第 14 屆彰化基督教醫院國際基因體醫學研討會-罕見疾病臨床診斷治療及照護

會議時間：2020 年 12 月 26 日（星期六）

會議地點：彰基國際培訓中心望愛廳(彰化縣彰化市建寶街 20 號 B1)

主辦單位：彰化基督教醫院 罕病防治中心，血管暨基因體研究中心，粒線體醫學暨自由基研究院

協辦單位：澳亞醫學科學研究學會台灣分會、台灣神經罕見疾病學會

繼續教育積分：台灣小兒神經醫學會 5 點，台灣神經學學會 2 學分，台灣遺傳諮詢學會 7.1 積分，中華民國人類遺傳學會 5.7 小時，臺灣諮商心理學會 8.1 積分，營養師繼續教育課程積分 專業 7.4 點/專業品質 0.7 點，護理師繼續教育課程積分 8.1 積分，專科護理師繼續教育課程積分 8.1 積分，醫事檢驗師繼續教育積分 8.4 積分，西醫師繼續教育 8.1 積分

2020.12.1 起，出入八大類場所應佩戴口罩

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【指揮中心醫療應變訊息-疾管署】2020.12.22

為因應 COVID-19(武漢肺炎)疫情發展及秋冬防疫專案實施，指揮中心調整「醫療照護工作人員 COVID-19 擴大採檢及個案處理流程」，以強化醫療院所針對醫療照護人員之健康監測。擴大採檢個案比照社區監測通報採檢，採檢後不再列入自主健康管理對象，於退燒超過 24 小時（未使用退燒藥）且相關症狀緩解後，不需等待檢驗結果陰性，即可返回上班。

配合前述流程修訂，另訂定「醫療照護工作人員 COVID-19 擴大採檢後注意事項」，擴大採檢個案於採檢後 3 日內（採檢次日為第 1 日）或所有症狀尚未完全消失前，避免與嚴重免疫功能低下之病人或服務對象接觸。

相關指引已更新並公布於衛生福利部疾病管制署全球資訊網/嚴重特殊傳染性肺炎/醫療照護機構感染管制相關指引項下，供各界參考依循

各位參加研討會之人員，配合政府規定量體溫，戴口罩，勤洗手，若體溫超過 37.5℃將無法入場(休息 5 分鐘再測量第二次)。

緣起：

彰化基督教醫院罕病團隊關懷照顧及研究始於 2010 年神經內科罕病特別門診及基因醫學部擴大臨床基因檢測，分別為劉青山博士及陳明博士加入彰基服務團隊，因此展開罕見疾病病患之全人醫療服務，陸續成立彰基罕病關懷及研究團隊、遺傳諮詢中心、臨床心理諮商中心、罕病防治中心，罕病照護委員會；106 年 12 月開始承接國民健康署「罕見疾病照護服務計畫」，從關懷病人與家屬之角度，以個案管理模式，依據「罕見疾病及罕見遺傳疾病缺陷照護服務辦法」所定服務範圍及方法，不止提供衛服部核准之罕見疾病個案及家屬相關服務，同時亦提供尚未核准之罕病個案及家屬相關服務，期能達到政府「罕見疾病防治及藥物法」、「安寧緩和法」及「病人自主權利法」、加強照顧罕見疾病病人及家屬、防治罕見疾病發生之目的。

本計畫執行國健署派案之院內外個案，經過罕見疾病病人或其法定代理人同意後，由本院個管師及相關專業醫師及醫事人員評估，瞭解病人及家屬是否有需疾病之瞭解、心理支持、生育關懷、照護諮詢等四大服務之需求，進而由個管或相關醫師及醫事針對其需求提供服務或轉介。同時針對他院之罕病病患患者及家屬提供電話關懷及諮商，亦提供非本院罕病照顧醫師有關之電子報及最新罕病消息或教育課程，同時每月提供罕病照護團隊之教育課程，因著重視照護關懷成效，亦成立彰基罕見疾病關懷小組。

會議目的：

依衛福部國健署計畫罕見疾病照護服務計畫，提供罕見疾病者照護服務，涵蓋全國之罕見疾病患者，建立四大服務目標，我們會議之主軸也將放在

(1)醫療團隊如何提供病患及家屬有關疾病之瞭解

(2)醫療團隊如何提供病人及家屬有關心理層面支持

(3)醫療團隊如何提供病人及家屬有關生育關懷之諮商

(4)醫療團隊如何提供病人及家屬有關照護諮詢

本次的研討會，也會邀請研究學者及醫療院所代表，讓大家群聚在這專業的平台上，透過專家分享彰基及其他院所之罕病關懷、治療、遺傳諮詢、緩和醫療、營養、運動、生涯規劃。

研討會籌辦委員：

Congress President:

陳穆寬 教授 彰化基督教醫院院長

Congress Vice President:

劉青山 教授 彰化基督教醫院副院長

陳明 教授 彰化基督教醫院醫療長

Congress Secretary-General:

張瑞芝 博士 彰化基督教醫院 血管暨基因體研究中心副主任

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Chairpersons:

魏耀揮 教授 彰化基督教醫院

蘇矢立 主任 彰化基督教醫院

趙美琴 教授 彰化基督教醫院

蔡玲貞 主任 彰化基督教醫院

Organizers:

彰化基督教醫院 罕病防治中心，血管暨基因體研究中心，粒線體醫學暨自由基研究院

Co-Organizers:

澳亞醫學科學研究學會台灣分會

台灣神經罕見疾病學會

※會議議程：

時間	題目	講員	引言人
9:00-9:10	歡迎詞	彰基 陳穆寬院長/彰基 魏耀揮教授	
9:10-9:40	神經肌肉罕病學思歷程	高醫 鐘育志校長	彰基 趙美琴教授
9:40-10:10	罕見疾病照護在奇美	奇美 林秀娟教授	彰基 楊仁宏教授
10:10-10:40	彰基/高醫罕見疾病之分享	彰基 趙美琴教授	彰基 李佳儒醫師
10:40-10:55	休息(大合照)		
10:55-11:25	台北榮總神經罕病之照護及研究	北榮 李宜中教授	彰基 陳大成主任
11:25-11:55	高醫治療神經肌肉疾病之分享	高醫 梁文貞教授	高醫 鐘育志校長
11:55-12:25	台大罕見疾病之分享	臺大 李妮鍾教授	彰基 吳鴻明教授
12:25-13:00	午餐/演講 Suppression of Mitophagy by maladaptive cell signaling pathways in the progression of mitochondrial disease (Michael R Duchen, Ph.D; Department of Cell and Developmental Biology, University College London, UK.)		彰基 魏耀揮教授
12:25-13:00	澳亞醫學科學研究學會台灣分會第三屆第四次理監事會(尊榮 A 廳)		
13:00-13:30	澳亞醫學科學研究學會台灣分會第三屆第一次會員大會(尊榮 A 廳)		
13:30-13:50	罕見疾病基金會的使命與任務	陳冠如執行長	彰基 張明裕主任
13:50-14:10	海洋性貧血的過去、現在與未來	彰基 王士忠主任	彰基 趙美琴教授
14:10-14:30	營養學與神經退化疾病	中山醫大劉凱莉教授	彰基 張瑞芝主任
14:30-14:50	中醫在罕見疾病治療的角色	中國醫大謝慶良教授	彰基 邱重閔醫師
14:50-15:10	可治療性之小兒神經罕病	彰基 張通銘主任	義大 李介元醫師
15:10-15:25	休息		
15:25-15:40	罕病照護計畫成果分享	彰基 蔡玲貞主任	奇美 林秀娟教授
15:40-15:55	小兒罕病之復健醫學	彰基 廖淑芬主任	彰基 劉森永主任
15:55-16:10	以 Raxone (Idebenone)治療雷伯氏視神經萎縮症之臨床經驗	彰基 陳彥廷醫師	彰基 劉青山教授
16:10-16:25	無症狀罕見疾病患者心理諮商	彰基 蕭真真主任	彰基 張正辰主任
16:25-16:40	光線與健康-遠紅外線	中山醫大王祖興教授	中山醫大劉凱莉教授
16:40-16:55	McCune Albright 氏症候群分享	彰基 吳怡磊主任	彰基 蘇矢立主任
16:55-17:10	Q&A 賦歸(學分簽退)		彰基 劉青山教授
17:30	晚宴-怡園會館 2F 茉莉廳(講員與座長)		

鐘育志, MD,DM Sci

EDUCATION

1993	D. M. Sci.	Tokyo Women's Medical University, Japan
1984	Master of Medicine	Kaohsiung Medical College, Taiwan
1979	Doctor of Medicine	Kaohsiung Medical College, Taiwan

CURRENT POSITION & PROFESSIONAL BACKGROUND

2018/8- present	President, Kaohsiung Medical University (KMU), Kaohsiung, Taiwan
2017/6- present	President, Taiwan SMA Families, Kaohsiung, Taiwan
2015/8- present	Professor, Graduate Institute of Clinical Medicine, College of Medicine, KMU
1984/8- present	Visiting Staff, Departments of Pediatrics and Laboratory Medicine, KMU Hospital, Kaohsiung, Taiwan

RESEARCH INTEREST(S)

- Pediatrics
- Pediatric Neurology
- Neuromuscular Disease

Honors and Awards(or Selected Publications)

Awards:

Taiwan Child Neurology Society Award (2020)

Taiwan Pediatric Association Award (2013)

Award for Contribution to Pediatric Education (2011)

The 3rd National Golden Palm Tree Award (2005)

Selected Publications:

1. Po-Jui Hsu; Horng-Dar Wang; Yung-Che Tseng; Shao-Wei Pan; Bonifasius Putera Sampurna; Yuh-Jyh Jong*; Chiou-Hwa Yuh*. L-Carnitine Ameliorates Congenital Myopathy in a Tropomyosin 3 de novo Mutation Transgenic Zebrafish. J Biomed Sci 2020 (In Press).
2. Shan-Fu Ou, Che-Sheng Ho, Wang-Tso Lee, Kuang-Lin Lin, Cynthia C Jones, Yuh-Jyh Jong*, SMA Study Group. Natural history in spinal muscular atrophy Type I in Taiwanese population: A longitudinal study. Brain Dev 2021;43(1):127-34. (SCI).
3. Wen-Chen Liang, Yuh-Jyh Jong*, Chien-Hua Wang, Chen-Hua Wang, Xia Tian, Wan-Zi Chen, Tzu-Min Kan, Narihiro Minami, Ichizo Nishino, Lee-Jun C Wong. Clinical, pathological, imaging, and genetic characterization in a Taiwanese cohort with limb-girdle muscular dystrophy. Orphanet J Rare Dis 2020;15(1):160. (SCI).
4. Wen-Chen Liang, Chen-Hua Wang, Wan-Zi Chen, Yun-Ting Kuo, Hsiu-Fen Lin, Shigeaki Suzuki, Ichizo Nishino, Yuh-Jyh Jong*. Treatment experience of Taiwanese patients with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase myopathy. Kaohsiung J Med Sci 2020;36(8):649-55. (SCI).
5. Shen PC, Lu CC, Liang WC, Tien YC, Jong YJ, Lu YM, Liu ZM, Shih CL, Chou SH. Predictors

for Deformity Progression in a Spinal Muscular Atrophy Cohort After Scoliosis Correction Surgery. Clin Spine Surg 2020;33(8):E407-14. (SCI).

6. De Vivo DC1, Bertini E, Swoboda KJ, Hwu WL, Crawford TO, Finkel RS, Kirschner J, Kuntz NL, Parsons JA, Ryan MM, Butterfield RJ, Topaloglu H, Ben-Omran T, Sansone VA, Jong YJ, Shu F, Staropoli JF, Kerr D, Sandrock AW, Stebbins C, Petrillo M, Braley G, Johnson K, Foster R, Gheuens S, Bhan I, Reyna SP, Fradette S, Farwell W; NURTURE Study Group. Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscul Disord 2019;29(11):842-56. (SCI).
7. Yang TW, Lee WH, Tu SJ, Huang WC, Chen HM, Sun TH, Tsai MC, Wang CC, Chen HY, Huang CC, Shiu BH, Yang TL, Huang HT, Chou YP, Chou CH, Huang YR, Sun YR, Liang C, Lin FM, Ho SY, Chen WL, Yang SF, Ueng KC, Huang HD, Huang CN, Jong YJ*, Lin CC. Enterotype-based Analysis of Gut Microbiota along the Conventional Adenoma-Carcinoma Colorectal Cancer Pathway, Sci Rep. 2019;9(1):10923. (SCI).
8. Lin CY, Lee CH, Chuang YH, Lee JY, Chiu YY, Wu Lee YH, Jong YJ, Hwang JK, Huang SH, Chen LC, Wu CH, Tu SH, Ho YS, Yang JM. Membrane protein-regulated networks across human cancers, Nat Commun 2019;10(1):3131. (SCI).
9. Chen TH , Liang WC, Chen IC, Liu YC, Hsu JH, Jong YJ. Combined noninvasive ventilation and mechanical insufflator–exsufflator for acute respiratory failure in patients with neuromuscular disease: effectiveness and outcome predictors. Ther Adv Respir Dis 2019;13:1753466619875928 (SCI).
10. Wang CC, Chen CA, Jong YJ, Kou HS. Specific Gene Capture Combined with Restriction-Fragment Release for Directly Fluorescent Genotyping of Single-Nucleotide Polymorphisms in Diagnosing Spinal Muscular Atrophy. Anal Chem 2018;90(19):11599-606. (SCI).
11. Chang YA, Weng SL, Yang SF, Chou CH, Huang WC, Tu SJ, Chang TH, Huang CN, Jong YJ, Huang HD. A three-microRNA signature as a potential biomarker for the early detection of oral cancer. Int J Mol Sci 2018;19(3):758. (SCI).
12. Yang SF, Huang HD, Fan WL, Jong YJ, Chen MK, Huang CN, Chuang CY, Kuo YL, Chung WH, Su SC. Compositional and functional variations of oral microbiota associated with the mutational changes in oral cancer. Oral Oncol 2018;77:1-8. (SCI).
13. Liang WC, Uruha A, Suzuki S, Murakami N, Takeshita E, Chen WZ, Jong YJ, Endo Y, Komaki H, Fujii T, Kawano Y, Mori-Yoshimura M, Oya Y, Xi J, Zhu W, Zhao C, Watanabe Y, Ikemoto K, Nishikawa A, Hamanaka K, Mitsuhashi S, Suzuki N, Nishino I*. Pediatric necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl -coenzyme A reductase antibodies. Rheumatology (Oxford) 2017;56(2):287-93. (SCI).
14. Liang WC, Lin YF, Liu TY, Chang SC, Chen BH, Nishino I, Jong YJ*. Neurite growth could be impaired by ETFDH mutation but restored by mitochondrial cofactors. Muscle Nerve 2017;56:479-85. (SCI).
15. Tseng WL, Chou ST, Chiang HC, Wang ML, Chien CS, Chen KH, Leu HB, Wang CY, Chang YL, Liu YY, Jong YJ, Lin SZ, Chiou SH, Lin SJ, Yu WC*, Lin S. Imbalanced production of reactive oxygen species and mitochondrial antioxidant-SOD2 in Fabry disease-specific human induced pluripotent stem cell-differentiated vascular endothelial cells. Cell Transplant 2017;26(3):513-27. (SCI).
16. Liu TY, Chen YC, Jong YJ, Tsai HJ, Lee CC, Chang YS, Chang JG*, Chang YF. Muscle developmental defects in heterogeneous nuclear Ribonucleoprotein A1 knockout mice. Open Biol 2017;7(1):160303. (SCI).
17. Chen YC, Chang JG, Liu TY, Jong YJ, Cheng WL, Yuo CY*. Securinine enhances SMN2 exon 7 inclusion in spinal muscular atrophy cells. Biomed Pharmacother 2017;88:708-14. (SCI).
18. Liang WC, Tian X, You CY, Chen WZ, Kan TM, Su YN, Nishino I, Wong LC, Jong YJ*. Comprehensive target capture/next-generation sequencing as a second-tier diagnostic approach for congenital muscular dystrophy in Taiwan. PLoS One 2017;12(2):e0170517. (SCI).
19. Chiou GY, Yang TW, Huang CC, Tang CY, Yen JY, Tsai MC, Chen HY, Fadhilah N, Lin CC*, Jong YJ*. Musashi-1 promotes a cancer stem cell lineage and chemoresistance in colorectal cancer cells. Sci Rep 2017;7(1):2172. (SCI).
20. Chien YH, Chiang SC, Weng WC, Lee NC, Lin CJ, Hsieh WS, Lee WT, Jong YJ, Ko TM, Hwu WL*. Presymptomatic diagnosis of spinal muscular atrophy through newborn screening.

J Pediatr 2017;190:124-9. e1. (SCI).

21. Tseng YT, Jong YJ, Liang WF, Chang FR, Lo YC*. The water extract of Liuwei dihuang possesses multi-protective properties on neurons and muscle tissue against deficiency of survival motor neuron protein. Phytomedicine 2017;34:97-105. (SCI).
22. Chen TH, Hsu JH, Jong YJ*. Noninvasive airway approaches for acute neuromuscular respiratory failure in emergency departments. Pediatr Pulmonol 2017;52(10):E55-7. (SCI).
23. Victor RG, Sweeney HL, Finkel R, McDonald CM, Byrne B, Eagle M, Goemans N, Vandenborne K, Dubrovsky AL, Topaloglu H, Miceli MC, Furlong P, Landry J, Elashoff R, Cox D, Tadalafil DMD Study Group. A phase 3 randomized placebo-controlled trial of tadalafil for Duchenne muscular dystrophy. Neurology 2017;89(17):1811-20. (SCI).
24. Finkel RS, Mercuri E, Darras BT, Connolly AM, Kuntz NL, Kirschner J, Chiriboga CA, Saito K, Servais L, Tizzano E, Topaloglu H, Tulinius M, Montes J, Glanzman AM, Bishop K, Zhong ZJ, Gheuens S, Bennett CF, Schneider E, Farwell W, De Vivo DC, ENDEAR Study Group. Nusinersen versus sham control in infantile-onset spinal muscular atrophy. N Engl J Med. 2017;377(18):1723-32. (SCI).
25. Lee WH, Chen HM, Yang SF, Liang C, Peng CY, Lin FM, Tsai LL, Wu BC, Hsin CH, Chuang CY, Yang T, Yang TL, Ho SY, Chen WL, Ueng KC, Huang HD, Huang CN, Jong YJ*. Bacterial alterations in salivary microbiota and their association in oral cancer. Sci Rep 2017;7(1):16540. (SCI)

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神經肌肉罕病學思歷程

My Research Journey of Neuromuscular Rare Diseases

Yuh-Jyh Jong

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Graduate Institute of Clinical Medicine, College of Medicine, Kaohsiung Medical University, Kaohsiung, Taiwan

Abstract

Neuromuscular diseases (NMDs) can be caused by autoimmune disorders, genetic/hereditary disorders, some forms of the collagen disorder, exposure to environmental chemicals and poisoning which includes heavy metal poisoning. Inherited neuromuscular diseases (INMDs) are genetically and clinically heterogeneous diseases, mainly involving spinal motor neurons, neuromuscular junctions, nerves, and muscles. The majority of INMDs are hereditary, degenerative, rare, delayed diagnosis and under-care.

Deep phenotyping, including clinical examinations, biochemistry, neurophysiology, muscle imaging, muscle histopathology and immunochemistry; deep genotyping including Sanger confirmation that variant is analytically valid and segregates with disease from candidate genes by comprehensive target capture/next-generation sequencing (NGS), computational validation with candidate protein modeling, and experimental validation with cell model system for INMD mutated gene expression; and multidisciplinary care, including genetic counseling, standard of care, available clinical trials, and expanded access program, are the cornerstones of precision medicine of NMDs.

In this presentation, we will share our reflection of caring patients with NMDs.

《回議程》

林秀娟, MD

EDUCATION

1980	國立台灣大學醫學院醫學系

CURRENT POSITION & PROFESSIONAL BACKGROUND

2012-present	奇美醫療財團法人奇美醫院講座教授兼遺傳諮詢中心主任 成功大學醫學院醫學系小兒科兼任教授
2007-2012	成功大學醫學院附設醫院副院長
2003-2005	行政院衛生署國民健康局局長
2002-2003	行政院衛生署國民健康局副局長
1996-1999	成功大學醫學院醫學系小兒科主任　附設醫院小兒部主任
1995-present	成功大學醫學院醫學系教授
1985-1986	Fellow, Department of Pediatrics, Division of Medical Genetics, Rutgers Medical School, New Brunswick, New Jersey
1984-1985	Fellow, Department of Medical Genetics, Johns Hopkins Hospital, Baltimore, Maryland
1980-1984	台灣大學醫學院附設醫院住院醫師

RESEARCH INTEREST(S)

- 小兒科(遺傳新陳代謝&內分泌)
- 醫學倫理學
- 醫學教育

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罕見疾病照護在奇美

林秀娟
奇美醫療財團法人奇美醫院

Abstract
本院遺傳諮詢中心自 106 年 12 月起承接「106 年-108 年罕見疾病照護服務計畫」，於 109 年繼續執行罕見疾病照護服務計畫。跨科部組成「罕見疾病照護服務」執行團隊；成員包括醫師、護理師、藥劑師、社工師、營養師、復健師、心理師等，團隊成員依「罕見疾病照護工作手冊」組成核心工作小組及各服務項目之聯絡窗口。此外奇美醫療體系亦同時具有醫學中心、區域醫院(柳營奇美醫院)及地區醫院(佳里奇美醫院)，體系內 3 家醫院有良好的垂直整合並有居家醫療及長照系統之網絡；且與雲嘉南地區之各層級醫療院所有良好合作關係，根據個案的需求進行照護服務。因此雖然本計畫服務對象多為其他醫院通報的個案，而且個案居住地不只是台南還包含雲林、嘉義地區，執行上相對困難，藉由整合醫療、長照、社區等各方面資源，讓罕見疾病病人及家庭能得到良好的照護服務。藉此研討會分享執行心得並向各院請益。

《回議程》

趙美琴, MD

EDUCATION

1966-1973 Kaohsiung Medical College

CURRENT POSITION & PROFESSIONAL BACKGROUND

2015/8 -present Chief of Division of Genetics and Metabolism
Children Hospital of ChangHua Christian Hospital

RESEARCH INTEREST(S)

- Clinical cytogenetics
- Molecular genetics
- Genetic counseling

Honors and Awards(or Selected Publications)

1. Impaired glucose homeostasis and a novel HLCS pathogenic variant in holocarboxylase synthetase deficiency: a report of two cases and brief review. J Pediatr Endocrinol Metab . 2020 Nov 26;33(11):1481-1486.
2. Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985-2019) Orphanet J Rare Dis . 2020 Nov 7;15(1):314.
3. Congenital generalized lipodystrophy in Taiwan. J Formos Med Assoc . 2019 Jan;118(1 Pt 1):142-147.
4. Functional independence of Taiwanese children with Prader-Willi syndrome. Am J Med Genet A. 2018 Jun;176(6):1309-1314.
5. Association study of LIN28B in girls with precocious puberty. J Pediatr Endocrinol Metab . 2017 May 24;30(6):663-667.

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彰基/高醫罕見疾病之分享

趙美琴[1]，陳冠容[1]，吳信儒[1]，陳明[2]，李美慧[2]，李涵薇[2]，蕭惠彬[3]，王禎鞠[3]，郭佩雯[3]
[1]彰化基督教兒童醫院　兒童遺傳及新陳代謝科
[2]彰化基督教醫院　遺傳諮詢中心
[3]高雄醫學大學附設醫院　小兒科部，遺傳新陳代謝及內分泌科，遺傳諮詢中心

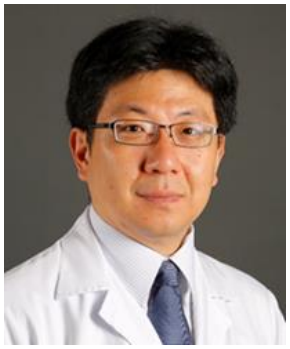
Abstract

罕見疾病在國內的定義為盛行率小於萬分之一，且具有診療困難性的疾病。我國已於 2000 年通過「罕見疾病防治及藥物法」，以維護這群少數弱勢病人的權益。高雄醫學大學附設醫院於 1990 年成立「優生保健諮詢中心」，彰化基督教醫院亦於 1996 年成立「醫學遺傳中心」，並分別於 2000 年與 2004 年更名為「遺傳諮詢中心」，以提供產前與產後各類遺傳疾病之諮詢與追蹤治療。

罕見疾病大致可分成先天畸形、先天性代謝異常、各類器官異常（如神經、呼吸、消化、泌尿、皮膚、肌肉骨骼、內分泌、血液及免疫疾病等）。這些病患往往需要終身照顧，而各類疾病在不同時期的表現也有所差別，因此特別需要遺傳專科醫師的全方位照顧。舉例而言，小胖威利症候群在幼兒時期會因為低張力而生長與發展遲緩，而到了成人時期卻往往苦於過度肥胖與代謝症候群。

由於多數的罕見疾病具有遺傳性，因此疾病的基因確診極為重要。從產前羊水、絨毛膜取樣，到產後血液染色體或致病基因的檢查、新陳代謝檢驗等，而隨著全基因定序的普及化，許多過去僅有臨床診斷的案例，現在也都能順利找到致病基因，讓這些罕見疾病患者與家庭能得到更詳盡的遺傳諮詢與治療。

《回議程》



李宜中, MD, PhD

EDUCATION

2004/9-2008/5	Ph.D.	Institute of Clinical Medicine, National Yang-Ming University
1989/10-1996/7	M.D.	Department of Medicine, National Yang-Ming University

CURRENT POSITION & PROFESSIONAL BACKGROUND

2018/1-present	Fellow, American Academy of Neurology
2018/1-present	Chairperson, Division of Clinical Neurosciences, Department of Life Science, Ministry of Science and Technology, Taiwan
2017/8-present	Chief, Division of Peripheral Nervous System Disorders, Neurological Institute, Taipei Veterans General Hospital
2017/5-present	Chairperson, Neurogenetic Sections, Taiwan Neurological Society
2016/8-present	Adjunct Professor, Department of Neurology, National Yang-Ming University School of Medicine

RESEARCH INTEREST(S)

Dr. Lee's research and clinical interests are to find out why and how the patients get inherited neurological diseases, with an emphasis on inherited peripheral neuropathy, amyotrophic lateral sclerosis (ALS) and inherited cerebral small vessel diseases. His lab has identified several novel disease genes for inherited neuropathy and elucidated the genetic and phenotypic features of hereditary diseases in Taiwan, including familial ALS, Charcot-Marie-Tooth disease, Hereditary Spastic Paraplegia, and CADASIL.

Honors and Awards(or Selected Publications)

1. YC Liao, YC Hu, Chung CP, Wang YF, Guo YC, Tsai YS, **Lee YC.*** Intracerebral hemorrhage in CADASIL: prevalence, clinical and neuroimaging features and risk factors. Stroke 2020 (accepted)
2. **Lee YC**, Chung CP, Chang MH, Wang SJ, Liao YC*. NOTCH3 cysteine-altering variant is an important risk factor for stroke in Taiwanese population. Neurology 2020;94(1):e87-e96.
3. **Lee YC**, Chung CP, Chao NC, Fuh JL, Chang FC, Soong BW, Liao YC*. Characterization of heterozygous HTRA1 mutations in Taiwanese patients with cerebral small vessel disease. Stroke 2018;49:1593-1601.
4. Tsai PC, Soong BW, Mademan I, Huang YH, Liu CR, Hsiao CT, Wu HT, Liu TT, Liu YT, Tseng YT, Lin KP, Yang UC, Chung KW, Choi BO, Nicholson GA, Kennerson ML, Chan CC, De Jonghe P, Cheng TH, Liao YC*, Züchner S, Baets J, **Lee YC***. A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain 2017;140:1252-66.

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台北榮總神經罕病之照護及研究

李宜中 [1,2]

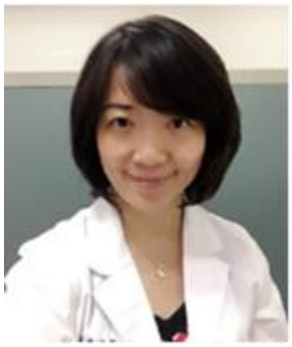
[1] 臺北榮民總醫院周邊神經科主任

[2] 國立陽明大學醫學院神經學科教授

Abstract

神經罕見疾病佔我們國家所公告的罕見疾病項目內的大宗，而神經罕見疾病的臨床照護中最基本的一件事情就是正確的診斷，但大部分的神經罕見疾病都與基因突變相關，需要靠基因檢測來正確地診斷或分型。目前神經罕病的病友，要申請健保的重大傷病身份都需要罕見疾病的通報先通過，而重大傷病的身份對許多罕病病友的照護與經濟負擔都相當重要。所以許多與基因突變相關的神經罕病雖然能以臨床徵兆或生化檢測來確定診斷，但基因檢測對他們仍然是非常重要的。台北榮總周邊神經科神經基因實驗室從宋秉文教授創立以來，秉持著為神經罕病病人服務的使命感，長年來為數千位各類神經罕病病友進行基因檢測，而未讓病友自行負擔費用，同時也匯集許多神經罕見疾病的基因診斷經驗與研究機會。在本次報告中，我將與各位先進介紹我們對於小腦脊髓運動失調症 (spinocerebellar ataxia)、肌萎縮性脊髓側索硬化症 (Amyotrophic lateral sclerosis)、遺傳性痙攣性截癱 (Hereditary Spastic Paraplegia)、家族性類澱粉神經病變 (Familial amyloidotic polyneuropathy)、與 Charcot-Marie-Tooth disease 的罕病診斷照護經驗與相關本土研究結果。

《回議程》



梁文貞, MD

EDUCATION

2018-present	Ph.D.	Graduate Institute of Medicine, Kaohsiung Medical University
2004-2007	B.S.	Graduate Institute of Medicine, Kaohsiung Medical University
1992-1999	M.D.	Faculty of Medicine, Kaohsiung Medical University

CURRENT POSITION & PROFESSIONAL BACKGROUND

2012-present	Assistant Professor, Department of Pediatrics, School of Medicine, College of Medicine, Kaohsiung Medical University
2007-2010	Research Fellow, Department of Neuromuscular Research, National Institute of Neurology, National Center of Neurology and Psychiatry
2004-present	Attending physician, Department of Pediatrics, Kaohsiung Medical University Hospital

RESEARCH INTEREST(S)

● Neuromuscular diseases; Neurogenetics; Animal models

Honors and Awards(or Selected Publications)

1. **Liang WC**, Jong YJ*, Wang CH, Wang CH, Tian X, Chen WZ, Kan TM, Minami N, Nishino I, Wong LC. Clinical, pathological, imaging, and genetic characterization in a Taiwanese cohort with limb-girdle muscular dystrophy. Orphanet J Rare Dis 2020 Jun 23;15(1):160.
2. **Liang WC**, Tian X, You CY, Chen WZ, Kan TM, Su YN, Nishino I, Wong LJC, Jong YJ*. Comprehensive target capture/next generation sequencing as a second-tier diagnostic approach for congenital muscular dystrophy in Taiwan. PLoS One 2017;12(2):e0170517.
3. **Liang WC**, Lin YF, Liu TY, Chang SC, Chen BH, Nishino I, Jong YJ*. Neurite growth could be impaired by ETFDH mutation but restored by mitochondrial cofactors. Muscle Nerve 2017;56:479-485.
4. **Liang WC#**, Uruha A#, Suzuki S, Murakami N, Takeshita E, Chen WZ, Jong YJ, Endo Y, Komaki H, Fujii T, Kawano Y, Mori-Yoshimura M, Oya Y, Xi J, Zhu W, Zhao C, Watanabe Y, Ikemoto K, Nishikawa A, Hamanaka K, Mitsuhashi S, Suzuki N, Nishino I*. Pediatric necrotizing myopathy associated with anti-3-hydroxy-3-methylglutaryl -coenzyme A reductase antibodies. Rheumatology (Oxford) 2017;56:287-293.
5. Elsevier membership prize of the 21st annual meeting of World Muscle Society in Granada, Spain (Poster presentation) 2016
6. Best Poster Award of the 15th scientific meeting of Asian and Oceanian Myology Center in Hsinchu, Taiwan (Poster presentation) 2016
7. The Young Investigator Award of the 218th scientific meeting of Taiwan Pediatric Association in Taipei, Taiwan (Platform presentation)
8. **Liang WC**, Mitsuhashi H, Keduka E, Nonaka I, Noguchi S, Nishino I, Hayashi YK*. TMEM43 Mutations in Emery-Dreifuss Muscular Dystrophy-related Myopathy. Ann Neurol 2011;69:1005-1013.
9. The Young Investigator Award of the 50th Japanese Society of Child Neurology annual meeting in Tokyo, Japan (Poster presentation) 2008

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高醫治療神經肌肉疾病之分享

Multidisciplinary care for patients with neuromuscular diseases in KMUH

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[1]Department of Pediatrics Kaohsiung Medical University Hospital, Kaohsiung Medical University, Kaohsiung, Taiwan

[2]Translational Research Center of Neuromuscular Diseases, Kaohsiung Medical University Hospital, Kaohsiung Medical University, Kaohsiung, Taiwan

[3]Department of Laboratory Medicine, Kaohsiung Medical University Hospital, Kaohsiung Medical University, Kaohsiung, Taiwan

[4]Department of Pediatrics, School of Medicine, College of Medicine, Kaohsiung Medical University, Kaohsiung, Taiwan

[5]Graduate Institute of Clinical Medicine, College of Medicine, Kaohsiung Medical University, Kaohsiung, Taiwan

Abstract

Neuromuscular disease (NMD) is a disease entity consists of the disorders involved in lower motor neuron, peripheral nerve, neuromuscular junction and muscle. Most of them are hereditary diseases but some are required such as autoimmune-related myasthenia gravis and myositis. The clinical course of NMD is usually progressive which culminates in marked motor function impairment and complications involved in multiple systems. The NMDs thus result in the physical, psychogenic and economic burden of not only patients themselves, but their families and whole society. To date, effective therapy is available for only a few NMDs; patients and families therefore often drop regular medical visit and neglect the importance of standard care. In fact, even the most effective drugs could not reverse the majority of complications, such as scoliosis, joint contracture, respiratory failure etc. For that reason, multidisciplinary standard care provided by medical team together with patients' associations, non-profit organizations and Governmental bureaus could play an important and practical role to prevent, identify or ameliorate these complications. The final goal to be achieved is therefore to improve the life quality of patients and families, and to further reduce the waste of medical and social resource for taking care of severe complications.

《回議程》

李妮鍾, MD, PhD

EDUCATION

2014	Ph.D.	Graduate Institute of Clinical Medicine, College of Medicine, National Taiwan University
1999	M.D.	Medical College, National Yang-Ming University

CURRENT POSITION & PROFESSIONAL BACKGROUND

2016/8-present	Clinical associate professor, Department of Pediatrics, National Taiwan University Hospital, Taipei, Taiwan
2006-present	Attending Physician, Department of Medical Genetics and Pediatrics, NTUH

RESEARCH INTEREST(S)

- Molecular Genetics
- Gene therapy

Honors and Awards(or Selected Publications)

1. Wu ET, Hwu WL, Chien YH, Hsu C, Chen TF, Chen NQ, Chou HC, Tsao PN, Fan PC, Tsai IJ, Lin SP, Hsieh WS, Chang TM, Chen CN, Lee CH, Chou YY, Chiu PC, Tsai WH, Hsiung HC, Lai F, **Lee NC**. Critical Trio Exome Benefits In-Time Decision-Making for Pediatric Patients With Severe Illnesses. Pediatr Crit Care Med. 2019 Jul 1. (Corresponding author)
2. Yu MH, Tsang MH, Lai S, Ho MS, Tse DML, Willis B, Kwong AK, Chou YY, Lin SP, Quinzii CM, Hwu WL, Chien YH, Kuo PL, Chan VC, Tsoi C, Chong SC, Rodenburg RJT, Smeitink J, Mak CC, Yeung KS, Fung JL, Lam W, Hui J, **Lee NC**, Fung CW, Chung BH. Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. NPJ Genom Med. 2019 Aug 5;4:18. (Corresponding author)
3. **Lee NC**, Hwu WL, Muramatsu SI, Falk DJ, Byrne BJ, Cheng CH, Shih NC, Chang KL, Tsai LK, Chien YH. A Neuron-Specific Gene Therapy Relieves Motor Deficits in Pompe Disease Mice. Mol Neurobiol. 2018 Jun;55(6):5299-5309.

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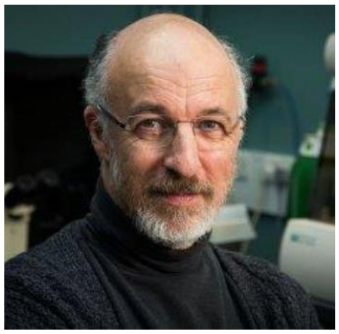
台大罕見疾病之分享

李妮鍾，簡穎秀，胡務亮
臺大醫院基因醫學部/小兒部

Abstract
臺大醫院之罕見病照護主要立基於基因醫學部。基因醫學部的前身優生保健諮詢中心於 1983 年 12 月 13 日由衛生署委託成立，迄今已逾 35 年。最初乃是依據行政院衛生署研擬完成之優生保健法施行細則草案而設，進行優生健康檢查、產前遺傳診斷、新生兒先天缺陷疾病篩檢及優生諮詢等服務。自 1987 年納入編制為常設醫療部門後更於 2001 年改組為目前之基因醫學部。2001 年引進先進的串聯質譜儀(Tandem mass)，只需採取小寶寶的一滴血便可同時檢測二十種以上的氨基酸代謝異常、脂肪酸代謝異常及有機酸血症，目前已成為先天代謝異常檢測之常規，並接軌新生兒代謝篩檢。此外，因此我們自 2005 年起推動了全世界第一個龐貝氏症的新生兒篩檢計畫，成功的證明經由新生兒篩檢確認的嬰兒型患者，不但心臟肥大的現象全部皆得以改善，連運動功能的改善，也全部達成，相較於因為臨床症狀表現後再開始治療的個案，的確可以大幅改善治療效果。這樣的成績可以維持，到 10 年的長期非依賴呼吸器的存活率仍為 100%，表示有極佳的生活品質，領先其他國家的治療成果。我們的研究結果無缺失的通過美國 FDA 實地查核，因此可以當作藥物上市新增適應症的結果。2010 年胡務亮教授開始全球首例的 AADC 基因治療之人道救援治療，利用 AAV 病毒載體直接注入病人的腦部，大幅改善患者運動功能與神經症狀。美國的 Agilis Biotherapeutics, LLC 於 2015 年年底和台灣大學簽署產學合作技術轉此治療，並於 2016 年 6 月得到美國 FDA 孤兒藥之認定。2017 年在科技部「建立以婦幼醫學為主軸的精準醫療專案計畫」支持下，成立了「高速次世代基因診斷團隊」，建立快速的次世代基因診斷平台，以診斷急重症兒童的遺傳性疾病，為兒童罕病的照護更向前邁進一步。

《回議程》



Michael R Duchen, PhD

EDUCATION

BA Physiology Oxford
MBBS, Oxford and London
PhD London
MRCP UK

CURRENT POSITION & PROFESSIONAL BACKGROUND

1999-present	Professor of Physiology, UCL
1992-1999	Reader in Physiology, UCL
1984-1992	Royal Society University research fellow UCL
1978-1981	Posts in clinical medicine St George's Hospital group

RESEARCH INTEREST(S)

Roles of mitochondrial dysfunction in human disease and pathways to mitochondrial dysfunction as potential therapeutic targets.

Honors and Awards(or Selected Publications)

1. Plotegher N, et al. (2020) GBA1 depletion compromises mitochondrial metabolism and sensitises neurons to calcium overload. Cell Death and Differentiation. 27 (5), 1588-1603.
2. Haythorne E, et al. (2019) Diabetes causes marked inhibition of mitochondrial metabolism in pancreatic β-cells. Nature Communications. 10(1), 1-17.
3. Pollard AE, et al. (2019) AMPK activation protects against diet induced obesity through Ucp1-independent thermogenesis in subcutaneous white adipose tissue. Nature Metabolism. 2019 Mar;1(3):340-349.
4. Maffioletti SM, et al. Three-Dimensional Human iPSC-Derived Artificial Skeletal Muscles Model Muscular Dystrophies and Enable Multilineage Tissue Engineering. Cell Reports, 2018 Apr 17;23(3):899-908.
5. Kon,S et al. (2017) "Cell competition with normal epithelial cells promotes apical extrusion of transformed cells through metabolic changes", Nature Cell Biology, 19(5):530-541.
6. Hawkins KE, et al. (2016) NRF2 orchestrates the metabolic shift during induced pluripotent stem cell reprogramming. Cell Reports, 14(8):1883-91.

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Suppression of Mitophagy by Maladaptive Cell Signaling Pathways in the Progression of Mitochondrial Disease

Michael R Duchen

Department of Cell and Developmental Biology, University College London, UK.

Abstract

Mutations of the mitochondrial genome (mtDNA) cause a range of profoundly debilitating clinical conditions for which treatment options are very limited. Most mtDNA diseases show heteroplasmy – tissues express both wild-type and mutant mtDNA. While the level of heteroplasmy broadly correlates with disease severity, the relationships between specific mtDNA mutations, heteroplasmy, disease phenotype and severity are not wellunderstood. We have found that the m.3243A>G mutation, the most prevalent mutation of mtDNA diseases and the major cause of mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes (MELAS), leads to metabolic changes which promote the upregulation of the PI3K-Akt-mTORC1 axis in patient-derived cells and tissues. Remarkably, pharmacological inhibition of PI3K, Akt, or mTORC1 activated mitophagy, reduced mtDNA mutant load and rescued cellular bioenergetics. The rescue was prevented by inhibition of mitophagy. These data suggest that the maladaptive activation of the PI3K-Akt-mTOR axis suppresses mitophagy and sustains the mutant mtDNA. The PI3K-Akt-mTORC1 axis thus represents a therapeutic target with translational potential that may benefit people suffering from the consequences of the m.3243A>G mutation.

《回議程》

陳冠如 執行長

EDUCATION

1997/9-1999/7 M.S. Institute of Genetics, National Yang-Ming University

CURRENT POSITION & PROFESSIONAL BACKGROUND

2017/4-present Executive Director, Taiwan Foundation for Rare Disorders

2012-presetn EGC member, MJ Biobank, MJ Health Research Foundation

2006-present Certified Genetic Counselor by Human Genetics Society

2000/11-present Genetic Counselor, Taiwan Foundation for Rare Disorders

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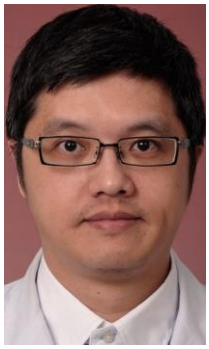
罕見疾病基金會的使命與任務

陳冠如
財團法人罕見疾病基金會

Abstract
「罕見疾病」這個名詞在 20 年年前並不存在，患者常為醫療體制及社福系統的孤兒。在病患家長創辦人陳莉茵女士及共同創辦人曾敏傑教授的發起之下，一連串社會運動，喚起台灣社會重視醫療弱勢族群的聲音，催生罕見疾病防治及藥物法，成為全世界第五個立法保障罕病醫療權益之國家。適逢罕病法執行 20 周年之際，本次專題將藉由回顧罕病法的發展，了解罕病基金會的使命與任務，並且從罕病家庭的改變，看見各界在罕病領域耕耘的成果。期望未來能依著病友家庭期待提供永續服務。

《回議程》



王士忠, MD

EDUCATION

2005/9-2009/7　私立長榮大學醫學研究所在職碩士班碩士
1988/-1995/6　私立中國醫藥大學中醫學系學士

CURRENT POSITION & PROFESSIONAL BACKGROUND

2018/7-present　彰化基督教醫院 教學部 主任
2018/1-present　彰化基督教兒童醫院 兒科部 部主任
2015/3-present　彰化基督教兒童醫院 兒童血液腫瘤科 主任
2015/2-present　彰化基督教兒童醫院 教學部 主任
2011/8-present　彰化基督教醫院 教學部 實習中心 主任
2010/7-2011/7　彰化基督教醫院 教學部 醫師教育中心 協同主任
2007/7-2015/2　彰化基督教醫院 兒童血液腫瘤科 主任
2006/6-2010/6　彰化基督教醫院 教學部 醫學教育中心 主任
2004/1-2007/6　彰化基督教醫院 兒童血液腫瘤科 主治醫師
2001/8-2003/7　國立台大醫院 兒童血液腫瘤科 研究醫師
2000/7-2001/7　彰化基督教醫院 兒科部 總醫師
1997/7-2000/6　彰化基督教醫院 兒科部 住院醫師

Honors and Awards(or Selected Publications)

1. M2-like polarization of THP-1 monocyte-derived macrophages under chronic iron overload. Kao JK, **Wang SC**, Ho LW, Huang SW, Lee CH, Lee MS, Yang RC, Shieh JJ.Ann Hematol. 2020 Mar;99(3):431-441.
2. Intravenous immunoglobulin therapy enhances suppressive regulatory T cells and decreases innate lymphoid cells in children with immune thrombocytopenia. **Wang SC**, Yang KD, Lin CY, Huang AY, Hsiao CC, Lin MT, Tsai YG. Pediatr Blood Cancer. 2020 Feb;67(2):e28075.
3. TP53 alterations in relapsed childhood acute lymphoblastic leukemia. Yu CH, Chang WT, Jou ST, Lin TK, Chang YH, Lin CY, Lin KH, Lu MY, Chen SH, Wu KH, **Wang SC**, Chang HH, Su YN, Hung CC, Lin DT, Chen HY, Yang YL. Cancer Sci. 2020 Jan;111(1):229-238.
4. Celastrol induces vincristine multidrug resistance oral cancer cell apoptosis by targeting JNK1/2 signaling pathway. Lin FZ, **Wang SC**, Hsi YT, Lo YS, Lin CC, Chuang YC, Lin SH, Hsieh MJ, Chen MK. Phytomedicine. 2019 Feb 15;54:1-8.
5. Outcome and prognosis of anaplastic large cell lymphoma in children: a report from the Taiwan Pediatric Oncology Group. Chen SH, Chen JS, Jou ST, Wu KH, Hung IJ, Sheen JM, Lu MY, Chen BW, Jaing TH, **Wang SC**, Lin MT, Chang TK, Liu HC, Yang CP. Leuk Lymphoma. 2019 Jan 15:1-8.
6. Treatment of childhood acute lymphoblastic leukemia with delayed first intrathecal therapy and omission of prophylactic cranial irradiation: Results of the TPOG-ALL-2002 study. Yeh TC, Liang DC, Hou JY, Jaing TH, Lin DT, Yang CP, Peng CT, Hung IJ, Lin KH, Hsiao CC, Jou ST, Chiou SS, Chen JS, **Wang SC**, Chang TK, Wu KH, Sheen JM, Yen HJ, Chen SH, Lu

MY, Li MJ, Chang TT, Huang TH, Chang YH, Chen SH, Yang YL, Chang HH, Chen BW, Lin PC, Cheng CN, Chao YH, Yang SH, Chao YY, Liu HC. Cancer. 2018 Dec 1;124(23):4538-4547.

7. Cantharidic acid induces apoptosis of human leukemic HL-60 cells via c-Jun N-terminal kinase-regulated caspase-8/-9/-3 activation pathway. **Wang SC**, Chow JM, Chien MH, Lin CW, Chen HY, Hsiao PC, Yang SF. Environ Toxicol. 2018 Apr;33(4):514-522. doi: 10.1002/tox.22537. Epub 2018 Jan 18.
8. Port catheter-associated Aureobasidium melanigenum fungemia. **Wang SC**, Lo HJ, Lin LJ, Chen CH. J Formos Med Assoc. 2018 Apr;117(4):346-347. doi: 10.1016/j.jfma.2017.06.009. Epub 2017 Jul 1.
9. Pediatric acute lymphoblastic leukemia with t(1;19)/TCF3-PBX1 in Taiwan. Yen HJ, Chen SH, Chang TY, Yang CP, Lin DT, Hung IJ, Lin KH, Chen JS, Hsiao CC, Chang TT, Chang TK, Peng CT, Lin MT, Jaing TH, Liu HC, Jou ST, Lu MY, Cheng CN, Sheen JM, Chiou SS, Hung GY, Wu KH, Yeh TC, **Wang SC**, Chen RL, Chang HH, Yang YL, Chen SH, Cheng SN, Chang YH, Chen BW, Hsieh YL, Huang FL, Ho WL, Wang JL, Chang CY, Chao YH, Lin PC, Chen YC, Liao YM, Lin TH, Shih LY, Liang DC. Pediatr Blood Cancer. 2017 Oct;64(10).
10. Treatment for childhood acute lymphoblastic leukemia in Taiwan: Taiwan Pediatric Oncology Group ALL-2002 study emphasizing optimal reinduction therapy and central nervous system preventive therapy without cranial radiation. Li MJ, Liu HC, Yen HJ, Jaing TH, Lin DT, Yang CP, Lin KH, Hung IJ, Jou ST, Lu MY, Hsiao CC, Peng CT, Chang TT, **Wang SC**, Lin MT, Chen JS, Chang TK, Hung GY, Wu KH, Yang YL, Chang HH, Chen SH, Yeh TC, Cheng CN, Lin PC, Chiou SS, Sheen JM, Cheng SN, Chen SH, Chang YH, Ho WL, Chao YH, Chen RL, Chen BW, Wang JL, Hsieh YL, Liao YM, Yang SH, Chang WH, Chao YY, Liang DC. Pediatr Blood Cancer. 2017 Feb;64(2):234-241.
11. Efficacy of rabbit antithymocyte globulin as first-line treatment of severe aplastic anemia: an Asian multicenter retrospective study. Chuncharunee S, Wong R, Rojnuckarin P, Chang CS, Chang KM, Lu MY, Hwang WL, Koh LP, Chen TY, Leung AY, Norasetthada L, **Wang SC**, Chang MC, Wu KH, Issaragrisil S. Int J Hematol. 2016 Oct;104(4):454-61.
12. Chronic Iron Overload Results in Impaired Bacterial Killing of THP-1 Derived Macrophage through the Inhibition of Lysosomal Acidification. Kao JK, **Wang SC**, Ho LW, Huang SW, Chang SH, Yang RC, Ke YY, Wu CY, Wang JY, Shieh JJ. PLoS One. 2016 May 31;11(5):e0156713.
13. Outcomes Following Discontinuation of E. coli l-Asparaginase Upon Severe Allergic Reactions in Children With Acute Lymphoblastic Leukemia. Yen HJ, Chang WH, Liu HC, Yeh TC, Hung GY, Wu KH, Peng CT, Chang YH, Chang TK, Hsiao CC, Sheen JM, Chao YH, Chang TT, Chiou SS, Lin PC, **Wang SC**, Lin MT, Ho WL, Chen YC, Liang DC. Pediatr Blood Cancer. 2016 Apr;63(4):665-70.
14. Effect of Heat Shock Treatment on Endothelial Cell Integrity after Histamine Challenge 熱休克對內皮細胞經組織胺刺激後的細胞完整性的影響 **王士忠(Shih-Chung Wang)**；楊瑞成(Rei-Cheng Yang)；張明裕(Ming-Yuh Chang)；呂宗禧(Tsong-Shi Lu)；曹龍彥(Long-Yen Tsao) The Changhua Journal of Medicine；14卷1期 (2016 / 03 / 01)，P1 – 11.
15. Hypersensitivity to mosquito bites as the primary clinical manifestation of an Epstein-Barr virus infection. Chiu TM, Lin YM, **Wang SC**, Tsai YG. J Microbiol Immunol Infect. 2016 Aug;49(4):613-6.
16. Bilateral Acute Proptosis as Initial Manifestation of Acute Myeloid Leukemia. Huang YC, **Wang SC**, Chen SN, Jou JR. Orbit. 2015;34(5):248-52.

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海洋性貧血的過去、現在與未來

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Abstract
海洋性貧血以前稱為地中海型貧血，1997 年 4 月衛生署統一稱海洋性貧血，是一種自體隱性遺傳的血液疾病，它是因為紅血球中的血紅素蛋白的合成缺陷所引起的先天性病變，因為血紅素蛋白的量失衡，使紅血球容易破裂導致貧血，是台灣地區常見的遺傳疾病之一，約有 5%的人為帶因者。海洋性貧血主要分為甲型（α型）和乙型（β型）。台灣地區甲型比乙型多。台灣的甲型海洋性貧血絕大部分（95%）為東南亞型基因缺失，而乙型海洋性貧血主要由四種突變所造成。嚴重型海洋性貧血以乙型為主，主要的治療有常規輸血與排鐵劑的使用，過去病人常會接受脾臟切除，現在已少施行。血液幹細胞移植可以治癒嚴重型海洋性貧血，但是必須有合適的捐贈者，同時有化學治療的毒性與移植物抗宿主反應的風險。基因治療(Lentiglobin)與 CRISPR/Cas9 技術(CTX001)的發展，增加除了血液幹細胞移植以外的治療選項，值得期待。藥物治療方面，除了過去用來增加胎兒血色素 HbF 的 5-azacytidine，hydroxyurea 之外，與 TGF β ligand 結合的 Luspatercept, Sotatercept 在臨床試驗上也顯示可以減少病人輸血的需求。
台灣在 1993 年開始實施全國性的海洋性貧血篩檢，大幅減少嚴重型海洋性貧血患者的出生，但是每年仍會有零星的個案出現，醫師與民眾都須持續加強宣導。全民健保的實施，讓台灣的嚴重型海洋性貧血病人可以接受常規的輸血與昂貴的排鐵劑治療，這些病人的平均餘命已經可以接近一般健康人，目前照顧的重點，在於如何進一步提升病人的生活品質，除了醫師的照護之外，需要更多更全面的團隊人員一齊投入。

《回議程》

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EDUCATION

1996/12-1999/12	P.D.	Department of Foods and Nutrition, Purdue University, USA
1994/8-1996/12	M.S.	Department of Foods and Nutrition, Purdue University, USA

CURRENT POSITION & PROFESSIONAL BACKGROUND

2010/2-present Professor, Department of Nutrition, Chung Shan Medical University, Taiwan

RESEARCH INTEREST(S)

- Cancer cachexia
- Machado-Joseph disease

Honors and Awards(or Selected Publications)

1. Wang SC, Sun HL, Hsu YH, Liu SH, Lii CK, Tsai CH, **Liu KL**, Huang CS, Li CC. α-Linolenic acid inhibits the migration of human triple-negative breast cancer cells by attenuating Twist1 expression and suppressing Twist1-mediated epithelial-mesenchymal transition. Biochem Pharmacol. 2020 Oct; 180:114152.
2. Chang LC, Sun HL, Tsai CH, Kuo CW, **Liu KL**, Lii CK, Huang CS, Li CC. 1,25(OH)2 D3 attenuates indoxyl sulfate-induced epithelial-to-mesenchymal cell transition via inactivation of PI3K/Akt/β-catenin signaling in renal tubular epithelial cells. Nutrition. 2020 Jan;69:110554.
3. Wang CL, Lin KP, Hsu GW, **Liu KL**, Guo CH. Altered Mineral Metabolism and Disequilibrium Between Calcification Promoters and Inhibitors in Chronic Hemodialysis Patients. Biol Trace Elem Res. 2020 Jan;193(1):14-22 .
4. Lin HC, Li CC, Yang YC, Chiu TH, **Liu KL**, Lii CK, Chen HW. Andrographis paniculata diterpenoids and ethanolic extract inhibit TNFα-induced ICAM-1 expression in EA.hy926 cells. Phytomedicine. 2019 Jan;52:157-167.
5. Wang TS, Lin CP, Chen YP, Chao MR, Li CC, **Liu KL**. CYP450-mediated mitochondrial ROS production involved in arecoline N-oxide-induced oxidative damage in liver cell lines. Environ Toxicol. 2018 Oct;33(10):1029-1038. 0.347.
6. Wu YL, Chang JC, Lin WY, Li CC, Hsieh M, Chen HW, Wang TS, Wu WT, Liu CS, **Liu KL**. Caffeic acid and resveratrol ameliorate cellular damage in cell and Drosophila models of spinocerebellar ataxia type 3 through upregulation of Nrf2 pathway. Free Radic Biol Med. 2018 Feb 1;115:309-317. 11.7
7. **Liu KL**, Kuo WC, Lin CY, Lii CK, Liu YL, Cheng YH, Tsai CW. Prevention of 4-hydroxynonenal-induced lipolytic activation by carnosic acid is related to the induction of glutathione S-transferase in 3T3-L1 adipocytes. Free Radic Biol Med. 2018 Apr 23;121:1-8.
8. Chang TY, **Liu KL**, Chang CS, Su CT, Chen SH, Lee YC, Chang JS. Ferric Citrate Supplementation Reduces Red-Blood-Cell Aggregation and Improves CD163+ Macrophage-Mediated Hemoglobin Metabolism in a Rat Model of High-Fat-Diet-Induced Obesity. Mol Nutr Food Res. 2018 Jan;62(2).
9. Liu PJ, **Liu KL**, Liao EC. Specific Immunoglobulin G4 and Immunoglobulin E Titers to Common Food Antigens in Sera of Children With Allergic Rhinitis. Altern Ther Health Med. 2018 Nov;24(6):38-45

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營養與神經退化疾病

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Abstract
神經退化性疾病 (Neurodegenerative diseases) 是最常使人衰弱的疾病之一，通常與基因突變，異常蛋白質蓄積，活性氧（reactive oxygen species）增加或大腦特定部位的神經元破壞有關。目前常見與神經退化性疾病的發作和進展相關機制包含，神經元線粒體的損害，細胞內 Ca2 + 超載，ROS 的產生不受控制，持續的炎症狀況。儘管這些信號傳導的起因可能有所不同，但因為毒物誘導的神經元損傷和神經退化性疾病的進展擁有相似的機制,從而增強了這些途徑作為介入策略的重要性。 我們經由對神經元損傷的潛在機制的理解， 設計可能潛在可延遲發作和/或減輕神經退化進展。食物（指未加工或加工的全部或可食用部分）中營養素與沒有營養成分的物質，對人類健康有積極影響，稱為類藥劑營養品(Nutraceuticals)。類藥劑營養品是由食物萃取濃縮出來的成分，沒有明確的定義或適當的法規。Dr. Stephen De Felice 提出了將營養與藥物結合的術語，類藥劑營養品是指食品或食品中的一部分具有醫療或健康益處，可用於疾病的預防或輔助治療。類藥劑營養品可以是來自食品，膳食補充劑，基因工程食品(genetically engineered food)，草藥產品或加工食品的純化物。 類藥劑營養品是天然存在的生物活性成分，由從不同的食物中萃取而得，而功能性食物(functional food)則是新鮮或加工食品指聲稱具有超出食物中營養素所提供基本營養功能外的健康促進及/或疾病預防的特性。本演說將以介入神經退化性疾病相關機制的角度，介紹類藥劑營養品及富含類藥劑營養品飲食型態，介紹如何由營養來預防及/或減緩神經退化性疾病發生與進展。

《回議程》

謝慶良, MD, PhD

EDUCATION

1991/4-1995/3　醫學博士　日本國立九州大學 腦神經病研究所
1971/9-1978/6　醫學士　中國醫藥大學 中醫學系

CURRENT POSITION & PROFESSIONAL BACKGROUND

2019/8-present　中國醫藥大學針灸研究所教授

RESEARCH INTEREST(S)

- Stroke, Epilepsy
- Chinese Medicine
- Effect and Mechanisms

Honors and Awards(or Selected Publications)

1. 109 年度中醫藥優良學術著作獎，中華民國中醫師公會全國聯合會，2020.10.25
2. 台灣癲癇醫學會癲癇研究論文獎佳作，台灣癲癇醫學會，2018.04.22
3. 推廣中醫藥學術貢獻卓著，中華民國中醫師公會全國聯合會，2017.10.22
4. 台彎癲癇醫學會癲癇研究論文獎-佳作，台彎癲癇醫學會，2015.03.29
5. 中醫藥臨床試驗與研究頂尖研究獎，衛生福利部卓越臨床試驗中心，2014.01.01

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中醫在罕見疾病治療之角色

謝慶良 [1,2]
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Abstract

罕見疾病大都屬於遺傳性或先天性疾病，發病形式以慢性進行性或反覆發作為主要。臨床表現各式各樣包括智能、發育、骨骼、肌肉等。中醫對於疾病的劃分歸類為心、肝、脾、肺和腎等五大系統，即所謂的五臟論。臨床上出現與智能、意識等相關之症狀歸屬於心系統（心主神明），與運動相關則歸屬於肝（肝屬筋），與遺傳、先天、發育和退化相關則屬於腎（腎主骨藏精），與肌肉相關則屬於脾（脾主肌），與呼吸、皮膚和毛髮相關則歸屬於肺（肺主氣和毛髮）。綜合上述，罕見疾病涉及到中醫的五臟。依傳統中醫理論，如疾病屬於遺傳性、先天性或退化性治療以補腎為主，如肌肉萎縮等以補脾為主。腎為先天之本，而脾為後天之本，先天不足可以後天脾來治療。因此，推測補脾和腎是治療罕見疾病的主軸。

根據我所知，罕見疾病使用中醫治療而獲得良好效果的報告甚少，僅有一些零星的個案報告。因此，雖然理論上中醫能用於罕見疾病的治療，但在實證上有待驗證，或許一些療法如太極拳、遠紅外線或針灸等可提供作為輔助治療緩和某些臨床症狀。

《回議程》

張通銘, MD

EDUCATION

交通大學生物科技學系博士班(進修中)
長榮大學醫學研究所碩士
台北醫學大學學士

CURRENT POSITION & PROFESSIONAL BACKGROUND

彰基嬰兒照顧中心主任

彰基兒童神經科主治醫師

台中榮總小兒部住院醫師

小兒神經研究醫師

台北榮總新竹分院小兒科主任

RESEARCH INTEREST(S)

- 兒童神經性疾病(癲癇、頭痛、妥瑞、注意力過動症)
- 兒童發展評估檢查及治療
- 兒童遺傳代謝疾病診斷（罕見疾病之診斷）
- 一般兒科

Honors and Awards(or Selected Publications)

1. Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985-2019). Lin HY, Lee CL, Chang CY, Chiu PC, Chien YH, Niu DM, Tsai FJ, Hwu WL, Lin SJ, Lin JL, Chao MC, Chang TM, Tsai WH, Wang TJ, Chuang CK, Lin SP. Orphanet J Rare Dis. 2020 Nov 7;15(1):314.
2. Ictal and interictal electroencephalographic findings can contribute to early diagnosis and prompt treatment in KCNQ2-associated epileptic encephalopathy. Lee IC, Chang MY, Liang JS, Chang TM. J Formos Med Assoc. 2020 Aug 27:S0929-6646(20)30388-0.
3. Cardiac Evaluation using Two-Dimensional Speckle-Tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. Lin HY, Chuang CK, Lee CL, Chen MR, Sung KT, Lin SM, Hou CJ, Niu DM, Chang TM, Hung CL, Lin SP. Diagnostics (Basel). 2020 Jan 23;10(2):62.
4. Delay Maturation in Occipital Lobe in Girls With Inattention Subtype of Attention-Deficit Hyperactivity Disorder. Chang TM, Yang RC, Chiang CT, Ouyang CS, Wu RC, Yu S, Lin LC. Clin EEG Neurosci. 2020 Sep;51(5):325-330.
5. Systemic hypertension followed by insidious stroke in a 12-year-old boy with childhood neurofibromatosis type 1 presenting with renal and cerebral artery vasculopathy. Lee ML, Chang TM, Yang RC, Yang AD, Chen M. Turk J Pediatr. 2019;61(4):629-634.
6. Impact of mycoplasma pneumonia infection on urticaria: A nationwide, population-based retrospective cohort study in Taiwan. Yong SB, Yeh WC, Wu HJ, Chen HH, Huang JY, Chang TM, Wei JC. PLoS One. 2019 Dec 20;14(12):e0226759.
7. Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with

Different Types of Mucopolysaccharidoses. Lin HY, Lee CL, Chiu PC, Niu DM, Tsai FJ, Hwu WL, Lin SJ, Lin JL, Chang TM, Chuang CK, Lin SP. Diagnostics (Basel). 2019 Oct 14;9(4):148

8. An At-Risk Population Screening Program for Mucopolysaccharidoses by Measuring Urinary Glycosaminoglycans in Taiwan. Lin HY, Lee CL, Lo YT, Tu RY, Chang YH, Chang CY, Chiu PC, Chang TM, Tsai WH, Niu DM, Chuang CK, Lin SP. Diagnostics (Basel). 2019 Oct 5;9(4):140.
9. Critical Trio Exome Benefits In-Time Decision-Making for Pediatric Patients With Severe Illnesses. Wu ET, Hwu WL, Chien YH, Hsu C, Chen TF, Chen NQ, Chou HC, Tsao PN, Fan PC, Tsai IJ, Lin SP, Hsieh WS, Chang TM, Chen CN, Lee CH, Chou YY, Chiu PC, Tsai WH, Hsiung HC, Lai F, Lee NC. Pediatr Crit Care Med. 2019 Nov;20(11):1021-1026.
10. Functional independence of Taiwanese patients with mucopolysaccharidoses. Lee CL, Lin HY, Chuang CK, Chiu HC, Tu RY, Huang YH, Hwu WL, Tsai FJ, Chiu PC, Niu DM, Chen YJ, Chao MC, Chang TM, Lin JL, Chang CY, Kao YC, Lin SP. Mol Genet Genomic Med. 2019 Aug;7(8):e790.
11. KCNQ2 mutations in childhood nonlesional epilepsy: Variable phenotypes and a novel mutation in a case series. Lee IC, Chang TM, Liang JS, Li SY. Mol Genet Genomic Med. 2019 Jul;7(7):e00816.
12. Cardiac characteristics and natural progression in Taiwanese patients with mucopolysaccharidosis III. Lin HY, Chen MR, Lin SM, Hung CL, Niu DM, Chang TM, Chuang CK, Lin SP. Orphanet J Rare Dis. 2019 Jun 13;14(1):140

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可治療性之小兒神經罕病

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彰化基督教醫院嬰兒照顧中心

Abstract
小兒神經相關的罕見疾病非常廣泛，廣義來說兒童罕病牽涉到包括發展遲緩，神經肌肉疾病，癲癇，及運動障礙(involuntary movement)者皆可列入小兒神經罕病，其中可治療的罕病仍屈指可數。過去，可治療之罕病以溶小體儲積症(Lysosomal storage disease)為主，包括跟肌肉發展有關的龐貝氏症 Pompe disease，跟動作發展遲緩有關的第 4 型黏多醣症(mucopolysaccroidosis:MPS)，與神經認知遲緩有關的第一、二、六型黏多醣症。此外，以廣泛性(global)發展遲緩相關的可治療罕病有 Cerebral Creatine Deficiency Syndrome。去年開始，神經肌肉有關的脊髓性肌肉萎縮症 spinal muscular atrophy:SMA 已開始臨床治療。癲癇相關的可治療小兒神經罕病，包括 CoQ4 deficiency, glucose transporter deficiency, Pyridoxine-dependent epilepsy 及離子通道相關癲癇等(sodium, potassium)。運動障礙相關包括原發性陣發性動作型運動不良症（paroxysmal kinesigenic dyskinesia；PKD），兒童交替性偏癱（Alternating Hemiplegia of Childhood AHC)，苯酮尿症（Phenylketouria, PKU)，及 AADC deficiency 等。拜次世代定序(NGS:Next Generation Sequencing)診斷速度日新月異之賜，臨床醫師如何精準且儘早診斷可治療之小兒神經罕病是當代刻不容緩的責任。

《回議程》

蔡玲貞, RD

EDUCATION

輔仁大學

中山醫學院營養科學研究所碩士

CURRENT POSITION & PROFESSIONAL BACKGROUND

彰化基督教醫療財團法人彰化基督教醫院　營養部 主任

彰化基督教醫療財團法人彰化基督教醫院　體系營養部 主任

中華民國營養師公會全聯會　理事長

台灣省營養師公會　理事長

中華民國糖尿病衛教學會　常務理事、理事

台灣營養學會　常務理事、理事

考選部營養師審議委員

中山醫學大學　兼任講師

弘光科技大學食品營養系　兼任講師

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分享罕病照護計畫成果

蔡玲貞
彰化基督教醫療財團法人彰化基督教醫院

Abstract
彰基承接 106 年國健署計畫，符合政府「罕見疾病防治及藥物法」、「安寧緩和法」及「病人自主權利法」，提供衛福部罕見疾病個案四大服務及相關服務，亦提供尚未核准之罕病個案及家屬相關服務、關懷及支持。一. 照護方式：個管師定期電訪關懷 、門診訪視 、急住院訪視、到宅訪視。二. 罕病照護團隊模式運作:透過完整資訊系統、轉介專業照護流程、提供病人豐富資訊、定期會議、罕見疾病介紹及個案討論之教育訓練等，於本院鼎力支持下，以強化成員照護之賦能及責任，達到照護服務之目的。本院提供罕病個案全人照護及協助社政資源連結，持續追蹤及關懷需求的滿足。

《回議程》

廖淑芬, MD

EDUCATION

1986-1993	M.D.	國立陽明醫學院醫學系

CURRENT POSITION & PROFESSIONAL BACKGROUND

present	彰化基督教醫院復健部協同主任
	彰化基督教兒童醫院兒童復健科主任
	彰化基督教醫院復健部兒童復健科主任
	彰化基督教醫院體系輔具中心協同主任
	彰化基督教癌症e醫癌症復健科主任
	彰化基督教兒童醫院聯合評估中心遲緩鑑定醫師
2014-present	弘光科技大學物治系兼任助理教授
2005/9-present	長榮長榮大學護理系兼任助理教授
2001-present	彰化基督教醫院復健科永久主治醫師
1993-1997	台北台北榮民總醫院復健科住院醫師

RESEARCH INTEREST(S)

●兒童復健及早期療育

●淋巴水腫/癌症復健

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小兒罕病之復健醫學

廖淑芬 [1,2]
[1] 彰化基督教醫院復健部
[2] 彰化基督教兒童醫院兒童復健科

Abstract
罕病有 80%和基因是有關係的，絕大多數是發生於產前，或兒童時期發病。所以，罕病的兒童復健和發展遲緩是息息相關的。WHO 在 2001 年提出「國際機能、失能和健康分類(International Classification of functioning, Disabilities ,and Health, ICF)」的概念，從「身體功能和身體結構」及「活動和參與」來評估兒童之健康與失能(disability)，和環境及個人因素互相相關。對於罕病的照護復健，從多領域(multidisciplinary), 界領域(interdisciplinary)到跨領域(transidisciplinary)的介入，隨著醫療及新科技的進步，兒童復健的療綱是一直在變化的，希望可以提供一個以病人為中心，更完整的照護，最終可以改善其 health-related quality of life (HRQOL)。

《回議程》

陳彥廷, MD

EDUCATION

2012-2020	Institute of Clinical Medicine, National Yang-Ming University
1998-2005	Doctor of Medicine(MD), School of Medicine, Kaohsiung Medical University

CURRENT POSITION & PROFESSIONAL BACKGROUND

2011/10- Present	Member of the Ophthalmological Society of Taiwan (2011/10/31起) Member of the Taiwan Academy of Ophthalmology (2011/10/31起)
2010/7-Present	Attending Doctor, Department of Ophthalmology, Changhua Christian Hospital (2010/7/1-2010/8/31, 2011/9/5起)
2009/10-2010/5	Fellow Doctor, Department of Ophthalmology, Linkou Chang Gung Memorial Hospital (2009/10/1-2010/5/31)
2008/7-2009/6	Chief Resident Doctor, Department of Ophthalmology, Changhua Christian Hospital (2008/7/1-2009/6/30)

RESEARCH INTEREST(S)

- Mitochondrial eye disorders
- Neurological ophthalmology
- Pediatric ophthalmology

Honors and Awards(or Selected Publications)

1. 2009-6-25 彰化基督教醫院優良臨床教師獎
2. 2011-12 彰化基督教醫院血管暨基因體研究中心服務醫療奉獻獎
3. 2019-12-13 中華民國眼科醫學會-傅堯安教授論文獎: The relationship between optic atrophy 1 polymorphism and normal tension glaucoma in Taiwan

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以 Raxone (Idebenone)治療雷伯氏視神經萎縮症之臨床經驗

YanTing Chen [1,2,3,4]

[1]Vascular and Genomic Research Center, Changhua Christian Hospital

[2]Departments of Ophthalmology, Changhua Christian Hospital

[3]Institute of Clinical Medicine, National Yang-Ming University

[4]Department of Optometry, Central Taiwan University of Science and Technology

Abstract

Purpose

Leber's Hereditary Optic Neuropathy (LHON) is a mitochondrial disease which causes severe visual loss in young people. Here we reviewed our LHON patients in Taiwan with Raxone® (Idebenone, Santhera) therapy for 12 months, and summary their changes in visual acuity, visual field, and retinal nerve fiber layer thickness.

Methods

This is a retrospective study which reviewed patients with genetically confirmed G11778A type LHON and received Raxone 900mg daily for at least 12 months. The primary endpoint is the changes of LogMar visual acuity (VA) between baseline and 12th months follow-up. The secondary endpoints are the evolution of visual field, and retinal nerve fiber layer (RNFL) thickness.

Results

Six G11778A LHON patients with the mean age of 23.2 years were included in this study. The mean duration between LHON onset and Raxone therapy initiation is 301 days. An improvement of LogMar VA was found at 12th month in 6 eyes (50.0 %), and a worsening was observed in 2 eyes (16.7 %). Mean LogMar VA changes from 1.610 to 1.197 during these 12 months. Even though 5 eyes (41.7%) showed decreased mean defect in Humphrey 30-2 visual field exam at 12th month, the difference from baseline is very small. Average mean defect of visual field changes from -23.65 to -23.54 during these 12 months. The thickness of RNFL of these patients was all decreased at 12th months. Mean RNFL thickness changes from 72.83 to 55.5 during these 12 months.

Conclusions

Our results showed that Raxone therapy may improve LHON patients' visual acuity in Taiwan. It took 3-12 months to observe the first sign of VA improvement in our cases. We also found that younger patients seem to have better prognosis with Raxone therapy. Even though half eyes in this study got VA improvement, the visual fields of these patients are still poor.

《回議程》

蕭貞貞, MA

EDUCATION

2019/9-present	M.A.	東海大學社會工作研究所(博士班)
2007/9-2010/6	M.A.	清華大學(原新竹教育大學)教育心理與諮商所
1997/8-2000/6	M.A.	台南神學院宗教社會工作研究所

CURRENT POSITION & PROFESSIONAL BACKGROUND

2020/11-present	彰化縣諮商心理公會理事
2020/7-present	彰化縣政府心理健康促進暨精神疾病防治諮詢委員
2019/9-2022/9	澳亞醫學科學研究學會台灣分會第三屆監事
2018/1-present	中華民國社會工作師公會全國聯合會 第六屆-第七屆監事
2016/9-present	亞洲大學社會工作學系 兼任助理教授級專業技術人員
2015/7-present	彰化基督教醫院 心理諮商中心 主任/諮商心理師
1988/7-2015/6	彰化基督教醫院 精神科心衛生專科社工師

RESEARCH INTEREST(S)

- 精神病患疾病管理與復原性適應
- 職場心理健康及復原力
- 司法心理衡鑑與非志願性案主心理治療

Honors and Awards(or Selected Publications)

1. 行政院衛生署頒發「九十二年度家庭暴力推動暨性侵害防治業務卓越貢獻獎」
2. 法務部 93 年及 94 年『推動家庭暴力與性侵害有功人員』
3. 中華團體心理治療學會 95 年及 100 年『光智學術獎』
4. 彰化縣政府 107 年家庭暴力防治工作有功人員
5. 自創媒材《聽我說卡》及《我的翅膀在哪裡繪本》應用於醫療場域團體課程之探究—以神經罕見疾病病友會為例。輔導季刊，53(2)，22-34

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無症狀罕見疾病患者心理諮商

蕭貞貞
彰化基督教醫院新心理諮商中心

Abstract

罕見疾病多具遺傳性、且具備當今醫學科技仍無法治癒之屬性。無症狀之親屬目睹至親發病、身亡，因中常帶著恐懼，擔心同樣的情形會否也發生在自己身上？繼而變得憂鬱、焦慮。雖然，當代的基因醫學已經可以協助這些無症狀親屬預先得知自己未來有無發病之可能，減少因為不確定性帶來的恐慌，甚至，也可以幫助無症狀者提前透過運動、營養、藥物保健來延緩退化。但是，在確定自己罹患罕見疾病後，有多少人可以挺過心理上的衝擊？

彰化基督教醫院的罕病照護團隊透過醫師、遺傳諮詢師、諮商心理師的跨領域合作，以人類遺傳學會公告之遺傳檢驗及諮詢一般倫理準則為基礎，針對無症狀求診者，設計基因檢測前的遺傳諮詢、心理社會風險評估，以及檢測後的壞消息告知、悲傷輔導服務，以「全程」的照護服務，預防心理脆弱性較高者暴露於得知診斷後的心理衝擊當中，並能夠接納悲傷，在往後的生活中與疾病和諧共處，恢復生活品質。三年多來，已服務 33 人次，未來我們也將繼續優化服務流程，期能提供患者更為完善的全人照護。

《回議程》

王祖興, PhD

EDUCATION

1991/9-1996/12	Ph.D.	Institute of Radiation Biology, Tsing Hua University (Taiwan)
1986/9-1988/6	MSc	Department of Biology, Tunghai University (Taiwan)
1982/9-1986/6	BSc	Department of Biology, Tunghai University (Taiwan)

CURRENT POSITION & PROFESSIONAL BACKGROUND

2017/8-present	Chief-Secretary. (Secretariat Office, Chung Shan Medical University (Taiwan)
2016/8-present	Dean, Office of Library & Information Services, Chung Shan Medical University (Taiwan)
2010/8-2016/7	Dean, College of Medical Science and Technology, Chung Shan Medical University (Taiwan)
2009/8-present	Professor. (Department of Biomedical Sciences, Chung Shan Medical University (Taiwan)
2009/8-present	Professor. (Department of Biomedical Sciences, Chung Shan Medical University (Taiwan)

RESEARCH INTEREST(S)

- Stress response in mammalian cells
- DNA damage & repair
- Natural antioxidant

Honors and Awards(or Selected Publications)

1. Wang TS, Lin CP, Chen YP, Chao MR, Li CC, Liu KL. CYP450-mediated mitochondrial ROS production involved in arecoline N-oxide-induced oxidative damage in liver cell lines. Environ Toxicol. 2018 Oct;33(10):1029-1038.
2. Tian ZH, Weng JT, Shih LJ, Siao AC, Chan TY, Tsuei YW, Kuo YC, Wang TS, Kao YH. Arecoline inhibits the growth of 3T3-L1 preadipocytes via AMP-activated protein kinase and reactive oxygen species pathways. PLoS One. 2018 Jul 16;13(7):e0200508.
3. Lee JS, Lin YY, Wang TS, Liu JY, Lin WW, Yang JJ. Antitumorigenic Effects of ZAKβ, an Alternative Splicing Isoform of ZAK. Chin J Physiol. 2018 Feb 28;61(1):25-34.
4. Wu YL, Chang JC, Lin WY, Li CC, Hsieh M, Chen HW, Wang TS, Wu WT, Liu CS, Liu KL. Caffeic acid and resveratrol ameliorate cellular damage in cell and Drosophila models of spinocerebellar ataxia type 3 through upregulation of Nrf2 pathway. Free Radic Biol Med. 2018 Feb 1;115:309-317.
5. Lee JS, Wang TS, Lin MC, Lin WW, Yang JJ. Inhibition of Curcumin on ZAKα Activity Resultant in Apoptosis and Anchorage-Independent Growth in Cancer Cells. Chin J Physiol. 2017 Oct 31;60(5):267-274.
6. Wu YL, Chang JC, Lin WY, Li CC, Hsieh M, Chen HW, Wang TS, Liu CS, Liu KL. Treatment with Caffeic Acid and Resveratrol Alleviates Oxidative Stress Induced Neurotoxicity in Cell and Drosophila Models of Spinocerebellar Ataxia Type3. Sci Rep. 2017 Sep 14;7(1):11641.
7. Huang YC, Tsai MS, Hsieh PC, Shih JH, Wang TS, Wang YC, Lin TH, Wang SH. Galangin

ameliorates cisplatin-induced nephrotoxicity by attenuating oxidative stress, inflammation and cell death in mice through inhibition of ERK and NF-kappaB signaling. Toxicol Appl Pharmacol. 2017 Aug 15;329:128-139.

8. Lii CK, Chang JW, Chen JJ, Chen HW, Liu KL, Yeh SL, Wang TS, Liu SH, Tsai CH, Li CC. Docosahexaenoic acid inhibits 12-O-tetradecanoylphorbol-13-acetate-induced fascin-1-dependent breast cancer cell migration by suppressing the PKCδ- and Wnt-1/β-catenin-mediated pathways. Oncotarget. 2016 May 3;7(18):25162-79.
9. Hu CW, Chang YJ, Hsu YW, Chen JL, Wang TS, Chao MR. Comprehensive analysis of the formation and stability of peroxynitrite-derived 8-nitroguanine by LC-MS/MS: Strategy for the quantitative analysis of cellular 8-nitroguanine. Free Radic Biol Med. 2016 Dec;101:348-355.
10. Lin BH, Tsai MH, Lii CK, Wang TS. IP3 and calcium signaling involved in the reorganization of the actin cytoskeleton and cell rounding induced by cigarette smoke extract in human endothelial cells. Environ Toxicol. 2016 Nov;31(11):1293-1306.

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光線與健康-遠紅外線

王祖興
生物醫學科學學系，中山醫學大學(台中，台灣)

Abstract
太陽光是人類生存的條件之一，長久以來生物在陽光的洗禮下，演化出許多特殊的生理活動，例如植物的光合作用、動物的視覺、晝夜節律及維生素 D 的製造。太陽光的組成中，包含有明顯破壞能力的紫外光(3%)、可見光(44%)及紅外光(53%)。常態下，這些組成在人類整體健康上各自發揮其角色，然而，在現今的「宅」生活方式，一般人普遍發生太陽光照射不足的現象，這與人類的平均體溫降低(37℃ 鯷 36.5℃)，甚至與健康趨勢走低之間，可能有密切的相關性，值得注意並探討。如何了解並運用太陽光的組成，輔助人類因不愛動、日照不足的低體溫、低免疫防護力現況，或是在疾病下的「祛邪扶正」，相對於藥物治療或侵入式治療， 遠紅外線照射是一安全、方便的自然輔助性療法，透過其熱效應及非熱效應，增加身體心血管系統的流運行，進而促進新陳代謝及免疫系統能力，或合併化學藥物或奈米顆粒刺激神經細胞或殺死癌細胞等間接作用，維護身體健康。雖然，臨床實務已將遠紅外線照射列入洗腎患者的血液廔管照護，但遠紅外線照射如何發揮對身體不同細胞的有利效果，以及其分子作用機制，至今都仍不清楚，亟待更多的研究了解並開發有效的照射模式，促進其人類的健康。

《回議程》

吳怡磊, MD

EDUCATION

1997-2005　M.D.　中國醫藥大學中西醫學士

CURRENT POSITION & PROFESSIONAL BACKGROUND

2018-present　台灣兒科醫學會青少年醫學委員

2012-present　彰化基督教兒童醫院兒童內分泌科主任

2011-present　中華民國醫用超音波會員

2011-present　糖尿病衛教學會衛教人員

2011-2012　台北馬偕紀念醫院兒童急救加護醫學主治醫師

2011-2012　台北馬偕紀念醫院小兒內分泌暨新陳代謝專科主治醫師

2009-2011　台北馬偕紀念醫院小兒內分泌暨新陳代謝專科研究醫師

2006-2011　台北馬偕紀念醫院小兒科住院醫師/總醫師

RESEARCH INTEREST(S)

● Pediatric endocrinology

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McCune Albright 氏症候群分享

吳怡磊
Department of pediatric endocrinology, Changhua Christian Children's Hospital, Changhua, Taiwan

Abstract
McCune-Albright syndrome (MAS) is a rare genetic disordered characterized by the triad of 1) skeletal lesions- mono/polyostotic fibrous dysplasia, 2) hyperfunctioning endocrinopathies-gonadotropic -independent precocious puberty, hyperthyroidism, growth hormone excess and hypercortisolism in neonatal period , and 3) skin hyperpigmentation- café-au-lait spots. The prevalence of MAS is estimated to be between 1 in 100,000 to 1 in 1,000,000, but rare cases reported in Taiwan.
MAS results from post zygotic gain-of-function mosaic somatic activating pathogenic variants in GNAS gene, which encodes the cAMP pathway-associated G-protein, Gsα. Affected tissues can include those derived from ectoderm, mesoderm, and endoderm, and commonly include skin, skeleton, and certain endocrine organs. The mutation is distributed in a mosaic pattern and presenting signs and symptoms are dependent on the time frame the mutation occurs. In bone, increased cAMP causes osteoblasts to differentiate into stromal cells while inhibiting further differentiation, resulting in fibrous dysplasia. Increased cAMP signaling in the skin results in stimulation of melanin production via alpha-MSH, resulting in café-au-lait macules. A variety of endocrine disorders, including hyperthyroidism, acromegaly, phosphate wasting, and Cushing syndrome are now considered as part of the endocrinopathies seen in this disorder. MAS is largely a clinical diagnosis. Genetic testing for GNAS mutation is commercially available, however, due to the mosaicism, false negatives do occur, and a positive result contributes little to the clinical management.

《回議程》

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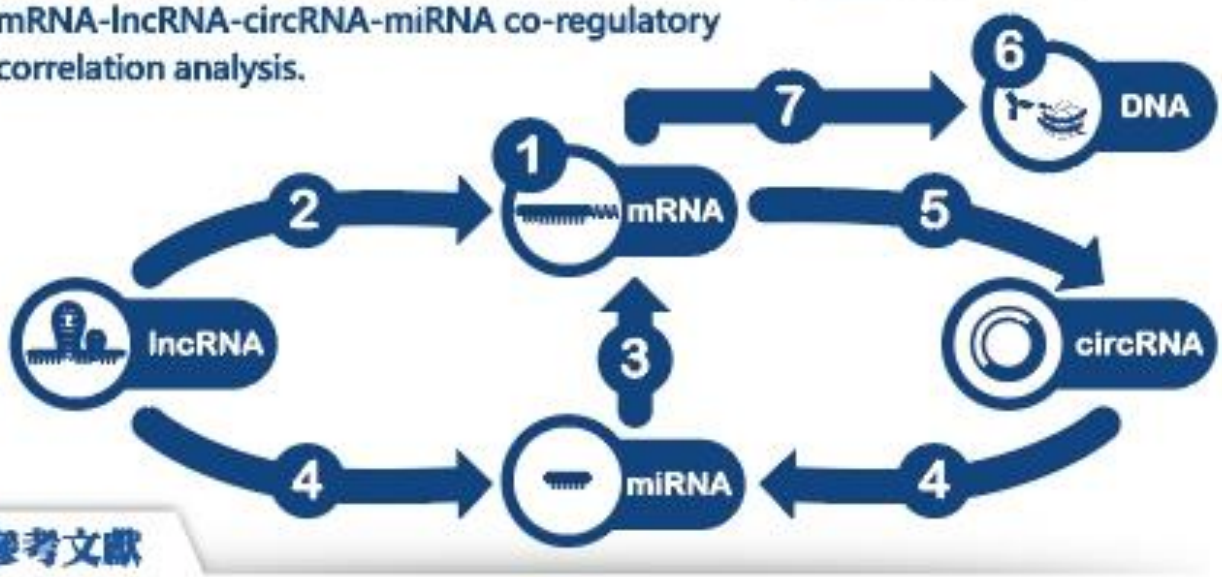
6. Epigenomics analysis ChIP-Seq
7. Joint analysis of mRNA and DNA

All mRNA-lncRNA-circRNA-miRNA co-regulatory correlation analysis.



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The Journal of Allergy and Clinical Immunology | Nucleic Acids Research | BMC Genomics | PLOS ONE

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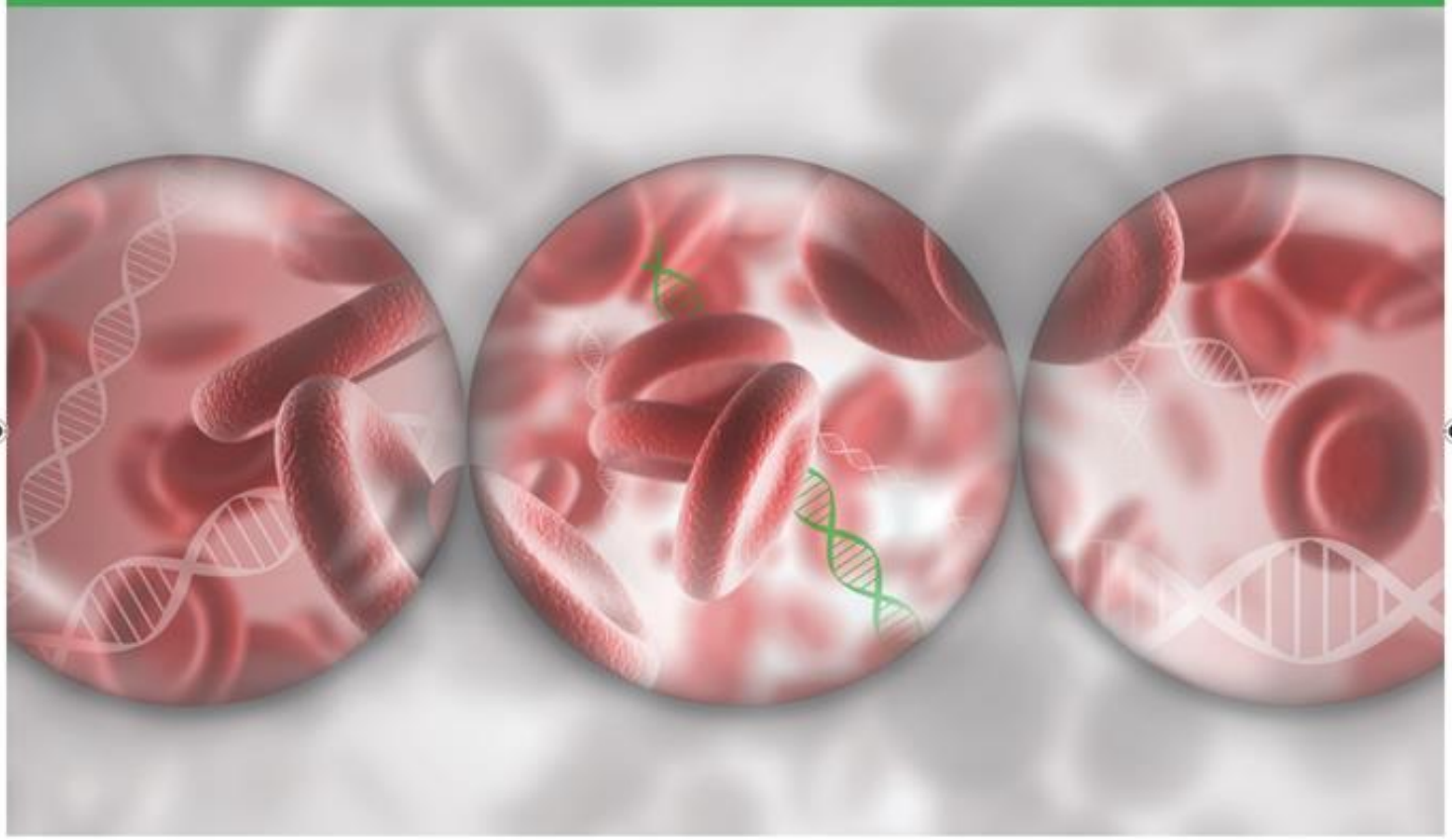

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