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神經醫學診療的新紀元：挑戰與突破

A New Era in Neurological Medicine: Challenges and Breakthroughs

時　　間：114 年 6 月 29 日(星期日) 08:45~12:10
地　　點：臺北榮民總醫院 致德樓第一會議室

08:45-08:50	***Opening Remarks***	王署君副院長 Shuu-Jiun Wang
	座長：賴資賢 主任 (Tzu-Hsien Lai)	
08:50-09:15	偏頭痛治療更新：CGRP 治療與性別考量 Migraine Management Update: CGRP Therapies and Sex Considerations	楊富吉主任 Fu-Chi Yang
09:15-09:20	Q&A	
	座長：傅中玲 主任 (Jong-Ling Fuh)	
09:20-09:45	阿茲海默症診斷與治療的最新進展 An Update on the Recent Progress in the Diagnosis and Treatment of Alzheimer's Disease	李威儒主任 Wei-Ju Lee
09:45-09:50	Q&A	
	座長：黃文成 主任 (Wen-Cheng Huang)	
09:50-10:15	腦出血之外科介入 The Role of Surgical Intervention for Intracerebral Hemorrhage	李政家醫師 Cheng-Chia Lee
10:15-10:20	Q&A	
10:20-10:30	***Coffee Break***	
	座長：陳柏霖 主任 (Po-Lin Chen)	
10:30-10:55	基因診斷在癲癇精準醫療的角色 The Role of Genetic Diagnosis in Precision Medicine of Epilepsy	劉祐岑主任 Yo-Tsen Liu
10:55-11:00	Q&A	
	座長：李宜中 教授 (Yi-Chung Lee)	
11:00-11:25	ALS 致病基因與治療的最新進展 New Frontiers in ALS Genetics and Treatments	季康揚醫師 Kang-Yang Jih
11:25-11:30	Q&A	

座長：劉康渡 主任 (Kang-Du Liou)

11:30-11:55	巴金森病的藥物與手術治療的最新進展 Pharmacological and Surgical Treatment of PD: An Update	陳俊宇醫師 Chun-Yu Chen
11:55-12:00	Q&A	
12:00-12:10	***Closing Remarks***	傅中玲主任 Jong-Ling Fuh

Migraine management update: CGRP therapies and sex considerations

偏頭痛治療更新：CGRP 治療與性別考量

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Migraine is a highly prevalent and disabling neurological disorder, characterized by a pronounced female preponderance and influenced by complex pathophysiological mechanisms. This presentation provides an updated overview of migraine biology, highlighting the pivotal role of calcitonin gene-related peptide (CGRP) in the onset and progression of migraine attacks. It addresses the evolution of migraine therapies, from traditional acute treatments—including triptans, analgesics, and nonsteroidal anti-inflammatory drugs—to innovative preventive strategies such as botulinum toxin type A and CGRP-targeted medications. Particular attention is given to the latest CGRP-related therapies, encompassing both monoclonal antibodies and small-molecule CGRP receptor antagonists. These novel agents show promise in reducing the frequency and intensity of migraine episodes, particularly among individuals who have not responded to or cannot tolerate conventional medications. Emerging evidence suggests that hormonal fluctuations—especially variations in estrogen and prolactin levels—may heighten migraine susceptibility in women. Clinical trial data underscore the potential for greater efficacy in females receiving CGRP-targeting therapies, with some differences observed in male patients. Consequently, personalized treatment approaches are essential, taking sex-specific responses and comorbidities into account when selecting therapy options.

An update on the recent progress in the diagnosis and treatment of Alzheimer's disease

阿茲海默症診斷與治療的最新進展

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Alzheimer's disease (AD) is a progressive neurodegenerative disorder characterized by cognitive decline and memory impairment, significantly impacting patients' quality of life and imposing a substantial burden on healthcare systems worldwide. Recent advancements in the diagnosis and treatment of AD have shown promising potential to enhance early detection and improve patient outcomes.

In diagnostics, blood-based biomarkers have emerged as a revolutionary approach, offering minimally invasive and cost-effective options for early detection and disease monitoring. Additionally, amyloid and tau positron emission tomography (PET) imaging has demonstrated high specificity and sensitivity in detecting pathological hallmarks of AD, enabling accurate diagnosis even in the preclinical stages. The integration of these advanced diagnostic techniques has significantly improved early identification and disease staging.

On the therapeutic front, the development of amyloid antibody treatments has marked a breakthrough in AD management. Monoclonal antibodies targeting amyloid-beta, such as Aducanumab and Lecanemab, have demonstrated the ability to reduce amyloid plaques and potentially slow cognitive decline. These therapies, combined with supportive care and lifestyle modifications, are shaping a more comprehensive and personalized approach to AD treatment.

This talk will provide an update on the recent progress in AD diagnosis and treatment, emphasizing the impact of blood-based biomarkers, amyloid and tau PET imaging, and amyloid antibody therapies on clinical practice.

The role of surgical intervention for intracerebral hemorrhage

腦出血之外科介入

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Minimally invasive hematoma evacuation techniques, such as stereotactic aspiration and endoscopic surgery, are increasingly used to treat intracerebral hemorrhage (ICH), especially in patients who may not be suitable candidates for traditional craniotomy. These methods offer the advantage of smaller incisions, reduced brain tissue disruption, and faster recovery times compared to open surgery. The primary goal of these procedures is to evacuate the hematoma, alleviate intracranial pressure (ICP), and improve neurological outcomes while minimizing surgical risks.

Stereotactic aspiration involves using a small-bore catheter inserted through the skull into the hematoma cavity, guided by stereotactic imaging or navigation systems. The process begins with preoperative imaging (CT or MRI) to identify the hematoma's location. Once the catheter is in place, the hematoma is aspirated through the catheter, often using a vacuum or syringe. This approach is particularly useful for deep-seated or small hematomas and is associated with lower complication rates, such as reduced risk of infection or brain tissue damage.

Endoscopic evacuation, on the other hand, involves inserting a rigid or flexible endoscope through a small incision. The surgeon visualizes the hematoma directly on the monitor and evacuates it using specialized instruments. This technique provides real-time imaging and is effective for more superficial hematomas, such as those in the basal ganglia or frontal lobe. Endoscopic surgery is advantageous in that it allows precise removal of the clot while preserving surrounding brain tissue, potentially improving recovery outcomes.

While minimally invasive techniques offer several benefits, they also have limitations. They may not be suitable for large hematomas or those located in inaccessible brain regions. In some cases, complete evacuation may not be achievable, requiring follow-up procedures or conversion to traditional surgery. Nonetheless, for appropriately selected patients, these approaches can significantly reduce surgical risks and shorten recovery times.

The role of genetic diagnosis in precision medicine of epilepsy

基因診斷在癲癇精準醫療的角色

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Epilepsy is one of the most common neurological diseases with more than 60 million people bearing the diagnosis worldwide. The rapid advancement of sequencing technology has led to the unraveling of genetic factors to be the underlying cause of diverse epilepsy syndromes in the past decade. Since the discovery of the first epilepsy gene, *CHRNA4*, nearly 1000 genes have been reported to be associated with epilepsy. The latest 2017 classification of epilepsy proposed by International League Against Epilepsy has adopted "genetic" as one of the main categories of epilepsies etiologies.

Genetic testing now plays a pivotal role in the clinical management of patients with epilepsy. Accurate genetic diagnosis may guide treatments such as disease-modifying therapies and/or the selection of antiseizure medications known to be effective or ineffective in certain epilepsy syndromes. Genetic diagnosis may also help to prognosticate and limit further investigations that have associated risks and cost. Further, genetic diagnoses may help identify or anticipate potential co-morbidities, allowing for optimization of treatment. With appropriate genetic diagnosis, genetic counseling for future pregnancies may be possible.

All kinds of genetic aberrations, including single nucleotide variants, copy number variations, chromosomal rearrangements, and mitochondrial diseases, are involved in diverse epilepsy syndromes. It is challenging to choose the proper genetic test for different phenotypes. Furthermore, it may be difficult to determine the clinical significance of a rare genomic variant. The interpretation and delivery of the genetic test results would generate significant impacts on the patients and their family. There are potential concerns about genetic testing, including psychological distress, social stigma and problems with health and life insurance.

I will share the experience of genetic diagnosis at Taipei Veterans General Hospital, a tertiary referral center in Taiwan. I will also introduce the general guidance on the circumstances in which genetic testing is indicated and test selection in order to guide optimal test appropriateness and benefit proposed by Taiwan Epilepsy Society.

New frontiers in ALS genetics and treatments

ALS 致病基因與治療的最新進展

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Background: Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative disease that involves degeneration of both upper and lower motor neurons. ALS patients typically experience progressive weakness in their limbs and later developed swallowing difficulties and respiratory failure. The average survival time of ALS is 3 to 5 years since disease onset. Disease-associated mutations can be found in approximately 15% of the ALS patients. However, the heredity of ALS was estimated to be around 40 to 60% based on analysis of twin data and a national registry. The discrepancy in ALS heredity has long been an enigma. We aimed to investigate the role of short tandem repeats in ALS genetics

Methods: We recruited 649 patients diagnosed with definite or probably ALS. All patients were tested for common disease genes, tandem repeat expansions and detail clinical characteristics were acquired after informed consent. The patients were followed up biannually for evaluation of ALSFRS-R score. 292 of them have received biannually ALSFRS-R score evaluations.

Results: The male and female ratio is 1.46. 12% of the patients exhibited bulbar onset. Average diagnosis delay since symptoms onset was 13.9 months. The most common genetic causes are *SOD1* and *C9ORF72*. On average, the functional outcome, evaluated by ALSFRS-R score, decline from 33.8 to 22.8 over the first year of diagnosis. Age of onset, presence of disease-causing genes and gender did not affect the rate of functional decline. Initial ALSFRS-R score, bulbar onset, BMI and older age of onset resulted in worse survival outcome. Serum neurofilament light chain level is inversely correlated to the first ALSFRS-R score.

Conclusion: Identification of genetic variants in ALS patients could lead to access of breakthrough genetic therapies. In addition to indel variants, tandem repeat expansions play an important role in ALS genetics.

Pharmacological and surgical treatment of Parkinson's disease: An update

巴金森病的藥物與手術治療的最新進展

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Parkinson's disease (PD) is a progressive neurodegenerative disorder characterized by both motor and non-motor symptoms due to dopamine depletion in the basal ganglia. While no cure exists, current treatment strategies aim to alleviate these symptoms and address complications arising from disease progression and long-term therapy.

Pharmacological treatments remain the cornerstone of PD management, with levodopa providing the most effective symptomatic relief. However, its long-term use is associated with motor fluctuations and dyskinesias, necessitating adjunctive therapies such as dopamine agonists, monoamine oxidase-B inhibitors, and catechol-O-methyltransferase inhibitors. Extended-release formulations, infusion therapies, and novel delivery systems have been developed to enhance drug efficacy and minimize side effects.

For patients with advanced PD experiencing motor fluctuations and medication-refractory symptoms, surgical interventions offer effective treatment options. Deep brain stimulation has revolutionized the management of PD by providing sustained symptom relief, improving motor function, and reducing medication requirements. More recently, high-intensity focused ultrasound has emerged as a non-invasive alternative for patients with tremor-dominant PD.

This talk will provide an in-depth exploration of the latest evidence supporting optimal PD management.