

本期企劃

從優生保健到精準醫療： 論遺傳諮詢師立法之 必要性與法制使命

From Eugenics to Precision Medicine:
The Necessity and Legal Mandate of
Genetic Counselor Legislation

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摘要

遺傳諮詢師是連結醫學遺傳學之資訊與民眾健康決策的重要橋樑。過往遺傳諮詢的服務對象以優生保健與婦幼衛生為主。現今，在精準醫療的浪潮下，其服務範圍擴展至疾病預防、藥物基因學與罕見疾病照護等多元領域。然而，隨著基因檢測走向商業化，專業遺傳諮詢的缺位與認證制度不足，已衍生民眾誤解、醫療爭議與潛在的倫理風險。本文透過比較歐美與亞洲主要國家在遺傳諮詢師的培育、認證與法規保障現況，分析台灣在法制化上的主要缺口，並提出具體政

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關鍵詞：基因檢測倫理（genetic testing ethics）、專業認證（professional certification）、精準醫療（precision medicine）、遺傳諮詢（genetic counseling）、遺傳諮詢師法（genetic counselor legislation）

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策建議，以期建構我國遺傳諮詢師專業之法制基礎，保障民眾基因權益，並鞏固國家醫療品質。

Genetic counselors serve as a critical bridge between medical genetics information and public health decision-making. Historically, genetic counseling services were primarily focused on eugenics and maternal-child health. Today, with the advent of precision medicine, their role has significantly expanded to encompass diverse fields such as disease prevention, pharmacogenomics, and rare disease care. However, as genetic testing becomes commercialized, the lack of professional genetic counseling and insufficient credentialing have led to public misunderstanding, medical disputes, and potential ethical risks. This article reviews the current status of genetic counselor training, certification, and legal protection in major Western and Asian countries, analyzes Taiwan's key gaps in legal system establishment, and proposes specific policy recommendations. The ultimate goal is to establish a legal foundation for the genetic counseling profession in Taiwan, ensuring the protection of citizens' genomic rights and fortifying national healthcare quality.

壹、緒論

台灣遺傳諮詢的起源可追溯至衛生福利部的「優生保健法」¹，主要目的在於協助民眾進行婚、孕前的遺傳風險評估與家庭計畫指導。此階段的遺傳諮詢以「預防遺傳病」與「減少罕病發生率」為導向，與公共衛生政策緊密相關²。

1 台灣衛生福利部「優生保健法」立法背景。

2 台灣公共衛生政策與遺傳疾病防治相關報告。

隨著人類基因體計畫（Human Genome Project, HGP）的完成及次世代基因定序（Next Generation Sequencing, NGS）技術的突飛猛進，遺傳諮詢師（Genetic Counselor, GC）所提供的專業服務，已從過往的優生保健業務，邁向精準醫療（Precision Medicine）的嶄新時代³。在這一關鍵轉變中，遺傳諮詢師的專業地位與法制保障顯得刻不容緩⁴，不僅關乎國民健康福祉，更將深刻影響台灣生醫產業及人民健康的發展。

遺傳諮詢的歷史可追溯至20世紀初的公共衛生與社會改革，當時曾與優生學（eugenics）運動緊密相連，旨在透過選育和（常為非自願的）絕育來改善人類種族⁵。

1947年Dr. Sheldon C. Reed首次提出「遺傳諮詢」一詞，將焦點從優生學轉向遺傳學⁶，說明某些遺傳物質的傳遞情

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- 3 Robin L. Bennett, Heather L. Hampel, Jessica B. Mandell & Joan H Marks, *Genetic Counselors: Translating Genomic Science into Clinical Practice*, 112(9) J CLIN INVEST 1274-1279 (2003). Yamamoto, T., & Nakashima, M. Genetic Counseling and Testing in Japan: Current Challenges and Future Directions, 8(1)JOURNAL OF HUMAN GENETICS e1537 (2020).
 - 4 National Society of Genetic Counselors. National Society of Genetic Counselors, n. d., <https://www.nsgc.org/>. GeneOnline.，職場新趨勢：了解遺傳諮詢師的6件事，2023年5月10日，<https://geneonline.news/6-things-about-genetic-counselors/>（瀏覽日期：2025年10月25日）。
 - 5 Yayoi Aizawa, Atsushi Watanabe, Atsushi Watanab & Kazuto Kato, *Institutional and Social Issues Surrounding Genetic Counselors in Japan : Current Challenges and Implications for the Global Community*, 12 Sec. ELSI IN SCIENCE AND GENETICS (2021); Hong, M. J., & Choi, E., DEVELOPMENT OF A COMPETENCY MODEL for *Genetic Counselors in Korea*. 20(2) JOURNAL OF KOREAN SOCIETY OF DIABETES 111-120 (2019).
 - 6 Gunda Schwaninger, Caroline Benjamin, Sabine Rudnik-Schöneborn & Johannes Zschocke, *The Genetic Counseling Profession in Austria: Stakeholders' Perspectives*, 30(3) JOURNAL OF GENETIC COUNSELING 861-871(2021). Alya A. Qari, Ameera S. Balobaid, Rifaat R. Rawashdeh & Moeenaldeen D. Al Sayed, *The Development of Genetic Counseling Services and Training Program in Saudi Arabia*, 22(6) JOURNAL OF GENETIC COUNSELING 835-838 (2013).