, with about 80% having cerebellar haemangioblastoma, over half with renal cell carcinoma, and others with tumours in the spine, retina, pancreas, and more. More than half of the patients underwent surgery, including removal of parts of the kidney, pancreas, and brain, with each patient having an average of three to four surgeries. Nearly half of the patients died during the study period, most due to VHL syndrome.

The study also found that VHL patients incur high medical expenses, particularly due to frequent hospital admissions and surgeries. The annualised per-patient cost for emergency and inpatient admission exceeded HK$138,000. Six patients required dialysis, resulting in a total cost of HK$28.8 million. The researchers emphasise the urgent need to improve the treatment and follow-up care for VHL patients in Hong Kong. VHL has been treated with surgery and radiotherapy to remove tumours, but these procedures can damage organ function, mobility, and vision. A novel treatment is now available that could potentially reduce the number of surgeries—a transformative treatment for VHL patients. At the time of publication, this drug had already been supported by the Samaritan Fund.[1]

Professor Chi-fai Ng, Tzu Leung Ho Professor of Urology of CUHK, explained that VHL patients carry a mutation on chromosome 3, increasing the risk of tumours and cysts in certain organs. If one parent has VHL disease, each child has a 50% of inheriting the condition. ‘VHL patients typically develop the disease at a young age, with the youngest being only 4 months old, and continue to grow tumours throughout their lives. Even after removal, tumours may recur—it’s like

carrying multiple ticking time bombs. Now, with treatment that may reduce the need for surgery and possibly avoid dialysis, there is new hope for patients. It may also help relieve overall healthcare burden.'

Given the VHL syndrome induced tumours may affect various of organs, its management requires collaboration across multiple specialties including but not limited to genetics, urology, neurosurgery, hepatobiliary and pancreatic surgery, oncology, endocrinology, and ophthalmology. The research team recommends a holistic multidisciplinary approach to care, and treatment plans should be discussed at multidisciplinary meetings to prioritise the most life-threatening and symptomatic conditions, with pre- and post-surgery support, timely monitoring, and appropriate follow-up. The team also recommends establishing local VHL registries to facilitate clinical trial enrolment, long-term follow-up and complication screening, and expand patient access to innovative treatments.

The article "Presentation, management, and clinical outcomes of von Hippel–Lindau syndrome" was published in the *Hong Kong Medical Journal*. https://doi.org/10.12809/hkmj2412496

新聞稿

# 中大研究揭示希林氏病沉重醫療負擔　促加強跨專科治療及支援

（香港，2025 年 8 月 28 日） — **希佩爾—林道病（簡稱「希林氏病」）是一種罕見的常染色體顯性遺傳病，可導致多個器官出現腫瘤。香港中文大學（中大）醫學院進行的研究首次詳細分析本港希林氏病患者的臨床情況，揭示患者一生經歷多次入院、手術、洗腎等程序，對醫療系統造成龐大負擔。團隊亦提出有新藥已證明可減少手術次數，並強調跨專科護理的重要性，以提升患者整體照護。研究已刊登於《香港醫學雜誌》。**

研究回顧 1993 至 2024 年間五所公立醫院的患者紀錄，識別出 32 名希林氏病患者。患者平均 28 歲發病，全部均診斷出腫瘤，約八成患者的腫瘤位於腦部、逾半位於腎臟，亦有脊椎、視網膜、胰臟等部位。逾半患者曾接受手術，包括切除部分腎臟、胰臟、腦部等，平均每人要接受三至四次手術。研究期間近半患者離世，大部分由希林氏病相關腫瘤導致。

研究亦發現希林氏病患者的醫療開支高昂，患者頻繁入院及接受手術，急症及住院費用每人每年成本超過 13.8 萬港元。六名患者需要洗腎，洗腎總開支累計達 2880 萬港元。研究團隊認為，本港急切需要改善希林氏病患者的治療和跟進護理。以往希林氏病治療以手術和電療移除腫瘤，但有機會損害患者器官功能、活動能力和視覺。目前有新藥可減少手術次數，是針對希林氏病的革命性藥物，研究發表之時，該藥已納入撒瑪利亞基金援助項目。[1]

中大醫學院外科學系何子樑泌尿科教授吳志輝表示，希林氏病患者的 3 號染色體出現突變，引致部分器官形成腫瘤和囊腫的風險增加，如果父母一方患有希林氏病，有 50%機會遺傳給子女。「希林氏病患者發病時普遍相當年輕，年紀最輕僅得 4 個月大，終其一生不斷生長腫瘤，切除後仍可能再出現，就像揹着多個計時炸彈。現時有藥物有望減少手術或能避免洗腎，對患者是新的希望，亦有機會減少整體醫療成本。」

希林氏病患者不同器官的腫瘤涉及多個專科，包括遺傳科、泌尿外科、神經外科、肝膽胰外科、臨床腫瘤科、內分泌科、眼科等。研究團隊建議就每位患者以跨專科會議商討治療方案，優先著手最危急的病況，配合手術前後的支援，以及適時監察病情變化及作適切跟進。團隊亦建議探討成立本地希林氏病患者登記制度，以便日後更多臨床研究、長期跟進和篩查不良反應，並制定政策讓患者更容易負擔創新藥物。

詳細內容可參閱原文《希林氏病的臨床表現、治療及結果》。

HONG KONG MEDICAL JOURNAL 香港醫學雜誌

**Reference**

1. Samaritan Fund. Items supported by the Samaritan Fund. Available from: https://www.ha.org.hk/haho/ho/sf/SF_Items_en.pdf. Accessed 18 Aug 2025.

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Source: Athena YH Lee, David KW Leung, CH Leung, et al. Presentation, management, and clinical outcomes of von Hippel–Lindau syndrome. Hong Kong Med J 2025;31:Epub 28 Aug 2025. https://doi.org/10.12809/hkmj2412496.