本期企劃

從代謝疾病到基因世代：新生兒篩檢法律議題之挑戰與台灣展望

From Metabolic Disorders to the Genomic Era: Legal Challenges of Newborn Screening and the Outlook for Taiwan

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摘 要

新生兒篩檢自1980年代在台灣以代謝疾病為核心推行，透過乾血滴檢體與串聯式質譜技術，建立了近乎百分之百涵蓋率的公共衛生防護網。然而，基因定序技術的快速進展，正推動篩檢進入「基因世代」，帶來資訊過量、疾病邊界模糊、心理社會衝擊等新挑戰，也使既有Wilson-Jungner原則的適用性受到質疑。基因世代的篩檢引發多重法律爭點，包括篩檢範圍如何界定、父母決定權與兒童最佳利益的平衡、同意程序的有效性、檢體與基因資料保存的規範，以及基

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關鍵詞：父母同意權（parental consent）、法律議題（legal issues）、基因定序（genomic sequencing）、新生兒篩檢（newborn screening）、隱私與歧視（privacy and discrimination）

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因資訊可能導致的歧視風險。國際經驗顯示，美國、英國與德國等國雖探索基因定序，但常規清單仍以代謝疾病為主，基因篩檢多停留於研究或試辦階段，反映政策的審慎態度。台灣同樣面臨基因世代帶來的法律與倫理挑戰，現行制度在篩檢範圍、同意與揭露程序、資料保存與隱私規範等方面仍存不足，但已有初步建議，如建立專責審議機制、調整同意內容、強化隱私保障與反歧視規範。未來如何進一步落實，將是台灣在守護新生兒健康與建構社會信任間的關鍵課題。

Since the 1980s, newborn screening (NBS) in Taiwan has centered on metabolic disorders, supported by dried blood spot collection and tandem mass spectrometry, establishing a nationwide safety net with near-universal coverage. However, the rapid advancement of genomic sequencing technologies has ushered NBS into the "genomic era," characterized by challenges of information overload, blurred disease boundaries, psychosocial impacts, and uncertainties that strain the applicability of the Wilson–Jungner criteria. These developments raise critical legal issues, including the scope of screening, the balance between parental authority and the child's best interests, the adequacy of consent procedures, governance of sample and data retention, and the prevention of genetic discrimination. Internationally, countries such as the United States, the United Kingdom, and Germany remain cautious, keeping genomic sequencing largely within pilot or research programs, reflecting a conservative policy stance. Taiwan faces similar legal and ethical challenges, and its current framework shows gaps in defining screening scope, consent and disclosure mechanisms, and privacy protections. Nonetheless, preliminary recommendations have been

proposed, including the establishment of dedicated review mechanisms, adjustments to consent content, and stronger safeguards for privacy and anti-discrimination. How these measures are implemented will determine Taiwan's ability to both safeguard newborn health and build social trust in the genomic era.

壹、引言

根據美國中央情報局《世界概況》資料顯示，台灣總生育率長期位居全球最低之列[1]，2024年更僅有約0.89[2]。在此少子化與高齡化並行的社會背景下，如何在鼓勵生育的同時，保障新生兒健康，成為國家公共衛生政策不可迴避的課題。

新生兒篩檢（newborn screening, NBS）是全球公認的重要公共衛生措施，能透過早期發現與介入，顯著降低新生兒與幼兒的死亡率與失能[3]。台灣自1980年代推動以代謝疾病為核心的篩檢制度，也已建立起全國性防護網，篩檢率將近100%[4]。然而，隨著基因定序技術的快速進展，新生兒篩檢正逐步邁向「基因世代」[5]。而基因世代的特性在於「不確定性」：疾

1 Total Fertility Rate Comparison - The World Factbook, https://www.cia.gov/the-world-factbook/field/total-fertility-rate/country-comparison/ (last visited Sep. 14, 2025).
2 國家發展委員會，國家發展委員會人口推估查詢系統，https://pop-proj.ndc.gov.tw/Custom_Fast_Statistics_Search.aspx?d=H10&m=83&n=232&sms=10362（瀏覽日期：2025年9月14日）
3 Scott D. Grosse et al., *Progress in Expanding Newborn Screening in the United States*, 110 (6) THE AMERICAN JOURNAL OF HUMAN GENETICS 1015, 2-3 (2023).
4 劉智淵，新生兒篩檢30年，衛生福利部國民健康署，2016年9月，9頁。
5 Ute Spiekerkoetter, David Bick, Richard Scott, Henrietta Hopkins, Tanja Krones, Edith Sky Gross & James R Bonham, *Genomic*