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陳明
SY7

Difficult Scenarios in Genetic Diagnosis and Counseling

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Genetic diagnosis and counseling becomes more and more challenging because of the rapid advancement of genotyping tools, it has undergone rocketing breakthrough in the past two decades. The speaker witnessed and participated its progress since his overseas studies back in Glasgow twenty years ago and had established a full spectrum of genetic diagnosis under one roof with the facilities of cytogenetics, molecular cytogenetics, molecular genetics (including Sanger, next generation sequencing). He developed in-house tests such as NIPT, PGT, WES, and now WGS services and his lab had awarded CAP accreditation since 2011, one of the first few labs in Asia-Pacific region of its kind and is the only member who was elected Fellow in the famous close-door academic elite "Fetoscopy Group" from Taiwan. The speaker also participated numerous patents of genomics and bioengineering in Taiwan, China, US, and EU. However, in this talk the speaker will not focus on "what he can do" or "how competent his lab is" but rather to demonstrate the "misdiagnosis" experience of his own to highlight the difficulties of genetic diagnosis and counseling. With this selfless sharing of his experience the speaker would like to explicit his view of this rapid changing field (in some senses, deeply infiltrated by the commercial parties), and to offer a more comprehensive and informative way for the clinical colleagues to have a more sensible attitude to hold when doing clinical practice and not to underscore its risks.

■專題演講──產科

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林芷伃
SY8

Expand genetic carrier screening and counseling

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　　帶因篩檢，顧名思義就是將沒有發病的基因突變帶因者找出來，目的是希望可以避免將基因疾病傳給下一代，或者是在早期發現患者，讓患者有機會及早接受治療。台灣的產前檢查在這二十年間進步得相當飛快，早在 2005 年，台灣就開始大規模地篩檢「脊髓性肌肉萎縮症帶因者」，後來更推出「X 染色體脆折症」帶因篩檢，甚至是「新生兒聽力基因篩檢」，末不是跑在世界的先端。但是考量到人力與時間，過去的基因檢測的數目受到限制和價格居高不下。然而，隨著基因診斷技術的進步，next generation sequencing 的技術普及與價格愈來愈親民，在 2021 年美國基因暨基因體醫學會與美國婦產科醫學會提出臨床建議，認為所有孕婦或是準備懷孕者，都應該被提供帶因率高於 1/200 的隱性遺傳疾病以及性聯遺傳疾病的篩檢機會，她們的配偶也可以同時接受這些隱性遺傳疾病的篩檢。台灣的大範圍帶因篩檢也在 2020 年就開始慢慢推廣，我們發現，有高達七成的受檢者會驗出至少一種隱性遺傳疾病的帶因，甚至有將近 6%的受檢者身上帶有三種以上的隱性遺傳疾病帶因。透過這樣的大範圍基因篩檢，我們可以在孕期或者懷孕早期就發現帶有相同隱性遺傳疾病的帶因者夫妻。在過去，這些家庭往往都是生下第一個發病的患童後，才會發現這些遺傳疾病;現在，經過遺傳諮詢後，這些夫妻可以透過胚胎著床前基因診斷或是早期的絨毛膜穿刺或羊膜穿刺來確認胎兒的基因型。及早發現這些隱性遺傳的問題，一方面有機會讓這些疾病不會造成家庭的負擔或悲劇，另一方面，也是讓這些未來可能的發病者持續接受適當的醫療追蹤與及早治療的機會，造福許多家庭。

施景中
SY9

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ChatGPT in obstetrics and gynecology

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ChatGPT is a natural language generation system that use deep neural networks to generate coherent and fluent writings depending on input. It is one of the most advanced and adaptable natural language processing models, and it has been used to a variety of fields including conversation systems, text summarization, text completion, and creative writing.

In this session, I will present an updated overview of the ChatGPT literature in the field of obstetrics and gynecology. One of the problems of obstetrics and gynecology is providing patients with individualized and timely information, particularly during pregnancy and childbirth. Patients may have a variety of issues, questions, preferences, and expectations regarding their health and treatment alternatives. Furthermore, during pregnancy, birth, or gynecologic surgery, individuals may suffer worry, stress, discomfort, or mental anguish.

Obstetricians and gynecologists can utilize ChatGPT as a tool to help them with patient education and assistance. ChatGPT may provide conversational and captivating messages that can provide answers to frequently asked queries, pertinent information, an explanation of a medical word, practical advice, or emotional support. The tone and style of ChatGPT may also be changed to accommodate the patient's temperament, personality, and educational background.

Instead than replacing real physicians and nurses, ChatGPT is designed to support them and improve the quality of their care. Consistent, approachable, individualized, and interesting information and assistance are all things ChatGPT can offer. By taking care of some of the typical or repetitive activities, ChatGPT can help lessen the effort and stress on the medical personnel. Last but not least, ChatGPT still needs improvement and has several drawbacks. It's possible that ChatGPT doesn't always provide precise or pertinent messages for every circumstance or patient. Concerns around data privacy, permission, responsibility, or accountability may also arise with ChatGPT.

許晉婕

SY10

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　　　林口長庚醫院婦產部產科研修醫師
　　　台大分醫所遺傳諮詢組碩士(進修中)

產婦重大生產事故與死亡之原因分析與處置建議

孕產婦於周產期死亡常導致家庭重大創傷，並影響新生兒的成長與照顧。

WHO 希望在 2030 年前將全球周產期死亡率(maternal mortality ratio, MMR)降至每十萬活產數小於 70 位母親死亡，目前國際上成績最優秀的國家為澳洲及紐西蘭(MMR=4)，而台灣近十年平均 MMR 為 11.3，尚有進步空間。此外，近年來由於大眾晚婚晚生，生育年齡不斷上升，高危險妊娠的比率亦大幅增加。國內出生率雖年年下降，但周產期死亡率卻節節上升，由十年前的 8.5 爬升至民國 110 年的 14.0，平均每年約有 21 位孕產婦死亡。

本講題將回顧林口長庚醫院近六年之生育事故通報及救濟紀錄，呈現孕產婦死亡率及死亡原因，並討論幾近死亡(near-death)或發生重大併發症的急重危症案例。希望藉由案例討論，強調在搶救孕產婦性命的關頭，跨團隊、跨科部、甚至是跨院合作的重要性。最後，針對常見的孕產婦急重危症(產後大出血、子癇症/子癇前症、肺栓塞、孕產婦突發性心跳停止)給予處置及轉診建議。期待能強化同業的臨床反射，練習臨危不亂、處變不驚，於最短的時間組織團隊，搶救生命。

陳宜雍
SY11

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胎兒與新生兒死亡之原因分析與處置建議

　　隨著社會演變，晚婚少生子現象愈益明顯，台灣新生兒數屢創新低，去年全年新生兒數僅 13 萬 8986 人，因此維護胎兒及新生兒健康就變得更加重要。 然而生產過程存在不可預期性的風險，為共同承擔婦女生產的風險，我國自民國 105 年開始施行「生產事故救濟條例」，其中胎兒及新生兒死亡也納入救濟範圍。本次內容將依據過往生產事故救濟審議案件，統整胎兒及新生兒死亡個案並加以分析，提供作為第一線照護胎兒及新生兒的婦產科醫師參考，一起為增進母嬰健康努力。

蘇美慈
SY12

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子癲前症新觀點:診斷及治療

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Preeclampsia is a severe gestational hypertensive disorder that leads to maternal multiple organ dysfunction and adverse fetal outcomes. Contemporary evidence suggests preeclampsia is a two-stage disease. The first stage is an early pregnancy asymptomatic stage, resulting from poor placentation due to abnormal trophoblast invasion and spiral artery remodeling. The second stage of the disease in later gestation is characterized by a placental ischemia/reperfusion injury and a maternal immunemediated response, which leads to an angiogenic imbalance, immune-mediated exaggerated inflammatory response, and endothelial cell dysfunction. The overall consequence of this cascade results in the clinical manifestations of preeclampsia.

There has been increasing research momentum to identify new therapeutic agents for the prevention or treatment of preeclampsia, drugs that can affect the underlying disease pathophysiology. This talk will provide new insighs of pathoetiologies of preeclampsia and review some potential drugs that are ongoing or registered for preeclampsia-associated clinical trials, such as pravastatin, proton-pump inhibitors, metformin, micronutrients, and biologics. With reassuring and positive findings from pilot studies and strong biological plausibility, some candidate or repurposed drugs may be a preventative or therapeutic agent for preeclampsia in the near future and provide a delicated care for maternal-fetal medicine.