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解碼肥厚的心：肥厚性心肌病變、法布瑞氏症與心肌類澱粉沉積症的最新臨床進展

Decoding the Thickened Heart: A Contemporary Update on Hypertrophic Cardiomyopathy, Fabry Disease, and Cardiac Amyloidosis

時　　間：115 年 6 月 27 日(星期六) 13:00~17:15
地　　點：臺北榮民總醫院 致德樓第四會議室

13:00-13:05	***Opening Remarks***	余文鍾主任 Wen-Chung Yu
	座長：余文鍾 主任 (Wen-Chung Yu)	
13:05-13:35	巔峰對話：肥厚性心肌病變的現代治療新視界 At the Forefront: Modern Management of Hypertrophic Cardiomyopathy	Martin S. Maron (美國)
13:35-13:40	***Q&A***	余文鍾主任 Wen-Chung Yu
	座長：王俊力 教授 (Chun-Li Wang)	
13:40-14:10	心牆背後的真相：左心室肥大的診斷迷霧 Behind the Thickened Wall: Deciphering LVH in Cardiomyopathy	郭泠醫師 Ling Kuo
	座長：洪崇烈 教授 (Chung-Lieh Hung)	
14:10-14:40	偉大的模仿者：法布瑞氏症—臺灣實戰經驗 The Great Imitator: Facing Fabry Disease - Taiwan Experience	張皓智醫師 Hao-Chih Chang
14:40-15:00	***Panel Discussion: All speakers & moderators*** 蔡惟全 Wei-Chuan Tsai、黃群耀 Chun-Yao Huang	
15:00-15:20	***Healthy Break***	
	座長：黃群耀 教授 (Chun-Yao Huang)	
15:20-15:50	站在診斷的十字路口：心臟類澱粉沉積症概論 At the Crossroads: A Comprehensive Overview of Cardiac Amyloidosis	陳美綾醫師 Mei-Ling Chen

座長：洪明銳 教授 (Ming-Jui Hung)

15:50-16:20	向右走：轉運蛋白類澱粉心肌病變的新藥策略 The Right Path: Navigating the Modern Therapies of ATT "R" Cardiomyopathy	林彥宏教授 Yen-Hung Lin

座長：黃聖懿 醫師 (Shang-Yi Huang)

16:20-16:50	向左走：輕鏈類澱粉心肌病變的多專科治療 The Left Path: Racing Against Time in A "L" Cardiomyopathy Management	蔡淳光醫師 Chun-Kuang Tsai
16:50-17:10	***Panel Discussion: All speakers & moderators*** 賴志泓 Chih-Hung Lai、王玟樺 Wen-Hwa Wang	
17:10-17:15	***Closing Remarks***	余文鍾主任 Wen-Chung Yu

At the forefront: Modern management of hypertrophic cardiomyopathy

巔峰對話：肥厚性心肌病變的現代治療新視界

Martin S. Maron

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Hypertrophic cardiomyopathy (HCM) has evolved into a highly treatable disease with near-normal longevity. This lecture reviews contemporary paradigms in risk stratification, imaging, and therapeutics reshaping its natural history. Sudden cardiac death (SCD) prevention relies on ACC/AHA guidelines. Key markers—family history of SCD, unexplained syncope, massive LVH, apical aneurysms, and sensitivity in risk stratification. Furthermore, CMR is indispensable; an LGE scar burden of ≥15% serves as a critical threshold guiding ICD decisions. Distinguishing obstructive from non-obstructive phenotypes is paramount. Upright exercise echocardiography remains the gold standard, revealing that nearly 70% of patients exhibit dynamic LVOT obstruction. For symptomatic obstructive patients, while myectomy and alcohol ablation remain excellent options, cardiac myosin inhibitors have revolutionized medical therapy. Mavacamten and the next-generation agent Aficamten robustly reduce gradients, improve peak VO2, and down-refer patients from invasive interventions. Notably, Aficamten features pharmacological advantages, including a shorter half-life for faster titration, fewer drug interactions, and less transient systolic dysfunction. Lastly, the lecture highlights a low threshold for early anticoagulation in HCM patients with atrial fibrillation, independent of CHA2DS2-VASc scores, to mitigate stroke risk. Future horizons, including multi-drug targeted therapies for non-obstructive HCM (metabolic modulators, SGLT2 inhibitors) and AI to refine diagnostic specificity, are also discussed to optimize global survival.

Behind the thickened wall: Deciphering LVH in cardiomyopathy

心牆背後的真相：左心室肥大的診斷迷霧

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Left ventricular hypertrophy (LVH) represents a common yet diagnostically challenging phenotype encountered in contemporary cardiology. While hypertrophic cardiomyopathy (HCM) remains the most recognized cause of unexplained LVH, several infiltrative and phenocopy conditions, including cardiac amyloidosis and Fabry cardiomyopathy can present with similar morphologic features but substantially different prognostic and therapeutic implications. Other differential diagnosis including. This lecture, "Behind the Thickened Wall: Deciphering LVH in Cardiomyopathy," will provide a practical multimodality imaging–based approach to the differential diagnosis of LVH, emphasizing the integration of clinical clues, electrocardiography, echocardiography, cardiovascular magnetic resonance (CMR), nuclear imaging, and emerging artificial intelligence applications.

The great imitator: Facing Fabry Disease – Taiwan experience

偉大的模仿者：法布瑞氏症—臺灣實戰經驗

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Fabry disease is increasingly recognized in Taiwan and is no longer considered exceptionally rare. Data from nationwide newborn screening programs identified approximately 1 affected male infant in every 1,600 births. The predominant genotype in Taiwan is the IVS4+919G>A mutation, which is associated with a late-onset phenotype characterized predominantly by cardiac involvement. With growing awareness among cardiologists, an increasing number of patients with unexplained left ventricular hypertrophy (LVH) have been diagnosed with Fabry disease. Fabry cardiomyopathy may present with asymmetric LVH, closely mimicking sarcomeric hypertrophic cardiomyopathy (HCM), thereby posing substantial diagnostic challenges in daily clinical practice. At Taipei Veterans General Hospital, we have established the largest adult Fabry disease cohort in Taiwan. This prospective observational cohort includes more than 300 patients with longitudinal follow-up extending beyond 10 years. The cohort encompasses asymptomatic mutation carriers as well as patients with overt cardiac involvement, with and without enzyme replacement therapy (ERT).

In this lecture, I will highlight the Taiwan experience in Fabry disease, focusing on the overlapping phenotypic features between Fabry cardiomyopathy and HCM, including similarities in morphology and cardiovascular risk profiles. I will also discuss the early detection of subclinical cardiac involvement using speckle-tracking echocardiography, refined risk stratification based on LVH severity and myocardial mechanical function, and pragmatic imaging tools for monitoring treatment responsiveness during ERT in relation to the underlying inflammatory and fibrotic myocardial status. These findings may help establish a more precise risk stratification framework for Fabry cardiomyopathy, analogous to the well-established approach used in HCM, while also providing practical guidance for the management and longitudinal monitoring of patients undergoing lifelong ERT.

At the crossroads: A comprehensive overview of cardiac amyloidosis

站在診斷的十字路口：心臟類澱粉沉積症概論

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Cardiac amyloidosis has emerged from a historically underrecognized disorder to a major cause of heart failure, arrhythmia, and increased mortality, particularly among older adults. Advances in multimodality imaging, biomarker assessment, genetic testing, and disease-modifying therapies have transformed the diagnostic and therapeutic landscape, placing clinicians “at the crossroads” of evolving diagnostic algorithms and expanding treatment opportunities. This lecture provides a comprehensive overview of cardiac amyloidosis, with a focus on the two predominant subtypes: light-chain (AL) amyloidosis and transthyretin amyloidosis (ATTR), including both hereditary and wild-type forms.

This presentation will review the pathophysiology and clinical manifestations of cardiac amyloid infiltration, emphasizing red-flag features that facilitate early recognition in patients presenting with unexplained left ventricular hypertrophy, heart failure with preserved ejection fraction, conduction abnormalities, or extracardiac manifestations. Contemporary diagnostic strategies integrating echocardiography, cardiac magnetic resonance imaging, nuclear scintigraphy, biomarkers, and tissue confirmation will be discussed, alongside practical approaches to differentiating AL from ATTR amyloidosis.

The right path: Navigating the modern therapies of ATT “R” cardiomyopathy

向右走：轉運蛋白類澱粉心肌病變的新藥策略

Yen-Hung Lin

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Transthyretin amyloid cardiomyopathy (ATTR-CM) is caused by myocardial deposition of misfolded transthyretin protein. It is classified into 2 groups by the genetics of Transthyretin amyloidosis (ATTR): wild-type (ATTRwt) or hereditary (hATTR or ATTRm). ATTR-CM, irrespective of genotype, is an unrecognized mechanism underlying HFpEF. It was reported wild-type TTR might be an underdiagnosed cause of HFpEF. However, the prevalence of wild-type ATTR among patients with HFpEF is not well-established in Taiwan and Asia.

In recent years, various pharmacological treatments have been investigated, focusing on different therapeutic targets. These targets include reducing transthyretin production, preventing the dissociation of the transthyretin tetramer, and the depleting transthyretin amyloid deposits.

Transthyretin stabilizers including tafamidis and acoramidis, have been investigated for their efficacy in preventing the dissociation of transthyretin tetramers into monomers, thereby mitigating the progression of ATTR-CM. These medications were studied in in Tafamidis in Transthyretin Cardiomyopathy Clinical Trial (ATTR-ACT) and Efficacy and Safety of acoramidis in Subjects With Transthyretin Amyloid Cardiomyopathy (ATTRibute-CM). Transthyretin mRNA silencers, including patisiran and vutrisiran, function as siRNA to inhibit the transcription of transthyretin mRNA into protein, thereby delaying the progression of ATTR-CM. Patisiran was studied in the Patisiran Treatment in Patients with Transthyretin Cardiac Amyloidosis (APOLLO-B) trial8, and vutrisiran in the Patients with Transthyretin Amyloidosis with Cardiomyopathy (HELIOS-B) trial.

The speech will cover the rationale, major clinical trials, current recommendation, and future perspectives in ATTR-CM treatment.

The left path: Racing against time in a “L” cardiomyopathy management

向左走：輕鏈類澱粉心肌病變的多專科治療

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Systemic light-chain (AL) amyloidosis with cardiac involvement is a hematologic emergency in which delayed diagnosis and treatment markedly worsen survival. Patients often present with heart failure, unexplained ventricular wall thickening, edema, renal dysfunction, neuropathy, or multisystem symptoms that mimic common cardiovascular diseases. Management therefore depends on rapid recognition, urgent confirmation of diagnosis, and immediate therapy. A multidisciplinary approach involving cardiology, hematology, nephrology, pathology, and supportive care teams is essential to shorten time to treatment and prevent irreversible organ damage. Diagnostic evaluation should include serum and urine monoclonal protein studies, serum free light chain assay, cardiac biomarkers, echocardiography, and tissue biopsy with Congo red staining followed by definitive amyloid typing. Cardiac magnetic resonance imaging may further support diagnosis when clinically feasible. Once AL amyloidosis is confirmed, prompt plasma cell–directed therapy is critical. Daratumumab-based regimens, particularly daratumumab, bortezomib, cyclophosphamide, and dexamethasone, have become preferred frontline treatment because they can induce rapid and deep hematologic responses, which are especially important in advanced cardiac disease. However, treatment must be individualized, as fragile patients may require reduced dosing, sequential drug introduction, and close monitoring for hypotension, arrhythmia, infection, neuropathy, and worsening heart failure. Supportive management differs from standard heart failure care and often relies on careful diuresis, salt and fluid restriction, rhythm surveillance, and cautious use of conventional heart failure medications because many are poorly tolerated. Serial monitoring of free light chains, NT-proBNP, renal function, and clinical status is necessary to guide response-adapted care. In summary, successful management of “L” cardiomyopathy requires racing against time through early suspicion, rapid diagnosis, immediate clone-directed therapy, and coordinated multidisciplinary care.