

精準醫學趨勢下 醫療性基因檢測同意書 重要條款

Key Elements of the Consent Forms
for Clinical Genetic Testing in the
Precision Medicine Era

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摘要

為縮小醫病資訊落差、維護民眾權利，本文參考國際學術界、產業界基因檢測同意書，列舉並討論重要條款如下：一、檢測項目範圍；二、檢測目的與利益；三、檢測方法、流程及限制；四、替代診斷方法；五、檢測報告內容、結果類別；六、臨床意義不明的基因變異；七、無預期資訊、偶然發現、未經請求的發現；八、風險；九、隱私與保密；十、是否告知家屬；十一、病歷保存；十二、檢體保存；十三、資料分享；十四、再接觸；十五、本人死亡後資料、檢體處理方式；十六、檢測志願性；十七、撤回同意／變更意願／不知情權。

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關鍵詞：同意書（consent form）、知情同意（informed consent）、病人自主權利法（Patient Right to Autonomy Act）、基因檢測（genetic testing）、精準醫學（precision medicine）

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Genetic testing is a vital tool in precision medicine. To address the information gap between healthcare institutions and patients while protecting patients' rights, this article examines the design of genetic testing consent forms proposed by international experts, outlining and discussing key clauses as follows: 1. Scope; 2. Benefits; 3. Procedure and Limitations; 4. Alternative Diagnostic Methods; 5. What is Reported, Types of Results; 6. Variants of Uncertain Significance (VUS); 7. Unexpected Information/Incidental Findings/Unsolicited Findings; 8. Risks; 9. Privacy and Confidentiality; 10. To inform relatives or not; 11. Health Records; 12. Sample Storage; 13. Data Sharing; 14. Recontact; 15. In Case of Death; 16. Voluntary; 17. Withdrawal/Change of Mind/Right not to Know.

壹、基因檢測之價值與類型

精準醫學（Precision Medicine）在現代醫療扮演日益重要角色，是指根據個人基因、環境及生活形態個別差異進行治療及預防疾病。¹精準醫學可為個人量身訂做治療方案而改變醫學。藉由在正確時間提供正確治療，醫療照護得以更精確且更有效率，同時為病人提供更高品質照護。而將基因體與其他分子資訊納入常規醫療照護對於精準醫學醫療系統至關重要。²

目前精準醫學在預防、診斷、治療等臨床應用方面，主

1 LISTER HILL NATIONAL CENTER FOR BIOMEDICAL COMMUNICATIONS, U.S. NATIONAL LIBRARY OF MEDICINE, NATIONAL INSTITUTES OF HEALTH & DEPARTMENT OF HEALTH & HUMAN SERVICES., PRECISION MEDICINE 3 (2016).

2 Victor J Dzau & Geoffrey S Ginsburg, *Realizing the Full Potential of Precision Medicine in Health and Health Care*, 316 JAMA 1659, 1659 (2016).