初福傑
SY21

現職：台北長庚醫院婦產科 主治醫師
經歷：台北長庚醫院婦產科 臨床細胞遺傳學研究員
台北長庚醫院婦產科 研究醫師
林口長庚醫院婦產部 住院醫師

帶因篩檢

Fu-Chieh Chu, MD
Department of Obstetrics and Gynecology, Taipei Chang Gung Memorial Hospital, Taipei, Taiwan

單基因遺傳疾病的遺傳模式可分為體染色體顯性，體染色體隱性以及性聯遺傳。目前已知的單基因遺傳疾病約一萬種以上，全球的總發生率約為 1/100, 大約造成 20%的新生兒死亡率及住院原因。隱性遺傳疾病的帶因者是完全沒有症狀的健康人，但是會有較高的風險生下罹患隱性遺傳疾病的孩子。帶因篩檢 (carrier screening) 則是幫助我們在孕前或是產前了解夫妻雙方是否為隱性遺傳疾病的帶因者，不僅了解未來自己孩子的患病風險，也可以根據本身的宗教信仰和價值觀，來決定產前進一步的遺傳檢測項目。

由於基因的普及檢測牽涉倫理道德規範的考量，在 2015 年時，ACMG, ACOG, NSGC, SMFM 共同提出一份意見聲明，針對帶因篩檢所帶來的基因體世代倫理議題，強調基因檢測的過程前後都需接受遺傳諮詢，資料的保密作業，專業人員的培訓，對於好壞基因的價值判斷，甚至是對於疾病的污名化的議題。

在 2017 年時，美國婦產科醫學會 ACOG 則提出針對帶因篩檢更進一步的意見聲明，認為隱性遺傳疾病的檢測應該不再受限於種族背景，任何的種族都可以考慮同時接受多種疾病的帶因篩檢，可接受泛種族帶因篩檢 (panethnic carrier screening) 和多疾病帶因篩檢 (expanded carrier screening) 做為孕前產前帶因篩檢的選擇。納入篩檢的隱性遺傳疾病要有較高的帶因率，明確的臨床表現造成嚴重身體或認知功能的傷害，以及會在嬰幼兒時期就發作的疾病。另外 ACOG 也強調帶因篩檢不能和現有的新生兒篩檢兩者劃上等號。針對泛種族帶因率較高的的疾病，則是每一位孕婦都應該做檢測，包含脊髓型肌肉萎縮症，囊腫性纖維化，和血球異常疾病。

如果帶因篩檢檢測出為體染色體隱性遺傳疾病的帶因者，進一步應該檢測其配偶同樣疾病的帶因篩檢。如果夫妻雙方都是同一疾病的帶因者時，則下一代的孩子都有 1/4 的風險會同時遺傳到兩個異常基因而罹患隱性遺傳疾病。所以雙帶因的夫妻在確定懷孕後的產前檢測可以安排絨毛膜採檢或羊膜穿刺來確定胎兒是否罹病。或於準備懷孕前進行人工生殖的輔助，安排孕前胚胎著床前基因檢測 (preimplantation genetic diagnosis, PGD)，以確認胚胎是否罹患隱性遺傳疾病。

X 染色體性聯隱性遺傳疾病的遺傳模式則和體染色體不完全一樣。男性只要有一個 X 染色體致病基因異常就會罹患 X 染色體隱性遺傳疾病。而女性如果只有一個 X 染色體致病基因異常則為沒有症狀的帶因者。X 染色體性聯隱性遺傳疾病的篩檢只需要針對女性進行即可，如果檢測出女方為性聯隱性遺傳疾病的帶因者時，下一步直接對胎兒或胚胎進行確認。

現階段國內由政府所補助的常規帶因者檢測只有海洋性貧血帶因者的篩檢，針對台灣種族帶因率較高的隱性遺傳疾病，包含脊髓型肌肉萎縮症和 X 染色體脆折症，產檢醫師也應建議每位孕婦進行篩檢。現今台灣社會越來越多的多種族融合且進步，隱性遺傳疾病的帶因也可能更加多樣化。以預防醫學的角度來看，產科醫師需協助孕育健康的下一代，減少嚴重先天異常疾病的發生，也協助進行早期診斷與及時治療來爭取未來治療的黃金時間。

林苾伃
SY22

現職：台大醫學院婦產科副教授
台大醫院婦產部訪問學者
現職：台大醫院基因醫學部 研修醫師
台大醫院婦產部 研修醫師
台大醫院新竹分院婦產部 主治醫師

Chromosomal Microarray Analysis: for selected or unselected women with amniocentesis?

Shin-Yu, Lin
Dept. of OBS&GYN, National Taiwan University Hospital

Chromosomal microarray analysis can identify chromosomal aneuploidy and other large changes in the structure of chromosomes that would otherwise be identified by standard karyotype analysis, as well as submicroscopic abnormalities that are too small to be detected by traditional modalities. In 2016, The ACOG and the Society for Maternal– Fetal Medicine made the following recommendations and conclusions for the use of chromosomal microarray analysis in prenatal diagnosis—"This test typically can replace the need for fetal karyotype in cases with fetal sonographic anomalies." Down syndrome risk is increased with maternal age. However, most submicroscopic genetic changes identified by chromosomal microarray analysis are not associated with increasing maternal age; therefore, the use of this test can be considered for all women, regardless of age. I will talk about the pros and cons when applying CMA in prenatal amniocentesis.

應宗和
SY23

現職：中山醫學大學醫學系 副教授 / 婦產學科主任
台灣婦產科醫學會理事
台灣周產期醫學會常務理事

Thyroid function screening in pregnancy: risk factor-based or universal?

Hyperthyroidism occurs in 0.2– 0.7% of pregnancies. Inadequately treated maternal hyperthyroidism is associated with (1). Greater risk preeclampsia with severe features (2). Maternal heart failure (3). Thyroid storm than treated controlled maternal thyrotoxicosis. Inadequately treated hyperthyroidism for fetal outcome is associated with an increase in (1). Medically indicated preterm deliveries (2). Low birth weight (3). Miscarriage, and stillbirth.

Overt hypothyroidism complicates 0.2– 1.0% of pregnancies. May be indistinguishable from common signs or symptoms of pregnancy, such as (1). Fatigue, constipation, cold intolerance, (2). Muscle cramps, and weight gain (3). Edema, dry skin, hair loss, and a prolonged relaxation phase of deep tendon reflexes. Adverse perinatal outcomes such as (1). Spontaneous abortion, preeclampsia, preterm birth (2). Abruptio placentae, and stillbirth (3). Low birth weight and impaired neuropsychologic development.

Universal screening for thyroid disease in pregnancy is not recommended by The American College of Obstetricians and Gynecologists, the Endocrine Society, and the American Association of Clinical Endocrinologists. But indicated testing of thyroid function should be performed in women. Such as (1). A personal or family history of thyroid disease (2). Type 1 diabetes mellitus, or clinical suspicion of thyroid disease.

The American Thyroid Association currently finds that there are insufficient data to recommend for or against universal thyroid screening.

[published erratum appears in Thyroid 2017;27:1212] Thyroid 2017;27:315– 89. (Level III)

葛菁如
SY24

現職：高醫附設醫院婦產部 主治醫師
經歷：高醫附設醫院婦產部 研修醫師
高醫附設醫院婦產部 總醫師
高醫附設醫院婦產部 住院醫師

Shall We Screen for Vitamin D Deficiency and Iron Deficiency Anemia at the First Antenatal Examination?

Chin-Ru Ker, MD
Department of OB/GYN, Kaohsiung Medical University Hospital, Kaohsiung, Taiwan

Undesirable perinatal outcomes including gestational diabetes, pre-eclampsia, preterm birth, low birth weight, stillbirths and Cesarean section rates are found to be associated with low vitamin D level, which also directly influence vitamin D status and bone health in the neonates. Most practice guidelines around the world suggest routine supplement of colecalciferol 400 units daily for all women and endorse a stepwise approach to access vitamin D status. At booking appointments, pregnant women should be accessed for risk factors such as vegetarian, limited sun exposure, dark skin, veiled customs, ethnic minorities, office workers, night shifters and body mass index greater than 30-40 kg/m2. If present, 1000 units of colecalciferol daily should be given and serum level of 25-hydroxy vitamin D level is measured to guide further management. However, the supporting reports are inconsistent and largely heterogenous in study designs, seasonality, latitudes, body mass index, vitamin D supplementation and laboratory methodology. Recommendation of vitamin D supplementation beyond that contained in a prenatal vitamin should await more robust evidence from better designed studies.

Iron deficiency anemia in pregnancy is linked to poor gestational weight gain, fetal growth restriction, preterm delivery, delivery complications and depression in the mother; the newborns can also be iron deficient with impairments in cognition and neurodevelopment. Current practices in the United States, United kingdom, Australia and Canada are agreeable in screening for anemia with full blood count at booking appointments. If anemic (Hgb< 110g/L), a trial of oral iron supplement (60-100mg per day) is usually initiated, of which a lack of response prompts subsequent investigations for other causes. Serum ferritin testing should be reserved for possible hemoglobinopathies, anemia of infection, vitamin B12 or folic acid deficiency, unexplained iron deficiency or suspected chronic blood loss. At present, there is insufficient evidence to recommend for routine screening for iron status or routine iron supplementation in pregnant women. Higher quality evidence from randomized control trials and economic studies are needed to assess the clinical and cost effectiveness of such practices.

王伊蕾
SY25

現職：幸福婦產科診所負責人
經歷：前台北榮民總醫院婦產科專科醫師
前竹東榮民醫院婦產科主治醫師
前桃園榮民醫院婦產科主治醫師
前台北榮總不孕生殖科臨床研究員

CS rates only 2%? 再談無國界醫生如何幫助阿富汗產婦

幸福婦產科診所　王伊蕾醫師

在醫療資源不足的地方，無國界醫生(Medecins Sans Frontieres, MSF)希望能盡量減少剖腹生產的機會，以減少日後婦女因戰爭或天災，必須在家生產，產生子宮破裂，植入性胎盤，產後出血等致命合併症的可能。WHO 的統計數據也顯示，如果剖腹生產率超過 10%，無助於減少胎兒或母親的死亡率。

2017 年統計，阿富汗霍斯特母嬰親善醫院(Khost Maternity Hospital, KMH)全年生產數 22856 人次，剖腹生產 513 人次，剖腹生產率只有 2.24%。這是如何做到的呢? 以最簡陋的設備，挑戰最艱困的病例。台灣第一個婦產專科無國界醫生分享在阿富汗的第一手觀察紀錄 。

吳佩臻
SY26

現職：台兒診所/台兒中山集英聯合診所主治醫師
經歷：台北馬偕婦產部住院醫師
台北馬偕生育保健科研修醫師
台兒診所研修醫師

Symposium First Trimester Anatomical Screening: Nuchal Translucency and Beyond

It is well documented that a large nuchal translucency is associated with increased risk of structural anomalies and genetic syndromes, even in the absence of aneuploidy. Focusing only on screening for chromosomal anomalies and postponing the anatomical assessment to the second trimester may result in delaying the detection of major structural malformations.
The performance of early anatomical scan for most abnormalities ultimately depends on their association with easily detectable markers and a policy decision as to the objectives of the scan.
Which Anomalies Should be Targeted in Early Pregnancy? [2019 ISUOG]

1. Nearly always detectable (approximately 90-100%):
   (1) Severe CNS anomalies (anencephaly, alobar holoprosencephaly, encephalocele)
   (2) Ectopic cordis
   (3) Abdominal wall defects (omphalocele, gastroschisis, limb-body wall complex / body stalk anomaly, megacystis)
2. Potentially detectable (approximately 2-90%):
   (1) Congenital diaphragmatic hernia
   (2) Major heart defects (TGA, DORV, CoA, HLHS, septal defects)
   (3) Spina bifilar
   (4) MCDK
   (5) Skeletal disorders (lethal skeletal dysplasia, limb reduction, polydactyly)
3. Virtually undetectable (<2%):
   (1) Cerebellar hypoplasia
   (2) Agenesis of corpus callosum
   (3) Echogenic lung lesions (CPAM, extralobar BPS)
   (4) GI disorders (duodenal artesian, bowel obstruction, anal atresia)
   (5) Mild renal anomalies (duplex kidneys, hyponephrosis)
   (6) Ovarian cysts
   (7) Fetal tumors

Early suspicion of lethal or severe fetal abnormalities allows not only for change in the diagnostic pathway (e.g. invasive genetic testing in preference to cfDNA), but also for timely reproductive choices and optimised clinical management. In these patients, early decision-making has the additional benefit of being associated with lower long-term psychological morbidity.